Supplementary Material for

Predicting the functional effects of voltage-gated potassium channel missense variants with multi-task learning

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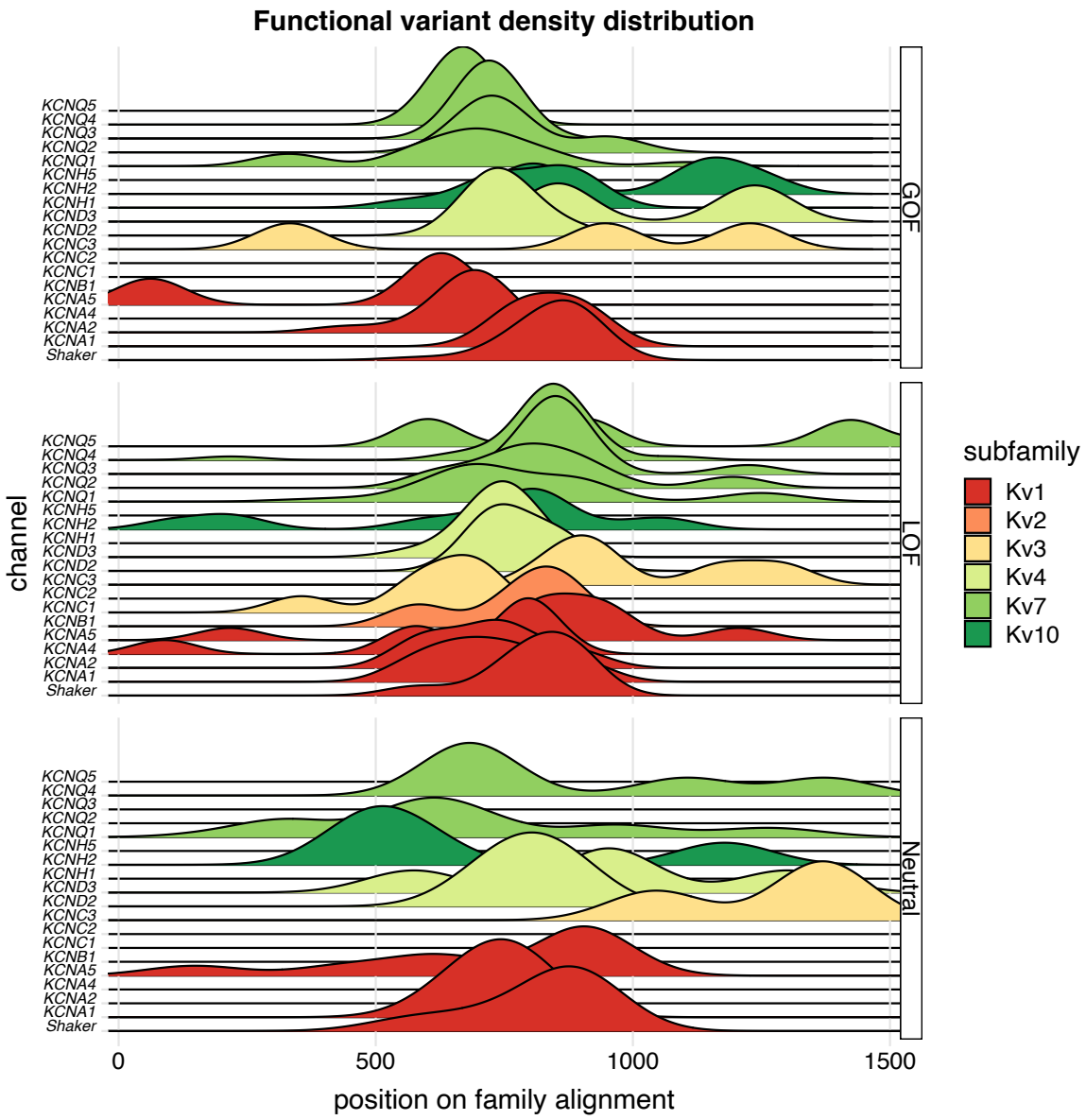
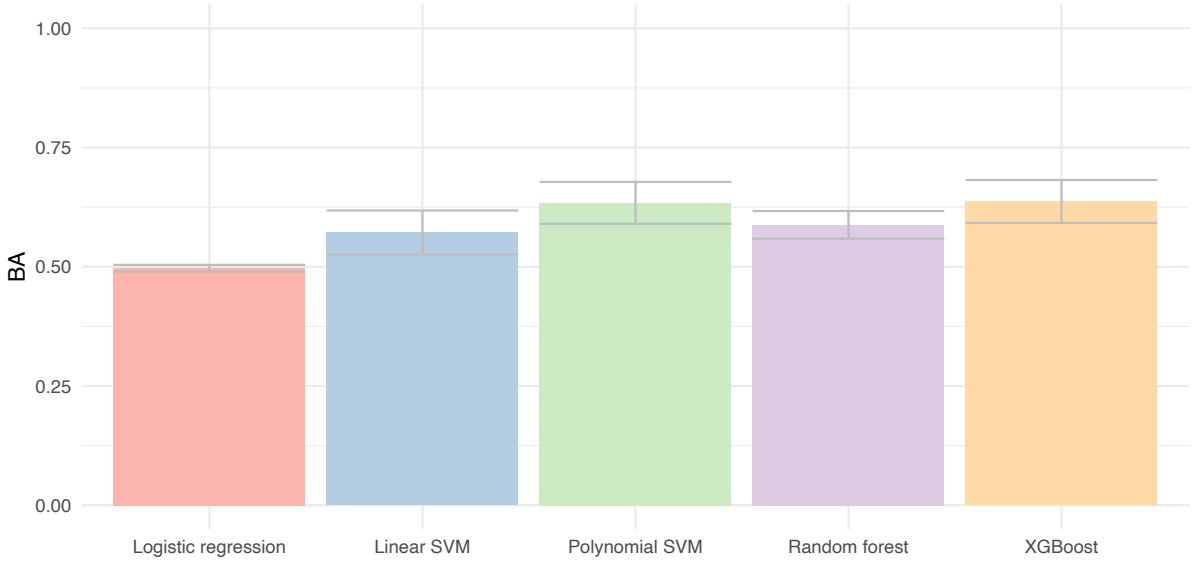
[†] These authors were equally contributing PIs of this work.

Contents



Supplementary Figure 1. Functional variant density distribution along the channel superfamily multiple sequence alignment. Variants are grouped by functional effect (GOF, LOF, and neutral) and coloured by channel subfamily membership.



Supplementary Figure 2. Performance of simple algorithms (logistic regression) and state-of-the-art algorithms that were used in previous work (XGBoost).[1] Models were implemented through the tidymodels framework.[2] Model hyperparameter tuning and performance estimation were carried out with nested k-fold cross validation. Logistic regression, BA 0.497 ± 0.007. Linear SVM, BA 0.572 ± 0.046. Polynomial SVM, BA 0.634 ± 0.044. Random Forest, BA 0.588 ± 0.029. XGBoost, BA 0.637 ± 0.047. Abbreviations: BA – balanced accuracy.

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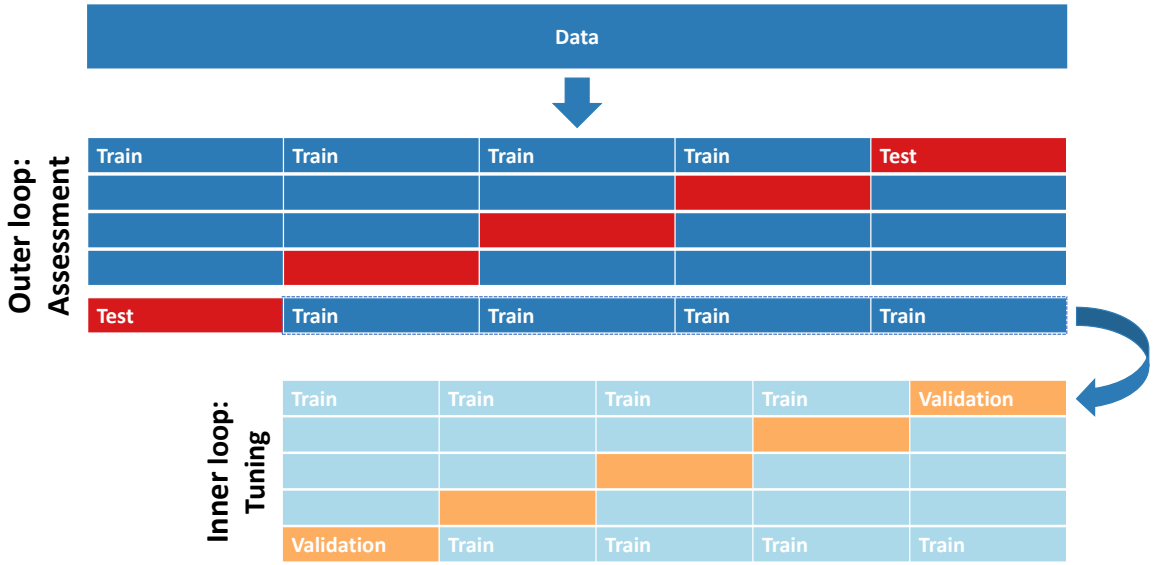


Supplementary Figure 3. Graphical representation of the nested k-fold cross-validation procedure used during model selection (hyperparameter tuning) and model assessment.

Algorithm 1: Nested k-fold cross-validation

Input: Data set S, set of hyperparameters C

Output: Average error for best parameter combination

K ← Randomly split S into outer_folds folds

FOR i = 1 **TO** outer_folds *# outer loop*

outer_test ← $k_i \in K$

outer_train ← K \ k_i

FOR hyperparameter **IN** C *# inner loop*

H ← Randomly split outer_train into inner_folds folds

FOR j = 1 **TO** inner_folds

inner_test ← $h_j \in H$

inner_train ← H \ h_j

model ← Train model on inner_train with hyperparameter

e_j ← Compute test error of model on inner_test

ENDFOR

$\bar{e}$ ← Compute average e_j (j = 1, 2, ..., inner_folds)

ENDFOR

best_hyperparameter ← select hyperparameter with minimum $\bar{e}$

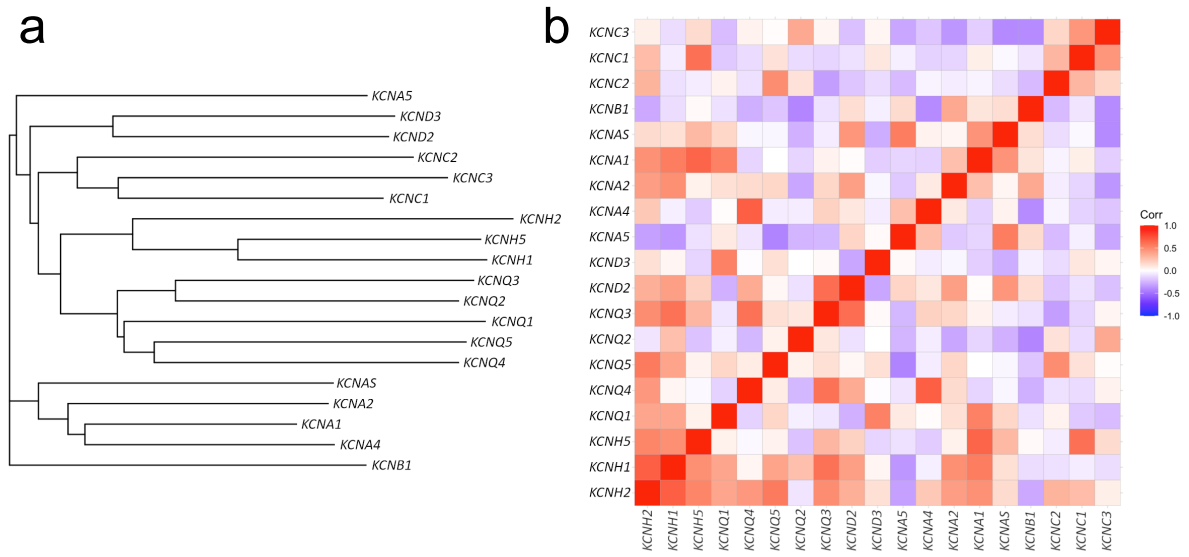
model ← Train model with hyperparameter on outer_train

f_i ← Compute test error of model on outer_test

ENDFOR

RETURN $\bar{f}$ ← Compute average f_i (i = 1, 2, ..., outer_folds)

Supplementary Figure 4. Pseudocode representation of the nested k-fold cross-validation procedure used during model selection (hyperparameter tuning) and model assessment.



Supplementary Figure 5. *a*: Task hierarchy T derived from MUSCLE multiple sequence alignment. *b*: Visualization of the task similarity matrix K_t as a correlation plot, where positive values (shown in red) correspond to a lower pairwise distance and higher pairwise similarity, and vice versa.

Feature	Description	Data type	p_{GOF}	p_{LOF}	$p_{Neutral}$	Reference
aa_blosum62	amino acid substitution value according to the midrange Blocks Substitution Matrix (BLOSUM62)	numerical	0.495	**0.020**	**0.029**	[1]
aa_blomap_aa1_dim1	encoding of the physicochemical properties of the original amino acid using a Naïve Bayes classifier, first encoding dimension	numerical	0.955	0.858	0.495	[2]
aa_blomap_aa1_dim2	encoding of the physicochemical properties of the original amino acid using a Naïve Bayes classifier, second encoding dimension	numerical	0.879	0.408	0.449	[2]
aa_blomap_aa1_dim3	encoding of the physicochemical properties of the original amino acid using a Naïve Bayes classifier, third encoding dimension	numerical	0.879	0.655	0.919	[2]
aa_blomap_aa1_dim4	encoding of the physicochemical properties of the original amino acid using a Naïve Bayes classifier, fourth encoding dimension	numerical	0.879	0.323	0.367	[2]
aa_blomap_aa1_dim5	encoding of the physicochemical properties of the original amino acid using a Naïve Bayes classifier, fifth encoding dimension	numerical	0.910	0.651	0.840	[2]
aa_blomap_aa2_dim1	encoding of the physicochemical properties of the substituted amino acid using a Naïve Bayes classifier, first encoding dimension	numerical	0.980	0.907	0.670	[2]
aa_blomap_aa2_dim2	encoding of the physicochemical properties of the substituted amino acid using a Naïve Bayes classifier, second encoding dimension	numerical	0.897	0.458	0.488	[2]
aa_blomap_aa2_dim3	encoding of the physicochemical properties of the substituted amino acid using a Naïve Bayes classifier, third encoding dimension	numerical	0.910	0.614	0.703	[2]
aa_blomap_aa2_dim4	encoding of the physicochemical properties of the substituted amino acid using a Naïve Bayes classifier, fourth encoding dimension	numerical	0.910	0.648	0.110	[2]
aa_blomap_aa2_dim5	encoding of the physicochemical properties of the substituted amino acid using a Naïve Bayes classifier, fifth encoding dimension	numerical	0.505	0.161	0.421	[2]
aa_braun_aa1_E1	encoding of the physicochemical properties of the original amino acid using multidimensional scaling, first encoding dimension	numerical	0.989	0.931	0.762	[3]
aa_braun_aa1_E2	encoding of the physicochemical properties of the original amino acid using multidimensional scaling, second encoding dimension	numerical	0.997	0.537	0.231	[3]
aa_braun_aa1_E3	encoding of the physicochemical properties of the original amino acid using multidimensional scaling, third encoding dimension	numerical	0.997	0.968	0.941	[3]
aa_braun_aa1_E4	encoding of the physicochemical properties of the original amino acid using multidimensional scaling, fourth encoding dimension	numerical	0.910	0.740	0.941	[3]
aa_braun_aa1_E5	encoding of the physicochemical properties of the original amino acid using multidimensional scaling, fifth encoding dimension	numerical	0.669	0.651	0.919	[3]
aa_braun_aa2_E1	encoding of the physicochemical properties of the substituted amino acid using multidimensional scaling, first encoding dimension	numerical	0.712	0.305	0.423	[3]
aa_braun_aa2_E2	encoding of the physicochemical properties of the substituted amino acid using multidimensional scaling, second encoding dimension	numerical	0.578	0.931	0.137	[3]
aa_braun_aa2_E3	encoding of the physicochemical properties of the substituted amino acid using multidimensional scaling, third encoding dimension	numerical	0.381	0.072	0.386	[3]
aa_braun_aa2_E4	encoding of the physicochemical properties of the substituted amino acid using multidimensional scaling, fourth encoding dimension	numerical	0.955	0.931	0.623	[3]

aa_braun_aa2_E5	encoding of the physicochemical properties of the substituted amino acid using multidimensional scaling, fifth encoding dimension	numerical	0.669	0.648	0.931	[3]
aa_grantham	Grantham distance between the original and substituted amino acids, based on composition, polarity and residue volume	numerical	0.955	0.502	0.066	[4]
aa_hphob_pca1	change in hydrophobicity by amino acid substitution using 98 hydrophobicity scales and principal component analysis, first component	numerical	0.955	0.740	0.794	[5]
aa_hphob_pca2	change in hydrophobicity by amino acid substitution using 98 hydrophobicity scales and principal component analysis, first component	numerical	0.980	0.621	0.442	[5]
aa_hphob_pca3	change in hydrophobicity by amino acid substitution using 98 hydrophobicity scales and principal component analysis, first component	numerical	0.980	0.855	0.873	[5]
str_np_rsa	change in residue relative accessible surface area predicted by NetSurfP – 2.0	numerical	0.381	**<0.001**	**<0.001**	[6]
str_np_asa	change in residue accessible surface area predicted by NetSurfP – 2.0	numerical	0.381	**0.012**	0.054	[6]
str_paraz	amino acid conservation across gene paralogs (Parazscore)	numerical	0.076	**0.017**	0.832	[7]
str_iupred	energy estimation-based structural state prediction for ordered and disordered residues by IUPred2	numerical	0.989	0.059	**<0.001**	[8]
str_anchor	energy estimation-based structural state prediction for disordered binding regions by ANCHOR2	numerical	0.669	**0.012**	**0.002**	[8]
str_asa	change in residue accessible surface area predicted by PROF via PredictProtein	numerical	0.997	0.858	0.747	[9]
str_rsa	change in residue relative accessible surface area predicted by PROF via PredictProtein	numerical	0.955	0.931	0.595	[9]
str_helix	prediction of topology for helical transmembrane proteins by PHDhtm_top via PredictProtein, network output for helix	numerical	0.796	0.305	**0.001**	[9]
str_loop	prediction of topology for helical transmembrane proteins by PHDhtm_top via PredictProtein, network output for loop	numerical	0.780	0.310	**0.001**	[9]
str_nors	prediction of protein disorder by NORSnet via PredictProtein	numerical	0.879	0.261	**0.001**	[9,10]
str_profbval	prediction of protein disorder by PROFbval via PredictProtein	numerical	0.414	**0.002**	**0.001**	[9,11]
str_ucon	prediction of protein disorder by Ucon via PredictProtein	numerical	0.268	0.323	**<0.001**	[9,12]
str_isis	prediction of residue involvement in protein-protein interaction sites by ISIS	numerical	0.381	**<0.001**	**<0.001**	[13]
str_consurf	probabilistic positional evolutionary conservation estimates by ConSurf via PredictProtein	numerical	0.505	**0.002**	**<0.001**	[9,14]
ecdf	relative variant position along sequence length (empirical distribution function)	numerical	0.268	0.124	0.941	–

cid	relative variant position on the gene family multiple sequence alignment via MUSCLE with UpgmaMax clustering	numerical	0.712	0.855	0.641	[15]
str_pbie_b	state of residue relative accessible surface area predicted by PROF via PredictProtein, buried	categorical	0.955	0.561	0.451	[9,11]
str_pbie_e	state of residue relative accessible surface area predicted by PROF via PredictProtein, exposed	categorical	0.997	0.655	0.423	[9,11]
str_pbie_i	state of residue relative accessible surface area predicted by PROF via PredictProtein, intermediate	categorical	0.980	0.430	0.200	[9,11]
str_prhl_H	maximum score model prediction of topology for helical transmembrane proteins by PHDhtm_top via PredictProtein, helical membrane	categorical	0.967	0.716	0.363	[9]
str_prhl_L	maximum score model prediction of topology for helical transmembrane proteins by PHDhtm_top via PredictProtein, no helical transmembrane	categorical	0.967	0.716	0.363	[9]
str_pito_i	predicted topology of transmembrane regions by PHDhtm_top via PredictProtein, loop inside	categorical	0.505	**0.014**	**0.010**	[9]
str_pito_o	predicted topology of transmembrane regions by PHDhtm_top via PredictProtein, loop outside	categorical	0.394	0.053	0.190	[9]
str_pito_T	predicted topology of transmembrane regions by PHDhtm_top via PredictProtein, transmembrane	categorical	0.967	0.716	0.363	[9]
str_top_cytoplasmic	position on protein topology via UniProt, cytoplasmic	categorical	0.997	**0.009**	**<0.001**	[16]
str_top_extracellular	position on protein topology via UniProt, cytoplasmic	categorical	0.952	0.609	0.592	[16]
str_top_porehelix	position on protein topology via UniProt, cytoplasmic	categorical	0.268	**0.002**	**0.029**	[16]
str_top_S1	position on protein topology via UniProt, segment S1	categorical	0.381	0.931	0.106	[16]
str_top_S2	position on protein topology via UniProt, segment S2	categorical	0.381	0.323	0.994	[16]
str_top_S3	position on protein topology via UniProt, segment S3	categorical	0.935	0.968	0.623	[16]
str_top_S4	position on protein topology via UniProt, segment S4	categorical	0.381	0.949	**0.048**	[16]
str_top_S5	position on protein topology via UniProt, segment S5	categorical	0.669	0.124	0.200	[16]
str_top_S6	position on protein topology via UniProt, segment S6	categorical	**0.009**	0.117	0.114	[16]
str_exp_calmodulin	expert and literature-based annotation of variant position within functional domains or motifs, calmodulin binding site	categorical	0.955	0.848	0.941	[17-26]
str_exp_gating	expert and literature-based annotation of variant position within functional domains or motifs, gating charges or countercharges	categorical	0.381	0.957	0.085	

str_exp_none	expert and literature-based annotation of variant position within functional domains or motifs, none	categorical	0.381	0.968	**0.023**	
str_exp_pvp	expert and literature-based annotation of variant position within functional domains or motifs, PVP motif	categorical	0.381	0.813	0.229	
str_exp_S4S5linker	expert and literature-based annotation of variant position within functional domains or motifs, S4-S5 linker	categorical	0.505	0.502	0.941	
str_exp_selectivityfilter	expert and literature-based annotation of variant position within functional domains or motifs, selectivity filter	categorical	0.381	0.059	0.328	
str_exp_selfassociation	expert and literature-based annotation of variant position within functional domains or motifs, self-association site	categorical	0.381	0.322	0.873	
str_np_q3_C	3-class secondary structure classification (SS3) by NetSurfP – 2.0 and DSSP	categorical	0.952	**0.012**	**<0.001**	[6,27]
str_np_q3_E	3-class secondary structure classification (SS3) by NetSurfP – 2.0 and DSSP	categorical	0.381	0.117	0.488	[6,27]
str_np_q3_H	3-class secondary structure classification (SS3) by NetSurfP – 2.0 and DSSP	categorical	0.997	0.053	**<0.001**	[6,27]
str_np_q8_B	8-class secondary structure classification (SS8) by NetSurfP – 2.0 and DSSP	categorical	0.997	0.813	0.603	[6,27]
str_np_q8_C	8-class secondary structure classification (SS8) by NetSurfP – 2.0 and DSSP	categorical	0.405	**<0.001**	**<0.001**	[6,27]
str_np_q8_E	8-class secondary structure classification (SS8) by NetSurfP – 2.0 and DSSP	categorical	0.381	0.470	0.623	[6,27]
str_np_q8_G	8-class secondary structure classification (SS8) by NetSurfP – 2.0 and DSSP	categorical	0.450	0.502	0.873	[6,27]
str_np_q8_H	8-class secondary structure classification (SS8) by NetSurfP – 2.0 and DSSP	categorical	0.910	0.083	**<0.001**	[6,27]
str_np_q8_I	8-class secondary structure classification (SS8) by NetSurfP – 2.0 and DSSP	categorical	0.935	0.072	**0.010**	[6,27]
str_np_q8_S	8-class secondary structure classification (SS8) by NetSurfP – 2.0 and DSSP	categorical	0.709	0.619	0.992	[6,27]
str_np_q8_T	8-class secondary structure classification (SS8) by NetSurfP – 2.0 and DSSP	categorical	0.381	0.074	0.387	[6,27]

Supplementary Table 1: List of features used in model training, sorted by name and appearance in the training data set. Group differences were compared using the Wilcoxon–Mann–Whitney test two-sample test, with a one-versus-all approach for three classes. p-values were adjusted for multiple comparisons using the Benjamini-Hochberg method. Significant p-values after correction are shown in bold and indicate a non-directional difference in group distribution (i.e., a discriminative feature).

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Gene	Original amino acid	Position	Substituted amino acid	Net overall change in biophysical function	Change in voltage-dependence of activation [$\Delta V_{1/2}$, mV]	Change in slope factor of activation [Δk]	Change in time constant of deactivation [$\Delta \tau$, ms]	Change in slope factor of (fast) inactivation [Δk]	Change in voltage-dependence of (fast) inactivation [$\Delta V_{1/2}$, mV]	Change in amplitude of peak current or conductance [nS, pA/pF]	Loss of cell current (total loss-of-function)	Change in expression level	Comment	DOI
KCNA1	A	242	P	LOF	-4.8	-0.4	4.2			0		none		10.1002/1531-8249(200010)48:4<647::AID-ANA12>3.0.CO;2-Q.
KCNA1	A	395	L	LOF	3									10.1113/jphysiol.2009.174730
KCNA1	C	185	W	LOF	61.2					-41		none	faster recovery from slow inactivation	10.3389/fphys.2014.00525
KCNA1	C	185	W	LOF	0.8	-1.8				-35				10.1136/jnnp-2012-304131
KCNA1	E	283	K	LOF	4.4	1.6	3.5			49			slower activation	10.1016/j.mcn.2017.06.006
KCNA1	E	325	D	LOF	60.4	9.4	-19.3			-0.96		reduced		10.1523/JNEUROSCI.18-08-02842.1998
KCNA1	E	325	D	LOF	21.8	4.3	19.9							10.1096/fasebj.13.11.1335
KCNA1	F	184	C	LOF	28	0	6			-2.5		reduced	normal tau of inactivation	10.1096/fasebj.13.11.1335
KCNA1	F	184	C	LOF	25.9	-1	4.6			-0.85				10.1523/JNEUROSCI.18-08-02842.1998
KCNA1	F	232	L	LOF	3									10.1113/jphysiol.2009.174730
KCNA1	F	249	C	LOF	0					-2		reduced		10.1007/s10048-016-0486-0
KCNA1	F	249	I	LOF	-0.6	-1.9	21.9			-0.99		reduced		10.1523/JNEUROSCI.18-08-02842.1998
KCNA1	F	303	V	LOF	12	3	-4	1	4	-6.3				10.1038/s41598-017-03041-z
KCNA1	F	411	L	LOF								not expressed		10.1113/jphysiol.2009.174730

KCNA1	F	411	Y	LOF								not expressed		10.1113/jphysiol.2009.174730
KCNA1	F	414	C	Mixed	2.2	1.3	-2.3			-0.7			Faster recovery from inactivation, faster inactivation	10.1016/j.neuroscience.2008.09.022
KCNA1	F	414	L	GOF	-17								Two phases in the I-V curve	10.1113/jphysiol.2009.174730
KCNA1	F	414	S	LOF	14.9					-79.9		none	Two phases in the I-V curve	10.1212/WNL.0b013e3181ea9ee3
KCNA1	F	414	Y	LOF	13								Two phases in the I-V curve	10.1113/jphysiol.2009.174730
KCNA1	G	311	D	LOF						-0.5		reduced		10.3389/fneur.2018.00587
KCNA1	G	311	S	LOF	31.7	5.3	-0.4			-0.75		reduced		10.1016/S0014-5793(98)00814-X
KCNA1	G	316	A	Neutral	0									10.1113/jphysiol.2009.174730
KCNA1	I	177	L	LOF						-0.92		increased		10.1074/jbc.274.17.11487.
KCNA1	I	177	N	LOF	59	1.7	-28			-0.94				10.1007/s00424-002-0962-2
KCNA1	I	177	P	LOF						-0.06		reduced		10.1074/jbc.274.17.11487.
KCNA1	I	177	R	LOF						-0.97				10.1074/jbc.274.17.11487.
KCNA1	I	262	L	LOF						-2.67		increased		10.1016/j.abb.2012.05.006
KCNA1	I	262	M	LOF						-172		none		10.1002/mus.24242
KCNA1	I	262	T	LOF	12.8	1.1		-0.3	9.5	-1.95		reduced		10.1038/srep19378
KCNA1	I	262	T	LOF	12.8	1.1		-0.3	9.5	-1.95		reduced		10.1016/j.abb.2012.05.006
KCNA1	I	314	L	LOF								not expressed		10.1113/jphysiol.2009.174730
KCNA1	I	330	V	LOF	4									10.1113/jphysiol.2009.174730
KCNA1	I	400	V	GOF									20fold increase in recovery rate	10.1038/nsmb825
KCNA1	I	407	L	LOF	52									10.1113/jphysiol.2009.174730
KCNA1	I	407	M	LOF	21.4					-32.7				10.1136/jnnp-2012-304131
KCNA1	L	305	I	LOF	53									10.1113/jphysiol.2009.174730
KCNA1	L	312	M	LOF	12									10.1113/jphysiol.2009.174730

KCNA1	L	315	V	LOF	9									10.1113/jphysiol.2009.174730
KCNA1	L	319	R	LOF	12	0		0	21.5	-140.3				10.1093/hmg/ddx430
KCNA1	L	319	V	GOF	-4	0								10.1113/jphysiol.2009.174730
KCNA1	L	326	V	GOF	-23									10.1113/jphysiol.2009.174730
KCNA1	L	328	V	LOF						-209		none		10.1159/000488954
KCNA1	L	329	I	LOF										10.1016/j.abb.2012.05.006
KCNA1	L	329	V	Unclear									does not saturate	10.1113/jphysiol.2009.174730
KCNA1	L	333	V	GOF	-10									10.1113/jphysiol.2009.174730
KCNA1	L	398	A	LOF								not expressed		10.1113/jphysiol.2009.174730
KCNA1	M	323	L	Neutral	0	0								10.1113/jphysiol.2009.174730
KCNA1	N	255	A	Mixed	-12.1	1.6			-13.2					10.1074/jbc.M109.041517
KCNA1	N	255	D	LOF	31					-326		none		10.1172/JCI36948
KCNA1	N	255	E	LOF										10.1074/jbc.M109.041517
KCNA1	N	255	H	Mixed	-4.4	0.2			-6.1					10.1074/jbc.M109.041517
KCNA1	N	255	K	LOF	24	0		0	-22	-124.66				10.1093/hmg/ddx430
KCNA1	N	255	Q	LOF							no current			10.1074/jbc.M109.041517
KCNA1	N	255	T	Mixed	-21.1	-0.9			-13.4					10.1074/jbc.M109.041517
KCNA1	N	255	V	Mixed	-16.1	1.8			-13.3					10.1074/jbc.M109.041517
KCNA1	P	244	H	Neutral	-0.7	0.3	0.7			0		none		10.1002/1531-8249(200010)48:4<647::AID-ANA12>3.0.CO;2-Q
KCNA1	R	167	M	LOF	55.6					-41.6		none		10.1136/jnnp-2012-304131
KCNA1	R	239	S	LOF						0		reduced		10.1523/JNEUROSCI.18-08-02842.1998
KCNA1	R	307	C	LOF	11.4					-64.7		none		10.1212/WNL.0b013e3181ea9ee3

KCNA1	R	324	T	Mixed	20	0	0	0	20	-57		none	slow activation (2-4fold)	10.1002/mds.26737
KCNA1	S	180	L	LOF						-0.94				10.1074/jbc.274.17.11487.
KCNA1	S	180	P	LOF						-0.89				10.1074/jbc.274.17.11487.
KCNA1	S	180	R	LOF						-0.94				10.1074/jbc.274.17.11487.
KCNA1	S	342	I	LOF						-2.87		none		10.1016/j.abb.2012.05.006
KCNA1	S	342	T	LOF						-2.41		increased		10.1016/j.abb.2012.05.006
KCNA1	T	226	A	LOF	14.3	-2.1	28.4	0	0	-0.95		reduced		10.1016/S0014-5793(98)00814-X
KCNA1	T	226	K	LOF						-0.34				10.1007/s10048-006-0071-z
KCNA1	T	226	M	LOF	14.8	-2	42.2	0	0	-0.95				10.1016/S0014-5793(98)00814-X
KCNA1	T	226	R	LOF	-25	-3.4	5			-3.7		none		10.1111/j.1749-6632.1999.tb11310.x
KCNA1	T	226	R	LOF	-25	-3.4	5			-3.7		none		10.1093/brain/122.5.817
KCNA1	T	318	I	LOF								not expressed		10.1113/jphysiol.2009.174730
KCNA1	V	174	F	LOF	35.1	-2	3.2			-0.92			faster inactivation	10.1523/JNEUROSCI.18-08-02842.1998
KCNA1	V	229	I	LOF	54.1					-23.6				10.1212/WNL.0b013e3181b87959
KCNA1	V	404	I	LOF	11.3	1.9	7.2			0		none		10.1002/1531-8249(200010)48:4<647::AID-ANA12>3.0.CO;2-Q
KCNA1	V	406	L	GOF	-30									10.1113/jphysiol.2009.174730
KCNA1	V	408	A	LOF	1	0	-16.8			-1.4		reduced	faster inactivation	10.1046/j.1460-9568.1999.00659.x
KCNA1	V	408	A	LOF	0.4	-0.6	-20.7			-0.5			faster inactivation	10.1523/JNEUROSCI.18-08-02842.1998
KCNA1	V	408	A	LOF	0.9	1.9	-3.6							10.1096/fasebj.13.11.1335
KCNA1	V	408	L	LOF								none		10.1113/jphysiol.2009.174730

KCNA1	V	408	L	LOF								not expressed		10.1002/mds.22467
KCNA2	E	157	K	GOF	-10	3.5		0.8	1.9	3.7				10.1093/brain/awx184
KCNA2	E	183	A	LOF	30	0				0				10.1016/j.bpj.2012.03.032
KCNA2	E	183	D	LOF						0	no current			10.1016/j.bpj.2012.03.032
KCNA2	E	236	A	GOF	-15					0				10.1016/j.bpj.2012.03.032
KCNA2	E	236	D	GOF	-15	0				0				10.1016/j.bpj.2012.03.032
KCNA2	E	236	L	Unclear	0	0				0				10.1016/j.bpj.2012.03.032
KCNA2	F	180	W	LOF							no current			10.1016/j.bpj.2012.03.032
KCNA2	F	180	Y	GOF	0									10.1016/j.bpj.2012.03.032
KCNA2	G	398	C	LOF	0	0			0	-0.92				10.1093/brain/awx184
KCNA2	I	263	T	LOF	9	4.7		-1.3	-0.2	-0.87		none		10.1038/ng.3239
KCNA2	K	306	Q	LOF	2									10.1016/j.bpj.2012.03.032
KCNA2	K	322	A	LOF	30	0								10.1016/j.bpj.2012.03.032
KCNA2	K	322	L	LOF						0				10.1016/j.bpj.2012.03.032
KCNA2	L	290	R	GOF	-9.9	5.2		-2.1	-23.1	1.6				10.1093/brain/awx184
KCNA2	L	293	H	GOF	-46.5	-2.1		-1.3	-58.4	-0.1				10.1093/brain/awx184
KCNA2	L	298	F	GOF	-43.2	3.03			0	12.3		none		10.1038/ng.3239
KCNA2	L	328	V	Mixed	-19.7	3.3		-0.6	-2.9	-0.68				10.1093/brain/awx184
KCNA2	N	207	Q	LOF	6.1	3.3	110			0			Tau fast of inactivation significantly increased	10.1016/j.brainres.2007.01.092
KCNA2	P	405	L	LOF	-1	0		0.2	-4.5	-0.86		none		10.1038/ng.3239
KCNA2	Q	357	R	Neutral	-2.3	-0.4		-0.4	-1.9	-0.1				10.1093/brain/awx184
KCNA2	R	294	H	LOF	6.2	4.2			0	-0.9				10.1002/ana.24762
KCNA2	R	294	Q	LOF	-15									10.1085/jgp.201411300

KCNA2	R	294	Q	GOF	25									10.1016/j.bpj.2012.03.032
KCNA2	R	297	Q	GOF	-20	0						none		10.1085/jgp.201411300
KCNA2	R	297	Q	GOF	-38.3	1.7			0	6.4				10.1016/j.bpj.2012.03.032
KCNA2	R	297	Q	GOF	-20									10.1038/ng.3239
KCNA2	R	300	N	GOF	-5									10.1085/jgp.201411300
KCNA2	R	300	Q	LOF						0				10.1016/j.bpj.2012.03.032
KCNA2	R	303	Q	GOF	0	0								10.1085/jgp.201411300
KCNA2	R	303	Q	LOF						0				10.1016/j.bpj.2012.03.032
KCNA2	T	252	C	GOF	-32.8	2								10.1529/biophysj.107.116160
KCNA2	T	252	D	GOF	-9	3.7								10.1529/biophysj.107.116160
KCNA2	T	252	K	GOF	-29.1	4.6				0				10.1529/biophysj.107.116160
KCNA2	T	252	R	GOF	-30	4.9								10.1529/biophysj.107.116160
KCNA2	T	374	A	Mixed	-19.9	0.7		-1.1	-2.7	-0.86				10.1093/brain/awx184
Shaker	A	355	K	Mixed	10	0		0	10					10.1074/jbc.M111.237792
Shaker	A	355	R	Mixed	10	0		0	10					10.1074/jbc.M111.237792
Shaker	A	359	K	Mixed	25	0			30					10.1074/jbc.M111.237792
Shaker	A	359	R	Mixed	25	0		0	25					10.1074/jbc.M111.237792
Shaker	A	391	V	GOF	-15.7									10.1016/s0092-8674(02)01013-9
Shaker	A	413	V	LOF	1									10.1016/s0092-8674(02)01013-9
Shaker	A	413	W	LOF	12.4									10.1016/j.neuron.2006.10.005
Shaker	A	417	V	GOF	-0.9									10.1016/s0092-8674(02)01013-9
Shaker	A	417	W	LOF							no current			10.1016/j.neuron.2006.10.005
Shaker	A	419	V	GOF	-2.5									10.1016/s0092-8674(02)01013-9
Shaker	A	432	V	GOF	-3.5									10.1016/s0092-8674(02)01013-9

Shaker	A	436	V	GOF	-18									10.1016/s0092-8674(02)01013-9
Shaker	A	463	C	LOF	14							none		10.1085/jgp.112.2.243
Shaker	A	463	V	LOF	-0.9									10.1016/s0092-8674(02)01013-9
Shaker	A	463	W	LOF							no current			10.1016/j.neuron.2006.10.005
Shaker	A	465	V	GOF	-19.8									10.1016/s0092-8674(02)01013-9
Shaker	A	471	D	Unclear	-6.5								Two phases in the I-V curve	10.1085/jgp.200285 69.
Shaker	A	471	V	LOF								not expressed		10.1016/s0092-8674(02)01013-9
Shaker	A	471	W	LOF	6.7									10.1085/jgp.200285 69.
Shaker	C	462	A	Neutral	0.5									10.1016/s0092-8674(02)01013-9
Shaker	C	462	W	LOF							no current			10.1016/j.neuron.2006.10.005
Shaker	D	316	N	LOF	67									10.1016/0896-6273(95)90276-7
Shaker	D	431	A	GOF	-2.7									10.1016/s0092-8674(02)01013-9
Shaker	D	447	N	LOF							no current			10.1016/0896-6273(95)90276-7
Shaker	E	283	Q	LOF	78	10.6								10.1016/0896-6273(95)90276-7
Shaker	E	293	Q	GOF	-17	0.9								10.1016/0896-6273(95)90276-7
Shaker	E	395	A	GOF	-33.7									10.1016/s0092-8674(02)01013-9
Shaker	E	395	W	LOF	210.9									10.1016/j.neuron.2006.10.005
Shaker	E	422	A	GOF	-0.7									10.1016/s0092-8674(02)01013-9
Shaker	E	488	C	Neutral	0									10.1085/jgp.200286 11.
Shaker	F	244	W	LOF	40									10.1085/jgp.201210827
Shaker	F	370	M	GOF	-22.1	-2.3								10.1085/jgp.111.3.421
Shaker	F	401	A	Unclear									two phases in the I-V curve	10.1016/s0092-8674(02)01013-9
Shaker	F	402	A	Unclear									two phases in the I-V curve	10.1016/s0092-8674(02)01013-9

Shaker	F	404	A	LOF	1.9									10.1016/s0092-8674(02)01013-9
Shaker	F	410	A	GOF	-1.9									10.1016/s0092-8674(02)01013-9
Shaker	F	416	A	GOF	-4.4									10.1016/s0092-8674(02)01013-9
Shaker	F	416	A	LOF	4.3									10.1016/j.neuron.2006.10.005
Shaker	F	425	A	GOF	-1.6									10.1016/s0092-8674(02)01013-9
Shaker	F	426	A	GOF	-4.2									10.1016/s0092-8674(02)01013-9
Shaker	F	433	A	LOF	1.6									10.1016/s0092-8674(02)01013-9
Shaker	F	481	A	LOF								not expressed		10.1016/s0092-8674(02)01013-9
Shaker	F	481	D	LOF								not expressed		10.1085/jgp.200285 69.
Shaker	F	481	W	GOF	-13.2									10.1085/jgp.200285 69.
Shaker	F	484	A	Unclear									two phases in the I-V curve	10.1016/s0092-8674(02)01013-9
Shaker	F	484	A	GOF	-8.3									10.1085/jgp.200285 69.
Shaker	F	484	C	LOF	13.4									10.1085/jgp.200286 11.
Shaker	F	484	D	GOF	-22.1									10.1085/jgp.200285 69.
Shaker	F	484	W	GOF	-8.5									10.1085/jgp.200285 69.
Shaker	G	397	A	LOF	0.8									10.1016/s0092-8674(02)01013-9
Shaker	G	397	W	GOF	-14.8									10.1016/j.neuron.2006.10.005
Shaker	G	406	A	LOF	10.9									10.1016/s0092-8674(02)01013-9
Shaker	G	420	A	LOF	6.2									10.1016/s0092-8674(02)01013-9
Shaker	G	452	A	LOF	2									10.1016/s0092-8674(02)01013-9
Shaker	G	455	A	LOF	1									10.1016/s0092-8674(02)01013-9
Shaker	G	455	W	LOF							no current			10.1016/j.neuron.2006.10.005
Shaker	G	459	A	GOF	-14.3									10.1016/s0092-8674(02)01013-9

Shaker	G	466	A	LOF								not expressed		10.1016/s0092-8674(02)01013-9
Shaker	G	466	W	LOF							no current			10.1016/j.neuron.2006.10.005
Shaker	H	486	A	LOF	2.5									10.1016/s0092-8674(02)01013-9
Shaker	H	486	C	GOF	-5.9									10.1085/jgp.20028611.
Shaker	I	241	A	LOF	25									10.1085/jgp.201210827
Shaker	I	241	C	LOF	12									10.1085/jgp.201210827
Shaker	I	241	F	LOF	30									10.1085/jgp.201210827
Shaker	I	241	G	LOF	20									10.1085/jgp.201210827
Shaker	I	241	L	GOF	-5									10.1085/jgp.201210827
Shaker	I	241	M	LOF	9									10.1085/jgp.201210827
Shaker	I	241	N	LOF	40									10.1085/jgp.201210827
Shaker	I	241	Q	LOF	20									10.1085/jgp.201210827
Shaker	I	241	S	LOF	20									10.1085/jgp.201210827
Shaker	I	241	T	GOF	-10									10.1085/jgp.201210827
Shaker	I	241	V	Neutral	0									10.1085/jgp.201210827
Shaker	I	241	W	LOF	45									10.1085/jgp.201210827
Shaker	I	241	Y	LOF	50									10.1085/jgp.201210827
Shaker	I	372	L	LOF	54.5	7.6								10.1085/jgp.111.3.421
Shaker	I	400	A	GOF	-2.6									10.1016/s0092-8674(02)01013-9
Shaker	I	405	A	LOF	3.5									10.1016/s0092-8674(02)01013-9
Shaker	I	405	W	LOF	55.5									10.1016/j.neuron.2006.10.005
Shaker	I	429	A	LOF	17.2									10.1016/s0092-8674(02)01013-9
Shaker	I	457	A	LOF	6.5									10.1016/s0092-8674(02)01013-9

Shaker	I	457	W	LOF	16.7									10.1016/j.neuron.2006.10.005
Shaker	I	464	A	GOF	-2.5									10.1016/s0092-8674(02)01013-9
Shaker	I	464	W	LOF							no current			10.1016/j.neuron.2006.10.005
Shaker	I	470	A	GOF	-1.5									10.1085/jgp.200285 69.
Shaker	I	470	A	LOF	12.6									10.1016/s0092-8674(02)01013-9
Shaker	I	470	D	LOF								not expressed		10.1085/jgp.200285 69.
Shaker	I	470	W	GOF	-16.6									10.1085/jgp.200285 69.
Shaker	I	470	W	GOF	-16.6									10.1016/j.neuron.2006.10.005
Shaker	I	477	A	GOF	-29.2									10.1016/s0092-8674(02)01013-9
Shaker	I	477	A	GOF	-17.2									10.1085/jgp.200308905
Shaker	I	477	D	LOF								not expressed		10.1085/jgp.200285 69.
Shaker	I	477	W	GOF	-29.9									10.1085/jgp.200308905
Shaker	K	374	Q	LOF								not expressed		10.1016/0896-6273(95)90276-7
Shaker	K	374	Q	LOF							no current			10.1038/349305a0.
Shaker	K	374	R	LOF	2	-1		1.5	-2					10.1038/349305a0.
Shaker	K	380	Q	LOF	5	1		-0.4	-4					10.1038/349305a0.
Shaker	K	380	R	GOF	-6	-3		-0.7	-3					10.1038/349305a0.
Shaker	K	390	A	GOF	-6.3									10.1016/s0092-8674(02)01013-9
Shaker	K	427	A	GOF	-5.9									10.1016/s0092-8674(02)01013-9
Shaker	K	456	A	LOF								not expressed		10.1016/s0092-8674(02)01013-9
Shaker	K	456	W	LOF							no current			10.1016/j.neuron.2006.10.005
Shaker	L	358	F	Mixed	10	0		0	10					10.1074/jbc.M111.237792
Shaker	L	358	K	Mixed	-30	2.47		1.87	-30					10.1074/jbc.M111.237792

Shaker	L	358	Q	Mixed	-30	2.09		2.99	-30					10.1074/jbc.M111.237792
Shaker	L	358	R	Mixed	-30	2.54		1.72	-30					10.1074/jbc.M111.237792
Shaker	L	361	E	Mixed	-20	5.3		3.6	-40				fast activation	10.1113/jphysiol.2007.131490
Shaker	L	361	F	Neutral	0	-2.2		-0.5	0					10.1113/jphysiol.2007.131490
Shaker	L	361	K	Mixed	-60	4		4.4	-76				fast activation	10.1113/jphysiol.2007.131490
Shaker	L	361	Q	Mixed	-30	1.6		3.6	-40				fast activation	10.1113/jphysiol.2007.131490
Shaker	L	361	R	Mixed	-60	3		1.7	-71				fast activation	10.1113/jphysiol.2007.131490
Shaker	L	366	A	GOF	-40.3									10.1016/s0092-8674(02)01013-9
Shaker	L	396	A	Unclear									two phases in the I-V curve	10.1016/s0092-8674(02)01013-9
Shaker	L	398	W	LOF								not expressed		10.1016/j.neuron.2006.10.005
Shaker	L	398	W	LOF	51.6									10.1016/s0092-8674(02)01013-9
Shaker	L	399	A	Unclear									two phases in the I-V curve	10.1016/s0092-8674(02)01013-9
Shaker	L	403	A	LOF	6.5									10.1016/s0092-8674(02)01013-9
Shaker	L	409	A	LOF	13									10.1016/s0092-8674(02)01013-9
Shaker	L	409	W	LOF	32.2									10.1016/j.neuron.2006.10.005
Shaker	L	461	A	GOF	-8.5									10.1016/s0092-8674(02)01013-9
Shaker	L	461	W	GOF	-1.7									10.1016/j.neuron.2006.10.005
Shaker	L	468	A	LOF	18.2									10.1016/s0092-8674(02)01013-9
Shaker	L	468	W	GOF	-17.1									10.1016/j.neuron.2006.10.005
Shaker	L	472	A	LOF	12.6									10.1016/s0092-8674(02)01013-9
Shaker	L	472	A	LOF	25.6									10.1085/jgp.20028569.
Shaker	L	472	D	LOF								not expressed		10.1085/jgp.20028569.
Shaker	L	472	W	LOF	80.9									10.1016/j.neuron.2006.10.005

Shaker	L	472	W	LOF	80.9									10.1085/jgp.2002 8569.
Shaker	M	356	K	Mixed	25	0		0	25					10.1074/jbc.M111 .237792
Shaker	M	356	R	Mixed	20	0		0	20					10.1074/jbc.M111 .237792
Shaker	M	393	A	LOF	20.1									10.1016/s0092- 8674(02)01013-9
Shaker	M	393	W	GOF	-13.4									10.1016/j.neuron. 2006.10.005
Shaker	M	448	A	Neutral	-0.2									10.1016/s0092- 8674(02)01013-9
Shaker	N	353	R	LOF	10	0		0	0	0				10.1074/jbc.M111 .237792
Shaker	N	423	A	GOF	-0.7									10.1016/s0092- 8674(02)01013-9
Shaker	N	480	A	LOF								not expressed		10.1016/s0092- 8674(02)01013-9
Shaker	N	480	A	LOF	80.5									10.1085/jgp.2002 8569.
Shaker	N	480	D	GOF	-18									10.1085/jgp.2002 8569.
Shaker	N	480	W	LOF	35.8									10.1085/jgp.2002 8569.
Shaker	N	482	A	GOF	-9.5									10.1016/s0092- 8674(02)01013-9
Shaker	N	482	A	GOF	-6.1									10.1085/jgp.2002 8569.
Shaker	N	482	C	LOF	5.4									10.1085/jgp.2002 8611.
Shaker	N	482	D	GOF	-2									10.1085/jgp.2002 8569.
Shaker	N	482	W	GOF	-17.3									10.1085/jgp.2002 8569.
Shaker	P	430	A	LOF								not expressed		10.1016/s0092- 8674(02)01013-9
Shaker	P	430	G	LOF							no current			10.1038/349700a0
Shaker	P	450	A	LOF								not expressed		10.1016/s0092- 8674(02)01013-9
Shaker	P	450	G	LOF							no current			10.1038/349700a0
Shaker	P	473	A	LOF							no current			10.1016/s0092- 8674(02)01013-9
Shaker	P	473	D	LOF								not expressed		10.1085/jgp.2002 8569.
Shaker	P	473	W	LOF							no current			10.1085/jgp.2002 8569.

Shaker	P	475	A	LOF							no current			10.1016/j.bpj.2013.11.025
Shaker	P	475	A	LOF	101									10.1085/jgp.200285569.
Shaker	P	475	C	LOF	176.5									10.1085/jgp.200308905.
Shaker	P	475	D	GOF	-68									10.1085/jgp.200308905.
Shaker	P	475	E	GOF	-65.6									10.1085/jgp.200308905.
Shaker	P	475	F	LOF	8.9									10.1085/jgp.200308905.
Shaker	P	475	G	LOF	75.1									10.1085/jgp.200285569.
Shaker	P	475	G	LOF	75.1									10.1016/j.bpj.2013.11.025
Shaker	P	475	H	GOF	-45.5									10.1085/jgp.200308905.
Shaker	P	475	I	LOF	94.4									10.1085/jgp.200308905.
Shaker	P	475	K	GOF	-63.7									10.1085/jgp.200308905.
Shaker	P	475	M	LOF	18.6									10.1085/jgp.200308905.
Shaker	P	475	N	GOF	-53.1									10.1085/jgp.200308905.
Shaker	P	475	Q	GOF	-64.7									10.1085/jgp.200308905.
Shaker	P	475	D	GOF	-42.3									10.1085/jgp.200308905.
Shaker	P	475	S	LOF	225.5									10.1085/jgp.200285569.
Shaker	P	475	T	LOF	15.5									10.1085/jgp.200308905.
Shaker	P	475	V	LOF	95.3									10.1085/jgp.200308905.
Shaker	P	475	W	GOF	-18.9									10.1085/jgp.200308905.
Shaker	P	475	Y	GOF	-17.8									10.1085/jgp.200308905.
Shaker	R	297	K	Mixed	8	-1		3.5	14					10.1038/349305a0.
Shaker	R	362	E	LOF	16	4.2								10.1085/jgp.111.3.421
Shaker	R	362	K	LOF	11	-2		-0.8	12					10.1038/349305a0.

Shaker	R	362	Q	Mixed	19	1		4.2	22					10.1038/349305a0.
Shaker	R	365	K	Mixed	7	2		0.2	4					10.1038/349305a0.
Shaker	R	365	Q	Mixed	-11	1		2.9	-14					10.1038/349305a0.
Shaker	R	368	K	LOF	7	2		-0.3	-8					10.1038/349305a0.
Shaker	R	368	Q	LOF	51	6		12.1	55					10.1016/s0006-3495(94)80783-0
Shaker	R	368	Q	LOF	108									10.1038/349305a0.
Shaker	R	371	K	Mixed	62	-4		3.9	64					10.1038/349305a0.
Shaker	R	371	Q	Neutral	-1									10.1016/s0006-3495(94)80783-0
Shaker	R	371	Q	Mixed	-17	3		0.4	-14					10.1038/349305a0.
Shaker	R	377	K	LOF	31	4		10.1	27					10.1016/s0006-3495(94)80783-0
Shaker	R	377	K	LOF	36									10.1038/349305a0.
Shaker	R	377	Q	LOF							no current			10.1038/349305a0.
Shaker	R	377	Q	LOF										10.1016/0896-6273(95)90276-7
Shaker	R	394	A	LOF	7.8									10.1016/s0092-8674(02)01013-9
Shaker	R	394	W	LOF	23.3									10.1016/j.neuron.2006.10.005
Shaker	R	487	C	GOF	-4									10.1085/jgp.200208611.
Shaker	S	376	T	LOF	20	6.1								10.1085/jgp.111.3.421
Shaker	S	392	A	LOF								not expressed		10.1016/s0092-8674(02)01013-9
Shaker	S	411	A	LOF	1									10.1016/s0092-8674(02)01013-9
Shaker	S	411	W	LOF							no current			10.1016/j.neuron.2006.10.005
Shaker	S	412	A	LOF	1.7									10.1016/s0092-8674(02)01013-9
Shaker	S	412	W	LOF								reduced		10.1016/j.neuron.2006.10.005
Shaker	S	421	A	GOF	-1.7									10.1016/s0092-8674(02)01013-9

Shaker	S	424	A	LOF	3.6									10.1016/s0092-8674(02)01013-9
Shaker	S	428	A	LOF	15									10.1016/s0092-8674(02)01013-9
Shaker	S	460	A	GOF	-10.4									10.1016/s0092-8674(02)01013-9
Shaker	S	460	W	LOF								reduced		10.1016/j.neuron.2006.10.005
Shaker	S	479	A	LOF	1.7									10.1016/s0092-8674(02)01013-9
Shaker	S	479	A	Neutral	0.2									10.1085/jgp.200285 69.
Shaker	S	479	D	GOF	-27.5									10.1085/jgp.200285 69.
Shaker	S	479	W	LOF	85.5									10.1085/jgp.200285 69.
Shaker	T	439	A	LOF								not expressed		10.1016/s0092-8674(02)01013-9
Shaker	T	439	A	LOF							no current			10.1038/349700a0
Shaker	T	439	S	LOF							no current			10.1038/349700a0
Shaker	T	441	A	LOF	9.2									10.1016/s0092-8674(02)01013-9
Shaker	T	441	S	Neutral									inactivation state stabilized, 50% decrease of recovery	10.1016/S0006-3495(95)80206-7
Shaker	T	441	S	Unclear									care: may change ion selectivity!	10.1038/349700a0
Shaker	T	442	A	LOF	-25.8						no current			10.1016/s0092-8674(02)01013-9
Shaker	T	442	A	GOF	-26									10.1038/349700a0
Shaker	T	442	S	Mixed	-21								inactivation state destabilized, c-Type inactivation	10.1016/S0006-3495(95)80206-7
Shaker	T	442	S	Mixed	-20.5				-22				care: may change ion selectivity!	10.1038/349700a0
Shaker	T	449	A	GOF	-5									10.1016/s0092-8674(02)01013-9
Shaker	T	469	A	GOF	-19.5									10.1016/s0092-8674(02)01013-9
Shaker	T	469	A	GOF	-5.8									10.1085/jgp.200285 69.
Shaker	T	469	D	LOF								not expressed		10.1085/jgp.200285 69.

Shaker	T	469	W	GOF	-6.2									10.1085/jgp.2002 8569.
Shaker	T	469	W	GOF	-6.2									10.1016/j.neuron. 2006.10.005
Shaker	T	489	C	Neutral	-0.7						no current			10.1085/jgp.2002 8611.
Shaker	V	369	I	LOF	17.8	2.9								10.1085/jgp.111.3 .421
Shaker	V	407	A	GOF	-4.4									10.1016/s0092- 8674(02)01013-9
Shaker	V	408	A	LOF	9.7									10.1016/s0092- 8674(02)01013-9
Shaker	V	408	W	LOF	32.6									10.1016/j.neuron. 2006.10.005
Shaker	V	414	A	LOF	4									10.1016/s0092- 8674(02)01013-9
Shaker	V	414	W	LOF							no current			10.1016/j.neuron. 2006.10.005
Shaker	V	437	A	GOF	-17.8									10.1016/s0092- 8674(02)01013-9
Shaker	V	443	A	LOF								not expressed		10.1016/s0092- 8674(02)01013-9
Shaker	V	451	A	GOF	-1									10.1016/s0092- 8674(02)01013-9
Shaker	V	453	A	LOF	2									10.1016/s0092- 8674(02)01013-9
Shaker	V	453	W	GOF	-4									10.1016/j.neuron. 2006.10.005
Shaker	V	458	A	GOF	-21									10.1016/s0092- 8674(02)01013-9
Shaker	V	458	W	LOF	24									10.1016/j.neuron. 2006.10.005
Shaker	V	467	A	GOF	-7.9									10.1016/s0092- 8674(02)01013-9
Shaker	V	474	A	GOF	-17.1									10.1085/jgp.2002 8569.
Shaker	V	474	D	LOF								not expressed		10.1085/jgp.2002 8569.
Shaker	V	474	W	LOF								not expressed		10.1085/jgp.2002 8569.
Shaker	V	474	W	LOF							no current			10.1016/j.neuron. 2006.10.005
Shaker	V	476	A	GOF	-35.5									10.1016/s0092- 8674(02)01013-9
Shaker	V	476	A	GOF	-22.9									10.1085/jgp.2002 8569.

Shaker	V	476	W	GOF	-22.9									10.1085/jgp.20028569.
Shaker	V	478	A	GOF	-11.4									10.1085/jgp.20028569.
Shaker	V	478	D	LOF								not expressed		10.1085/jgp.20028569.
Shaker	V	478	W	LOF							no current			10.1085/jgp.20028569.
Shaker	W	434	A	LOF								not expressed		10.1016/s0092-8674(02)01013-9
Shaker	W	434	F	LOF							no current			10.1085/jgp.109.6.779
Shaker	W	434	F	LOF							no current			10.1016/0896-6273(93)90190-3
Shaker	W	434	S	LOF							no current			10.1038/349700a0
Shaker	W	435	A	LOF								not expressed		10.1016/s0092-8674(02)01013-9
Shaker	W	435	S	LOF							no current			10.1038/349700a0
Shaker	W	454	A	GOF	-2									10.1016/s0092-8674(02)01013-9
Shaker	W	454	A	LOF	6									10.1016/j.neuron.2006.10.005
Shaker	Y	483	A	Unclear									two phases in the I-V curve	10.1016/s0092-8674(02)01013-9
Shaker	Y	483	A	GOF	-4.9									10.1085/jgp.20028569.
Shaker	Y	483	C	GOF	-4.4									10.1085/jgp.20028611.
Shaker	Y	483	D	GOF	-11.6									10.1085/jgp.20028569.
Shaker	Y	483	W	Neutral	-0.6									10.1085/jgp.20028569.
Shaker	Y	485	A	LOF	3.5									10.1016/s0092-8674(02)01013-9
Shaker	Y	485	A	LOF	4.4									10.1085/jgp.20028569.
Shaker	Y	485	A	LOF	4.4									10.1085/jgp.20028569.
Shaker	Y	485	C	LOF	1.2									10.1085/jgp.20028611.
Shaker	Y	485	W	GOF	-13.4									10.1085/jgp.20028569.
KCNA4	R	89	Q	LOF						-3				10.1136/jmedgenet-2015-103637
KCNA4	T	330	S	Neutral						139			surface expression 65%	10.1074/jbc.M708921200

KCNA4	T	330	K	LOF						-1053		reduced	surface expression 21%, variant fails to inactivate	10.1074/jbc.M708921200
KCNA4	T	330	A	LOF						-1234		reduced	surface expression 9%	10.1074/jbc.M708921200
KCNA4	E	502	V	LOF						-1251		reduced	surface expression 7%	10.1074/jbc.M708921200
KCNA4	E	502	D	LOF								reduced	surface expression 30%	10.1074/jbc.M708921200
KCNA4	E	502	T	LOF						-1252		reduced	surface expression 28%	10.1074/jbc.M708921200
KCNA4	E	502	K	LOF						-1225		reduced	surface expression 9%	10.1074/jbc.M708921200
KCNA5	A	115	V	Neutral	-0.6		-1.6			13				10.1016/j.clpt.2004.10.008
KCNA5	A	251	T	Neutral	0.7		1.4			24				10.1016/j.clpt.2004.10.008
KCNA5	P	307	S	Neutral	-1.2		-0.9			18				10.1016/j.clpt.2004.10.008
KCNA5	P	310	L	Neutral	-3.1		-1.4			-8				10.1016/j.clpt.2004.10.008
KCNA5	P	532	L	Neutral	-0.5		-9.9			11			drug resistant	10.1016/j.clpt.2004.10.008
KCNA5	R	578	K	LOF	9.1		-2.3			17			drug resistant	10.1016/j.clpt.2004.10.008
KCNA5	P	532	L	Neutral	-0.4		-8.06			-6				10.1172/JCI23741
KCNA5	P	532	G	Neutral	5		-4.24			1				10.1172/JCI23741
KCNA5	P	532	A	Neutral	3.4		-10.01			-5				10.1172/JCI23741
KCNA5	P	532	E	Neutral	2.66		-0.73			-8				10.1172/JCI23741
KCNA5	P	532	M	LOF	20.49		0.1			6				10.1172/JCI23741
KCNA5	P	532	S	Neutral	9.69		-0.63			-15				10.1172/JCI23741
KCNA5	E	48	G	GOF	-3.77	-0.16		0.3	2.33	175.2				10.1093/eurheartj/ehs442
KCNA5	A	305	T	GOF	-5.36	-0.54		-0.64	4.46	233.5				10.1093/eurheartj/ehs442
KCNA5	D	322	H	GOF	-4.7	-2		-0.83	7.85	438.9				10.1093/eurheartj/ehs442
KCNA5	Y	155	C	LOF	-0.08	-2.91		-0.39	3.39	-246				10.1093/eurheartj/ehs442
KCNA5	P	488	S	LOF	-0.18	-0.31		0.01	2.99	-218				10.1093/eurheartj/ehs442
KCNA5	D	469	E	LOF	4.82	-0.21				-474.1				10.1093/eurheartj/ehs442

KCNA5	T	527	M	LOF						-2			reduced current, exact value not available	10.3760/CMA.J.ISSN.0376-2491.2010.16.007
KCNA5	T	527	M	LOF						-28.45				10.1038/jhg.2009.26
KCNA5	A	576	V	LOF						-52.28				10.1038/jhg.2009.26
KCNA5	E	610	K	LOF						-57.73				10.1038/jhg.2009.26
KCNA5	H	463	R	LOF	1.5	-1.4				-437				10.1161/CIRCEP.114.002519
KCNA5	T	527	M	GOF	-4.6	-0.2				353				10.1161/CIRCEP.114.002519
KCNB1	R	306	C	LOF	0.8	10.4				0			slow activation, severe reduction of repetitive firing	10.1038/srep15199
KCNB1	G	401	R	LOF	7.2	7				-0.9			fast inactivation (92.7 ms vs 145.7 ms), reduced repetitive firing	10.1038/srep15199
KCNB1	I	199	F	LOF	9	-4.5		1.4	3.1	-50.8				10.1212/NXG.0000000000000198
KCNB1	V	378	A	GOF								reduced	loss of ion selectivity, faster inactivation, large tail current	10.1085/jgp.201511444
KCNB1	S	347	R	LOF	6.9	1.5				-160			loss of ion selectivity, slower activation, conductance estimated from figure	10.1002/ana.24263
KCNB1	G	379	R	LOF	5.4	0.8				-280			loss of ion selectivity, slower activation, conductance estimated from figure	10.1002/ana.24263
KCNB1	T	374	I	LOF	6	1.3				-280			loss of ion selectivity, slower activation, conductance estimated from figure	10.1002/ana.24263
KCNB1	S	202	F	LOF	8.6	9.3		4.1	10	0			high throughput assay, current density estimated from figure	10.1002/ana.25607

KCNB1	T	210	K	LOF						-0.99				10.1002/ana.25607
KCNB1	T	210	M	LOF	0.4	0		0.2	-3.7	0		reduced		10.1002/ana.25607
KCNB1	R	306	C	LOF						-0.98				10.1002/ana.25607
KCNB1	R	312	C	LOF						-0.95				10.1002/ana.25607
KCNB1	R	312	H	LOF						-0.4				10.1002/ana.25607
KCNB1	R	325	Q	Neutral	-3.4	-0.1		-0.4	-3.8	0		reduced		10.1002/ana.25607
KCNB1	R	325	W	GOF	-7	3.2		-0.4	-12.3	0				10.1002/ana.25607
KCNB1	E	330	D	LOF	-5.1	2.4		1.1	-0.8	0		reduced		10.1002/ana.25607
KCNB1	W	370	R	LOF	3.8	3.5		0.6	0.7	-0.9		reduced		10.1002/ana.25607
KCNB1	V	378	A	LOF	-0.2	0.4		-0.7	-1.8	0		reduced		10.1002/ana.25607
KCNB1	V	378	L	LOF	1.5	3.1		1.2	-6.8	-0.5		reduced		10.1002/ana.25607
KCNB1	G	381	R	LOF	-2.1	1.3		0	-1.6	0		reduced		10.1002/ana.25607
KCNB1	P	385	T	LOF	0.6	2		0.6	-1	-0.9		reduced		10.1002/ana.25607
KCNB1	K	391	N	LOF	-2.1	-0.3		-0.4	-1.8	0		reduced		10.1002/ana.25607
KCNB1	F	416	L	LOF	4.2	2.7		1	0.1	-0.9		reduced		10.1002/ana.25607
KCNB1	S	457	R	GOF	-8.4									10.1002/ana.25607
KCNC1	R	320	H	LOF						-1				10.1038/ng.3144
KCNC1	A	421	V	LOF						-1	no current		homozygous variant has no current above background	10.1002/acn3.50822
KCNC1	R	317	H	LOF						-1	no current			10.1002/acn3.50822
KCNC1	Q	339	X	LOF						-1	no current			10.1002/acn3.50822
KCNC1	Q	492	X	LOF	-8	-0.1				-0.5				10.1002/acn3.50822
KCNC1B	A	513	V	Neutral	-4.8	-1.8				0				10.1002/acn3.50822
KCNC1	C	208	Y	LOF						-1				10.1002/acn3.50799

KCNC1	T	399	M	LOF						-1			dominant negative	10.1002/acn3.50799
KCNC1	A	421	V	LOF						-1			dominant negative	10.1002/acn3.50799
KCNC1B	R	320	H	LOF	5.3					0			slower activation, other complex effects	10.1111/epi.16867
KCNC1	W	392	F	LOF						-1	no current			10.1111/epi.16867
KCNC2	D	167	Y	GOF						-0.5			hyperpolarized shift of the voltage dependence of activation and altered slope factor, but no exact values provided	10.1055/s-0040-1710524
KCNC3	R	420	H	LOF						-0.9			currents estimated from figure	10.1038/ng1758
KCNC3	F	448	L	GOF	-12.4	0	11.3						label uncertain, mutation increases open state probability, but also increases interspike interval	10.1038/ng1758
KCNC3	R	423	H	LOF						-1			currents estimated from figure	10.1002/humu.21165
KCNC3	R	366	H	LOF						-0.9			currents estimated from figure	10.1002/humu.21165
KCNC3	R	420	H	LOF						-1				10.1113/jphysiol.2012.228205
KCNC3	R	423	H	GOF	-16.5	1.6				-0.92				10.1113/jphysiol.2012.228205
KCNC3	F	448	L	GOF	-16.5	0.6	78.7							10.1113/jphysiol.2012.228205
KCNC3	W	495	F	LOF							no current		abolished ionic conductance through the central pore	10.1113/jphysiol.2012.228205
KCNC3	R	420	H	LOF								reduced	additional data on post-translational modifications	10.1016/j.nbd.2014.08.020
KCNC3	D	129	N	GOF	-9.25	-2.1				877			labels uncertain and not provided by author	10.1371/journal.pone.0116599
KCNC3	D	477	N	Neutral	-0.67	-2				872				10.1371/journal.pone.0116599

KCNC3	V	535	M	GOF	-31.06	2.7				706				10.1371/journal.pone.0116599
KCNC3	S	591	G	LOF	16.34	-2.7				-2050				10.1371/journal.pone.0116599
KCNC3	G	643	S	Neutral	-6.36	0.1				1025				10.1371/journal.pone.0116599
KCNC3	P	645	R	Neutral	-5.72	0.4				-725				10.1371/journal.pone.0116599
KCNC3	D	746	N	Neutral	1.84	-1.8				840				10.1371/journal.pone.0116599
KCNC3	T	428	I	LOF						-11.9			currents estimated from figure, slower activation	10.1093/brain/awv117
KCND2	E	323	K	LOF									pre-print, data not released yet	10.1093/hmg/ddab192
KCND2	P	403	A	Unclear									pre-print, data not released yet	10.1093/hmg/ddab192
KCND2	V	404	L	Unclear									pre-print, data not released yet	10.1093/hmg/ddab192
KCND2	V	404	M	Unclear									pre-print, data not released yet	10.1093/hmg/ddab192
KCND2	V	404	M	GOF									significantly slowed inactivation	10.1093/hmg/ddu056
KCND2	G	309	A	GOF	-9.2	1.9		-0.7	-7.8	7.7				10.1093/hmg/ddab192
KCND2	L	310	A	LOF	123.5	41.6		6.2	47.2	-2.4				10.1093/hmg/ddab192
KCND2	R	311	A	LOF	18.6	2.5		-0.1	18.7	0.1				10.1093/hmg/ddab192
KCND2	I	312	A	GOF	6.3	0.3		1.8	-5.4	10.1				10.1093/hmg/ddab192
KCND2	L	313	A	LOF	4.5	4.6		1.1	-10.6	-0.5				10.1093/hmg/ddab192
KCND2	G	314	A	Neutral										10.1093/hmg/ddab192
KCND2	Y	315	A	GOF	1.2	2.5		-0.4	4.1	6.3				10.1093/hmg/ddab192
KCND2	T	316	A	LOF	4.2	2.5		0.9	-5.8	1.6				10.1093/hmg/ddab192
KCND2	L	317	A	LOF	14.1	3.5		1.7	-5.7	1.3				10.1093/hmg/ddab192
KCND2	K	318	A	LOF	7.7	3.7		-0.5	-0.8	1.6				10.1093/hmg/ddab192
KCND2	S	319	A	LOF						-3.57				10.1093/hmg/ddab192

KCND2	C	320	A	LOF	13.6	4.1		-0.9	1	1.5				10.1093/hmg/ddab192
KCND2	C	320	S	GOF	-0.3	10.8		-1	-0.5	9.1				10.1093/hmg/ddab192
KCND2	A	321	V	GOF	-16.7	0.7		-0.9	-1.5	1.4				10.1093/hmg/ddab192
KCND2	S	322	A	LOF	13.2	-0.2		-0.1	14.2	4				10.1093/hmg/ddab192
KCND2	E	323	A	GOF	-10.1	-1.2		0.9	-12.2	0.5				10.1093/hmg/ddab192
KCND2	L	324	A	LOF						-3.64				10.1093/hmg/ddab192
KCND2	G	325	A	GOF	0.9	0		0.4	2	7.4				10.1093/hmg/ddab192
KCND2	F	326	A	LOF	2.9	0.6		1	21	-1.5				10.1093/hmg/ddab192
KCND2	L	327	A	Neutral										10.1093/hmg/ddab192
KCND2	L	328	A	LOF						-3.65				10.1093/hmg/ddab192
KCND2	F	329	A	Neutral										10.1093/hmg/ddab192
KCND2	V	397	A	LOF	122.5	85.6				-2.9				10.1093/hmg/ddab192
KCND2	I	398	A	Neutral										10.1093/hmg/ddab192
KCND2	A	399	V	LOF						-3.6				10.1093/hmg/ddab192
KCND2	L	400	A	LOF	-4.6	9.8		5	-25.2	-2.5				10.1093/hmg/ddab192
KCND2	P	401	A	LOF						-3.4				10.1093/hmg/ddab192
KCND2	V	402	A	Unclear	-6.6	3		3.9	7.7	-1.4				10.1093/hmg/ddab192
KCND2	P	403	A	Neutral										10.1093/hmg/ddab192
KCND2	V	404	A	Unclear	6.1	4.6		4.3	-28.3	1				10.1093/hmg/ddab192
KCND2	I	405	A	Neutral										10.1093/hmg/ddab192
KCND2	V	406	A	Neutral										10.1093/hmg/ddab192
KCND2	S	407	A	GOF	0.1	0.7		0	5.9	5.8				10.1093/hmg/ddab192
KCND2	N	408	A	LOF	21.4	2.5		-0.2	10.7	-1.1				10.1093/hmg/ddab192

KCND2	F	409	A	LOF						-3.3				10.1093/hmg/ddab192
KCND2	S	410	A	LOF	5.2	1.7		0.4	1.6	0.3				10.1093/hmg/ddab192
KCND2	R	411	A	Unclear	1	1.3		0.6	3.7	1.5				10.1093/hmg/ddab192
KCND2	I	412	A	Unclear	1	3.1		0	-13.3	0.6				10.1093/hmg/ddab192
KCND2	Y	413	A	Unclear	2	1.6		1.1	4.3	1.2				10.1093/hmg/ddab192
KCND3	Y	241	C	LOF									reduced amplitude, dominant negative	own data provided by Uli Hedrich
KCND3	V	338	E	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	G	345	V	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	S	347	W	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	T	352	P	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	W	359	G	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	G	371	R	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	M	373	I	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	V	374	A	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	G	384	S	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	S	390	N	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	R	431	C	LOF									reduced amplitude	own data provided by Uli Hedrich
KCND3	C	317	Y	LOF									reduced amplitude, right shift of activation, left shift of inactivation	own data provided by Uli Hedrich
KCND3	P	375	S	LOF									reduced amplitude, right shift of activation, no change in inactivation	own data provided by Uli Hedrich
KCND3	T	377	M	LOF									reduced amplitude, right	own data provided by Uli Hedrich

													shift of activation, no change in inactivation	
KCND3	S	283	F	Unclear									reduced amplitude, right shift of activation and inactivation	own data provided by Uli Hedrich
KCND3	R	290	Q	Unclear									reduced amplitude, left shift of activation and inactivation, slowed macroscopic decay and decovery	own data provided by Uli Hedrich
KCND3	G	391	S	Unclear									reduced amplitude, slowed macroscopic delay	own data provided by Uli Hedrich
KCND3	G	306	A	Unclear									increased amplitude, slowed decay and recovery	own data provided by Uli Hedrich
KCND3	T	361	S	Unclear									increased amplitude, slowed recovery	own data provided by Uli Hedrich
KCND3	R	431	H	GOF									increased amplitude, fast recovery	own data provided by Uli Hedrich
KCND3	L	450	F	GOF									increased amplitude	own data provided by Uli Hedrich
KCND3	A	564	P	GOF									increased amplitude, slowed decay	own data provided by Uli Hedrich
KCND3	G	600	R	GOF									increased amplitude, slowed decay	own data provided by Uli Hedrich
KCND3	K	241	R	Neutral										own data provided by Uli Hedrich
KCND3	S	530	F	Neutral										own data provided by Uli Hedrich
KCND3	S	530	P	Neutral										own data provided by Uli Hedrich
KCND3	P	633	S	Neutral										own data provided by Uli Hedrich
KCNH1	K	217	N	GOF	-40.2	4.3	3.14			116				10.1038/ng.3153
KCNH1	L	489	F	GOF	-36.4	2.7	3.41			-74				10.1038/ng.3153

KCNH1	I	494	V	GOF	-33.9	4.1	2.55			90				10.1038/ng.3153
KCNH1	Q	503	R	GOF	-49.1	2.3	3.53			-555				10.1038/ng.3153
KCNH1	G	348	R	GOF	-43.5	1.7								10.1038/ng.3282
KCNH1	I	467	V	GOF	-60.5	3.6								10.1038/ng.3282
KCNH1	L	352	V	GOF	-94.3	1.9								10.1038/ng.3282
KCNH1	S	325	Y	GOF	-47	1.8								10.1038/ng.3282
KCNH1	V	356	L	GOF	-18.6	-0.6								10.1038/ng.3282
KCNH1	G	469	R	GOF									complex functional effect	10.1038/ng.3282
KCNH2	Y	611	H	LOF								not expressed	trafficking defect	10.1074/jbc.M502327200
KCNH2	Y	611	H	LOF								not expressed	ER retained	10.1074/jbc.273.33.21061
KCNH2	V	822	M	LOF								not expressed	ER retained	10.1074/jbc.273.33.21061
KCNH2	I	593	R	LOF							no current		unable to form functional channels	10.1074/jbc.273.33.21061
KCNH2	G	628	S	LOF							no current		unable to form functional channels	10.1074/jbc.273.33.21061
KCNH2	G	628	S	LOF							no current			10.1073/pnas.93.5.2208
KCNH2	A	561	V	LOF							no current			10.1073/pnas.93.5.2208
KCNH2	N	470	D	LOF	-18	-1.1				-599				10.1073/pnas.93.5.2208
KCNH2	T	474	I	LOF	3.1	0.1	0	1.2	-2.7	-321			dominant negative	10.1161/01.res.83.4.415
KCNH2	A	614	V	LOF	3.1	-1.7	0	0.3	-9.3	-645			dominant negative	10.1161/01.res.83.4.415
KCNH2	V	630	L	LOF	0.6	-2.6	0	3	-22.2	-978			dominant negative, faster inactivation	10.1161/01.res.83.4.415
KCNH2	A	558	P	LOF			0			-92		reduced	dominant trafficking defect, fast inactivation	10.1172/JCI35337
KCNH2	I	31	S	LOF								reduced	large temperature-dependent trafficking study	10.1161/CIRCULATIONAHA.105.570200
KCNH2	R	328	C	Neutral	0	0	0			14.1				10.1161/CIRCULATIONAHA.105.570200

KCNH2	P	347	S	Neutral	0	0	0			-18.6				10.1161/CIRCULATIONAHA.105.570200
KCNH2	T	421	M	LOF									altered gating and permeation	10.1161/CIRCULATIONAHA.105.570200
KCNH2	A	422	T	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	T	436	M	Neutral	0	0	0			-29.1				10.1161/CIRCULATIONAHA.105.570200
KCNH2	D	456	Y	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	N	470	D	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	T	474	I	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	R	534	C	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	A	561	T	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	A	561	V	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	H	562	P	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	I	571	L	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	G	572	S	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	I	593	R	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	P	596	R	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	G	601	S	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200

KCNH2	Y	611	H	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	V	612	L	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	A	614	V	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	T	623	I	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	G	628	S	LOF									altered gating and permeation	10.1161/CIRCULATIONAHA.105.570200
KCNH2	N	629	D	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	N	629	S	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	V	630	A	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	V	630	L	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	F	640	V	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	R	752	W	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	F	805	C	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	S	818	L	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	V	822	M	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	R	823	W	LOF								reduced		10.1161/CIRCULATIONAHA.105.570200
KCNH2	R	922	W	Neutral	0	0	0			5				10.1161/CIRCULATIONAHA.105.570200

KCNH2	N	588	K	GOF									complex mechanism	10.1161/01.CIR.0000109482.92774.3A
KCNH2	K	897	T	GOF	-7.1	-0.1		-1	-0.6	0			faster activation, faster deactivation, no change in recovery, interesting "functional polymorphism"	10.1016/S0008-6363(03)00342-0
KCNH2	R	752	Q	LOF							no current		homozygote patient, healthy heterozygote parents, no functional data for heterozygous conditions available	10.1203/01.PDR.0000059750.17002.B6
KCNH2	L	552	S	LOF	-8.4		-503			0			faster activation , another rare homozygote	10.1016/S0735-1097(00)00636-7
KCNH2	T	65	P	LOF	0	0						reduced	faster activation, faster deactivation, trafficking defect	10.1074/jbc.M206569200
KCNH2	R	784	W	LOF	13.3					-28.1				10.1161/01.CIR.0000014448.19052.4C
KCNH2	A	490	T	LOF	0	0				-34				10.1002/1096-8628(20010201)98:4<348::AID-AJMG1109>3.0.CO;2-A
KCNH2	F	29	L	LOF									faster deactivation	10.1074/jbc.274.15.10113
KCNH2	N	33	T	LOF	7.5								faster deactivation, slower activation	10.1074/jbc.274.15.10113
KCNH2	G	53	R	LOF									faster deactivation	10.1074/jbc.274.15.10113
KCNH2	R	56	Q	LOF	11.3	0.2							faster deactivation, slower activation	10.1074/jbc.274.15.10113
KCNH2	C	66	G	LOF									faster deactivation	10.1074/jbc.274.15.10113
KCNH2	H	70	R	LOF	0	0							faster deactivation	10.1074/jbc.274.15.10113

KCNH2	A	78	P	LOF									faster deactivation	10.1074/jbc.274.15.10113
KCNH2	L	86	R	LOF	0	0							faster deactivation	10.1074/jbc.274.15.10113
KCNH2	V	612	L	LOF	1	-1						reduced	decrease in surface expression, variable dominant-negative effect	10.1161/hc3501.093815
KCNH2	T	613	M	LOF	2	0.5						reduced	decrease in surface expression, variable dominant-negative effect	10.1161/hc3501.093815
KCNH2	L	615	V	LOF	-2	-0.8						reduced	decrease in surface expression, variable dominant-negative effect	10.1161/hc3501.093815
KCNH2	K	28	E	LOF	-2.7	-0.8	-102	-1.1	7.8					10.1074/jbc.M110.205948
KCNH2	N	33	T	LOF	3.5	1.4	-75	-3.9	11.5					10.1074/jbc.M110.205948
KCNH2	R	56	Q	LOF	-3.3	1.7	-144	-7.8	26.6					10.1074/jbc.M110.205948
KCNH2	M	124	R	LOF	0	-0.1	-105	-3.3	13.6					10.1074/jbc.M110.205948
KCNH2	F	29	L	LOF	0.3	0.6	-109							10.1074/jbc.M110.205948
KCNH2	G	53	R	Neutral	3.2	1.1								10.1074/jbc.M110.205948
KCNH2	H	70	R	Neutral	1.5	1								10.1074/jbc.M110.205948
KCNH2	A	78	P	LOF	3.1	1.6								10.1074/jbc.M110.205948
KCNH2	Y	43	C	LOF								not expressed		10.1074/jbc.M110.205948
KCNH2	C	66	G	LOF							no current			10.1074/jbc.M110.205948
KCNH2	L	86	R	LOF								not expressed		10.1074/jbc.M110.205948
KCNH2	T	618	I	GOF						5.14				10.1016/j.yjmcc.2010.11.017
KCNH2	N	985	S	GOF						0.45				10.1016/j.cardiores.2005.06.027

KCNH2	G	873	S	GOF						0.64				10.1016/j.cardiores.2005.06.027
KCNH5	R	327	H	GOF	-64.9	-0.2				0.7			faster activation	10.1523/JNEUROSCI.2307-13.2013
KCNQ1	V	100	I	Neutral	-2.03	0.62	-24.55			7.43			start - high throughput data by Carlos	10.1161/CIRCGEN.118.002345
KCNQ1	A	102	S	GOF	-3.56	0.76	11.98			103.28				10.1161/CIRCGEN.118.002345
KCNQ1	T	104	I	LOF						-40				10.1161/CIRCGEN.118.002345
KCNQ1	T	104	S	Neutral	-2.87	0.91	-17.34			-10.39				10.1161/CIRCGEN.118.002345
KCNQ1	H	105	N	Neutral	1.53	-0.03	-32.32			-15.81				10.1161/CIRCGEN.118.002345
KCNQ1	H	105	L	GOF	-3					22				10.1161/CIRCGEN.118.002345
KCNQ1	H	105	Y	Neutral	3.68	-0.98	-35.75			28.95				10.1161/CIRCGEN.118.002345
KCNQ1	V	106	I	Neutral	-1.43	-0.24	16.23			11.82				10.1161/CIRCGEN.118.002345
KCNQ1	Q	107	H	LOF						-32				10.1161/CIRCGEN.118.002345
KCNQ1	R	109	L	GOF						74				10.1161/CIRCGEN.118.002345
KCNQ1	V	110	I	LOF	1.53	0.25	4.63			-42.27				10.1161/CIRCGEN.118.002345
KCNQ1	Y	111	C	LOF						-57.93				10.1161/CIRCGEN.118.002345
KCNQ1	L	114	P	LOF						-57.25				10.1161/CIRCGEN.118.002345
KCNQ1	E	115	G	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	T	118	S	LOF	5.24	-1.61	8.45			-13.71				10.1161/CIRCGEN.118.002345
KCNQ1	G	119	R	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	C	122	Y	LOF	15.09	-4.4	-194.53			-58.85				10.1161/CIRCGEN.118.002345
KCNQ1	V	124	I	Neutral	-0.72	0.45	-2.76			-4.76				10.1161/CIRCGEN.118.002345
KCNQ1	Y	125	D	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	H	126	L	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	F	127	L	LOF	5.5					-0.2				10.1161/CIRCGEN.118.002345

KCNQ1	A	128	T	Neutral	-1.18	0.43	-20.63			-14.83				10.1161/CIRCGEN.118.002345
KCNQ1	V	129	I	Neutral	5.5									10.1161/CIRCGEN.118.002345
KCNQ1	L	131	P	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	I	132	L	Neutral	2.11	-0.14	245.28			6.56				10.1161/CIRCGEN.118.002345
KCNQ1	V	133	I	Neutral										10.1161/CIRCGEN.118.002345
KCNQ1	L	134	P	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	V	135	A	Neutral	-2.38	0.24	82.06			-12.2				10.1161/CIRCGEN.118.002345
KCNQ1	V	135	I	Neutral	-4.29	0.93	-16			0				10.1161/CIRCGEN.118.002345
KCNQ1	C	136	F	GOF	-6									10.1161/CIRCGEN.118.002345
KCNQ1	S	140	R	LOF						-47				10.1161/CIRCGEN.118.002345
KCNQ1	T	144	A	GOF	-8									10.1161/CIRCGEN.118.002345
KCNQ1	E	146	K	LOF	7									10.1161/CIRCGEN.118.002345
KCNQ1	A	149	V	Neutral	0.86	-0.59	-22.49			3.74				10.1161/CIRCGEN.118.002345
KCNQ1	A	150	V	Neutral	1.1	0.27	-95.43			-27.47				10.1161/CIRCGEN.118.002345
KCNQ1	A	150	G	LOF	7									10.1161/CIRCGEN.118.002345
KCNQ1	A	150	T	LOF						-20				10.1161/CIRCGEN.118.002345
KCNQ1	T	153	M	LOF						-17				10.1161/CIRCGEN.118.002345
KCNQ1	E	160	K	LOF						-59.01				10.1161/CIRCGEN.118.002345
KCNQ1	V	162	M	LOF	6									10.1161/CIRCGEN.118.002345
KCNQ1	T	169	M	LOF						-16				10.1161/CIRCGEN.118.002345
KCNQ1	R	174	C	LOF						-56.42				10.1161/CIRCGEN.118.002345
KCNQ1	R	174	H	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	R	174	L	LOF							no current			10.1161/CIRCGEN.118.002345

KCNQ1	W	176	R	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	G	179	A	LOF						-49.7				10.1161/CIRCGEN.118.002345
KCNQ1	G	179	S	LOF			-34.54			-51.15				10.1161/CIRCGEN.118.002345
KCNQ1	G	186	C	LOF						-47.3				10.1161/CIRCGEN.118.002345
KCNQ1	L	187	P	Neutral										10.1161/CIRCGEN.118.002345
KCNQ1	G	189	A	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	G	189	E	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	R	190	P	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	R	192	C	LOF						-42				10.1161/CIRCGEN.118.002345
KCNQ1	R	195	P	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	R	195	Q	LOF						-27				10.1161/CIRCGEN.118.002345
KCNQ1	P	197	L	GOF						31				10.1161/CIRCGEN.118.002345
KCNQ1	P	197	S	LOF	16									10.1161/CIRCGEN.118.002345
KCNQ1	K	196	T	LOF	15.59	-5.2	-206.01			-45.11				10.1161/CIRCGEN.118.002345
KCNQ1	D	202	G	LOF							no current			10.1161/CIRCGEN.118.002345
KCNQ1	I	204	F	LOF	13.1	-2.59	210.13			-46.62				10.1161/CIRCGEN.118.002345
KCNQ1	V	207	M	Neutral	1.59	0.31	-72.48			-6.92				10.1161/CIRCGEN.118.002345
KCNQ1	S	209	P	GOF	-2.6	1.28				-12.17				10.1161/CIRCGEN.118.002345
KCNQ1	M	210	I	LOF						-19				10.1161/CIRCGEN.118.002345
KCNQ1	V	215	G	LOF	8									10.1161/CIRCGEN.118.002345
KCNQ1	K	218	E	LOF						-26				10.1161/CIRCGEN.118.002345
KCNQ1	S	225	W	GOF	-13									10.1161/CIRCGEN.118.002345
KCNQ1	I	227	L	LOF	11									10.1161/CIRCGEN.118.002345

KCNQ1	R	231	C	LOF						-29.47				10.1161/CIRCGEN.118.002345
KCNQ1	F	279	I	LOF	1.85	0.08	-118.06			-21.52				10.1161/CIRCGEN.118.002345
KCNQ1	A	300	T	LOF	-15.96	2.9	-29.57			-44.6			mild, LQT phenotype only in hom	10.1161/CIRCGEN.118.002345
KCNQ1	Q	234	P	LOF						-50.43				10.1161/CIRCGEN.118.002345
KCNQ1	L	236	P	LOF						-50.35				10.1161/CIRCGEN.118.002345
KCNQ1	L	236	R	LOF						-51.34				10.1161/CIRCGEN.118.002345
KCNQ1	V	241	I	LOF	5.24	-2.26	-144.16			-29.35				10.1161/CIRCGEN.118.002345
KCNQ1	A	344	V	LOF	17.14	-5.66	499.69			-46.61				10.1161/CIRCGEN.118.002345
KCNQ1	G	245	R	LOF						-50.73			end throughput data by Carlos	10.1161/CIRCGEN.118.002345
KCNQ1	A	46	T	Neutral						0			start - dataset Q1VarPred	10.1161/CIRCGENETICS.117.001754
KCNQ1	V	110	I	LOF	30					-0.6				10.1161/CIRCGENETICS.117.001754
KCNQ1	Y	111	C	LOF						-1		not expressed		10.1161/CIRCGENETICS.117.001754
KCNQ1	L	114	P	LOF						-1		not expressed		10.1161/CIRCGENETICS.117.001754
KCNQ1	P	117	L	LOF						-1		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	S	140	G	GOF						0.5				10.1161/CIRCGENETICS.117.001754
KCNQ1	V	141	M	GOF						2				10.1161/CIRCGENETICS.117.001754
KCNQ1	Q	147	R	LOF						-0.4				10.1161/CIRCGENETICS.117.001754
KCNQ1	G	168	R	LOF						-0.95				10.1161/CIRCGENETICS.117.001754

KCNQ1	R	174	C	LOF	17					-0.53				10.1161/CIRCGENETICS.117.001754
KCNQ1	A	178	T	LOF	45		-0.14			-0.59		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	G	179	S	GOF						-0.46				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	190	Q	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	L	191	P	LOF						-0.78				10.1161/CIRCGENETICS.117.001754
KCNQ1	F	193	L	LOF						-0.2			slow activation	10.1161/CIRCGENETICS.117.001754
KCNQ1	D	202	E	LOF			-0.67			-0.89				10.1161/CIRCGENETICS.117.001754
KCNQ1	D	202	H	LOF	16.6		-0.74			-0.59				10.1161/CIRCGENETICS.117.001754
KCNQ1	D	202	N	LOF	23.8		-0.74			-0.59				10.1161/CIRCGENETICS.117.001754
KCNQ1	D	202	A	Neutral	12									10.1085/jgp.200910351
KCNQ1	D	202	M	LOF	24									10.1085/jgp.200910351
KCNQ1	D	202	K	LOF	22									10.1085/jgp.200910351
KCNQ1	I	204	F	LOF	53.3		-0.57			-0.77			extremely slow activation	10.1161/CIRCGENETICS.117.001754
KCNQ1	I	204	M	LOF	36.1		-0.35			-0.66				10.1161/CIRCGENETICS.117.001754
KCNQ1	I	204	N	LOF	32.9		-0.3							10.1161/CIRCGENETICS.117.001754
KCNQ1	I	204	A	LOF	18.3									10.1085/jgp.200910351
KCNQ1	V	205	L	Neutral	-17.7									10.1085/jgp.200910351
KCNQ1	V	205	A	LOF	50.3									10.1085/jgp.200910351

KCNQ1	V	205	M	LOF	20		-0.58			-0.64				10.1161/CIRCGENETICS.117.001754
KCNQ1	V	207	M	Neutral	7.1		0.2			-0.07				10.1161/CIRCGENETICS.117.001754
KCNQ1	S	209	F	GOF	-48.7					-0.65				10.1161/CIRCGENETICS.117.001754
KCNQ1	S	209	M	GOF	-40.7									10.1085/jgp.200910351
KCNQ1	S	209	A	GOF	-33.7									10.1085/jgp.200910351
KCNQ1	S	209	L	GOF	-47.7									10.1085/jgp.200910351
KCNQ1	S	209	P	GOF	-42.4		4.7			1				10.1161/CIRCGENETICS.117.001754
KCNQ1	V	215	M	LOF	20.2					-0.59				10.1161/CIRCGENETICS.117.001754
KCNQ1	V	215	A	Neutral	-9.7									10.1085/jgp.200910351
KCNQ1	S	225	L	LOF	11					-0.9				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	231	C	LOF						-0.95				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	231	H	LOF	40					-0.85				10.1161/CIRCGENETICS.117.001754
KCNQ1	I	235	N	LOF						-0.9				10.1161/CIRCGENETICS.117.001754
KCNQ1	L	236	R	LOF	54					-1		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	R	243	C	LOF	67					-0.88				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	243	H	LOF						-0.87				10.1161/CIRCGENETICS.117.001754
KCNQ1	W	248	R	LOF						-1				10.1161/CIRCGENETICS.117.001754

KCNQ1	L	251	P	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	V	254	M	LOF	41.5					-0.93				10.1161/CIRCGENETICS.117.001754
KCNQ1	H	258	R	GOF	-44					-0.95		reduced	slow activation	10.1161/CIRCGENETICS.117.001754
KCNQ1	R	259	C	LOF	10					-0.7				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	259	H	GOF	1					1				10.1161/CIRCGENETICS.117.001754
KCNQ1	E	261	D	LOF						-0.91				10.1161/CIRCGENETICS.117.001754
KCNQ1	E	261	K	LOF						-0.95				10.1161/CIRCGENETICS.117.001754
KCNQ1	T	265	I	LOF	8					0			slow activation	10.1161/CIRCGENETICS.117.001754
KCNQ1	G	269	D	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	G	269	S	LOF	70.7		-0.6			-0.85		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	G	272	V	LOF	10					-0.66				10.1161/CIRCGENETICS.117.001754
KCNQ1	F	275	S	LOF	27		1			-0.66		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	S	277	L	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	F	279	I	GOF	-25		0			0.5				10.1161/CIRCGENETICS.117.001754
KCNQ1	Y	281	C	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	A	283	T	LOF	9					-0.8				10.1161/CIRCGENETICS.117.001754

KCNQ1	F	296	S	GOF	-10					-0.88				10.1161/CIRCGENETICS.117.001754
KCNQ1	A	300	T	GOF	-19					-0.85				10.1161/CIRCGENETICS.117.001754
KCNQ1	A	302	V	LOF						-0.95				10.1161/CIRCGENETICS.117.001754
KCNQ1	W	305	S	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	V	307	L	GOF	-18					0.3				10.1161/CIRCGENETICS.117.001754
KCNQ1	V	310	I	LOF	60					-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	I	313	K	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	G	314	S	LOF						-0.88				10.1161/CIRCGENETICS.117.001754
KCNQ1	Y	315	C	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	Y	315	S	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	G	316	E	LOF						-0.82				10.1161/CIRCGENETICS.117.001754
KCNQ1	P	320	A	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	P	320	H	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	T	322	A	LOF						-1		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	T	322	M	LOF						-1		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	G	325	R	LOF						-1				10.1161/CIRCGENETICS.117.001754

KCNQ1	S	338	F	LOF	12					-0.95				10.1161/CIRCGENETICS.117.001754
KCNQ1	F	339	S	LOF	1					-0.96				10.1161/CIRCGENETICS.117.001754
KCNQ1	A	341	V	LOF	60		-0.71			-0.94			slow activation	10.1161/CIRCGENETICS.117.001754
KCNQ1	L	342	F	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	P	343	S	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	A	344	V	LOF	40					0				10.1161/CIRCGENETICS.117.001754
KCNQ1	Q	357	R	LOF	20		0			-0.73		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	R	360	G	LOF						-0.8				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	366	P	LOF	24.1					-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	366	Q	LOF	29					-0.78				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	366	W	LOF	39.2					-0.7		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	A	371	T	LOF	21.9					-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	S	373	P	LOF	37.9					-0.95		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	W	379	R	LOF						-1		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	R	380	S	LOF						-0.67				10.1161/CIRCGENETICS.117.001754
KCNQ1	T	391	I	Neutral						-0.15				10.1161/CIRCGENETICS.117.001754

KCNQ1	W	392	R	LOF	28.3					-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	K	393	M	LOF						-0.67				10.1161/CIRCGENETICS.117.001754
KCNQ1	K	393	N	Neutral						0				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	397	Q	Neutral						-0.1		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	R	397	W	LOF			0			-0.6				10.1161/CIRCGENETICS.117.001754
KCNQ1	V	417	M	Neutral						0				10.1161/CIRCGENETICS.117.001754
KCNQ1	P	448	R	GOF						1				10.1161/CIRCGENETICS.117.001754
KCNQ1	H	455	Y	LOF						-0.57				10.1161/CIRCGENETICS.117.001754
KCNQ1	M	520	R	LOF						-1		not expressed		10.1161/CIRCGENETICS.117.001754
KCNQ1	Y	522	S	LOF	7					-0.9		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	A	525	T	LOF	22		0.08			-0.64		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	R	533	W	Neutral	13.9					-0.28				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	539	W	LOF	33.9		-0.59			-0.83				10.1161/CIRCGENETICS.117.001754
KCNQ1	S	546	L	LOF	50.7		-0.19			-0.75				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	555	C	LOF	60					-0.75				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	555	H	LOF	50		-0.28			-0.88				10.1161/CIRCGENETICS.117.001754

KCNQ1	K	557	E	LOF						-1				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	562	M	LOF	43.3		0.07			-0.57				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	583	H	Neutral						0				10.1161/CIRCGENETICS.117.001754
KCNQ1	G	589	D	LOF	33					-0.85		reduced		10.1161/CIRCGENETICS.117.001754
KCNQ1	A	590	T	LOF	10					-0.55				10.1161/CIRCGENETICS.117.001754
KCNQ1	R	594	Q	LOF	60					-0.95				10.1161/CIRCGENETICS.117.001754
KCNQ1	D	611	Y	Neutral						0				10.1161/CIRCGENETICS.117.001754
KCNQ1	L	619	M	LOF						-0.99				10.1161/CIRCGENETICS.117.001754
KCNQ1	G	643	S	LOF	1.1		-0.28			-0.65			end - dataset Q1VarPred	10.1161/CIRCGENETICS.117.001754
KCNQ2	R	333	Q	LOF	-1.4	0.1				-0.3				10.1093/brain/awg286
KCNQ2	M	208	V	LOF	-0.1	0.1				-0.15				10.1093/brain/awg286
KCNQ2	T	359	K	LOF	6.2	1.4				-672			slower activation	10.1016/j.neulet.2009.06.064
KCNQ2	N	258	S	LOF	1	0				-218		reduced		10.1002/humu.21554
KCNQ2	I	205	V	LOF	24					-0.1			slowing of activation and deactivation	10.1002/ana.24080
KCNQ2	R	213	Q	LOF	61					-0.4			slowing of activation and deactivation	10.1002/ana.24080
KCNQ2	A	265	P	LOF						-0.65				10.1002/ana.24080
KCNQ2	T	274	M	LOF						-0.8				10.1002/ana.24080
KCNQ2	G	290	D	LOF	10					-0.68				10.1002/ana.24080

KCNQ2	M	518	V	LOF						0				10.1002/ana.24080
KCNQ2	R	532	W	LOF	17.1					0		reduced	slower activation, faster deactivation	10.1002/ana.24080
KCNQ2	R	144	Q	GOF	-20.2	5.6				6				10.1523/JNEUROSCI.4423-14.2015
KCNQ2	R	201	C	GOF						29			loss of time dependence of activation	10.1523/JNEUROSCI.4423-14.2015
KCNQ2	R	201	H	GOF	-29.4	13.1				20				10.1523/JNEUROSCI.4423-14.2015
KCNQ2	D	172	R	LOF	14.7									10.1523/JNEUROSCI.4423-14.2015
KCNQ2	D	172	C	LOF	22.7									10.1523/JNEUROSCI.4423-14.2015
KCNQ2	E	130	R	LOF	86.7									10.1523/JNEUROSCI.4423-14.2015
KCNQ2	E	130	C	LOF	27.7									10.1523/JNEUROSCI.4423-14.2015
KCNQ2	E	140	R	LOF								not expressed		10.1523/JNEUROSCI.4423-14.2015
KCNQ2	E	140	C	LOF								not expressed		10.1523/JNEUROSCI.4423-14.2015
KCNQ2	L	351	F	LOF	-0.8	0.7				-9			currents estimated from figure	10.1016/j.bbadis.2015.06.012
KCNQ2	L	351	V	GOF	-2.8	-1.5				0			currents estimated from figure	10.1016/j.bbadis.2015.06.012
KCNQ2	Y	362	C	GOF	-3	-0.3				12			currents estimated from figure	10.1016/j.bbadis.2015.06.012
KCNQ2	R	553	Q	LOF	0.7	-1.1				-13			currents estimated from figure	10.1016/j.bbadis.2015.06.012
KCNQ2	W	344	R	LOF							no current			10.1016/j.bbadis.2015.06.012
KCNQ2	E	130	R	LOF	84.5	20				-4.2				10.3390/ijms20143382
KCNQ2	E	140	Q	LOF	71.1	10.1				-13.6			faster deactivation	10.3390/ijms20143382
KCNQ2	E	140	R	LOF						-32.4				10.3390/ijms20143382
KCNQ2	D	172	R	LOF	13.1	5.8				-20				10.3390/ijms20143382
KCNQ2	R	207	E	LOF	68.8	7				-1.4				10.3390/ijms20143382
KCNQ2	R	210	E	LOF	74.1	11.4				40.5				10.3390/ijms20143382
KCNQ2	R	210	D	LOF	225	27.3				-6.3				10.3390/ijms20143382

KCNQ2	R	213	D	LOF						-33				10.3390/ijms20143382
KCNQ2	P	285	T	LOF	1.4	-0.6				-8.2				10.1038/s41598-020-70212-w
KCNQ2	S	247	L	LOF	2.9	-1.6				-14		reduced		10.1038/s41598-020-70212-w
KCNQ2	S	247	W	LOF	3.4	-3.1				-16.5		reduced		10.1038/s41598-020-70212-w
KCNQ2	V	175	L	GOF	-15.4	0.35				45			activation twice as fast, deactivation and ion selectivity unchanged	10.1111/epi.13366
KCNQ2	R	198	Q	GOF	-29.3	5.6				23.7			slower deactivation	10.1111/epi.13601
KCNQ2	R	198	D	GOF						13.7			completely abolished voltage-dependent gating	10.1111/epi.13601
KCNQ2	R	198	E	GOF						37.7			completely abolished voltage-dependent gating	10.1111/epi.13601
KCNQ2	E	130	K	LOF						-57.3			homomeric channels w/o current	10.1111/epi.14609
KCNQ2	L	243	F	LOF	-0.7	-0.3				-20.9			homomeric channels w/o current	10.1111/epi.14609
KCNQ2	W	270	R	LOF	-7.3	-2.2				-8.1			homomeric channels w/o current	10.1111/epi.14609
KCNQ2	G	281	R	LOF						-56			homomeric channels w/o current	10.1111/epi.14609
KCNQ2	R	207	W	LOF	15.8					-0.75			slow activation, current estimated from figure	10.1073/pnas.211431298
KCNQ2	R	214	W	LOF	0					-0.75			slow activation, current estimated from figure	10.1073/pnas.211431298
KCNQ2	R	198	Q	GOF	-26.5	2.7				-15				10.1529/biophysj.107.128371
KCNQ2	R	201	Q	GOF						-4			significant loss of time-dependent kinetics (linear I/V, leak channel)	10.1529/biophysj.107.128371
KCNQ2	R	207	Q	LOF	22.6	0.6				-3				10.1529/biophysj.107.128371
KCNQ2	R	210	Q	LOF	39.5	6.2				-10				10.1529/biophysj.107.128371

KCNQ2	R	213	Q	LOF	64.3	2.4				-9				10.1529/biophysj.107.128371
KCNQ2	R	214	Q	LOF	34	2.6				-17				10.1529/biophysj.107.128371
KCNQ2	R	207	W	LOF	29.5	0							altered tau activation	10.1529/biophysj.107.128371
KCNQ2	R	214	W	LOF	12.5	3.3							activation kinetics estimated from figure	10.1529/biophysj.107.128371
KCNQ2	G	271	V	LOF	3.1	2.53				-80.98			slower activation	10.1016/j.brainres.2015.04.060
KCNQ2	R	325	G	LOF						-27.7			additional data on PIP modulation	10.1038/srep38167
KCNQ2	A	294	V	LOF	0	0	0			-0.83			subcellular mislocalization	10.1016/j.nbd.2015.04.017
KCNQ2	A	294	G	Neutral						-0.3				10.1016/j.nbd.2015.04.017
KCNQ2	E	119	G	LOF	6.8	0.3				-0.2			mild slowing of activation	10.1113/jphysiol.2007.143826
KCNQ2	S	122	L	LOF	6	2.6				-0.1			current estimated from figure, slower activation	10.1016/j.nbd.2006.06.011
KCNQ2	T	114	A	LOF	24.8		-13.7			-0.45			slower activation	10.1007/s00424-012-1184-x
KCNQ2	K	526	N	LOF	11.4	2.6				-2.7				10.1212/01.WNL.0000132979.08394.6D
KCNQ2	L	203	P	LOF	24.4	-0.7				251.3			slow activation	10.1038/s41598-020-61697-6
KCNQ2	L	268	F	LOF	-4.9	-0.4				-689.7		reduced		10.1038/s41598-020-61697-6
KCNQ2	K	552	T	LOF	0.8	-1.5				-786.6		reduced		10.1038/s41598-020-61697-6
KCNQ2	R	553	L	LOF	-4.5	-0.8				-65.3		reduced	interesting PIP2 binding effects	10.1038/s41598-020-61697-6
KCNQ2	Y	284	C	LOF						-0.25				10.1038/25367
KCNQ2	G	310	V	LOF						-0.2				10.1038/25367
KCNQ2	A	306	T	LOF						-0.3				10.1038/25367
KCNQ2	G	279	S	LOF						-13			dominant negative	10.1038/25367
KCNQ3	N	468	S	Neutral	0.6	1.9				-0.1			currents estimated from figure	10.1093/brain/awg286
KCNQ3	D	305	G	LOF	0.8	1.7	0			-0.4				10.1093/brain/awg286
KCNQ3	R	227	Q	GOF	-5.9	1.1				-32.2			homozygotes have extreme GOF	10.1002/ana.25522

KCNQ3	R	230	C	GOF	-6.3	1.7				-24.7				10.1002/ana.25522
KCNQ3	R	230	S	GOF	-5.4	1.4				-16.8				10.1002/ana.25522
KCNQ3	R	230	H	GOF	-5.9	0.6				-10.5				10.1002/ana.25522
KCNQ3	M	240	R	LOF	45.8	11.4				-56.2				10.3389/fphys.2020.01040
KCNQ3	W	309	R	LOF	7.5	1.9				-0.6			currents estimated, slower activation	10.1007/s00232-008-9097-5
KCNQ3	W	309	F	LOF									data only for homozygote	10.1007/s00232-008-9097-5
KCNQ3	W	309	Y	LOF									data only for homozygote	10.1007/s00232-008-9097-5
KCNQ3	G	310	V	LOF						-0.2			homozygote w/o current	10.1038/25367
KCNQ3	T	313	I	LOF	4.2	-0.8							homomeric channels non-functional	10.1002/epi4.12438
KCNQ3	T	313	M	LOF									strong reduction of peak current, no exact value given	10.1002/epi4.12438
KCNQ3	V	279	F	LOF	0	0				-0.88				10.1159/000447461
KCNQ3	R	230	K	GOF	-48.5	0.7								10.1085/jgp.201812221
KCNQ3	R	230	H	GOF	-78.1	4.9								10.1085/jgp.201812221
KCNQ3	V	359	L	LOF	7	-0.4				-13.3				10.1007/s12035-018-0883-5
KCNQ3	D	542	N	LOF	2.6	-1.9				-21.5				10.1007/s12035-018-0883-5
KCNQ4	G	285	S	LOF						-0.9				10.1016/s0092-8674(00)80556-5
KCNQ4	G	296	S	LOF	0	0				-0.9		reduced		10.1007/s00439-007-0447-7
KCNQ4	L	274	H	LOF								reduced		10.1074/jbc.M110.179010
KCNQ4	W	276	S	LOF	3	0				-142		reduced	slower activation	10.1074/jbc.M110.179010
KCNQ4	L	281	S	LOF								reduced	faster activation	10.1074/jbc.M110.179010
KCNQ4	G	285	C	LOF								reduced	faster activation, faster deactivation	10.1074/jbc.M110.179010

KCNQ4	G	296	S	LOF								reduced	faster activation, faster deactivation	10.1074/jbc.M110.179010
KCNQ4	G	321	S	LOF	0	0	0			-0.7			faster activation, current estimated from figure	10.1074/jbc.M110.179010
KCNQ4	F	182	L	Neutral	0	0				0			faster activation, faster deactivation	10.1074/jbc.M110.179010
KCNQ4	W	276	S	LOF						-0.9				10.1111/j.1476-5381.2011.01697.x
KCNQ4	L	281	S	LOF						-0.9				10.1111/j.1476-5381.2011.01697.x
KCNQ4	G	285	S	LOF						-0.9				10.1111/j.1476-5381.2011.01697.x
KCNQ4	G	296	S	LOF						-0.9				10.1111/j.1476-5381.2011.01697.x
KCNQ4	L	274	H	LOF								reduced		10.1111/jcmm.12080
KCNQ4	W	276	S	LOF								reduced		10.1111/jcmm.12080
KCNQ4	L	281	S	LOF						-13.11		reduced		10.1111/jcmm.12080
KCNQ4	G	285	C	LOF								reduced		10.1111/jcmm.12080
KCNQ4	G	285	S	LOF								reduced		10.1111/jcmm.12080
KCNQ4	G	296	S	LOF						-13.79		reduced		10.1111/jcmm.12080
KCNQ4	G	321	S	LOF						-14.09		reduced		10.1111/jcmm.12080
KCNQ4	D	266	Y	LOF						-0.3				10.1038/s41598-018-34876-9
KCNQ4	L	47	P	LOF	0	0				-60			current estimated from figure	10.1002/humu.23698
KCNQ4	N	264	S	LOF						-96.9				10.1038/s12276-019-0300-9
KCNQ4	S	269	F	LOF						-103				10.1038/s12276-019-0300-9
KCNQ4	S	273	A	LOF						-74.2				10.1038/s12276-019-0300-9
KCNQ4	T	278	A	LOF						-87				10.1038/s12276-019-0300-9
KCNQ4	R	433	W	LOF						-71.6				10.1038/s12276-019-0300-9

KCNQ4	L	281	M	LOF						-77.5				10.1038/s12276-019-0300-9
KCNQ4	L	295	P	LOF						-70.7				10.1038/s12276-019-0300-9
KCNQ4	H	455	Q	Neutral						-14.4				10.1038/s12276-019-0300-9
KCNQ4	W	267	S	LOF						-84.8				10.1038/s12276-019-0300-9
KCNQ4	S	680	F	Neutral						0				10.1016/j.heares.2020.107884
KCNQ4	E	260	K	LOF						-54.1		reduced		10.1016/j.heares.2020.107884
KCNQ4	P	291	S	LOF						-34.3		reduced		10.1016/j.heares.2020.107884
KCNQ4	V	230	E	LOF						-46.8				10.1016/j.heares.2020.107884
KCNQ4	D	262	V	LOF						-43.6				10.1016/j.heares.2020.107884
KCNQ4	Y	270	H	LOF						-35.8				10.1016/j.heares.2020.107884
KCNQ4	W	275	R	LOF						-56				10.1016/j.heares.2020.107884
KCNQ4	G	287	R	LOF						-41.1				10.1016/j.heares.2020.107884
KCNQ4	P	291	L	LOF						-41.1				10.1016/j.heares.2020.107884
KCNQ4	Y	160	A	LOF							no current			10.3389/fphys.2020.604134
KCNQ4	G	162	V	LOF							no current			10.3389/fphys.2020.604134
KCNQ4	W	163	A	Neutral										10.3389/fphys.2020.604134
KCNQ4	Q	164	A	Neutral										10.3389/fphys.2020.604134
KCNQ4	C	156	A	GOF	-15.7								faster activation	10.3389/fphys.2020.604134
KCNQ4	C	157	A	GOF	-11.4								faster activation	10.3389/fphys.2020.604134
KCNQ4	C	158	V	GOF	-12.4								faster activation	10.3389/fphys.2020.604134
KCNQ4	R	159	A	GOF	-17.5								faster activation	10.3389/fphys.2020.604134
KCNQ4	R	161	A	GOF	-14.8								faster activation	10.3389/fphys.2020.604134
KCNQ5	R	359	C	LOF						-595				10.1101/2021.04.20.21255696

KCNQ5	L	692	V	LOF	0.4	5.33				-508.6				10.1101/2021.04.20.21255696
KCNQ5	Q	735	R	LOF	2.56	1.36				-481.3				10.1101/2021.04.20.21255696
KCNQ5	F	165	I	LOF	6.49	-1.02				52.7				10.1101/2021.04.20.21255696
KCNQ5	L	926	S	LOF	3.78	-0.31				5.2				10.1101/2021.04.20.21255696
KCNQ5	V	145	G	LOF	7.4	7.6				-0.5		reduced	slow activation	10.1016/j.ajhg.2017.05.016
KCNQ5	L	341	I	LOF	3.3	-3.8				-0.45		reduced	slow activation	10.1016/j.ajhg.2017.05.016
KCNQ5	S	448	I	GOF	-0.4	1.5				0.3			slow activation	10.1016/j.ajhg.2017.05.016
KCNQ5	P	369	R	GOF	-30.8	-3.2				-0.3			fast activation, slow deactivation	10.1016/j.ajhg.2017.05.016

Supplementary Table 2. Non-synonymous variants in voltage-gated potassium channels used in training and their overall effect on ion channel function (label). The label is based on the original authors' assessment and our independent external validation, with the experimental data shown used to verify the label. For each variant, we assessed whether coexpression yielded significant differences of these parameters in comparison to the wildtype. Hyperpolarizing shifts of the activation curve, depolarizing shifts in the inactivation curve and an increase of peak current amplitude were labeled as gain-of-function. Trafficking defects, depolarizing shifts in the activation curve, hyperpolarizing shifts in the inactivation curve and decrease of peak current amplitude were labeled as loss-of-function. If there were conflicting results, we ranked trafficking defects above gating kinetics above peak current. Variants that did not significantly differ in these regards were labeled as neutral. Abbreviations: LOF – loss-of-function; GOF – gain-of-function.

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gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA1	1	M	L	NM_000217.3(KCNA1):c.1A>T (p.Met1Leu)	Episodic ataxia type 1	Uncertain significance(Last reviewed: May 27, 2019)	criteria provided, single submitter	VCV000946822	12	4911379	946822	926653	rs776861490	Neutral	0.662	0.221	0.117
KCNA1	1	M	R	NM_000217.3(KCNA1):c.2T>G (p.Met1Arg)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Nov 6, 2019)	criteria provided, single submitter	VCV000967035	12	4911380	967035	956894	rs1947350138	Neutral	0.683	0.192	0.124
KCNA1	1	M	V	NM_000217.3(KCNA1):c.1A>G (p.Met1Val)	Episodic ataxia type 1	Benign(Last reviewed: Mar 26, 2018)	criteria provided, single submitter	VCV000883201	12	4911379	883201	869942	rs776861490	Neutral	0.656	0.224	0.12
KCNA1	4	M	I	NM_000217.3(KCNA1):c.12G>T (p.Met4Ile)	not provided	Uncertain significance(Last reviewed: Dec 20, 2019)	criteria provided, single submitter	VCV000586087	12	4911390	586087	577272	rs773128496	Neutral	0.585	0.292	0.123
KCNA1	7	E	K	NM_000217.3(KCNA1):c.19G>A (p.Glu7Lys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000883202	12	4911397	883202	869943	rs529968149	Neutral	0.617	0.228	0.155
KCNA1	10	D	E	NM_000217.3(KCNA1):c.30C>G (p.Asp10Glu)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Dec 3, 2018)	criteria provided, single submitter	VCV000642028	12	4911408	642028	641181	rs1471834737	Neutral	0.626	0.232	0.142
KCNA1	13	S	L	NM_000217.3(KCNA1):c.38C>T (p.Ser13Leu)	not provided	Uncertain significance(Last reviewed: Jul 18, 2019)	criteria provided, single submitter	VCV001316112	12	4911416	1316112	1308514	NA	Neutral	0.568	0.29	0.142
KCNA1	15	A	S	NM_000217.3(KCNA1):c.43G>T (p.Ala15Ser)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Apr 6, 2020)	criteria provided, single submitter	VCV000961458	12	4911421	961458	948051	rs777276806	Neutral	0.647	0.216	0.137
KCNA1	17	G	R	NM_000217.3(KCNA1):c.49G>C (p.Gly17Arg)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jul 31, 2019)	criteria provided, single submitter	VCV000955397	12	4911427	955397	948052	rs1324506346	Neutral	0.669	0.192	0.138
KCNA1	18	H	P	NM_000217.3(KCNA1):c.53A>C (p.His18Pro)	not provided|Episodic ataxia type 1	Uncertain significance(Last reviewed: Jul 17, 2019)	criteria provided, multiple submitters, no conflicts	VCV000374425	12	4911431	374425	361311	rs367921276	Neutral	0.642	0.224	0.134
KCNA1	20	Q	H	NM_000217.3(KCNA1):c.60G>C (p.Gln20His)	not provided|Episodic ataxia type 1	Conflicting interpretations of pathogenicity(Last reviewed: Sep 1, 2021)	criteria provided, conflicting interpretations	VCV000447612	12	4911438	447612	441551	rs201504073	Neutral	0.606	0.254	0.141
KCNA1	21	D	N	NM_000217.3(KCNA1):c.61G>A (p.Asp21Asn)	not provided	Uncertain significance(Last reviewed: Jun 5, 2019)	criteria provided, single submitter	VCV000804963	12	4911439	804963	793434	rs747465523	Neutral	0.611	0.238	0.15
KCNA1	24	Y	H	NM_000217.3(KCNA1):c.70T>C (p.Tyr24His)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV000950047	12	4911448	950047	926654	rs1224258529	Neutral	0.664	0.218	0.117
KCNA1	26	R	W	NM_000217.3(KCNA1):c.76C>T (p.Arg26Trp)	not specified	Uncertain significance(Last reviewed: Oct 5, 2015)	criteria provided, single submitter	VCV000435546	12	4911454	435546	429429	rs373645838	Neutral	0.617	0.259	0.124
KCNA1	32	D	H	NM_000217.3(KCNA1):c.94G>C (p.Asp32His)	not provided	Uncertain significance(Last reviewed: Sep 6, 2019)	criteria provided, single submitter	VCV001318467	12	4911472	1318467	1308251	NA	Neutral	0.568	0.274	0.158

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA1	32	D	Y	NM_000217.3(KCNA1):c.94G>T (p.Asp32Tyr)	not provided	Uncertain significance(Last reviewed: Nov 19, 2021)	criteria provided, single submitter	VCV001316825	12	4911472	1316825	1307919	NA	Neutral	0.549	0.291	0.16
KCNA1	46	L	M	NM_000217.3(KCNA1):c.136C>A (p.Leu46Met)	Episodic ataxia type 1|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Dec 1, 2020)	criteria provided, conflicting interpretations	VCV000639858	12	4911514	639858	641182	rs149959487	LOF	0.842	0.086	0.072
KCNA1	46	L	P	NM_000217.3(KCNA1):c.137T>C (p.Leu46Pro)	not provided	Uncertain significance(Last reviewed: May 29, 2019)	criteria provided, single submitter	VCV001191407	12	4911515	1191407	1181016	NA	LOF	0.848	0.082	0.07
KCNA1	60	N	S	NM_000217.3(KCNA1):c.179A>G (p.Asn60Ser)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Mar 2, 2020)	criteria provided, single submitter	VCV001003091	12	4911557	1003091	995242	rs752067203	LOF	0.717	0.153	0.129
KCNA1	71	R	H	NM_000217.3(KCNA1):c.212G>A (p.Arg71His)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000883997	12	4911590	883997	869944	rs1947351910	LOF	0.754	0.155	0.091
KCNA1	75	P	H	NM_000217.3(KCNA1):c.224C>A (p.Pro75His)	Episodic ataxia type 1|Inborn genetic diseases	Uncertain significance(Last reviewed: Jun 28, 2019)	criteria provided, multiple submitters, no conflicts	VCV000463862	12	4911602	463862	462249	rs897263951	LOF	0.752	0.127	0.121
KCNA1	92	I	V	NM_000217.3(KCNA1):c.274A>G (p.Ile92Val)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000883998	12	4911652	883998	869945	rs962399769	LOF	0.827	0.09	0.083
KCNA1	97	Q	K	NM_000217.3(KCNA1):c.289C>A (p.Gln97Lys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Nov 6, 2019)	criteria provided, single submitter	VCV000942661	12	4911667	942661	936146	rs1166033590	LOF	0.844	0.079	0.076
KCNA1	104	R	S	NM_000217.3(KCNA1):c.312G>T (p.Arg104Ser)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Mar 13, 2019)	criteria provided, single submitter	VCV000863541	12	4911690	863541	839993	rs1947352517	LOF	0.751	0.147	0.102
KCNA1	106	V	A	NM_000217.3(KCNA1):c.317T>C (p.Val106Ala)	Myokymia|Episodic ataxia type 1	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000309144	12	4911695	309144	332921	rs886049510	Neutral	0.7	0.191	0.108
KCNA1	106	V	I	NM_000217.3(KCNA1):c.316G>A (p.Val106Ile)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Feb 27, 2018)	criteria provided, single submitter	VCV001056453	12	4911694	1056453	1047988	NA	Neutral	0.642	0.242	0.116
KCNA1	114	S	F	NM_000217.3(KCNA1):c.341C>T (p.Ser114Phe)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jan 25, 2020)	criteria provided, single submitter	VCV000661566	12	4911719	661566	641183	rs1591627522	LOF	0.742	0.157	0.1
KCNA1	121	E	K	NM_000217.3(KCNA1):c.361G>A (p.Glu121Lys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV001014549	12	4911739	1014549	1010473	rs1463513823	LOF	0.753	0.104	0.143
KCNA1	124	E	K	NM_000217.3(KCNA1):c.370G>A (p.Glu124Lys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Aug 3, 2017)	criteria provided, single submitter	VCV000463863	12	4911748	463863	462507	rs1555085687	LOF	0.772	0.12	0.108
KCNA1	138	K	Q	NM_000217.3(KCNA1):c.412A>C (p.Lys138Gln)	Myokymia|Episodic ataxia type 1	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000309145	12	4911790	309145	317395	rs886049511	Neutral	0.624	0.227	0.15

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA1	139	E	K	NM_000217.3(KCNA1):c.415G>A (p.Glu139Lys)	not provided	Uncertain significance(Last reviewed: Oct 1, 2018)	criteria provided, single submitter	VCV000623795	12	4911793	623795	612952	rs1565433107	LOF	0.686	0.161	0.153
KCNA1	141	E	K	NM_000217.3(KCNA1):c.421G>A (p.Glu141Lys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Feb 12, 2020)	criteria provided, single submitter	VCV000948787	12	4911799	948787	926655	rs934102011	Neutral	0.652	0.192	0.156
KCNA1	149	Y	C	NM_000217.3(KCNA1):c.446A>G (p.Tyr149Cys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Feb 2, 2018)	criteria provided, single submitter	VCV000880707	12	4911824	880707	869946	rs1947353699	LOF	0.816	0.102	0.081
KCNA1	151	R	C	NM_000217.3(KCNA1):c.451C>T (p.Arg151Cys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV001023031	12	4911829	1023031	1010474	rs1947353744	LOF	0.826	0.086	0.088
KCNA1	159	Y	H	NM_000217.3(KCNA1):c.475T>C (p.Tyr159His)	not provided|Episodic ataxia type 1	Uncertain significance(Last reviewed: Sep 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000586091	12	4911853	586091	577273	rs764645312	LOF	0.766	0.136	0.098
KCNA1	166	A	P	NM_000217.3(KCNA1):c.496G>C (p.Ala166Pro)	not specified	Uncertain significance(Last reviewed: May 3, 2017)	criteria provided, single submitter	VCV000447611	12	4911874	447611	441554	rs1179635654	LOF	0.902	0.045	0.053
KCNA1	170	A	T	NM_000217.3(KCNA1):c.508G>A (p.Ala170Thr)	not provided	Uncertain significance(Last reviewed: Oct 1, 2019)	criteria provided, single submitter	VCV000872570	12	4911886	872570	859985	rs1947354204	LOF	0.892	0.028	0.08
KCNA1	174	V	F	NM_000217.3(KCNA1):c.520G>T (p.Val174Phe)	Inborn genetic diseases|Episodic ataxia type 1	Pathogenic(Last reviewed: Dec 26, 2018)	criteria provided, single submitter	VCV000013482	12	4911898	13482	28521	rs104894349	LOF	0.875	0.024	0.101
KCNA1	174	V	I	NM_000217.3(KCNA1):c.520G>A (p.Val174Ile)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Feb 23, 2020)	criteria provided, single submitter	VCV000959065	12	4911898	959065	948053	rs104894349	LOF	0.823	0.044	0.132
KCNA1	177	I	N	NM_000217.3(KCNA1):c.530T>A (p.Ile177Asn)	Episodic ataxia type 1	Pathogenic(Last reviewed: Apr 1, 1998)	no assertion criteria provided	VCV000013488	12	4911908	13488	28527	rs267607195	LOF	0.94	0.01	0.05
KCNA1	184	F	C	NM_000217.3(KCNA1):c.551T>G (p.Phe184Cys)	Episodic ataxia type 1	Pathogenic(Last reviewed: Dec 1, 1995)	no assertion criteria provided	VCV000013484	12	4911929	13484	28523	rs104894357	LOF	0.917	0.021	0.061
KCNA1	187	E	K	NM_000217.3(KCNA1):c.559G>A (p.Glu187Lys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Mar 12, 2018)	criteria provided, single submitter	VCV000573452	12	4911937	573452	565532	rs1565433190	LOF	0.911	0.02	0.069
KCNA1	195	D	E	NM_000217.3(KCNA1):c.585C>G (p.Asp195Glu)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Mar 7, 2020)	criteria provided, single submitter	VCV000855826	12	4911963	855826	839994	rs1947354740	LOF	0.76	0.105	0.135
KCNA1	199	T	M	NM_000217.3(KCNA1):c.596C>T (p.Thr199Met)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV000835844	12	4911974	835844	839995	rs1207573200	Neutral	0.638	0.205	0.156
KCNA1	200	G	A	NM_000217.3(KCNA1):c.599G>C (p.Gly200Ala)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Dec 4, 2017)	criteria provided, single submitter	VCV000532541	12	4911977	532541	527204	rs1411072418	Neutral	0.698	0.17	0.132
KCNA1	204	R	H	NM_000217.3(KCNA1):c.611G>A (p.Arg204His)	Episodic ataxia type 1|not provided	Conflicting interpretations of pathogenicity(Last	criteria provided, conflicting	VCV000290682	12	4911989	290682	274919	rs2229000	LOF	0.794	0.12	0.086

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Dec 3, 2020)	interpretations										
KCNA1	214	S	F	NM_000217.3(KCNA1):c.641C>T (p.Ser214Phe)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jan 21, 2020)	criteria provided, single submitter	VCV000998939	12	4912019	998939	995243	rs764561596	LOF	0.728	0.122	0.15
KCNA1	215	N	I	NM_000217.3(KCNA1):c.644A>T (p.Asn215Ile)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jul 20, 2020)	criteria provided, single submitter	VCV001017234	12	4912022	1017234	1010475	rs762016621	LOF	0.734	0.116	0.15
KCNA1	218	T	R	NM_000217.3(KCNA1):c.653C>G (p.Thr218Arg)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jun 29, 2020)	criteria provided, single submitter	VCV000955267	12	4912031	955267	948054	rs1280516411	LOF	0.81	0.05	0.14
KCNA1	223	I	V	NM_000217.3(KCNA1):c.667A>G (p.Ile223Val)	Episodic ataxia type 1	Uncertain significance(Last reviewed: May 20, 2020)	criteria provided, single submitter	VCV001006491	12	4912045	1006491	995244	rs146948558	LOF	0.849	0.045	0.106
KCNA1	224	V	L	NM_000217.3(KCNA1):c.670G>T (p.Val224Leu)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Sep 11, 2020)	criteria provided, single submitter	VCV001043157	12	4912048	1043157	1030984	rs1947355480	LOF	0.846	0.041	0.113
KCNA1	226	T	A	NM_000217.3(KCNA1):c.676A>G (p.Thr226Ala)	Episodic ataxia type 1	Pathogenic(Last reviewed: Aug 16, 2012)	no assertion criteria provided	VCV000013486	12	4912054	13486	28525	rs104894354	LOF	0.882	0.032	0.085
KCNA1	226	T	K	NM_000217.3(KCNA1):c.677C>A (p.Thr226Lys)	Myokymia 1	Pathogenic(Last reviewed: Apr 1, 2007)	no assertion criteria provided	VCV000013493	12	4912055	13493	28532	rs28933383	LOF	0.909	0.02	0.071
KCNA1	226	T	M	NM_000217.3(KCNA1):c.677C>T (p.Thr226Met)	Episodic ataxia type 1	Pathogenic(Last reviewed: Aug 16, 2012)	no assertion criteria provided	VCV000021127	12	4912055	21127	33979	rs28933383	LOF	0.891	0.028	0.082
KCNA1	226	T	R	NM_000217.3(KCNA1):c.677C>G (p.Thr226Arg)	not provided|Episodic ataxia/myokymia syndrome|Episodic ataxia type 1	Pathogenic(Last reviewed: Nov 16, 2021)	criteria provided, multiple submitters, no conflicts	VCV000013492	12	4912055	13492	28531	rs28933383	LOF	0.912	0.019	0.069
KCNA1	234	F	L	NM_000217.3(KCNA1):c.702C>A (p.Phe234Leu)	not provided	Uncertain significance(Last reviewed: Jun 4, 2021)	criteria provided, single submitter	VCV001317352	12	4912080	1317352	1306296	NA	LOF	0.865	0.041	0.093
KCNA1	239	R	S	NM_000217.3(KCNA1):c.715C>A (p.Arg239Ser)	Episodic ataxia type 1	Pathogenic(Last reviewed: Dec 1, 1995)	no assertion criteria provided	VCV000013481	12	4912093	13481	28520	rs104894348	LOF	0.916	0.02	0.064
KCNA1	242	A	P	NM_000217.3(KCNA1):c.724G>C (p.Ala242Pro)	not provided|Myokymia 1	Pathogenic(Last reviewed: Oct 14, 2016)	criteria provided, single submitter	VCV000013489	12	4912102	13489	28528	rs28933381	LOF	0.877	0.06	0.063
KCNA1	244	P	H	NM_000217.3(KCNA1):c.731C>A (p.Pro244His)	Myokymia 1	Pathogenic(Last reviewed: Oct 1, 2000)	no assertion criteria provided	VCV000013490	12	4912109	13490	28529	rs28933382	Neutral	0.485	0.403	0.111
KCNA1	249	F	I	NM_000217.3(KCNA1):c.745T>A (p.Phe249Ile)	Episodic ataxia type 1	Pathogenic(Last reviewed: Dec 1, 1995)	no assertion criteria provided	VCV000013483	12	4912123	13483	28522	rs104894356	LOF	0.855	0.046	0.099
KCNA1	253	I	F	NM_000217.3(KCNA1):c.757A>T (p.Ile253Phe)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Sep 17, 2019)	criteria provided, single submitter	VCV000955199	12	4912135	955199	948056	rs1947356316	LOF	0.853	0.029	0.118
KCNA1	253	I	V	NM_000217.3(KCNA1):c.757A>G (p.Ile253Val)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Sep 19, 2019)	criteria provided, single submitter	VCV000964532	12	4912135	964532	948055	rs1947356316	LOF	0.807	0.042	0.152

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA1	255	N	D	NM_000217.3(KCNA1):c.763A>G (p.Asn255Asp)	Myokymia 1 with hypomagnesemia	Pathogenic(Last reviewed: Jan 1, 2010)	no assertion criteria provided	VCV000013494	12	4912141	13494	28533	rs121918067	LOF	0.905	0.023	0.071
KCNA1	265	Y	C	NM_000217.3(KCNA1):c.794A>G (p.Tyr265Cys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Mar 20, 2020)	criteria provided, single submitter	VCV000846128	12	4912172	846128	839996	rs1947356506	LOF	0.927	0.021	0.052
KCNA1	268	T	K	NM_000217.3(KCNA1):c.803C>A (p.Thr268Lys)	not provided	Uncertain significance(Last reviewed: Sep 16, 2018)	no assertion criteria provided	VCV000591423	12	4912181	591423	582414	rs1565433356	LOF	0.909	0.021	0.07
KCNA1	279	N	K	NM_000217.3(KCNA1):c.837C>A (p.Asn279Lys)	not provided	Uncertain significance(Last reviewed: Dec 31, 2018)	criteria provided, single submitter	VCV000804964	12	4912215	804964	793435	rs1591627786	Neutral	0.668	0.178	0.154
KCNA1	291	L	H	NM_000217.3(KCNA1):c.872T>A (p.Leu291His)	not provided	Uncertain significance(Last reviewed: Mar 21, 2017)	criteria provided, single submitter	VCV000424183	12	4912250	424183	408684	rs1064796844	LOF	0.927	0.01	0.063
KCNA1	295	R	H	NM_000217.3(KCNA1):c.884G>A (p.Arg295His)	not provided	Uncertain significance(Last reviewed: Sep 11, 2017)	criteria provided, single submitter	VCV000586092	12	4912262	586092	577274	rs778463081	LOF	0.905	0.017	0.078
KCNA1	296	L	F	NM_000217.3(KCNA1):c.888G>T (p.Leu296Phe)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Dec 5, 2020)	criteria provided, single submitter	VCV001029721	12	4912266	1029721	1017632	rs1315748120	LOF	0.835	0.023	0.142
KCNA1	296	L	F	NM_000217.3(KCNA1):c.888G>T (p.Leu296Phe)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Dec 5, 2020)	criteria provided, single submitter	VCV001029721	12	4912266	1029721	1017632	rs1315748120	LOF	0.835	0.023	0.142
KCNA1	296	L	F	NM_000217.3(KCNA1):c.888G>C (p.Leu296Phe)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Nov 13, 2019)	criteria provided, single submitter	VCV000966172	12	4912266	966172	956895	rs1315748120	LOF	0.835	0.023	0.142
KCNA1	296	L	F	NM_000217.3(KCNA1):c.888G>C (p.Leu296Phe)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Nov 13, 2019)	criteria provided, single submitter	VCV000966172	12	4912266	966172	956895	rs1315748120	LOF	0.835	0.023	0.142
KCNA1	298	R	T	NM_000217.3(KCNA1):c.893G>C (p.Arg298Thr)	not provided	Uncertain significance(Last reviewed: Jun 1, 2021)	criteria provided, single submitter	VCV001176598	12	4912271	1176598	1166054	NA	LOF	0.914	0.021	0.066
KCNA1	305	L	F	NM_000217.3(KCNA1):c.913C>T (p.Leu305Phe)	not provided|Episodic ataxia type 1	Likely pathogenic(Last reviewed: Oct 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000447614	12	4912291	447614	441556	rs1555085761	LOF	0.869	0.035	0.096
KCNA1	306	S	C	NM_000217.3(KCNA1):c.917C>G (p.Ser306Cys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jul 27, 2020)	criteria provided, single submitter	VCV001009332	12	4912295	1009332	995246	rs1947357313	LOF	0.891	0.046	0.062
KCNA1	311	G	C	NM_000217.3(KCNA1):c.931G>T (p.Gly311Cys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Aug 28, 2019)	criteria provided, single submitter	VCV000957833	12	4912309	957833	948057	rs1947357459	Neutral	0.728	0.185	0.086
KCNA1	312	L	I	NM_000217.3(KCNA1):c.934C>A (p.Leu312Ile)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Nov 18, 2019)	criteria provided, single submitter	VCV000834847	12	4912312	834847	839997	rs1947357501	Neutral	0.656	0.226	0.117
KCNA1	314	I	T	NM_000217.3(KCNA1):c.941T>C (p.Ile314Thr)	not provided|Episodic ataxia type 1	Likely	criteria	VCV000427198	12	4912319	427198	415337	rs1085308020	LOF	0.795	0.118	0.087

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						pathogenic(Last reviewed: Jul 26, 2018)	provided, multiple submitters, no conflicts										
KCNA1	322	S	R	NM_000217.3(KCNA1):c.964A>C (p.Ser322Arg)	Episodic ataxia type 1	Uncertain significance(Last reviewed: May 3, 2018)	criteria provided, single submitter	VCV000577805	12	4912342	577805	571880	rs1565433425	LOF	0.782	0.125	0.094
KCNA1	325	E	D	NM_000217.3(KCNA1):c.975G>C (p.Glu325Asp)	Episodic ataxia type 1	Pathogenic(Last reviewed: Dec 1, 1995)	no assertion criteria provided	VCV000013485	12	4912353	13485	28524	rs104894353	LOF	0.897	0.034	0.069
KCNA1	331	F	C	NM_000217.3(KCNA1):c.992T>G (p.Phe331Cys)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jan 29, 2020)	criteria provided, single submitter	VCV001042632	12	4912370	1042632	1030985	rs1947357713	LOF	0.91	0.015	0.075
KCNA1	332	F	S	NM_000217.3(KCNA1):c.995T>C (p.Phe332Ser)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Oct 21, 2020)	criteria provided, single submitter	VCV001052052	12	4912373	1052052	1047989	NA	LOF	0.917	0.013	0.07
KCNA1	352	A	P	NM_000217.3(KCNA1):c.1054G>C (p.Ala352Pro)	not provided	Uncertain significance(Last reviewed: Jul 1, 2016)	criteria provided, single submitter	VCV000374743	12	4912432	374743	361629	rs1057519226	LOF	0.733	0.139	0.128
KCNA1	365	W	R	NM_000217.3(KCNA1):c.1093T>C (p.Trp365Arg)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Apr 29, 2020)	criteria provided, single submitter	VCV001060322	12	4912471	1060322	1047990	NA	LOF	0.943	0.011	0.046
KCNA1	376	G	R	NM_000217.3(KCNA1):c.1126G>C (p.Gly376Arg)	not provided|Episodic ataxia type 1	Uncertain significance(Last reviewed: Oct 31, 2017)	criteria provided, single submitter	VCV000453118	12	4912504	453118	445033	rs1555085786	LOF	0.897	0.038	0.065
KCNA1	384	G	R	NM_000217.3(KCNA1):c.1150G>A (p.Gly384Arg)	Episodic ataxia type 1	Uncertain significance(Last reviewed: May 15, 2019)	criteria provided, single submitter	VCV000863534	12	4912528	863534	839998	rs1947358468	LOF	0.899	0.03	0.071
KCNA1	395	A	S	NM_000217.3(KCNA1):c.1183G>T (p.Ala395Ser)	Episodic ataxia type 1	Likely pathogenic(Last reviewed: Mar 14, 2017)	criteria provided, single submitter	VCV000431378	12	4912561	431378	424900	rs1135401950	LOF	0.882	0.02	0.097
KCNA1	396	G	R	NM_000217.3(KCNA1):c.1186G>C (p.Gly396Arg)	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy	Likely pathogenic(Last reviewed: May 19, 2021)	criteria provided, single submitter	VCV001098715	12	4912564	1098715	1087424	NA	LOF	0.933	0.014	0.053
KCNA1	396	G	V	NM_000217.3(KCNA1):c.1187G>T (p.Gly396Val)	Episodic kinesigenic dyskinesia	Likely pathogenic(Last reviewed: May 19, 2021)	criteria provided, single submitter	VCV001098714	12	4912565	1098714	1087425	NA	LOF	0.911	0.024	0.065
KCNA1	401	A	V	NM_000217.3(KCNA1):c.1202C>T (p.Ala401Val)	Inborn genetic diseases|not provided	Pathogenic/Likely pathogenic(Last reviewed: Aug 16, 2019)	criteria provided, multiple submitters, no conflicts	VCV000208748	12	4912580	208748	205276	rs797044929	LOF	0.801	0.027	0.172
KCNA1	403	P	T	NM_000217.3(KCNA1):c.1207C>A (p.Pro403Thr)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Aug 1, 2018)	criteria provided, single submitter	VCV000644532	12	4912585	644532	641184	rs1591627924	LOF	0.908	0.027	0.065
KCNA1	404	V	I	NM_000217.3(KCNA1):c.1210G>A (p.Val404Ile)	Episodic ataxia type 1	Pathogenic(Last reviewed: Oct 1, 2000)	no assertion criteria provided	VCV000013487	12	4912588	13487	28526	rs104894355	LOF	0.853	0.046	0.101
KCNA1	405	P	A	NM_000217.3(KCNA1):c.1213C>G (p.Pro405Ala)	Inborn genetic diseases	Likely pathogenic(Last reviewed: Jan 30,	criteria provided, single submitter	VCV000985808	12	4912591	985808	973859	rs1947358808	LOF	0.901	0.032	0.066

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)											
KCNA1	405	P	L	NM_000217.3(KCNA1):c.1214C>T (p.Pro405Leu)	not provided	Pathogenic(Last reviewed: Dec 22, 2016)	criteria provided, single submitter	VCV000447609	12	4912592	447609	441557	rs1555085798	LOF	0.896	0.036	0.068
KCNA1	408	V	A	NM_000217.3(KCNA1):c.1223T>C (p.Val408Ala)	Episodic ataxia type 1	Pathogenic(Last reviewed: Jun 25, 2015)	no assertion criteria provided	VCV000013480	12	4912601	13480	28519	rs104894352	LOF	0.859	0.02	0.121
KCNA1	408	V	M	NM_000217.3(KCNA1):c.1222G>A (p.Val408Met)	not provided	Uncertain significance(Last reviewed: Jul 24, 2015)	criteria provided, single submitter	VCV000282028	12	4912600	282028	266265	rs113994117	LOF	0.844	0.023	0.133
KCNA1	429	H	Q	NM_000217.3(KCNA1):c.1287C>G (p.His429Gln)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Jun 17, 2020)	criteria provided, single submitter	VCV001059224	12	4912665	1059224	1047991	NA	Neutral	0.653	0.207	0.14
KCNA1	430	V	L	NM_000217.3(KCNA1):c.1288G>C (p.Val430Leu)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Mar 26, 2020)	criteria provided, single submitter	VCV001058791	12	4912666	1058791	1047992	NA	Neutral	0.558	0.312	0.13
KCNA1	442	S	T	NM_000217.3(KCNA1):c.1325G>C (p.Ser442Thr)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Sep 8, 2019)	criteria provided, single submitter	VCV000934436	12	4912703	934436	936147	rs1947359738	Neutral	0.57	0.27	0.159
KCNA1	443	R	G	NM_000217.3(KCNA1):c.1327C>G (p.Arg443Gly)	not specified|Episodic ataxia type 1|not provided	Benign/Likely benign(Last reviewed: Oct 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000195042	12	4912705	195042	192204	rs150849316	Neutral	0.615	0.261	0.124
KCNA1	443	R	H	NM_000217.3(KCNA1):c.1328G>A (p.Arg443His)	Episodic ataxia type 1	Uncertain significance(Last reviewed: May 21, 2020)	criteria provided, single submitter	VCV001056962	12	4912706	1056962	1047993	NA	Neutral	0.557	0.306	0.138
KCNA1	449	M	T	NM_000217.3(KCNA1):c.1346T>C (p.Met449Thr)	not provided	Uncertain significance(Last reviewed: May 1, 2018)	criteria provided, single submitter	VCV000586089	12	4912724	586089	577276	rs1481224137	Neutral	0.637	0.236	0.127
KCNA1	455	M	I	NM_000217.3(KCNA1):c.1365G>T (p.Met455Ile)	not provided	Uncertain significance(Last reviewed: Jan 24, 2020)	criteria provided, single submitter	VCV001195360	12	4912743	1195360	1184716	NA	Neutral	0.566	0.325	0.109
KCNA1	455	M	K	NM_000217.3(KCNA1):c.1364T>A (p.Met455Lys)	Episodic ataxia|Myokymia	Uncertain significance(Last reviewed: Jun 14, 2016)	criteria provided, single submitter	VCV000309147	12	4912742	309147	331464	rs886049512	Neutral	0.721	0.172	0.107
KCNA1	455	M	V	NM_000217.3(KCNA1):c.1363A>G (p.Met455Val)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Aug 15, 2020)	criteria provided, single submitter	VCV001021989	12	4912741	1021989	1010476	rs1173585397	Neutral	0.579	0.312	0.109
KCNA1	460	D	Y	NM_000217.3(KCNA1):c.1378G>T (p.Asp460Tyr)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Feb 14, 2020)	criteria provided, single submitter	VCV001055177	12	4912756	1055177	1047994	NA	Neutral	0.599	0.281	0.119
KCNA1	461	M	T	NM_000217.3(KCNA1):c.1382T>C (p.Met461Thr)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Apr 8, 2020)	criteria provided, single submitter	VCV001039751	12	4912760	1039751	1030986	rs767068909	Neutral	0.688	0.214	0.097
KCNA1	466	A	D	NM_000217.3(KCNA1):c.1397C>A (p.Ala466Asp)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Dec 18, 2018)	criteria provided, single submitter	VCV000642349	12	4912775	642349	641185	rs1303950325	LOF	0.733	0.163	0.104

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA1	472	N	S	NM_000217.3(KCNA1):c.1415A>G (p.Asn472Ser)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Dec 11, 2019)	criteria provided, single submitter	VCV000849888	12	4912793	849888	839999	rs150612442	Neutral	0.595	0.25	0.155
KCNA1	481	A	V	NM_000217.3(KCNA1):c.1442C>T (p.Ala481Val)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV001052029	12	4912820	1052029	1047995	NA	Neutral	0.555	0.308	0.136
KCNA1	483	Q	H	NM_000217.3(KCNA1):c.1449A>T (p.Gln483His)	not provided|Episodic ataxia type 1	Uncertain significance(Last reviewed: Aug 11, 2019)	criteria provided, multiple submitters, no conflicts	VCV000586090	12	4912827	586090	577277	rs372539672	Neutral	0.569	0.289	0.142
KCNA1	485	C	S	NM_000217.3(KCNA1):c.1453T>A (p.Cys485Ser)	Episodic ataxia type 1	Uncertain significance(Last reviewed: May 25, 2019)	criteria provided, single submitter	VCV000949579	12	4912831	949579	926656	rs1470787858	Neutral	0.681	0.2	0.119
KCNA1	486	V	I	NM_000217.3(KCNA1):c.1456G>A (p.Val486Ile)	Episodic ataxia type 1	Uncertain significance(Last reviewed: Oct 6, 2020)	criteria provided, single submitter	VCV001061129	12	4912834	1061129	1047996	NA	Neutral	0.58	0.299	0.121
KCNA1	489	S	N	NM_000217.3(KCNA1):c.1466G>A (p.Ser489Asn)	not provided	Uncertain significance(Last reviewed: May 5, 2017)	criteria provided, single submitter	VCV000430064	12	4912844	430064	421928	rs1131691765	Neutral	0.653	0.207	0.14
KCNA2	8	P	L	NM_004974.4(KCNA2):c.23C>T (p.Pro8Leu)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Feb 2, 2020)	criteria provided, single submitter	VCV001008882	1	110604760	1008882	986803	rs1487522107	Neutral	0.588	0.237	0.175
KCNA2	12	A	V	NM_004974.4(KCNA2):c.35C>T (p.Ala12Val)	not provided|Epileptic encephalopathy, early infantile, 32	Conflicting interpretations of pathogenicity(Last reviewed: May 25, 2021)	criteria provided, conflicting interpretations	VCV000663661	1	110604748	663661	626578	rs372822052	Neutral	0.585	0.23	0.185
KCNA2	15	L	V	NM_004974.4(KCNA2):c.43C>G (p.Leu15Val)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jul 25, 2020)	criteria provided, single submitter	VCV001020683	1	110604740	1020683	1002022	rs1649522982	Neutral	0.66	0.203	0.138
KCNA2	16	P	R	NM_004974.4(KCNA2):c.47C>G (p.Pro16Arg)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: May 4, 2019)	criteria provided, single submitter	VCV000952381	1	110604736	952381	921567	rs1649522718	Neutral	0.55	0.257	0.193
KCNA2	16	P	S	NM_004974.4(KCNA2):c.46C>T (p.Pro16Ser)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 30, 2018)	criteria provided, single submitter	VCV000663887	1	110604737	663887	626577	rs1570754155	Neutral	0.549	0.26	0.192
KCNA2	23	Y	C	NM_004974.4(KCNA2):c.68A>G (p.Tyr23Cys)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 25, 2018)	criteria provided, single submitter	VCV000476053	1	110604715	476053	447049	rs753829876	Neutral	0.663	0.203	0.134
KCNA2	27	A	T	NM_004974.4(KCNA2):c.79G>A (p.Ala27Thr)	Epileptic encephalopathy, early infantile, 32	Likely benign(Last reviewed: Nov 6, 2019)	criteria provided, single submitter	VCV001087753	1	110604704	1087753	1066022	NA	Neutral	0.543	0.235	0.221
KCNA2	43	R	W	NM_004974.4(KCNA2):c.127C>T (p.Arg43Trp)	not provided	Uncertain significance(Last reviewed: Feb 6, 2020)	criteria provided, single submitter	VCV001304041	1	110604656	1304041	1294316	NA	LOF	0.818	0.094	0.088
KCNA2	57	T	I	NM_004974.4(KCNA2):c.170C>T (p.Thr57Ile)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Sep 27, 2019)	criteria provided, single submitter	VCV000962503	1	110604613	962503	941369	rs1649517174	GOF	0.606	0.1	0.294

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA2	65	R	Q	NM_004974.4(KCNA2):c.194G>A (p.Arg65Gln)	Epileptic encephalopathy, early infantile, 32|not provided	Uncertain significance(Last reviewed: Oct 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV001006374	1	110604589	1006374	986802	rs1649516324	LOF	0.731	0.154	0.114
KCNA2	73	R	Q	NM_004974.4(KCNA2):c.218G>A (p.Arg73Gln)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 25, 2019)	criteria provided, single submitter	VCV000581262	1	110604565	581262	556838	rs373042266	GOF	0.52	0.093	0.388
KCNA2	79	D	N	NM_004974.4(KCNA2):c.235G>A (p.Asp79Asn)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 28, 2019)	criteria provided, single submitter	VCV000951627	1	110604548	951627	921566	rs747844549	GOF	0.573	0.079	0.348
KCNA2	79	D	Y	NM_004974.4(KCNA2):c.235G>T (p.Asp79Tyr)	Epileptic encephalopathy, early infantile, 32|not provided	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV001036616	1	110604548	1036616	1022530	rs747844549	GOF	0.578	0.077	0.344
KCNA2	80	R	Q	NM_004974.4(KCNA2):c.239G>A (p.Arg80Gln)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Dec 27, 2019)	criteria provided, single submitter	VCV000852778	1	110604544	852778	822501	rs1649513909	GOF	0.547	0.085	0.368
KCNA2	80	R	W	NM_004974.4(KCNA2):c.238C>T (p.Arg80Trp)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Mar 26, 2018)	criteria provided, single submitter	VCV000581223	1	110604545	581223	556836	rs1448937059	GOF	0.628	0.096	0.276
KCNA2	82	R	C	NM_004974.4(KCNA2):c.244C>T (p.Arg82Cys)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 30, 2017)	criteria provided, single submitter	VCV000542665	1	110604539	542665	515007	rs1433727837	GOF	0.672	0.065	0.262
KCNA2	97	R	Q	NM_004974.4(KCNA2):c.290G>A (p.Arg97Gln)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jul 13, 2017)	criteria provided, single submitter	VCV000976219	1	110604493	976219	964122	rs1194485302	GOF	0.495	0.142	0.363
KCNA2	100	R	Q	NM_004974.4(KCNA2):c.299G>A (p.Arg100Gln)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV001025134	1	110604484	1025134	1002021	rs1649509246	GOF	0.537	0.106	0.357
KCNA2	117	E	D	NM_004974.4(KCNA2):c.351G>T (p.Glu117Asp)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Dec 13, 2019)	criteria provided, single submitter	VCV000854988	1	110604432	854988	822500	rs1649507257	GOF	0.515	0.115	0.371
KCNA2	122	A	T	NM_004974.4(KCNA2):c.364G>A (p.Ala122Thr)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jan 11, 2020)	criteria provided, single submitter	VCV000998665	1	110604419	998665	986800	rs1366378482	GOF	0.614	0.055	0.331
KCNA2	141	P	H	NM_004974.4(KCNA2):c.422C>A (p.Pro141His)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jul 1, 2020)	criteria provided, single submitter	VCV001010084	1	110604361	1010084	986799	rs1649502580	GOF	0.553	0.112	0.335
KCNA2	150	W	R	NM_004974.4(KCNA2):c.448T>C (p.Trp150Arg)	Epileptic encephalopathy, early infantile, 32|not provided	Uncertain significance(Last reviewed: Mar 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV001016452	1	110604335	1016452	1002020	rs1649500367	GOF	0.617	0.053	0.33
KCNA2	154	E	Q	NM_004974.4(KCNA2):c.460G>C (p.Glu154Gln)	not provided	Uncertain significance(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV001306994	1	110604323	1306994	1295943	NA	GOF	0.573	0.042	0.385
KCNA2	156	P	S	NM_004974.4(KCNA2):c.466C>T (p.Pro156Ser)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 3, 2020)	criteria provided, single submitter	VCV000862623	1	110604317	862623	822499	rs1649498927	GOF	0.616	0.065	0.319

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA2	178	V	I	NM_004974.4(KCNA2):c.532G>A (p.Val178Ile)	not provided	Uncertain significance(Last reviewed: Jul 3, 2019)	criteria provided, single submitter	VCV001306817	1	110604251	1306817	1295767	NA	GOF	0.64	0.067	0.292
KCNA2	179	S	N	NM_004974.4(KCNA2):c.536G>A (p.Ser179Asn)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 23, 2020)	criteria provided, single submitter	VCV001059155	1	110604247	1059155	1039347	NA	GOF	0.501	0.041	0.458
KCNA2	182	L	M	NM_004974.4(KCNA2):c.544C>A (p.Leu182Met)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV001005091	1	110604239	1005091	986798	rs367662144	GOF	0.655	0.062	0.284
KCNA2	183	E	K	NM_004974.4(KCNA2):c.547G>A (p.Glu183Lys)	Epileptic encephalopathy, early infantile, 32|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Nov 1, 2019)	criteria provided, conflicting interpretations	VCV000542661	1	110604236	542661	514988	rs1553181398	GOF	0.672	0.044	0.284
KCNA2	189	R	W	NM_004974.4(KCNA2):c.565C>T (p.Arg189Trp)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 24, 2018)	criteria provided, single submitter	VCV000569307	1	110604218	569307	556976	rs770611476	LOF	0.82	0.096	0.084
KCNA2	197	G	D	NM_004974.4(KCNA2):c.590G>A (p.Gly197Asp)	Seizures	Uncertain significance(Last reviewed: Apr 4, 2017)	criteria provided, single submitter	VCV000588883	1	110604193	588883	578709	rs752985457	Neutral	0.592	0.247	0.161
KCNA2	198	S	G	NM_004974.4(KCNA2):c.592A>G (p.Ser198Gly)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Feb 20, 2020)	criteria provided, single submitter	VCV001019596	1	110604191	1019596	1002019	rs1649489553	Neutral	0.586	0.256	0.158
KCNA2	204	T	I	NM_004974.4(KCNA2):c.611C>T (p.Thr204Ile)	Epileptic encephalopathy, early infantile, 32	Likely benign(Last reviewed: Jun 7, 2020)	criteria provided, single submitter	VCV000948207	1	110604172	948207	921565	rs1421276694	Neutral	0.615	0.216	0.169
KCNA2	209	T	A	NM_004974.4(KCNA2):c.625A>G (p.Thr209Ala)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jul 18, 2019)	criteria provided, single submitter	VCV000956614	1	110604158	956614	941368	rs1649486218	Neutral	0.578	0.242	0.18
KCNA2	209	T	S	NM_004974.4(KCNA2):c.625A>T (p.Thr209Ser)	not provided	Uncertain significance(Last reviewed: Jan 7, 2020)	criteria provided, single submitter	VCV001311940	1	110604158	1311940	1302471	NA	Neutral	0.556	0.264	0.18
KCNA2	211	G	R	NM_004974.4(KCNA2):c.631G>A (p.Gly211Arg)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jul 17, 2018)	criteria provided, single submitter	VCV000582691	1	110604152	582691	556585	rs149727427	Neutral	0.605	0.227	0.168
KCNA2	232	W	R	NM_004974.4(KCNA2):c.694T>A (p.Trp232Arg)	Inborn genetic diseases	Uncertain significance(Last reviewed: May 1, 2017)	criteria provided, single submitter	VCV000521733	1	110604089	521733	511150	rs1553181370	GOF	0.691	0.066	0.243
KCNA2	232	W	S	NM_004974.4(KCNA2):c.695G>C (p.Trp232Ser)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Feb 26, 2019)	criteria provided, single submitter	VCV000639637	1	110604088	639637	626574	rs1570753354	LOF	0.725	0.064	0.211
KCNA2	234	S	P	NM_004974.4(KCNA2):c.700T>C (p.Ser234Pro)	not provided	Uncertain significance(Last reviewed: Jul 14, 2020)	criteria provided, single submitter	VCV001313077	1	110604083	1313077	1303338	NA	GOF	0.611	0.054	0.335
KCNA2	246	S	I	NM_004974.4(KCNA2):c.737G>T (p.Ser246Ile)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 19, 2019)	criteria provided, single submitter	VCV000971175	1	110604046	971175	952010	rs1649481724	GOF	0.568	0.07	0.361
KCNA2	247	K	E	NM_004974.4(KCNA2):c.739A>G (p.Lys247Glu)	not provided	Uncertain significance(Last	criteria provided, single	VCV001303863	1	110604044	1303863	1294138	NA	GOF	0.588	0.059	0.353

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Sep 4, 2020)	submitter										
KCNA2	249	G	S	NM_004974.4(KCNA2):c.745G>A (p.Gly249Ser)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 8, 2019)	criteria provided, single submitter	VCV000850411	1	110604038	850411	822497	rs746926057	GOF	0.557	0.07	0.373
KCNA2	252	T	A	NM_004974.4(KCNA2):c.754A>G (p.Thr252Ala)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jul 10, 2020)	criteria provided, single submitter	VCV000948350	1	110604029	948350	921564	rs772053786	GOF	0.5	0.051	0.449
KCNA2	254	I	N	NM_004974.4(KCNA2):c.761T>A (p.Ile254Asn)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Dec 24, 2019)	criteria provided, single submitter	VCV000849504	1	110604022	849504	822496	rs1649480102	GOF	0.556	0.049	0.395
KCNA2	256	N	I	NM_004974.4(KCNA2):c.767A>T (p.Asn256Ile)	not provided	Uncertain significance(Last reviewed: Aug 27, 2019)	criteria provided, single submitter	VCV001307977	1	110604016	1307977	1297924	NA	GOF	0.697	0.036	0.267
KCNA2	259	D	A	NM_004974.4(KCNA2):c.776A>C (p.Asp259Ala)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 6, 2020)	criteria provided, single submitter	VCV001010948	1	110604007	1010948	986797	rs1649479585	GOF	0.662	0.037	0.301
KCNA2	261	V	A	NM_004974.4(KCNA2):c.782T>C (p.Val261Ala)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 2, 2019)	criteria provided, single submitter	VCV000845684	1	110604001	845684	822495	rs1649479037	GOF	0.669	0.058	0.273
KCNA2	263	I	T	NM_004974.4(KCNA2):c.788T>C (p.Ile263Thr)	Epileptic encephalopathy, early infantile, 32	Likely pathogenic(Last reviewed: Dec 19, 2019)	criteria provided, single submitter	VCV000190326	1	110603995	190326	188153	rs786205231	GOF	0.674	0.049	0.278
KCNA2	280	D	N	NM_004974.4(KCNA2):c.838G>A (p.Asp280Asn)	not provided	Uncertain significance(Last reviewed: Jul 17, 2019)	criteria provided, single submitter	VCV001306971	1	110603945	1306971	1295920	NA	Neutral	0.499	0.333	0.168
KCNA2	281	A	T	NM_004974.4(KCNA2):c.841G>A (p.Ala281Thr)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: May 3, 2018)	criteria provided, single submitter	VCV000581469	1	110603942	581469	556583	rs753926939	Neutral	0.56	0.258	0.181
KCNA2	284	G	D	NM_004974.4(KCNA2):c.851G>A (p.Gly284Asp)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jul 2, 2018)	criteria provided, single submitter	VCV000578279	1	110603932	578279	556974	rs1246925002	LOF	0.631	0.147	0.222
KCNA2	284	G	R	NM_004974.4(KCNA2):c.850G>C (p.Gly284Arg)	not provided	Uncertain significance(Last reviewed: Jan 3, 2022)	criteria provided, single submitter	VCV001326483	1	110603933	1326483	1316843	NA	Neutral	0.596	0.163	0.242
KCNA2	288	M	V	NM_004974.4(KCNA2):c.862A>G (p.Met288Val)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV001026363	1	110603921	1026363	1002018	rs1486839913	GOF	0.646	0.064	0.29
KCNA2	290	L	P	NM_004974.4(KCNA2):c.869T>C (p.Leu290Pro)	not provided	Likely pathogenic(Last reviewed: Apr 6, 2017)	criteria provided, single submitter	VCV000426735	1	110603914	426735	414722	rs1085307768	GOF	0.508	0.046	0.446
KCNA2	290	L	R	NM_004974.4(KCNA2):c.869T>G (p.Leu290Arg)	Inborn genetic diseases	Likely pathogenic(Last reviewed: Jul 16, 2018)	criteria provided, single submitter	VCV000985630	1	110603914	985630	973123	rs1085307768	GOF	0.428	0.048	0.525
KCNA2	294	R	H	NM_004974.4(KCNA2):c.881G>A (p.Arg294His)	Inborn genetic diseases|not provided|Epileptic encephalopathy, early infantile, 32	Pathogenic/Likely pathogenic(Last reviewed: Jul 8, 2021)	criteria provided, multiple submitters, no	VCV000280584	1	110603902	280584	263933	rs886041761	GOF	0.492	0.046	0.462

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNA2	297	R	Q	NM_004974.4(KCNA2):c.890G>A (p.Arg297Gln)	Inborn genetic diseases|not provided|Epileptic encephalopathy, early infantile, 32|Epileptic encephalopathy, early infantile, 1| Neurodevelopmental disorder	Pathogenic(Last reviewed: Oct 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000190328	1	110603893	190328	188155	rs786205232	GOF	0.476	0.047	0.478
KCNA2	297	R	W	NM_004974.4(KCNA2):c.889C>T (p.Arg297Trp)	Epileptic encephalopathy, early infantile, 32|not provided	Pathogenic(Last reviewed: Oct 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000986989	1	110603894	986989	974944	rs1649473972	GOF	0.604	0.043	0.353
KCNA2	298	L	F	NM_004974.4(KCNA2):c.894G>T (p.Leu298Phe)	Epileptic encephalopathy, early infantile, 32|not provided	Pathogenic(Last reviewed: Mar 1, 2017)	criteria provided, single submitter	VCV000190327	1	110603889	190327	188154	rs876657390	GOF	0.455	0.051	0.494
KCNA2	299	V	L	NM_004974.4(KCNA2):c.895G>C (p.Val299Leu)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: May 26, 2017)	criteria provided, single submitter	VCV000476054	1	110603888	476054	447041	rs1553181334	GOF	0.484	0.051	0.465
KCNA2	300	R	S	NM_004974.4(KCNA2):c.900A>T (p.Arg300Ser)	Epileptic encephalopathy, early infantile, 32	Likely pathogenic(Last reviewed: Jul 23, 2020)	criteria provided, single submitter	VCV001066046	1	110603883	1066046	1054728	NA	GOF	0.507	0.045	0.448
KCNA2	302	F	L	NM_004974.4(KCNA2):c.906T>G (p.Phe302Leu)	Epileptic encephalopathy, early infantile, 1	Likely pathogenic(Last reviewed: Jan 1, 2020)	no assertion criteria provided	VCV001174095	1	110603877	1174095	1163172	NA	GOF	0.536	0.057	0.407
KCNA2	307	L	W	NM_004974.4(KCNA2):c.920T>G (p.Leu307Trp)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Feb 27, 2020)	criteria provided, single submitter	VCV001004548	1	110603863	1004548	986796	rs1649471614	GOF	0.543	0.048	0.41
KCNA2	309	R	G	NM_004974.4(KCNA2):c.925A>G (p.Arg309Gly)	not provided	Uncertain significance(Last reviewed: Oct 26, 2020)	criteria provided, single submitter	VCV001313559	1	110603858	1313559	1303820	NA	GOF	0.685	0.055	0.26
KCNA2	310	H	R	NM_004974.4(KCNA2):c.929A>G (p.His310Arg)	Epileptic encephalopathy, early infantile, 32	Likely pathogenic(Last reviewed: Nov 9, 2018)	no assertion criteria provided	VCV000975835	1	110603854	975835	964109	rs1649470988	GOF	0.494	0.065	0.441
KCNA2	320	T	I	NM_004974.4(KCNA2):c.959C>T (p.Thr320Ile)	Epileptic encephalopathy, early infantile, 32	Likely pathogenic(Last reviewed: Jan 1, 2018)	criteria provided, single submitter	VCV000495271	1	110603824	495271	486730	rs1553181323	GOF	0.652	0.047	0.3
KCNA2	321	L	F	NM_004974.4(KCNA2):c.961C>T (p.Leu321Phe)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 25, 2018)	criteria provided, single submitter	VCV000583343	1	110603822	583343	556554	rs1557732226	GOF	0.696	0.06	0.245
KCNA2	329	A	V	NM_004974.4(KCNA2):c.*1204C>T	not provided	Benign(Last reviewed: Nov 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000586061	1	110602079	586061	576390	rs115258190	GOF	0.58	0.042	0.378
KCNA2	330	L	H	NM_004974.4(KCNA2):c.989T>A (p.Leu330His)	not provided	Uncertain significance(Last reviewed: Aug 20, 2019)	criteria provided, single submitter	VCV001307978	1	110603794	1307978	1297925	NA	GOF	0.528	0.04	0.432
KCNA2	330	L	R	NM_004974.4(KCNA2):c.989T>G (p.Leu330Arg)	not provided	Likely pathogenic(Last reviewed: Nov 18, 2016)	criteria provided, single submitter	VCV000390269	1	110603794	390269	364357	rs1057523703	GOF	0.544	0.045	0.411
KCNA2	331	M	V	NM_004974.4(KCNA2):c.*1209A>G	not provided	Uncertain significance(Last	criteria provided, single	VCV001298434	1	110602074	1298434	1288296	NA	GOF	0.569	0.052	0.379

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Jul 1, 2021)	submitter										
KCNA2	333	F	L	NM_004974.4(KCNA2):c.997T>C (p.Phe333Leu)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 23, 2018)	criteria provided, single submitter	VCV000575248	1	110603786	575248	556552	rs1557732183	GOF	0.618	0.057	0.325
KCNA2	338	G	E	NM_004974.4(KCNA2):c.1013G>A (p.Gly338Glu)	Epileptic encephalopathy, early infantile, 32|not provided	Likely pathogenic(Last reviewed: Jun 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000542660	1	110603770	542660	515037	rs1553181301	GOF	0.64	0.038	0.322
KCNA2	341	L	P	NM_004974.4(KCNA2):c.1022T>C (p.Leu341Pro)	not provided	Uncertain significance(Last reviewed: Jul 1, 2020)	criteria provided, single submitter	VCV001306680	1	110603761	1306680	1296623	NA	GOF	0.641	0.05	0.309
KCNA2	343	D	N	NM_004974.4(KCNA2):c.*1245G>A	not provided	Likely benign(Last reviewed: Jan 3, 2020)	criteria provided, single submitter	VCV000392819	1	110602038	392819	364351	rs756992381	GOF	0.576	0.037	0.387
KCNA2	343	S	F	NM_004974.4(KCNA2):c.1028C>T (p.Ser343Phe)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Feb 11, 2018)	criteria provided, single submitter	VCV000578773	1	110603755	578773	556834	rs1557732150	GOF	0.652	0.03	0.318
KCNA2	354	R	Q	NM_004974.4(KCNA2):c.1061G>A (p.Arg354Gln)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV000855444	1	110603722	855444	822494	rs374827915	Neutral	0.457	0.379	0.164
KCNA2	355	E	D	NM_004974.4(KCNA2):c.1065G>T (p.Glu355Asp)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV001038969	1	110603718	1038969	1022528	rs1308544471	Neutral	0.497	0.319	0.184
KCNA2	356	S	C	NM_004974.4(KCNA2):c.1067C>G (p.Ser356Cys)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Nov 7, 2019)	criteria provided, single submitter	VCV000939630	1	110603716	939630	929953	rs1317841788	GOF	0.656	0.116	0.229
KCNA2	362	P	A	NM_004974.4(KCNA2):c.1084C>G (p.Pro362Ala)	not provided	Likely pathogenic(Last reviewed: Aug 15, 2016)	criteria provided, single submitter	VCV000430451	1	110603699	430451	421156	rs1131691974	GOF	0.691	0.034	0.275
KCNA2	370	V	I	NM_004974.4(KCNA2):c.1108G>A (p.Val370Ile)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV000934618	1	110603675	934618	929952	rs756214647	GOF	0.622	0.05	0.328
KCNA2	373	T	I	NM_004974.4(KCNA2):c.1118C>T (p.Thr373Ile)	not provided|Epileptic encephalopathy, early infantile, 32	Conflicting interpretations of pathogenicity(Last reviewed: Dec 30, 2019)	criteria provided, conflicting interpretations	VCV000452594	1	110603665	452594	442578	rs1553181282	LOF	0.719	0.047	0.234
KCNA2	374	T	A	NM_004974.4(KCNA2):c.1120A>G (p.Thr374Ala)	Epileptic encephalopathy, early infantile, 32	Pathogenic(Last reviewed: May 28, 2019)	criteria provided, multiple submitters, no conflicts	VCV000559647	1	110603663	559647	550344	rs1553181280	LOF	0.769	0.066	0.165
KCNA2	376	G	A	NM_004974.4(KCNA2):c.1127G>C (p.Gly376Ala)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jun 1, 2018)	criteria provided, single submitter	VCV000577200	1	110603656	577200	556550	rs1557732017	LOF	0.825	0.061	0.114
KCNA2	379	D	H	NM_004974.4(KCNA2):c.1135G>C (p.Asp379His)	not provided	Likely pathogenic(Last reviewed: Nov 18, 2019)	criteria provided, single submitter	VCV001196855	1	110603648	1196855	1186033	NA	LOF	0.775	0.129	0.096
KCNA2	382	P	L	NM_004974.4(KCNA2):c.1145C>T (p.Pro382Leu)	not provided|Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 5,	criteria provided, multiple	VCV000579198	1	110603638	579198	556581	rs777008812	LOF	0.743	0.062	0.195

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2021)	submitters, no conflicts										
KCNA2	385	I	T	NM_004974.4(KCNA2):c.1154T>C (p.Ile385Thr)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Nov 7, 2017)	criteria provided, single submitter	VCV000542662	1	110603629	542662	515024	rs1197586006	GOF	0.607	0.071	0.322
KCNA2	392	S	F	NM_004974.4(KCNA2):c.1175C>T (p.Ser392Phe)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 2, 2020)	criteria provided, single submitter	VCV001060438	1	110603608	1060438	1039345	NA	GOF	0.614	0.035	0.351
KCNA2	395	A	V	NM_004974.4(KCNA2):c.1184C>T (p.Ala395Val)	not provided	Uncertain significance(Last reviewed: Feb 1, 2017)	criteria provided, single submitter	VCV000806185	1	110603599	806185	794368	rs1570752776	GOF	0.654	0.045	0.302
KCNA2	399	V	G	NM_004974.4(KCNA2):c.1196T>G (p.Val399Gly)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jul 1, 2020)	criteria provided, single submitter	VCV001019183	1	110603587	1019183	1002017	rs1649457707	GOF	0.64	0.045	0.316
KCNA2	399	V	M	NM_004974.4(KCNA2):c.1195G>A (p.Val399Met)	KCN2A-Related Disorder|not provided|Epileptic encephalopathy, early infantile, 32	Likely pathogenic(Last reviewed: Aug 28, 2017)	criteria provided, single submitter	VCV000420845	1	110603588	420845	404879	rs1064794738	GOF	0.536	0.047	0.417
KCNA2	401	T	I	NM_004974.4(KCNA2):c.1202C>T (p.Thr401Ile)	not provided	Pathogenic(Last reviewed: Jun 25, 2021)	criteria provided, single submitter	VCV000430391	1	110603581	430391	421155	rs1131691939	GOF	0.604	0.036	0.36
KCNA2	402	I	T	NM_004974.4(KCNA2):c.1205T>C (p.Ile402Thr)	not provided	Uncertain significance(Last reviewed: Aug 7, 2019)	criteria provided, single submitter	VCV001307633	1	110603578	1307633	1297581	NA	GOF	0.544	0.044	0.412
KCNA2	403	A	V	NM_004974.4(KCNA2):c.1208C>T (p.Ala403Val)	Epileptic encephalopathy, early infantile, 32|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jan 1, 2020)	criteria provided, conflicting interpretations	VCV000871870	1	110603575	871870	858825	rs1649457143	GOF	0.578	0.042	0.38
KCNA2	404	L	I	NM_004974.4(KCNA2):c.1210T>A (p.Leu404Ile)	Epileptic encephalopathy, early infantile, 32	Likely pathogenic(Last reviewed: Feb 24, 2020)	criteria provided, single submitter	VCV001068304	1	110603573	1068304	1054727	NA	GOF	0.596	0.056	0.348
KCNA2	405	P	L	NM_004974.4(KCNA2):c.1214C>T (p.Pro405Leu)	Epileptic encephalopathy, early infantile, 32|not provided	Pathogenic/Likely pathogenic(Last reviewed: Nov 17, 2021)	criteria provided, multiple submitters, no conflicts	VCV000190325	1	110603569	190325	188152	NA	LOF	0.863	0.038	0.1
KCNA2	405	P	R	NM_004974.4(KCNA2):c.1214C>G (p.Pro405Arg)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Nov 4, 2019)	criteria provided, single submitter	VCV000966763	1	110603569	966763	952009	rs876657389	LOF	0.801	0.046	0.153
KCNA2	406	V	F	NM_004974.4(KCNA2):c.1216G>T (p.Val406Phe)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Apr 5, 2018)	criteria provided, single submitter	VCV000542666	1	110603567	542666	515018	rs1553181257	LOF	0.778	0.047	0.175
KCNA2	407	P	A	NM_004974.4(KCNA2):c.1219C>G (p.Pro407Ala)	Epileptic encephalopathy, early infantile, 32|not provided	Likely pathogenic(Last reviewed: Sep 18, 2019)	criteria provided, multiple submitters, no conflicts	VCV000623699	1	110603564	623699	612492	rs1557731896	LOF	0.834	0.038	0.129
KCNA2	407	P	R	NM_004974.4(KCNA2):c.1220C>G (p.Pro407Arg)	Epileptic encephalopathy, early infantile, 32	Likely pathogenic(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV001067683	1	110603563	1067683	1054726	NA	LOF	0.79	0.044	0.166
KCNA2	407	P	S	NM_004974.4(KCNA2):c.1219C>T (p.Pro407Ser)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last	criteria provided, single	VCV000999097	1	110603564	999097	986795	rs1557731896	LOF	0.81	0.038	0.152

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: May 14, 2020)	submitter										
KCNA2	408	V	A	NM_004974.4(KCNA2):c.1223T>C (p.Val408Ala)	Epileptic encephalopathy, early infantile, 32	Pathogenic(Last reviewed: Jan 1, 2014)	criteria provided, single submitter	VCV000666316	1	110603560	666316	653844	rs1570752696	GOF	0.574	0.048	0.378
KCNA2	408	V	G	NM_004974.4(KCNA2):c.1223T>G (p.Val408Gly)	Epileptic encephalopathy, early infantile, 32	Likely pathogenic(Last reviewed: Mar 28, 2020)	criteria provided, single submitter	VCV000853263	1	110603560	853263	822493	rs1570752696	GOF	0.656	0.046	0.298
KCNA2	414	N	K	NM_004974.4(KCNA2):c.1242C>A (p.Asn414Lys)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 30, 2020)	criteria provided, single submitter	VCV001002622	1	110603541	1002622	986794	rs749520613	GOF	0.624	0.047	0.329
KCNA2	414	N	K	NM_004974.4(KCNA2):c.1242C>A (p.Asn414Lys)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 30, 2020)	criteria provided, single submitter	VCV001002622	1	110603541	1002622	986794	rs749520613	GOF	0.624	0.047	0.329
KCNA2	414	N	K	NM_004974.4(KCNA2):c.1242C>G (p.Asn414Lys)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Aug 29, 2018)	criteria provided, single submitter	VCV000662361	1	110603541	662361	626572	rs749520613	GOF	0.624	0.047	0.329
KCNA2	414	N	K	NM_004974.4(KCNA2):c.1242C>G (p.Asn414Lys)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Aug 29, 2018)	criteria provided, single submitter	VCV000662361	1	110603541	662361	626572	rs749520613	GOF	0.624	0.047	0.329
KCNA2	416	F	L	NM_004974.4(KCNA2):c.1248C>A (p.Phe416Leu)	not provided|Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 12, 2020)	criteria provided, multiple submitters, no conflicts	VCV001061921	1	110603535	1061921	1039344	NA	GOF	0.697	0.059	0.245
KCNA2	417	Y	C	NM_004974.4(KCNA2):c.1250A>G (p.Tyr417Cys)	Epileptic encephalopathy, early infantile, 32|not provided	Uncertain significance(Last reviewed: Oct 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000623698	1	110603533	623698	612491	rs1557731858	LOF	0.737	0.059	0.204
KCNA2	419	R	Q	NM_004974.4(KCNA2):c.1256G>A (p.Arg419Gln)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jun 21, 2019)	criteria provided, single submitter	VCV000476047	1	110603527	476047	447086	rs1296710118	GOF	0.607	0.091	0.302
KCNA2	419	R	W	NM_004974.4(KCNA2):c.1255C>T (p.Arg419Trp)	not provided	Uncertain significance(Last reviewed: Dec 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV001254658	1	110603528	1254658	1244591	NA	GOF	0.642	0.1	0.259
KCNA2	435	C	S	NM_004974.4(KCNA2):c.1304G>C (p.Cys435Ser)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Oct 10, 2017)	criteria provided, single submitter	VCV000542664	1	110603479	542664	515005	rs1553181236	Neutral	0.629	0.178	0.193
KCNA2	435	C	Y	NM_004974.4(KCNA2):c.1304G>A (p.Cys435Tyr)	not provided	Uncertain significance(Last reviewed: Jul 10, 2019)	criteria provided, single submitter	VCV001306936	1	110603479	1306936	1295885	NA	Neutral	0.638	0.175	0.187
KCNA2	452	T	S	NM_004974.4(KCNA2):c.1354A>T (p.Thr452Ser)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Jun 22, 2020)	criteria provided, single submitter	VCV001021350	1	110603429	1021350	1002016	rs765999960	Neutral	0.443	0.328	0.229
KCNA2	458	Y	H	NM_004974.4(KCNA2):c.1372T>C (p.Tyr458His)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, single submitter	VCV001016870	1	110603411	1016870	1002015	rs1649446402	Neutral	0.556	0.283	0.161
KCNA2	459	M	V	NM_004974.4(KCNA2):c.1375A>G	Epileptic encephalopathy, early infantile, 32	Uncertain	criteria	VCV001341871	1	110603408	1341871	1333118	NA	Neutral	0.554	0.269	0.177

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Met459Val)		significance(Last reviewed: Feb 23, 2021)	provided, single submitter										
KCNA2	460	E	D	NM_004974.4(KCNA2):c.1380G>T (p.Glu460Asp)	Epileptic encephalopathy, early infantile, 32	Likely benign(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV000833855	1	110603403	833855	821810	rs1649445890	Neutral	0.506	0.307	0.188
KCNA2	462	Q	R	NM_004974.4(KCNA2):c.1385A>G (p.Gln462Arg)	Epileptic encephalopathy, early infantile, 32	Likely benign(Last reviewed: Nov 13, 2020)	criteria provided, single submitter	VCV001132372	1	110603398	1132372	1109274	NA	Neutral	0.457	0.34	0.202
KCNA2	464	G	S	NM_004974.4(KCNA2):c.1390G>A (p.Gly464Ser)	not provided	Uncertain significance(Last reviewed: Apr 8, 2019)	criteria provided, single submitter	VCV001308309	1	110603393	1308309	1296930	NA	Neutral	0.515	0.266	0.219
KCNA2	478	K	R	NM_004974.4(KCNA2):c.1433A>G (p.Lys478Arg)	not provided	Uncertain significance(Last reviewed: Dec 17, 2018)	criteria provided, single submitter	VCV000804950	1	110603350	804950	792820	rs1243868138	Neutral	0.442	0.372	0.186
KCNA2	480	A	T	NM_004974.4(KCNA2):c.1438G>A (p.Ala480Thr)	not provided	Uncertain significance(Last reviewed: May 16, 2019)	criteria provided, single submitter	VCV001305909	1	110603345	1305909	1297180	NA	Neutral	0.524	0.278	0.199
KCNA2	482	C	S	NM_004974.4(KCNA2):c.1445G>C (p.Cys482Ser)	not provided	Uncertain significance(Last reviewed: Sep 19, 2019)	criteria provided, single submitter	VCV001312361	1	110603338	1312361	1301743	NA	Neutral	0.624	0.207	0.169
KCNA2	489	Y	C	NM_004974.4(KCNA2):c.1466A>G (p.Tyr489Cys)	Epileptic encephalopathy, early infantile, 32	Uncertain significance(Last reviewed: Mar 18, 2020)	criteria provided, single submitter	VCV000999752	1	110603317	999752	986793	rs149776127	Neutral	0.62	0.22	0.16
KCNA2	499	V	A	NM_004974.4(KCNA2):c.1496T>C (p.Val499Ala)	Intellectual disability|Seizures	Uncertain significance(Last reviewed: Nov 18, 2019)	no assertion criteria provided	VCV000977395	1	110603287	977395	965458	rs1017816495	Neutral	0.59	0.217	0.193
KCNA4	89	R	Q	NM_002233.4(KCNA4):c.266G>A (p.Arg89Gln)	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum	Uncertain significance(Last reviewed: Sep 25, 2019)	criteria provided, single submitter	VCV000599408	11	30012413	599408	590745	rs779101828	LOF	0.822	0.084	0.094
KCNA4	608	K	N	NM_002233.4(KCNA4):c.1824G>C (p.Lys608Asn)	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum	Uncertain significance(Last reviewed: Sep 25, 2019)	criteria provided, single submitter	VCV001028009	11	30010855	1028009	1017444	rs200225575	Neutral	0.668	0.228	0.104
KCNA5	1	M	V	NM_002234.4(KCNA5):c.1A>G (p.Met1Val)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Nov 20, 2019)	criteria provided, single submitter	VCV000858451	12	5044148	858451	840004	rs889711431	Neutral	0.574	0.274	0.152
KCNA5	5	L	P	NM_002234.4(KCNA5):c.14T>C (p.Leu5Pro)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jul 9, 2019)	criteria provided, single submitter	VCV000960087	12	5044161	960087	948059	rs1862740772	Neutral	0.557	0.296	0.146
KCNA5	10	N	D	NM_002234.4(KCNA5):c.28A>G (p.Asn10Asp)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Oct 4, 2018)	criteria provided, single submitter	VCV000662585	12	5044175	662585	641186	rs894679405	Neutral	0.524	0.256	0.221
KCNA5	21	E	K	NM_002234.4(KCNA5):c.61G>A (p.Glu21Lys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jul 2, 2020)	criteria provided, single submitter	VCV001006528	12	5044208	1006528	995247	rs1862741663	Neutral	0.475	0.219	0.307
KCNA5	23	R	Q	NM_002234.4(KCNA5):c.68G>A (p.Arg23Gln)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Apr 11, 2020)	criteria provided, single submitter	VCV001001400	12	5044215	1001400	995248	rs1197530568	Neutral	0.529	0.214	0.257
KCNA5	24	A	S	NM_002234.4(KCNA5):c.70G>T (p.Ala24Ser)	Atrial fibrillation, familial, 7	Uncertain	criteria	VCV000957605	12	5044217	957605	948060	rs1862741867	Neutral	0.378	0.3	0.322

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						significance(Last reviewed: Nov 5, 2019)	provided, single submitter										
KCNA5	27	G	S	NM_002234.4(KCNA5):c.79G>A (p.Gly27Ser)	not provided|Atrial fibrillation, familial, 7	Conflicting interpretations of pathogenicity(Last reviewed: Nov 13, 2020)	criteria provided, conflicting interpretations	VCV000469598	12	5044226	469598	462511	rs201238766	Neutral	0.406	0.308	0.286
KCNA5	29	A	D	NM_002234.4(KCNA5):c.86C>A (p.Ala29Asp)	Atrial fibrillation, familial, 7	Likely benign(Last reviewed: Nov 11, 2020)	criteria provided, single submitter	VCV000697538	12	5044233	697538	688005	rs763281934	Neutral	0.441	0.248	0.311
KCNA5	31	G	A	NM_002234.4(KCNA5):c.92G>C (p.Gly31Ala)	not provided	Uncertain significance(Last reviewed: Dec 13, 2018)	criteria provided, single submitter	VCV001207644	12	5044239	1207644	1198391	NA	Neutral	0.404	0.345	0.25
KCNA5	31	G	R	NM_002234.4(KCNA5):c.91G>C (p.Gly31Arg)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Dec 19, 2019)	criteria provided, single submitter	VCV000855013	12	5044238	855013	840005	rs1862742154	Neutral	0.493	0.289	0.218
KCNA5	31	G	V	NM_002234.4(KCNA5):c.92G>T (p.Gly31Val)	not provided|Atrial fibrillation, familial, 7	Benign/Likely benign(Last reviewed: Mar 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000309332	12	5044239	309332	317551	rs61737395	Neutral	0.416	0.363	0.221
KCNA5	33	E	V	NM_002234.4(KCNA5):c.98A>T (p.Glu33Val)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Oct 29, 2021)	criteria provided, multiple submitters, no conflicts	VCV000537313	12	5044245	537313	527200	rs71584818	Neutral	0.412	0.274	0.313
KCNA5	36	C	S	NM_002234.4(KCNA5):c.106T>A (p.Cys36Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jan 31, 2020)	criteria provided, single submitter	VCV000566847	12	5044253	566847	566861	rs910072879	Neutral	0.482	0.289	0.229
KCNA5	39	T	P	NM_002234.4(KCNA5):c.115A>C (p.Thr39Pro)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 21, 2020)	criteria provided, single submitter	VCV000842980	12	5044262	842980	840006	rs1299972539	Neutral	0.439	0.254	0.307
KCNA5	47	K	R	NM_002234.4(KCNA5):c.140A>G (p.Lys47Arg)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Apr 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000309333	12	5044287	309333	317552	rs886049573	Neutral	0.573	0.187	0.24
KCNA5	48	E	G	NM_002234.4(KCNA5):c.143A>G (p.Glu48Gly)	Atrial fibrillation, familial, 7	Pathogenic(Last reviewed: May 1, 2013)	no assertion criteria provided	VCV000127137	12	5044290	127137	132635	rs587777336	GOF	0.43	0.108	0.462
KCNA5	52	K	N	NM_002234.4(KCNA5):c.156G>T (p.Lys52Asn)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 16, 2019)	criteria provided, single submitter	VCV000957629	12	5044303	957629	948061	rs768347248	GOF	0.513	0.156	0.332
KCNA5	63	G	V	NM_002234.4(KCNA5):c.188G>T (p.Gly63Val)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Jul 25, 2019)	criteria provided, multiple submitters, no conflicts	VCV000640925	12	5044335	640925	641187	rs768062067	Neutral	0.431	0.358	0.21
KCNA5	66	P	S	NM_002234.4(KCNA5):c.196C>T (p.Pro66Ser)	not provided|Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000665674	12	5044343	665674	641188	rs368699451	Neutral	0.41	0.297	0.293
KCNA5	72	D	H	NM_002234.4(KCNA5):c.214G>C (p.Asp72His)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jan 30,	criteria provided, single submitter	VCV000945889	12	5044361	945889	926657	rs758310507	Neutral	0.445	0.233	0.322

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)											
KCNA5	74	G	E	NM_002234.4(KCNA5):c.221G>A (p.Gly74Glu)	Atrial fibrillation, familial, 7	Likely benign(Last reviewed: Sep 30, 2020)	criteria provided, single submitter	VCV000537319	12	5044368	537319	527208	rs555008698	Neutral	0.485	0.298	0.217
KCNA5	76	R	L	NM_002234.4(KCNA5):c.227G>T (p.Arg76Leu)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 25, 2020)	criteria provided, single submitter	VCV001061754	12	5044374	1061754	1047998	NA	Neutral	0.475	0.308	0.218
KCNA5	77	P	R	NM_002234.4(KCNA5):c.230C>G (p.Pro77Arg)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 5, 2019)	criteria provided, single submitter	VCV000653091	12	5044377	653091	641189	rs1003591437	Neutral	0.468	0.336	0.196
KCNA5	77	P	S	NM_002234.4(KCNA5):c.229C>T (p.Pro77Ser)	not provided|Atrial fibrillation, familial, 7	Benign/Likely benign(Last reviewed: Apr 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000469590	12	5044376	469590	462519	rs202083721	Neutral	0.404	0.354	0.242
KCNA5	84	E	A	NM_002234.4(KCNA5):c.251A>C (p.Glu84Ala)	Atrial fibrillation, familial, 7|not provided	Conflicting interpretations of pathogenicity(Last reviewed: May 12, 2020)	criteria provided, conflicting interpretations	VCV000191456	12	5044398	191456	189345	rs377036305	GOF	0.427	0.188	0.385
KCNA5	87	R	Q	NM_002234.4(KCNA5):c.260G>A (p.Arg87Gln)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Jan 14, 2022)	criteria provided, multiple submitters, no conflicts	VCV000565628	12	5044407	565628	566866	rs71537801	Neutral	0.54	0.221	0.239
KCNA5	90	R	W	NM_002234.4(KCNA5):c.268C>T (p.Arg90Trp)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Mar 23, 2018)	criteria provided, single submitter	VCV000883692	12	5044415	883692	870026	rs764215137	Neutral	0.556	0.233	0.211
KCNA5	116	S	F	NM_002234.4(KCNA5):c.347C>T (p.Ser116Phe)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 3, 2019)	criteria provided, single submitter	VCV000955024	12	5044494	955024	948062	rs962908688	Neutral	0.388	0.43	0.182
KCNA5	127	S	F	NM_002234.4(KCNA5):c.380_381delinsTT (p.Ser127Phe)	not provided	Uncertain significance(Last reviewed: Sep 20, 2019)	criteria provided, single submitter	VCV001302378	12	5044527 - 5044528	1302378	1292659	NA	Neutral	0.687	0.199	0.114
KCNA5	128	G	R	NM_002234.4(KCNA5):c.382G>C (p.Gly128Arg)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jul 6, 2017)	criteria provided, single submitter	VCV000469593	12	5044529	469593	463140	rs766137868	LOF	0.795	0.113	0.092
KCNA5	130	R	H	NM_002234.4(KCNA5):c.389G>A (p.Arg130His)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Apr 27, 2017)	criteria provided, single submitter	VCV000881348	12	5044536	881348	870027	rs751342646	LOF	0.795	0.128	0.076
KCNA5	139	A	E	NM_002234.4(KCNA5):c.416C>A (p.Ala139Glu)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 27, 2019)	criteria provided, single submitter	VCV000961260	12	5044563	961260	948063	rs1351785009	Neutral	0.623	0.211	0.166
KCNA5	148	D	N	NM_002234.4(KCNA5):c.442G>A (p.Asp148Asn)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Jun 25, 2021)	criteria provided, multiple submitters, no conflicts	VCV000958518	12	5044589	958518	948064	rs773318785	LOF	0.629	0.16	0.211
KCNA5	150	A	V	NM_002234.4(KCNA5):c.449C>T (p.Ala150Val)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Oct 7, 2019)	criteria provided, single submitter	VCV000930576	12	5044596	930576	919448	rs1862749143	LOF	0.756	0.157	0.086
KCNA5	154	R	S	NM_002234.4(KCNA5):c.460C>A (p.Arg154Ser)	not provided	Uncertain significance(Last	criteria provided, single	VCV000499403	12	5044607	499403	490827	rs756977657	LOF	0.803	0.119	0.079

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Jan 16, 2017)	submitter										
KCNA5	154	R	S	NM_002234.4(KCNA5):c.460C>A (p.Arg154Ser)	not provided	Uncertain significance(Last reviewed: Jan 16, 2017)	criteria provided, single submitter	VCV000499403	12	5044607	499403	490827	rs756977657	LOF	0.803	0.119	0.079
KCNA5	154	R	S	NM_002234.4(KCNA5):c.459_460delinsAA (p.Arg154Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jan 26, 2019)	criteria provided, single submitter	VCV000849518	12	5044606 - 5044607	849518	840008	rs1862749418	LOF	0.803	0.119	0.079
KCNA5	154	R	S	NM_002234.4(KCNA5):c.459_460delinsAA (p.Arg154Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jan 26, 2019)	criteria provided, single submitter	VCV000849518	12	5044606 - 5044607	849518	840008	rs1862749418	LOF	0.803	0.119	0.079
KCNA5	155	Y	C	NM_002234.4(KCNA5):c.464A>G (p.Tyr155Cys)	Atrial fibrillation, familial, 7|not provided	Conflicting interpretations of pathogenicity(Last reviewed: May 28, 2019)	criteria provided, conflicting interpretations	VCV000191457	12	5044611	191457	189346	rs202117321	LOF	0.855	0.053	0.092
KCNA5	157	D	E	NM_002234.4(KCNA5):c.471C>G (p.Asp157Glu)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 31, 2020)	criteria provided, single submitter	VCV001004369	12	5044618	1004369	995249	rs200395850	LOF	0.793	0.127	0.08
KCNA5	166	D	A	NM_002234.4(KCNA5):c.497A>C (p.Asp166Ala)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Dec 19, 2019)	criteria provided, multiple submitters, no conflicts	VCV000537306	12	5044644	537306	527454	rs748629738	LOF	0.687	0.128	0.185
KCNA5	174	G	A	NM_002234.4(KCNA5):c.521_522delinsCC (p.Gly174Ala)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 26, 2018)	criteria provided, single submitter	VCV000572604	12	5044668 - 5044669	572604	568088	rs1565465275	Neutral	0.671	0.205	0.124
KCNA5	181	S	Y	NM_002234.4(KCNA5):c.542C>A (p.Ser181Tyr)	not provided	Uncertain significance(Last reviewed: Feb 10, 2020)	criteria provided, single submitter	VCV001314974	12	5044689	1314974	1305235	NA	LOF	0.683	0.145	0.171
KCNA5	182	G	R	NM_002234.4(KCNA5):c.544G>A (p.Gly182Arg)	not provided|Atrial fibrillation, familial, 7	Conflicting interpretations of pathogenicity(Last reviewed: Nov 5, 2020)	criteria provided, conflicting interpretations	VCV000469594	12	5044691	469594	463145	rs755408841	LOF	0.816	0.085	0.1
KCNA5	184	R	H	NM_002234.4(KCNA5):c.551G>A (p.Arg184His)	not provided	Uncertain significance(Last reviewed: Jun 24, 2013)	criteria provided, single submitter	VCV000191458	12	5044698	191458	189347	rs375080039	LOF	0.678	0.138	0.184
KCNA5	187	R	K	NM_002234.4(KCNA5):c.560G>A (p.Arg187Lys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV000968432	12	5044707	968432	956896	rs1862751394	LOF	0.732	0.11	0.158
KCNA5	188	P	A	NM_002234.4(KCNA5):c.562C>G (p.Pro188Ala)	not provided	Uncertain significance(Last reviewed: Nov 1, 2018)	criteria provided, single submitter	VCV000806778	12	5044709	806778	796786	rs907032325	Neutral	0.696	0.166	0.138
KCNA5	190	N	K	NM_002234.4(KCNA5):c.570C>G (p.Asn190Lys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Apr 1, 2019)	criteria provided, single submitter	VCV000844475	12	5044717	844475	840009	rs12720444	LOF	0.754	0.106	0.14
KCNA5	201	R	C	NM_002234.4(KCNA5):c.601C>T (p.Arg201Cys)	not provided	Uncertain significance(Last reviewed: Jun 1, 2018)	criteria provided, single submitter	VCV000806779	12	5044748	806779	796787	rs764783669	Neutral	0.606	0.254	0.14

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA5	201	R	H	NM_002234.4(KCNA5):c.602G>A (p.Arg201His)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000881350	12	5044749	881350	870029	rs765380398	LOF	0.702	0.137	0.161
KCNA5	206	G	A	NM_002234.4(KCNA5):c.617G>C (p.Gly206Ala)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 6, 2020)	criteria provided, single submitter	VCV001011111	12	5044764	1011111	995250	rs200813950	Neutral	0.599	0.22	0.181
KCNA5	207	D	N	NM_002234.4(KCNA5):c.619G>A (p.Asp207Asn)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Apr 18, 2019)	criteria provided, single submitter	VCV000840414	12	5044766	840414	840010	rs757340322	Neutral	0.664	0.15	0.186
KCNA5	210	M	T	NM_002234.4(KCNA5):c.629T>C (p.Met210Thr)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 31, 2019)	criteria provided, single submitter	VCV000658568	12	5044776	658568	641191	rs1228879118	Neutral	0.527	0.319	0.154
KCNA5	211	E	D	NM_002234.4(KCNA5):c.633G>C (p.Glu211Asp)	Atrial fibrillation, familial, 7|not specified| Primary pulmonary hypertension 1	Conflicting interpretations of pathogenicity(Last reviewed: Dec 6, 2020)	criteria provided, conflicting interpretations	VCV000191459	12	5044780	191459	189348	rs35853292	Neutral	0.669	0.173	0.158
KCNA5	211	E	Q	NM_002234.4(KCNA5):c.631G>C (p.Glu211Gln)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 26, 2020)	criteria provided, single submitter	VCV001010338	12	5044778	1010338	995251	rs1862753153	Neutral	0.661	0.174	0.165
KCNA5	212	R	C	NM_002234.4(KCNA5):c.634C>T (p.Arg212Cys)	Atrial fibrillation, familial, 7|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Nov 25, 2020)	criteria provided, conflicting interpretations	VCV000191460	12	5044781	191460	189349	rs77281462	Neutral	0.55	0.313	0.137
KCNA5	214	R	C	NM_002234.4(KCNA5):c.640C>T (p.Arg214Cys)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Jan 11, 2021)	criteria provided, multiple submitters, no conflicts	VCV000661693	12	5044787	661693	641192	rs3197074	Neutral	0.609	0.257	0.134
KCNA5	223	E	K	NM_002234.4(KCNA5):c.667G>A (p.Glu223Lys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Oct 25, 2018)	criteria provided, single submitter	VCV000537307	12	5044814	537307	527216	rs367992628	Neutral	0.673	0.156	0.172
KCNA5	230	N	H	NM_002234.4(KCNA5):c.688A>C (p.Asn230His)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 30, 2020)	criteria provided, single submitter	VCV001035260	12	5044835	1035260	1030987	rs1283845498	LOF	0.659	0.121	0.22
KCNA5	230	N	S	NM_002234.4(KCNA5):c.689A>G (p.Asn230Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 27, 2020)	criteria provided, single submitter	VCV001042694	12	5044836	1042694	1030988	rs1431311668	LOF	0.652	0.132	0.216
KCNA5	234	R	L	NM_002234.4(KCNA5):c.701G>T (p.Arg234Leu)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000191461	12	5044848	191461	189350	rs371521698	Neutral	0.631	0.199	0.17
KCNA5	234	R	S	NM_002234.4(KCNA5):c.700C>A (p.Arg234Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 14, 2020)	criteria provided, single submitter	VCV001034629	12	5044847	1034629	1030989	rs747919632	LOF	0.732	0.105	0.164
KCNA5	235	Q	H	NM_002234.4(KCNA5):c.705G>T (p.Gln235His)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jan 21, 2020)	criteria provided, single submitter	VCV000855542	12	5044852	855542	840011	rs773235814	LOF	0.701	0.112	0.187
KCNA5	236	V	M	NM_002234.4(KCNA5):c.706G>A (p.Val236Met)	Atrial fibrillation, familial, 7	Uncertain significance(Last	criteria provided, single	VCV000638821	12	5044853	638821	641193	rs762579966	Neutral	0.498	0.357	0.145

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Aug 7, 2018)	submitter										
KCNA5	251	A	T	NM_002234.4(KCNA5):c.751G>A (p.Ala251Thr)	not specified|not provided|Atrial fibrillation, familial, 7	Benign/Likely benign(Last reviewed: Dec 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000191570	12	5044898	191570	189351	rs12720442	Neutral	0.485	0.403	0.111
KCNA5	252	I	L	NM_002234.4(KCNA5):c.754A>C (p.Ile252Leu)	not provided	Uncertain significance(Last reviewed: Jun 13, 2019)	criteria provided, single submitter	VCV001306517	12	5044901	1306517	1296460	NA	Neutral	0.453	0.471	0.076
KCNA5	252	I	V	NM_002234.4(KCNA5):c.754A>G (p.Ile252Val)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 26, 2016)	criteria provided, single submitter	VCV000469597	12	5044901	469597	462525	rs1555100234	Neutral	0.441	0.485	0.074
KCNA5	253	A	T	NM_002234.4(KCNA5):c.757G>A (p.Ala253Thr)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 24, 2020)	criteria provided, single submitter	VCV000842461	12	5044904	842461	840012	rs1343401682	Neutral	0.571	0.345	0.085
KCNA5	257	V	A	NM_002234.4(KCNA5):c.770T>C (p.Val257Ala)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 11, 2020)	criteria provided, single submitter	VCV000998896	12	5044917	998896	995252	rs1565465513	Neutral	0.576	0.367	0.057
KCNA5	259	V	F	NM_002234.4(KCNA5):c.775G>T (p.Val259Phe)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 30, 2020)	criteria provided, single submitter	VCV000574748	12	5044922	574748	566871	rs528511896	Neutral	0.599	0.323	0.078
KCNA5	260	I	F	NM_002234.4(KCNA5):c.778A>T (p.Ile260Phe)	not provided	Uncertain significance(Last reviewed: May 24, 2019)	criteria provided, single submitter	VCV001305998	12	5044925	1305998	1297269	NA	Neutral	0.644	0.302	0.055
KCNA5	266	T	I	NM_002234.4(KCNA5):c.797C>T (p.Thr266Ile)	Atrial fibrillation, familial, 7|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 6, 2020)	criteria provided, conflicting interpretations	VCV000579528	12	5044944	579528	568090	rs369750762	Neutral	0.606	0.3	0.094
KCNA5	275	F	S	NM_002234.4(KCNA5):c.824T>C (p.Phe275Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 2, 2019)	criteria provided, single submitter	VCV000953579	12	5044971	953579	948065	rs1183786437	Neutral	0.625	0.226	0.149
KCNA5	276	R	K	NM_002234.4(KCNA5):c.827G>A (p.Arg276Lys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 17, 2019)	criteria provided, single submitter	VCV000943244	12	5044974	943244	936149	rs762170694	LOF	0.722	0.091	0.187
KCNA5	283	R	C	NM_002234.4(KCNA5):c.847C>T (p.Arg283Cys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 23, 2019)	criteria provided, multiple submitters, no conflicts	VCV000309337	12	5044994	309337	331616	rs886049575	Neutral	0.47	0.353	0.176
KCNA5	283	R	H	NM_002234.4(KCNA5):c.848G>A (p.Arg283His)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 28, 2017)	criteria provided, single submitter	VCV000469599	12	5044995	469599	463016	rs1020769441	Neutral	0.505	0.294	0.202
KCNA5	285	P	A	NM_002234.4(KCNA5):c.853C>G (p.Pro285Ala)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 24, 2019)	criteria provided, single submitter	VCV000834438	12	5045000	834438	840013	rs1862757483	Neutral	0.371	0.455	0.174
KCNA5	287	A	L	NM_002234.4(KCNA5):c.859_860delinsTT (p.Ala287Leu)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Oct 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000596455	12	5045006 - 5045007	596455	587516	rs1565465582	Neutral	0.392	0.401	0.208

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA5	287	A	S	NM_002234.4(KCNA5):c.859G>T (p.Ala287Ser)	Atrial fibrillation, familial, 7	Likely benign(Last reviewed: Dec 31, 2019)	criteria provided, single submitter	VCV000723488	12	5045006	723488	738695	rs149582940	Neutral	0.389	0.336	0.275
KCNA5	287	A	V	NM_002234.4(KCNA5):c.860C>T (p.Ala287Val)	Atrial fibrillation, familial, 7	Likely benign(Last reviewed: Dec 31, 2019)	criteria provided, single submitter	VCV000723489	12	5045007	723489	738696	rs144246051	Neutral	0.379	0.389	0.232
KCNA5	293	A	T	NM_002234.4(KCNA5):c.877G>A (p.Ala293Thr)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 13, 2017)	criteria provided, single submitter	VCV000537310	12	5045024	537310	527460	rs898525645	Neutral	0.366	0.35	0.284
KCNA5	296	P	S	NM_002234.4(KCNA5):c.886C>T (p.Pro296Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 15, 2020)	criteria provided, single submitter	VCV001034637	12	5045033	1034637	1030990	rs1172777549	Neutral	0.376	0.434	0.189
KCNA5	299	N	K	NM_002234.4(KCNA5):c.897C>G (p.Asn299Lys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 17, 2020)	criteria provided, single submitter	VCV001051622	12	5045044	1051622	1048000	NA	Neutral	0.533	0.263	0.204
KCNA5	300	G	C	NM_002234.4(KCNA5):c.898G>T (p.Gly300Cys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Dec 8, 2016)	criteria provided, single submitter	VCV000469601	12	5045045	469601	462286	rs148708451	Neutral	0.407	0.423	0.17
KCNA5	300	G	S	NM_002234.4(KCNA5):c.898G>A (p.Gly300Ser)	Atrial fibrillation, familial, 7|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Sep 22, 2020)	criteria provided, conflicting interpretations	VCV000191462	12	5045045	191462	189352	rs148708451	Neutral	0.414	0.371	0.216
KCNA5	302	G	Q	NM_002234.4(KCNA5):c.904_905delinsCA (p.Gly302Gln)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Dec 27, 2019)	criteria provided, single submitter	VCV000859386	12	5045051 - 5045052	859386	840014	rs1862758690	Neutral	0.44	0.33	0.23
KCNA5	304	M	T	NM_002234.4(KCNA5):c.911T>C (p.Met304Thr)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 12, 2019)	criteria provided, single submitter	VCV000469602	12	5045058	469602	462528	rs745760298	Neutral	0.386	0.338	0.276
KCNA5	305	A	T	NM_002234.4(KCNA5):c.913G>A (p.Ala305Thr)	not provided|Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Mar 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000127136	12	5045060	127136	132634	rs199794307	Neutral	0.353	0.311	0.336
KCNA5	306	P	Q	NM_002234.4(KCNA5):c.917C>A (p.Pro306Gln)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 17, 2020)	criteria provided, single submitter	VCV001005634	12	5045064	1005634	995253	rs527534559	Neutral	0.447	0.367	0.187
KCNA5	307	P	S	NM_002234.4(KCNA5):c.919C>T (p.Pro307Ser)	Atrial fibrillation, familial, 7|not specified|not provided|altered potassium channel function	Conflicting interpretations of pathogenicity(Last reviewed: Dec 29, 2020)	criteria provided, conflicting interpretations	VCV000191463	12	5045066	191463	189353	rs17215409	Neutral	0.424	0.395	0.181
KCNA5	309	G	D	NM_002234.4(KCNA5):c.926G>A (p.Gly309Asp)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: Jul 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000852918	12	5045073	852918	840015	rs369120527	Neutral	0.523	0.249	0.227
KCNA5	310	P	L	NM_002234.4(KCNA5):c.929C>T (p.Pro310Leu)	not provided|Atrial fibrillation, familial, 7|not specified	Benign/Likely benign(Last reviewed: Jan 12, 2021)	criteria provided, multiple submitters, no conflicts	VCV000191571	12	5045076	191571	189354	rs17215402	Neutral	0.445	0.397	0.158
KCNA5	321	A	T	NM_002234.4(KCNA5):c.961G>A (p.Ala321Thr)	Atrial fibrillation, familial, 7|not provided	Likely benign(Last reviewed: Aug 6, 2020)	criteria provided, single submitter	VCV000537314	12	5045108	537314	527754	rs765142607	GOF	0.573	0.124	0.303

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA5	321	A	V	NM_002234.4(KCNA5):c.962C>T (p.Ala321Val)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 4, 2020)	criteria provided, single submitter	VCV001015513	12	5045109	1015513	1010477	rs529780345	GOF	0.53	0.192	0.278
KCNA5	322	D	E	NM_002234.4(KCNA5):c.966C>G (p.Asp322Glu)	not provided	Uncertain significance(Last reviewed: Jun 24, 2013)	criteria provided, single submitter	VCV000191464	12	5045113	191464	189355	rs199525305	LOF	0.715	0.058	0.227
KCNA5	322	D	H	NM_002234.4(KCNA5):c.964G>C (p.Asp322His)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Apr 18, 2017)	criteria provided, single submitter	VCV000469603	12	5045111	469603	463153	rs139614200	GOF	0.645	0.065	0.29
KCNA5	322	D	N	NM_002234.4(KCNA5):c.964G>A (p.Asp322Asn)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Dec 26, 2019)	criteria provided, single submitter	VCV000836547	12	5045111	836547	840016	rs139614200	GOF	0.693	0.058	0.249
KCNA5	323	P	R	NM_002234.4(KCNA5):c.968C>G (p.Pro323Arg)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 27, 2020)	criteria provided, single submitter	VCV001042033	12	5045115	1042033	1030991	rs1333918540	LOF	0.787	0.069	0.143
KCNA5	323	P	S	NM_002234.4(KCNA5):c.967C>T (p.Pro323Ser)	not provided	Uncertain significance(Last reviewed: Dec 8, 2016)	criteria provided, single submitter	VCV000498922	12	5045114	498922	490346	rs376977376	LOF	0.768	0.081	0.151
KCNA5	347	P	S	NM_002234.4(KCNA5):c.1039C>T (p.Pro347Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Oct 3, 2019)	criteria provided, single submitter	VCV000469581	12	5045186	469581	463158	rs752143941	LOF	0.838	0.098	0.064
KCNA5	348	S	N	NM_002234.4(KCNA5):c.1043G>A (p.Ser348Asn)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 6, 2020)	criteria provided, single submitter	VCV001024450	12	5045190	1024450	1010478	rs755829081	LOF	0.769	0.053	0.178
KCNA5	349	K	E	NM_002234.4(KCNA5):c.1045A>G (p.Lys349Glu)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 19, 2018)	criteria provided, single submitter	VCV000565316	12	5045192	565316	565534	rs1565465758	LOF	0.816	0.057	0.127
KCNA5	354	R	W	NM_002234.4(KCNA5):c.1060C>T (p.Arg354Trp)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Dec 29, 2019)	criteria provided, single submitter	VCV000861735	12	5045207	861735	840017	rs746268474	Neutral	0.687	0.158	0.156
KCNA5	379	Q	P	NM_002234.4(KCNA5):c.1136A>C (p.Gln379Pro)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 5, 2020)	criteria provided, single submitter	VCV001051710	12	5045283	1051710	1048001	NA	Neutral	0.551	0.206	0.243
KCNA5	380	Q	E	NM_002234.4(KCNA5):c.1138C>G (p.Gln380Glu)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jul 16, 2020)	criteria provided, single submitter	VCV001042253	12	5045285	1042253	1030992	rs1448543777	LOF	0.663	0.144	0.193
KCNA5	381	P	T	NM_002234.4(KCNA5):c.1141C>A (p.Pro381Thr)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jan 15, 2020)	criteria provided, single submitter	VCV000946819	12	5045288	946819	926658	rs774114620	Neutral	0.437	0.407	0.156
KCNA5	383	G	D	NM_002234.4(KCNA5):c.1148_1149inv (p.Gly383Asp)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 25, 2020)	criteria provided, single submitter	VCV001036384	12	5045295 - 5045296	1036384	1030993	NA	Neutral	0.506	0.302	0.192
KCNA5	384	G	R	NM_002234.4(KCNA5):c.1150G>A (p.Gly384Arg)	Atrial fibrillation, familial, 7|not provided|not specified	Benign/Likely benign(Last reviewed: Oct 19, 2020)	criteria provided, multiple submitters, no conflicts	VCV000191572	12	5045297	191572	189356	rs76708779	Neutral	0.549	0.266	0.185

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA5	384	G	R	NM_002234.4(KCNA5):c.1150G>A (p.Gly384Arg)	Atrial fibrillation, familial, 7|not provided|not specified	Benign/Likely benign(Last reviewed: Oct 19, 2020)	criteria provided, multiple submitters, no conflicts	VCV000191572	12	5045297	191572	189356	rs76708779	Neutral	0.549	0.266	0.185
KCNA5	384	G	R	NM_002234.4(KCNA5):c.1149_1150inv (p.Gly384Arg)	Atrial fibrillation, familial, 7	Benign(Last reviewed: Dec 4, 2020)	criteria provided, single submitter	VCV000469582	12	5045296 - 5045297	469582	463166	NA	Neutral	0.549	0.266	0.185
KCNA5	384	G	R	NM_002234.4(KCNA5):c.1149_1150inv (p.Gly384Arg)	Atrial fibrillation, familial, 7	Benign(Last reviewed: Dec 4, 2020)	criteria provided, single submitter	VCV000469582	12	5045296 - 5045297	469582	463166	NA	Neutral	0.549	0.266	0.185
KCNA5	385	G	R	NM_002234.4(KCNA5):c.1153G>C (p.Gly385Arg)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Mar 14, 2019)	criteria provided, single submitter	VCV000864049	12	5045300	864049	840018	rs1175569776	Neutral	0.554	0.263	0.184
KCNA5	397	A	T	NM_002234.4(KCNA5):c.1189G>A (p.Ala397Thr)	not provided	Uncertain significance(Last reviewed: Jun 1, 2017)	criteria provided, single submitter	VCV000806780	12	5045336	806780	796788	rs1246627857	Neutral	0.638	0.206	0.156
KCNA5	397	A	V	NM_002234.4(KCNA5):c.1190C>T (p.Ala397Val)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 31, 2020)	criteria provided, single submitter	VCV001026646	12	5045337	1026646	1010479	rs748288439	Neutral	0.534	0.309	0.158
KCNA5	400	R	Q	NM_002234.4(KCNA5):c.1199G>A (p.Arg400Gln)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, single submitter	VCV001012073	12	5045346	1012073	995254	rs914933506	LOF	0.877	0.048	0.075
KCNA5	409	R	C	NM_002234.4(KCNA5):c.1225C>T (p.Arg409Cys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 14, 2020)	criteria provided, single submitter	VCV001018732	12	5045372	1018732	1010480	rs763570659	LOF	0.794	0.122	0.084
KCNA5	429	A	V	NM_002234.4(KCNA5):c.1286C>T (p.Ala429Val)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 7, 2019)	criteria provided, single submitter	VCV000853630	12	5045433	853630	840019	rs745704941	Neutral	0.605	0.206	0.189
KCNA5	435	G	R	NM_002234.4(KCNA5):c.1303G>A (p.Gly435Arg)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Mar 18, 2019)	criteria provided, single submitter	VCV000837177	12	5045450	837177	840020	rs1862765898	LOF	0.832	0.102	0.066
KCNA5	438	I	V	NM_002234.4(KCNA5):c.1312A>G (p.Ile438Val)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 29, 2020)	criteria provided, single submitter	VCV001063528	12	5045459	1063528	1048002	NA	Neutral	0.57	0.371	0.059
KCNA5	443	I	M	NM_002234.4(KCNA5):c.1329C>G (p.Ile443Met)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 26, 2021)	criteria provided, single submitter	VCV000997830	12	5045476	997830	985548	rs147209278	Neutral	0.594	0.323	0.083
KCNA5	443	I	V	NM_002234.4(KCNA5):c.1327A>G (p.Ile443Val)	not provided|Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000469583	12	5045474	469583	462538	rs370591031	Neutral	0.531	0.39	0.079
KCNA5	452	V	I	NM_002234.4(KCNA5):c.1354G>A (p.Val452Ile)	Atrial fibrillation, familial, 7|Arrhythmia	Uncertain significance(Last reviewed: Jan 1, 2017)	criteria provided, single submitter	VCV000523450	12	5045501	523450	514020	rs1486417435	Neutral	0.545	0.367	0.089
KCNA5	452	V	L	NM_002234.4(KCNA5):c.1354G>C (p.Val452Leu)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 25, 2018)	criteria provided, single submitter	VCV000574366	12	5045501	574366	566872	rs1486417435	Neutral	0.563	0.342	0.095
KCNA5	455	A	T	NM_002234.4(KCNA5):c.1363G>A	Atrial fibrillation, familial, 7	Uncertain	criteria	VCV001055852	12	5045510	1055852	1048003	NA	LOF	0.723	0.121	0.156

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Ala455Thr)		significance(Last reviewed: May 21, 2020)	provided, single submitter										
KCNA5	465	S	C	NM_002234.4(KCNA5):c.1394C>G (p.Ser465Cys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Oct 20, 2020)	criteria provided, single submitter	VCV001022595	12	5045541	1022595	1010481	rs148812702	LOF	0.632	0.143	0.225
KCNA5	467	I	T	NM_002234.4(KCNA5):c.1400T>C (p.Ile467Thr)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 17, 2018)	criteria provided, single submitter	VCV000537312	12	5045547	537312	527767	rs746800603	Neutral	0.707	0.161	0.132
KCNA5	496	V	M	NM_002234.4(KCNA5):c.1486G>A (p.Val496Met)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 20, 2020)	criteria provided, single submitter	VCV001026851	12	5045633	1026851	1010482	rs757749649	Neutral	0.565	0.295	0.14
KCNA5	498	S	L	NM_002234.4(KCNA5):c.1493C>T (p.Ser498Leu)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Apr 21, 2020)	criteria provided, single submitter	VCV001055873	12	5045640	1055873	1048004	NA	Neutral	0.72	0.145	0.134
KCNA5	503	A	T	NM_002234.4(KCNA5):c.1507G>A (p.Ala503Thr)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Feb 8, 2020)	criteria provided, single submitter	VCV001036453	12	5045654	1036453	1030995	rs140711603	Neutral	0.611	0.277	0.112
KCNA5	505	V	A	NM_002234.4(KCNA5):c.1514T>C (p.Val505Ala)	not provided	Uncertain significance(Last reviewed: Feb 7, 2019)	criteria provided, single submitter	VCV001304443	12	5045661	1304443	1294718	NA	Neutral	0.57	0.327	0.103
KCNA5	522	F	S	NM_002234.4(KCNA5):c.1565T>C (p.Phe522Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 25, 2019)	criteria provided, single submitter	VCV000579216	12	5045712	579216	565541	rs1565466043	Neutral	0.617	0.242	0.141
KCNA5	525	R	W	NM_002234.4(KCNA5):c.1573C>T (p.Arg525Trp)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 21, 2020)	criteria provided, single submitter	VCV000944575	12	5045720	944575	926659	rs376656991	Neutral	0.501	0.344	0.155
KCNA5	527	T	M	NM_002234.4(KCNA5):c.1580C>T (p.Thr527Met)	Atrial fibrillation, familial, 7|not provided	Uncertain significance(Last reviewed: May 9, 2019)	criteria provided, multiple submitters, no conflicts	VCV000013470	12	5045727	13470	28509	rs121908591	Neutral	0.528	0.333	0.139
KCNA5	530	E	K	NM_002234.4(KCNA5):c.1588G>A (p.Glu530Lys)	not provided|Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jul 13, 2020)	criteria provided, multiple submitters, no conflicts	VCV000848914	12	5045735	848914	840021	rs915470188	Neutral	0.711	0.162	0.127
KCNA5	532	P	L	NM_002234.4(KCNA5):c.1595C>T (p.Pro532Leu)	not provided|Atrial fibrillation, familial, 7	Conflicting interpretations of pathogenicity(Last reviewed: Dec 14, 2020)	criteria provided, conflicting interpretations	VCV000469586	12	5045742	469586	462542	rs17221812	Neutral	0.419	0.492	0.089
KCNA5	546	P	S	NM_002234.4(KCNA5):c.1636C>T (p.Pro546Ser)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Mar 12, 2020)	criteria provided, single submitter	VCV001023092	12	5045783	1023092	1010483	rs142289285	Neutral	0.422	0.457	0.12
KCNA5	554	R	Q	NM_002234.4(KCNA5):c.1661G>A (p.Arg554Gln)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Nov 13, 2019)	criteria provided, single submitter	VCV000966389	12	5045808	966389	956897	rs71581016	Neutral	0.528	0.323	0.148
KCNA5	557	S	N	NM_002234.4(KCNA5):c.1670G>A (p.Ser557Asn)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Aug 26, 2020)	criteria provided, single submitter	VCV001014743	12	5045817	1014743	1010484	rs201466058	Neutral	0.529	0.308	0.163

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNA5	558	G	R	NM_002234.4(KCNA5):c.1672G>A (p.Gly558Arg)	Atrial fibrillation, familial, 7	Conflicting interpretations of pathogenicity(Last reviewed: Nov 21, 2020)	criteria provided, conflicting interpretations	VCV000469587	12	5045819	469587	463167	rs201328038	Neutral	0.562	0.282	0.156
KCNA5	561	G	R	NM_002234.4(KCNA5):c.1681G>C (p.Gly561Arg)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Sep 7, 2017)	criteria provided, single submitter	VCV000537309	12	5045828	537309	527210	rs1440776773	Neutral	0.596	0.257	0.147
KCNA5	568	G	V	NM_002234.4(KCNA5):c.1703G>T (p.Gly568Val)	not provided|Heart disease|Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jul 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000374033	12	5045850	374033	360948	rs71581017	Neutral	0.445	0.437	0.118
KCNA5	568	G	W	NM_002234.4(KCNA5):c.1702G>T (p.Gly568Trp)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Oct 29, 2018)	criteria provided, single submitter	VCV000648351	12	5045849	648351	641194	rs1281374886	Neutral	0.478	0.405	0.117
KCNA5	576	A	T	NM_002234.4(KCNA5):c.1726G>A (p.Ala576Thr)	not provided	Uncertain significance(Last reviewed: May 2, 2019)	criteria provided, single submitter	VCV001305421	12	5045873	1305421	1298015	NA	Neutral	0.466	0.343	0.192
KCNA5	576	A	V	NM_002234.4(KCNA5):c.1727C>T (p.Ala576Val)	Atrial fibrillation, familial, 7	Pathogenic(Last reviewed: May 1, 2009)	no assertion criteria provided	VCV000013471	12	5045874	13471	28510	rs121908592	Neutral	0.439	0.387	0.173
KCNA5	577	R	Q	NM_002234.4(KCNA5):c.1730G>A (p.Arg577Gln)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Oct 14, 2019)	criteria provided, single submitter	VCV000537311	12	5045877	537311	527211	rs139805781	Neutral	0.653	0.227	0.12
KCNA5	578	R	K	NM_002234.4(KCNA5):c.1733G>A (p.Arg578Lys)	Atrial fibrillation, familial, 7|not specified| Brugada syndrome 1|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Dec 4, 2020)	criteria provided, conflicting interpretations	VCV000191573	12	5045880	191573	189357	rs12720445	Neutral	0.707	0.184	0.109
KCNA5	586	C	G	NM_002234.4(KCNA5):c.1756T>G (p.Cys586Gly)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 4, 2020)	criteria provided, single submitter	VCV001017278	12	5045903	1017278	1010485	rs1862773506	Neutral	0.551	0.324	0.124
KCNA5	597	R	Q	NM_002234.4(KCNA5):c.1790G>A (p.Arg597Gln)	Atrial fibrillation, familial, 7|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 25, 2021)	criteria provided, conflicting interpretations	VCV000537308	12	5045937	537308	527217	rs201342234	Neutral	0.637	0.228	0.135
KCNA5	597	R	W	NM_002234.4(KCNA5):c.1789C>T (p.Arg597Trp)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Apr 28, 2017)	criteria provided, single submitter	VCV000882944	12	5045936	882944	870030	rs759691488	Neutral	0.479	0.394	0.127
KCNA5	609	R	P	NM_002234.4(KCNA5):c.1826G>C (p.Arg609Pro)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 29, 2017)	criteria provided, single submitter	VCV000469588	12	5045973	469588	462545	rs991950771	Neutral	0.654	0.244	0.102
KCNA5	609	R	Q	NM_002234.4(KCNA5):c.1826G>A (p.Arg609Gln)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: May 7, 2019)	criteria provided, single submitter	VCV000665819	12	5045973	665819	641195	rs991950771	Neutral	0.729	0.173	0.098
KCNA5	610	E	K	NM_002234.4(KCNA5):c.1828G>A (p.Glu610Lys)	Atrial fibrillation, familial, 7	Pathogenic(Last reviewed: May 1, 2009)	no assertion criteria provided	VCV000013472	12	5045975	13472	28511	rs121908593	LOF	0.763	0.139	0.098
KCNA5	611	T	K	NM_002234.4(KCNA5):c.1832C>A (p.Thr611Lys)	Atrial fibrillation, familial, 7	Uncertain significance(Last reviewed: Jun 7, 2018)	criteria provided, single submitter	VCV000566746	12	5045979	566746	566879	rs765166524	Neutral	0.714	0.183	0.103

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNB1	2	P	R	NM_004975.4(KCNB1):c.5C>G (p.Pro2Arg)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Oct 14, 2020)	criteria provided, single submitter	VCV001167120	20	49482476	1167120	1158898	NA	Neutral	0.71	0.174	0.116
KCNB1	2	P	S	NM_004975.4(KCNB1):c.4C>T (p.Pro2Ser)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Dec 27, 2019)	criteria provided, single submitter	VCV000833790	20	49482477	833790	822172	rs1054441474	Neutral	0.7	0.184	0.116
KCNB1	7	K	E	NM_004975.4(KCNB1):c.19A>G (p.Lys7Glu)	Epileptic encephalopathy, early infantile, 26| Seizures	Uncertain significance(Last reviewed: Aug 2, 2019)	criteria provided, multiple submitters, no conflicts	VCV000577816	20	49482462	577816	571303	rs936778119	Neutral	0.675	0.2	0.125
KCNB1	24	V	M	NM_004975.4(KCNB1):c.70G>A (p.Val24Met)	not provided	Uncertain significance(Last reviewed: Nov 2, 2020)	criteria provided, single submitter	VCV001313585	20	49482411	1313585	1303846	NA	LOF	0.768	0.134	0.098
KCNB1	25	R	Q	NM_004975.4(KCNB1):c.74_75delinsAA (p.Arg25Gln)	not provided	Uncertain significance(Last reviewed: May 17, 2021)	criteria provided, single submitter	VCV001309158	20	49482406 - 49482407	1309158	1299858	NA	LOF	0.73	0.16	0.11
KCNB1	28	A	V	NM_004975.4(KCNB1):c.83C>T (p.Ala28Val)	Epileptic encephalopathy, early infantile, 26|not provided	Uncertain significance(Last reviewed: Jun 18, 2019)	criteria provided, multiple submitters, no conflicts	VCV000936998	20	49482398	936998	939007	rs769852170	LOF	0.753	0.136	0.111
KCNB1	38	G	A	NM_004975.4(KCNB1):c.113G>C (p.Gly38Ala)	not provided	Uncertain significance(Last reviewed: Jun 26, 2017)	criteria provided, single submitter	VCV000432932	20	49482368	432932	426343	rs1555801618	LOF	0.861	0.067	0.072
KCNB1	41	A	V	NM_004975.4(KCNB1):c.122C>T (p.Ala41Val)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Dec 6, 2019)	criteria provided, single submitter	VCV000855507	20	49482359	855507	848478	rs1980525336	LOF	0.803	0.115	0.082
KCNB1	42	H	Q	NM_004975.4(KCNB1):c.126C>G (p.His42Gln)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Dec 13, 2018)	criteria provided, single submitter	VCV000580878	20	49482355	580878	575116	rs1568658526	LOF	0.805	0.115	0.08
KCNB1	43	E	G	NM_004975.4(KCNB1):c.128A>G (p.Glu43Gly)	Epileptic encephalopathy	Likely pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633619	20	49482353	633619	622069	rs1568658507	LOF	0.795	0.12	0.085
KCNB1	81	N	I	NM_004975.4(KCNB1):c.242A>T (p.Asn81Ile)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jan 19, 2021)	no assertion criteria provided	VCV001174488	20	49482239	1174488	1163751	NA	LOF	0.812	0.089	0.098
KCNB1	114	S	R	NM_004975.4(KCNB1):c.342C>A (p.Ser114Arg)	not provided	Uncertain significance(Last reviewed: Oct 20, 2020)	criteria provided, single submitter	VCV001312383	20	49482139	1312383	1301765	NA	LOF	0.848	0.064	0.087
KCNB1	137	Y	F	NM_004975.4(KCNB1):c.410A>T (p.Tyr137Phe)	not provided	Uncertain significance(Last reviewed: Oct 13, 2020)	criteria provided, single submitter	VCV001162860	20	49482071	1162860	1152899	NA	LOF	0.812	0.092	0.096
KCNB1	144	M	I	NM_004975.4(KCNB1):c.432G>A (p.Met144Ile)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV000950197	20	49482049	950197	929222	rs1179147119	LOF	0.799	0.116	0.085
KCNB1	154	T	I	NM_004975.4(KCNB1):c.461C>T (p.Thr154Ile)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Aug 21, 2020)	criteria provided, single submitter	VCV001110401	20	49482020	1110401	1107109	NA	LOF	0.783	0.11	0.107
KCNB1	157	E	D	NM_004975.4(KCNB1):c.471G>T (p.Glu157Asp)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last	criteria provided, single	VCV000582652	20	49482010	582652	571295	rs144926751	Neutral	0.727	0.168	0.105

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Apr 2, 2018)	submitter										
KCNB1	189	K	N	NM_004975.4(KCNB1):c.567G>C (p.Lys189Asn)	not provided	Uncertain significance(Last reviewed: Dec 1, 2019)	criteria provided, single submitter	VCV000872734	20	49481914	872734	860652	rs1980508600	LOF	0.845	0.077	0.078
KCNB1	193	I	K	NM_004975.4(KCNB1):c.578T>A (p.Ile193Lys)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Oct 8, 2018)	criteria provided, single submitter	VCV000475267	20	49374982	475267	470457	rs1555889171	LOF	0.862	0.07	0.068
KCNB1	199	I	F	NM_004975.4(KCNB1):c.595A>T (p.Ile199Phe)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000803613	20	49374965	803613	791988	rs1601072041	LOF	0.864	0.047	0.09
KCNB1	199	I	S	NM_004975.4(KCNB1):c.596T>G (p.Ile199Ser)	not provided	Uncertain significance(Last reviewed: Oct 7, 2019)	criteria provided, single submitter	VCV001303788	20	49374964	1303788	1294063	NA	LOF	0.887	0.047	0.066
KCNB1	200	V	I	NM_004975.4(KCNB1):c.598G>A (p.Val200Ile)	not provided	Likely benign(Last reviewed: Mar 23, 2018)	criteria provided, single submitter	VCV000429380	20	49374962	429380	422324	rs748573940	LOF	0.846	0.078	0.075
KCNB1	202	S	C	NM_004975.4(KCNB1):c.605C>G (p.Ser202Cys)	not provided	Likely pathogenic(Last reviewed: Apr 1, 2020)	criteria provided, single submitter	VCV000932381	20	49374955	932381	920972	rs1060499607	LOF	0.875	0.044	0.081
KCNB1	202	S	F	NM_004975.4(KCNB1):c.605C>T (p.Ser202Phe)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Apr 8, 2016)	no assertion criteria provided	VCV000417935	20	49374955	417935	404844	rs1060499607	LOF	0.861	0.042	0.097
KCNB1	209	N	K	NM_004975.4(KCNB1):c.627C>A (p.Asn209Lys)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: May 21, 2020)	criteria provided, single submitter	VCV000943082	20	49374933	943082	939006	rs1984276555	LOF	0.848	0.067	0.085
KCNB1	210	T	M	NM_004975.4(KCNB1):c.629C>T (p.Thr210Met)	developmental encephalopathy with epilepsy|KCNB1-related disorder|not provided|Epileptic encephalopathy, early infantile, 26	Pathogenic/Likely pathogenic(Last reviewed: Dec 30, 2021)	criteria provided, multiple submitters, no conflicts	VCV000542057	20	49374931	542057	533574	rs1555889162	LOF	0.856	0.05	0.094
KCNB1	210	T	R	NM_004975.4(KCNB1):c.629C>G (p.Thr210Arg)	Epileptic encephalopathy	Pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633620	20	49374931	633620	622068	rs1555889162	LOF	0.857	0.056	0.087
KCNB1	218	D	E	NM_004975.4(KCNB1):c.654T>G (p.Asp218Glu)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Sep 1, 2020)	criteria provided, single submitter	VCV001056665	20	49374906	1056665	1051744	NA	LOF	0.778	0.128	0.094
KCNB1	221	G	S	NM_004975.4(KCNB1):c.661G>A (p.Gly221Ser)	not provided	Uncertain significance(Last reviewed: Oct 1, 2019)	criteria provided, single submitter	VCV001308882	20	49374899	1308882	1300910	NA	LOF	0.814	0.099	0.087
KCNB1	235	V	M	NM_004975.4(KCNB1):c.703G>A (p.Val235Met)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jan 17, 2019)	criteria provided, single submitter	VCV000863356	20	49374857	863356	848477	rs1315753296	LOF	0.843	0.077	0.081
KCNB1	246	L	R	NM_004975.4(KCNB1):c.737T>G (p.Leu246Arg)	Epileptic encephalopathy, early infantile, 26	Conflicting interpretations of pathogenicity(Last reviewed: Jul 21, 2020)	criteria provided, conflicting interpretations	VCV000647049	20	49374823	647049	648764	rs1601071839	LOF	0.857	0.067	0.076
KCNB1	247	R	K	NM_004975.4(KCNB1):c.740G>A (p.Arg247Lys)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Mar 13,	criteria provided, single submitter	VCV000852956	20	49374820	852956	848476	rs1984270863	LOF	0.837	0.078	0.086

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)											
KCNB1	251	S	L	NM_004975.4(KCNB1):c.752C>T (p.Ser251Leu)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Apr 14, 2020)	criteria provided, single submitter	VCV001016853	20	49374808	1016853	1014172	rs1984270019	LOF	0.842	0.063	0.095
KCNB1	252	P	S	NM_004975.4(KCNB1):c.754C>T (p.Pro252Ser)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Aug 10, 2020)	criteria provided, single submitter	VCV000998500	20	49374806	998500	999055	rs1984269566	LOF	0.842	0.08	0.078
KCNB1	271	L	P	NM_004975.4(KCNB1):c.812T>C (p.Leu271Pro)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: May 29, 2019)	criteria provided, single submitter	VCV000572716	20	49374748	572716	573574	rs1569017375	LOF	0.865	0.07	0.065
KCNB1	272	P	S	NM_004975.4(KCNB1):c.814C>T (p.Pro272Ser)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000803612	20	49374746	803612	791987	rs1601071747	LOF	0.888	0.052	0.06
KCNB1	281	E	K	NM_004975.4(KCNB1):c.841G>A (p.Glu281Lys)	not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 19, 2020)	criteria provided, conflicting interpretations	VCV000809258	20	49374719	809258	797994	rs1601071708	LOF	0.786	0.117	0.097
KCNB1	286	V	M	NM_004975.4(KCNB1):c.856G>A (p.Val286Met)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Sep 1, 2020)	criteria provided, single submitter	VCV000946306	20	49374704	946306	929221	rs1194019938	LOF	0.815	0.09	0.095
KCNB1	293	R	C	NM_004975.4(KCNB1):c.877C>T (p.Arg293Cys)	Epileptic encephalopathy, early infantile, 26	Conflicting interpretations of pathogenicity(Last reviewed: Feb 26, 2021)	criteria provided, conflicting interpretations	VCV000940052	20	49374683	940052	939005	rs1984264748	LOF	0.824	0.071	0.106
KCNB1	297	Q	R	NM_004975.4(KCNB1):c.890A>G (p.Gln297Arg)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, single submitter	VCV001040672	20	49374670	1040672	1034745	rs749981323	LOF	0.835	0.077	0.088
KCNB1	300	R	C	NM_004975.4(KCNB1):c.898C>T (p.Arg300Cys)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jan 1, 2019)	criteria provided, single submitter	VCV000983032	20	49374662	983032	971148	rs202170806	LOF	0.865	0.042	0.094
KCNB1	303	R	Q	NM_004975.4(KCNB1):c.908G>A (p.Arg303Gln)	Epileptic encephalopathy, early infantile, 26|not provided	Likely pathogenic(Last reviewed: Apr 9, 2019)	criteria provided, multiple submitters, no conflicts	VCV000373805	20	49374652	373805	360423	rs1057518621	LOF	0.851	0.066	0.083
KCNB1	306	R	C	NM_004975.4(KCNB1):c.916C>T (p.Arg306Cys)	Intellectual disability|Epileptic encephalopathy, early infantile, 26|developmental encephalopathy with epilepsy|Myoclonic absence seizure|Epileptic encephalopathy|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Mar 15, 2021)	criteria provided, conflicting interpretations	VCV000449693	20	49374644	449693	446266	rs1555889130	LOF	0.862	0.042	0.095
KCNB1	306	R	H	NM_004975.4(KCNB1):c.917G>A (p.Arg306His)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Jan 1, 2019)	criteria provided, single submitter	VCV000982573	20	49374643	982573	971147	rs1984262011	LOF	0.848	0.062	0.09
KCNB1	307	I	V	NM_004975.4(KCNB1):c.919A>G (p.Ile307Val)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Aug 30, 2020)	criteria provided, single submitter	VCV001057900	20	49374641	1057900	1051742	NA	LOF	0.851	0.077	0.073
KCNB1	311	A	T	NM_004975.4(KCNB1):c.931G>A (p.Ala311Thr)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jan 20, 2020)	criteria provided, single submitter	VCV001056800	20	49374629	1056800	1051741	NA	LOF	0.844	0.073	0.082

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNB1	312	R	C	NM_004975.4(KCNB1):c.934C>T (p.Arg312Cys)	Epileptic encephalopathy, early infantile, 26| Inborn genetic diseases|Intellectual disability| not provided	Pathogenic/Likely pathogenic(Last reviewed: Feb 2, 2022)	criteria provided, multiple submitters, no conflicts	VCV000265207	20	49374626	265207	260231	NA	LOF	0.857	0.046	0.097
KCNB1	312	R	H	NM_004975.4(KCNB1):c.935G>A (p.Arg312His)	Intellectual disability|Early infantile epileptic encephalopathy with suppression bursts|not provided|Delayed speech and language development|Seizures|Epileptic encephalopathy|Difficulty walking|Intellectual disability|Generalized hypotonia|Epileptic encephalopathy|developmental encephalopathy with epilepsy	Pathogenic/Likely pathogenic(Last reviewed: Sep 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000523478	20	49374625	523478	514105	rs1555889127	LOF	0.841	0.067	0.092
KCNB1	313	H	R	NM_004975.4(KCNB1):c.938A>G (p.His313Arg)	not provided	Likely pathogenic(Last reviewed: Jun 7, 2016)	criteria provided, single submitter	VCV000384177	20	49374622	384177	378313	rs1057521887	LOF	0.837	0.078	0.085
KCNB1	313	H	Y	NM_004975.4(KCNB1):c.937C>T (p.His313Tyr)	not provided	Pathogenic(Last reviewed: Oct 12, 2020)	criteria provided, single submitter	VCV000451654	20	49374623	451654	446265	rs918313461	LOF	0.827	0.075	0.097
KCNB1	314	S	A	NM_004975.4(KCNB1):c.940T>G (p.Ser314Ala)	not provided	Likely pathogenic(Last reviewed: Jul 1, 2019)	criteria provided, single submitter	VCV000872733	20	49374620	872733	860651	rs1984260160	LOF	0.854	0.066	0.08
KCNB1	319	S	F	NM_004975.4(KCNB1):c.956C>T (p.Ser319Phe)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Dec 1, 2019)	no assertion criteria provided	VCV000973169	20	49374604	973169	961494	rs1984259606	LOF	0.679	0.13	0.191
KCNB1	319	S	Y	NM_004975.4(KCNB1):c.956C>A (p.Ser319Tyr)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Jun 26, 2020)	criteria provided, single submitter	VCV001067365	20	49374604	1067365	1056615	NA	LOF	0.68	0.143	0.176
KCNB1	321	G	S	NM_004975.4(KCNB1):c.961G>A (p.Gly321Ser)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Apr 8, 2020)	criteria provided, single submitter	VCV001068345	20	49374599	1068345	1056614	NA	Neutral	0.691	0.191	0.118
KCNB1	323	T	I	NM_004975.4(KCNB1):c.968C>T (p.Thr323Ile)	Epileptic encephalopathy	Likely pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633622	20	49374592	633622	622066	rs1569017257	LOF	0.693	0.135	0.172
KCNB1	325	R	Q	NM_004975.4(KCNB1):c.974G>A (p.Arg325Gln)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Apr 14, 2020)	criteria provided, single submitter	VCV001041245	20	49374586	1041245	1034744	rs759765009	Neutral	0.439	0.396	0.165
KCNB1	325	R	W	NM_004975.4(KCNB1):c.973C>T (p.Arg325Trp)	Inborn genetic diseases|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jul 16, 2019)	criteria provided, conflicting interpretations	VCV000985215	20	49374587	985215	974197	rs1276378675	GOF	0.443	0.197	0.359
KCNB1	329	N	S	NM_004975.4(KCNB1):c.986A>G (p.Asn329Ser)	Epileptic encephalopathy, early infantile, 26	Conflicting interpretations of pathogenicity(Last reviewed: Jul 22, 2021)	criteria provided, conflicting interpretations	VCV000959038	20	49374574	959038	951127	rs1984257387	Neutral	0.643	0.233	0.124
KCNB1	330	E	D	NM_004975.4(KCNB1):c.990G>C (p.Glu330Asp)	not provided	Likely pathogenic(Last reviewed: Oct 3, 2015)	criteria provided, single submitter	VCV000429607	20	49374570	429607	422323	rs1131691489	Neutral	0.652	0.231	0.117
KCNB1	330	E	K	NM_004975.4(KCNB1):c.988G>A (p.Glu330Lys)	Inborn genetic diseases	Likely pathogenic(Last reviewed: Mar 17, 2017)	criteria provided, single submitter	VCV000521643	20	49374572	521643	512459	rs1555889114	Neutral	0.643	0.234	0.123
KCNB1	332	G	V	NM_004975.4(KCNB1):c.995G>T (p.Gly332Val)	not provided	Likely	criteria	VCV000451423	20	49374565	451423	446264	rs1555889110	LOF	0.876	0.053	0.071

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						pathogenic(Last reviewed: Jan 13, 2021)	provided, multiple submitters, no conflicts										
KCNB1	334	L	P	NM_004975.4(KCNB1):c.1001T>C (p.Leu334Pro)	Epileptic encephalopathy|Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, multiple submitters, no conflicts	VCV000559895	20	49374559	559895	550642	rs1555889108	LOF	0.872	0.061	0.067
KCNB1	336	L	F	NM_004975.4(KCNB1):c.1006C>T (p.Leu336Phe)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Feb 2, 2020)	criteria provided, single submitter	VCV001057172	20	49374554	1057172	1051740	NA	LOF	0.862	0.058	0.08
KCNB1	339	A	P	NM_004975.4(KCNB1):c.1015G>C (p.Ala339Pro)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Feb 17, 2019)	criteria provided, single submitter	VCV000860413	20	49374545	860413	848475	rs1984255406	LOF	0.864	0.071	0.066
KCNB1	347	S	R	NM_004975.4(KCNB1):c.1041C>G (p.Ser347Arg)	Epileptic encephalopathy|Epileptic encephalopathy, early infantile, 26	Pathogenic(Last reviewed: Sep 19, 2020)	criteria provided, multiple submitters, no conflicts	VCV000475255	20	49374519	475255	470956	NA	LOF	0.859	0.049	0.092
KCNB1	347	S	R	NM_004975.4(KCNB1):c.1041C>G (p.Ser347Arg)	Epileptic encephalopathy|Epileptic encephalopathy, early infantile, 26	Pathogenic(Last reviewed: Sep 19, 2020)	criteria provided, multiple submitters, no conflicts	VCV000475255	20	49374519	475255	470956	NA	LOF	0.859	0.049	0.092
KCNB1	347	S	R	NM_004975.4(KCNB1):c.1041C>A (p.Ser347Arg)	Inborn genetic diseases|Epileptic encephalopathy, early infantile, 26	Pathogenic/Likely pathogenic(Last reviewed: Mar 2, 2018)	criteria provided, multiple submitters, no conflicts	VCV000156533	20	49374519	156533	166304	rs587777848	LOF	0.859	0.049	0.092
KCNB1	347	S	R	NM_004975.4(KCNB1):c.1041C>A (p.Ser347Arg)	Inborn genetic diseases|Epileptic encephalopathy, early infantile, 26	Pathogenic/Likely pathogenic(Last reviewed: Mar 2, 2018)	criteria provided, multiple submitters, no conflicts	VCV000156533	20	49374519	156533	166304	rs587777848	LOF	0.859	0.049	0.092
KCNB1	349	V	F	NM_004975.4(KCNB1):c.1045G>T (p.Val349Phe)	Epileptic encephalopathy, early infantile, 26| Epileptic encephalopathy	Pathogenic(Last reviewed: Oct 29, 2019)	criteria provided, multiple submitters, no conflicts	VCV000633624	20	49374515	633624	622064	rs1569017205	LOF	0.861	0.055	0.084
KCNB1	351	F	S	NM_004975.4(KCNB1):c.1052T>C (p.Phe351Ser)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jul 13, 2020)	criteria provided, single submitter	VCV001055078	20	49374508	1055078	1051739	NA	LOF	0.867	0.066	0.067
KCNB1	353	E	K	NM_004975.4(KCNB1):c.1057G>A (p.Glu353Lys)	not provided	Likely pathogenic(Last reviewed: Jan 26, 2017)	criteria provided, single submitter	VCV000392897	20	49374503	392897	377080	rs1057524688	LOF	0.861	0.063	0.076
KCNB1	367	S	F	NM_004975.4(KCNB1):c.1100C>T (p.Ser367Phe)	Inborn genetic diseases	Likely pathogenic(Last reviewed: Nov 23, 2018)	criteria provided, single submitter	VCV000985970	20	49374460	985970	974196	rs1984252463	LOF	0.867	0.043	0.09
KCNB1	369	W	R	NM_004975.4(KCNB1):c.1105T>C (p.Trp369Arg)	Epileptic encephalopathy, early infantile, 26| Epileptic encephalopathy	Conflicting interpretations of pathogenicity(Last reviewed: Mar 1, 2019)	criteria provided, conflicting interpretations	VCV000633625	20	49374455	633625	622063	rs1569017174	LOF	0.865	0.063	0.071
KCNB1	370	W	R	NM_004975.4(KCNB1):c.1108T>C (p.Trp370Arg)	not provided	Pathogenic(Last reviewed: May 3, 2016)	criteria provided, single submitter	VCV000280560	20	49374452	280560	264751	rs886041743	LOF	0.864	0.054	0.082

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNB1	372	T	I	NM_004975.4(KCNB1):c.1115C>T (p.Thr372Ile)	Intellectual disability	Pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633626	20	49374445	633626	622061	rs1569017160	LOF	0.84	0.056	0.104
KCNB1	372	T	N	NM_004975.4(KCNB1):c.1115C>A (p.Thr372Asn)	Intellectual disability	Likely pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633627	20	49374445	633627	622062	rs1569017160	LOF	0.854	0.066	0.08
KCNB1	374	T	I	NM_004975.4(KCNB1):c.1121C>T (p.Thr374Ile)	Epileptic encephalopathy, early infantile, 26|not provided	Pathogenic(Last reviewed: Nov 12, 2018)	criteria provided, single submitter	VCV000156534	20	49374439	156534	166305	rs587777849	LOF	0.857	0.048	0.095
KCNB1	375	M	V	NM_004975.4(KCNB1):c.1123A>G (p.Met375Val)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jul 8, 2019)	criteria provided, single submitter	VCV000957470	20	49374437	957470	951126	rs1984250919	LOF	0.861	0.058	0.081
KCNB1	377	T	I	NM_004975.4(KCNB1):c.1130C>T (p.Thr377Ile)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Aug 8, 2019)	criteria provided, single submitter	VCV000941236	20	49374430	941236	939004	rs1569017148	LOF	0.868	0.047	0.086
KCNB1	377	T	N	NM_004975.4(KCNB1):c.1130C>A (p.Thr377Asn)	Epileptic encephalopathy	Pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633628	20	49374430	633628	622060	rs1569017148	LOF	0.878	0.049	0.074
KCNB1	378	V	A	NM_004975.4(KCNB1):c.1133T>C (p.Val378Ala)	not provided	Pathogenic(Last reviewed: Dec 1, 2017)	criteria provided, single submitter	VCV000872732	20	49374427	872732	860650	rs1984250089	LOF	0.872	0.047	0.081
KCNB1	378	V	L	NM_004975.4(KCNB1):c.1132G>C (p.Val378Leu)	not provided|Epileptic encephalopathy	Pathogenic(Last reviewed: Oct 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000633629	20	49374428	633629	622059	rs1569017143	LOF	0.856	0.048	0.096
KCNB1	379	G	E	NM_004975.4(KCNB1):c.1136G>A (p.Gly379Glu)	not provided	Pathogenic(Last reviewed: Aug 12, 2019)	criteria provided, single submitter	VCV001199933	20	49374424	1199933	1192247	NA	LOF	0.872	0.047	0.08
KCNB1	379	G	R	NM_004975.4(KCNB1):c.1135G>A (p.Gly379Arg)	Epileptic encephalopathy, early infantile, 26	Pathogenic(Last reviewed: Oct 1, 2014)	no assertion criteria provided	VCV000156535	20	49374425	156535	166306	rs587777850	LOF	0.858	0.046	0.096
KCNB1	379	G	V	NM_004975.4(KCNB1):c.1136G>T (p.Gly379Val)	Epileptic encephalopathy, early infantile, 26	Pathogenic(Last reviewed: Nov 19, 2018)	criteria provided, single submitter	VCV001031489	20	49374424	1031489	1022009	rs1984249721	LOF	0.878	0.045	0.077
KCNB1	380	Y	C	NM_004975.4(KCNB1):c.1139A>G (p.Tyr380Cys)	Epileptic encephalopathy, early infantile, 26| Epileptic encephalopathy	Conflicting interpretations of pathogenicity(Last reviewed: Mar 1, 2019)	criteria provided, conflicting interpretations	VCV000633630	20	49374421	633630	622058	rs1569017123	LOF	0.869	0.06	0.072
KCNB1	381	G	E	NM_004975.4(KCNB1):c.1142G>A (p.Gly381Glu)	Epileptic encephalopathy, early infantile, 26	Pathogenic(Last reviewed: May 12, 2018)	criteria provided, single submitter	VCV000576946	20	49374418	576946	572937	rs1569017114	LOF	0.87	0.048	0.081
KCNB1	382	D	H	NM_004975.4(KCNB1):c.1144G>C (p.Asp382His)	not provided	Pathogenic(Last reviewed: Aug 15, 2017)	criteria provided, single submitter	VCV000451018	20	49374416	451018	446263	rs1555889090	LOF	0.866	0.063	0.071
KCNB1	382	D	N	NM_004975.4(KCNB1):c.1144G>A (p.Asp382Asn)	Epileptic encephalopathy	Likely pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633631	20	49374416	633631	622057	rs1555889090	LOF	0.866	0.067	0.067
KCNB1	382	D	V	NM_004975.4(KCNB1):c.1145A>T (p.Asp382Val)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Nov 14, 2019)	criteria provided, single submitter	VCV000943655	20	49374415	943655	939003	rs1984248540	LOF	0.866	0.064	0.071
KCNB1	385	P	L	NM_004975.4(KCNB1):c.1154C>T (p.Pro385Leu)	not provided	Uncertain significance(Last reviewed: Jun 27, 2019)	criteria provided, single submitter	VCV001302569	20	49374406	1302569	1292850	NA	LOF	0.878	0.05	0.072

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNB1	385	P	T	NM_004975.4(KCNB1):c.1153C>A (p.Pro385Thr)	Epileptic encephalopathy, early infantile, 26| Early infantile epileptic encephalopathy with suppression bursts	Pathogenic/Likely pathogenic(Last reviewed: Dec 1, 2019)	no assertion criteria provided	VCV000417907	20	49374407	417907	404843	rs1060499592	LOF	0.883	0.047	0.071
KCNB1	394	G	R	NM_004975.4(KCNB1):c.1180G>A (p.Gly394Arg)	Epileptic encephalopathy	Likely pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633632	20	49374380	633632	622056	rs1569017073	LOF	0.883	0.044	0.073
KCNB1	395	G	R	NM_004975.4(KCNB1):c.1183G>A (p.Gly395Arg)	Epileptic encephalopathy, early infantile, 26| Intellectual disability|Epileptic encephalopathy	Conflicting interpretations of pathogenicity(Last reviewed: Aug 3, 2020)	criteria provided, conflicting interpretations	VCV000545438	20	49374377	545438	535710	rs959316981	LOF	0.862	0.055	0.083
KCNB1	397	C	F	NM_004975.4(KCNB1):c.1190G>T (p.Cys397Phe)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Oct 19, 2020)	no assertion criteria provided	VCV000984381	20	49374370	984381	972450	NA	LOF	0.873	0.055	0.072
KCNB1	397	C	Y	NM_004975.4(KCNB1):c.1190G>A (p.Cys397Tyr)	Epileptic encephalopathy, early infantile, 26|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jul 11, 2018)	criteria provided, conflicting interpretations	VCV000452347	20	49374370	452347	446262	rs1555889084	LOF	0.875	0.052	0.073
KCNB1	401	G	R	NM_004975.4(KCNB1):c.1201G>A (p.Gly401Arg)	Epileptic encephalopathy	Pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633633	20	49374359	633633	622055	rs1569017045	LOF	0.87	0.046	0.084
KCNB1	402	V	A	NM_004975.4(KCNB1):c.1205T>C (p.Val402Ala)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jun 22, 2018)	criteria provided, single submitter	VCV001031490	20	49374355	1031490	1022008	rs1984245487	LOF	0.875	0.058	0.068
KCNB1	404	V	M	NM_004975.4(KCNB1):c.1210G>A (p.Val404Met)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Nov 26, 2019)	criteria provided, single submitter	VCV000857377	20	49374350	857377	848474	rs1984244898	LOF	0.867	0.057	0.077
KCNB1	406	A	V	NM_004975.4(KCNB1):c.1217C>T (p.Ala406Val)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Feb 14, 2020)	criteria provided, multiple submitters, no conflicts	VCV000659148	20	49374343	659148	648762	rs1601071099	LOF	0.872	0.053	0.074
KCNB1	408	P	S	NM_004975.4(KCNB1):c.1222C>T (p.Pro408Ser)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Sep 26, 2019)	no assertion criteria provided	VCV000800948	20	49374338	800948	788939	rs1601071085	LOF	0.883	0.059	0.058
KCNB1	409	I	T	NM_004975.4(KCNB1):c.1226T>C (p.Ile409Thr)	Intellectual disability	Likely pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633634	20	49374334	633634	622054	rs1569017025	LOF	0.877	0.065	0.058
KCNB1	413	V	I	NM_004975.4(KCNB1):c.1237G>A (p.Val413Ile)	Epileptic encephalopathy, early infantile, 26	Conflicting interpretations of pathogenicity(Last reviewed: Sep 9, 2020)	criteria provided, conflicting interpretations	VCV000982681	20	49374323	982681	971146	rs1984243311	LOF	0.846	0.068	0.086
KCNB1	414	N	D	NM_004975.4(KCNB1):c.1240A>G (p.Asn414Asp)	Marfanoid habitus and intellectual disability	Likely pathogenic	criteria provided, single submitter	VCV000689696	20	49374320	689696	677391	rs1601071071	LOF	0.815	0.102	0.083
KCNB1	416	F	L	NM_004975.4(KCNB1):c.1248C>A (p.Phe416Leu)	Epileptic encephalopathy	Pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633635	20	49374312	633635	622053	rs1569017015	LOF	0.84	0.067	0.093
KCNB1	416	F	L	NM_004975.4(KCNB1):c.1248C>A (p.Phe416Leu)	Epileptic encephalopathy	Pathogenic(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000633635	20	49374312	633635	622053	rs1569017015	LOF	0.84	0.067	0.093
KCNB1	416	F	L	NM_004975.4(KCNB1):c.1246T>C	Epileptic encephalopathy, early infantile, 26	Likely	criteria	VCV000859281	20	49374314	859281	848473	rs1984242860	LOF	0.84	0.067	0.093

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Phe416Leu)		pathogenic(Last reviewed: Dec 24, 2019)	provided, single submitter										
KCNB1	416	F	L	NM_004975.4(KCNB1):c.1246T>C (p.Phe416Leu)	Epileptic encephalopathy, early infantile, 26	Likely pathogenic(Last reviewed: Dec 24, 2019)	criteria provided, single submitter	VCV000859281	20	49374314	859281	848473	rs1984242860	LOF	0.84	0.067	0.093
KCNB1	417	S	P	NM_004975.4(KCNB1):c.1249T>C (p.Ser417Pro)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Aug 29, 2020)	criteria provided, single submitter	VCV001052355	20	49374311	1052355	1051738	NA	LOF	0.847	0.071	0.082
KCNB1	422	E	A	NM_004975.4(KCNB1):c.1265A>C (p.Glu422Ala)	not provided	Likely pathogenic(Last reviewed: Dec 13, 2017)	criteria provided, single submitter	VCV000421307	20	49374295	421307	410767	rs1064795048	LOF	0.745	0.151	0.104
KCNB1	422	E	D	NM_004975.4(KCNB1):c.1266G>C (p.Glu422Asp)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: May 23, 2018)	criteria provided, single submitter	VCV000626121	20	49374294	626121	614475	rs755843579	LOF	0.741	0.162	0.097
KCNB1	432	R	W	NM_004975.4(KCNB1):c.1294C>T (p.Arg432Trp)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Aug 20, 2019)	criteria provided, single submitter	VCV000833960	20	49374266	833960	822171	rs752596402	Neutral	0.658	0.198	0.144
KCNB1	446	V	I	NM_004975.4(KCNB1):c.1336G>A (p.Val446Ile)	not provided|Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Sep 19, 2020)	criteria provided, single submitter	VCV000580254	20	49374224	580254	572935	rs776968332	LOF	0.785	0.12	0.095
KCNB1	448	M	I	NM_004975.4(KCNB1):c.1344G>A (p.Met448Ile)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Nov 20, 2019)	criteria provided, single submitter	VCV000833523	20	49374216	833523	822170	rs1984239766	LOF	0.692	0.109	0.198
KCNB1	449	N	D	NM_004975.4(KCNB1):c.1345A>G (p.Asn449Asp)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Sep 29, 2019)	criteria provided, single submitter	VCV000953558	20	49374215	953558	951125	rs1984239652	GOF	0.644	0.099	0.258
KCNB1	452	D	G	NM_004975.4(KCNB1):c.1355A>G (p.Asp452Gly)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jun 19, 2019)	criteria provided, single submitter	VCV000948656	20	49374205	948656	929220	rs760931963	LOF	0.692	0.109	0.199
KCNB1	456	R	G	NM_004975.4(KCNB1):c.1366C>G (p.Arg456Gly)	Epileptic encephalopathy, early infantile, 26|not provided	Uncertain significance(Last reviewed: Dec 30, 2021)	criteria provided, multiple submitters, no conflicts	VCV000934441	20	49374194	934441	939002	rs368043123	LOF	0.71	0.114	0.176
KCNB1	456	R	Q	NM_004975.4(KCNB1):c.1367G>A (p.Arg456Gln)	Epileptic encephalopathy, early infantile, 26|not provided	Benign/Likely benign(Last reviewed: Jun 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000385978	20	49374193	385978	377076	rs761082551	GOF	0.661	0.102	0.237
KCNB1	457	S	R	NM_004975.4(KCNB1):c.1371C>G (p.Ser457Arg)	not provided	Uncertain significance(Last reviewed: Apr 13, 2020)	criteria provided, single submitter	VCV001190347	20	49374189	1190347	1181870	NA	GOF	0.467	0.062	0.471
KCNB1	463	I	T	NM_004975.4(KCNB1):c.1388T>C (p.Ile463Thr)	Epileptic encephalopathy, early infantile, 26|not provided	Benign/Likely benign(Last reviewed: Dec 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000475257	20	49374172	475257	470948	rs145421532	GOF	0.671	0.104	0.225
KCNB1	465	V	I	NM_004975.4(KCNB1):c.1393G>A (p.Val465Ile)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV000568029	20	49374167	568029	571291	rs1294696493	LOF	0.699	0.111	0.19
KCNB1	466	E	D	NM_004975.4(KCNB1):c.1398G>C (p.Glu466Asp)	Intellectual disability	Likely benign(Last reviewed: Jan 1, 2019)	no assertion criteria provided	VCV000975410	20	49374162	975410	963909	rs1984236966	LOF	0.689	0.109	0.202

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNB1	482	H	N	NM_004975.4(KCNB1):c.1444C>A (p.His482Asn)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Mar 26, 2020)	criteria provided, single submitter	VCV001017488	20	49374116	1017488	1014171	rs1171133807	Neutral	0.692	0.197	0.111
KCNB1	482	H	Q	NM_004975.4(KCNB1):c.1446C>A (p.His482Gln)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Oct 21, 2019)	criteria provided, single submitter	VCV000970573	20	49374114	970573	958857	rs750701951	Neutral	0.691	0.197	0.112
KCNB1	486	N	K	NM_004975.4(KCNB1):c.1458C>G (p.Asn486Lys)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Sep 7, 2018)	criteria provided, single submitter	VCV000665812	20	49374102	665812	648761	rs1388412501	Neutral	0.655	0.22	0.124
KCNB1	502	K	N	NM_004975.4(KCNB1):c.1506G>T (p.Lys502Asn)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Oct 28, 2020)	criteria provided, single submitter	VCV001145952	20	49374054	1145952	1149512	NA	Neutral	0.65	0.228	0.123
KCNB1	510	G	V	NM_004975.4(KCNB1):c.1529G>T (p.Gly510Val)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Dec 4, 2019)	criteria provided, single submitter	VCV000864105	20	49374031	864105	848472	rs767889610	Neutral	0.668	0.214	0.118
KCNB1	517	S	L	NM_004975.4(KCNB1):c.1550C>T (p.Ser517Leu)	Epileptic encephalopathy, early infantile, 26|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 5, 2020)	criteria provided, conflicting interpretations	VCV001169000	20	49374010	1169000	1158896	NA	Neutral	0.659	0.218	0.123
KCNB1	526	V	I	NM_004975.4(KCNB1):c.1576G>A (p.Val526Ile)	Epileptic encephalopathy, early infantile, 26|not provided	Benign(Last reviewed: Nov 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000382765	20	49373984	382765	378112	rs140932636	Neutral	0.704	0.191	0.105
KCNB1	532	M	V	NM_004975.4(KCNB1):c.1594A>G (p.Met532Val)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Feb 14, 2020)	criteria provided, single submitter	VCV000870207	20	49373966	870207	858356	rs1984228484	Neutral	0.704	0.192	0.103
KCNB1	533	Y	H	NM_004975.4(KCNB1):c.1597T>C (p.Tyr533His)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Sep 25, 2019)	criteria provided, single submitter	VCV000833544	20	49373963	833544	822169	rs1984228232	Neutral	0.71	0.193	0.098
KCNB1	536	M	L	NM_004975.4(KCNB1):c.1606A>T (p.Met536Leu)	Epileptic encephalopathy, early infantile, 26|not provided	Uncertain significance(Last reviewed: Sep 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000938424	20	49373954	938424	939001	rs1984227856	Neutral	0.711	0.186	0.102
KCNB1	541	S	Y	NM_004975.4(KCNB1):c.1622C>A (p.Ser541Tyr)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Sep 19, 2019)	criteria provided, single submitter	VCV000833843	20	49373938	833843	822168	rs374460751	Neutral	0.67	0.208	0.122
KCNB1	543	P	H	NM_004975.4(KCNB1):c.1628C>A (p.Pro543His)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jun 9, 2020)	criteria provided, single submitter	VCV001062724	20	49373932	1062724	1051737	NA	Neutral	0.648	0.234	0.118
KCNB1	550	S	L	NM_004975.4(KCNB1):c.1649C>T (p.Ser550Leu)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV001164941	20	49373911	1164941	1158895	NA	Neutral	0.64	0.235	0.124
KCNB1	552	A	T	NM_004975.4(KCNB1):c.1654G>A (p.Ala552Thr)	not provided	Likely benign(Last reviewed: Aug 25, 2020)	criteria provided, single submitter	VCV001215098	20	49373906	1215098	1205081	NA	Neutral	0.637	0.226	0.137
KCNB1	552	A	V	NM_004975.4(KCNB1):c.1655C>T (p.Ala552Val)	not provided|Epileptic encephalopathy, early infantile, 26	Benign/Likely benign(Last reviewed: Jul 16, 2020)	criteria provided, multiple submitters, no conflicts	VCV000646121	20	49373905	646121	648760	rs370974891	Neutral	0.65	0.218	0.132
KCNB1	565	I	V	NM_004975.4(KCNB1):c.1693A>G (p.Ile565Val)	Epileptic encephalopathy, early infantile, 26|not provided	Conflicting interpretations of	criteria provided,	VCV000391756	20	49373867	391756	377065	rs761452916	Neutral	0.707	0.191	0.102

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						pathogenicity(Last reviewed: May 28, 2021)	conflicting interpretations										
KCNB1	569	V	I	NM_004975.4(KCNB1):c.1705G>A (p.Val569Ile)	Epileptic encephalopathy, early infantile, 26|not provided	Benign/Likely benign(Last reviewed: Dec 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000653828	20	49373855	653828	648759	rs376490656	Neutral	0.67	0.214	0.116
KCNB1	575	R	C	NM_004975.4(KCNB1):c.1723C>T (p.Arg575Cys)	Epileptic encephalopathy, early infantile, 26|not provided	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, multiple submitters, no conflicts	VCV000392965	20	49373837	392965	377063	rs947355756	Neutral	0.657	0.219	0.125
KCNB1	575	R	H	NM_004975.4(KCNB1):c.1724G>A (p.Arg575His)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV000833871	20	49373836	833871	822167	rs544069848	Neutral	0.619	0.251	0.13
KCNB1	575	R	L	NM_004975.4(KCNB1):c.1724G>T (p.Arg575Leu)	not provided	Uncertain significance(Last reviewed: Dec 17, 2019)	criteria provided, single submitter	VCV001311373	20	49373836	1311373	1299430	NA	Neutral	0.637	0.236	0.127
KCNB1	576	T	K	NM_004975.4(KCNB1):c.1727C>A (p.Thr576Lys)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV000475258	20	49373833	475258	469411	rs1355278265	Neutral	0.655	0.225	0.121
KCNB1	593	S	T	NM_004975.4(KCNB1):c.1778G>C (p.Ser593Thr)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jan 18, 2020)	criteria provided, single submitter	VCV001055120	20	49373782	1055120	1051736	NA	Neutral	0.718	0.174	0.108
KCNB1	613	P	S	NM_004975.4(KCNB1):c.1837C>T (p.Pro613Ser)	not specified|Epileptic encephalopathy, early infantile, 26|not provided	Benign(Last reviewed: Dec 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000383226	20	49373723	383226	378294	rs112735799	Neutral	0.651	0.234	0.114
KCNB1	616	T	I	NM_004975.4(KCNB1):c.1847C>T (p.Thr616Ile)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Oct 8, 2019)	criteria provided, single submitter	VCV000833862	20	49373713	833862	822166	rs2229006	Neutral	0.636	0.236	0.128
KCNB1	616	T	S	NM_004975.4(KCNB1):c.1847C>G (p.Thr616Ser)	not provided|not specified|Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Dec 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000380823	20	49373713	380823	378110	rs2229006	Neutral	0.623	0.249	0.128
KCNB1	618	G	V	NM_004975.4(KCNB1):c.1853G>T (p.Gly618Val)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Nov 20, 2019)	criteria provided, single submitter	VCV000853160	20	49373707	853160	848471	rs771932289	Neutral	0.671	0.207	0.122
KCNB1	621	A	S	NM_004975.4(KCNB1):c.1861G>T (p.Ala621Ser)	not provided|Epileptic encephalopathy, early infantile, 26	Benign/Likely benign(Last reviewed: Jul 23, 2021)	criteria provided, multiple submitters, no conflicts	VCV000708213	20	49373699	708213	728714	rs201960228	Neutral	0.638	0.232	0.131
KCNB1	621	A	V	NM_004975.4(KCNB1):c.1862C>T (p.Ala621Val)	not provided|Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Dec 30, 2019)	criteria provided, multiple submitters, no conflicts	VCV000661163	20	49373698	661163	648758	rs770069345	Neutral	0.644	0.227	0.128
KCNB1	627	R	L	NM_004975.4(KCNB1):c.1880G>T (p.Arg627Leu)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Aug 2, 2020)	criteria provided, single submitter	VCV001167105	20	49373680	1167105	1158894	NA	Neutral	0.644	0.232	0.124
KCNB1	627	R	Q	NM_004975.4(KCNB1):c.1880G>A (p.Arg627Gln)	not provided|Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: May 4, 2021)	criteria provided, multiple submitters, no	VCV000392453	20	49373680	392453	379715	rs754927664	Neutral	0.632	0.247	0.121

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNB1	627	R	W	NM_004975.4(KCNB1):c.1879C>T (p.Arg627Trp)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Oct 10, 2019)	criteria provided, single submitter	VCV000833641	20	49373681	833641	822165	rs554351105	Neutral	0.638	0.23	0.132
KCNB1	631	G	C	NM_004975.4(KCNB1):c.1891G>T (p.Gly631Cys)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: May 6, 2020)	criteria provided, single submitter	VCV000833954	20	49373669	833954	822164	rs1433173706	Neutral	0.687	0.198	0.115
KCNB1	633	S	N	NM_004975.4(KCNB1):c.1898G>A (p.Ser633Asn)	not provided	Uncertain significance(Last reviewed: Oct 27, 2017)	criteria provided, single submitter	VCV000453029	20	49373662	453029	446261	rs750807577	Neutral	0.64	0.231	0.129
KCNB1	636	R	T	NM_004975.4(KCNB1):c.1907G>C (p.Arg636Thr)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jun 21, 2020)	criteria provided, single submitter	VCV001053563	20	49373653	1053563	1051735	NA	Neutral	0.629	0.245	0.125
KCNB1	645	D	E	NM_004975.4(KCNB1):c.1935T>G (p.Asp645Glu)	not provided	Uncertain significance(Last reviewed: Sep 20, 2019)	criteria provided, single submitter	VCV001219470	20	49373625	1219470	1209448	NA	Neutral	0.649	0.225	0.126
KCNB1	646	A	D	NM_004975.4(KCNB1):c.1937C>A (p.Ala646Asp)	not provided	Uncertain significance(Last reviewed: Aug 1, 2019)	criteria provided, single submitter	VCV000872731	20	49373623	872731	860649	rs1247080453	Neutral	0.663	0.21	0.126
KCNB1	646	A	T	NM_004975.4(KCNB1):c.1936G>A (p.Ala646Thr)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: May 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000851269	20	49373624	851269	848470	rs1447311755	Neutral	0.643	0.227	0.13
KCNB1	654	I	F	NM_004975.4(KCNB1):c.1960A>T (p.Ile654Phe)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Apr 4, 2019)	criteria provided, single submitter	VCV000947824	20	49373600	947824	929219	rs1276898684	Neutral	0.689	0.198	0.113
KCNB1	655	E	K	NM_004975.4(KCNB1):c.1963G>A (p.Glu655Lys)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Dec 3, 2018)	criteria provided, single submitter	VCV000639890	20	49373597	639890	648757	rs769948178	Neutral	0.641	0.237	0.122
KCNB1	661	M	V	NM_004975.4(KCNB1):c.1981A>G (p.Met661Val)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Jun 7, 2020)	criteria provided, single submitter	VCV000953176	20	49373579	953176	951123	rs1392516083	Neutral	0.68	0.208	0.111
KCNB1	666	P	L	NM_004975.4(KCNB1):c.1997C>T (p.Pro666Leu)	not provided|Epileptic encephalopathy, early infantile, 26	Benign/Likely benign(Last reviewed: Oct 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000385555	20	49373563	385555	379714	rs201212125	Neutral	0.716	0.18	0.105
KCNB1	670	R	Q	NM_004975.4(KCNB1):c.2009G>A (p.Arg670Gln)	Epileptic encephalopathy, early infantile, 26|not provided	Likely benign(Last reviewed: Aug 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000576599	20	49373551	576599	571290	rs753575587	Neutral	0.661	0.225	0.115
KCNB1	675	N	S	NM_004975.4(KCNB1):c.2024A>G (p.Asn675Ser)	not provided	Uncertain significance(Last reviewed: Jan 1, 2021)	criteria provided, single submitter	VCV001013457	20	49373536	1013457	1001170	rs1186529246	Neutral	0.719	0.177	0.104
KCNB1	677	M	V	NM_004975.4(KCNB1):c.2029A>G (p.Met677Val)	not provided	Uncertain significance(Last reviewed: Jan 11, 2021)	criteria provided, single submitter	VCV001302405	20	49373531	1302405	1292686	NA	Neutral	0.677	0.211	0.112
KCNB1	687	V	I	NM_004975.4(KCNB1):c.2059G>A (p.Val687Ile)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Nov 23, 2020)	criteria provided, single submitter	VCV000833949	20	49373501	833949	822163	rs372263564	Neutral	0.712	0.183	0.105

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNB1	688	L	P	NM_004975.4(KCNB1):c.2063T>C (p.Leu688Pro)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Dec 19, 2018)	criteria provided, single submitter	VCV000645917	20	49373497	645917	648756	rs1601069788	Neutral	0.725	0.176	0.099
KCNB1	690	M	T	NM_004975.4(KCNB1):c.2069T>C (p.Met690Thr)	not provided	Uncertain significance(Last reviewed: Dec 3, 2019)	criteria provided, single submitter	VCV001310973	20	49373491	1310973	1300354	NA	Neutral	0.684	0.207	0.109
KCNB1	693	D	E	NM_004975.4(KCNB1):c.2079C>A (p.Asp693Glu)	not provided	Uncertain significance(Last reviewed: Nov 12, 2019)	criteria provided, single submitter	VCV001310109	20	49373481	1310109	1301960	NA	Neutral	0.695	0.199	0.106
KCNB1	697	N	K	NM_004975.4(KCNB1):c.2091C>G (p.Asn697Lys)	not provided	Uncertain significance(Last reviewed: Sep 7, 2017)	criteria provided, single submitter	VCV000451846	20	49373469	451846	446260	rs746382408	Neutral	0.634	0.242	0.124
KCNB1	698	R	Q	NM_004975.4(KCNB1):c.2093G>A (p.Arg698Gln)	Epileptic encephalopathy, early infantile, 26|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 11, 2020)	criteria provided, conflicting interpretations	VCV000938351	20	49373467	938351	939000	rs1449919002	Neutral	0.632	0.246	0.122
KCNB1	698	R	W	NM_004975.4(KCNB1):c.2092C>T (p.Arg698Trp)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jan 25, 2020)	criteria provided, single submitter	VCV001007454	20	49373468	1007454	999054	rs779379680	Neutral	0.646	0.227	0.127
KCNB1	700	S	N	NM_004975.4(KCNB1):c.2099G>A (p.Ser700Asn)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Nov 7, 2019)	criteria provided, single submitter	VCV000961786	20	49373461	961786	951122	rs1984201256	Neutral	0.651	0.228	0.121
KCNB1	702	A	E	NM_004975.4(KCNB1):c.2105C>A (p.Ala702Glu)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Mar 23, 2019)	criteria provided, single submitter	VCV000576853	20	49373455	576853	572927	rs144379782	Neutral	0.673	0.209	0.117
KCNB1	702	A	V	NM_004975.4(KCNB1):c.2105C>T (p.Ala702Val)	not provided|Epileptic encephalopathy, early infantile, 26	Benign/Likely benign(Last reviewed: Oct 26, 2020)	criteria provided, multiple submitters, no conflicts	VCV000762094	20	49373455	762094	773152	rs144379782	Neutral	0.661	0.218	0.121
KCNB1	706	A	S	NM_004975.4(KCNB1):c.2116G>T (p.Ala706Ser)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Sep 15, 2020)	criteria provided, single submitter	VCV001098263	20	49373444	1098263	1085397	NA	Neutral	0.677	0.211	0.112
KCNB1	706	A	T	NM_004975.4(KCNB1):c.2116G>A (p.Ala706Thr)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Feb 17, 2018)	criteria provided, single submitter	VCV000581484	20	49373444	581484	575115	rs779750227	Neutral	0.674	0.211	0.115
KCNB1	712	T	M	NM_004975.4(KCNB1):c.2135C>T (p.Thr712Met)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Aug 6, 2018)	criteria provided, single submitter	VCV000660973	20	49373425	660973	648755	rs775588653	Neutral	0.679	0.202	0.119
KCNB1	715	D	H	NM_004975.4(KCNB1):c.2143G>C (p.Asp715His)	Epileptic encephalopathy, early infantile, 26|not provided	Uncertain significance(Last reviewed: Jun 9, 2021)	criteria provided, multiple submitters, no conflicts	VCV000647767	20	49373417	647767	648754	rs1601069538	Neutral	0.664	0.217	0.119
KCNB1	718	V	M	NM_004975.4(KCNB1):c.2152G>A (p.Val718Met)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Dec 27, 2017)	criteria provided, single submitter	VCV000542056	20	49373408	542056	534143	rs774976039	Neutral	0.691	0.2	0.109
KCNB1	727	T	S	NM_004975.4(KCNB1):c.2180C>G (p.Thr727Ser)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: May 3, 2020)	criteria provided, single submitter	VCV001004111	20	49373380	1004111	999053	rs778296598	Neutral	0.641	0.24	0.118

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNB1	734	P	H	NM_004975.4(KCNB1):c.2201C>A (p.Pro734His)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV001006665	20	49373359	1006665	999052	rs780940515	Neutral	0.65	0.234	0.116
KCNB1	734	P	S	NM_004975.4(KCNB1):c.2200C>T (p.Pro734Ser)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: May 31, 2017)	criteria provided, single submitter	VCV000475261	20	49373360	475261	469403	rs1214997887	Neutral	0.647	0.237	0.115
KCNB1	735	P	A	NM_004975.4(KCNB1):c.2203C>G (p.Pro735Ala)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Apr 1, 2018)	criteria provided, single submitter	VCV000572894	20	49373357	572894	571285	rs267605983	Neutral	0.669	0.216	0.114
KCNB1	735	P	L	NM_004975.4(KCNB1):c.2204C>T (p.Pro735Leu)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Sep 30, 2019)	criteria provided, single submitter	VCV000964616	20	49373356	964616	958856	rs1984193843	Neutral	0.681	0.206	0.114
KCNB1	736	R	Q	NM_004975.4(KCNB1):c.2207G>A (p.Arg736Gln)	not provided	Likely benign(Last reviewed: Nov 15, 2019)	criteria provided, single submitter	VCV001191617	20	49373353	1191617	1181869	NA	Neutral	0.663	0.22	0.117
KCNB1	736	R	W	NM_004975.4(KCNB1):c.2206C>T (p.Arg736Trp)	not provided	Uncertain significance(Last reviewed: Oct 1, 2018)	criteria provided, single submitter	VCV000809256	20	49373354	809256	797992	rs754428005	Neutral	0.675	0.206	0.119
KCNB1	742	T	K	NM_004975.4(KCNB1):c.2225C>A (p.Thr742Lys)	Epileptic encephalopathy, early infantile, 26	Likely benign(Last reviewed: Jun 7, 2020)	criteria provided, single submitter	VCV001083688	20	49373335	1083688	1085396	NA	Neutral	0.65	0.229	0.12
KCNB1	745	A	V	NM_004975.4(KCNB1):c.2234C>T (p.Ala745Val)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Feb 24, 2020)	criteria provided, single submitter	VCV000845087	20	49373326	845087	848467	rs1984191107	Neutral	0.722	0.171	0.107
KCNB1	750	A	V	NM_004975.4(KCNB1):c.2249C>T (p.Ala750Val)	not provided|Epileptic encephalopathy, early infantile, 26	Conflicting interpretations of pathogenicity(Last reviewed: Jul 21, 2020)	criteria provided, conflicting interpretations	VCV000848107	20	49373311	848107	848466	rs750157634	Neutral	0.688	0.197	0.115
KCNB1	758	A	S	NM_004975.4(KCNB1):c.2272G>T (p.Ala758Ser)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Oct 23, 2020)	criteria provided, single submitter	VCV000951533	20	49373288	951533	929218	rs774577428	Neutral	0.66	0.226	0.114
KCNB1	758	A	T	NM_004975.4(KCNB1):c.2272G>A (p.Ala758Thr)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Oct 26, 2020)	criteria provided, single submitter	VCV000567109	20	49373288	567109	571278	rs774577428	Neutral	0.656	0.228	0.116
KCNB1	762	D	N	NM_004975.4(KCNB1):c.2284G>A (p.Asp762Asn)	not provided	Uncertain significance(Last reviewed: Oct 1, 2020)	criteria provided, single submitter	VCV001013456	20	49373276	1013456	1001169	rs1277329633	Neutral	0.666	0.22	0.114
KCNB1	771	D	V	NM_004975.4(KCNB1):c.2312A>T (p.Asp771Val)	not provided	Uncertain significance(Last reviewed: May 23, 2018)	criteria provided, single submitter	VCV000546484	20	49373248	546484	537003	rs1304300872	Neutral	0.637	0.241	0.122
KCNB1	772	S	Y	NM_004975.4(KCNB1):c.2315C>A (p.Ser772Tyr)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Nov 3, 2021)	criteria provided, single submitter	VCV001319065	20	49373245	1319065	1309318	NA	Neutral	0.645	0.229	0.126
KCNB1	774	P	T	NM_004975.4(KCNB1):c.2320C>A (p.Pro774Thr)	Epileptic encephalopathy, early infantile, 26|not provided	Benign(Last reviewed: Nov 21, 2020)	criteria provided, single submitter	VCV000568166	20	49373240	568166	573568	rs748597382	Neutral	0.644	0.245	0.111
KCNB1	775	P	A	NM_004975.4(KCNB1):c.2323C>G (p.Pro775Ala)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Oct 1, 2020)	criteria provided, single submitter	VCV000947951	20	49373237	947951	929217	rs142461221	Neutral	0.652	0.24	0.108

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNB1	775	P	H	NM_004975.4(KCNB1):c.2324C>A (p.Pro775His)	Epileptic encephalopathy, early infantile, 26|not provided	Uncertain significance(Last reviewed: Jun 26, 2018)	criteria provided, multiple submitters, no conflicts	VCV000430040	20	49373236	430040	422322	rs530927636	Neutral	0.65	0.237	0.113
KCNB1	775	P	R	NM_004975.4(KCNB1):c.2324C>G (p.Pro775Arg)	Epileptic encephalopathy, early infantile, 26|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 8, 2020)	criteria provided, conflicting interpretations	VCV000425270	20	49373236	425270	413604	rs530927636	Neutral	0.666	0.221	0.113
KCNB1	776	K	R	NM_004975.4(KCNB1):c.2327A>G (p.Lys776Arg)	Epileptic encephalopathy, early infantile, 26|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 21, 2020)	criteria provided, conflicting interpretations	VCV000578493	20	49373233	578493	571276	rs756982426	Neutral	0.663	0.22	0.117
KCNB1	777	S	G	NM_004975.4(KCNB1):c.2329A>G (p.Ser777Gly)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Oct 26, 2020)	criteria provided, single submitter	VCV001009790	20	49373231	1009790	999051	rs1984182896	Neutral	0.659	0.227	0.114
KCNB1	781	S	N	NM_004975.4(KCNB1):c.2342G>A (p.Ser781Asn)	not provided	Likely benign(Last reviewed: Apr 1, 2020)	criteria provided, single submitter	VCV000932380	20	49373218	932380	920971	rs1271782193	Neutral	0.639	0.234	0.127
KCNB1	782	T	I	NM_004975.4(KCNB1):c.2345C>T (p.Thr782Ile)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jul 9, 2020)	criteria provided, single submitter	VCV001022623	20	49373215	1022623	1014170	rs751639377	Neutral	0.649	0.232	0.119
KCNB1	782	T	S	NM_004975.4(KCNB1):c.2345C>G (p.Thr782Ser)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Oct 21, 2020)	criteria provided, single submitter	VCV001002068	20	49373215	1002068	999050	rs751639377	Neutral	0.636	0.245	0.119
KCNB1	784	P	L	NM_004975.4(KCNB1):c.2351C>T (p.Pro784Leu)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: May 2, 2018)	criteria provided, single submitter	VCV000576386	20	49373209	576386	575114	rs766596568	Neutral	0.663	0.229	0.108
KCNB1	788	T	K	NM_004975.4(KCNB1):c.2363C>A (p.Thr788Lys)	Epileptic encephalopathy, early infantile, 26|not provided	Benign(Last reviewed: Aug 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000542062	20	49373197	542062	533646	rs140932985	Neutral	0.656	0.227	0.118
KCNB1	788	T	M	NM_004975.4(KCNB1):c.2363C>T (p.Thr788Met)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Dec 17, 2019)	criteria provided, single submitter	VCV000833692	20	49373197	833692	822162	rs140932985	Neutral	0.646	0.237	0.117
KCNB1	792	S	L	NM_004975.4(KCNB1):c.2375C>T (p.Ser792Leu)	not provided	Likely benign(Last reviewed: Dec 18, 2020)	criteria provided, single submitter	VCV001198556	20	49373185	1198556	1188869	NA	Neutral	0.652	0.23	0.118
KCNB1	798	E	Q	NM_004975.4(KCNB1):c.2392G>C (p.Glu798Gln)	Epileptic encephalopathy, early infantile, 26|not provided	Uncertain significance(Last reviewed: Jun 26, 2018)	criteria provided, multiple submitters, no conflicts	VCV000430043	20	49373168	430043	422321	rs1131691752	Neutral	0.655	0.224	0.121
KCNB1	804	T	A	NM_004975.4(KCNB1):c.2410A>G (p.Thr804Ala)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Aug 17, 2019)	criteria provided, single submitter	VCV000957516	20	49373150	957516	951121	rs1262864745	Neutral	0.649	0.234	0.117
KCNB1	804	T	I	NM_004975.4(KCNB1):c.2411C>T (p.Thr804Ile)	not specified	Uncertain significance(Last reviewed: Nov 30, 2016)	criteria provided, single submitter	VCV000373635	20	49373149	373635	360467	rs1049874069	Neutral	0.657	0.224	0.119
KCNB1	806	P	L	NM_004975.4(KCNB1):c.2417C>T (p.Pro806Leu)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Nov 5, 2019)	criteria provided, single submitter	VCV000833869	20	49373143	833869	822161	rs1249019020	Neutral	0.706	0.195	0.1
KCNB1	806	P	S	NM_004975.4(KCNB1):c.2416C>T	Epileptic encephalopathy, early infantile, 26	Uncertain	criteria	VCV000940834	20	49373144	940834	938999	rs1471856482	Neutral	0.704	0.2	0.097

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Pro806Ser)		significance(Last reviewed: Sep 18, 2019)	provided, single submitter										
KCNB1	814	I	M	NM_004975.4(KCNB1):c.2442T>G (p.Ile814Met)	Seizures|Epileptic encephalopathy, early infantile, 26|not provided	Benign(Last reviewed: Nov 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000392430	20	49373118	392430	378077	rs565025643	Neutral	0.739	0.168	0.093
KCNB1	819	A	T	NM_004975.4(KCNB1):c.2455G>A (p.Ala819Thr)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Oct 30, 2019)	criteria provided, single submitter	VCV000833994	20	49373105	833994	822160	rs373423740	Neutral	0.65	0.229	0.121
KCNB1	819	A	V	NM_004975.4(KCNB1):c.2456C>T (p.Ala819Val)	Epileptic encephalopathy, early infantile, 26|not provided	Likely benign(Last reviewed: Aug 25, 2018)	criteria provided, single submitter	VCV000740544	20	49373104	740544	757587	rs777645076	Neutral	0.66	0.222	0.118
KCNB1	820	L	F	NM_004975.4(KCNB1):c.2460G>T (p.Leu820Phe)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV001006183	20	49373100	1006183	999049	rs1984174923	Neutral	0.692	0.199	0.109
KCNB1	824	G	D	NM_004975.4(KCNB1):c.2471G>A (p.Gly824Asp)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Dec 23, 2019)	criteria provided, single submitter	VCV000833699	20	49373089	833699	822159	rs780469478	Neutral	0.678	0.206	0.115
KCNB1	825	P	S	NM_004975.4(KCNB1):c.2473C>T (p.Pro825Ser)	Epileptic encephalopathy, early infantile, 26|not provided|not specified	Benign(Last reviewed: Dec 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000382142	20	49373087	382142	378277	rs34467662	Neutral	0.654	0.227	0.119
KCNB1	825	P	S	NM_004975.4(KCNB1):c.2473C>T (p.Pro825Ser)	Epileptic encephalopathy, early infantile, 26|not provided|not specified	Benign(Last reviewed: Dec 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000382142	20	49373087	382142	378277	rs34467662	Neutral	0.654	0.227	0.119
KCNB1	825	P	S	NM_004975.4(KCNB1):c.2472_2473delinsTT (p.Pro825Ser)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jul 10, 2020)	criteria provided, single submitter	VCV001062047	20	49373087 - 49373088	1062047	1051734	NA	Neutral	0.654	0.227	0.119
KCNB1	825	P	S	NM_004975.4(KCNB1):c.2472_2473delinsTT (p.Pro825Ser)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jul 10, 2020)	criteria provided, single submitter	VCV001062047	20	49373087 - 49373088	1062047	1051734	NA	Neutral	0.654	0.227	0.119
KCNB1	838	S	P	NM_004975.4(KCNB1):c.2512T>C (p.Ser838Pro)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Jan 11, 2020)	criteria provided, single submitter	VCV001020008	20	49373048	1020008	1014169	rs1984172893	Neutral	0.716	0.18	0.104
KCNB1	839	P	L	NM_004975.4(KCNB1):c.2516C>T (p.Pro839Leu)	Epileptic encephalopathy, early infantile, 26|not specified	Likely benign(Last reviewed: Jan 11, 2018)	criteria provided, multiple submitters, no conflicts	VCV000475262	20	49373044	475262	471425	rs761070687	Neutral	0.722	0.182	0.096
KCNB1	839	P	T	NM_004975.4(KCNB1):c.2515C>A (p.Pro839Thr)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Sep 18, 2020)	criteria provided, single submitter	VCV001045643	20	49373045	1045643	1034742	rs1984172647	Neutral	0.718	0.187	0.095
KCNB1	841	V	I	NM_004975.4(KCNB1):c.2521G>A (p.Val841Ile)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Oct 15, 2020)	criteria provided, single submitter	VCV001170471	20	49373039	1170471	1158892	NA	Neutral	0.736	0.169	0.094
KCNB1	842	R	C	NM_004975.4(KCNB1):c.2524C>T (p.Arg842Cys)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV000998443	20	49373036	998443	999048	rs758964585	Neutral	0.68	0.21	0.11
KCNB1	842	R	H	NM_004975.4(KCNB1):c.2525G>A (p.Arg842His)	Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Oct 5,	criteria provided, single	VCV001166389	20	49373035	1166389	1158891	NA	Neutral	0.647	0.242	0.111

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)	submitter										
KCNB1	846	G	V	NM_004975.4(KCNB1):c.2537G>T (p.Gly846Val)	Epileptic encephalopathy, early infantile, 26	Uncertain significance(Last reviewed: Sep 17, 2019)	criteria provided, single submitter	VCV000958081	20	49373023	958081	951120	rs1386449842	Neutral	0.692	0.199	0.108
KCNB1	853	T	I	NM_004975.4(KCNB1):c.2558C>T (p.Thr853Ile)	not provided	Uncertain significance(Last reviewed: Jan 1, 2020)	criteria provided, single submitter	VCV000872730	20	49373002	872730	860648	rs1001982708	Neutral	0.699	0.194	0.107
KCNB1	857	S	N	NM_004975.4(KCNB1):c.2570G>A (p.Ser857Asn)	not provided|not specified|Epileptic encephalopathy, early infantile, 26	Benign(Last reviewed: Dec 6, 2020)	criteria provided, multiple submitters, no conflicts	VCV000380824	20	49372990	380824	378076	rs34280195	Neutral	0.68	0.211	0.109
KCNC1	4	G	E	NM_001112741.2(KCNC1):c.11G>A (p.Gly4Glu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jun 22, 2019)	criteria provided, single submitter	VCV000931566	11	17736013	931566	919344	rs1848761561	Neutral	0.662	0.172	0.166
KCNC1	23	R	H	NM_001112741.2(KCNC1):c.68G>A (p.Arg23His)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Aug 25, 2019)	criteria provided, single submitter	VCV000938579	11	17736070	938579	935449	rs1158193192	LOF	0.799	0.066	0.135
KCNC1	31	G	R	NM_001112741.2(KCNC1):c.91G>C (p.Gly31Arg)	not provided	Uncertain significance(Last reviewed: Sep 28, 2019)	criteria provided, single submitter	VCV001308752	11	17736093	1308752	1300780	NA	LOF	0.773	0.097	0.13
KCNC1	41	D	N	NM_001112741.2(KCNC1):c.121G>A (p.Asp41Asn)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Apr 10, 2018)	criteria provided, single submitter	VCV000567485	11	17736123	567485	564515	rs1192156090	Neutral	0.693	0.145	0.162
KCNC1	44	S	N	NM_001112741.2(KCNC1):c.131G>A (p.Ser44Asn)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Aug 30, 2018)	criteria provided, single submitter	VCV000653011	11	17736133	653011	639846	rs773455390	LOF	0.79	0.119	0.091
KCNC1	45	H	Y	NM_001112741.2(KCNC1):c.133C>T (p.His45Tyr)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV000864788	11	17736135	864788	838159	rs1470613305	Neutral	0.684	0.146	0.17
KCNC1	46	F	L	NM_001112741.2(KCNC1):c.138C>G (p.Phe46Leu)	not specified|Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Oct 25, 2019)	criteria provided, multiple submitters, no conflicts	VCV000928891	11	17736140	928891	917083	rs1848763448	LOF	0.748	0.134	0.119
KCNC1	50	P	S	NM_001112741.2(KCNC1):c.148C>T (p.Pro50Ser)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV001036073	11	17736150	1036073	1030143	rs763076681	LOF	0.72	0.138	0.142
KCNC1	59	R	H	NM_001112741.2(KCNC1):c.176G>A (p.Arg59His)	not provided	Uncertain significance(Last reviewed: Mar 19, 2019)	criteria provided, single submitter	VCV001302178	11	17736178	1302178	1292460	NA	LOF	0.781	0.114	0.105
KCNC1	72	R	L	NM_001112741.2(KCNC1):c.215G>T (p.Arg72Leu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 13, 2020)	criteria provided, single submitter	VCV001031095	11	17736217	1031095	1017430	rs1848764021	LOF	0.831	0.055	0.114
KCNC1	75	K	E	NM_001112741.2(KCNC1):c.223A>G (p.Lys75Glu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Apr 23, 2018)	criteria provided, single submitter	VCV000582490	11	17736225	582490	565569	rs1565152713	LOF	0.775	0.121	0.104
KCNC1	94	W	C	NM_001112741.2(KCNC1):c.282G>T (p.Trp94Cys)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last	criteria provided, single	VCV000982618	11	17736284	982618	970931	rs1848764853	LOF	0.845	0.074	0.082

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Jan 1, 2019)	submitter										
KCNC1	131	A	D	NM_001112741.2(KCNC1):c.392C>A (p.Ala131Asp)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV001024248	11	17736394	1024248	1009600	rs1848765901	Neutral	0.66	0.188	0.152
KCNC1	132	D	N	NM_001112741.2(KCNC1):c.394G>A (p.Asp132Asn)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jan 16, 2018)	criteria provided, single submitter	VCV000570244	11	17736396	570244	567099	rs770445113	Neutral	0.558	0.215	0.228
KCNC1	137	D	N	NM_001112741.2(KCNC1):c.409G>A (p.Asp137Asn)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: May 12, 2019)	criteria provided, single submitter	VCV000847604	11	17736411	847604	838160	rs1848766389	Neutral	0.617	0.208	0.175
KCNC1	141	D	Y	NM_001112741.2(KCNC1):c.421G>T (p.Asp141Tyr)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, single submitter	VCV000661955	11	17736423	661955	639847	rs376363769	Neutral	0.627	0.203	0.17
KCNC1	143	G	D	NM_001112741.2(KCNC1):c.428G>A (p.Gly143Asp)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jun 30, 2020)	criteria provided, single submitter	VCV001014282	11	17736430	1014282	1009601	rs1848766892	Neutral	0.626	0.205	0.169
KCNC1	145	G	R	NM_001112741.2(KCNC1):c.433G>C (p.Gly145Arg)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jul 26, 2017)	criteria provided, single submitter	VCV000475354	11	17736435	475354	461316	rs759773311	Neutral	0.639	0.191	0.17
KCNC1	147	D	E	NM_001112741.2(KCNC1):c.441C>G (p.Asp147Glu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 10, 2020)	criteria provided, single submitter	VCV000852716	11	17736443	852716	838161	rs1279299321	Neutral	0.608	0.196	0.197
KCNC1	148	E	K	NM_001112741.2(KCNC1):c.442G>A (p.Glu148Lys)	not provided	Uncertain significance(Last reviewed: Nov 21, 2019)	criteria provided, single submitter	VCV001310491	11	17736444	1310491	1301195	NA	Neutral	0.663	0.188	0.149
KCNC1	162	D	E	NM_001112741.2(KCNC1):c.486T>A (p.Asp162Glu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Dec 18, 2019)	criteria provided, single submitter	VCV000930835	11	17736488	930835	919345	rs1436435735	Neutral	0.678	0.178	0.144
KCNC1	164	R	W	NM_001112741.2(KCNC1):c.490C>T (p.Arg164Trp)	Epilepsy, progressive myoclonic 7	Likely pathogenic(Last reviewed: Oct 2, 2021)	criteria provided, single submitter	VCV001320194	11	17736492	1320194	1310439	NA	Neutral	0.663	0.181	0.156
KCNC1	165	P	L	NM_001112741.2(KCNC1):c.494C>T (p.Pro165Leu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Dec 3, 2018)	criteria provided, single submitter	VCV000475356	11	17736496	475356	460935	rs749790557	Neutral	0.643	0.195	0.162
KCNC1	166	G	D	NM_001112741.2(KCNC1):c.497G>A (p.Gly166Asp)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Apr 25, 2019)	criteria provided, single submitter	VCV000843064	11	17736499	843064	838162	rs1848768219	LOF	0.69	0.137	0.173
KCNC1	167	G	C	NM_001112741.2(KCNC1):c.499G>T (p.Gly167Cys)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 12, 2020)	criteria provided, single submitter	VCV000567985	11	17736501	567985	567101	rs1274575581	LOF	0.733	0.122	0.145
KCNC1	167	G	D	NM_001112741.2(KCNC1):c.500G>A (p.Gly167Asp)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Aug 11, 2020)	criteria provided, single submitter	VCV001055648	11	17736502	1055648	1047130	NA	LOF	0.736	0.109	0.155
KCNC1	171	R	C	NM_001112741.2(KCNC1):c.511C>T (p.Arg171Cys)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Apr 22,	criteria provided, single submitter	VCV001020341	11	17736513	1020341	1009602	rs1286632713	LOF	0.754	0.117	0.129

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)											
KCNC1	175	R	C	NM_001112741.2(KCNC1):c.523C>T (p.Arg175Cys)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV000650855	11	17736525	650855	639848	rs1197911307	LOF	0.799	0.077	0.124
KCNC1	183	P	R	NM_001112741.2(KCNC1):c.548C>G (p.Pro183Arg)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Oct 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000849585	11	17736550	849585	838163	rs1848768884	LOF	0.818	0.068	0.114
KCNC1	185	S	L	NM_001112741.2(KCNC1):c.554C>T (p.Ser185Leu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Apr 18, 2019)	criteria provided, single submitter	VCV000575538	11	17736556	575538	565572	rs1565152898	LOF	0.836	0.055	0.109
KCNC1	188	Y	C	NM_001112741.2(KCNC1):c.563A>G (p.Tyr188Cys)	Seizures	Uncertain significance(Last reviewed: Sep 16, 2016)	criteria provided, single submitter	VCV000588275	11	17736565	588275	579707	rs1027286161	LOF	0.817	0.085	0.098
KCNC1	204	I	T	NM_001112741.2(KCNC1):c.611T>C (p.Ile204Thr)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jan 30, 2018)	criteria provided, single submitter	VCV000542107	11	17771705	542107	526059	rs1554991304	LOF	0.881	0.049	0.07
KCNC1	208	C	Y	NM_001112741.2(KCNC1):c.623G>A (p.Cys208Tyr)	not provided	Likely pathogenic(Last reviewed: Oct 23, 2020)	criteria provided, single submitter	VCV000987044	11	17771717	987044	975309	rs1849231544	LOF	0.858	0.044	0.098
KCNC1	226	N	I	NM_001112741.2(KCNC1):c.677A>T (p.Asn226Ile)	not provided|Epilepsy, progressive myoclonic 7| Seizures	Likely benign(Last reviewed: Sep 16, 2020)	criteria provided, multiple submitters, no conflicts	VCV000578014	11	17771771	578014	567113	rs73424032	LOF	0.793	0.118	0.089
KCNC1	227	V	I	NM_001112741.2(KCNC1):c.679G>A (p.Val227Ile)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jan 31, 2019)	criteria provided, single submitter	VCV000844665	11	17771773	844665	838164	rs1590106427	LOF	0.747	0.136	0.117
KCNC1	228	R	H	NM_001112741.2(KCNC1):c.683G>A (p.Arg228His)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Oct 10, 2019)	criteria provided, single submitter	VCV000845316	11	17771777	845316	838165	rs200873319	LOF	0.732	0.148	0.119
KCNC1	228	R	L	NM_001112741.2(KCNC1):c.683G>T (p.Arg228Leu)	Epilepsy, progressive myoclonic 7|not provided	Benign/Likely benign(Last reviewed: Nov 19, 2020)	criteria provided, multiple submitters, no conflicts	VCV000475358	11	17771777	475358	460938	rs200873319	LOF	0.739	0.143	0.118
KCNC1	231	T	A	NM_001112741.2(KCNC1):c.691A>G (p.Thr231Ala)	Epilepsy, progressive myoclonic 7	Likely pathogenic(Last reviewed: Mar 29, 2018)	criteria provided, single submitter	VCV000568146	11	17771785	568146	570411	rs1565162623	LOF	0.791	0.078	0.131
KCNC1	231	T	M	NM_001112741.2(KCNC1):c.692C>T (p.Thr231Met)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Dec 28, 2017)	criteria provided, single submitter	VCV000542103	11	17771786	542103	526150	rs1554991314	LOF	0.788	0.077	0.135
KCNC1	232	Q	H	NM_001112741.2(KCNC1):c.696A>C (p.Gln232His)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Oct 19, 2017)	criteria provided, single submitter	VCV000475359	11	17771790	475359	460939	rs775885483	LOF	0.795	0.118	0.087
KCNC1	234	R	H	NM_001112741.2(KCNC1):c.701G>A (p.Arg234His)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Mar 15, 2018)	criteria provided, single submitter	VCV000542109	11	17771795	542109	526508	rs763794003	LOF	0.794	0.118	0.088
KCNC1	236	Y	H	NM_001112741.2(KCNC1):c.706T>C	Epilepsy, progressive myoclonic 7	Uncertain	criteria	VCV001051707	11	17771800	1051707	1047131	NA	LOF	0.765	0.118	0.118

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Tyr236His)		significance(Last reviewed: Feb 27, 2020)	provided, single submitter										
KCNC1	237	R	Q	NM_001112741.2(KCNC1):c.710G>A (p.Arg237Gln)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, single submitter	VCV001045848	11	17771804	1045848	1030145	rs200015927	Neutral	0.714	0.157	0.129
KCNC1	237	R	W	NM_001112741.2(KCNC1):c.709C>T (p.Arg237Trp)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 18, 2020)	criteria provided, single submitter	VCV000578002	11	17771803	578002	570413	rs150993414	LOF	0.725	0.152	0.123
KCNC1	248	I	T	NM_001112741.2(KCNC1):c.743T>C (p.Ile248Thr)	not provided	Uncertain significance(Last reviewed: Jul 1, 2018)	criteria provided, single submitter	VCV000623766	11	17771837	623766	612903	rs1565162643	LOF	0.877	0.053	0.07
KCNC1	259	E	K	NM_001112741.2(KCNC1):c.775G>A (p.Glu259Lys)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Apr 18, 2019)	criteria provided, single submitter	VCV000944058	11	17771869	944058	926164	rs1849234046	LOF	0.865	0.046	0.089
KCNC1	263	R	H	NM_001112741.2(KCNC1):c.788G>A (p.Arg263His)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jul 12, 2018)	criteria provided, single submitter	VCV000570230	11	17771882	570230	564518	rs775163829	LOF	0.885	0.035	0.08
KCNC1	269	N	S	NM_001112741.2(KCNC1):c.806A>G (p.Asn269Ser)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 6, 2020)	criteria provided, single submitter	VCV001058747	11	17771900	1058747	1047132	NA	LOF	0.76	0.083	0.157
KCNC1	283	F	L	NM_001112741.2(KCNC1):c.847T>C (p.Phe283Leu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Dec 16, 2020)	criteria provided, single submitter	VCV001333779	11	17771941	1333779	1324549	NA	LOF	0.835	0.079	0.086
KCNC1	287	L	M	NM_001112741.2(KCNC1):c.859C>A (p.Leu287Met)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Oct 29, 2020)	criteria provided, single submitter	VCV001035028	11	17771953	1035028	1030146	rs1849234887	LOF	0.848	0.071	0.082
KCNC1	306	V	L	NM_001112741.2(KCNC1):c.916G>T (p.Val306Leu)	not provided	Uncertain significance(Last reviewed: May 25, 2017)	criteria provided, single submitter	VCV000430428	11	17772010	430428	421844	rs369830206	LOF	0.858	0.055	0.087
KCNC1	316	V	M	NM_001112741.2(KCNC1):c.946G>A (p.Val316Met)	not provided	Uncertain significance(Last reviewed: Apr 5, 2021)	criteria provided, single submitter	VCV001314562	11	17772040	1314562	1304823	NA	LOF	0.87	0.048	0.082
KCNC1	317	R	H	NM_001112741.2(KCNC1):c.950G>A (p.Arg317His)	Inborn genetic diseases	Uncertain significance(Last reviewed: Oct 16, 2017)	criteria provided, single submitter	VCV000522132	11	17772044	522132	511905	rs1554991338	LOF	0.867	0.033	0.1
KCNC1	320	R	H	NM_001112741.2(KCNC1):c.959G>A (p.Arg320His)	Epilepsy, progressive myoclonic 7	Pathogenic(Last reviewed: Jan 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000162519	11	17772053	162519	172192	rs727502818	LOF	0.852	0.031	0.116
KCNC1	365	Y	C	NM_001112741.2(KCNC1):c.1094A>G (p.Tyr365Cys)	not provided	Uncertain significance(Last reviewed: Mar 14, 2019)	criteria provided, single submitter	VCV001307988	11	17772188	1307988	1297935	NA	LOF	0.879	0.049	0.072
KCNC1	385	K	T	NM_001112741.2(KCNC1):c.1154A>C (p.Lys385Thr)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jul 5, 2017)	criteria provided, single submitter	VCV000475346	11	17772248	475346	461324	rs1554991360	LOF	0.79	0.078	0.132
KCNC1	408	P	L	NM_001112741.2(KCNC1):c.1223C>T (p.Pro408Leu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 24,	criteria provided, single submitter	VCV000961788	11	17772317	961788	947378	rs1355766508	LOF	0.86	0.049	0.092

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)											
KCNC1	418	A	S	NM_001112741.2(KCNC1):c.1252G>T (p.Ala418Ser)	Epilepsy, progressive myoclonic 7|not provided	Uncertain significance(Last reviewed: Sep 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV001036828	11	17772346	1036828	1030147	rs1849239426	LOF	0.881	0.045	0.074
KCNC1	421	A	T	NM_001112741.2(KCNC1):c.1261G>A (p.Ala421Thr)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Apr 5, 2018)	criteria provided, single submitter	VCV000566539	11	17772355	566539	564523	rs1565162836	LOF	0.884	0.037	0.079
KCNC1	421	A	V	NM_001112741.2(KCNC1):c.1262C>T (p.Ala421Val)	Epilepsy, progressive myoclonic 7|not provided	Pathogenic/Likely pathogenic(Last reviewed: Sep 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000488536	11	17772356	488536	481404	rs1554991378	LOF	0.869	0.042	0.089
KCNC1	425	V	E	NM_001112741.2(KCNC1):c.1274T>A (p.Val425Glu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Nov 3, 2021)	criteria provided, single submitter	VCV001319331	11	17772368	1319331	1309584	NA	LOF	0.891	0.044	0.065
KCNC1	425	V	M	NM_001112741.2(KCNC1):c.1273G>A (p.Val425Met)	not provided	Likely pathogenic(Last reviewed: Apr 5, 2017)	criteria provided, single submitter	VCV000426760	11	17772367	426760	415258	rs1085307785	LOF	0.864	0.049	0.086
KCNC1	432	V	L	NM_001112741.2(KCNC1):c.1294G>C (p.Val432Leu)	Epilepsy, progressive myoclonic 7	Likely pathogenic(Last reviewed: Oct 19, 2018)	criteria provided, single submitter	VCV000813805	11	17772388	813805	802001	NA	LOF	0.853	0.056	0.091
KCNC1	432	V	M	NM_001112741.2(KCNC1):c.1294G>A (p.Val432Met)	Inborn genetic diseases|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jun 29, 2021)	criteria provided, conflicting interpretations	VCV000392871	11	17772388	392871	373997	rs1057524670	LOF	0.863	0.05	0.087
KCNC1	434	V	L	NM_001112741.2(KCNC1):c.1300G>C (p.Val434Leu)	Epilepsy, progressive myoclonic 7|not provided	Uncertain significance(Last reviewed: May 28, 2019)	criteria provided, multiple submitters, no conflicts	VCV000835488	11	17772394	835488	838168	rs1849240404	LOF	0.853	0.056	0.091
KCNC1	441	M	L	NM_001112741.2(KCNC1):c.1321A>T (p.Met441Leu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Apr 9, 2020)	criteria provided, single submitter	VCV000848761	11	17772415	848761	838169	rs1849240546	LOF	0.835	0.063	0.102
KCNC1	462	R	W	NM_001112741.2(KCNC1):c.1384C>T (p.Arg462Trp)	not provided	Uncertain significance(Last reviewed: Oct 30, 2020)	criteria provided, single submitter	VCV001256593	11	17772478	1256593	1246522	NA	Neutral	0.593	0.24	0.167
KCNC1	464	P	L	NM_001112741.2(KCNC1):c.1391C>T (p.Pro464Leu)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV001026165	11	17772485	1026165	1009603	rs899899700	Neutral	0.56	0.252	0.188
KCNC1	467	G	R	NM_001112741.2(KCNC1):c.1399G>C (p.Gly467Arg)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jul 27, 2018)	criteria provided, single submitter	VCV000645528	11	17772493	645528	639849	rs1477026732	Neutral	0.583	0.202	0.215
KCNC1	470	N	S	NM_001112741.2(KCNC1):c.1409A>G (p.Asn470Ser)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Aug 14, 2019)	criteria provided, single submitter	VCV000931673	11	17772503	931673	919346	rs143588903	GOF	0.545	0.202	0.253
KCNC1	474	S	C	NM_001112741.2(KCNC1):c.1421C>G (p.Ser474Cys)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Feb 1, 2020)	criteria provided, single submitter	VCV000851860	11	17772515	851860	838170	rs1554991422	Neutral	0.614	0.21	0.176

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNC1	474	S	Y	NM_001112741.2(KCNC1):c.1421C>A (p.Ser474Tyr)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Mar 21, 2017)	criteria provided, single submitter	VCV000475349	11	17772515	475349	461328	rs1554991422	Neutral	0.614	0.19	0.196
KCNC1	475	V	I	NM_001112741.2(KCNC1):c.1423G>A (p.Val475Ile)	not provided	Uncertain significance(Last reviewed: Aug 13, 2019)	criteria provided, single submitter	VCV001307853	11	17772517	1307853	1297800	NA	Neutral	0.669	0.195	0.136
KCNC1	476	V	I	NM_001112741.2(KCNC1):c.1426G>A (p.Val476Ile)	Seizures|Epilepsy, progressive myoclonic 7|not specified|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Sep 17, 2020)	criteria provided, conflicting interpretations	VCV000218659	11	17772520	218659	215431	rs146311746	Neutral	0.661	0.201	0.138
KCNC1	477	N	S	NM_001112741.2(KCNC1):c.1430A>G (p.Asn477Ser)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV000850956	11	17772524	850956	838171	rs1849241725	Neutral	0.519	0.257	0.224
KCNC1	478	S	A	NM_001112741.2(KCNC1):c.1432T>G (p.Ser478Ala)	not provided	Uncertain significance(Last reviewed: Sep 17, 2019)	criteria provided, single submitter	VCV001312325	11	17772526	1312325	1301707	NA	Neutral	0.568	0.23	0.202
KCNC1	480	H	N	NM_001112741.2(KCNC1):c.1438C>A (p.His480Asn)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Sep 5, 2017)	criteria provided, single submitter	VCV000542106	11	17772532	542106	526065	rs1554991425	Neutral	0.553	0.24	0.207
KCNC1	484	Q	E	NM_001112741.2(KCNC1):c.1450C>G (p.Gln484Glu)	Epilepsy, progressive myoclonic 7|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jul 11, 2019)	criteria provided, conflicting interpretations	VCV000475351	11	17772544	475351	461345	rs376505218	Neutral	0.553	0.237	0.21
KCNC1	484	Q	R	NM_001112741.2(KCNC1):c.1451A>G (p.Gln484Arg)	Epilepsy, progressive myoclonic 7	Benign(Last reviewed: Dec 31, 2019)	criteria provided, single submitter	VCV000708688	11	17772545	708688	724342	rs770586397	Neutral	0.536	0.231	0.234
KCNC1	502	D	G	NM_001112741.2(KCNC1):c.1505A>G (p.Asp502Gly)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Sep 14, 2018)	criteria provided, single submitter	VCV000650375	11	17779456	650375	639850	rs1590109843	Neutral	0.66	0.208	0.132
KCNC1	502	G	N	NM_001112741.2(KCNC1):c.1504G>A (p.Asp502Asn)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Sep 21, 2019)	criteria provided, single submitter	VCV000960153	11	17772598	960153	947379	rs1849242580	Neutral	0.661	0.2	0.139
KCNC1	510	A	V	NM_001112741.2(KCNC1):c.1529C>T (p.Ala510Val)	Epilepsy, progressive myoclonic 7	Uncertain significance(Last reviewed: Jan 7, 2020)	criteria provided, single submitter	VCV000946569	11	17779480	946569	926165	rs1849322367	Neutral	0.631	0.208	0.161
KCNC2	382	F	C	NM_139137.4(KCNC2):c.1145T>G (p.Phe382Cys)	not provided	Uncertain significance(Last reviewed: Feb 1, 2020)	criteria provided, single submitter	VCV000916302	12	75050860	916302	904574	rs1881091856	LOF	0.861	0.055	0.084
KCNC2	469	V	L	NM_139137.4(KCNC2):c.1405G>T (p.Val469Leu)	KCNC2-related disorder	Uncertain significance(Last reviewed: Apr 7, 2021)	criteria provided, single submitter	VCV001172543	12	75050600	1172543	1161647	NA	LOF	0.858	0.051	0.09
KCNC2	470	P	S	NM_139137.4(KCNC2):c.1408C>T (p.Pro470Ser)	not provided	Uncertain significance(Last reviewed: Sep 1, 2019)	criteria provided, single submitter	VCV000872705	12	75050597	872705	860011	rs1881066861	LOF	0.883	0.033	0.084
KCNC3	8	S	W	NM_004977.3(KCNC3):c.23C>G (p.Ser8Trp)	not provided|not specified	Conflicting interpretations of pathogenicity(Last reviewed: Apr 14, 2017)	criteria provided, conflicting interpretations	VCV000447623	19	50329060	447623	442247	rs761806977	Neutral	0.653	0.2	0.147

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNC3	16	A	T	NM_004977.3(KCNC3):c.46G>A (p.Ala16Thr)	not provided	Likely benign(Last reviewed: Jul 31, 2016)	criteria provided, single submitter	VCV000374785	19	50329037	374785	361671	rs1057519247	Neutral	0.622	0.23	0.148
KCNC3	24	P	L	NM_004977.3(KCNC3):c.71C>T (p.Pro24Leu)	not provided	Conflicting interpretations of pathogenicity(Last reviewed: Sep 19, 2018)	criteria provided, conflicting interpretations	VCV000718964	19	50329012	718964	728360	rs775818633	Neutral	0.616	0.234	0.15
KCNC3	41	Q	H	NM_004977.3(KCNC3):c.123G>T (p.Gln41His)	not provided|not specified	Benign/Likely benign(Last reviewed: May 4, 2021)	criteria provided, multiple submitters, no conflicts	VCV000284725	19	50328960	284725	268962	rs185017345	Neutral	0.603	0.249	0.148
KCNC3	43	P	S	NM_004977.3(KCNC3):c.127C>T (p.Pro43Ser)	not specified	Benign(Last reviewed: Apr 13, 2017)	criteria provided, single submitter	VCV000447619	19	50328956	447619	442246	rs928573902	Neutral	0.554	0.272	0.174
KCNC3	47	G	D	NM_004977.3(KCNC3):c.140G>A (p.Gly47Asp)	Spinocerebellar ataxia type 13	Uncertain significance(Last reviewed: Jun 16, 2020)	criteria provided, single submitter	VCV001029909	19	50328943	1029909	1018622	rs2037143763	Neutral	0.603	0.249	0.147
KCNC3	63	D	G	NM_004977.3(KCNC3):c.188A>G (p.Asp63Gly)	Spinocerebellar ataxia type 13|not provided|not specified	Benign(Last reviewed: Aug 19, 2021)	criteria provided, multiple submitters, no conflicts	VCV000193297	19	50328895	193297	190461	rs375912738	Neutral	0.564	0.26	0.176
KCNC3	136	N	D	NM_004977.3(KCNC3):c.634A>G (p.Asn212Asp)	not provided	Uncertain significance(Last reviewed: Nov 6, 2019)	criteria provided, single submitter	VCV001309681	19	50328449	1309681	1299056	NA	LOF	0.8	0.111	0.089
KCNC3	141	A	T	NM_004977.3(KCNC3):c.649G>A (p.Ala217Thr)	not provided	Benign(Last reviewed: Dec 31, 2019)	criteria provided, single submitter	VCV000712000	19	50328434	712000	728359	rs373416318	LOF	0.788	0.059	0.153
KCNC3	257	P	T	NM_004977.3(KCNC3):c.769C>A (p.Pro257Thr)	Inborn genetic diseases|not provided	Uncertain significance(Last reviewed: May 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000521572	19	50328314	521572	512432	rs1232558368	Neutral	0.48	0.307	0.213
KCNC3	271	F	L	NM_004977.3(KCNC3):c.1041C>G (p.Phe347Leu)	not provided	Uncertain significance(Last reviewed: Feb 1, 2021)	criteria provided, single submitter	VCV001256130	19	50323912	1256130	1246060	NA	LOF	0.749	0.079	0.172
KCNC3	331	G	R	NM_004977.3(KCNC3):c.991G>A (p.Gly331Arg)	not provided|Spinocerebellar ataxia type 13	Uncertain significance(Last reviewed: Jul 12, 2019)	criteria provided, multiple submitters, no conflicts	VCV000586086	19	50323962	586086	577817	rs1305901422	Neutral	0.53	0.325	0.144
KCNC3	332	D	G	NM_004977.3(KCNC3):c.1223A>G (p.Asp408Gly)	Spinocerebellar ataxia type 13	Likely pathogenic(Last reviewed: Jul 2, 2019)	criteria provided, single submitter	VCV000976029	19	50323730	976029	964525	rs2037067131	Neutral	0.604	0.255	0.141
KCNC3	337	I	T	NM_004977.3(KCNC3):c.1010T>C (p.Ile337Thr)	not provided	Uncertain significance(Last reviewed: Aug 6, 2018)	criteria provided, single submitter	VCV000391603	19	50323943	391603	377895	rs1057524161	Neutral	0.674	0.186	0.14
KCNC3	380	A	T	NM_004977.3(KCNC3):c.1366G>A (p.Ala456Thr)	not provided	Uncertain significance(Last reviewed: Sep 16, 2019)	criteria provided, single submitter	VCV001311117	19	50323587	1311117	1300498	NA	LOF	0.781	0.073	0.146
KCNC3	387	V	A	NM_004977.3(KCNC3):c.1160T>C (p.Val387Ala)	not provided	Uncertain significance(Last reviewed: Aug 1, 2019)	criteria provided, single submitter	VCV000871011	19	50323793	871011	860601	rs2037067867	LOF	0.714	0.056	0.23

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNC3	399	S	L	NM_004977.3(KCNC3):c.1196C>T (p.Ser399Leu)	not provided|not specified	Conflicting interpretations of pathogenicity(Last reviewed: Mar 24, 2020)	criteria provided, conflicting interpretations	VCV000447618	19	50323757	447618	442244	rs1437049667	LOF	0.754	0.12	0.126
KCNC3	410	L	P	NM_004977.3(KCNC3):c.1229T>C (p.Leu410Pro)	not provided	Uncertain significance(Last reviewed: Dec 31, 2018)	criteria provided, single submitter	VCV000804959	19	50323724	804959	793807	rs1601098675	LOF	0.844	0.056	0.1
KCNC3	420	R	H	NM_004977.3(KCNC3):c.1259G>A (p.Arg420His)	not provided|Spinocerebellar ataxia type 13	Pathogenic(Last reviewed: Dec 28, 2018)	criteria provided, multiple submitters, no conflicts	VCV000013473	19	50323694	13473	28512	rs104894699	LOF	0.864	0.036	0.1
KCNC3	423	R	H	NM_004977.3(KCNC3):c.1268G>A (p.Arg423His)	Spinocerebellar ataxia type 13|not provided| Inborn genetic diseases	Pathogenic(Last reviewed: Feb 10, 2022)	criteria provided, multiple submitters, no conflicts	VCV000208671	19	50323685	208671	205316	rs797044872	LOF	0.85	0.038	0.112
KCNC3	426	T	M	NM_004977.3(KCNC3):c.1505C>T (p.Thr502Met)	not provided	Uncertain significance(Last reviewed: Apr 26, 2019)	criteria provided, single submitter	VCV001305210	19	50323448	1305210	1295485	NA	LOF	0.846	0.036	0.118
KCNC3	428	T	I	NM_004977.3(KCNC3):c.1283C>T (p.Thr428Ile)	Spinocerebellar ataxia type 13	Pathogenic(Last reviewed: Jul 20, 2016)	no assertion criteria provided	VCV000245604	19	50323670	245604	245205	rs879253883	LOF	0.842	0.036	0.122
KCNC3	448	F	L	NM_004977.3(KCNC3):c.1344C>G (p.Phe448Leu)	not provided	Pathogenic(Last reviewed: Nov 3, 2016)	no assertion criteria provided	VCV000545048	19	50323609	545048	535339	rs104894700	GOF	0.473	0.053	0.474
KCNC3	448	F	L	NM_004977.3(KCNC3):c.1344C>G (p.Phe448Leu)	not provided	Pathogenic(Last reviewed: Nov 3, 2016)	no assertion criteria provided	VCV000545048	19	50323609	545048	535339	rs104894700	GOF	0.473	0.053	0.474
KCNC3	448	F	L	NM_004977.3(KCNC3):c.1344C>A (p.Phe448Leu)	Spinocerebellar ataxia type 13	Pathogenic(Last reviewed: Apr 1, 2006)	no assertion criteria provided	VCV000013474	19	50323609	13474	28513	rs104894700	GOF	0.473	0.053	0.474
KCNC3	448	F	L	NM_004977.3(KCNC3):c.1344C>A (p.Phe448Leu)	Spinocerebellar ataxia type 13	Pathogenic(Last reviewed: Apr 1, 2006)	no assertion criteria provided	VCV000013474	19	50323609	13474	28513	rs104894700	GOF	0.473	0.053	0.474
KCNC3	477	D	N	NM_004977.3(KCNC3):c.1429G>A (p.Asp477Asn)	not provided	Uncertain significance(Last reviewed: May 4, 2015)	criteria provided, single submitter	VCV000195233	19	50323524	195233	192394	rs148033381	Neutral	0.451	0.398	0.151
KCNC3	490	P	S	NM_004977.3(KCNC3):c.1696C>T (p.Pro566Ser)	not provided	Uncertain significance(Last reviewed: Feb 4, 2020)	criteria provided, single submitter	VCV001211248	19	50323257	1211248	1201246	NA	LOF	0.838	0.065	0.097
KCNC3	509	M	V	NM_004977.3(KCNC3):c.1525A>G (p.Met509Val)	not specified	Uncertain significance(Last reviewed: Apr 28, 2017)	criteria provided, single submitter	VCV000447621	19	50323428	447621	442242	rs1555781403	LOF	0.738	0.096	0.166
KCNC3	514	G	S	NM_004977.3(KCNC3):c.1768G>A (p.Gly590Ser)	Toe walking|not provided	Uncertain significance(Last reviewed: Feb 17, 2021)	criteria provided, multiple submitters, no conflicts	VCV000916249	19	50323185	916249	904687	rs1167185763	LOF	0.747	0.137	0.116
KCNC3	528	V	G	NM_004977.3(KCNC3):c.1583T>G (p.Val528Gly)	Spinocerebellar ataxia type 13	Uncertain significance(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000803574	19	50323370	803574	791945	rs1601098237	LOF	0.697	0.088	0.215
KCNC3	539	A	V	NM_004977.3(KCNC3):c.1844C>T (p.Ala615Val)	not provided	Uncertain significance(Last	criteria provided,	VCV000804961	19	50323109	804961	793805	rs1432486680	GOF	0.655	0.059	0.286

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Oct 22, 2019)	multiple submitters, no conflicts										
KCNC3	541	P	L	NM_004977.3(KCNC3):c.1850C>T (p.Pro617Leu)	Spinocerebellar ataxia type 13	Uncertain significance(Last reviewed: Dec 18, 2020)	criteria provided, single submitter	VCV001064691	19	50323103	1064691	1052974	NA	LOF	0.7	0.119	0.182
KCNC3	549	R	S	NM_004977.3(KCNC3):c.1875G>T (p.Arg625Ser)	not provided	Benign(Last reviewed: Nov 6, 2018)	criteria provided, single submitter	VCV000804962	19	50323078	804962	793804	rs537912602	Neutral	0.673	0.162	0.165
KCNC3	550	M	V	NM_004977.3(KCNC3):c.1648A>G (p.Met550Val)	not provided	Uncertain significance(Last reviewed: Aug 30, 2021)	criteria provided, single submitter	VCV000424001	19	50323305	424001	410666	rs1064796737	LOF	0.66	0.136	0.205
KCNC3	565	R	W	NM_004977.3(KCNC3):c.1693C>T (p.Arg565Trp)	not provided	Uncertain significance(Last reviewed: Oct 20, 2019)	criteria provided, single submitter	VCV000424152	19	50323260	424152	410665	rs750349505	Neutral	0.557	0.292	0.15
KCNC3	587	P	L	NM_004977.3(KCNC3):c.1760C>T (p.Pro587Leu)	not provided	Benign(Last reviewed: Mar 30, 2018)	criteria provided, single submitter	VCV000586083	19	50323193	586083	577812	rs554197864	Neutral	0.53	0.338	0.133
KCNC3	588	N	S	NM_004977.3(KCNC3):c.1991A>G (p.Asn664Ser)	not provided	Likely benign(Last reviewed: Jul 1, 2020)	criteria provided, single submitter	VCV000995164	19	50320772	995164	983184	rs199833067	Neutral	0.503	0.35	0.148
KCNC3	591	S	G	NM_004977.3(KCNC3):c.1771A>G (p.Ser591Gly)	not specified|not provided	Benign(Last reviewed: Nov 26, 2020)	criteria provided, multiple submitters, no conflicts	VCV000710785	19	50323182	710785	728357	rs549394447	Neutral	0.608	0.257	0.135
KCNC3	599	E	K	NM_004977.3(KCNC3):c.2023G>A (p.Glu675Lys)	not provided	Uncertain significance(Last reviewed: Jan 12, 2021)	criteria provided, single submitter	VCV000995165	19	50320740	995165	983183	rs758477949	Neutral	0.409	0.394	0.197
KCNC3	643	G	V	NM_004977.3(KCNC3):c.2156G>T (p.Gly719Val)	not provided	Uncertain significance(Last reviewed: Aug 27, 2019)	criteria provided, single submitter	VCV001307976	19	50320607	1307976	1297923	NA	Neutral	0.46	0.381	0.158
KCNC3	647	P	L	NM_004977.3(KCNC3):c.1940C>T (p.Pro647Leu)	not specified	Uncertain significance(Last reviewed: Jan 5, 2016)	criteria provided, single submitter	VCV000435548	19	50323013	435548	430262	rs375737637	Neutral	0.495	0.355	0.149
KCNC3	652	E	K	NM_004977.3(KCNC3):c.1954G>A (p.Glu652Lys)	not specified	Uncertain significance(Last reviewed: Aug 2, 2016)	criteria provided, single submitter	VCV000435547	19	50322999	435547	430261	rs1555781339	Neutral	0.485	0.372	0.143
KCNC3	673	A	T	NM_004977.3(KCNC3):c.2245G>A (p.Ala749Thr)	not provided	Uncertain significance(Last reviewed: Sep 1, 2021)	criteria provided, single submitter	VCV001298784	19	50320275	1298784	1288646	NA	Neutral	0.534	0.322	0.144
KCNC3	693	I	M	NM_004977.3(KCNC3):c.2079C>G (p.Ile693Met)	not provided	Likely pathogenic(Last reviewed: Oct 6, 2016)	criteria provided, single submitter	VCV000422460	19	50320684	422460	410664	rs1064795793	Neutral	0.623	0.251	0.125
KCNC3	698	R	H	NM_004977.3(KCNC3):c.2093G>A (p.Arg698His)	not provided|not specified	Benign(Last reviewed: Sep 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000447622	19	50320670	447622	442241	rs144957827	Neutral	0.475	0.368	0.158
KCND2	11	F	L	NM_012281.3(KCND2):c.31T>C (p.Phe11Leu)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Apr 25,	criteria provided, single submitter	VCV000569893	7	120274663	569893	561244	rs142392153	Neutral	0.673	0.167	0.16

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2018)											
KCND2	11	F	L	NM_012281.3(KCND2):c.31T>C (p.Phe11Leu)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Apr 25, 2018)	criteria provided, single submitter	VCV000569893	7	120274663	569893	561244	rs142392153	Neutral	0.673	0.167	0.16
KCND2	11	F	L	NM_012281.3(KCND2):c.33T>G (p.Phe11Leu)	Early myoclonic encephalopathy	Likely benign(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV000947786	7	120274665	947786	924622	rs35950053	Neutral	0.673	0.167	0.16
KCND2	11	F	L	NM_012281.3(KCND2):c.33T>G (p.Phe11Leu)	Early myoclonic encephalopathy	Likely benign(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV000947786	7	120274665	947786	924622	rs35950053	Neutral	0.673	0.167	0.16
KCND2	21	P	A	NM_012281.3(KCND2):c.61C>G (p.Pro21Ala)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Sep 9, 2019)	criteria provided, single submitter	VCV000954629	7	120274693	954629	945352	rs1799145590	LOF	0.742	0.064	0.194
KCND2	22	V	L	NM_012281.3(KCND2):c.64G>C (p.Val22Leu)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Feb 22, 2020)	criteria provided, single submitter	VCV000648141	7	120274696	648141	635565	rs746823484	LOF	0.759	0.146	0.095
KCND2	26	P	S	NM_012281.3(KCND2):c.76C>T (p.Pro26Ser)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 3, 2020)	criteria provided, single submitter	VCV001008472	7	120274708	1008472	991980	rs373082898	LOF	0.739	0.159	0.102
KCND2	39	Q	E	NM_012281.3(KCND2):c.115C>G (p.Gln39Glu)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Apr 10, 2019)	criteria provided, single submitter	VCV000947519	7	120274747	947519	924623	rs1272011851	Neutral	0.629	0.187	0.184
KCND2	44	V	M	NM_012281.3(KCND2):c.130G>A (p.Val44Met)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Feb 27, 2020)	criteria provided, single submitter	VCV001025284	7	120274762	1025284	1007134	rs1799147199	LOF	0.794	0.122	0.084
KCND2	51	R	C	NM_012281.3(KCND2):c.151C>T (p.Arg51Cys)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, single submitter	VCV001018504	7	120274783	1018504	1007135	rs751823176	LOF	0.772	0.13	0.098
KCND2	63	P	L	NM_012281.3(KCND2):c.188C>T (p.Pro63Leu)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 21, 2020)	criteria provided, single submitter	VCV001038912	7	120274820	1038912	1027661	rs1799148358	Neutral	0.658	0.192	0.15
KCND2	99	Y	C	NM_012281.3(KCND2):c.296A>G (p.Tyr99Cys)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 4, 2018)	criteria provided, single submitter	VCV000639013	7	120274928	639013	635566	rs1584706364	LOF	0.672	0.124	0.204
KCND2	100	R	L	NM_012281.3(KCND2):c.299G>T (p.Arg100Leu)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Dec 27, 2018)	criteria provided, single submitter	VCV000662969	7	120274931	662969	635567	rs377746178	GOF	0.618	0.049	0.333
KCND2	108	R	C	NM_012281.3(KCND2):c.322C>T (p.Arg108Cys)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Dec 13, 2018)	criteria provided, single submitter	VCV000654476	7	120274954	654476	635568	rs763189187	LOF	0.715	0.101	0.184
KCND2	108	R	H	NM_012281.3(KCND2):c.323G>A (p.Arg108His)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 16, 2020)	criteria provided, single submitter	VCV001036551	7	120274955	1036551	1027662	rs1799151446	LOF	0.707	0.07	0.223
KCND2	109	H	N	NM_012281.3(KCND2):c.325C>A (p.His109Asn)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 16, 2020)	criteria provided, single submitter	VCV001063447	7	120274957	1063447	1044599	NA	GOF	0.637	0.092	0.271
KCND2	120	A	D	NM_012281.3(KCND2):c.359C>A	Early myoclonic encephalopathy	Uncertain	criteria	VCV001043092	7	120274991	1043092	1027663	rs1199722034	GOF	0.638	0.062	0.3

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Ala120Asp)		significance(Last reviewed: Mar 26, 2020)	provided, single submitter										
KCND2	129	I	V	NM_012281.3(KCND2):c.385A>G (p.Ile129Val)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Nov 13, 2018)	criteria provided, single submitter	VCV000645514	7	120275017	645514	635569	rs1448870371	LOF	0.713	0.156	0.131
KCND2	197	V	I	NM_012281.3(KCND2):c.589G>A (p.Val197Ile)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 3, 2020)	criteria provided, single submitter	VCV000941432	7	120275221	941432	933611	rs750056826	Neutral	0.585	0.256	0.159
KCND2	216	I	V	NM_012281.3(KCND2):c.646A>G (p.Ile216Val)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Aug 9, 2018)	criteria provided, single submitter	VCV000652645	7	120275278	652645	635570	rs373390097	Neutral	0.689	0.191	0.12
KCND2	218	E	Q	NM_012281.3(KCND2):c.652G>C (p.Glu218Gln)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Dec 10, 2018)	criteria provided, single submitter	VCV000663034	7	120275284	663034	635571	rs1584706614	GOF	0.675	0.084	0.24
KCND2	235	A	V	NM_012281.3(KCND2):c.704C>T (p.Ala235Val)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Sep 6, 2019)	criteria provided, single submitter	VCV000956493	7	120275336	956493	945353	rs1398907255	LOF	0.796	0.126	0.078
KCND2	249	A	V	NM_012281.3(KCND2):c.746C>T (p.Ala249Val)	Early myoclonic encephalopathy	Likely benign(Last reviewed: Nov 14, 2020)	criteria provided, single submitter	VCV000460205	7	120275378	460205	456380	rs146220085	LOF	0.693	0.102	0.206
KCND2	330	S	L	NM_012281.3(KCND2):c.989C>T (p.Ser330Leu)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Sep 27, 2018)	criteria provided, single submitter	VCV000657270	7	120275621	657270	635572	rs1584706842	LOF	0.764	0.105	0.13
KCND2	333	M	V	NM_012281.3(KCND2):c.997A>G (p.Met333Val)	not provided	Uncertain significance(Last reviewed: Feb 28, 2020)	criteria provided, single submitter	VCV001299437	7	120275629	1299437	1289384	NA	Neutral	0.747	0.18	0.074
KCND2	352	S	T	NM_012281.3(KCND2):c.1055G>C (p.Ser352Thr)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, single submitter	VCV000859284	7	120275687	859284	832904	rs765958038	GOF	0.677	0.037	0.286
KCND2	395	V	A	NM_012281.3(KCND2):c.1184T>C (p.Val395Ala)	not provided	Uncertain significance(Last reviewed: Oct 10, 2019)	criteria provided, single submitter	VCV001308937	7	120732971	1308937	1300965	NA	Neutral	0.486	0.329	0.185
KCND2	403	P	A	NM_012281.3(KCND2):c.1207C>G (p.Pro403Ala)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Mar 9, 2020)	criteria provided, single submitter	VCV001005965	7	120732994	1005965	991981	rs1792826036	LOF	0.847	0.055	0.097
KCND2	403	P	L	NM_012281.3(KCND2):c.1208C>T (p.Pro403Leu)	not provided|Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV001009567	7	120732995	1009567	991982	rs750457269	LOF	0.861	0.055	0.084
KCND2	404	V	M	NM_012281.3(KCND2):c.1210G>A (p.Val404Met)	not provided|Early myoclonic encephalopathy	Pathogenic(Last reviewed: Feb 28, 2020)	criteria provided, single submitter	VCV000144007	7	120732997	144007	153738	rs587777631	Neutral	0.516	0.384	0.1
KCND2	413	Y	C	NM_012281.3(KCND2):c.1238A>G (p.Tyr413Cys)	not provided	Uncertain significance(Last reviewed: Nov 8, 2017)	criteria provided, single submitter	VCV000560272	7	120733025	560272	551298	rs1562923143	Neutral	0.65	0.175	0.175
KCND2	434	R	W	NM_012281.3(KCND2):c.1300C>T (p.Arg434Trp)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 9,	criteria provided, single submitter	VCV001019937	7	120741555	1019937	1007137	rs376029594	Neutral	0.726	0.174	0.101

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)											
KCND2	435	A	V	NM_012281.3(KCND2):c.1304C>T (p.Ala435Val)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Jun 3, 2020)	criteria provided, single submitter	VCV001042710	7	120741559	1042710	1027664	rs1484410590	Neutral	0.708	0.188	0.104
KCND2	441	A	T	NM_012281.3(KCND2):c.1321G>A (p.Ala441Thr)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Feb 4, 2019)	criteria provided, single submitter	VCV000575507	7	120741576	575507	566333	rs1417852990	Neutral	0.703	0.19	0.107
KCND2	442	N	S	NM_012281.3(KCND2):c.1325A>G (p.Asn442Ser)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Jun 1, 2020)	criteria provided, single submitter	VCV000529706	7	120741580	529706	522090	rs957617040	Neutral	0.704	0.189	0.107
KCND2	471	G	R	NM_012281.3(KCND2):c.1411G>C (p.Gly471Arg)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Jan 28, 2020)	criteria provided, single submitter	VCV000640257	7	120742546	640257	635573	rs757922549	Neutral	0.716	0.177	0.107
KCND2	472	S	F	NM_012281.3(KCND2):c.1415C>T (p.Ser472Phe)	Early myoclonic encephalopathy	Likely benign(Last reviewed: Nov 21, 2020)	criteria provided, single submitter	VCV000767427	7	120742550	767427	699780	rs146058924	Neutral	0.719	0.178	0.103
KCND2	473	S	T	NM_012281.3(KCND2):c.1418G>C (p.Ser473Thr)	Early myoclonic encephalopathy	Likely benign(Last reviewed: Feb 24, 2020)	criteria provided, single submitter	VCV000857319	7	120742553	857319	832905	rs140087318	Neutral	0.698	0.195	0.107
KCND2	504	M	V	NM_012281.3(KCND2):c.1510A>G (p.Met504Val)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Jan 27, 2019)	criteria provided, single submitter	VCV000836847	7	120745822	836847	832906	rs1792998586	Neutral	0.537	0.286	0.177
KCND2	511	R	H	NM_012281.3(KCND2):c.1532G>A (p.Arg511His)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Jan 26, 2020)	criteria provided, single submitter	VCV000998545	7	120745844	998545	991983	rs35460901	Neutral	0.708	0.186	0.106
KCND2	512	P	S	NM_012281.3(KCND2):c.1534C>T (p.Pro512Ser)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Apr 13, 2018)	criteria provided, single submitter	VCV000568350	7	120745846	568350	566337	rs372198586	Neutral	0.698	0.199	0.104
KCND2	521	S	L	NM_012281.3(KCND2):c.1562C>T (p.Ser521Leu)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Oct 7, 2019)	criteria provided, single submitter	VCV000936111	7	120745874	936111	933612	rs1792999457	Neutral	0.713	0.185	0.103
KCND2	533	R	Q	NM_012281.3(KCND2):c.1598G>A (p.Arg533Gln)	Early myoclonic encephalopathy	Likely benign(Last reviewed: Jun 13, 2020)	criteria provided, single submitter	VCV000574695	7	120745910	574695	566340	rs367713278	Neutral	0.719	0.178	0.102
KCND2	539	R	H	NM_012281.3(KCND2):c.1616G>A (p.Arg539His)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Jul 29, 2020)	criteria provided, single submitter	VCV000529715	7	120745928	529715	522091	rs376772376	Neutral	0.718	0.181	0.101
KCND2	543	A	V	NM_012281.3(KCND2):c.1628C>T (p.Ala543Val)	Early myoclonic encephalopathy	Uncertain significance(Last reviewed: Jun 26, 2018)	criteria provided, single submitter	VCV000568878	7	120745940	568878	561319	rs1168875214	Neutral	0.714	0.186	0.101
KCND3	2	A	E	NM_001378969.1(KCND3):c.5C>A (p.Ala2Glu)	Spinocerebellar ataxia type 19/22| Cardiovascular phenotype|not specified|not provided|Inborn genetic diseases	Conflicting interpretations of pathogenicity(Last reviewed: Nov 20, 2020)	criteria provided, conflicting interpretations	VCV000372391	1	111982722	372391	359196	rs201340369	LOF	0.854	0.044	0.102
KCND3	13	R	Q	NM_001378969.1(KCND3):c.38G>A (p.Arg13Gln)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Dec 12, 2017)	criteria provided, single submitter	VCV000518761	1	111982689	518761	509074	rs1339082402	LOF	0.863	0.04	0.097
KCND3	15	A	V	NM_001378969.1(KCND3):c.44C>T	not provided	Uncertain	criteria	VCV000586067	1	111982683	586067	576402	rs866544148	LOF	0.839	0.051	0.109

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Ala15Val)		significance(Last reviewed: Nov 13, 2017)	provided, single submitter										
KCND3	28	P	T	NM_001378969.1(KCND3):c.82C>A (p.Pro28Thr)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jan 31, 2020)	criteria provided, single submitter	VCV001017799	1	111982645	1017799	1002043	rs1675024873	LOF	0.71	0.117	0.173
KCND3	30	A	D	NM_001378969.1(KCND3):c.89C>A (p.Ala30Asp)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: May 5, 2020)	criteria provided, single submitter	VCV000465169	1	111982638	465169	447055	rs1307934269	LOF	0.661	0.144	0.195
KCND3	31	P	S	NM_001378969.1(KCND3):c.91C>T (p.Pro31Ser)	Spinocerebellar ataxia type 19/22|not provided	Uncertain significance(Last reviewed: Feb 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000452259	1	111982636	452259	442581	rs1403997481	LOF	0.704	0.12	0.175
KCND3	37	R	Q	NM_001378969.1(KCND3):c.110G>A (p.Arg37Gln)	not provided	Uncertain significance(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV001316993	1	111982617	1316993	1306811	NA	Neutral	0.626	0.148	0.226
KCND3	86	R	G	NM_001378969.1(KCND3):c.256C>G (p.Arg86Gly)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Oct 17, 2019)	criteria provided, single submitter	VCV000967961	1	111982471	967961	952015	rs754042199	LOF	0.786	0.085	0.129
KCND3	86	R	Q	NM_001378969.1(KCND3):c.257G>A (p.Arg86Gln)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jun 11, 2019)	criteria provided, single submitter	VCV000689484	1	111982470	689484	677121	rs1571941606	LOF	0.775	0.08	0.146
KCND3	116	D	N	NM_001378969.1(KCND3):c.346G>A (p.Asp116Asn)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Oct 23, 2019)	criteria provided, single submitter	VCV000961544	1	111982381	961544	941383	rs766584545	LOF	0.832	0.088	0.08
KCND3	119	A	D	NM_001378969.1(KCND3):c.356C>A (p.Ala119Asp)	not provided	Uncertain significance(Last reviewed: Jun 7, 2021)	criteria provided, single submitter	VCV001256140	1	111982371	1256140	1246070	NA	LOF	0.884	0.027	0.088
KCND3	129	G	A	NM_001378969.1(KCND3):c.386G>C (p.Gly129Ala)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Sep 20, 2019)	criteria provided, single submitter	VCV000940586	1	111982341	940586	929964	rs1675001550	LOF	0.842	0.043	0.115
KCND3	133	Y	H	NM_001378969.1(KCND3):c.397T>C (p.Tyr133His)	not specified	Uncertain significance(Last reviewed: Dec 20, 2016)	criteria provided, single submitter	VCV000447627	1	111982330	447627	440345	rs1553187539	LOF	0.855	0.044	0.101
KCND3	136	Y	C	NM_001378969.1(KCND3):c.407A>G (p.Tyr136Cys)	not provided	Uncertain significance(Last reviewed: Nov 26, 2019)	criteria provided, single submitter	VCV000995134	1	111982320	995134	982355	rs1675000214	LOF	0.872	0.044	0.084
KCND3	144	A	T	NM_001378969.1(KCND3):c.430G>A (p.Ala144Thr)	not provided	Uncertain significance(Last reviewed: May 7, 2019)	criteria provided, single submitter	VCV000804955	1	111982297	804955	792825	rs143933558	LOF	0.734	0.097	0.169
KCND3	145	E	K	NM_001378969.1(KCND3):c.433G>A (p.Glu145Lys)	not provided	Uncertain significance(Last reviewed: Aug 22, 2018)	criteria provided, single submitter	VCV000586066	1	111982294	586066	576400	rs1557768261	LOF	0.741	0.083	0.176
KCND3	149	D	G	NM_001378969.1(KCND3):c.446A>G (p.Asp149Gly)	not specified|Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jun 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000447628	1	111982281	447628	440344	rs1217571134	Neutral	0.657	0.16	0.183

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCND3	150	D	N	NM_001378969.1(KCND3):c.448G>A (p.Asp150Asn)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Mar 4, 2019)	criteria provided, single submitter	VCV000838739	1	111982279	838739	822538	rs1488410733	LOF	0.785	0.12	0.095
KCND3	166	R	C	NM_001378969.1(KCND3):c.496C>T (p.Arg166Cys)	not provided	Uncertain significance(Last reviewed: Dec 18, 2019)	criteria provided, single submitter	VCV001315984	1	111982231	1315984	1306242	NA	LOF	0.878	0.038	0.084
KCND3	172	A	T	NM_001378969.1(KCND3):c.514G>A (p.Ala172Thr)	Cardiovascular phenotype	Uncertain significance(Last reviewed: May 16, 2017)	criteria provided, single submitter	VCV000519447	1	111982213	519447	509071	rs149488365	LOF	0.88	0.031	0.089
KCND3	185	F	L	NM_001378969.1(KCND3):c.553T>C (p.Phe185Leu)	not provided	Uncertain significance(Last reviewed: Feb 9, 2018)	criteria provided, single submitter	VCV000586068	1	111982174	586068	576399	rs1219355757	LOF	0.835	0.058	0.106
KCND3	204	T	M	NM_001378969.1(KCND3):c.611C>T (p.Thr204Met)	Spinocerebellar ataxia type 19/22	Likely pathogenic(Last reviewed: Jul 12, 2021)	criteria provided, single submitter	VCV001027415	1	111982116	1027415	1015157	NA	LOF	0.869	0.029	0.103
KCND3	209	T	M	NM_001378969.1(KCND3):c.626C>T (p.Thr209Met)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Jan 20, 2017)	criteria provided, single submitter	VCV000519385	1	111982101	519385	509070	rs771878661	LOF	0.739	0.102	0.159
KCND3	214	K	R	NM_001378969.1(KCND3):c.641A>G (p.Lys214Arg)	not specified|not provided|Cardiovascular phenotype|Spinocerebellar ataxia type 19/22	Conflicting interpretations of pathogenicity(Last reviewed: Jun 17, 2021)	criteria provided, conflicting interpretations	VCV000222665	1	111982086	222665	224176	rs142744204	LOF	0.735	0.098	0.167
KCND3	223	S	P	NM_001378969.1(KCND3):c.667T>C (p.Ser223Pro)	not specified	Uncertain significance(Last reviewed: Sep 30, 2016)	criteria provided, single submitter	VCV000447629	1	111982060	447629	440343	rs778053540	LOF	0.859	0.054	0.086
KCND3	258	V	I	NM_001378969.1(KCND3):c.772G>A (p.Val258Ile)	not provided	Uncertain significance(Last reviewed: Oct 18, 2019)	criteria provided, single submitter	VCV001316355	1	111981955	1316355	1308757	NA	LOF	0.878	0.042	0.079
KCND3	262	I	S	NM_001378969.1(KCND3):c.785T>G (p.Ile262Ser)	not provided	Uncertain significance(Last reviewed: Jan 14, 2021)	criteria provided, single submitter	VCV001317441	1	111981942	1317441	1306385	NA	LOF	0.896	0.029	0.075
KCND3	269	P	R	NM_001378969.1(KCND3):c.806C>G (p.Pro269Arg)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jan 12, 2021)	criteria provided, single submitter	VCV001333928	1	111981921	1333928	1324698	NA	LOF	0.905	0.023	0.072
KCND3	273	G	S	NM_001378969.1(KCND3):c.817G>A (p.Gly273Ser)	Brugada syndrome	Uncertain significance(Last reviewed: Sep 14, 2015)	criteria provided, single submitter	VCV000222666	1	111981910	222666	224175	rs869025444	LOF	0.889	0.029	0.082
KCND3	280	E	K	NM_001378969.1(KCND3):c.838G>A (p.Glu280Lys)	not provided	Uncertain significance(Last reviewed: Jul 19, 2019)	criteria provided, single submitter	VCV001316178	1	111981889	1316178	1308580	NA	LOF	0.841	0.046	0.114
KCND3	290	R	Q	NM_001378969.1(KCND3):c.869G>A (p.Arg290Gln)	Spinocerebellar ataxia type 19/22	Likely pathogenic(Last reviewed: Mar 2, 2018)	criteria provided, single submitter	VCV000976123	1	111981858	976123	964123	rs1674966041	LOF	0.901	0.016	0.083
KCND3	290	R	W	NM_001378969.1(KCND3):c.868C>T (p.Arg290Trp)	KCND3-Related Disorder	not provided	no assertion provided	VCV001339841	1	111981859	1339841	1331051	NA	LOF	0.895	0.021	0.084
KCND3	293	R	H	NM_001378969.1(KCND3):c.878G>A (p.Arg293His)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last	criteria provided, single	VCV001038793	1	111981849	1038793	1022544	rs1674965278	LOF	0.84	0.089	0.071

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Aug 3, 2020)	submitter										
KCND3	304	S	P	NM_001378969.1(KCND3):c.910T>C (p.Ser304Pro)	not provided	Likely pathogenic(Last reviewed: Oct 23, 2020)	criteria provided, single submitter	VCV000987028	1	111981817	987028	974945	rs1674962966	LOF	0.882	0.027	0.09
KCND3	313	T	A	NM_001378969.1(KCND3):c.937A>G (p.Thr313Ala)	not specified	Uncertain significance(Last reviewed: Jun 21, 2017)	criteria provided, single submitter	VCV000447630	1	111981790	447630	440342	rs1553187267	LOF	0.903	0.021	0.076
KCND3	317	C	Y	NM_001378969.1(KCND3):c.950G>A (p.Cys317Tyr)	Spinocerebellar ataxia type 19/22	Pathogenic(Last reviewed: Apr 20, 2019)	criteria provided, single submitter	VCV000626316	1	111981777	626316	614651	rs1571939905	LOF	0.875	0.025	0.1
KCND3	328	L	R	NM_001378969.1(KCND3):c.983T>G (p.Leu328Arg)	not provided	Uncertain significance(Last reviewed: Apr 12, 2019)	criteria provided, single submitter	VCV001315586	1	111981744	1315586	1305846	NA	LOF	0.911	0.023	0.066
KCND3	338	V	E	NM_001378969.1(KCND3):c.1013T>A (p.Val338Glu)	Spinocerebellar ataxia type 19/22	Pathogenic(Last reviewed: Apr 20, 2019)	criteria provided, single submitter	VCV000626317	1	111981714	626317	614650	rs1571939827	LOF	0.857	0.046	0.097
KCND3	343	E	K	NM_001378969.1(KCND3):c.1027G>A (p.Glu343Lys)	not provided	Uncertain significance(Last reviewed: Jan 16, 2018)	criteria provided, single submitter	VCV000586062	1	111981700	586062	576394	rs1557767012	LOF	0.918	0.019	0.063
KCND3	345	G	V	NM_001378969.1(KCND3):c.1034G>T (p.Gly345Val)	Spinocerebellar ataxia type 19/22	Pathogenic(Last reviewed: Jun 27, 2014)	criteria provided, single submitter	VCV000211216	1	111981693	211216	206701	rs797045634	LOF	0.871	0.029	0.1
KCND3	350	K	N	NM_001378969.1(KCND3):c.1050G>T (p.Lys350Asn)	not provided	Uncertain significance(Last reviewed: Sep 1, 2021)	criteria provided, single submitter	VCV001298437	1	111981677	1298437	1288299	NA	LOF	0.738	0.089	0.172
KCND3	351	F	L	NM_001378969.1(KCND3):c.1051T>C (p.Phe351Leu)	not provided	Likely pathogenic(Last reviewed: May 3, 2016)	criteria provided, single submitter	VCV000421079	1	111981676	421079	404882	rs1064794895	LOF	0.77	0.076	0.154
KCND3	352	T	A	NM_001378969.1(KCND3):c.1054A>G (p.Thr352Ala)	not specified	Uncertain significance(Last reviewed: Dec 20, 2016)	criteria provided, single submitter	VCV000447625	1	111981673	447625	440341	rs397515476	LOF	0.867	0.043	0.09
KCND3	352	T	P	NM_001378969.1(KCND3):c.1054A>C (p.Thr352Pro)	not provided|Spinocerebellar ataxia type 19/22	Pathogenic(Last reviewed: Oct 23, 2020)	criteria provided, single submitter	VCV000066062	1	111981673	66062	76966	rs397515476	LOF	0.858	0.044	0.098
KCND3	354	I	V	NM_001378969.1(KCND3):c.1060A>G (p.Ile354Val)	not provided	Uncertain significance(Last reviewed: Sep 1, 2021)	criteria provided, single submitter	VCV001298436	1	111981667	1298436	1288298	NA	LOF	0.87	0.071	0.06
KCND3	355	P	S	NM_001378969.1(KCND3):c.1063C>T (p.Pro355Ser)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Nov 3, 2021)	criteria provided, single submitter	VCV001319064	1	111981664	1319064	1309317	NA	LOF	0.864	0.065	0.071
KCND3	357	S	L	NM_001378969.1(KCND3):c.1070C>T (p.Ser357Leu)	Spinocerebellar ataxia type 19/22	Likely pathogenic(Last reviewed: May 15, 2019)	criteria provided, single submitter	VCV000827788	1	111981657	827788	815974	rs867628133	LOF	0.852	0.074	0.074
KCND3	357	S	P	NM_001378969.1(KCND3):c.1069T>C (p.Ser357Pro)	not provided	Uncertain significance(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV001316217	1	111981658	1316217	1308619	NA	LOF	0.864	0.064	0.072
KCND3	357	S	W	NM_001378969.1(KCND3):c.1070C>G (p.Ser357Trp)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last	criteria provided, single	VCV001004161	1	111981657	1004161	986820	rs867628133	LOF	0.849	0.076	0.075

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Oct 20, 2020)	submitter										
KCND3	359	W	L	NM_001378969.1(KCND3):c.1076G>T (p.Trp359Leu)	Seizures|Intellectual disability	Likely pathogenic	criteria provided, single submitter	VCV000816626	1	111981651	816626	804831	rs1571939623	LOF	0.865	0.06	0.075
KCND3	368	L	M	NM_001378969.1(KCND3):c.1102C>A (p.Leu368Met)	not provided	Uncertain significance(Last reviewed: Jul 1, 2021)	criteria provided, single submitter	VCV001298435	1	111981625	1298435	1288297	NA	LOF	0.907	0.031	0.062
KCND3	371	G	R	NM_001378969.1(KCND3):c.1111G>A (p.Gly371Arg)	not provided|Spinocerebellar ataxia type 19/22	Pathogenic/Likely pathogenic(Last reviewed: Feb 3, 2021)	criteria provided, multiple submitters, no conflicts	VCV000383943	1	111787102	383943	364367	rs1057521793	LOF	0.859	0.043	0.098
KCND3	373	M	I	NM_001378969.1(KCND3):c.1119G>A (p.Met373Ile)	Variant of unknown significance	Uncertain significance(Last reviewed: Dec 1, 2012)	no assertion criteria provided	VCV000066063	1	111787094	66063	76967	rs397515477	LOF	0.892	0.027	0.081
KCND3	375	P	L	NM_001378969.1(KCND3):c.1124C>T (p.Pro375Leu)	Inborn genetic diseases	Uncertain significance(Last reviewed: Dec 28, 2017)	criteria provided, single submitter	VCV000985855	1	111787089	985855	973124	rs1664633885	LOF	0.821	0.058	0.121
KCND3	375	P	S	NM_001378969.1(KCND3):c.1123C>T (p.Pro375Ser)	Spinocerebellar ataxia type 19/22	Pathogenic(Last reviewed: Apr 20, 2019)	criteria provided, single submitter	VCV000626318	1	111787090	626318	614649	rs1571636508	LOF	0.838	0.042	0.12
KCND3	377	T	M	NM_001378969.1(KCND3):c.1130C>T (p.Thr377Met)	not provided|Spinocerebellar ataxia type 19/22	Pathogenic(Last reviewed: Oct 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000626319	1	111787083	626319	614648	rs1571636501	LOF	0.855	0.049	0.095
KCND3	384	G	S	NM_001378969.1(KCND3):c.1150G>A (p.Gly384Ser)	not provided	Likely pathogenic(Last reviewed: Dec 11, 2019)	criteria provided, single submitter	VCV000995131	1	111787063	995131	982353	rs1664632655	LOF	0.861	0.045	0.095
KCND3	385	S	P	NM_001378969.1(KCND3):c.1153T>C (p.Ser385Pro)	Spinocerebellar ataxia type 19/22	Pathogenic	no assertion criteria provided	VCV000375399	1	111787060	375399	362133	rs1057519453	LOF	0.872	0.053	0.075
KCND3	390	S	N	NM_001378969.1(KCND3):c.1169G>A (p.Ser390Asn)	Variant of unknown significance	Uncertain significance(Last reviewed: Dec 1, 2012)	no assertion criteria provided	VCV000066064	1	111787044	66064	76968	rs397515478	LOF	0.905	0.017	0.079
KCND3	392	V	I	NM_001378969.1(KCND3):c.1174G>A (p.Val392Ile)	Spinocerebellar ataxia type 19/22|Brugada syndrome 9|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Nov 23, 2021)	criteria provided, conflicting interpretations	VCV000192255	1	111787039	192255	190052	rs786205867	LOF	0.864	0.045	0.09
KCND3	399	V	L	NM_001378969.1(KCND3):c.1195G>C (p.Val399Leu)	not provided	Likely pathogenic(Last reviewed: Dec 17, 2015)	criteria provided, single submitter	VCV000372829	1	111787018	372829	359195	rs1057518007	LOF	0.825	0.092	0.083
KCND3	419	R	H	NM_001378969.1(KCND3):c.1256G>A (p.Arg419His)	not provided	Uncertain significance(Last reviewed: Mar 7, 2018)	criteria provided, single submitter	VCV000393083	1	111786957	393083	364361	rs774338559	LOF	0.669	0.147	0.184
KCND3	426	R	H	NM_001378969.1(KCND3):c.1277G>A (p.Arg426His)	not provided	Uncertain significance(Last reviewed: May 10, 2019)	criteria provided, single submitter	VCV000804951	1	111780784	804951	792824	rs760274429	LOF	0.681	0.132	0.187
KCND3	431	R	H	NM_001378969.1(KCND3):c.1292G>A (p.Arg431His)	Cardiovascular phenotype|Spinocerebellar ataxia type 19/22|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Feb 10, 2021)	criteria provided, conflicting interpretations	VCV000519484	1	111780769	519484	509066	rs771703569	GOF	0.459	0.073	0.468

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCND3	438	S	L	NM_001378969.1(KCND3):c.1313C>T (p.Ser438Leu)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Nov 3, 2021)	criteria provided, single submitter	VCV001319063	1	111780748	1319063	1309316	NA	Neutral	0.459	0.214	0.327
KCND3	438	S	W	NM_001378969.1(KCND3):c.1313C>G (p.Ser438Trp)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: May 16, 2017)	criteria provided, single submitter	VCV000465164	1	111780748	465164	447138	rs1172444288	Neutral	0.466	0.219	0.316
KCND3	446	R	C	NM_001378969.1(KCND3):c.1336C>T (p.Arg446Cys)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Oct 8, 2017)	criteria provided, single submitter	VCV000533725	1	111780725	533725	514985	rs756087542	GOF	0.546	0.132	0.322
KCND3	447	N	D	NM_001378969.1(KCND3):c.1339A>G (p.Asn447Asp)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jul 13, 2018)	criteria provided, single submitter	VCV000663733	1	111780722	663733	626612	rs1571626928	GOF	0.509	0.159	0.332
KCND3	450	L	F	NM_001378969.1(KCND3):c.1348C>T (p.Leu450Phe)	Cardiovascular phenotype|Brugada syndrome 9| Spinocerebellar ataxia type 19/22|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Dec 1, 2021)	criteria provided, conflicting interpretations	VCV000192253	1	111780713	192253	190050	rs150401343	GOF	0.444	0.092	0.464
KCND3	452	E	K	NM_001378969.1(KCND3):c.1354G>A (p.Glu452Lys)	not provided|Primary dilated cardiomyopathy| not specified|Spinocerebellar ataxia type 19/22	Benign/Likely benign(Last reviewed: May 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000447626	1	111780707	447626	440340	rs200532657	GOF	0.478	0.185	0.338
KCND3	457	T	M	NM_001378969.1(KCND3):c.1370C>T (p.Thr457Met)	Brugada syndrome 9|not provided	Uncertain significance(Last reviewed: Apr 20, 2018)	criteria provided, single submitter	VCV000432538	1	111780691	432538	425302	rs199637120	Neutral	0.376	0.318	0.306
KCND3	463	E	K	NM_001378969.1(KCND3):c.1387G>A (p.Glu463Lys)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Sep 19, 2019)	criteria provided, single submitter	VCV000958292	1	111780299	958292	941382	rs1571626257	Neutral	0.446	0.324	0.23
KCND3	471	L	F	NM_001378969.1(KCND3):c.1411C>T (p.Leu471Phe)	not provided	Uncertain significance(Last reviewed: Oct 11, 2018)	criteria provided, multiple submitters, no conflicts	VCV000423997	1	111780275	423997	404881	rs1064796735	LOF	0.643	0.139	0.219
KCND3	476	H	R	NM_001378969.1(KCND3):c.1427A>G (p.His476Arg)	not provided|Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jul 15, 2020)	criteria provided, multiple submitters, no conflicts	VCV000804952	1	111780259	804952	792823	rs1571626155	LOF	0.742	0.065	0.194
KCND3	486	T	A	NM_001378969.1(KCND3):c.1456A>G (p.Thr486Ala)	not specified|not provided|Spinocerebellar ataxia type 19/22	Conflicting interpretations of pathogenicity(Last reviewed: Dec 3, 2020)	criteria provided, conflicting interpretations	VCV000415286	1	111780230	415286	390769	rs149008060	LOF	0.721	0.095	0.184
KCND3	493	V	G	NM_001378969.1(KCND3):c.1478T>G (p.Val493Gly)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Sep 29, 2020)	criteria provided, single submitter	VCV001041440	1	111778476	1041440	1022543	rs1216457569	Neutral	0.68	0.147	0.173
KCND3	499	S	C	NM_001378969.1(KCND3):c.1496C>G (p.Ser499Cys)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jun 7, 2019)	criteria provided, single submitter	VCV000570216	1	111778458	570216	556978	rs976664434	LOF	0.635	0.156	0.209
KCND3	501	R	Q	NM_001378969.1(KCND3):c.1502G>A (p.Arg501Gln)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Dec 31, 2019)	criteria provided, single submitter	VCV000837098	1	111778452	837098	822537	rs764928165	Neutral	0.602	0.176	0.222
KCND3	515	M	L	NM_001378969.1(KCND3):c.1543A>C	Cardiovascular phenotype	Uncertain	criteria	VCV000519397	1	111777249	519397	509064	rs369907159	LOF	0.663	0.132	0.205

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Met515Leu)		significance(Last reviewed: Feb 8, 2017)	provided, single submitter										
KCND3	515	P	T	NM_001378969.1(KCND3):c.1600C>A (p.Pro534Thr)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Mar 12, 2020)	criteria provided, single submitter	VCV001014492	1	111777192	1014492	1002042	rs1447493103	GOF	0.598	0.117	0.285
KCND3	517	E	Q	NM_001378969.1(KCND3):c.1549G>C (p.Glu517Gln)	not provided	Uncertain significance(Last reviewed: Dec 21, 2018)	criteria provided, single submitter	VCV000804953	1	111777243	804953	792822	rs202110939	GOF	0.571	0.111	0.318
KCND3	525	M	V	NM_001378969.1(KCND3):c.1573A>G (p.Met525Val)	Cardiovascular phenotype|not specified| Spinocerebellar ataxia type 19/22|not provided	Benign/Likely benign(Last reviewed: Mar 27, 2021)	criteria provided, multiple submitters, no conflicts	VCV000519434	1	111777219	519434	509063	rs145890206	GOF	0.461	0.2	0.339
KCND3	526	T	I	NM_001378969.1(KCND3):c.1634C>T (p.Thr545Ile)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jul 30, 2020)	criteria provided, single submitter	VCV001059490	1	111777158	1059490	1039359	NA	Neutral	0.4	0.324	0.275
KCND3	531	R	H	NM_001378969.1(KCND3):c.1649G>A (p.Arg550His)	not provided	Uncertain significance(Last reviewed: Sep 4, 2020)	criteria provided, single submitter	VCV000995132	1	111777143	995132	982352	rs151164490	Neutral	0.446	0.209	0.345
KCND3	534	P	L	NM_001378969.1(KCND3):c.1601C>T (p.Pro534Leu)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Feb 7, 2020)	criteria provided, single submitter	VCV000465166	1	111777191	465166	446985	rs1553235768	Neutral	0.476	0.243	0.281
KCND3	547	R	H	NM_001378969.1(KCND3):c.1697G>A (p.Arg566His)	not provided	Uncertain significance(Last reviewed: May 20, 2019)	criteria provided, single submitter	VCV000804954	1	111777095	804954	792821	rs761867267	GOF	0.517	0.111	0.372
KCND3	549	R	C	NM_001378969.1(KCND3):c.1645C>T (p.Arg549Cys)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Jun 22, 2017)	criteria provided, single submitter	VCV000519470	1	111777147	519470	509062	rs1294150954	Neutral	0.57	0.149	0.281
KCND3	549	R	H	NM_001378969.1(KCND3):c.1646G>A (p.Arg549His)	Cardiovascular phenotype|Spinocerebellar ataxia type 19/22	Conflicting interpretations of pathogenicity(Last reviewed: Jul 12, 2021)	criteria provided, conflicting interpretations	VCV000264530	1	111777146	264530	257907	rs35027371	GOF	0.461	0.167	0.372
KCND3	556	T	M	NM_001378969.1(KCND3):c.1724C>T (p.Thr575Met)	not provided	Uncertain significance(Last reviewed: Mar 1, 2017)	criteria provided, single submitter	VCV000806186	1	111777068	806186	794371	rs775768536	GOF	0.412	0.191	0.396
KCND3	568	R	H	NM_001378969.1(KCND3):c.1703G>A (p.Arg568His)	Spinocerebellar ataxia type 19/22|not provided	Uncertain significance(Last reviewed: Nov 12, 2020)	criteria provided, multiple submitters, no conflicts	VCV000533722	1	111777089	533722	515008	rs200212002	GOF	0.52	0.175	0.304
KCND3	570	M	T	NM_001378969.1(KCND3):c.1709T>C (p.Met570Thr)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jun 23, 2017)	criteria provided, single submitter	VCV000465167	1	111777083	465167	447091	rs1553235743	GOF	0.473	0.176	0.351
KCND3	576	L	W	NM_001378969.1(KCND3):c.1784T>G (p.Leu595Trp)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Dec 5, 2018)	criteria provided, single submitter	VCV000655552	1	111776261	655552	626611	rs1483036958	LOF	0.698	0.142	0.16
KCND3	581	G	R	NM_001378969.1(KCND3):c.1798G>A (p.Gly600Arg)	Cardiovascular phenotype|Brugada syndrome 9| Spinocerebellar ataxia type 19/22|not provided	Uncertain significance(Last reviewed: Apr 7, 2021)	criteria provided, multiple submitters, no	VCV000192254	1	111776247	192254	190051	rs149344567	GOF	0.479	0.111	0.41

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCND3	581	S	C	NM_001378969.1(KCND3):c.1741A>T (p.Ser581Cys)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Apr 10, 2018)	criteria provided, single submitter	VCV000577665	1	111777051	577665	556840	rs1420542041	GOF	0.532	0.182	0.286
KCND3	586	L	V	NM_001378969.1(KCND3):c.1756C>G (p.Leu586Val)	Brugada syndrome 9|Cardiovascular phenotype| Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: May 30, 2019)	criteria provided, multiple submitters, no conflicts	VCV000519297	1	111777036	519297	509061	rs778053688	GOF	0.477	0.19	0.333
KCND3	590	R	H	NM_001378969.1(KCND3):c.1769G>A (p.Arg590His)	Spinocerebellar ataxia type 19/22|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 17, 2019)	criteria provided, conflicting interpretations	VCV000586063	1	111776276	586063	576393	rs186194682	Neutral	0.468	0.197	0.335
KCND3	603	A	S	NM_001378969.1(KCND3):c.1864G>T (p.Ala622Ser)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jun 7, 2020)	criteria provided, single submitter	VCV001057886	1	111776181	1057886	1039358	NA	Neutral	0.405	0.316	0.279
KCND3	603	A	V	NM_001378969.1(KCND3):c.1865C>T (p.Ala622Val)	Spinocerebellar ataxia type 19/22	Likely benign(Last reviewed: Jun 17, 2020)	criteria provided, single submitter	VCV001140881	1	111776180	1140881	1109308	NA	Neutral	0.412	0.308	0.281
KCND3	608	G	R	NM_001378969.1(KCND3):c.1879G>A (p.Gly627Arg)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Dec 20, 2018)	criteria provided, single submitter	VCV000647173	1	111776166	647173	626610	rs372362132	GOF	0.524	0.108	0.368
KCND3	611	R	Q	NM_001378969.1(KCND3):c.1889G>A (p.Arg630Gln)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, single submitter	VCV000934670	1	111776156	934670	929963	rs774713377	LOF	0.818	0.106	0.077
KCND3	617	I	V	NM_001378969.1(KCND3):c.1849A>G (p.Ile617Val)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Sep 28, 2017)	criteria provided, single submitter	VCV000533723	1	111776196	533723	515041	rs948125814	GOF	0.595	0.141	0.264
KCND3	629	N	S	NM_001378969.1(KCND3):c.1943A>G (p.Asn648Ser)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Jul 15, 2019)	criteria provided, single submitter	VCV000954104	1	111776102	954104	941381	rs766562520	Neutral	0.377	0.434	0.189
KCND3	630	V	A	NM_001378969.1(KCND3):c.1946T>C (p.Val649Ala)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Dec 31, 2018)	criteria provided, single submitter	VCV001027158	1	111776099	1027158	1002041	rs760907112	GOF	0.585	0.17	0.246
KCND3	635	A	T	NM_001378969.1(KCND3):c.1960G>A (p.Ala654Thr)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Sep 16, 2019)	criteria provided, single submitter	VCV000934368	1	111776085	934368	929962	rs774711788	Neutral	0.381	0.415	0.204
KCND3	639	N	K	NM_001378969.1(KCND3):c.1917C>A (p.Asn639Lys)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Apr 12, 2017)	criteria provided, single submitter	VCV000465168	1	111776128	465168	447051	rs777172603	Neutral	0.49	0.329	0.181
KCND3	642	I	F	NM_001378969.1(KCND3):c.1924A>T (p.Ile642Phe)	Spinocerebellar ataxia type 19/22|Inborn genetic diseases	Uncertain significance(Last reviewed: Nov 21, 2017)	criteria provided, multiple submitters, no conflicts	VCV000533724	1	111776121	533724	514977	rs754759010	Neutral	0.542	0.231	0.228
KCND3	645	I	T	NM_001378969.1(KCND3):c.1934T>C (p.Ile645Thr)	Spinocerebellar ataxia type 19/22	Uncertain significance(Last reviewed: Feb 27, 2018)	criteria provided, single submitter	VCV000579054	1	111776111	579054	556587	rs1557929628	Neutral	0.563	0.211	0.226
KCNH1	3	M	I	NM_172362.3(KCNH1):c.9G>A (p.Met3Ile)	not provided	Uncertain significance(Last	criteria provided, single	VCV000806343	1	211133937	806343	794560	rs752274336	GOF	0.656	0.109	0.235

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: May 1, 2018)	submitter										
KCNH1	58	A	E	NM_172362.3(KCNH1):c.173C>A (p.Ala58Glu)	not provided	Uncertain significance(Last reviewed: Jul 1, 2020)	criteria provided, single submitter	VCV000932653	1	211107284	932653	920635	rs1691367220	GOF	0.615	0.055	0.33
KCNH1	82	K	R	NM_172362.3(KCNH1):c.245A>G (p.Lys82Arg)	Intellectual disability	Likely benign(Last reviewed: Jan 1, 2019)	no assertion criteria provided	VCV000975412	1	211103561	975412	963470	rs1558605689	GOF	0.531	0.062	0.407
KCNH1	84	R	Q	NM_172362.3(KCNH1):c.251G>A (p.Arg84Gln)	not provided	Likely benign(Last reviewed: Apr 1, 2020)	criteria provided, single submitter	VCV001205093	1	211103555	1205093	1192762	NA	GOF	0.556	0.053	0.391
KCNH1	95	F	C	NM_172362.3(KCNH1):c.284T>G (p.Phe95Cys)	not provided	Uncertain significance(Last reviewed: Jul 1, 2018)	criteria provided, single submitter	VCV000806342	1	211103522	806342	794559	rs1571649012	LOF	0.862	0.055	0.083
KCNH1	152	A	P	NM_172362.3(KCNH1):c.454G>C (p.Ala152Pro)	not provided	Uncertain significance(Last reviewed: Mar 11, 2019)	criteria provided, single submitter	VCV001307611	1	211082884	1307611	1298881	NA	GOF	0.626	0.081	0.292
KCNH1	178	V	I	NM_172362.3(KCNH1):c.532G>A (p.Val178Ile)	not provided	Uncertain significance(Last reviewed: Sep 9, 2019)	criteria provided, single submitter	VCV001312106	1	211082806	1312106	1302637	NA	LOF	0.601	0.137	0.262
KCNH1	182	S	T	NM_172362.3(KCNH1):c.544T>A (p.Ser182Thr)	Zimmermann-Laband syndrome 1	Uncertain significance(Last reviewed: Nov 3, 2021)	criteria provided, single submitter	VCV001319807	1	211082794	1319807	1310056	NA	GOF	0.513	0.101	0.386
KCNH1	199	K	R	NM_172362.3(KCNH1):c.596A>G (p.Lys199Arg)	KCNH1-related phenotype	Likely pathogenic(Last reviewed: Jun 22, 2018)	criteria provided, single submitter	VCV000560720	1	211019219	560720	551755	rs1558570102	GOF	0.526	0.095	0.378
KCNH1	217	K	N	NM_172362.3(KCNH1):c.651G>C (p.Lys217Asn)	Temple-Baraitser syndrome	Pathogenic(Last reviewed: Jan 1, 2015)	no assertion criteria provided	VCV000162523	1	211019164	162523	172196	rs727502822	GOF	0.484	0.026	0.49
KCNH1	225	L	F	NM_172362.3(KCNH1):c.675G>T (p.Leu225Phe)	not provided	Uncertain significance(Last reviewed: Sep 16, 2018)	no assertion criteria provided	VCV000591989	1	211019140	591989	582978	rs1558570068	GOF	0.596	0.036	0.368
KCNH1	288	R	H	NM_172362.3(KCNH1):c.863G>A (p.Arg288His)	not provided	Uncertain significance(Last reviewed: Apr 11, 2019)	criteria provided, single submitter	VCV001308467	1	211018952	1308467	1297088	NA	GOF	0.523	0.027	0.45
KCNH1	294	T	M	NM_172362.3(KCNH1):c.881C>T (p.Thr294Met)	not provided	Likely pathogenic(Last reviewed: Dec 5, 2018)	criteria provided, single submitter	VCV000633559	1	211018934	633559	621937	rs1558569964	GOF	0.566	0.026	0.408
KCNH1	297	V	M	NM_172362.3(KCNH1):c.889G>A (p.Val297Met)	not provided	Uncertain significance(Last reviewed: Jun 11, 2019)	criteria provided, single submitter	VCV001306445	1	211018926	1306445	1296388	NA	GOF	0.69	0.043	0.268
KCNH1	325	S	Y	NM_172362.3(KCNH1):c.1055C>A (p.Ser352Tyr)	Zimmermann-Laband syndrome 1	Pathogenic	no assertion criteria provided	VCV000183419	1	210920047	183419	181521	rs730882172	GOF	0.469	0.059	0.472
KCNH1	333	R	C	NM_172362.3(KCNH1):c.1078C>T (p.Arg360Cys)	not provided	Pathogenic(Last reviewed: Jan 19, 2022)	criteria provided, single submitter	VCV001190684	1	210920024	1190684	1179170	NA	GOF	0.599	0.123	0.278
KCNH1	341	R	K	NM_172362.3(KCNH1):c.1022G>A (p.Arg341Lys)	not provided	Uncertain significance(Last reviewed: Aug 8, 2019)	criteria provided, single submitter	VCV001307585	1	211018793	1307585	1298855	NA	GOF	0.532	0.117	0.351
KCNH1	345	G	A	NM_172362.3(KCNH1):c.1034G>C	Zimmermann-Laband syndrome 1|Temple-	Uncertain	criteria	VCV000587447	1	210920068	587447	578375	rs1558526097	GOF	0.539	0.061	0.401

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Gly345Ala)	Baraitser syndrome	significance(Last reviewed: Aug 7, 2018)	provided, single submitter										
KCNH1	348	G	R	NM_172362.3(KCNH1):c.1123G>A (p.Gly375Arg)	Zimmermann-Laband syndrome 1	Pathogenic(Last reviewed: Jun 1, 2015)	no assertion criteria provided	VCV000183415	1	210919979	183415	181520	rs730882174	GOF	0.451	0.03	0.519
KCNH1	352	L	V	NM_172362.3(KCNH1):c.1135C>G (p.Leu379Val)	Zimmermann-Laband syndrome 1	Pathogenic(Last reviewed: Jun 1, 2015)	no assertion criteria provided	VCV000183416	1	210919967	183416	181519	rs730882176	GOF	0.477	0.045	0.478
KCNH1	354	K	N	NM_172362.3(KCNH1):c.1062A>C (p.Lys354Asn)	not provided	Likely pathogenic(Last reviewed: Apr 12, 2016)	criteria provided, single submitter	VCV000420958	1	210920040	420958	405027	rs1064794817	GOF	0.467	0.023	0.509
KCNH1	356	V	L	NM_172362.3(KCNH1):c.1147G>C (p.Val383Leu)	Zimmermann-Laband syndrome 1	Pathogenic	no assertion criteria provided	VCV000183417	1	210919955	183417	181518	rs730882173	GOF	0.477	0.045	0.478
KCNH1	356	V	Y	NM_002238.3(KCNH1):c.[1066G>C;974C>A]	Zimmermann-Laband syndrome 1	Pathogenic(Last reviewed: Jun 1, 2015)	no assertion criteria provided	VCV000203434	1|1	210919955	203434	181518|181521	rs730882173|rs730882172	GOF	0.476	0.036	0.488
KCNH1	357	R	Q	NM_172362.3(KCNH1):c.1070G>A (p.Arg357Gln)	Intellectual disability, severe|Abnormal facial shape|Seizures|not provided|KCNH1-related disorders|Inborn genetic diseases|Zimmermann-Laband syndrome 1|Temple-Baraitser syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jan 3, 2022)	criteria provided, multiple submitters, no conflicts	VCV000279981	1	210920032	279981	263996	rs886041300	GOF	0.389	0.02	0.591
KCNH1	367	A	T	NM_172362.3(KCNH1):c.1180G>A (p.Ala394Thr)	Zimmermann-Laband syndrome 1|Temple-Baraitser syndrome	Uncertain significance	criteria provided, single submitter	VCV001339058	1	210919922	1339058	1330101	NA	GOF	0.458	0.042	0.5
KCNH1	379	L	P	NM_172362.3(KCNH1):c.1136T>C (p.Leu379Pro)	not provided|Neurodevelopmental disorder	Pathogenic/Likely pathogenic(Last reviewed: Oct 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000235383	1	210919966	235383	237069	rs878853026	LOF	0.738	0.025	0.238
KCNH1	469	G	R	NM_172362.3(KCNH1):c.1486G>A (p.Gly496Arg)	Zimmermann-Laband syndrome 1|See cases|not provided|Inborn genetic diseases	Pathogenic/Likely pathogenic(Last reviewed: Jan 6, 2022)	criteria provided, multiple submitters, no conflicts	VCV000183418	1	210804143	183418	181517	rs730882175	GOF	0.481	0.033	0.485
KCNH1	471	V	A	NM_172362.3(KCNH1):c.1493T>C (p.Val498Ala)	Temple-Baraitser syndrome	Uncertain significance(Last reviewed: Dec 4, 2020)	criteria provided, single submitter	VCV001285479	1	210804136	1285479	1275322	NA	GOF	0.652	0.035	0.313
KCNH1	489	L	F	NM_172362.3(KCNH1):c.1465C>T (p.Leu489Phe)	not provided	Pathogenic(Last reviewed: Aug 22, 2017)	criteria provided, single submitter	VCV000449572	1	210804164	449572	442707	rs1553345948	GOF	0.478	0.042	0.48
KCNH1	489	L	F	NM_172362.3(KCNH1):c.1465C>T (p.Leu489Phe)	not provided	Pathogenic(Last reviewed: Aug 22, 2017)	criteria provided, single submitter	VCV000449572	1	210804164	449572	442707	rs1553345948	GOF	0.478	0.042	0.48
KCNH1	489	L	F	NM_172362.3(KCNH1):c.1546C>T (p.Leu516Phe)	Temple-Baraitser syndrome	Pathogenic(Last reviewed: Jan 1, 2015)	no assertion criteria provided	VCV000162521	1	210804083	162521	172194	rs727502820	GOF	0.478	0.042	0.48
KCNH1	489	L	F	NM_172362.3(KCNH1):c.1546C>T (p.Leu516Phe)	Temple-Baraitser syndrome	Pathogenic(Last reviewed: Jan 1, 2015)	no assertion criteria provided	VCV000162521	1	210804083	162521	172194	rs727502820	GOF	0.478	0.042	0.48
KCNH1	493	T	N	NM_172362.3(KCNH1):c.1478C>A (p.Thr493Asn)	not provided	Likely pathogenic(Last reviewed: Jul 1, 2018)	criteria provided, single submitter	VCV000807914	1	210804151	807914	794558	rs1574266171	GOF	0.56	0.023	0.417
KCNH1	494	I	V	NM_172362.3(KCNH1):c.1480A>G (p.Ile494Val)	Temple-Baraitser syndrome|Zimmermann-Laband syndrome 1|not provided|Zimmermann-Laband syndrome 1|Temple-Baraitser syndrome	Pathogenic(Last reviewed: Aug 4, 2017)	criteria provided, single submitter	VCV000162520	1	210804149	162520	172193	rs727502819	GOF	0.475	0.045	0.479
KCNH1	502	F	S	NM_172362.3(KCNH1):c.1505T>C	Inborn genetic diseases	Uncertain	criteria	VCV000521999	1	210804124	521999	511216	rs1553345934	GOF	0.702	0.024	0.274

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Phe502Ser)		significance(Last reviewed: Aug 7, 2017)	provided, single submitter										
KCNH1	503	Q	R	NM_172362.3(KCNH1):c.1508A>G (p.Gln503Arg)	Temple-Baraitser syndrome	Pathogenic(Last reviewed: Jan 1, 2015)	no assertion criteria provided	VCV000162522	1	210804121	162522	172195	rs727502821	GOF	0.448	0.033	0.519
KCNH1	514	V	L	NM_172362.3(KCNH1):c.1621G>C (p.Val541Leu)	not provided	Uncertain significance(Last reviewed: Dec 9, 2019)	criteria provided, single submitter	VCV001310940	1	210804008	1310940	1300321	NA	GOF	0.529	0.073	0.398
KCNH1	520	R	Q	NM_172362.3(KCNH1):c.1559G>A (p.Arg520Gln)	Zimmermann-Laband syndrome 1	Likely pathogenic(Last reviewed: Aug 6, 2018)	criteria provided, single submitter	VCV001034355	1	210804070	1034355	1019279	rs1684483227	GOF	0.527	0.037	0.436
KCNH1	531	I	V	NM_172362.3(KCNH1):c.1672A>G (p.Ile558Val)	not provided	Uncertain significance(Last reviewed: Jun 25, 2019)	criteria provided, single submitter	VCV001180372	1	210797751	1180372	1168809	NA	GOF	0.579	0.068	0.352
KCNH1	546	R	H	NM_172362.3(KCNH1):c.1718G>A (p.Arg573His)	not provided	Uncertain significance(Last reviewed: Aug 4, 2020)	criteria provided, single submitter	VCV001195149	1	210797705	1195149	1182832	NA	GOF	0.559	0.055	0.386
KCNH1	556	R	Q	NM_172362.3(KCNH1):c.1748G>A (p.Arg583Gln)	not provided	Uncertain significance(Last reviewed: Oct 12, 2020)	criteria provided, single submitter	VCV001313312	1	210797675	1313312	1303573	NA	GOF	0.595	0.031	0.374
KCNH1	569	V	M	NM_172362.3(KCNH1):c.1705G>A (p.Val569Met)	Inborn genetic diseases	Likely pathogenic(Last reviewed: Apr 27, 2015)	criteria provided, single submitter	VCV000520561	1	210797718	520561	511215	rs1553344875	GOF	0.531	0.046	0.423
KCNH1	573	R	C	NM_172362.3(KCNH1):c.1717C>T (p.Arg573Cys)	Zimmermann-Laband syndrome 1	Uncertain significance(Last reviewed: May 10, 2019)	criteria provided, single submitter	VCV000930399	1	210797706	930399	918596	rs1684345922	GOF	0.661	0.038	0.302
KCNH1	626	T	I	NM_172362.3(KCNH1):c.1958C>T (p.Thr653Ile)	Zimmermann-Laband syndrome 1	Uncertain significance(Last reviewed: Oct 2, 2019)	criteria provided, single submitter	VCV000930362	1	210775502	930362	918595	rs1683842802	LOF	0.873	0.05	0.078
KCNH1	658	T	M	NM_172362.3(KCNH1):c.2054C>T (p.Thr685Met)	Temple-Baraitser syndrome|not provided	Uncertain significance(Last reviewed: Jun 4, 2021)	criteria provided, multiple submitters, no conflicts	VCV001213775	1	210775406	1213775	1203768	NA	GOF	0.572	0.075	0.353
KCNH1	709	P	A	NM_172362.3(KCNH1):c.2206C>G (p.Pro736Ala)	Temple-Baraitser syndrome	Uncertain significance(Last reviewed: Oct 3, 2019)	criteria provided, single submitter	VCV001029267	1	210684045	1029267	1015634	rs1681345673	LOF	0.737	0.12	0.143
KCNH1	711	R	W	NM_172362.3(KCNH1):c.2212C>T (p.Arg738Trp)	Inborn genetic diseases	Uncertain significance(Last reviewed: Jul 31, 2018)	criteria provided, single submitter	VCV000986004	1	210684039	986004	973171	rs908699888	LOF	0.753	0.112	0.134
KCNH1	728	E	D	NM_172362.3(KCNH1):c.2265G>C (p.Glu755Asp)	Temple-Baraitser syndrome	Likely benign	no assertion criteria provided	VCV000689343	1	210683986	689343	676935	rs776649794	Neutral	0.651	0.209	0.139
KCNH1	732	R	Q	NM_172362.3(KCNH1):c.2276G>A (p.Arg759Gln)	not provided	Benign/Likely benign(Last reviewed: Sep 3, 2019)	criteria provided, multiple submitters, no conflicts	VCV000755041	1	210683975	755041	761535	rs141905481	Neutral	0.665	0.185	0.15
KCNH1	734	D	E	NM_172362.3(KCNH1):c.2202C>A (p.Asp734Glu)	not provided	Uncertain significance(Last reviewed: Jan 8,	criteria provided, single submitter	VCV000852617	1	210684049	852617	823310	rs1226108094	Neutral	0.656	0.2	0.145

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)											
KCNH1	749	A	G	NM_172362.3(KCNH1):c.2327C>G (p.Ala776Gly)	not provided	Benign/Likely benign(Last reviewed: Dec 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV001124856	1	210683924	1124856	1109980	NA	Neutral	0.669	0.196	0.136
KCNH1	764	R	H	NM_172362.3(KCNH1):c.2372G>A (p.Arg791His)	not provided	Benign(Last reviewed: Jan 22, 2021)	criteria provided, single submitter	VCV001247790	1	210683879	1247790	1237744	NA	Neutral	0.685	0.188	0.127
KCNH1	785	A	T	NM_172362.3(KCNH1):c.2434G>A (p.Ala812Thr)	not provided	Benign/Likely benign(Last reviewed: Feb 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000717772	1	210683817	717772	718601	rs759775665	GOF	0.486	0.188	0.326
KCNH1	792	S	T	NM_172362.3(KCNH1):c.2455T>A (p.Ser819Thr)	not provided	Benign(Last reviewed: Dec 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000707060	1	210683796	707060	690478	rs139659266	GOF	0.4	0.173	0.427
KCNH1	794	C	Y	NM_172362.3(KCNH1):c.2462G>A (p.Cys821Tyr)	not provided	Likely benign(Last reviewed: May 21, 2020)	criteria provided, single submitter	VCV001181381	1	210683789	1181381	1170657	NA	Neutral	0.549	0.192	0.259
KCNH1	799	G	V	NM_172362.3(KCNH1):c.2477G>T (p.Gly826Val)	not provided	Benign(Last reviewed: Aug 9, 2019)	criteria provided, single submitter	VCV001250452	1	210683774	1250452	1239539	NA	GOF	0.485	0.206	0.309
KCNH1	801	G	R	NM_172362.3(KCNH1):c.2482G>A (p.Gly828Arg)	not provided|Zimmermann-Laband syndrome 1| Temple-Baraitser syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Mar 19, 2021)	criteria provided, conflicting interpretations	VCV001216384	1	210683769	1216384	1206364	NA	GOF	0.52	0.163	0.317
KCNH1	802	G	D	NM_172362.3(KCNH1):c.2486G>A (p.Gly829Asp)	not provided	Uncertain significance(Last reviewed: Jul 17, 2019)	criteria provided, single submitter	VCV001304074	1	210683765	1304074	1294349	NA	Neutral	0.581	0.174	0.245
KCNH1	841	A	V	NM_172362.3(KCNH1):c.2603C>T (p.Ala868Val)	not provided	Uncertain significance(Last reviewed: Nov 1, 2019)	criteria provided, single submitter	VCV000807913	1	210683648	807913	794557	rs773735492	Neutral	0.498	0.218	0.284
KCNH1	896	Q	P	NM_172362.3(KCNH1):c.2768A>C (p.Gln923Pro)	not provided	Benign/Likely benign(Last reviewed: Dec 31, 2019)	criteria provided, multiple submitters, no conflicts	VCV000778789	1	210683483	778789	696435	rs140939826	GOF	0.495	0.193	0.312
KCNH1	903	R	G	NM_172362.3(KCNH1):c.2788A>G (p.Arg930Gly)	not provided	Uncertain significance(Last reviewed: Jun 10, 2019)	criteria provided, single submitter	VCV001306379	1	210683463	1306379	1296322	NA	Neutral	0.548	0.216	0.236
KCNH1	915	A	T	NM_172362.3(KCNH1):c.2824G>A (p.Ala942Thr)	not provided	Likely benign(Last reviewed: Aug 14, 2020)	criteria provided, single submitter	VCV001213227	1	210683427	1213227	1203221	NA	GOF	0.482	0.162	0.356
KCNH1	954	S	A	NM_172362.3(KCNH1):c.2941T>G (p.Ser981Ala)	Intellectual disability	Likely benign(Last reviewed: Jan 1, 2019)	no assertion criteria provided	VCV000975411	1	210683310	975411	963469	rs1681318890	LOF	0.693	0.099	0.209
KCNH1	961	A	S	NM_172362.3(KCNH1):c.2962G>T (p.Ala988Ser)	not provided	Benign/Likely benign(Last reviewed: Oct 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000710791	1	210683289	710791	718600	rs371262198	GOF	0.53	0.156	0.314
KCNH2	1	M	L	NM_000238.4(KCNH2):c.1A>T (p.Met1Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067354	7	150977913	67354	78250	rs199473036	Neutral	0.562	0.248	0.19

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	2	A	V	NM_000238.4(KCNH2):c.1128+1792C>T	Long QT syndrome	Uncertain significance(Last reviewed: Aug 31, 2016)	criteria provided, single submitter	VCV000405348	7	150955499	405348	396180	rs1060500667	Neutral	0.52	0.291	0.189
KCNH2	6	G	R	NM_000238.4(KCNH2):c.1128+1803G>A	Arrhythmia	Likely benign(Last reviewed: Apr 30, 2018)	criteria provided, single submitter	VCV000630104	7	150955488	630104	619280	rs553291120	LOF	0.704	0.082	0.214
KCNH2	6	G	W	NM_000238.4(KCNH2):c.1128+1803G>T	Arrhythmia	Likely benign(Last reviewed: Jul 26, 2019)	criteria provided, single submitter	VCV000925674	7	150955488	925674	915425	rs553291120	LOF	0.698	0.124	0.179
KCNH2	8	A	V	NM_000238.4(KCNH2):c.1128+1810C>T	Short QT syndrome 1|Long QT syndrome 2|Long QT syndrome|Arrhythmia|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 28, 2020)	criteria provided, conflicting interpretations	VCV000379235	7	150955481	379235	369621	rs972201049	Neutral	0.636	0.252	0.112
KCNH2	9	A	V	NM_000238.4(KCNH2):c.26C>T (p.Ala9Val)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 4, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200592	7	150977888	200592	197374	rs775317201	LOF	0.769	0.128	0.103
KCNH2	10	P	L	NM_000238.4(KCNH2):c.29C>T (p.Pro10Leu)	Long QT syndrome	Uncertain significance(Last reviewed: May 2, 2019)	criteria provided, single submitter	VCV000946979	7	150977885	946979	924765	rs1383782501	LOF	0.733	0.142	0.125
KCNH2	11	Q	R	NM_000238.4(KCNH2):c.32A>G (p.Gln11Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 28, 2020)	criteria provided, single submitter	VCV001063296	7	150977882	1063296	1044817	NA	LOF	0.703	0.098	0.199
KCNH2	12	G	V	NM_000238.4(KCNH2):c.1128+1822G>T	not provided|Long QT syndrome	Likely benign(Last reviewed: Feb 9, 2018)	criteria provided, single submitter	VCV000697451	7	150955469	697451	689864	rs781494018	LOF	0.773	0.099	0.128
KCNH2	12	G	W	NM_000238.4(KCNH2):c.1128+1821G>T	Arrhythmia	Likely benign(Last reviewed: Dec 3, 2018)	criteria provided, single submitter	VCV000926092	7	150955470	926092	915893	rs1801336913	LOF	0.745	0.09	0.165
KCNH2	13	T	N	NM_000238.4(KCNH2):c.38C>A (p.Thr13Asn)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000923223	7	150977876	923223	910811	rs758978727	LOF	0.808	0.087	0.105
KCNH2	15	L	Q	NM_000238.4(KCNH2):c.44T>A (p.Leu15Gln)	not provided	Uncertain significance(Last reviewed: Mar 20, 2018)	criteria provided, single submitter	VCV000200594	7	150977870	200594	197372	rs794728418	LOF	0.894	0.036	0.07
KCNH2	15	L	R	NM_000238.4(KCNH2):c.44T>G (p.Leu15Arg)	not provided	Uncertain significance(Last reviewed: Sep 16, 2020)	criteria provided, single submitter	VCV001206671	7	150977870	1206671	1197659	NA	LOF	0.881	0.033	0.085
KCNH2	15	R	W	NM_000238.4(KCNH2):c.1128+1830C>T	Long QT syndrome|Arrhythmia	Likely benign(Last reviewed: Nov 29, 2019)	criteria provided, multiple submitters, no conflicts	VCV000700755	7	150955461	700755	689863	rs778819756	LOF	0.853	0.047	0.1
KCNH2	16	D	A	NM_000238.4(KCNH2):c.47A>C (p.Asp16Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067507	7	150977867	67507	78403	rs199472825	LOF	0.86	0.042	0.097
KCNH2	17	R	S	NM_000238.4(KCNH2):c.1128+1838G>C	Arrhythmia	Likely benign(Last reviewed: Dec 16, 2018)	criteria provided, single submitter	VCV000921782	7	150955453	921782	915887	rs1310476998	LOF	0.883	0.045	0.072
KCNH2	19	I	F	NM_000238.4(KCNH2):c.55A>T (p.Ile19Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 19, 2018)	criteria provided, single submitter	VCV000527023	7	150977859	527023	522804	rs1554431444	LOF	0.852	0.058	0.089

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	19	I	S	NM_000238.4(KCNH2):c.56T>G (p.Ile19Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 27, 2019)	criteria provided, single submitter	VCV000949641	7	150977858	949641	924764	rs1802016246	LOF	0.891	0.034	0.074
KCNH2	20	R	G	NM_000238.4(KCNH2):c.58C>G (p.Arg20Gly)	not provided|Congenital long QT syndrome	Uncertain significance(Last reviewed: Apr 22, 2020)	criteria provided, single submitter	VCV000067513	7	150977856	67513	78409	rs199473486	LOF	0.804	0.071	0.126
KCNH2	21	G	D	NM_000238.4(KCNH2):c.1128+1849G>A	Long QT syndrome	Uncertain significance(Last reviewed: Dec 29, 2017)	criteria provided, single submitter	VCV000526911	7	150955442	526911	522779	rs1167016668	LOF	0.848	0.034	0.118
KCNH2	22	F	S	NM_000238.4(KCNH2):c.65T>C (p.Phe22Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067517	7	150977849	67517	78413	rs199472826	LOF	0.881	0.043	0.076
KCNH2	22	R	Q	NM_000238.4(KCNH2):c.1128+1852G>A	Long QT syndrome|Arrhythmia	Likely benign(Last reviewed: Oct 16, 2018)	criteria provided, multiple submitters, no conflicts	VCV000527075	7	150955439	527075	523141	rs780068180	LOF	0.874	0.037	0.089
KCNH2	22	R	W	NM_000238.4(KCNH2):c.1128+1851C>T	not provided|Long QT syndrome	Likely benign(Last reviewed: Jul 20, 2018)	criteria provided, single submitter	VCV000728449	7	150955440	728449	744317	rs1584860856	LOF	0.835	0.053	0.111
KCNH2	23	V	M	NM_000238.4(KCNH2):c.1128+1854G>A	Long QT syndrome|not provided	Likely benign(Last reviewed: Apr 25, 2018)	criteria provided, single submitter	VCV000701801	7	150955437	701801	689862	rs995080056	LOF	0.776	0.089	0.135
KCNH2	25	R	Q	NM_000238.4(KCNH2):c.1128+1861G>A	Arrhythmia	Likely benign(Last reviewed: Jul 3, 2018)	criteria provided, single submitter	VCV000630562	7	150955430	630562	619374	rs41311009	LOF	0.731	0.105	0.164
KCNH2	25	R	W	NM_000238.4(KCNH2):c.1128+1860C>T	not provided|Arrhythmia|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Nov 5, 2020)	criteria provided, conflicting interpretations	VCV000661053	7	150955431	661053	635915	rs767264288	Neutral	0.682	0.151	0.168
KCNH2	26	S	G	NM_000238.4(KCNH2):c.76A>G (p.Ser26Gly)	Arrhythmia	Uncertain significance(Last reviewed: Nov 18, 2020)	criteria provided, single submitter	VCV000928116	7	150977838	928116	910809	rs1802015752	LOF	0.75	0.117	0.133
KCNH2	26	S	I	NM_000238.4(KCNH2):c.77G>T (p.Ser26Ile)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067528	7	150974941	67528	78424	rs199472827	Neutral	0.688	0.164	0.148
KCNH2	27	R	H	NM_000238.4(KCNH2):c.80G>A (p.Arg27His)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jun 29, 2019)	criteria provided, conflicting interpretations	VCV000200540	7	150974938	200540	197368	rs199472828	LOF	0.749	0.119	0.132
KCNH2	27	R	P	NM_000238.4(KCNH2):c.80G>C (p.Arg27Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067530	7	150974938	67530	78426	rs199472828	LOF	0.766	0.104	0.129
KCNH2	27	V	L	NM_000238.4(KCNH2):c.1128+1866G>T	Arrhythmia	Likely benign(Last reviewed: Jul 2, 2019)	criteria provided, single submitter	VCV000921664	7	150955425	921664	915423	rs764462112	Neutral	0.714	0.164	0.122
KCNH2	28	K	E	NM_000238.4(KCNH2):c.82A>G (p.Lys28Glu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067532	7	150974936	67532	78428	rs199472829	LOF	0.855	0.05	0.095
KCNH2	28	R	C	NM_000238.4(KCNH2):c.1128+1869C>T	Long QT syndrome|Arrhythmia	Likely benign(Last reviewed: Jun 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000695712	7	150955422	695712	685211	rs62492438	LOF	0.866	0.054	0.08
KCNH2	28	R	H	NM_000238.4(KCNH2):c.1128+1870G>A	Arrhythmia	Likely benign(Last reviewed: Nov 8, 2018)	criteria provided, single submitter	VCV000922307	7	150955421	922307	915889	rs1219643674	LOF	0.864	0.048	0.088
KCNH2	29	F	L	NM_000238.4(KCNH2):c.87C>A (p.Phe29Leu)	Congenital long QT syndrome|Long QT	Pathogenic(Last	criteria	VCV000067538	7	150974931	67538	78434	rs199472830	LOF	0.856	0.048	0.096

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					syndrome	reviewed: Jun 29, 2020)	provided, single submitter										
KCNH2	29	F	S	NM_000238.4(KCNH2):c.86T>C (p.Phe29Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067536	7	150974932	67536	78432	rs199472831	LOF	0.859	0.046	0.095
KCNH2	29	I	F	NM_000238.4(KCNH2):c.1128+1872A>T	Arrhythmia	Likely benign(Last reviewed: May 20, 2019)	criteria provided, single submitter	VCV000918543	7	150955419	918543	916160	rs1801332643	LOF	0.867	0.046	0.087
KCNH2	30	I	T	NM_000238.4(KCNH2):c.89T>C (p.Ile30Thr)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Apr 11, 2018)	criteria provided, single submitter	VCV000067541	7	150974929	67541	78437	rs199472832	LOF	0.858	0.045	0.097
KCNH2	31	I	M	NM_000238.4(KCNH2):c.93C>G (p.Ile31Met)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 14, 2019)	criteria provided, single submitter	VCV000839172	7	150974925	839172	833364	rs745885744	LOF	0.865	0.045	0.09
KCNH2	31	I	N	NM_000238.4(KCNH2):c.92T>A (p.Ile31Asn)	Long QT syndrome 2	Likely pathogenic(Last reviewed: Feb 14, 2018)	criteria provided, single submitter	VCV000520423	7	150974926	520423	511026	rs199472833	LOF	0.864	0.045	0.091
KCNH2	31	I	S	NM_000238.4(KCNH2):c.92T>G (p.Ile31Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067546	7	150974926	67546	78442	rs199472833	LOF	0.859	0.045	0.096
KCNH2	31	I	T	NM_000238.4(KCNH2):c.92T>C (p.Ile31Thr)	not provided|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Jan 12, 2021)	criteria provided, single submitter	VCV000067545	7	150974926	67545	78441	rs199472833	LOF	0.858	0.044	0.098
KCNH2	32	A	T	NM_000238.4(KCNH2):c.94G>A (p.Ala32Thr)	Congenital long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 6, 2020)	criteria provided, single submitter	VCV000067549	7	150974924	67549	78445	rs199472834	LOF	0.867	0.045	0.088
KCNH2	32	A	V	NM_000238.4(KCNH2):c.95C>T (p.Ala32Val)	Long QT syndrome 2	Pathogenic(Last reviewed: Oct 12, 2019)	criteria provided, single submitter	VCV000869439	7	150974923	869439	857648	rs1801941580	LOF	0.869	0.047	0.084
KCNH2	33	N	T	NM_000238.4(KCNH2):c.98A>C (p.Asn33Thr)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Apr 29, 2021)	criteria provided, conflicting interpretations	VCV000067553	7	150974920	67553	78449	rs199473487	LOF	0.852	0.051	0.097
KCNH2	33	V	M	NM_000238.4(KCNH2):c.1128+1884G>A	Long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Feb 16, 2019)	criteria provided, conflicting interpretations	VCV000654625	7	150955407	654625	651783	rs765679790	LOF	0.831	0.078	0.091
KCNH2	34	A	V	NM_000238.4(KCNH2):c.1128+1888C>T	Long QT syndrome	Likely benign(Last reviewed: Jul 21, 2017)	criteria provided, single submitter	VCV000456881	7	150955403	456881	457103	rs1031478932	LOF	0.863	0.039	0.098
KCNH2	36	E	D	NM_000238.4(KCNH2):c.1128+1895G>C	Long QT syndrome	Uncertain significance(Last reviewed: Apr 19, 2017)	criteria provided, single submitter	VCV000405347	7	150955396	405347	396175	rs1060500666	LOF	0.832	0.047	0.12
KCNH2	39	C	R	NM_000238.4(KCNH2):c.115T>C (p.Cys39Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 10, 2017)	criteria provided, single submitter	VCV000456883	7	150974903	456883	456891	rs757491162	LOF	0.81	0.038	0.152
KCNH2	40	A	V	NM_000238.4(KCNH2):c.119C>T (p.Ala40Val)	not provided	Likely pathogenic(Last reviewed: Sep 30, 2013)	criteria provided, single submitter	VCV000200544	7	150974899	200544	197364	rs794728407	LOF	0.871	0.047	0.082
KCNH2	41	V	A	NM_000238.4(KCNH2):c.122T>C (p.Val41Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067169	7	150974896	67169	78065	rs731506	LOF	0.867	0.044	0.089

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	41	V	F	NM_000238.4(KCNH2):c.121G>T (p.Val41Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067167	7	150974897	67167	78063	rs199472835	LOF	0.873	0.044	0.083
KCNH2	41	V	L	NM_000238.4(KCNH2):c.121G>C (p.Val41Leu)	not provided	Likely pathogenic(Last reviewed: Apr 9, 2015)	criteria provided, single submitter	VCV000372589	7	150974897	372589	359813	rs199472835	LOF	0.868	0.046	0.086
KCNH2	42	I	N	NM_000238.4(KCNH2):c.125T>A (p.Ile42Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067172	7	150974893	67172	78068	rs199473488	LOF	0.868	0.045	0.086
KCNH2	42	I	T	NM_000238.4(KCNH2):c.125T>C (p.Ile42Thr)	not provided	Pathogenic(Last reviewed: Mar 15, 2013)	criteria provided, single submitter	VCV000200546	7	150974893	200546	197363	rs199473488	LOF	0.863	0.044	0.093
KCNH2	43	D	E	NM_000238.4(KCNH2):c.1149C>A (p.Asp383Glu)	Arrhythmia	Uncertain significance(Last reviewed: Nov 9, 2018)	criteria provided, single submitter	VCV000925167	7	150952833	925167	910780	rs201995634	LOF	0.867	0.054	0.079
KCNH2	43	Y	C	NM_000238.4(KCNH2):c.128A>G (p.Tyr43Cys)	Congenital long QT syndrome|not provided|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: May 21, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067182	7	150974890	67182	78078	rs199472836	LOF	0.857	0.047	0.096
KCNH2	43	Y	D	NM_000238.4(KCNH2):c.127T>G (p.Tyr43Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067177	7	150974891	67177	78073	rs199472837	LOF	0.872	0.046	0.082
KCNH2	44	C	F	NM_000238.4(KCNH2):c.131G>T (p.Cys44Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067186	7	150974887	67186	78082	rs199473489	LOF	0.87	0.052	0.078
KCNH2	44	C	W	NM_000238.4(KCNH2):c.132C>G (p.Cys44Trp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067187	7	150974886	67187	78083	rs199472838	LOF	0.869	0.055	0.075
KCNH2	44	C	Y	NM_000238.4(KCNH2):c.131G>A (p.Cys44Tyr)	not provided	Likely pathogenic(Last reviewed: Jul 23, 2021)	criteria provided, single submitter	VCV001214199	7	150974887	1214199	1204184	NA	LOF	0.869	0.051	0.08
KCNH2	45	N	K	NM_000238.4(KCNH2):c.135C>A (p.Asn45Lys)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 6, 2018)	criteria provided, single submitter	VCV000569898	7	150974883	569898	564168	rs1422251865	LOF	0.812	0.027	0.161
KCNH2	45	N	T	NM_000238.4(KCNH2):c.134A>C (p.Asn45Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 5, 2019)	criteria provided, single submitter	VCV000949254	7	150974884	949254	924763	rs1801939758	LOF	0.854	0.037	0.109
KCNH2	45	N	Y	NM_000238.4(KCNH2):c.133A>T (p.Asn45Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067189	7	150974885	67189	78085	rs199472839	LOF	0.833	0.038	0.129
KCNH2	46	D	E	NM_000238.4(KCNH2):c.138C>G (p.Asp46Glu)	Arrhythmia	Uncertain significance(Last reviewed: May 30, 2019)	criteria provided, single submitter	VCV000921751	7	150974880	921751	910805	rs752503743	LOF	0.846	0.021	0.134
KCNH2	46	D	G	NM_000238.4(KCNH2):c.137A>G (p.Asp46Gly)	not provided	Uncertain significance(Last reviewed: Mar 25, 2021)	criteria provided, single submitter	VCV001303967	7	150974881	1303967	1294242	NA	LOF	0.865	0.026	0.109
KCNH2	46	D	N	NM_000238.4(KCNH2):c.136G>A (p.Asp46Asn)	not provided	Likely pathogenic(Last reviewed: Mar 9, 2015)	criteria provided, single submitter	VCV000200551	7	150974882	200551	197361	rs794728408	LOF	0.856	0.022	0.122
KCNH2	47	G	C	NM_000238.4(KCNH2):c.139G>T (p.Gly47Cys)	not provided|Long QT syndrome 2	Pathogenic/Likely pathogenic(Last reviewed: Dec 19, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200553	7	150974879	200553	197360	rs794728409	LOF	0.886	0.023	0.091
KCNH2	47	G	V	NM_000238.4(KCNH2):c.140G>T (p.Gly47Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067195	7	150974878	67195	78091	rs199473490	LOF	0.882	0.023	0.096

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	49	C	F	NM_000238.4(KCNH2):c.146G>T (p.Cys49Phe)	Long QT syndrome 2	Likely pathogenic(Last reviewed: Nov 26, 2019)	criteria provided, single submitter	VCV000972694	7	150974872	972694	961001	rs199472840	LOF	0.852	0.036	0.112
KCNH2	49	C	W	NM_000238.4(KCNH2):c.147C>G (p.Cys49Trp)	not provided	Pathogenic(Last reviewed: Jun 19, 2013)	criteria provided, single submitter	VCV000200555	7	150974871	200555	197359	rs794728410	LOF	0.85	0.033	0.117
KCNH2	49	C	Y	NM_000238.4(KCNH2):c.146G>A (p.Cys49Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067203	7	150974872	67203	78099	rs199472840	LOF	0.864	0.028	0.108
KCNH2	50	E	D	NM_000238.4(KCNH2):c.150G>T (p.Glu50Asp)	short QT syndrome	not provided	no assertion provided	VCV000067211	7	150974868	67211	78107	rs199472841	LOF	0.818	0.044	0.138
KCNH2	50	E	Q	NM_000238.4(KCNH2):c.148G>C (p.Glu50Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 9, 2019)	criteria provided, single submitter	VCV000935728	7	150974870	935728	933786	rs1801938958	LOF	0.81	0.047	0.142
KCNH2	52	C	W	NM_000238.4(KCNH2):c.156C>G (p.Cys52Trp)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 10, 2020)	criteria provided, single submitter	VCV000972150	7	150974862	972150	955098	rs754921704	LOF	0.827	0.045	0.128
KCNH2	53	G	D	NM_000238.4(KCNH2):c.158G>A (p.Gly53Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067217	7	150974860	67217	78113	rs199473491	LOF	0.867	0.023	0.11
KCNH2	53	G	R	NM_000238.4(KCNH2):c.157G>C (p.Gly53Arg)	Congenital long QT syndrome|Arrhythmia|not provided	Pathogenic/Likely pathogenic(Last reviewed: Oct 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067215	7	150974861	67215	78111	rs199472842	LOF	0.828	0.023	0.148
KCNH2	53	G	S	NM_000238.4(KCNH2):c.157G>A (p.Gly53Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067214	7	150974861	67214	78110	rs199472842	LOF	0.872	0.026	0.102
KCNH2	54	R	C	NM_000238.4(KCNH2):c.1180C>T (p.Arg394Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 18, 2020)	criteria provided, single submitter	VCV000855813	7	150952802	855813	833343	rs1325776019	LOF	0.788	0.099	0.113
KCNH2	54	R	H	NM_000238.4(KCNH2):c.1181G>A (p.Arg394His)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Feb 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000565408	7	150952801	565408	566581	rs781140785	LOF	0.837	0.068	0.095
KCNH2	54	Y	H	NM_000238.4(KCNH2):c.160T>C (p.Tyr54His)	Prolonged QT interval|Sudden cardiac death|Congenital long QT syndrome	Uncertain significance(Last reviewed: Mar 25, 2019)	criteria provided, single submitter	VCV000067223	7	150974858	67223	78119	rs199472843	LOF	0.841	0.069	0.09
KCNH2	55	S	L	NM_000238.4(KCNH2):c.164C>T (p.Ser55Leu)	Long QT syndrome|Congenital long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 9, 2020)	criteria provided, conflicting interpretations	VCV000067224	7	150974854	67224	78120	rs199472844	LOF	0.78	0.103	0.117
KCNH2	56	R	L	NM_000238.4(KCNH2):c.167G>T (p.Arg56Leu)	not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Dec 10, 2020)	criteria provided, conflicting interpretations	VCV000200557	7	150974851	200557	197356	rs199472845	LOF	0.852	0.041	0.107
KCNH2	56	R	Q	NM_000238.4(KCNH2):c.167G>A (p.Arg56Gln)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Aug 9, 2018)	criteria provided, single submitter	VCV000067228	7	150974851	67228	78124	rs199472845	LOF	0.875	0.028	0.098
KCNH2	57	A	P	NM_000238.4(KCNH2):c.169G>C (p.Ala57Pro)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jul 22, 2019)	criteria provided, single submitter	VCV000067240	7	150974849	67240	78136	rs199472846	LOF	0.882	0.04	0.078
KCNH2	57	A	T	NM_000238.4(KCNH2):c.169G>A (p.Ala57Thr)	Long QT syndrome	Uncertain significance(Last	criteria provided, single	VCV000654924	7	150974849	654924	635930	rs199472846	LOF	0.879	0.047	0.074

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Sep 21, 2018)	submitter										
KCNH2	57	A	V	NM_000238.4(KCNH2):c.170C>T (p.Ala57Val)	not provided	Likely pathogenic(Last reviewed: Sep 3, 2014)	criteria provided, single submitter	VCV000200775	7	150974848	200775	197355	rs794728493	LOF	0.86	0.058	0.082
KCNH2	57	R	C	NM_000238.4(KCNH2):c.1189C>T (p.Arg397Cys)	Arrhythmia|Long QT syndrome|Cardiovascular phenotype	Uncertain significance(Last reviewed: Oct 28, 2018)	criteria provided, multiple submitters, no conflicts	VCV000405343	7	150952793	405343	395706	rs1060500663	LOF	0.85	0.049	0.102
KCNH2	57	R	H	NM_000238.4(KCNH2):c.1190G>A (p.Arg397His)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jun 7, 2021)	criteria provided, multiple submitters, no conflicts	VCV000851410	7	150952792	851410	833342	rs368817970	LOF	0.881	0.033	0.086
KCNH2	58	E	A	NM_000238.4(KCNH2):c.173A>C (p.Glu58Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067256	7	150974845	67256	78152	rs199472847	LOF	0.861	0.04	0.099
KCNH2	58	E	D	NM_000238.4(KCNH2):c.174G>C (p.Glu58Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067260	7	150974844	67260	78156	rs199473492	LOF	0.858	0.029	0.113
KCNH2	58	E	G	NM_000238.4(KCNH2):c.173A>G (p.Glu58Gly)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: Jan 14, 2021)	criteria provided, single submitter	VCV000067257	7	150974845	67257	78153	rs199472847	LOF	0.864	0.038	0.099
KCNH2	58	E	K	NM_000238.4(KCNH2):c.172G>A (p.Glu58Lys)	not provided|Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Aug 30, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067254	7	150974846	67254	78150	rs199473413	LOF	0.837	0.029	0.134
KCNH2	62	R	Q	NM_000238.4(KCNH2):c.185G>A (p.Arg62Gln)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: May 10, 2019)	criteria provided, single submitter	VCV000067298	7	150974833	67298	78194	rs199473664	LOF	0.801	0.072	0.127
KCNH2	63	P	H	NM_000238.4(KCNH2):c.188C>A (p.Pro63His)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Sep 24, 2021)	criteria provided, conflicting interpretations	VCV000200559	7	150974830	200559	197354	rs766379103	LOF	0.801	0.086	0.112
KCNH2	64	C	W	NM_000238.4(KCNH2):c.192C>G (p.Cys64Trp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067338	7	150974826	67338	78234	rs199473414	LOF	0.789	0.071	0.14
KCNH2	64	C	Y	NM_000238.4(KCNH2):c.191G>A (p.Cys64Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067335	7	150974827	67335	78231	rs199473415	LOF	0.799	0.063	0.138
KCNH2	65	T	P	NM_000238.4(KCNH2):c.193A>C (p.Thr65Pro)	Long QT syndrome 2|Congenital long QT syndrome	Pathogenic(Last reviewed: Dec 13, 2002)	no assertion criteria provided	VCV000014434	7	150974825	14434	29473	rs121912511	LOF	0.828	0.079	0.092
KCNH2	65	T	S	NM_000238.4(KCNH2):c.194C>G (p.Thr65Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 17, 2016)	criteria provided, single submitter	VCV000237295	7	150974824	237295	240004	rs878853772	LOF	0.803	0.104	0.092
KCNH2	66	C	G	NM_000238.4(KCNH2):c.196T>G (p.Cys66Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067351	7	150974822	67351	78247	rs199473416	LOF	0.856	0.048	0.096
KCNH2	66	C	R	NM_000238.4(KCNH2):c.196T>C (p.Cys66Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 3, 2017)	criteria provided, single submitter	VCV000456900	7	150974822	456900	456496	rs199473416	LOF	0.858	0.06	0.082
KCNH2	66	C	Y	NM_000238.4(KCNH2):c.197G>A (p.Cys66Tyr)	Long QT syndrome 2	Likely pathogenic(Last reviewed: May 18, 2018)	criteria provided, single submitter	VCV000545688	7	150974821	545688	536180	rs1554430943	LOF	0.856	0.06	0.084

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	67	D	G	NM_000238.4(KCNH2):c.200A>G (p.Asp67Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV001047548	7	150974818	1047548	1027904	rs1801935588	LOF	0.815	0.099	0.086
KCNH2	68	F	C	NM_000238.4(KCNH2):c.203T>G (p.Phe68Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 18, 2019)	criteria provided, single submitter	VCV000940257	7	150974815	940257	933785	rs1801935420	LOF	0.854	0.075	0.071
KCNH2	68	F	L	NM_000238.4(KCNH2):c.202T>C (p.Phe68Leu)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: May 10, 2014)	criteria provided, single submitter	VCV000067355	7	150974816	67355	78251	rs199473417	LOF	0.837	0.09	0.072
KCNH2	69	L	P	NM_000238.4(KCNH2):c.206T>C (p.Leu69Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067358	7	150974812	67358	78254	rs199473665	LOF	0.873	0.061	0.066
KCNH2	69	L	Q	NM_000238.4(KCNH2):c.206T>A (p.Leu69Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 12, 2016)	criteria provided, single submitter	VCV000520445	7	150974812	520445	511088	rs199473665	LOF	0.871	0.061	0.068
KCNH2	69	V	M	NM_000238.4(KCNH2):c.1225G>A (p.Val409Met)	Wolff-Parkinson-White pattern|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Oct 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000456884	7	150952757	456884	457546	rs539146547	LOF	0.848	0.079	0.073
KCNH2	70	H	N	NM_000238.4(KCNH2):c.208C>A (p.His70Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067362	7	150974810	67362	78258	rs199473418	LOF	0.796	0.093	0.111
KCNH2	70	H	R	NM_000238.4(KCNH2):c.209A>G (p.His70Arg)	Long QT syndrome|Congenital long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last reviewed: Jun 29, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067363	7	150974809	67363	78259	rs199473419	LOF	0.773	0.08	0.147
KCNH2	71	D	N	NM_000238.4(KCNH2):c.1231G>A (p.Asp411Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 27, 2019)	criteria provided, single submitter	VCV000941613	7	150952751	941613	933774	rs1801230141	LOF	0.823	0.061	0.116
KCNH2	71	G	E	NM_000238.4(KCNH2):c.212G>A (p.Gly71Glu)	Cardiovascular phenotype	Likely pathogenic(Last reviewed: Oct 27, 2015)	criteria provided, single submitter	VCV000264521	7	150974806	264521	258468	rs886039183	LOF	0.823	0.05	0.127
KCNH2	71	G	R	NM_000238.4(KCNH2):c.211G>A (p.Gly71Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 30, 2019)	criteria provided, single submitter	VCV000948713	7	150974807	948713	924762	rs199473420	LOF	0.781	0.048	0.171
KCNH2	71	G	R	NM_000238.4(KCNH2):c.211G>A (p.Gly71Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 30, 2019)	criteria provided, single submitter	VCV000948713	7	150974807	948713	924762	rs199473420	LOF	0.781	0.048	0.171
KCNH2	71	G	R	NM_000238.4(KCNH2):c.211G>C (p.Gly71Arg)	Long QT syndrome|Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Feb 11, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067366	7	150974807	67366	78262	rs199473420	LOF	0.781	0.048	0.171
KCNH2	71	G	R	NM_000238.4(KCNH2):c.211G>C (p.Gly71Arg)	Long QT syndrome|Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Feb 11, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067366	7	150974807	67366	78262	rs199473420	LOF	0.781	0.048	0.171
KCNH2	71	G	W	NM_000238.4(KCNH2):c.211G>T (p.Gly71Trp)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 19, 2019)	criteria provided, single submitter	VCV000853484	7	150974807	853484	833362	rs199473420	LOF	0.783	0.076	0.141
KCNH2	72	P	L	NM_000238.4(KCNH2):c.215C>T (p.Pro72Leu)	Congenital long QT syndrome|not provided	Pathogenic(Last	criteria	VCV000067370	7	150974803	67370	78266	rs199473421	LOF	0.815	0.075	0.109

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Jul 3, 2019)	provided, single submitter										
KCNH2	72	P	Q	NM_000238.4(KCNH2):c.215C>A (p.Pro72Gln)	Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 2|Cardiovascular phenotype|not provided	Pathogenic/Likely pathogenic(Last reviewed: Oct 3, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067368	7	150974803	67368	78264	rs199473421	LOF	0.842	0.051	0.108
KCNH2	72	P	R	NM_000238.4(KCNH2):c.215C>G (p.Pro72Arg)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jun 22, 2019)	criteria provided, single submitter	VCV000067369	7	150974803	67369	78265	rs199473421	LOF	0.806	0.045	0.149
KCNH2	72	P	S	NM_000238.4(KCNH2):c.214C>T (p.Pro72Ser)	not provided	Pathogenic(Last reviewed: Apr 12, 2014)	criteria provided, single submitter	VCV000200564	7	150974804	200564	197353	rs794728411	LOF	0.851	0.06	0.089
KCNH2	74	T	A	NM_000238.4(KCNH2):c.220A>G (p.Thr74Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV001040732	7	150974798	1040732	1027903	rs199473666	LOF	0.882	0.044	0.074
KCNH2	74	T	M	NM_000238.4(KCNH2):c.221C>T (p.Thr74Met)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jul 11, 2018)	criteria provided, conflicting interpretations	VCV000067377	7	150974797	67377	78273	rs199473422	LOF	0.872	0.049	0.079
KCNH2	74	T	P	NM_000238.4(KCNH2):c.220A>C (p.Thr74Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067375	7	150974798	67375	78271	rs199473666	LOF	0.887	0.035	0.077
KCNH2	74	T	R	NM_000238.4(KCNH2):c.221C>G (p.Thr74Arg)	Long QT syndrome|Congenital long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Dec 13, 2019)	criteria provided, conflicting interpretations	VCV000067376	7	150974797	67376	78272	rs199473422	LOF	0.851	0.032	0.117
KCNH2	75	Q	E	NM_000238.4(KCNH2):c.223C>G (p.Gln75Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 31, 2020)	criteria provided, single submitter	VCV001023341	7	150974795	1023341	1007360	rs1801933977	LOF	0.832	0.064	0.104
KCNH2	76	R	L	NM_000238.4(KCNH2):c.227G>T (p.Arg76Leu)	Arrhythmia	Uncertain significance(Last reviewed: Oct 30, 2018)	criteria provided, single submitter	VCV000629614	7	150974791	629614	617357	rs868054429	Neutral	0.691	0.174	0.135
KCNH2	78	A	P	NM_000238.4(KCNH2):c.232G>C (p.Ala78Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067386	7	150974786	67386	78282	rs199472848	LOF	0.886	0.04	0.074
KCNH2	78	A	T	NM_000238.4(KCNH2):c.232G>A (p.Ala78Thr)	not provided	Likely pathogenic(Last reviewed: Jun 22, 2021)	criteria provided, single submitter	VCV001197726	7	150974786	1197726	1187214	NA	LOF	0.884	0.046	0.071
KCNH2	78	A	V	NM_000238.4(KCNH2):c.233C>T (p.Ala78Val)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 16, 2019)	criteria provided, single submitter	VCV000957483	7	150974785	957483	945531	rs1801933488	LOF	0.864	0.056	0.08
KCNH2	79	A	P	NM_000238.4(KCNH2):c.235G>C (p.Ala79Pro)	not provided	Uncertain significance(Last reviewed: Dec 2, 2019)	criteria provided, single submitter	VCV000200778	7	150974783	200778	197352	rs794728494	LOF	0.88	0.048	0.072
KCNH2	79	A	S	NM_000238.4(KCNH2):c.235G>T (p.Ala79Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 23, 2017)	criteria provided, single submitter	VCV000456907	7	150974783	456907	457116	rs794728494	LOF	0.878	0.052	0.069
KCNH2	79	A	T	NM_000238.4(KCNH2):c.235G>A (p.Ala79Thr)	Arrhythmia|not specified	Uncertain significance(Last reviewed: Apr 5, 2021)	criteria provided, multiple submitters, no conflicts	VCV000987798	7	150974783	987798	975792	rs794728494	LOF	0.875	0.056	0.069

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	80	A	P	NM_000238.4(KCNH2):c.238G>C (p.Ala80Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067392	7	150974780	67392	78288	rs199473037	LOF	0.854	0.063	0.083
KCNH2	80	A	T	NM_000238.4(KCNH2):c.238G>A (p.Ala80Thr)	not provided	Uncertain significance(Last reviewed: Dec 27, 2019)	criteria provided, single submitter	VCV001311334	7	150974780	1311334	1300715	NA	LOF	0.841	0.081	0.078
KCNH2	80	A	V	NM_000238.4(KCNH2):c.239C>T (p.Ala80Val)	Sudden cardiac death	not provided	no assertion provided	VCV000067395	7	150974779	67395	78291	rs199473493	LOF	0.817	0.098	0.085
KCNH2	81	Q	H	NM_000238.4(KCNH2):c.243G>C (p.Gln81His)	Long QT syndrome 1|Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Mar 7, 2018)	criteria provided, multiple submitters, no conflicts	VCV000067399	7	150974775	67399	78295	rs199472849	LOF	0.864	0.045	0.091
KCNH2	82	I	T	NM_000238.4(KCNH2):c.245T>C (p.Ile82Thr)	Long QT syndrome 2	Likely pathogenic(Last reviewed: Jun 5, 2017)	criteria provided, single submitter	VCV000585220	7	150974773	585220	576259	rs1563189895	LOF	0.897	0.038	0.064
KCNH2	83	A	S	NM_000238.4(KCNH2):c.247G>T (p.Ala83Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 11, 2018)	criteria provided, single submitter	VCV000643651	7	150974771	643651	635929	rs1554430909	LOF	0.881	0.042	0.077
KCNH2	84	Q	R	NM_000238.4(KCNH2):c.251A>G (p.Gln84Arg)	Long QT syndrome 2	Uncertain significance(Last reviewed: Oct 6, 2021)	criteria provided, single submitter	VCV001299593	7	150974767	1299593	1289647	NA	LOF	0.821	0.043	0.136
KCNH2	85	A	P	NM_000238.4(KCNH2):c.253G>C (p.Ala85Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067410	7	150974765	67410	78306	rs199472850	LOF	0.9	0.028	0.072
KCNH2	85	A	T	NM_000238.4(KCNH2):c.253G>A (p.Ala85Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 14, 2018)	criteria provided, single submitter	VCV000456912	7	150974765	456912	456887	rs199472850	LOF	0.897	0.032	0.07
KCNH2	85	A	V	NM_000238.4(KCNH2):c.254C>T (p.Ala85Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067411	7	150974764	67411	78307	rs199473494	LOF	0.885	0.039	0.076
KCNH2	86	L	P	NM_000238.4(KCNH2):c.257T>C (p.Leu86Pro)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Dec 31, 2020)	criteria provided, single submitter	VCV000067413	7	150974761	67413	78309	rs199472851	LOF	0.897	0.039	0.065
KCNH2	86	L	R	NM_000238.4(KCNH2):c.257T>G (p.Leu86Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067414	7	150974761	67414	78310	rs199472851	LOF	0.883	0.031	0.085
KCNH2	87	L	P	NM_000238.4(KCNH2):c.260T>C (p.Leu87Pro)	Congenital long QT syndrome|not provided	Likely pathogenic	no assertion criteria provided	VCV000067418	7	150974758	67418	78314	rs199473495	LOF	0.842	0.071	0.087
KCNH2	91	E	D	NM_000238.4(KCNH2):c.273G>T (p.Glu91Asp)	not provided	Uncertain significance(Last reviewed: May 16, 2019)	criteria provided, single submitter	VCV001305882	7	150974745	1305882	1297153	NA	LOF	0.728	0.079	0.193
KCNH2	92	R	C	NM_000238.4(KCNH2):c.274C>T (p.Arg92Cys)	Long QT syndrome 2|not provided|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Jan 20, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200573	7	150974744	200573	197348	rs563611707	LOF	0.867	0.054	0.079
KCNH2	92	R	L	NM_000238.4(KCNH2):c.275G>T (p.Arg92Leu)	not provided	Uncertain significance(Last reviewed: Jun 25, 2019)	criteria provided, single submitter	VCV000200575	7	150974743	200575	197347	rs794728413	LOF	0.87	0.053	0.077
KCNH2	92	R	P	NM_000238.4(KCNH2):c.275G>C (p.Arg92Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 26, 2019)	criteria provided, single submitter	VCV000947667	7	150974743	947667	924761	rs794728413	LOF	0.87	0.054	0.076
KCNH2	93	K	R	NM_000238.4(KCNH2):c.278A>G (p.Lys93Arg)	Long QT syndrome|not provided|Arrhythmia|Cardiovascular phenotype	Likely benign(Last reviewed: Nov 15,	criteria provided,	VCV000413325	7	150974740	413325	395497	rs780197027	LOF	0.857	0.058	0.085

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)	multiple submitters, no conflicts										
KCNH2	94	V	G	NM_000238.4(KCNH2):c.281T>G (p.Val94Gly)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Sep 10, 2018)	criteria provided, single submitter	VCV000067441	7	150974737	67441	78337	rs199472852	LOF	0.874	0.048	0.078
KCNH2	94	V	L	NM_000238.4(KCNH2):c.280G>C (p.Val94Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 9, 2020)	criteria provided, single submitter	VCV001052852	7	150974738	1052852	1044815	NA	LOF	0.867	0.047	0.086
KCNH2	94	V	M	NM_000238.4(KCNH2):c.280G>A (p.Val94Met)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 3, 2020)	criteria provided, single submitter	VCV001055911	7	150974738	1055911	1044816	NA	LOF	0.87	0.044	0.086
KCNH2	95	E	A	NM_000238.4(KCNH2):c.284A>C (p.Glu95Ala)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 15, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200578	7	150974734	200578	197346	rs794728414	LOF	0.867	0.054	0.08
KCNH2	96	I	T	NM_000238.4(KCNH2):c.287T>C (p.Ile96Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067448	7	150974731	67448	78344	rs199472853	LOF	0.86	0.044	0.096
KCNH2	96	I	V	NM_000238.4(KCNH2):c.286A>G (p.Ile96Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067446	7	150974732	67446	78342	rs199473496	LOF	0.863	0.048	0.089
KCNH2	97	A	G	NM_000238.4(KCNH2):c.290C>G (p.Ala97Gly)	not provided	Uncertain significance(Last reviewed: May 16, 2012)	criteria provided, single submitter	VCV000200580	7	150974728	200580	197345	rs794728415	LOF	0.873	0.051	0.076
KCNH2	98	F	C	NM_000238.4(KCNH2):c.293T>G (p.Phe98Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 29, 2019)	criteria provided, single submitter	VCV000946662	7	150974725	946662	924760	rs1801930040	LOF	0.859	0.046	0.095
KCNH2	99	Y	C	NM_000238.4(KCNH2):c.296A>G (p.Tyr99Cys)	not provided	Likely pathogenic(Last reviewed: Jan 21, 2020)	criteria provided, single submitter	VCV000200582	7	150974722	200582	197344	rs199472854	LOF	0.86	0.046	0.094
KCNH2	99	Y	S	NM_000238.4(KCNH2):c.296A>C (p.Tyr99Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067457	7	150974722	67457	78353	rs199472854	LOF	0.866	0.044	0.089
KCNH2	100	P	L	NM_000238.4(KCNH2):c.1319C>T (p.Pro440Leu)	Arrhythmia|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Apr 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067185	7	150952663	67185	78081	rs199473509	LOF	0.792	0.099	0.109
KCNH2	100	R	G	NM_000238.4(KCNH2):c.298C>G (p.Arg100Gly)	not provided|Cardiovascular phenotype|Congenital long QT syndrome|Long QT syndrome 2/3, digenic	Conflicting interpretations of pathogenicity(Last reviewed: Nov 28, 2017)	criteria provided, conflicting interpretations	VCV000014442	7	150974720	14442	29481	rs121912515	LOF	0.822	0.075	0.103
KCNH2	100	R	L	NM_000238.4(KCNH2):c.299G>T (p.Arg100Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 28, 2018)	criteria provided, single submitter	VCV000657216	7	150974719	657216	635927	rs199472855	LOF	0.778	0.109	0.114
KCNH2	100	R	Q	NM_000238.4(KCNH2):c.299G>A (p.Arg100Gln)	Congenital long QT syndrome|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jun 30, 2020)	criteria provided, conflicting interpretations	VCV000067460	7	150974719	67460	78356	rs199472855	LOF	0.821	0.069	0.11
KCNH2	100	R	W	NM_000238.4(KCNH2):c.298C>T (p.Arg100Trp)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Oct 3, 2019)	criteria provided, single submitter	VCV000067459	7	150974720	67459	78355	rs121912515	LOF	0.78	0.102	0.118

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	101	K	E	NM_000238.4(KCNH2):c.301A>G (p.Lys101Glu)	Congenital long QT syndrome|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Aug 22, 2020)	criteria provided, conflicting interpretations	VCV000067462	7	150974717	67462	78358	rs199472856	LOF	0.805	0.042	0.153
KCNH2	101	K	N	NM_000238.4(KCNH2):c.303A>T (p.Lys101Asn)	not provided	Pathogenic(Last reviewed: Apr 28, 2014)	criteria provided, single submitter	VCV000200590	7	150974715	200590	197342	rs794728417	LOF	0.807	0.046	0.147
KCNH2	101	K	R	NM_000238.4(KCNH2):c.302A>G (p.Lys101Arg)	not provided	Uncertain significance(Last reviewed: Feb 15, 2021)	criteria provided, single submitter	VCV001256427	7	150974716	1256427	1246356	NA	LOF	0.777	0.044	0.18
KCNH2	102	D	A	NM_000238.4(KCNH2):c.305A>C (p.Asp102Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067464	7	150974713	67464	78360	rs199472857	LOF	0.777	0.056	0.168
KCNH2	102	D	V	NM_000238.4(KCNH2):c.305A>T (p.Asp102Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067465	7	150974713	67465	78361	rs199472857	LOF	0.761	0.064	0.175
KCNH2	104	E	D	NM_000238.4(KCNH2):c.1332G>T (p.Glu444Asp)	not provided	not provided	no assertion provided	VCV000067188	7	150952650	67188	78084	rs9770044	LOF	0.779	0.061	0.16
KCNH2	104	E	D	NM_000238.4(KCNH2):c.1332G>T (p.Glu444Asp)	not provided	not provided	no assertion provided	VCV000067188	7	150952650	67188	78084	rs9770044	LOF	0.779	0.061	0.16
KCNH2	104	E	D	NM_000238.4(KCNH2):c.1332G>C (p.Glu444Asp)	Arrhythmia	Uncertain significance(Last reviewed: Jul 25, 2019)	criteria provided, single submitter	VCV000927767	7	150952650	927767	910772	rs9770044	LOF	0.779	0.061	0.16
KCNH2	104	E	D	NM_000238.4(KCNH2):c.1332G>C (p.Glu444Asp)	Arrhythmia	Uncertain significance(Last reviewed: Jul 25, 2019)	criteria provided, single submitter	VCV000927767	7	150952650	927767	910772	rs9770044	LOF	0.779	0.061	0.16
KCNH2	104	E	K	NM_000238.4(KCNH2):c.1330G>A (p.Glu444Lys)	Arrhythmia|Long QT syndrome|Long QT syndrome 2|not provided	Uncertain significance(Last reviewed: Mar 24, 2021)	criteria provided, multiple submitters, no conflicts	VCV000191472	7	150952652	191472	189262	rs201268831	LOF	0.744	0.061	0.195
KCNH2	104	S	R	NM_000238.4(KCNH2):c.312C>G (p.Ser104Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 18, 2017)	criteria provided, single submitter	VCV000527024	7	150959732	527024	522474	rs1554428245	LOF	0.734	0.068	0.199
KCNH2	106	F	L	NM_000238.4(KCNH2):c.318C>A (p.Phe106Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067478	7	150959726	67478	78374	rs199473497	LOF	0.862	0.047	0.091
KCNH2	106	F	Y	NM_000238.4(KCNH2):c.317T>A (p.Phe106Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067477	7	150959727	67477	78373	rs199472858	LOF	0.869	0.046	0.084
KCNH2	108	A	S	NM_000238.4(KCNH2):c.1342G>T (p.Ala448Ser)	Arrhythmia|Long QT syndrome 2|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Oct 26, 2020)	criteria provided, conflicting interpretations	VCV000448973	7	150952640	448973	442552	rs767723985	LOF	0.87	0.045	0.085
KCNH2	108	A	T	NM_000238.4(KCNH2):c.1342G>A (p.Ala448Thr)	Arrhythmia	Uncertain significance(Last reviewed: Nov 4, 2019)	criteria provided, single submitter	VCV000924051	7	150952640	924051	910771	rs767723985	LOF	0.87	0.044	0.086
KCNH2	108	C	R	NM_000238.4(KCNH2):c.322T>C (p.Cys108Arg)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jan 30, 2019)	criteria provided, single submitter	VCV000067482	7	150959722	67482	78378	rs199472859	LOF	0.862	0.054	0.084
KCNH2	109	L	R	NM_000238.4(KCNH2):c.326T>G (p.Leu109Arg)	Long QT syndrome 2|not provided	Benign(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000067484	7	150959718	67484	78380	rs199473498	LOF	0.866	0.05	0.084
KCNH2	111	D	N	NM_000238.4(KCNH2):c.331G>A (p.Asp111Asn)	Arrhythmia	Uncertain significance(Last reviewed: Aug 21,	criteria provided, single submitter	VCV000921508	7	150959713	921508	910797	rs1801502139	LOF	0.868	0.052	0.079

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)											
KCNH2	111	D	V	NM_000238.4(KCNH2):c.332A>T (p.Asp111Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067489	7	150959712	67489	78385	rs199472860	LOF	0.87	0.053	0.077
KCNH2	111	P	L	NM_000238.4(KCNH2):c.1352C>T (p.Pro451Leu)	Long QT syndrome|Long QT syndrome 2| Congenital long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Mar 23, 2021)	criteria provided, conflicting interpretations	VCV000067190	7	150952630	67190	78086	rs199472902	LOF	0.873	0.051	0.076
KCNH2	113	A	T	NM_000238.4(KCNH2):c.1357G>A (p.Ala453Thr)	Arrhythmia	Uncertain significance(Last reviewed: Mar 24, 2020)	criteria provided, single submitter	VCV000919895	7	150952625	919895	910770	rs1801221411	LOF	0.869	0.044	0.087
KCNH2	114	P	S	NM_000238.4(KCNH2):c.340C>T (p.Pro114Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067494	7	150959704	67494	78390	rs199472861	LOF	0.875	0.048	0.077
KCNH2	115	V	G	NM_000238.4(KCNH2):c.344T>G (p.Val115Gly)	not provided	Pathogenic(Last reviewed: Apr 10, 2014)	criteria provided, single submitter	VCV000200276	7	150959700	200276	197339	rs794728351	LOF	0.873	0.05	0.077
KCNH2	115	V	M	NM_000238.4(KCNH2):c.343G>A (p.Val115Met)	Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 2	Uncertain significance(Last reviewed: Sep 25, 2020)	criteria provided, single submitter	VCV000067496	7	150959701	67496	78392	rs150988911	LOF	0.871	0.046	0.083
KCNH2	116	D	Y	NM_000238.4(KCNH2):c.1366G>T (p.Asp456Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067191	7	150952616	67191	78087	rs199473510	LOF	0.862	0.06	0.078
KCNH2	118	E	K	NM_000238.4(KCNH2):c.352G>A (p.Glu118Lys)	not provided	Uncertain significance(Last reviewed: Jun 1, 2017)	criteria provided, single submitter	VCV000810212	7	150959692	810212	795991	rs746600107	LOF	0.709	0.071	0.221
KCNH2	118	E	Q	NM_000238.4(KCNH2):c.352G>C (p.Glu118Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 24, 2020)	criteria provided, single submitter	VCV001000649	7	150959692	1000649	992200	rs746600107	LOF	0.717	0.076	0.207
KCNH2	119	D	H	NM_000238.4(KCNH2):c.355G>C (p.Asp119His)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Oct 31, 2019)	criteria provided, multiple submitters, no conflicts	VCV000405352	7	150959689	405352	396198	rs376308069	LOF	0.729	0.073	0.198
KCNH2	120	D	Y	NM_000238.4(KCNH2):c.1378G>T (p.Asp460Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067192	7	150952604	67192	78088	rs199472903	LOF	0.783	0.087	0.129
KCNH2	121	A	V	NM_000238.4(KCNH2):c.362C>T (p.Ala121Val)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 8, 2020)	criteria provided, single submitter	VCV001054947	7	150959682	1054947	1044814	NA	LOF	0.73	0.16	0.11
KCNH2	122	M	V	NM_000238.4(KCNH2):c.1384A>G (p.Met462Val)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 28, 2020)	criteria provided, single submitter	VCV001002624	7	150952598	1002624	992198	rs1801220156	LOF	0.864	0.047	0.089
KCNH2	123	F	L	NM_000238.4(KCNH2):c.1387T>C (p.Phe463Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067193	7	150952595	67193	78089	rs199472904	LOF	0.86	0.048	0.092
KCNH2	124	I	T	NM_000238.4(KCNH2):c.1391T>C (p.Ile464Thr)	Arrhythmia	Uncertain significance(Last reviewed: Dec 10, 2018)	criteria provided, single submitter	VCV000919164	7	150952591	919164	910769	rs1801219752	LOF	0.861	0.044	0.095
KCNH2	124	M	R	NM_000238.4(KCNH2):c.371T>G (p.Met124Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067501	7	150959673	67501	78397	rs199472862	LOF	0.857	0.048	0.096
KCNH2	124	M	T	NM_000238.4(KCNH2):c.371T>C (p.Met124Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067500	7	150959673	67500	78396	rs199472862	LOF	0.861	0.045	0.095
KCNH2	125	F	C	NM_000238.4(KCNH2):c.374T>G (p.Phe125Cys)	Congenital long QT syndrome|Cardiovascular phenotype	Uncertain significance(Last reviewed: Aug 5, 2015)	criteria provided, single submitter	VCV000067502	7	150959670	67502	78398	rs199473499	LOF	0.862	0.046	0.092

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	125	F	L	NM_000238.4(KCNH2):c.373T>C (p.Phe125Leu)	Long QT syndrome	Uncertain significance(Last reviewed: May 30, 2019)	criteria provided, single submitter	VCV000934591	7	150959671	934591	933784	rs1801500008	LOF	0.861	0.047	0.092
KCNH2	125	V	A	NM_000238.4(KCNH2):c.1394T>C (p.Val465Ala)	Hypertrophic cardiomyopathy|Arrhythmia	Uncertain significance(Last reviewed: Jan 8, 2014)	criteria provided, single submitter	VCV000374073	7	150952588	374073	360890	rs1057518876	LOF	0.867	0.044	0.089
KCNH2	126	D	Y	NM_000238.4(KCNH2):c.1396G>T (p.Asp466Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067194	7	150952586	67194	78090	rs199473511	LOF	0.875	0.05	0.076
KCNH2	126	I	N	NM_000238.4(KCNH2):c.377T>A (p.Ile126Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 19, 2019)	criteria provided, single submitter	VCV000859531	7	150959667	859531	833361	rs1801499760	LOF	0.867	0.045	0.089
KCNH2	127	L	F	NM_000238.4(KCNH2):c.379C>T (p.Leu127Phe)	not provided	Likely pathogenic(Last reviewed: Feb 4, 2014)	criteria provided, single submitter	VCV000200278	7	150959665	200278	197336	rs794728352	LOF	0.871	0.046	0.083
KCNH2	128	N	S	NM_000238.4(KCNH2):c.383A>G (p.Asn128Ser)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Apr 14, 2017)	criteria provided, multiple submitters, no conflicts	VCV000191473	7	150959661	191473	189263	rs200343670	LOF	0.87	0.049	0.081
KCNH2	129	F	L	NM_000238.4(KCNH2):c.387C>G (p.Phe129Leu)	not provided	Likely pathogenic(Last reviewed: Nov 7, 2019)	criteria provided, single submitter	VCV000200280	7	150959657	200280	197335	rs764831888	LOF	0.861	0.047	0.092
KCNH2	130	E	K	NM_000238.4(KCNH2):c.388G>A (p.Glu130Lys)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jun 29, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067503	7	150959656	67503	78399	rs199472863	LOF	0.859	0.055	0.086
KCNH2	131	V	L	NM_000238.4(KCNH2):c.391G>T (p.Val131Leu)	not provided|Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Feb 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000927974	7	150959653	927974	910795	rs142590566	LOF	0.869	0.047	0.084
KCNH2	131	V	M	NM_000238.4(KCNH2):c.391G>A (p.Val131Met)	Arrhythmia	Uncertain significance(Last reviewed: Aug 20, 2019)	criteria provided, single submitter	VCV000920295	7	150959653	920295	910796	rs142590566	LOF	0.871	0.044	0.085
KCNH2	132	R	P	NM_000238.4(KCNH2):c.1415G>C (p.Arg472Pro)	not provided	Likely pathogenic(Last reviewed: Jun 8, 2012)	criteria provided, single submitter	VCV000200340	7	150952567	200340	197281	rs794728369	LOF	0.815	0.095	0.09
KCNH2	133	M	T	NM_000238.4(KCNH2):c.398T>C (p.Met133Thr)	Long QT syndrome|Long QT syndrome 2|not provided	Uncertain significance(Last reviewed: Oct 14, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200282	7	150959646	200282	197334	rs794728353	Neutral	0.683	0.216	0.101
KCNH2	133	T	I	NM_000238.4(KCNH2):c.1418C>T (p.Thr473Ile)	not specified	Likely pathogenic(Last reviewed: Jul 24, 2018)	criteria provided, single submitter	VCV000811073	7	150952564	811073	799503	rs199472905	Neutral	0.626	0.279	0.095
KCNH2	133	T	N	NM_000238.4(KCNH2):c.1418C>A (p.Thr473Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067197	7	150952564	67197	78093	rs199472905	Neutral	0.716	0.182	0.102
KCNH2	133	T	P	NM_000238.4(KCNH2):c.1417A>C (p.Thr473Pro)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: Sep 7, 2021)	no assertion criteria provided	VCV000067196	7	150952565	67196	78092	rs199473512	Neutral	0.736	0.17	0.093
KCNH2	134	T	I	NM_000238.4(KCNH2):c.1421C>T	Congenital long QT syndrome|not provided	Pathogenic(Last	criteria	VCV000067198	7	150952561	67198	78094	rs199472906	Neutral	0.426	0.471	0.103

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Thr474Ile)		reviewed: Oct 10, 2014)	provided, single submitter										
KCNH2	135	Y	C	NM_000238.4(KCNH2):c.1424A>G (p.Tyr475Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067199	7	150952558	67199	78095	rs199472907	Neutral	0.452	0.378	0.17
KCNH2	136	V	I	NM_000238.4(KCNH2):c.1426G>A (p.Val476Ile)	Congenital long QT syndrome|Long QT syndrome|not specified|Arrhythmia	Uncertain significance(Last reviewed: Apr 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067200	7	150952556	67200	78096	rs199472908	Neutral	0.462	0.384	0.154
KCNH2	137	M	V	NM_000238.4(KCNH2):c.409A>G (p.Met137Val)	Short QT syndrome 1	Uncertain significance(Last reviewed: Mar 16, 2020)	criteria provided, single submitter	VCV000930960	7	150959635	930960	919088	rs1801498381	Neutral	0.397	0.438	0.165
KCNH2	139	G	A	NM_000238.4(KCNH2):c.416G>C (p.Gly139Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 27, 2020)	criteria provided, single submitter	VCV001018296	7	150959628	1018296	1007358	rs1451114925	Neutral	0.431	0.293	0.277
KCNH2	141	P	L	NM_000238.4(KCNH2):c.422C>T (p.Pro141Leu)	Long QT syndrome|Hypertrophic cardiomyopathy|Arrhythmia|Cardiovascular phenotype|not provided|not specified|Long QT syndrome 2	Conflicting interpretations of pathogenicity(Last reviewed: Feb 25, 2021)	criteria provided, conflicting interpretations	VCV000067504	7	150959622	67504	78400	rs199472864	Neutral	0.404	0.365	0.23
KCNH2	141	P	S	NM_000238.4(KCNH2):c.421C>T (p.Pro141Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 4, 2020)	criteria provided, single submitter	VCV001045443	7	150959623	1045443	1027902	rs1563172835	Neutral	0.412	0.34	0.249
KCNH2	142	A	T	NM_000238.4(KCNH2):c.424G>A (p.Ala142Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 29, 2020)	criteria provided, single submitter	VCV001025220	7	150959620	1025220	1007357	rs767514448	Neutral	0.392	0.408	0.2
KCNH2	143	V	F	NM_000238.4(KCNH2):c.1447G>T (p.Val483Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 24, 2019)	criteria provided, single submitter	VCV000949825	7	150952535	949825	924756	rs755834128	Neutral	0.422	0.417	0.161
KCNH2	144	D	V	NM_000238.4(KCNH2):c.431A>T (p.Asp144Val)	Long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Sep 10, 2020)	criteria provided, conflicting interpretations	VCV000526910	7	150959613	526910	522702	rs146284716	Neutral	0.358	0.337	0.305
KCNH2	145	T	I	NM_000238.4(KCNH2):c.434C>T (p.Thr145Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV001047559	7	150959610	1047559	1027901	rs1801497068	Neutral	0.331	0.484	0.185
KCNH2	147	G	A	NM_000238.4(KCNH2):c.1460G>C (p.Gly487Ala)	Arrhythmia	Uncertain significance(Last reviewed: Feb 11, 2019)	criteria provided, single submitter	VCV000921335	7	150952522	921335	910766	rs1801215399	Neutral	0.418	0.286	0.297
KCNH2	147	G	S	NM_000238.4(KCNH2):c.1459G>A (p.Gly487Ser)	not specified|Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 19, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200342	7	150952523	200342	197279	rs562875924	GOF	0.424	0.255	0.321
KCNH2	147	H	R	NM_000238.4(KCNH2):c.440A>G (p.His147Arg)	not provided|Long QT syndrome|Long QT syndrome 2|Short QT syndrome 1	Uncertain significance(Last reviewed: Dec 11, 2018)	criteria provided, multiple submitters, no conflicts	VCV000200285	7	150959604	200285	197333	rs768938134	GOF	0.443	0.229	0.328
KCNH2	148	R	C	NM_000238.4(KCNH2):c.1462C>T (p.Arg488Cys)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Mar 8, 2021)	criteria provided, multiple submitters, no conflicts	VCV001018981	7	150952520	1018981	1007351	rs281865155	Neutral	0.439	0.331	0.231

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	148	R	Q	NM_000238.4(KCNH2):c.443G>A (p.Arg148Gln)	Arrhythmia|Long QT syndrome|not provided	Uncertain significance(Last reviewed: May 21, 2020)	criteria provided, multiple submitters, no conflicts	VCV000567306	7	150959601	567306	561406	rs374912424	Neutral	0.44	0.271	0.289
KCNH2	148	R	W	NM_000238.4(KCNH2):c.442C>T (p.Arg148Trp)	Cardiovascular phenotype|Conduction disorder of the heart|not specified|Long QT syndrome 2| Arrhythmia|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Dec 2, 2020)	criteria provided, conflicting interpretations	VCV000067505	7	150959602	67505	78401	rs139544114	Neutral	0.42	0.357	0.223
KCNH2	149	G	A	NM_000238.4(KCNH2):c.446G>C (p.Gly149Ala)	Congenital long QT syndrome|Long QT syndrome 2|Short QT syndrome 1|Long QT syndrome	Uncertain significance(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000067506	7	150959598	67506	78402	rs199472865	Neutral	0.496	0.267	0.237
KCNH2	149	G	D	NM_000238.4(KCNH2):c.446G>A (p.Gly149Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 9, 2019)	criteria provided, single submitter	VCV000863779	7	150959598	863779	833360	rs199472865	GOF	0.556	0.184	0.26
KCNH2	149	I	F	NM_000238.4(KCNH2):c.1465A>T (p.Ile489Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067201	7	150952517	67201	78097	rs199472909	Neutral	0.522	0.309	0.169
KCNH2	150	A	P	NM_000238.4(KCNH2):c.1468G>C (p.Ala490Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067202	7	150952514	67202	78098	rs28928905	Neutral	0.53	0.266	0.204
KCNH2	150	P	L	NM_000238.4(KCNH2):c.449C>T (p.Pro150Leu)	Arrhythmia	Uncertain significance(Last reviewed: Dec 15, 2020)	criteria provided, single submitter	VCV001171810	7	150959595	1171810	1160704	NA	Neutral	0.405	0.387	0.208
KCNH2	150	P	S	NM_000238.4(KCNH2):c.448C>T (p.Pro150Ser)	Ventricular tachycardia	Uncertain significance(Last reviewed: Nov 20, 2015)	criteria provided, single submitter	VCV000222667	7	150959596	222667	224346	rs869025445	Neutral	0.422	0.361	0.217
KCNH2	151	P	A	NM_000238.4(KCNH2):c.451C>G (p.Pro151Ala)	Long QT syndrome 2|Short QT syndrome 1|not provided|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Jun 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000405361	7	150959593	405361	396190	rs1060500674	Neutral	0.421	0.383	0.196
KCNH2	151	P	S	NM_000238.4(KCNH2):c.451C>T (p.Pro151Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 20, 2020)	criteria provided, single submitter	VCV001026596	7	150959593	1026596	1007356	rs1060500674	Neutral	0.432	0.359	0.209
KCNH2	151	V	I	NM_000238.4(KCNH2):c.1471G>A (p.Val491Ile)	Arrhythmia|not provided|Long QT syndrome| Cardiovascular phenotype|not specified	Likely benign(Last reviewed: Dec 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200344	7	150952511	200344	197278	rs374376640	Neutral	0.467	0.36	0.173
KCNH2	152	H	Y	NM_000238.4(KCNH2):c.1474C>T (p.His492Tyr)	Long QT syndrome 2|Congenital long QT syndrome|Arrhythmia	Likely pathogenic(Last reviewed: Mar 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067204	7	150952508	67204	78100	rs199472910	Neutral	0.393	0.395	0.212
KCNH2	152	T	I	NM_000238.4(KCNH2):c.455C>T (p.Thr152Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2019)	criteria provided, single submitter	VCV000940899	7	150959589	940899	933783	rs794728354	Neutral	0.365	0.47	0.165
KCNH2	152	T	S	NM_000238.4(KCNH2):c.455C>G (p.Thr152Ser)	Arrhythmia|not provided	Uncertain significance(Last reviewed: Jul 7, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200288	7	150959589	200288	197331	rs794728354	Neutral	0.413	0.412	0.175
KCNH2	153	Y	C	NM_000238.4(KCNH2):c.1478A>G (p.Tyr493Cys)	Congenital long QT syndrome|not provided|Long QT syndrome	Likely pathogenic(Last reviewed: Jan 15,	criteria provided, multiple	VCV000067206	7	150952504	67206	78102	rs199472911	Neutral	0.495	0.355	0.15

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2021)	submitters, no conflicts										
KCNH2	153	Y	F	NM_000238.4(KCNH2):c.1478A>T (p.Tyr493Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067207	7	150952504	67207	78103	rs199472911	Neutral	0.477	0.382	0.141
KCNH2	153	Y	S	NM_000238.4(KCNH2):c.1478A>C (p.Tyr493Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067205	7	150952504	67205	78101	rs199472911	Neutral	0.517	0.338	0.145
KCNH2	154	W	R	NM_000238.4(KCNH2):c.460T>C (p.Trp154Arg)	Arrhythmia	Uncertain significance(Last reviewed: Aug 8, 2019)	criteria provided, single submitter	VCV000628868	7	150959584	628868	617355	rs747591865	Neutral	0.637	0.2	0.163
KCNH2	159	L	R	NM_000238.4(KCNH2):c.1496T>G (p.Leu499Arg)	not specified|not provided	Uncertain significance(Last reviewed: Feb 27, 2020)	criteria provided, single submitter	VCV000200346	7	150952486	200346	197277	rs794728370	Neutral	0.598	0.213	0.188
KCNH2	160	A	S	NM_000238.4(KCNH2):c.478G>T (p.Ala160Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 11, 2019)	criteria provided, single submitter	VCV000955775	7	150958497	955775	945530	rs1210130846	Neutral	0.53	0.335	0.136
KCNH2	161	D	G	NM_000238.4(KCNH2):c.1502A>G (p.Asp501Gly)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Aug 24, 2012)	criteria provided, single submitter	VCV000067210	7	150952480	67210	78106	rs199473513	Neutral	0.527	0.317	0.156
KCNH2	161	D	H	NM_000238.4(KCNH2):c.1501G>C (p.Asp501His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067209	7	150952481	67209	78105	rs199472912	Neutral	0.479	0.36	0.161
KCNH2	161	D	N	NM_000238.4(KCNH2):c.1501G>A (p.Asp501Asn)	Congenital long QT syndrome|not provided|Long QT syndrome|Cardiovascular phenotype	Pathogenic/Likely pathogenic(Last reviewed: Mar 22, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067208	7	150952481	67208	78104	rs199472912	Neutral	0.543	0.283	0.174
KCNH2	164	R	H	NM_000238.4(KCNH2):c.491G>A (p.Arg164His)	Congenital long QT syndrome|Long QT syndrome 2|Long QT syndrome	Uncertain significance(Last reviewed: Mar 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067508	7	150958484	67508	78404	rs199472866	Neutral	0.619	0.221	0.16
KCNH2	166	I	V	NM_000238.4(KCNH2):c.1516A>G (p.Ile506Val)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 28, 2018)	criteria provided, single submitter	VCV000645833	7	150952466	645833	635911	rs773139548	Neutral	0.56	0.312	0.129
KCNH2	167	L	V	NM_000238.4(KCNH2):c.499C>G (p.Leu167Val)	not specified	Uncertain significance(Last reviewed: Mar 12, 2019)	criteria provided, single submitter	VCV000929250	7	150958476	929250	917005	rs1801462947	Neutral	0.645	0.253	0.102
KCNH2	167	P	L	NM_000238.4(KCNH2):c.1520C>T (p.Pro507Leu)	not provided	Uncertain significance(Last reviewed: Oct 1, 2020)	criteria provided, single submitter	VCV000200352	7	150952462	200352	197275	rs794728372	Neutral	0.597	0.288	0.115
KCNH2	167	P	S	NM_000238.4(KCNH2):c.1519C>T (p.Pro507Ser)	not provided	Uncertain significance(Last reviewed: Aug 25, 2017)	criteria provided, multiple submitters, no conflicts	VCV000200350	7	150952463	200350	197276	rs794728371	Neutral	0.65	0.249	0.101
KCNH2	169	D	N	NM_000238.4(KCNH2):c.1525G>A (p.Asp509Asn)	Long QT syndrome 2|Cardiovascular phenotype|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jul 3, 2018)	criteria provided, conflicting interpretations	VCV000200354	7	150952457	200354	197273	rs370637245	LOF	0.684	0.152	0.164
KCNH2	172	A	V	NM_000238.4(KCNH2):c.515C>T (p.Ala172Val)	not provided	Uncertain significance(Last reviewed: Nov 5, 2013)	criteria provided, single submitter	VCV000200290	7	150958460	200290	197329	rs794728355	Neutral	0.619	0.274	0.107
KCNH2	173	F	L	NM_000238.4(KCNH2):c.1537T>C (p.Phe513Leu)	Long QT syndrome	Uncertain significance(Last	criteria provided, single	VCV000567702	7	150952445	567702	564160	rs1563159718	Neutral	0.557	0.319	0.124

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: May 9, 2019)	submitter										
KCNH2	175	A	D	NM_000238.4(KCNH2):c.524C>A (p.Ala175Asp)	Long QT syndrome 2|not specified|Long QT syndrome	Likely benign(Last reviewed: Jul 31, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200211	7	150958451	200211	197328	rs776541110	Neutral	0.638	0.224	0.138
KCNH2	175	A	S	NM_000238.4(KCNH2):c.523G>T (p.Ala175Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 3, 2019)	criteria provided, single submitter	VCV000937283	7	150958452	937283	933782	rs1296505812	Neutral	0.555	0.327	0.118
KCNH2	175	A	T	NM_000238.4(KCNH2):c.523G>A (p.Ala175Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 19, 2018)	criteria provided, single submitter	VCV000571170	7	150958452	571170	561741	rs1296505812	Neutral	0.53	0.361	0.11
KCNH2	176	R	Q	NM_000238.4(KCNH2):c.527G>A (p.Arg176Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 23, 2020)	criteria provided, single submitter	VCV001010337	7	150958448	1010337	992199	rs1164359840	Neutral	0.472	0.328	0.2
KCNH2	176	R	W	NM_000238.4(KCNH2):c.526C>T (p.Arg176Trp)	not specified|not provided|Long QT syndrome 2| Cardiovascular phenotype|Long QT syndrome| Short QT syndrome 1	Conflicting interpretations of pathogenicity, risk factor(Last reviewed: Feb 2, 2022)	criteria provided, conflicting interpretations	VCV000067509	7	150958449	67509	78405	rs36210422	Neutral	0.42	0.404	0.176
KCNH2	177	E	A	NM_000238.4(KCNH2):c.530A>C (p.Glu177Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 4, 2020)	criteria provided, single submitter	VCV001045316	7	150958445	1045316	1027899	rs1801461414	Neutral	0.405	0.363	0.232
KCNH2	179	S	A	NM_000238.4(KCNH2):c.535T>G (p.Ser179Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 25, 2020)	criteria provided, single submitter	VCV001059858	7	150958440	1059858	1044813	NA	Neutral	0.439	0.404	0.156
KCNH2	181	R	Q	NM_000238.4(KCNH2):c.542G>A (p.Arg181Gln)	not specified|not provided|Long QT syndrome| Cardiovascular phenotype	Benign/Likely benign(Last reviewed: Nov 21, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067510	7	150958433	67510	78406	rs41308954	Neutral	0.397	0.328	0.275
KCNH2	182	G	R	NM_000238.4(KCNH2):c.1564G>A (p.Gly522Arg)	Arrhythmia|Congenital long QT syndrome	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV000067212	7	150951829	67212	78108	rs199473514	GOF	0.483	0.2	0.317
KCNH2	182	S	W	NM_000238.4(KCNH2):c.545C>G (p.Ser182Trp)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jun 24, 2019)	criteria provided, single submitter	VCV000405351	7	150958430	405351	396186	rs1057517742	Neutral	0.411	0.367	0.222
KCNH2	185	K	N	NM_000238.4(KCNH2):c.1575G>T (p.Lys525Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067213	7	150951818	67213	78109	rs199472913	Neutral	0.386	0.408	0.207
KCNH2	186	T	A	NM_000238.4(KCNH2):c.1576A>G (p.Thr526Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 15, 2020)	criteria provided, single submitter	VCV001038284	7	150951817	1038284	1027890	rs1801183927	Neutral	0.354	0.462	0.184
KCNH2	187	G	S	NM_000238.4(KCNH2):c.559G>A (p.Gly187Ser)	not provided	not provided	no assertion provided	VCV000067511	7	150958416	67511	78407	rs199472867	Neutral	0.45	0.284	0.265
KCNH2	188	A	T	NM_000238.4(KCNH2):c.562G>A (p.Ala188Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 25, 2020)	criteria provided, single submitter	VCV001035380	7	150958413	1035380	1027898	rs1273532552	Neutral	0.379	0.389	0.232
KCNH2	188	A	V	NM_000238.4(KCNH2):c.563C>T (p.Ala188Val)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Aug 9, 2020)	criteria provided, multiple submitters, no	VCV000200293	7	150958412	200293	197326	rs794728356	Neutral	0.367	0.408	0.224

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNH2	188	R	P	NM_000238.4(KCNH2):c.1583G>C (p.Arg528Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067216	7	150951810	67216	78112	rs199472914	Neutral	0.439	0.324	0.236
KCNH2	188	R	W	NM_000238.4(KCNH2):c.1582C>T (p.Arg528Trp)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Oct 30, 2018)	criteria provided, multiple submitters, no conflicts	VCV000220162	7	150951811	220162	221673	rs864622403	Neutral	0.381	0.41	0.209
KCNH2	189	G	V	NM_000238.4(KCNH2):c.566G>T (p.Gly189Val)	Long QT syndrome 2	Uncertain significance(Last reviewed: Dec 4, 2018)	criteria provided, single submitter	VCV000637978	7	150958409	637978	625802	rs1584866229	Neutral	0.447	0.319	0.234
KCNH2	190	A	T	NM_000238.4(KCNH2):c.568G>A (p.Ala190Thr)	Long QT syndrome|not specified|not provided| Long QT syndrome 2|Short QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Oct 14, 2020)	criteria provided, conflicting interpretations	VCV000067512	7	150958407	67512	78408	rs150817714	Neutral	0.475	0.364	0.162
KCNH2	191	P	L	NM_000238.4(KCNH2):c.572C>T (p.Pro191Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 11, 2020)	criteria provided, single submitter	VCV001027196	7	150958403	1027196	1007355	rs1345107784	Neutral	0.373	0.436	0.191
KCNH2	191	R	Q	NM_000238.4(KCNH2):c.1592G>A (p.Arg531Gln)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067219	7	150951801	67219	78115	rs199473515	Neutral	0.418	0.368	0.214
KCNH2	191	R	W	NM_000238.4(KCNH2):c.1591C>T (p.Arg531Trp)	Congenital long QT syndrome|not specified|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Aug 24, 2021)	criteria provided, conflicting interpretations	VCV000067218	7	150951802	67218	78114	rs199472915	Neutral	0.375	0.444	0.181
KCNH2	192	L	P	NM_000238.4(KCNH2):c.1595T>C (p.Leu532Pro)	Long QT syndrome 1	Likely pathogenic(Last reviewed: Jan 8, 2019)	criteria provided, single submitter	VCV000978356	7	150951798	978356	966439	rs1801182500	Neutral	0.694	0.194	0.112
KCNH2	193	A	V	NM_000238.4(KCNH2):c.578C>T (p.Ala193Val)	not provided	Uncertain significance(Last reviewed: Jul 17, 2019)	criteria provided, single submitter	VCV001304544	7	150958397	1304544	1294819	NA	Neutral	0.513	0.383	0.104
KCNH2	193	V	G	NM_000238.4(KCNH2):c.1598T>G (p.Val533Gly)	not specified	Benign(Last reviewed: Jun 24, 2013)	criteria provided, single submitter	VCV000191574	7	150951795	191574	189261	rs202102276	Neutral	0.657	0.238	0.105
KCNH2	193	V	L	NM_000238.4(KCNH2):c.1597G>C (p.Val533Leu)	Arrhythmia	Uncertain significance(Last reviewed: Mar 16, 2021)	criteria provided, single submitter	VCV001332036	7	150951796	1332036	1322756	NA	Neutral	0.547	0.341	0.112
KCNH2	194	R	C	NM_000238.4(KCNH2):c.1600C>T (p.Arg534Cys)	Congenital long QT syndrome|not provided|Long QT syndrome	Pathogenic(Last reviewed: Oct 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067220	7	150951793	67220	78116	rs199472916	Neutral	0.515	0.351	0.134
KCNH2	194	R	G	NM_000238.4(KCNH2):c.1600C>G (p.Arg534Gly)	Long QT syndrome 2	Uncertain significance(Last reviewed: Apr 27, 2017)	criteria provided, single submitter	VCV000908910	7	150951793	908910	897671	rs199472916	Neutral	0.599	0.275	0.126
KCNH2	194	R	L	NM_000238.4(KCNH2):c.1601G>T (p.Arg534Leu)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Feb 22, 2020)	criteria provided, single submitter	VCV000067221	7	150951792	67221	78117	rs199473516	Neutral	0.482	0.394	0.124
KCNH2	194	R	S	NM_000238.4(KCNH2):c.1600C>A (p.Arg534Ser)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jul 23, 2018)	criteria provided, conflicting interpretations	VCV000200728	7	150951793	200728	197270	rs199472916	Neutral	0.539	0.346	0.114

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	194	V	M	NM_000238.4(KCNH2):c.580G>A (p.Val194Met)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jun 25, 2019)	criteria provided, multiple submitters, no conflicts	VCV000640808	7	150958395	640808	635926	rs1293129095	Neutral	0.552	0.346	0.101
KCNH2	195	V	L	NM_000238.4(KCNH2):c.1603G>T (p.Val535Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 14, 2018)	criteria provided, single submitter	VCV000570842	7	150951790	570842	561728	rs375872367	Neutral	0.486	0.389	0.125
KCNH2	195	V	M	NM_000238.4(KCNH2):c.1603G>A (p.Val535Met)	not provided|Long QT syndrome 2|Long QT syndrome|Cardiovascular phenotype	Uncertain significance(Last reviewed: Apr 15, 2019)	criteria provided, multiple submitters, no conflicts	VCV000161256	7	150951790	161256	171108	rs375872367	Neutral	0.465	0.419	0.115
KCNH2	197	D	N	NM_000238.4(KCNH2):c.589G>A (p.Asp197Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 31, 2019)	criteria provided, single submitter	VCV000952715	7	150958386	952715	924759	rs1396987764	Neutral	0.449	0.325	0.226
KCNH2	197	R	Q	NM_000238.4(KCNH2):c.1610G>A (p.Arg537Gln)	not provided	Likely pathogenic(Last reviewed: Dec 30, 2016)	criteria provided, single submitter	VCV000200360	7	150951783	200360	197269	rs794728373	Neutral	0.401	0.41	0.189
KCNH2	197	R	W	NM_000238.4(KCNH2):c.1609C>T (p.Arg537Trp)	Arrhythmia|Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Apr 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067222	7	150951784	67222	78118	rs199472917	Neutral	0.374	0.468	0.157
KCNH2	198	K	E	NM_000238.4(KCNH2):c.1612A>G (p.Lys538Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 14, 2016)	criteria provided, single submitter	VCV000359311	7	150951781	359311	305392	rs886062088	Neutral	0.492	0.326	0.182
KCNH2	201	R	H	NM_000238.4(KCNH2):c.1622G>A (p.Arg541His)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Jan 5, 2021)	criteria provided, multiple submitters, no conflicts	VCV000960653	7	150951771	960653	945524	rs1297393452	Neutral	0.349	0.442	0.209
KCNH2	201	T	M	NM_000238.4(KCNH2):c.602C>T (p.Thr201Met)	not provided	Uncertain significance(Last reviewed: Apr 23, 2013)	criteria provided, single submitter	VCV000200295	7	150958373	200295	197321	rs794728357	Neutral	0.277	0.549	0.174
KCNH2	203	A	T	NM_000238.4(KCNH2):c.607G>A (p.Ala203Thr)	not provided|Long QT syndrome|Cardiovascular phenotype	Uncertain significance(Last reviewed: Aug 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067514	7	150958368	67514	78410	rs199472868	Neutral	0.404	0.276	0.32
KCNH2	205	P	L	NM_000238.4(KCNH2):c.614C>T (p.Pro205Leu)	Short QT syndrome 1	Uncertain significance(Last reviewed: Jun 25, 2019)	no assertion criteria provided	VCV000829864	7	150958361	829864	818254	rs1326885330	Neutral	0.399	0.369	0.233
KCNH2	205	P	S	NM_000238.4(KCNH2):c.613C>T (p.Pro205Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 6, 2018)	criteria provided, single submitter	VCV000662184	7	150958362	662184	635925	rs939574819	Neutral	0.401	0.344	0.255
KCNH2	206	S	I	NM_000238.4(KCNH2):c.617_618delinsTT (p.Ser206Ile)	not specified|Long QT syndrome	Uncertain significance(Last reviewed: Oct 12, 2020)	criteria provided, multiple submitters, no conflicts	VCV000405339	7	150958357 - 150958358	405339	395495	rs1060500660	Neutral	0.35	0.441	0.209
KCNH2	207	A	V	NM_000238.4(KCNH2):c.1640C>T (p.Ala547Val)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Dec 3, 2019)	criteria provided, multiple submitters, no	VCV000852580	7	150951753	852580	833338	rs1473812774	Neutral	0.367	0.418	0.214

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNH2	209	V	M	NM_000238.4(KCNH2):c.1645G>A (p.Val549Met)	Arrhythmia	Uncertain significance(Last reviewed: Nov 24, 2019)	criteria provided, single submitter	VCV000919780	7	150951748	919780	910760	rs1257206721	Neutral	0.43	0.389	0.182
KCNH2	210	L	P	NM_000238.4(KCNH2):c.629T>C (p.Leu210Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 15, 2019)	criteria provided, single submitter	VCV000938315	7	150958346	938315	933781	rs1243787663	Neutral	0.58	0.246	0.174
KCNH2	211	F	S	NM_000238.4(KCNH2):c.1652T>C (p.Phe551Ser)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000392546	7	150951741	392546	369335	rs1057524535	Neutral	0.591	0.254	0.155
KCNH2	212	L	S	NM_000238.4(KCNH2):c.1655T>C (p.Leu552Ser)	Long QT syndrome 2|Long QT syndrome| Cardiovascular phenotype|Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Apr 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067225	7	150951738	67225	78121	rs199472918	Neutral	0.537	0.282	0.182
KCNH2	213	D	G	NM_000238.4(KCNH2):c.638A>G (p.Asp213Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 28, 2019)	criteria provided, single submitter	VCV000971664	7	150958337	971664	955097	rs531677961	Neutral	0.407	0.305	0.288
KCNH2	214	M	T	NM_000238.4(KCNH2):c.1661T>C (p.Met554Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 30, 2019)	criteria provided, single submitter	VCV000642793	7	150951732	642793	635905	rs1584854609	Neutral	0.386	0.387	0.227
KCNH2	215	C	Y	NM_000238.4(KCNH2):c.1664G>A (p.Cys555Tyr)	not provided	Likely pathogenic(Last reviewed: May 18, 2016)	criteria provided, single submitter	VCV000372574	7	150951729	372574	359636	rs1057517866	Neutral	0.564	0.249	0.186
KCNH2	215	V	G	NM_000238.4(KCNH2):c.644T>G (p.Val215Gly)	not provided	not provided	no assertion provided	VCV000067515	7	150958331	67515	78411	rs199473500	Neutral	0.596	0.261	0.143
KCNH2	216	T	I	NM_000238.4(KCNH2):c.647C>T (p.Thr216Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 9, 2018)	criteria provided, single submitter	VCV000582107	7	150958328	582107	566595	rs1563170569	Neutral	0.331	0.462	0.207
KCNH2	218	A	E	NM_000238.4(KCNH2):c.1673C>A (p.Ala558Glu)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: Jan 22, 2021)	criteria provided, single submitter	VCV000067226	7	150951720	67226	78122	rs199472919	Neutral	0.542	0.274	0.184
KCNH2	218	M	V	NM_000238.4(KCNH2):c.652A>G (p.Met218Val)	not provided|Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Apr 13, 2018)	criteria provided, single submitter	VCV000067516	7	150958323	67516	78412	rs199472869	Neutral	0.457	0.38	0.163
KCNH2	219	D	N	NM_000238.4(KCNH2):c.655G>A (p.Asp219Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 23, 2018)	criteria provided, single submitter	VCV000663570	7	150958320	663570	635924	rs1584865954	Neutral	0.407	0.275	0.318
KCNH2	219	D	V	NM_000238.4(KCNH2):c.656A>T (p.Asp219Val)	Long QT syndrome 2|Long QT syndrome	Uncertain significance(Last reviewed: Jul 5, 2019)	criteria provided, single submitter	VCV000157662	7	150958319	157662	167508	rs587777907	Neutral	0.352	0.366	0.282
KCNH2	219	L	F	NM_000238.4(KCNH2):c.1675C>T (p.Leu559Phe)	not provided	Pathogenic(Last reviewed: Feb 5, 2014)	criteria provided, single submitter	VCV000200363	7	150951718	200363	197268	rs794728374	Neutral	0.45	0.373	0.177
KCNH2	219	L	H	NM_000238.4(KCNH2):c.1676T>A (p.Leu559His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067227	7	150951717	67227	78123	rs199472920	Neutral	0.489	0.313	0.198
KCNH2	220	I	V	NM_000238.4(KCNH2):c.1678A>G (p.Ile560Val)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 17,	criteria provided, single submitter	VCV001055024	7	150951715	1055024	1044809	NA	Neutral	0.482	0.339	0.179

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)											
KCNH2	221	A	P	NM_000238.4(KCNH2):c.1681G>C (p.Ala561Pro)	Cardiovascular phenotype|Congenital long QT syndrome	Pathogenic(Last reviewed: Feb 5, 2018)	criteria provided, single submitter	VCV000067230	7	150951712	67230	78126	rs199472921	Neutral	0.497	0.303	0.199
KCNH2	221	A	T	NM_000238.4(KCNH2):c.1681G>A (p.Ala561Thr)	Long QT syndrome|Cardiovascular phenotype|Inborn genetic diseases|Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Jun 15, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067229	7	150951712	67229	78125	rs199472921	Neutral	0.381	0.414	0.205
KCNH2	221	H	Q	NM_000238.4(KCNH2):c.663C>G (p.His221Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 29, 2018)	criteria provided, single submitter	VCV000647893	7	150958312	647893	635923	rs752896394	Neutral	0.406	0.359	0.235
KCNH2	222	H	P	NM_000238.4(KCNH2):c.1685A>C (p.His562Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067231	7	150951708	67231	78127	rs199472922	Neutral	0.475	0.322	0.203
KCNH2	222	H	Q	NM_000238.4(KCNH2):c.1686C>G (p.His562Gln)	not provided	Likely pathogenic(Last reviewed: Oct 21, 2016)	criteria provided, single submitter	VCV000379052	7	150951707	379052	369333	rs1057520477	Neutral	0.434	0.339	0.227
KCNH2	222	H	R	NM_000238.4(KCNH2):c.1685A>G (p.His562Arg)	Long QT syndrome|Congenital long QT syndrome	Pathogenic(Last reviewed: Oct 1, 2019)	criteria provided, single submitter	VCV000067232	7	150951708	67232	78128	rs199472922	Neutral	0.476	0.281	0.243
KCNH2	222	H	Y	NM_000238.4(KCNH2):c.1684C>T (p.His562Tyr)	not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Aug 1, 2019)	criteria provided, conflicting interpretations	VCV000200733	7	150951709	200733	197265	rs794728481	Neutral	0.386	0.406	0.208
KCNH2	222	V	M	NM_000238.4(KCNH2):c.664G>A (p.Val222Met)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 9, 2019)	criteria provided, single submitter	VCV000835460	7	150958311	835460	833356	rs1263535997	Neutral	0.453	0.387	0.159
KCNH2	223	W	C	NM_000238.4(KCNH2):c.1689G>T (p.Trp563Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067234	7	150951704	67234	78130	rs199473517	Neutral	0.567	0.263	0.169
KCNH2	223	W	C	NM_000238.4(KCNH2):c.1689G>T (p.Trp563Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067234	7	150951704	67234	78130	rs199473517	Neutral	0.567	0.263	0.169
KCNH2	223	W	C	NM_000238.4(KCNH2):c.1689G>C (p.Trp563Cys)	not provided	Likely pathogenic(Last reviewed: Jul 31, 2018)	criteria provided, single submitter	VCV000200368	7	150951704	200368	197264	rs199473517	Neutral	0.567	0.263	0.169
KCNH2	223	W	C	NM_000238.4(KCNH2):c.1689G>C (p.Trp563Cys)	not provided	Likely pathogenic(Last reviewed: Jul 31, 2018)	criteria provided, single submitter	VCV000200368	7	150951704	200368	197264	rs199473517	Neutral	0.567	0.263	0.169
KCNH2	223	W	G	NM_000238.4(KCNH2):c.1687T>G (p.Trp563Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067233	7	150951706	67233	78129	rs199472923	Neutral	0.601	0.231	0.167
KCNH2	224	G	R	NM_000238.4(KCNH2):c.670G>A (p.Gly224Arg)	not provided	Uncertain significance(Last reviewed: Jul 3, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200297	7	150958305	200297	197320	rs794728358	GOF	0.466	0.186	0.348
KCNH2	224	L	P	NM_000238.4(KCNH2):c.1691T>C (p.Leu564Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067235	7	150951702	67235	78131	rs199472924	Neutral	0.575	0.224	0.201
KCNH2	225	A	P	NM_000238.4(KCNH2):c.1693G>C (p.Ala565Pro)	not provided	Likely pathogenic(Last reviewed: Feb 2, 2015)	criteria provided, single submitter	VCV000200372	7	150951700	200372	197262	rs199473518	Neutral	0.526	0.276	0.199
KCNH2	225	A	S	NM_000238.4(KCNH2):c.1693G>T (p.Ala565Ser)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Feb 20,	criteria provided, single submitter	VCV000519393	7	150951700	519393	509866	rs199473518	Neutral	0.445	0.339	0.216

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2017)											
KCNH2	225	A	T	NM_000238.4(KCNH2):c.1693G>A (p.Ala565Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067236	7	150951700	67236	78132	rs199473518	Neutral	0.417	0.371	0.212
KCNH2	225	A	V	NM_000238.4(KCNH2):c.1694C>T (p.Ala565Val)	not provided	Likely pathogenic(Last reviewed: Oct 1, 2019)	criteria provided, single submitter	VCV000200735	7	150951699	200735	197261	rs794728482	Neutral	0.406	0.393	0.201
KCNH2	226	C	F	NM_000238.4(KCNH2):c.1697G>T (p.Cys566Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067239	7	150951696	67239	78135	rs199472925	Neutral	0.536	0.262	0.202
KCNH2	226	C	G	NM_000238.4(KCNH2):c.1696T>G (p.Cys566Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067237	7	150951697	67237	78133	rs199473038	Neutral	0.597	0.217	0.186
KCNH2	226	C	S	NM_000238.4(KCNH2):c.1697G>C (p.Cys566Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067238	7	150951696	67238	78134	rs199472925	Neutral	0.533	0.237	0.23
KCNH2	227	I	T	NM_000238.4(KCNH2):c.1700T>C (p.Ile567Thr)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Aug 31, 2018)	criteria provided, single submitter	VCV000067241	7	150951693	67241	78137	rs199473519	Neutral	0.458	0.301	0.242
KCNH2	228	A	P	NM_000238.4(KCNH2):c.682G>C (p.Ala228Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 30, 2020)	criteria provided, single submitter	VCV000526974	7	150958293	526974	523146	rs989191027	Neutral	0.634	0.191	0.175
KCNH2	228	A	V	NM_000238.4(KCNH2):c.683C>T (p.Ala228Val)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 10, 2018)	criteria provided, single submitter	VCV000659941	7	150958292	659941	635922	rs1184559298	Neutral	0.535	0.288	0.177
KCNH2	228	W	C	NM_000238.4(KCNH2):c.1704G>C (p.Trp568Cys)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Aug 8, 2018)	criteria provided, single submitter	VCV000067243	7	150951689	67243	78139	rs199472926	Neutral	0.652	0.205	0.143
KCNH2	228	W	C	NM_000238.4(KCNH2):c.1704G>C (p.Trp568Cys)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Aug 8, 2018)	criteria provided, single submitter	VCV000067243	7	150951689	67243	78139	rs199472926	Neutral	0.652	0.205	0.143
KCNH2	228	W	C	NM_000238.4(KCNH2):c.1704G>T (p.Trp568Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067244	7	150951689	67244	78140	rs199472926	Neutral	0.652	0.205	0.143
KCNH2	228	W	C	NM_000238.4(KCNH2):c.1704G>T (p.Trp568Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067244	7	150951689	67244	78140	rs199472926	Neutral	0.652	0.205	0.143
KCNH2	228	W	R	NM_000238.4(KCNH2):c.1702T>C (p.Trp568Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067242	7	150951691	67242	78138	rs199472927	LOF	0.681	0.141	0.178
KCNH2	228	W	S	NM_000238.4(KCNH2):c.1703G>C (p.Trp568Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 31, 2016)	criteria provided, single submitter	VCV000520444	7	150951690	520444	511087	rs1554425880	Neutral	0.668	0.179	0.153
KCNH2	229	Y	H	NM_000238.4(KCNH2):c.1705T>C (p.Tyr569His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067245	7	150951688	67245	78141	rs199473520	Neutral	0.556	0.27	0.174
KCNH2	230	A	S	NM_000238.4(KCNH2):c.1708G>T (p.Ala570Ser)	not provided	Likely pathogenic	no assertion criteria provided	VCV001284270	7	150951685	1284270	1274125	NA	Neutral	0.546	0.285	0.169
KCNH2	230	A	T	NM_000238.4(KCNH2):c.1708G>A (p.Ala570Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 17, 2020)	criteria provided, single submitter	VCV001043586	7	150951685	1043586	1027888	rs1727086844	Neutral	0.52	0.314	0.166
KCNH2	231	I	L	NM_000238.4(KCNH2):c.1711A>C (p.Ile571Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067246	7	150951682	67246	78142	rs199472928	Neutral	0.61	0.26	0.129
KCNH2	231	I	V	NM_000238.4(KCNH2):c.1711A>G (p.Ile571Val)	not provided|Long QT syndrome|Congenital long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Aug 11, 2017)	criteria provided, conflicting interpretations	VCV000067247	7	150951682	67247	78143	rs199472928	Neutral	0.615	0.253	0.132

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	231	R	Q	NM_000238.4(KCNH2):c.692G>A (p.Arg231Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 11, 2020)	criteria provided, single submitter	VCV000952892	7	150958283	952892	924758	rs1383803455	Neutral	0.57	0.254	0.176
KCNH2	232	G	A	NM_000238.4(KCNH2):c.1715G>C (p.Gly572Ala)	not provided	Likely pathogenic(Last reviewed: Dec 19, 2016)	criteria provided, single submitter	VCV000391858	7	150951678	391858	369040	rs199473423	Neutral	0.416	0.34	0.244
KCNH2	232	G	C	NM_000238.4(KCNH2):c.1714G>T (p.Gly572Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067249	7	150951679	67249	78145	rs9333649	Neutral	0.459	0.336	0.205
KCNH2	232	G	D	NM_000238.4(KCNH2):c.1715G>A (p.Gly572Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067250	7	150951678	67250	78146	rs199473423	Neutral	0.496	0.265	0.239
KCNH2	232	G	S	NM_000238.4(KCNH2):c.1714G>A (p.Gly572Ser)	not provided|Long QT syndrome 2|Long QT syndrome|Cardiovascular phenotype|Congenital long QT syndrome	Pathogenic(Last reviewed: Nov 8, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067248	7	150951679	67248	78144	rs9333649	Neutral	0.429	0.301	0.27
KCNH2	232	G	V	NM_000238.4(KCNH2):c.1715G>T (p.Gly572Val)	not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: May 4, 2016)	criteria provided, single submitter	VCV000067251	7	150951678	67251	78147	rs199473423	Neutral	0.421	0.34	0.239
KCNH2	232	R	C	NM_000238.4(KCNH2):c.694C>T (p.Arg232Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 27, 2020)	criteria provided, single submitter	VCV000855032	7	150958281	855032	833354	rs956828658	Neutral	0.468	0.353	0.179
KCNH2	232	R	L	NM_000238.4(KCNH2):c.695G>T (p.Arg232Leu)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Nov 18, 2015)	criteria provided, single submitter	VCV000264446	7	150958280	264446	258464	rs886039160	Neutral	0.412	0.398	0.19
KCNH2	233	A	S	NM_000238.4(KCNH2):c.697G>T (p.Ala233Ser)	Long QT syndrome|not provided	Likely benign(Last reviewed: May 13, 2020)	criteria provided, multiple submitters, no conflicts	VCV000527096	7	150958278	527096	522697	rs772037564	Neutral	0.422	0.28	0.298
KCNH2	233	A	T	NM_000238.4(KCNH2):c.697G>A (p.Ala233Thr)	Long QT syndrome	Uncertain significance(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000848897	7	150958278	848897	833353	rs772037564	Neutral	0.403	0.298	0.299
KCNH2	234	M	V	NM_000238.4(KCNH2):c.1720A>G (p.Met574Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067252	7	150951673	67252	78148	rs199473667	Neutral	0.505	0.31	0.184
KCNH2	235	E	G	NM_000238.4(KCNH2):c.1724A>G (p.Glu575Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067253	7	150951669	67253	78149	rs199473424	Neutral	0.483	0.305	0.212
KCNH2	235	V	A	NM_000238.4(KCNH2):c.704T>C (p.Val235Ala)	not provided	Uncertain significance	no assertion criteria provided	VCV001049705	7	150958271	1049705	1037760	NA	Neutral	0.487	0.334	0.179
KCNH2	236	G	D	NM_000238.4(KCNH2):c.707G>A (p.Gly236Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 6, 2020)	criteria provided, single submitter	VCV000952259	7	150958268	952259	924757	rs199472870	GOF	0.476	0.214	0.31
KCNH2	236	G	V	NM_000238.4(KCNH2):c.707G>T (p.Gly236Val)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Aug 4, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067518	7	150958268	67518	78414	rs199472870	Neutral	0.412	0.284	0.304
KCNH2	237	P	H	NM_000238.4(KCNH2):c.710C>A (p.Pro237His)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 8, 2018)	criteria provided, single submitter	VCV000526973	7	150958265	526973	522781	rs1554427821	Neutral	0.359	0.306	0.336
KCNH2	238	G	S	NM_000238.4(KCNH2):c.712G>A (p.Gly238Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067519	7	150958263	67519	78415	rs199473501	GOF	0.381	0.231	0.388
KCNH2	238	H	P	NM_000238.4(KCNH2):c.1733A>C (p.His578Pro)	not provided	Uncertain significance(Last	criteria provided, single	VCV000200378	7	150951660	200378	197258	rs794728376	Neutral	0.412	0.272	0.315

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: May 10, 2014)	submitter										
KCNH2	239	M	T	NM_000238.4(KCNH2):c.1736T>C (p.Met579Thr)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: May 31, 2020)	criteria provided, single submitter	VCV000067255	7	150951657	67255	78151	rs199473425	Neutral	0.392	0.317	0.291
KCNH2	240	D	H	NM_000238.4(KCNH2):c.1738G>C (p.Asp580His)	not provided	Likely pathogenic	no assertion criteria provided	VCV000191223	7	150951655	191223	189026	rs786205588	GOF	0.388	0.275	0.337
KCNH2	240	P	L	NM_000238.4(KCNH2):c.719C>T (p.Pro240Leu)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Jul 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200299	7	150958256	200299	197318	rs794728359	Neutral	0.413	0.333	0.254
KCNH2	241	P	L	NM_000238.4(KCNH2):c.722C>T (p.Pro241Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067520	7	150958253	67520	78416	rs199472871	Neutral	0.365	0.351	0.283
KCNH2	242	R	C	NM_000238.4(KCNH2):c.724C>T (p.Arg242Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 12, 2019)	criteria provided, single submitter	VCV000652662	7	150958251	652662	635921	rs199472872	Neutral	0.412	0.356	0.232
KCNH2	242	R	G	NM_000238.4(KCNH2):c.724C>G (p.Arg242Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067521	7	150958251	67521	78417	rs199472872	Neutral	0.437	0.309	0.253
KCNH2	242	R	L	NM_000238.4(KCNH2):c.1745G>T (p.Arg582Leu)	Congenital long QT syndrome|Long QT syndrome	Likely pathogenic(Last reviewed: Mar 25, 2020)	criteria provided, single submitter	VCV000067258	7	150951648	67258	78154	rs199473426	Neutral	0.372	0.372	0.257
KCNH2	242	R	Q	NM_000238.4(KCNH2):c.725_726delinsAA (p.Arg242Gln)	Long QT syndrome	Uncertain significance(Last reviewed: May 30, 2017)	criteria provided, single submitter	VCV000456938	7	150958249 - 150958250	456938	457576	rs1554427803	GOF	0.401	0.281	0.319
KCNH2	243	I	M	NM_000238.4(KCNH2):c.1749C>G (p.Ile583Met)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 25, 2020)	criteria provided, single submitter	VCV001023474	7	150951644	1023474	1007349	rs765893780	Neutral	0.456	0.369	0.175
KCNH2	243	I	V	NM_000238.4(KCNH2):c.1747A>G (p.Ile583Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067259	7	150951646	67259	78155	rs199473427	Neutral	0.485	0.331	0.184
KCNH2	243	S	R	NM_000238.4(KCNH2):c.729C>G (p.Ser243Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 19, 2018)	criteria provided, single submitter	VCV000648410	7	150958246	648410	635920	rs863224337	Neutral	0.411	0.281	0.308
KCNH2	244	A	E	NM_000238.4(KCNH2):c.731C>A (p.Ala244Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 6, 2019)	criteria provided, single submitter	VCV000969507	7	150958244	969507	955096	rs748762712	Neutral	0.509	0.271	0.22
KCNH2	244	A	V	NM_000238.4(KCNH2):c.731C>T (p.Ala244Val)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 31, 2018)	criteria provided, single submitter	VCV000405349	7	150958244	405349	395868	rs748762712	Neutral	0.413	0.365	0.222
KCNH2	244	G	C	NM_000238.4(KCNH2):c.1750G>T (p.Gly584Cys)	Congenital long QT syndrome|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 17, 2018)	criteria provided, conflicting interpretations	VCV000067263	7	150951643	67263	78159	rs199473428	Neutral	0.453	0.33	0.216
KCNH2	244	G	R	NM_000238.4(KCNH2):c.1750G>C (p.Gly584Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067262	7	150951643	67262	78158	rs199473428	Neutral	0.501	0.218	0.281
KCNH2	244	G	S	NM_000238.4(KCNH2):c.1750G>A (p.Gly584Ser)	Cardiovascular phenotype|Congenital long QT syndrome|Long QT syndrome 2|Short QT syndrome 1|not provided|Long QT syndrome|Arrhythmia|Long QT syndrome 2	Pathogenic/Likely pathogenic(Last reviewed: Oct 11, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067261	7	150951643	67261	78157	rs199473428	Neutral	0.44	0.288	0.272

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	244	G	V	NM_000238.4(KCNH2):c.1751G>T (p.Gly584Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067264	7	150951642	67264	78160	rs199473429	Neutral	0.424	0.326	0.25
KCNH2	245	W	C	NM_000238.4(KCNH2):c.1755G>T (p.Trp585Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067265	7	150951638	67265	78161	rs199473430	Neutral	0.532	0.295	0.173
KCNH2	245	W	L	NM_000238.4(KCNH2):c.1754G>T (p.Trp585Leu)	not provided	Likely pathogenic(Last reviewed: Feb 27, 2012)	criteria provided, single submitter	VCV000200739	7	150951639	200739	197256	rs794728484	Neutral	0.479	0.328	0.193
KCNH2	246	G	C	NM_000238.4(KCNH2):c.736G>T (p.Gly246Cys)	not specified	Likely benign(Last reviewed: May 11, 2015)	criteria provided, single submitter	VCV000379687	7	150958239	379687	371000	rs1057520689	Neutral	0.413	0.3	0.287
KCNH2	246	L	V	NM_000238.4(KCNH2):c.1756C>G (p.Leu586Val)	not provided	Uncertain significance(Last reviewed: Mar 9, 2020)	criteria provided, single submitter	VCV000993664	7	150951637	993664	981595	rs1801169246	Neutral	0.471	0.296	0.232
KCNH2	247	Q	R	NM_000238.4(KCNH2):c.740A>G (p.Gln247Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 25, 2020)	criteria provided, single submitter	VCV001057001	7	150958235	1057001	1044812	NA	GOF	0.408	0.233	0.359
KCNH2	248	N	D	NM_000238.4(KCNH2):c.1762A>G (p.Asn588Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067266	7	150951631	67266	78162	rs199473431	Neutral	0.475	0.284	0.241
KCNH2	250	G	V	NM_000238.4(KCNH2):c.1769G>T (p.Gly590Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067267	7	150951624	67267	78163	rs199472929	Neutral	0.395	0.299	0.306
KCNH2	251	P	A	NM_000238.4(KCNH2):c.751C>G (p.Pro251Ala)	not provided	not provided	no assertion provided	VCV000067522	7	150958224	67522	78418	rs199472873	Neutral	0.401	0.354	0.244
KCNH2	251	P	S	NM_000238.4(KCNH2):c.751C>T (p.Pro251Ser)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jan 30, 2018)	criteria provided, single submitter	VCV000067523	7	150958224	67523	78419	rs199472873	Neutral	0.403	0.319	0.278
KCNH2	252	R	G	NM_000238.4(KCNH2):c.754C>G (p.Arg252Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067524	7	150958221	67524	78420	rs199472874	Neutral	0.452	0.304	0.244
KCNH2	252	R	Q	NM_000238.4(KCNH2):c.755G>A (p.Arg252Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 12, 2020)	criteria provided, single submitter	VCV000180380	7	150958220	180380	178577	rs730880117	GOF	0.415	0.278	0.308
KCNH2	253	A	G	NM_000238.4(KCNH2):c.758C>G (p.Ala253Gly)	not provided	Uncertain significance(Last reviewed: Oct 27, 2014)	criteria provided, single submitter	VCV000200723	7	150958217	200723	197314	rs794728479	Neutral	0.478	0.303	0.219
KCNH2	253	I	K	NM_000238.4(KCNH2):c.1778T>A (p.Ile593Lys)	Long QT syndrome|Congenital long QT syndrome	Pathogenic(Last reviewed: Jul 25, 2017)	criteria provided, single submitter	VCV000067269	7	150951615	67269	78165	rs28928904	Neutral	0.576	0.235	0.188
KCNH2	253	I	T	NM_000238.4(KCNH2):c.1778T>C (p.Ile593Thr)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Feb 2, 2014)	criteria provided, single submitter	VCV000067270	7	150951615	67270	78166	rs28928904	Neutral	0.495	0.307	0.198
KCNH2	253	I	V	NM_000238.4(KCNH2):c.1777A>G (p.Ile593Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067268	7	150951616	67268	78164	rs199472930	Neutral	0.512	0.31	0.177
KCNH2	254	G	D	NM_000238.4(KCNH2):c.1781G>A (p.Gly594Asp)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Jul 31, 2019)	criteria provided, single submitter	VCV000067271	7	150951612	67271	78167	rs199472931	Neutral	0.462	0.274	0.264
KCNH2	254	G	R	NM_000238.4(KCNH2):c.1780G>C (p.Gly594Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 27, 2020)	criteria provided, single submitter	VCV001026973	7	150951613	1026973	1007348	rs1801167694	Neutral	0.465	0.249	0.286
KCNH2	254	H	Q	NM_000238.4(KCNH2):c.762C>G (p.His254Gln)	not provided	not provided	no assertion provided	VCV000067525	7	150958213	67525	78421	rs199473502	Neutral	0.404	0.321	0.275
KCNH2	255	K	E	NM_000238.4(KCNH2):c.1783A>G	Congenital long QT syndrome	not provided	no assertion	VCV000067272	7	150951610	67272	78168	rs199472932	Neutral	0.425	0.278	0.297

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Lys595Glu)			provided										
KCNH2	255	K	N	NM_000238.4(KCNH2):c.1785A>T (p.Lys595Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067273	7	150951608	67273	78169	rs199473521	GOF	0.384	0.284	0.333
KCNH2	256	P	A	NM_000238.4(KCNH2):c.1786C>G (p.Pro596Ala)	Long QT syndrome 2	Pathogenic(Last reviewed: Nov 4, 2015)	criteria provided, single submitter	VCV000218093	7	150951607	218093	214747	rs863225288	Neutral	0.38	0.406	0.215
KCNH2	256	P	H	NM_000238.4(KCNH2):c.1787C>A (p.Pro596His)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Jan 31, 2014)	criteria provided, single submitter	VCV000067274	7	150951606	67274	78170	rs199472933	Neutral	0.395	0.376	0.229
KCNH2	256	P	L	NM_000238.4(KCNH2):c.1787C>T (p.Pro596Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067276	7	150951606	67276	78172	rs199472933	Neutral	0.382	0.415	0.204
KCNH2	256	P	R	NM_000238.4(KCNH2):c.1787C>G (p.Pro596Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067275	7	150951606	67275	78171	rs199472933	Neutral	0.453	0.278	0.268
KCNH2	257	N	H	NM_000238.4(KCNH2):c.769A>C (p.Asn257His)	not provided	Uncertain significance(Last reviewed: Nov 12, 2018)	criteria provided, single submitter	VCV000067526	7	150958206	67526	78422	rs199472875	Neutral	0.347	0.333	0.32
KCNH2	257	Y	C	NM_000238.4(KCNH2):c.1790A>G (p.Tyr597Cys)	Long QT syndrome|Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: Nov 18, 2015)	criteria provided, multiple submitters, no conflicts	VCV000067277	7	150951603	67277	78173	rs199472934	Neutral	0.411	0.389	0.2
KCNH2	258	P	L	NM_000238.4(KCNH2):c.773C>T (p.Pro258Leu)	not provided	Uncertain significance(Last reviewed: Dec 19, 2016)	criteria provided, single submitter	VCV000495747	7	150958202	495747	487314	rs1367019959	Neutral	0.349	0.366	0.285
KCNH2	258	P	S	NM_000238.4(KCNH2):c.772C>T (p.Pro258Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV001060097	7	150958203	1060097	1044811	NA	GOF	0.346	0.302	0.352
KCNH2	259	D	N	NM_000238.4(KCNH2):c.775G>A (p.Asp259Asn)	not provided|Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: May 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067527	7	150958200	67527	78423	rs199472876	GOF	0.395	0.234	0.371
KCNH2	259	S	N	NM_000238.4(KCNH2):c.1796G>A (p.Ser599Asn)	Arrhythmia	Uncertain significance(Last reviewed: Dec 21, 2020)	criteria provided, single submitter	VCV001172277	7	150951597	1172277	1160697	NA	GOF	0.385	0.248	0.367
KCNH2	259	S	R	NM_000238.4(KCNH2):c.1797C>A (p.Ser599Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067278	7	150951596	67278	78174	rs199472935	GOF	0.39	0.233	0.377
KCNH2	260	A	P	NM_000238.4(KCNH2):c.778G>C (p.Ala260Pro)	Hypertrophic cardiomyopathy|Long QT syndrome	Uncertain significance(Last reviewed: Oct 17, 2018)	criteria provided, multiple submitters, no conflicts	VCV000427970	7	150958197	427970	419276	rs1220897906	Neutral	0.45	0.259	0.291
KCNH2	260	A	V	NM_000238.4(KCNH2):c.779C>T (p.Ala260Val)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 26, 2018)	criteria provided, single submitter	VCV000571094	7	150958196	571094	561740	rs1228602666	Neutral	0.365	0.344	0.292
KCNH2	261	G	C	NM_000238.4(KCNH2):c.1801G>T (p.Gly601Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067280	7	150951592	67280	78176	rs199472936	Neutral	0.394	0.331	0.275
KCNH2	261	G	D	NM_000238.4(KCNH2):c.1802G>A (p.Gly601Asp)	not provided	Likely pathogenic(Last reviewed: Jul 14, 2021)	criteria provided, single submitter	VCV000200730	7	150951591	200730	197255	rs794728480	GOF	0.445	0.223	0.332
KCNH2	261	G	S	NM_000238.4(KCNH2):c.1801G>A (p.Gly601Ser)	Long QT syndrome 2|Long QT syndrome|Cardiovascular phenotype|Congenital long QT syndrome	Pathogenic(Last reviewed: Oct 5, 2020)	criteria provided, multiple	VCV000067279	7	150951592	67279	78175	rs199472936	GOF	0.39	0.262	0.348

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							submitters, no conflicts										
KCNH2	262	L	P	NM_000238.4(KCNH2):c.1805T>C (p.Leu602Pro)	Long QT syndrome|Cardiovascular phenotype| not specified|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jun 8, 2020)	criteria provided, conflicting interpretations	VCV000234996	7	150951588	234996	236770	rs876661348	Neutral	0.525	0.258	0.217
KCNH2	263	G	D	NM_000238.4(KCNH2):c.1808G>A (p.Gly603Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 8, 2020)	criteria provided, single submitter	VCV000456894	7	150951585	456894	457544	rs1554425806	GOF	0.49	0.22	0.29
KCNH2	264	G	D	NM_000238.4(KCNH2):c.1811G>A (p.Gly604Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067282	7	150951582	67282	78178	rs199472937	Neutral	0.472	0.237	0.291
KCNH2	264	G	S	NM_000238.4(KCNH2):c.1810G>A (p.Gly604Ser)	Congenital long QT syndrome|Long QT syndrome|Inborn genetic diseases|not provided	Pathogenic(Last reviewed: Oct 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067281	7	150951583	67281	78177	rs199473522	Neutral	0.412	0.295	0.293
KCNH2	264	G	V	NM_000238.4(KCNH2):c.1811G>T (p.Gly604Val)	not provided	Likely pathogenic(Last reviewed: Jan 3, 2018)	criteria provided, single submitter	VCV000200741	7	150951582	200741	197254	rs199472937	Neutral	0.389	0.342	0.269
KCNH2	265	P	L	NM_000238.4(KCNH2):c.1814C>T (p.Pro605Leu)	Long QT syndrome 2|Long QT syndrome| Congenital long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jul 11, 2018)	criteria provided, conflicting interpretations	VCV000067284	7	150951579	67284	78180	rs199472938	Neutral	0.393	0.426	0.181
KCNH2	265	P	S	NM_000238.4(KCNH2):c.1813C>T (p.Pro605Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067283	7	150951580	67283	78179	rs199472939	Neutral	0.412	0.39	0.198
KCNH2	266	S	I	NM_000238.4(KCNH2):c.797G>T (p.Ser266Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV001060485	7	150958178	1060485	1044810	NA	Neutral	0.384	0.429	0.187
KCNH2	266	S	P	NM_000238.4(KCNH2):c.1816T>C (p.Ser606Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067285	7	150951577	67285	78181	rs199473523	Neutral	0.491	0.306	0.202
KCNH2	267	I	F	NM_000238.4(KCNH2):c.1819A>T (p.Ile607Phe)	not provided	Likely pathogenic(Last reviewed: Jan 2, 2015)	criteria provided, single submitter	VCV000418245	7	150951574	418245	407061	rs1064793146	Neutral	0.492	0.37	0.138
KCNH2	268	A	D	NM_000238.4(KCNH2):c.803C>A (p.Ala268Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 4, 2019)	criteria provided, single submitter	VCV000941477	7	150958172	941477	933780	rs1241943196	Neutral	0.552	0.263	0.185
KCNH2	269	D	A	NM_000238.4(KCNH2):c.1826A>C (p.Asp609Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 9, 2017)	criteria provided, single submitter	VCV000456895	7	150951567	456895	456493	rs199472940	Neutral	0.356	0.371	0.272
KCNH2	269	D	G	NM_000238.4(KCNH2):c.1826A>G (p.Asp609Gly)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Jun 7, 2019)	criteria provided, single submitter	VCV000067289	7	150951567	67289	78185	rs199472940	Neutral	0.419	0.31	0.27
KCNH2	269	D	H	NM_000238.4(KCNH2):c.1825G>C (p.Asp609His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067287	7	150951568	67287	78183	rs199472941	Neutral	0.37	0.337	0.293
KCNH2	269	D	N	NM_000238.4(KCNH2):c.1825G>A (p.Asp609Asn)	Congenital long QT syndrome|Long QT syndrome|not provided	Pathogenic(Last reviewed: Aug 22, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067286	7	150951568	67286	78182	rs199472941	Neutral	0.419	0.274	0.307
KCNH2	269	D	Y	NM_000238.4(KCNH2):c.1825G>T (p.Asp609Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067288	7	150951568	67288	78184	rs199472941	Neutral	0.351	0.361	0.289

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	269	R	W	NM_000238.4(KCNH2):c.805C>T (p.Arg269Trp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067529	7	150958170	67529	78425	rs199473503	Neutral	0.367	0.434	0.2
KCNH2	270	K	E	NM_000238.4(KCNH2):c.1828A>G (p.Lys610Glu)	Long QT syndrome	Likely pathogenic(Last reviewed: May 16, 2020)	criteria provided, single submitter	VCV001066309	7	150951565	1066309	1055669	NA	Neutral	0.49	0.328	0.182
KCNH2	271	R	H	NM_000238.4(KCNH2):c.812G>A (p.Arg271His)	not provided	Uncertain significance(Last reviewed: Jul 7, 2014)	criteria provided, single submitter	VCV000200304	7	150958163	200304	197312	rs794728361	Neutral	0.385	0.388	0.227
KCNH2	271	Y	C	NM_000238.4(KCNH2):c.1832A>G (p.Tyr611Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 22, 2020)	criteria provided, single submitter	VCV001019935	7	150951561	1019935	1007347	rs1801163462	Neutral	0.414	0.405	0.181
KCNH2	271	Y	D	NM_000238.4(KCNH2):c.1831T>G (p.Tyr611Asp)	Congenital long QT syndrome|Long QT syndrome	Likely pathogenic	no assertion criteria provided	VCV000067290	7	150951562	67290	78186	rs199472942	Neutral	0.499	0.296	0.205
KCNH2	272	V	L	NM_000238.4(KCNH2):c.1834G>T (p.Val612Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067291	7	150951559	67291	78187	rs199472943	Neutral	0.52	0.316	0.163
KCNH2	273	R	G	NM_000238.4(KCNH2):c.817C>G (p.Arg273Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 27, 2020)	criteria provided, single submitter	VCV001044178	7	150958158	1044178	1027897	rs552583527	Neutral	0.533	0.264	0.203
KCNH2	273	R	Q	NM_000238.4(KCNH2):c.818G>A (p.Arg273Gln)	SUDDEN INFANT DEATH SYNDROME	not provided	no assertion provided	VCV000067531	7	150958157	67531	78427	rs199472877	Neutral	0.509	0.259	0.233
KCNH2	273	T	M	NM_000238.4(KCNH2):c.1838C>T (p.Thr613Met)	Long QT syndrome|Cardiovascular phenotype|Short QT syndrome 1|not provided|Congenital long QT syndrome|Long QT syndrome 2	Pathogenic/Likely pathogenic(Last reviewed: Sep 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067292	7	150951555	67292	78188	rs199473524	Neutral	0.413	0.397	0.19
KCNH2	273	T	S	NM_000238.4(KCNH2):c.1837A>T (p.Thr613Ser)	not provided	Pathogenic(Last reviewed: Sep 12, 2014)	criteria provided, single submitter	VCV000200743	7	150951556	200743	197252	rs794728485	Neutral	0.47	0.321	0.209
KCNH2	275	L	F	NM_000238.4(KCNH2):c.1843C>T (p.Leu615Phe)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: Aug 6, 2020)	criteria provided, single submitter	VCV000067294	7	150951550	67294	78190	rs199472945	Neutral	0.629	0.252	0.119
KCNH2	275	L	V	NM_000238.4(KCNH2):c.1843C>G (p.Leu615Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067293	7	150951550	67293	78189	rs199472945	Neutral	0.634	0.245	0.121
KCNH2	275	S	R	NM_000238.4(KCNH2):c.823A>C (p.Ser275Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 8, 2020)	criteria provided, single submitter	VCV000577151	7	150958152	577151	566583	rs1268162009	Neutral	0.627	0.172	0.201
KCNH2	275	S	R	NM_000238.4(KCNH2):c.823A>C (p.Ser275Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 8, 2020)	criteria provided, single submitter	VCV000577151	7	150958152	577151	566583	rs1268162009	Neutral	0.627	0.172	0.201
KCNH2	275	S	R	NM_000238.4(KCNH2):c.825C>G (p.Ser275Arg)	Long QT syndrome 2	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000911914	7	150958150	911914	897676	rs1801442023	Neutral	0.627	0.172	0.201
KCNH2	275	S	R	NM_000238.4(KCNH2):c.825C>G (p.Ser275Arg)	Long QT syndrome 2	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000911914	7	150958150	911914	897676	rs1801442023	Neutral	0.627	0.172	0.201
KCNH2	276	Y	C	NM_000238.4(KCNH2):c.1847A>G (p.Tyr616Cys)	Congenital long QT syndrome|Long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last reviewed: Apr 12, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067295	7	150951546	67295	78191	rs199472946	Neutral	0.481	0.357	0.162
KCNH2	277	A	D	NM_000238.4(KCNH2):c.830C>A (p.Ala277Asp)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last	criteria provided, single	VCV000067533	7	150958145	67533	78429	rs199472878	Neutral	0.496	0.258	0.246

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Feb 2, 2020)	submitter										
KCNH2	277	A	S	NM_000238.4(KCNH2):c.829G>T (p.Ala277Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 22, 2019)	criteria provided, single submitter	VCV000834430	7	150958146	834430	833351	rs1801441796	Neutral	0.422	0.337	0.242
KCNH2	277	F	L	NM_000238.4(KCNH2):c.1849T>C (p.Phe617Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 6, 2019)	criteria provided, single submitter	VCV000207940	7	150951544	207940	204115	rs796052195	Neutral	0.468	0.349	0.184
KCNH2	277	F	L	NM_000238.4(KCNH2):c.1849T>C (p.Phe617Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 6, 2019)	criteria provided, single submitter	VCV000207940	7	150951544	207940	204115	rs796052195	Neutral	0.468	0.349	0.184
KCNH2	277	F	L	NM_000238.4(KCNH2):c.1851C>A (p.Phe617Leu)	not provided	Likely pathogenic(Last reviewed: Mar 30, 2017)	criteria provided, single submitter	VCV000392690	7	150951542	392690	369332	rs1057524595	Neutral	0.468	0.349	0.184
KCNH2	277	F	L	NM_000238.4(KCNH2):c.1851C>A (p.Phe617Leu)	not provided	Likely pathogenic(Last reviewed: Mar 30, 2017)	criteria provided, single submitter	VCV000392690	7	150951542	392690	369332	rs1057524595	Neutral	0.468	0.349	0.184
KCNH2	278	S	C	NM_000238.4(KCNH2):c.832A>T (p.Ser278Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 18, 2020)	criteria provided, single submitter	VCV001026541	7	150958143	1026541	1007353	rs1801441560	Neutral	0.395	0.416	0.189
KCNH2	278	T	I	NM_000238.4(KCNH2):c.1853C>T (p.Thr618Ile)	Long QT syndrome|short QT syndrome	Likely pathogenic(Last reviewed: May 10, 2017)	criteria provided, single submitter	VCV000067297	7	150951540	67297	78193	rs199472947	Neutral	0.399	0.422	0.179
KCNH2	278	T	S	NM_000238.4(KCNH2):c.1853C>G (p.Thr618Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067296	7	150951540	67296	78192	rs199472947	Neutral	0.434	0.374	0.191
KCNH2	279	V	M	NM_000238.4(KCNH2):c.835G>A (p.Val279Met)	SUDDEN INFANT DEATH SYNDROME	not provided	no assertion provided	VCV000067534	7	150958140	67534	78430	rs199472879	Neutral	0.562	0.287	0.151
KCNH2	280	R	C	NM_000238.4(KCNH2):c.838C>T (p.Arg280Cys)	Long QT syndrome	Uncertain significance(Last reviewed: May 18, 2019)	criteria provided, single submitter	VCV000943502	7	150958137	943502	933779	rs781669869	Neutral	0.395	0.395	0.21
KCNH2	280	S	G	NM_000238.4(KCNH2):c.1858A>G (p.Ser620Gly)	not provided	Pathogenic(Last reviewed: Mar 5, 2021)	criteria provided, single submitter	VCV001189275	7	150951535	1189275	1180304	NA	Neutral	0.42	0.342	0.237
KCNH2	280	S	N	NM_000238.4(KCNH2):c.1859G>A (p.Ser620Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 9, 2020)	criteria provided, single submitter	VCV001017668	7	150951534	1017668	1007346	rs1801161235	Neutral	0.415	0.317	0.268
KCNH2	281	S	N	NM_000238.4(KCNH2):c.1862G>A (p.Ser621Asn)	Congenital long QT syndrome|Primary familial hypertrophic cardiomyopathy|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Mar 18, 2020)	criteria provided, conflicting interpretations	VCV000067299	7	150951531	67299	78195	rs199472948	Neutral	0.413	0.283	0.304
KCNH2	281	S	R	NM_000238.4(KCNH2):c.1863C>G (p.Ser621Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067300	7	150951530	67300	78196	rs199472949	GOF	0.411	0.241	0.348
KCNH2	282	L	F	NM_000238.4(KCNH2):c.1864C>T (p.Leu622Phe)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Mar 15, 2017)	criteria provided, single submitter	VCV000067301	7	150951529	67301	78197	rs199473525	Neutral	0.547	0.322	0.131
KCNH2	283	T	I	NM_000238.4(KCNH2):c.1868C>T (p.Thr623Ile)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067302	7	150951525	67302	78198	rs199472950	Neutral	0.403	0.428	0.169
KCNH2	285	A	S	NM_000238.4(KCNH2):c.853G>T (p.Ala285Ser)	Long QT syndrome	Uncertain	criteria	VCV000359315	7	150958122	359315	310199	rs864622366	Neutral	0.455	0.34	0.205

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						significance(Last reviewed: Jun 14, 2016)	provided, single submitter										
KCNH2	285	A	T	NM_000238.4(KCNH2):c.853G>A (p.Ala285Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 29, 2015)	criteria provided, single submitter	VCV000220070	7	150958122	220070	221674	rs864622366	Neutral	0.429	0.378	0.193
KCNH2	285	V	A	NM_000238.4(KCNH2):c.1874T>C (p.Val625Ala)	Long QT syndrome 2|Long QT syndrome	Likely pathogenic(Last reviewed: Nov 26, 2015)	criteria provided, multiple submitters, no conflicts	VCV000222668	7	150951519	222668	224343	rs199472951	Neutral	0.461	0.378	0.161
KCNH2	285	V	E	NM_000238.4(KCNH2):c.1874T>A (p.Val625Glu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067303	7	150951519	67303	78199	rs199472951	Neutral	0.569	0.242	0.189
KCNH2	285	V	L	NM_000238.4(KCNH2):c.1873G>C (p.Val625Leu)	not provided	Likely pathogenic(Last reviewed: Jun 26, 2017)	criteria provided, single submitter	VCV000618184	7	150951520	618184	609660	rs1563156725	Neutral	0.467	0.367	0.167
KCNH2	286	G	A	NM_000238.4(KCNH2):c.1877G>C (p.Gly626Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067306	7	150951516	67306	78202	rs199472952	Neutral	0.406	0.337	0.258
KCNH2	286	G	D	NM_000238.4(KCNH2):c.1877G>A (p.Gly626Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067305	7	150951516	67305	78201	rs199472952	GOF	0.476	0.231	0.293
KCNH2	286	G	S	NM_000238.4(KCNH2):c.1876G>A (p.Gly626Ser)	Congenital long QT syndrome|not provided|Long QT syndrome 2	Pathogenic/Likely pathogenic(Last reviewed: Dec 27, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067304	7	150951517	67304	78200	rs199472953	Neutral	0.42	0.296	0.284
KCNH2	286	G	V	NM_000238.4(KCNH2):c.1877G>T (p.Gly626Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067307	7	150951516	67307	78203	rs199472952	Neutral	0.397	0.341	0.262
KCNH2	287	F	I	NM_000238.4(KCNH2):c.1879T>A (p.Phe627Ile)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067308	7	150951514	67308	78204	rs199472954	Neutral	0.445	0.404	0.151
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1881C>A (p.Phe627Leu)	Long QT syndrome|not provided	Pathogenic(Last reviewed: Apr 22, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200747	7	150951512	200747	197251	rs199473039	Neutral	0.448	0.399	0.152
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1881C>A (p.Phe627Leu)	Long QT syndrome|not provided	Pathogenic(Last reviewed: Apr 22, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200747	7	150951512	200747	197251	rs199473039	Neutral	0.448	0.399	0.152
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1881C>A (p.Phe627Leu)	Long QT syndrome|not provided	Pathogenic(Last reviewed: Apr 22, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200747	7	150951512	200747	197251	rs199473039	Neutral	0.448	0.399	0.152
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1881C>G (p.Phe627Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067310	7	150951512	67310	78206	rs199473039	Neutral	0.448	0.399	0.152
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1881C>G (p.Phe627Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067310	7	150951512	67310	78206	rs199473039	Neutral	0.448	0.399	0.152
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1881C>G (p.Phe627Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067310	7	150951512	67310	78206	rs199473039	Neutral	0.448	0.399	0.152
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1879T>C (p.Phe627Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067309	7	150951514	67309	78205	rs199472954	Neutral	0.448	0.399	0.152
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1879T>C (p.Phe627Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067309	7	150951514	67309	78205	rs199472954	Neutral	0.448	0.399	0.152
KCNH2	287	F	L	NM_000238.4(KCNH2):c.1879T>C (p.Phe627Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067309	7	150951514	67309	78205	rs199472954	Neutral	0.448	0.399	0.152

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	288	G	A	NM_000238.4(KCNH2):c.1883G>C (p.Gly628Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067311	7	150951510	67311	78207	rs199472955	Neutral	0.466	0.354	0.18
KCNH2	288	G	R	NM_000238.4(KCNH2):c.1882G>C (p.Gly628Arg)	not provided	Likely pathogenic(Last reviewed: May 18, 2020)	criteria provided, single submitter	VCV000200749	7	150951511	200749	197250	rs121912507	Neutral	0.557	0.189	0.254
KCNH2	288	G	V	NM_000238.4(KCNH2):c.1883G>T (p.Gly628Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067312	7	150951510	67312	78208	rs199472955	Neutral	0.462	0.347	0.191
KCNH2	288	I	T	NM_000238.4(KCNH2):c.863T>C (p.Ile288Thr)	Long QT syndrome 2	Uncertain significance(Last reviewed: Sep 12, 2018)	criteria provided, single submitter	VCV000598780	7	150958112	598780	589837	rs1563169608	Neutral	0.543	0.32	0.137
KCNH2	289	E	K	NM_000238.4(KCNH2):c.865G>A (p.Glu289Lys)	Long QT syndrome|not specified|not provided| Congenital long QT syndrome|Long QT syndrome 2|Long QT syndrome 2|Short QT syndrome 1|Cardiovascular phenotype	Uncertain significance(Last reviewed: Sep 23, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067535	7	150958110	67535	78431	rs199472880	GOF	0.421	0.256	0.323
KCNH2	289	N	D	NM_000238.4(KCNH2):c.1885A>G (p.Asn629Asp)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Apr 9, 2015)	criteria provided, single submitter	VCV000067313	7	150951508	67313	78209	rs199472956	Neutral	0.427	0.282	0.291
KCNH2	289	N	I	NM_000238.4(KCNH2):c.1886A>T (p.Asn629Ile)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067316	7	150951507	67316	78212	rs199472957	Neutral	0.323	0.429	0.249
KCNH2	289	N	K	NM_000238.4(KCNH2):c.1887C>A (p.Asn629Lys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067317	7	150951506	67317	78213	rs41307295	Neutral	0.393	0.278	0.329
KCNH2	289	N	S	NM_000238.4(KCNH2):c.1886A>G (p.Asn629Ser)	Congenital long QT syndrome|Long QT syndrome 2|Short QT syndrome 1|Long QT syndrome|not provided|Long QT syndrome 2	Pathogenic(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000067315	7	150951507	67315	78211	rs199472957	Neutral	0.358	0.357	0.285
KCNH2	289	N	T	NM_000238.4(KCNH2):c.1886A>C (p.Asn629Thr)	Congenital long QT syndrome|Long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last reviewed: Jul 31, 2016)	criteria provided, multiple submitters, no conflicts	VCV000067314	7	150951507	67314	78210	rs199472957	Neutral	0.329	0.399	0.272
KCNH2	290	A	P	NM_000238.4(KCNH2):c.868G>C (p.Ala290Pro)	not provided	Uncertain significance(Last reviewed: Oct 31, 2016)	criteria provided, single submitter	VCV000200306	7	150958107	200306	197308	rs532891158	Neutral	0.533	0.286	0.181
KCNH2	290	A	S	NM_000238.4(KCNH2):c.868G>T (p.Ala290Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 14, 2016)	criteria provided, single submitter	VCV000359314	7	150958107	359314	310317	rs532891158	Neutral	0.442	0.373	0.186
KCNH2	290	V	A	NM_000238.4(KCNH2):c.1889T>C (p.Val630Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067319	7	150951504	67319	78215	rs199473526	Neutral	0.454	0.396	0.15
KCNH2	290	V	I	NM_000238.4(KCNH2):c.1888G>A (p.Val630Ile)	Long QT syndrome|Long QT syndrome 2	Conflicting interpretations of pathogenicity(Last reviewed: Oct 6, 2020)	criteria provided, conflicting interpretations	VCV000222669	7	150951505	222669	224342	rs199472958	Neutral	0.471	0.373	0.156
KCNH2	290	V	L	NM_000238.4(KCNH2):c.1888G>C (p.Val630Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067318	7	150951505	67318	78214	rs199472958	Neutral	0.458	0.389	0.154
KCNH2	291	M	R	NM_000238.4(KCNH2):c.872T>G (p.Met291Arg)	Long QT syndrome 2|Short QT syndrome 1|Long QT syndrome|not provided	Uncertain significance(Last reviewed: Jul 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000374955	7	150958103	374955	361865	rs199472881	Neutral	0.637	0.185	0.177
KCNH2	291	M	T	NM_000238.4(KCNH2):c.872T>C (p.Met291Thr)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Jul 6, 2018)	criteria provided, single submitter	VCV000067537	7	150958103	67537	78433	rs199472881	Neutral	0.561	0.273	0.166
KCNH2	291	S	A	NM_000238.4(KCNH2):c.1891T>G	Congenital long QT syndrome	not provided	no assertion	VCV000067320	7	150951502	67320	78216	rs199472959	Neutral	0.479	0.338	0.183

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Ser631Ala)			provided										
KCNH2	292	P	A	NM_000238.4(KCNH2):c.1894C>G (p.Pro632Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV001054005	7	150951499	1054005	1044806	NA	Neutral	0.353	0.404	0.243
KCNH2	292	P	S	NM_000238.4(KCNH2):c.1894C>T (p.Pro632Ser)	Congenital long QT syndrome|Cardiovascular phenotype|not provided	Pathogenic/Likely pathogenic(Last reviewed: Sep 17, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067321	7	150951499	67321	78217	rs199473527	Neutral	0.36	0.371	0.269
KCNH2	292	R	H	NM_000238.4(KCNH2):c.875G>A (p.Arg292His)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 7, 2020)	criteria provided, single submitter	VCV001017664	7	150958100	1017664	1007352	rs1355952300	Neutral	0.369	0.384	0.247
KCNH2	293	A	F	NM_000238.4(KCNH2):c.877_878delinsTT (p.Ala293Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 2, 2019)	criteria provided, single submitter	VCV000963108	7	150958097 - 150958098	963108	945527	rs1801438942	Neutral	0.331	0.427	0.242
KCNH2	293	A	S	NM_000238.4(KCNH2):c.877G>T (p.Ala293Ser)	not provided	Uncertain significance(Last reviewed: Apr 1, 2014)	criteria provided, single submitter	VCV000200308	7	150958098	200308	197307	rs794728362	Neutral	0.36	0.342	0.298
KCNH2	293	A	T	NM_000238.4(KCNH2):c.877G>A (p.Ala293Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 27, 2020)	criteria provided, single submitter	VCV000864669	7	150958098	864669	833350	rs794728362	Neutral	0.339	0.373	0.288
KCNH2	293	N	D	NM_000238.4(KCNH2):c.1897A>G (p.Asn633Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067322	7	150951496	67322	78218	rs199472960	GOF	0.4	0.269	0.332
KCNH2	293	N	H	NM_000238.4(KCNH2):c.1897A>C (p.Asn633His)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 27, 2020)	criteria provided, single submitter	VCV001001814	7	150951496	1001814	992195	rs199472960	Neutral	0.333	0.333	0.333
KCNH2	293	N	I	NM_000238.4(KCNH2):c.1898A>T (p.Asn633Ile)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067324	7	150951495	67324	78220	rs199472961	Neutral	0.311	0.402	0.287
KCNH2	293	N	S	NM_000238.4(KCNH2):c.1898A>G (p.Asn633Ser)	Congenital long QT syndrome|Long QT syndrome|Cardiovascular phenotype|not provided|Long QT syndrome 2	Pathogenic/Likely pathogenic(Last reviewed: Jan 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067323	7	150951495	67323	78219	rs199472961	Neutral	0.339	0.318	0.342
KCNH2	294	G	R	NM_000238.4(KCNH2):c.880G>A (p.Gly294Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 19, 2019)	criteria provided, single submitter	VCV000838034	7	150958095	838034	833349	rs1801438642	GOF	0.42	0.202	0.378
KCNH2	294	G	V	NM_000238.4(KCNH2):c.881G>T (p.Gly294Val)	Long QT syndrome|SUDDEN INFANT DEATH SYNDROME	Uncertain significance(Last reviewed: Feb 14, 2019)	criteria provided, single submitter	VCV000067539	7	150958094	67539	78435	rs199473549	Neutral	0.359	0.328	0.313
KCNH2	294	T	A	NM_000238.4(KCNH2):c.1900A>G (p.Thr634Ala)	not provided	Pathogenic(Last reviewed: May 28, 2014)	criteria provided, single submitter	VCV000200391	7	150951493	200391	197249	rs794728377	Neutral	0.315	0.414	0.271
KCNH2	294	T	I	NM_000238.4(KCNH2):c.1901C>T (p.Thr634Ile)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Oct 25, 2020)	criteria provided, single submitter	VCV000067325	7	150951492	67325	78221	rs199472962	Neutral	0.306	0.441	0.253
KCNH2	295	N	D	NM_000238.4(KCNH2):c.1903A>G (p.Asn635Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067326	7	150951490	67326	78222	rs199472963	GOF	0.392	0.262	0.346
KCNH2	295	N	I	NM_000238.4(KCNH2):c.1904A>T (p.Asn635Ile)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Aug 6, 2019)	criteria provided, single submitter	VCV000067327	7	150951489	67327	78223	rs199472964	Neutral	0.309	0.388	0.303

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	295	N	K	NM_000238.4(KCNH2):c.1905C>G (p.Asn635Lys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067328	7	150951488	67328	78224	rs199472965	GOF	0.357	0.251	0.391
KCNH2	295	N	S	NM_000238.4(KCNH2):c.1904A>G (p.Asn635Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 7, 2018)	criteria provided, single submitter	VCV000200393	7	150951489	200393	197248	rs199472964	GOF	0.335	0.298	0.367
KCNH2	296	S	L	NM_000238.4(KCNH2):c.1907C>T (p.Ser636Leu)	not provided	Uncertain significance(Last reviewed: Feb 18, 2020)	criteria provided, single submitter	VCV001303526	7	150951486	1303526	1293803	NA	Neutral	0.329	0.375	0.296
KCNH2	297	E	D	NM_000238.4(KCNH2):c.1911G>C (p.Glu637Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067331	7	150951482	67331	78227	rs199472966	GOF	0.429	0.245	0.326
KCNH2	297	E	G	NM_000238.4(KCNH2):c.1910A>G (p.Glu637Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067330	7	150951483	67330	78226	rs199472967	Neutral	0.403	0.297	0.301
KCNH2	297	E	K	NM_000238.4(KCNH2):c.1909G>A (p.Glu637Lys)	Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Apr 26, 2018)	criteria provided, single submitter	VCV000067329	7	150951484	67329	78225	rs199472968	GOF	0.399	0.234	0.367
KCNH2	297	P	S	NM_000238.4(KCNH2):c.889C>T (p.Pro297Ser)	Long QT syndrome|not provided|Congenital long QT syndrome	Uncertain significance(Last reviewed: Sep 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067540	7	150958086	67540	78436	rs199472882	Neutral	0.347	0.338	0.315
KCNH2	298	K	E	NM_000238.4(KCNH2):c.1912A>G (p.Lys638Glu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067332	7	150951481	67332	78228	rs199473528	GOF	0.395	0.281	0.324
KCNH2	298	K	N	NM_000238.4(KCNH2):c.1914G>T (p.Lys638Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067333	7	150951479	67333	78229	rs199472969	GOF	0.354	0.293	0.352
KCNH2	298	K	R	NM_000238.4(KCNH2):c.1913A>G (p.Lys638Arg)	not provided	Pathogenic(Last reviewed: Jan 10, 2014)	criteria provided, single submitter	VCV000200395	7	150951480	200395	197246	rs794728378	GOF	0.385	0.252	0.363
KCNH2	299	P	L	NM_000238.4(KCNH2):c.896C>T (p.Pro299Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 4, 2018)	criteria provided, single submitter	VCV000641100	7	150958079	641100	635919	rs1584865137	Neutral	0.33	0.397	0.272
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1920C>A (p.Phe640Leu)	Congenital long QT syndrome|Long QT syndrome 2	Pathogenic(Last reviewed: Jan 1, 2013)	criteria provided, single submitter	VCV000067336	7	150951473	67336	78232	rs199472970	Neutral	0.438	0.349	0.213
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1920C>A (p.Phe640Leu)	Congenital long QT syndrome|Long QT syndrome 2	Pathogenic(Last reviewed: Jan 1, 2013)	criteria provided, single submitter	VCV000067336	7	150951473	67336	78232	rs199472970	Neutral	0.438	0.349	0.213
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1920C>A (p.Phe640Leu)	Congenital long QT syndrome|Long QT syndrome 2	Pathogenic(Last reviewed: Jan 1, 2013)	criteria provided, single submitter	VCV000067336	7	150951473	67336	78232	rs199472970	Neutral	0.438	0.349	0.213
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1920C>G (p.Phe640Leu)	not provided	Pathogenic(Last reviewed: Mar 3, 2017)	criteria provided, single submitter	VCV000423753	7	150951473	423753	407059	rs199472970	Neutral	0.438	0.349	0.213
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1920C>G (p.Phe640Leu)	not provided	Pathogenic(Last reviewed: Mar 3, 2017)	criteria provided, single submitter	VCV000423753	7	150951473	423753	407059	rs199472970	Neutral	0.438	0.349	0.213
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1920C>G (p.Phe640Leu)	not provided	Pathogenic(Last reviewed: Mar 3, 2017)	criteria provided, single submitter	VCV000423753	7	150951473	423753	407059	rs199472970	Neutral	0.438	0.349	0.213
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1918T>C (p.Phe640Leu)	Long QT syndrome 2|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Apr 25, 2018)	criteria provided, conflicting interpretations	VCV000200751	7	150951475	200751	197243	rs199473529	Neutral	0.438	0.349	0.213
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1918T>C (p.Phe640Leu)	Long QT syndrome 2|not provided	Conflicting interpretations of pathogenicity(Last	criteria provided, conflicting	VCV000200751	7	150951475	200751	197243	rs199473529	Neutral	0.438	0.349	0.213

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Apr 25, 2018)	interpretations										
KCNH2	300	F	L	NM_000238.4(KCNH2):c.1918T>C (p.Phe640Leu)	Long QT syndrome 2|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Apr 25, 2018)	criteria provided, conflicting interpretations	VCV000200751	7	150951475	200751	197243	rs199473529	Neutral	0.438	0.349	0.213
KCNH2	300	F	V	NM_000238.4(KCNH2):c.1918T>G (p.Phe640Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067334	7	150951475	67334	78230	rs199473529	Neutral	0.425	0.34	0.234
KCNH2	301	R	L	NM_000238.4(KCNH2):c.902G>T (p.Arg301Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067542	7	150958073	67542	78438	rs199472883	Neutral	0.383	0.35	0.268
KCNH2	301	S	F	NM_000238.4(KCNH2):c.1922C>T (p.Ser641Phe)	Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Apr 30, 2019)	criteria provided, single submitter	VCV000067337	7	150951471	67337	78233	rs199472971	Neutral	0.353	0.357	0.29
KCNH2	302	I	M	NM_000238.4(KCNH2):c.1926C>G (p.Ile642Met)	Long QT syndrome 2	not provided	no assertion provided	VCV000973054	7	150951467	973054	961283	rs1801154313	Neutral	0.399	0.425	0.176
KCNH2	303	A	G	NM_000238.4(KCNH2):c.908C>G (p.Ala303Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 14, 2019)	criteria provided, single submitter	VCV000957218	7	150958067	957218	945526	rs1165306519	Neutral	0.464	0.346	0.19
KCNH2	304	V	F	NM_000238.4(KCNH2):c.1930G>T (p.Val644Phe)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV000067340	7	150951463	67340	78236	rs199472972	Neutral	0.434	0.394	0.172
KCNH2	304	V	I	NM_000238.4(KCNH2):c.1930G>A (p.Val644Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV001009718	7	150951463	1009718	992194	rs199472972	Neutral	0.471	0.346	0.183
KCNH2	304	V	L	NM_000238.4(KCNH2):c.1930G>C (p.Val644Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067339	7	150951463	67339	78235	rs199472972	Neutral	0.456	0.363	0.181
KCNH2	305	M	I	NM_000238.4(KCNH2):c.1935G>A (p.Met645Ile)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Oct 11, 2018)	criteria provided, single submitter	VCV000067343	7	150951458	67343	78239	rs199472973	Neutral	0.429	0.411	0.161
KCNH2	305	M	L	NM_000238.4(KCNH2):c.1933A>T (p.Met645Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067342	7	150951460	67342	78238	rs199472974	Neutral	0.44	0.396	0.164
KCNH2	305	M	L	NM_000238.4(KCNH2):c.1933A>T (p.Met645Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067342	7	150951460	67342	78238	rs199472974	Neutral	0.44	0.396	0.164
KCNH2	305	M	L	NM_000238.4(KCNH2):c.1933A>C (p.Met645Leu)	Long QT syndrome 2	Pathogenic(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV000869431	7	150951460	869431	857647	rs199472974	Neutral	0.44	0.396	0.164
KCNH2	305	M	L	NM_000238.4(KCNH2):c.1933A>C (p.Met645Leu)	Long QT syndrome 2	Pathogenic(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV000869431	7	150951460	869431	857647	rs199472974	Neutral	0.44	0.396	0.164
KCNH2	305	M	V	NM_000238.4(KCNH2):c.1933A>G (p.Met645Val)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Nov 22, 2013)	criteria provided, single submitter	VCV000067341	7	150951460	67341	78237	rs199472974	Neutral	0.427	0.41	0.163
KCNH2	306	G	R	NM_000238.4(KCNH2):c.916G>C (p.Gly306Arg)	Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 2	Likely pathogenic(Last reviewed: Nov 10, 2017)	criteria provided, single submitter	VCV000067543	7	150958059	67543	78439	rs199472884	GOF	0.517	0.168	0.315
KCNH2	306	G	W	NM_000238.4(KCNH2):c.916G>T (p.Gly306Trp)	Long QT syndrome|not provided|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Dec 22, 2017)	criteria provided, multiple submitters, no conflicts	VCV000067544	7	150958059	67544	78440	rs199472884	Neutral	0.5	0.267	0.233
KCNH2	308	G	D	NM_000238.4(KCNH2):c.1943G>A (p.Gly648Asp)	not provided	Pathogenic(Last reviewed: Sep 9, 2011)	criteria provided, single submitter	VCV000200753	7	150951450	200753	197242	rs794728486	Neutral	0.486	0.283	0.231

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	308	G	S	NM_000238.4(KCNH2):c.1942G>A (p.Gly648Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067344	7	150951451	67344	78240	rs199472975	Neutral	0.409	0.361	0.229
KCNH2	308	M	R	NM_000238.4(KCNH2):c.923T>G (p.Met308Arg)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Nov 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000923222	7	150957496	923222	910793	rs372559632	Neutral	0.513	0.28	0.207
KCNH2	309	H	Q	NM_000238.4(KCNH2):c.927C>G (p.His309Gln)	not provided	Uncertain significance(Last reviewed: May 1, 2018)	criteria provided, single submitter	VCV000810210	7	150957492	810210	795989	rs775056804	Neutral	0.433	0.326	0.241
KCNH2	309	H	Y	NM_000238.4(KCNH2):c.925C>T (p.His309Tyr)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 13, 2017)	criteria provided, single submitter	VCV000526915	7	150957494	526915	522691	rs1241849013	Neutral	0.372	0.409	0.219
KCNH2	309	S	F	NM_000238.4(KCNH2):c.1946C>T (p.Ser649Phe)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: May 31, 2019)	criteria provided, single submitter	VCV000067346	7	150951120	67346	78242	rs199472976	Neutral	0.354	0.454	0.191
KCNH2	309	S	P	NM_000238.4(KCNH2):c.1945T>C (p.Ser649Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067345	7	150951448	67345	78241	rs199473530	Neutral	0.466	0.322	0.211
KCNH2	310	P	L	NM_000238.4(KCNH2):c.929C>T (p.Pro310Leu)	Arrhythmia	Uncertain significance(Last reviewed: Jan 25, 2021)	criteria provided, single submitter	VCV001332223	7	150957490	1332223	1322942	NA	Neutral	0.389	0.422	0.189
KCNH2	310	P	T	NM_000238.4(KCNH2):c.928C>A (p.Pro310Thr)	Arrhythmia	Uncertain significance(Last reviewed: Dec 10, 2018)	criteria provided, single submitter	VCV000925941	7	150957491	925941	910792	rs1801415549	Neutral	0.39	0.418	0.192
KCNH2	311	M	R	NM_000238.4(KCNH2):c.1952T>G (p.Met651Arg)	Long QT syndrome 2	Likely pathogenic(Last reviewed: Feb 9, 2017)	criteria provided, single submitter	VCV000427945	7	150951114	427945	419275	rs1554425569	Neutral	0.513	0.28	0.206
KCNH2	312	R	C	NM_000238.4(KCNH2):c.934C>T (p.Arg312Cys)	Arrhythmia|Long QT syndrome 2|Short QT syndrome 1|Long QT syndrome|Congenital long QT syndrome|not specified	Conflicting interpretations of pathogenicity(Last reviewed: Oct 14, 2020)	criteria provided, conflicting interpretations	VCV000067547	7	150957485	67547	78443	rs199472885	Neutral	0.381	0.458	0.162
KCNH2	312	R	H	NM_000238.4(KCNH2):c.935G>A (p.Arg312His)	Long QT syndrome|Arrhythmia|not provided	Uncertain significance(Last reviewed: Aug 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000456942	7	150957484	456942	457557	rs747313232	Neutral	0.368	0.469	0.163
KCNH2	312	Y	H	NM_000238.4(KCNH2):c.1954_1956delinsCAC (p.Tyr652His)	not provided	Uncertain significance(Last reviewed: Sep 24, 2021)	criteria provided, single submitter	VCV000200761	7	150951110 - 150951112	200761	197237	rs794728490	Neutral	0.411	0.442	0.148
KCNH2	314	G	D	NM_000238.4(KCNH2):c.941G>A (p.Gly314Asp)	Arrhythmia	Uncertain significance(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV000925690	7	150957478	925690	910789	rs1801414464	Neutral	0.586	0.195	0.219
KCNH2	314	G	S	NM_000238.4(KCNH2):c.940G>A (p.Gly314Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067548	7	150957479	67548	78444	rs199473504	Neutral	0.53	0.276	0.194
KCNH2	315	I	V	NM_000238.4(KCNH2):c.1963A>G (p.Ile655Val)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 30, 2016)	criteria provided, single submitter	VCV000405359	7	150951103	405359	395694	rs1060500673	Neutral	0.486	0.384	0.13
KCNH2	316	F	C	NM_000238.4(KCNH2):c.1967T>G (p.Phe656Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067347	7	150951099	67347	78243	rs199472977	Neutral	0.493	0.368	0.14
KCNH2	317	G	C	NM_000238.4(KCNH2):c.1969G>T	Congenital long QT syndrome	not provided	no assertion	VCV000067350	7	150951097	67350	78246	rs199472978	Neutral	0.467	0.32	0.213

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Gly657Cys)			provided										
KCNH2	317	G	R	NM_000238.4(KCNH2):c.1969G>C (p.Gly657Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067349	7	150951097	67349	78245	rs199472978	GOF	0.513	0.163	0.324
KCNH2	317	G	S	NM_000238.4(KCNH2):c.1969G>A (p.Gly657Ser)	Congenital long QT syndrome|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jun 7, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067348	7	150951097	67348	78244	rs199472978	Neutral	0.498	0.267	0.235
KCNH2	317	G	V	NM_000238.4(KCNH2):c.1970G>T (p.Gly657Val)	not provided	Likely pathogenic(Last reviewed: Jun 26, 2015)	criteria provided, single submitter	VCV000427076	7	150951096	427076	415087	rs1085307943	Neutral	0.471	0.308	0.22
KCNH2	317	N	S	NM_000238.4(KCNH2):c.950A>G (p.Asn317Ser)	Long QT syndrome 2|Short QT syndrome 1| Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Apr 30, 2019)	criteria provided, multiple submitters, no conflicts	VCV000405346	7	150957469	405346	395490	rs779027664	Neutral	0.461	0.312	0.227
KCNH2	318	N	S	NM_000238.4(KCNH2):c.1973A>G (p.Asn658Ser)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Dec 20, 2016)	criteria provided, conflicting interpretations	VCV000389141	7	150951093	389141	369597	rs1057523338	Neutral	0.522	0.288	0.19
KCNH2	320	S	L	NM_000238.4(KCNH2):c.959C>T (p.Ser320Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067550	7	150957460	67550	78446	rs199472886	Neutral	0.524	0.322	0.154
KCNH2	320	S	L	NM_000238.4(KCNH2):c.959C>T (p.Ser320Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067550	7	150957460	67550	78446	rs199472886	Neutral	0.524	0.322	0.154
KCNH2	320	S	L	NM_000238.4(KCNH2):c.1979C>T (p.Ser660Leu)	Long QT syndrome 2|Congenital long QT syndrome|Cardiovascular phenotype|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 30, 2020)	criteria provided, conflicting interpretations	VCV000067352	7	150951087	67352	78248	rs199472979	Neutral	0.524	0.322	0.154
KCNH2	320	S	L	NM_000238.4(KCNH2):c.1979C>T (p.Ser660Leu)	Long QT syndrome 2|Congenital long QT syndrome|Cardiovascular phenotype|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 30, 2020)	criteria provided, conflicting interpretations	VCV000067352	7	150951087	67352	78248	rs199472979	Neutral	0.524	0.322	0.154
KCNH2	320	S	W	NM_000238.4(KCNH2):c.959C>G (p.Ser320Trp)	Arrhythmia|not provided	Uncertain significance(Last reviewed: Feb 8, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200312	7	150957460	200312	197304	rs199472886	Neutral	0.532	0.288	0.18
KCNH2	321	A	V	NM_000238.4(KCNH2):c.1982C>T (p.Ala661Val)	Sudden cardiac arrest	Uncertain significance(Last reviewed: Apr 7, 2020)	no assertion criteria provided	VCV000977179	7	150951084	977179	965303	rs768519164	Neutral	0.479	0.365	0.156
KCNH2	321	D	Y	NM_000238.4(KCNH2):c.961G>T (p.Asp321Tyr)	Long QT syndrome 2|not provided	Uncertain significance(Last reviewed: Mar 4, 2019)	criteria provided, multiple submitters, no conflicts	VCV000359312	7	150957458	359312	305394	rs886062089	Neutral	0.475	0.295	0.229
KCNH2	322	I	T	NM_000238.4(KCNH2):c.1985T>C (p.Ile662Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067353	7	150951081	67353	78249	rs199472980	Neutral	0.559	0.308	0.133
KCNH2	323	D	N	NM_000238.4(KCNH2):c.967G>A (p.Asp323Asn)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV000067551	7	150957452	67551	78447	rs199472887	Neutral	0.565	0.232	0.204
KCNH2	325	V	L	NM_000238.4(KCNH2):c.973G>C (p.Val325Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 28, 2020)	criteria provided, single submitter	VCV000856033	7	150957446	856033	833348	rs149381387	Neutral	0.509	0.381	0.11

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	325	V	M	NM_000238.4(KCNH2):c.973G>A (p.Val325Met)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 8, 2017)	criteria provided, single submitter	VCV000526931	7	150957446	526931	522688	rs149381387	Neutral	0.485	0.415	0.1
KCNH2	326	R	C	NM_000238.4(KCNH2):c.976C>T (p.Arg326Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 29, 2020)	criteria provided, single submitter	VCV001040723	7	150957443	1040723	1027895	rs759079349	Neutral	0.428	0.428	0.144
KCNH2	326	R	H	NM_000238.4(KCNH2):c.977G>A (p.Arg326His)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 10, 2018)	criteria provided, single submitter	VCV000649300	7	150957442	649300	635918	rs1187921286	Neutral	0.436	0.43	0.134
KCNH2	327	Y	C	NM_000238.4(KCNH2):c.980A>G (p.Tyr327Cys)	Cardiovascular phenotype	Uncertain significance(Last reviewed: May 8, 2015)	criteria provided, single submitter	VCV000264025	7	150957439	264025	258467	rs886039013	Neutral	0.482	0.395	0.123
KCNH2	327	Y	H	NM_000238.4(KCNH2):c.979T>C (p.Tyr327His)	Arrhythmia	Uncertain significance(Last reviewed: Jul 15, 2021)	criteria provided, single submitter	VCV000629260	7	150957440	629260	617352	rs776431167	Neutral	0.514	0.371	0.115
KCNH2	328	R	C	NM_000238.4(KCNH2):c.982C>T (p.Arg328Cys)	Long QT syndrome|Arrhythmia|Cardiovascular phenotype|not specified|not provided|Long QT syndrome 2	Conflicting interpretations of pathogenicity(Last reviewed: Nov 21, 2020)	criteria provided, conflicting interpretations	VCV000067552	7	150957437	67552	78448	rs199473505	Neutral	0.466	0.401	0.134
KCNH2	328	R	H	NM_000238.4(KCNH2):c.983G>A (p.Arg328His)	Arrhythmia|Long QT syndrome|not provided	Uncertain significance(Last reviewed: Jul 21, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200315	7	150957436	200315	197303	rs747437736	Neutral	0.495	0.383	0.122
KCNH2	329	G	A	NM_000238.4(KCNH2):c.2006G>C (p.Gly669Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 7, 2018)	criteria provided, single submitter	VCV000647535	7	150951060	647535	635901	rs1584852286	Neutral	0.576	0.302	0.123
KCNH2	329	T	A	NM_000238.4(KCNH2):c.985A>G (p.Thr329Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 6, 2018)	criteria provided, single submitter	VCV000640482	7	150957434	640482	635917	rs946110595	Neutral	0.507	0.401	0.091
KCNH2	331	A	G	NM_000238.4(KCNH2):c.2012C>G (p.Ala671Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, single submitter	VCV001024776	7	150951054	1024776	1007345	rs779970711	Neutral	0.636	0.245	0.119
KCNH2	332	K	R	NM_000238.4(KCNH2):c.995A>G (p.Lys332Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 26, 2019)	criteria provided, single submitter	VCV000847270	7	150957424	847270	833347	rs1801410888	Neutral	0.529	0.211	0.26
KCNH2	332	R	C	NM_000238.4(KCNH2):c.2014C>T (p.Arg672Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 21, 2020)	criteria provided, single submitter	VCV001020103	7	150951052	1020103	1007344	rs769197005	Neutral	0.466	0.342	0.192
KCNH2	334	P	L	NM_000238.4(KCNH2):c.1001C>T (p.Pro334Leu)	Congenital long QT syndrome|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jan 29, 2019)	criteria provided, conflicting interpretations	VCV000067161	7	150957418	67161	78057	rs199472888	Neutral	0.603	0.274	0.123
KCNH2	337	M	T	NM_000238.4(KCNH2):c.2030T>C (p.Met677Thr)	Long QT syndrome	Uncertain significance(Last reviewed: May 27, 2019)	criteria provided, single submitter	VCV000939911	7	150951036	939911	933769	rs756870695	Neutral	0.575	0.318	0.107
KCNH2	337	T	A	NM_000238.4(KCNH2):c.1009A>G (p.Thr337Ala)	Arrhythmia	Uncertain significance(Last reviewed: Feb 6, 2020)	criteria provided, single submitter	VCV000927557	7	150957410	927557	910786	rs1584863947	Neutral	0.528	0.379	0.093

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	337	T	S	NM_000238.4(KCNH2):c.1010C>G (p.Thr337Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV001043131	7	150957409	1043131	1027894	rs41311018	Neutral	0.563	0.338	0.099
KCNH2	338	L	P	NM_000238.4(KCNH2):c.2033T>C (p.Leu678Pro)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Apr 30, 2016)	criteria provided, single submitter	VCV000067356	7	150951033	67356	78252	rs199472981	Neutral	0.725	0.174	0.1
KCNH2	338	L	R	NM_000238.4(KCNH2):c.2033T>G (p.Leu678Arg)	not provided	Uncertain significance(Last reviewed: Oct 5, 2021)	criteria provided, single submitter	VCV001330843	7	150951033	1330843	1321520	NA	LOF	0.724	0.145	0.131
KCNH2	339	N	K	NM_000238.4(KCNH2):c.1017C>G (p.Asn339Lys)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 7, 2019)	criteria provided, single submitter	VCV000942831	7	150957402	942831	933777	rs1801410026	Neutral	0.622	0.203	0.174
KCNH2	339	R	Q	NM_000238.4(KCNH2):c.2036G>A (p.Arg679Gln)	Arrhythmia	Uncertain significance(Last reviewed: Jan 1, 2020)	criteria provided, single submitter	VCV000922598	7	150951030	922598	910747	rs567628192	Neutral	0.587	0.289	0.124
KCNH2	340	F	L	NM_000238.4(KCNH2):c.1018T>C (p.Phe340Leu)	Arrhythmia	Uncertain significance(Last reviewed: Jun 14, 2021)	criteria provided, single submitter	VCV001332429	7	150957401	1332429	1323147	NA	Neutral	0.615	0.268	0.116
KCNH2	341	R	Q	NM_000238.4(KCNH2):c.2042G>A (p.Arg681Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 15, 2019)	criteria provided, single submitter	VCV000957302	7	150951024	957302	945522	rs1801131066	Neutral	0.571	0.305	0.124
KCNH2	342	D	V	NM_000238.4(KCNH2):c.1025A>T (p.Asp342Val)	Acquired long QT syndrome	not provided	no assertion provided	VCV000067162	7	150957394	67162	78058	rs199472889	Neutral	0.513	0.326	0.161
KCNH2	342	E	D	NM_000238.4(KCNH2):c.2046G>C (p.Glu682Asp)	Wolff-Parkinson-White pattern|Arrhythmia|not provided	Uncertain significance(Last reviewed: Nov 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000487626	7	150951020	487626	480695	rs1338579153	Neutral	0.62	0.185	0.194
KCNH2	345	R	C	NM_000238.4(KCNH2):c.2053C>T (p.Arg685Cys)	Long QT syndrome 2|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Nov 25, 2019)	criteria provided, conflicting interpretations	VCV000448974	7	150951013	448974	442551	rs778135438	Neutral	0.417	0.396	0.187
KCNH2	345	R	H	NM_000238.4(KCNH2):c.2054G>A (p.Arg685His)	Long QT syndrome 2	Uncertain significance	criteria provided, single submitter	VCV000979129	7	150951012	979129	967185	rs758751607	Neutral	0.432	0.371	0.196
KCNH2	346	D	N	NM_000238.4(KCNH2):c.1036G>A (p.Asp346Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 22, 2019)	criteria provided, single submitter	VCV000841013	7	150957383	841013	833346	rs1420621400	Neutral	0.569	0.215	0.216
KCNH2	347	H	Y	NM_000238.4(KCNH2):c.2059C>T (p.His687Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067357	7	150951007	67357	78253	rs199472982	Neutral	0.42	0.415	0.165
KCNH2	347	P	S	NM_000238.4(KCNH2):c.1039C>T (p.Pro347Ser)	Cardiovascular phenotype|not specified|Long QT syndrome 2|Arrhythmia|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Dec 7, 2020)	criteria provided, conflicting interpretations	VCV000067163	7	150957380	67163	78059	rs138776684	Neutral	0.45	0.398	0.152
KCNH2	348	F	Y	NM_000238.4(KCNH2):c.1043T>A (p.Phe348Tyr)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Sep 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200319	7	150957376	200319	197301	rs794728363	Neutral	0.729	0.15	0.122
KCNH2	351	S	L	NM_000238.4(KCNH2):c.1052C>T (p.Ser351Leu)	Long QT syndrome|Long QT syndrome 2	Conflicting interpretations of	criteria provided,	VCV000864050	7	150957367	864050	833345	rs759134380	Neutral	0.472	0.364	0.165

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						pathogenicity(Last reviewed: Dec 30, 2019)	conflicting interpretations										
KCNH2	352	P	R	NM_000238.4(KCNH2):c.1055C>G (p.Pro352Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 19, 2018)	criteria provided, single submitter	VCV001038936	7	150957364	1038936	1027893	rs1801408390	Neutral	0.517	0.202	0.281
KCNH2	352	P	S	NM_000238.4(KCNH2):c.1054C>T (p.Pro352Ser)	Arrhythmia	Uncertain significance(Last reviewed: Nov 16, 2018)	criteria provided, single submitter	VCV000918184	7	150957365	918184	910784	rs1801408471	Neutral	0.493	0.303	0.204
KCNH2	353	L	P	NM_000238.4(KCNH2):c.2078T>C (p.Leu693Pro)	Congenital long QT syndrome|Inborn genetic diseases|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 6, 2017)	criteria provided, conflicting interpretations	VCV000067359	7	150950988	67359	78255	rs199472983	Neutral	0.582	0.26	0.158
KCNH2	353	T	I	NM_000238.4(KCNH2):c.1058C>T (p.Thr353Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 28, 2020)	criteria provided, single submitter	VCV001047609	7	150957361	1047609	1027892	rs1801408187	Neutral	0.381	0.47	0.148
KCNH2	354	R	C	NM_000238.4(KCNH2):c.2080C>T (p.Arg694Cys)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jun 9, 2019)	criteria provided, multiple submitters, no conflicts	VCV000651990	7	150950986	651990	635900	rs765717858	Neutral	0.436	0.405	0.158
KCNH2	354	R	H	NM_000238.4(KCNH2):c.2081G>A (p.Arg694His)	not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Mar 14, 2020)	criteria provided, conflicting interpretations	VCV000200410	7	150950985	200410	197232	rs794728383	Neutral	0.462	0.379	0.16
KCNH2	356	R	C	NM_000238.4(KCNH2):c.1066C>T (p.Arg356Cys)	Arrhythmia	Uncertain significance(Last reviewed: Dec 3, 2020)	criteria provided, single submitter	VCV000925269	7	150957353	925269	910783	rs772689518	Neutral	0.587	0.245	0.168
KCNH2	356	R	C	NM_000238.4(KCNH2):c.1066C>T (p.Arg356Cys)	Arrhythmia	Uncertain significance(Last reviewed: Dec 3, 2020)	criteria provided, single submitter	VCV000925269	7	150957353	925269	910783	rs772689518	Neutral	0.587	0.245	0.168
KCNH2	356	R	C	NM_000238.4(KCNH2):c.2086C>T (p.Arg696Cys)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Nov 28, 2018)	criteria provided, single submitter	VCV000067360	7	150950980	67360	78256	rs199472984	Neutral	0.587	0.245	0.168
KCNH2	356	R	C	NM_000238.4(KCNH2):c.2086C>T (p.Arg696Cys)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Nov 28, 2018)	criteria provided, single submitter	VCV000067360	7	150950980	67360	78256	rs199472984	Neutral	0.587	0.245	0.168
KCNH2	356	R	H	NM_000238.4(KCNH2):c.1067G>A (p.Arg356His)	Cardiovascular phenotype|Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Mar 22, 2021)	criteria provided, multiple submitters, no conflicts	VCV000180382	7	150957352	180382	178575	rs730880118	Neutral	0.641	0.199	0.161
KCNH2	356	R	P	NM_000238.4(KCNH2):c.2087G>C (p.Arg696Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067361	7	150950979	67361	78257	rs199473531	Neutral	0.653	0.185	0.162
KCNH2	357	E	D	NM_000238.4(KCNH2):c.1071G>T (p.Glu357Asp)	Arrhythmia	Uncertain significance(Last reviewed: Aug 8, 2019)	criteria provided, single submitter	VCV000628850	7	150957348	628850	617350	rs762016259	LOF	0.674	0.14	0.186
KCNH2	358	E	D	NM_000238.4(KCNH2):c.2094G>T (p.Glu698Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 27, 2020)	criteria provided, single submitter	VCV001001384	7	150950972	1001384	992193	rs1801127916	LOF	0.76	0.122	0.118

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	358	I	N	NM_000238.4(KCNH2):c.1073T>A (p.Ile358Asn)	Arrhythmia	Uncertain significance(Last reviewed: Jun 8, 2020)	criteria provided, single submitter	VCV001172314	7	150957346	1172314	1160702	NA	LOF	0.803	0.109	0.088
KCNH2	359	E	D	NM_000238.4(KCNH2):c.2097G>T (p.Glu699Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 24, 2020)	criteria provided, single submitter	VCV001001725	7	150950969	1001725	992192	rs550680502	LOF	0.759	0.107	0.135
KCNH2	359	I	K	NM_000238.4(KCNH2):c.1076T>A (p.Ile359Lys)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 19, 2017)	criteria provided, single submitter	VCV000456877	7	150957343	456877	456861	rs1554427331	LOF	0.807	0.079	0.114
KCNH2	361	P	S	NM_000238.4(KCNH2):c.1081C>T (p.Pro361Ser)	Long QT syndrome	Uncertain significance(Last reviewed: May 18, 2017)	criteria provided, single submitter	VCV000456878	7	150957338	456878	457555	rs1293901043	Neutral	0.661	0.213	0.127
KCNH2	366	R	Q	NM_000238.4(KCNH2):c.1097G>A (p.Arg366Gln)	not provided|Arrhythmia	Uncertain significance(Last reviewed: Jun 18, 2021)	criteria provided, multiple submitters, no conflicts	VCV000918732	7	150957322	918732	910782	rs1372751640	LOF	0.718	0.139	0.142
KCNH2	366	S	C	NM_000238.4(KCNH2):c.2117C>G (p.Ser706Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067364	7	150950949	67364	78260	rs199472985	Neutral	0.634	0.242	0.123
KCNH2	366	S	F	NM_000238.4(KCNH2):c.2117C>T (p.Ser706Phe)	Acquired long QT syndrome	not provided	no assertion provided	VCV000067365	7	150950949	67365	78261	rs199472985	Neutral	0.633	0.224	0.143
KCNH2	367	T	A	NM_000238.4(KCNH2):c.1099A>G (p.Thr367Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV001047434	7	150957320	1047434	1027891	rs199472890	LOF	0.78	0.132	0.088
KCNH2	367	T	S	NM_000238.4(KCNH2):c.1099A>T (p.Thr367Ser)	not provided|Arrhythmia	Uncertain significance(Last reviewed: Aug 18, 2020)	criteria provided, single submitter	VCV000067164	7	150957320	67164	78060	rs199472890	LOF	0.788	0.118	0.094
KCNH2	368	T	A	NM_000238.4(KCNH2):c.2122A>G (p.Thr708Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 24, 2017)	criteria provided, single submitter	VCV000527003	7	150950944	527003	522687	rs1554425463	Neutral	0.723	0.165	0.113
KCNH2	371	I	V	NM_000238.4(KCNH2):c.2131A>G (p.Ile711Val)	Long QT syndrome|not provided|Arrhythmia| Congenital long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jun 7, 2021)	criteria provided, conflicting interpretations	VCV000067367	7	150950935	67367	78263	rs199473532	LOF	0.81	0.105	0.085
KCNH2	375	A	V	NM_000238.4(KCNH2):c.2144C>T (p.Ala715Val)	Arrhythmia|not specified|Long QT syndrome| Cardiovascular phenotype	Uncertain significance(Last reviewed: Jul 22, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200413	7	150950922	200413	197228	rs780656919	LOF	0.841	0.081	0.077
KCNH2	376	Q	E	NM_000238.4(KCNH2):c.1126C>G (p.Gln376Glu)	Long QT syndrome 2	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000909783	7	150957293	909783	897674	rs1801405757	LOF	0.824	0.078	0.097
KCNH2	381	P	L	NM_000238.4(KCNH2):c.2162C>T (p.Pro721Leu)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Feb 6, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067371	7	150950404	67371	78267	rs199472986	LOF	0.737	0.148	0.115
KCNH2	383	C	G	NM_000238.4(KCNH2):c.2167T>G (p.Cys723Gly)	Arrhythmia	Uncertain significance(Last reviewed: Mar 4, 2020)	criteria provided, single submitter	VCV000922895	7	150950399	922895	910745	rs199472987	LOF	0.794	0.081	0.125
KCNH2	383	C	R	NM_000238.4(KCNH2):c.2167T>C	not provided	not provided	no assertion	VCV000067372	7	150950399	67372	78268	rs199472987	LOF	0.775	0.06	0.165

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Cys723Arg)			provided										
KCNH2	383	C	Y	NM_000238.4(KCNH2):c.2168G>A (p.Cys723Tyr)	not provided	Uncertain significance(Last reviewed: Mar 3, 2016)	criteria provided, single submitter	VCV000200763	7	150950398	200763	197226	rs794728491	LOF	0.758	0.081	0.161
KCNH2	384	L	P	NM_000238.4(KCNH2):c.2171T>C (p.Leu724Pro)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Sep 19, 2019)	criteria provided, multiple submitters, no conflicts	VCV000527007	7	150950395	527007	523130	rs1554425307	LOF	0.807	0.111	0.082
KCNH2	388	I	F	NM_000238.4(KCNH2):c.2182A>T (p.Ile728Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067373	7	150950384	67373	78269	rs199473533	LOF	0.831	0.096	0.074
KCNH2	391	H	P	NM_000238.4(KCNH2):c.2192A>C (p.His731Pro)	not provided	Likely pathogenic(Last reviewed: Jan 31, 2014)	criteria provided, single submitter	VCV000200417	7	150950374	200417	197225	rs794728385	LOF	0.729	0.115	0.156
KCNH2	393	P	L	NM_000238.4(KCNH2):c.1178C>T (p.Pro393Leu)	Long QT syndrome	Uncertain significance(Last reviewed: May 19, 2019)	criteria provided, single submitter	VCV000933401	7	150952804	933401	933776	rs769814960	LOF	0.721	0.149	0.129
KCNH2	394	R	C	NM_000238.4(KCNH2):c.2200C>T (p.Arg734Cys)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: May 21, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200765	7	150950366	200765	197224	rs143072395	Neutral	0.685	0.168	0.148
KCNH2	394	R	G	NM_000238.4(KCNH2):c.2200C>G (p.Arg734Gly)	Arrhythmia	Uncertain significance(Last reviewed: Apr 15, 2021)	criteria provided, single submitter	VCV001332169	7	150950366	1332169	1322888	NA	LOF	0.746	0.111	0.144
KCNH2	395	S	A	NM_000238.4(KCNH2):c.2203T>G (p.Ser735Ala)	Arrhythmia	Uncertain significance(Last reviewed: Aug 5, 2021)	criteria provided, single submitter	VCV001332619	7	150950363	1332619	1323337	NA	LOF	0.755	0.145	0.1
KCNH2	395	S	L	NM_000238.4(KCNH2):c.2204C>T (p.Ser735Leu)	Long QT syndrome|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Mar 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067374	7	150950362	67374	78270	rs199472988	Neutral	0.728	0.162	0.11
KCNH2	396	H	Y	NM_000238.4(KCNH2):c.1186C>T (p.His396Tyr)	Arrhythmia	Uncertain significance(Last reviewed: Mar 16, 2021)	criteria provided, single submitter	VCV001332414	7	150952796	1332414	1323132	NA	LOF	0.785	0.096	0.119
KCNH2	399	H	R	NM_000238.4(KCNH2):c.2216A>G (p.His739Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 1, 2015)	criteria provided, single submitter	VCV000222670	7	150950350	222670	224341	rs869025446	LOF	0.878	0.045	0.077
KCNH2	400	I	N	NM_000238.4(KCNH2):c.1199T>A (p.Ile400Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067165	7	150952783	67165	78061	rs199472891	LOF	0.885	0.044	0.071
KCNH2	401	K	Q	NM_000238.4(KCNH2):c.2221A>C (p.Lys741Gln)	Arrhythmia	Uncertain significance(Last reviewed: Jun 11, 2019)	criteria provided, single submitter	VCV000923298	7	150950345	923298	910743	rs1801092981	LOF	0.876	0.035	0.089
KCNH2	401	K	R	NM_000238.4(KCNH2):c.2222A>G (p.Lys741Arg)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Dec 23, 2021)	criteria provided, multiple submitters, no conflicts	VCV000566154	7	150950344	566154	561718	rs538224282	LOF	0.856	0.03	0.113
KCNH2	402	H	R	NM_000238.4(KCNH2):c.1205A>G (p.His402Arg)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Mar 3, 2017)	criteria provided, single submitter	VCV000067166	7	150952777	67166	78062	rs199473506	LOF	0.88	0.015	0.105

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	404	R	P	NM_000238.4(KCNH2):c.2231G>C (p.Arg744Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 22, 2020)	criteria provided, single submitter	VCV001017667	7	150950335	1017667	1007343	rs1264176305	LOF	0.877	0.041	0.082
KCNH2	404	R	Q	NM_000238.4(KCNH2):c.2231G>A (p.Arg744Gln)	Arrhythmia	Uncertain significance(Last reviewed: Dec 10, 2020)	criteria provided, single submitter	VCV000924279	7	150950335	924279	910742	rs1264176305	LOF	0.87	0.037	0.093
KCNH2	406	A	S	NM_000238.4(KCNH2):c.2236G>T (p.Ala746Ser)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000222671	7	150950330	222671	224340	rs751681463	LOF	0.905	0.022	0.073
KCNH2	409	G	C	NM_000238.4(KCNH2):c.2245G>T (p.Gly749Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 12, 2017)	criteria provided, single submitter	VCV000405350	7	150950321	405350	396142	rs1060500668	LOF	0.9	0.023	0.077
KCNH2	409	G	V	NM_000238.4(KCNH2):c.2246G>T (p.Gly749Val)	Congenital long QT syndrome|not specified|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Mar 31, 2016)	criteria provided, conflicting interpretations	VCV000067378	7	150950320	67378	78274	rs199472989	LOF	0.885	0.028	0.087
KCNH2	409	V	E	NM_000238.4(KCNH2):c.1226T>A (p.Val409Glu)	Cardiovascular phenotype|not provided	Uncertain significance(Last reviewed: Jan 16, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200329	7	150952756	200329	197289	rs794728367	LOF	0.897	0.021	0.082
KCNH2	410	W	S	NM_000238.4(KCNH2):c.1229G>C (p.Trp410Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067168	7	150952753	67168	78064	rs199472892	LOF	0.924	0.017	0.059
KCNH2	412	R	W	NM_000238.4(KCNH2):c.2254C>T (p.Arg752Trp)	Long QT syndrome|Cardiovascular phenotype|Congenital long QT syndrome|not provided|Arrhythmia	Pathogenic/Likely pathogenic(Last reviewed: Mar 17, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067379	7	150950312	67379	78275	rs199472990	LOF	0.89	0.016	0.094
KCNH2	413	A	S	NM_000238.4(KCNH2):c.2257G>T (p.Ala753Ser)	Congenital long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jan 17, 2020)	criteria provided, single submitter	VCV000067380	7	150950309	67380	78276	rs199472991	LOF	0.882	0.029	0.089
KCNH2	413	L	P	NM_000238.4(KCNH2):c.1238T>C (p.Leu413Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067170	7	150952744	67170	78066	rs199472893	LOF	0.895	0.024	0.081
KCNH2	413	L	R	NM_000238.4(KCNH2):c.1238T>G (p.Leu413Arg)	Long QT syndrome 2	Likely pathogenic(Last reviewed: Aug 30, 2016)	criteria provided, single submitter	VCV000279588	7	150952744	279588	263859	rs199472893	LOF	0.885	0.021	0.094
KCNH2	414	I	V	NM_000238.4(KCNH2):c.1240A>G (p.Ile414Val)	Arrhythmia	Uncertain significance(Last reviewed: Mar 7, 2019)	criteria provided, single submitter	VCV000924916	7	150952742	924916	910776	rs1801229604	LOF	0.805	0.064	0.131
KCNH2	414	L	R	NM_000238.4(KCNH2):c.2261T>G (p.Leu754Arg)	not provided	Uncertain significance(Last reviewed: Apr 28, 2017)	criteria provided, single submitter	VCV000200767	7	150950305	200767	197223	rs794728492	LOF	0.895	0.019	0.086
KCNH2	415	A	T	NM_000238.4(KCNH2):c.2263G>A (p.Ala755Thr)	not provided	Likely pathogenic(Last reviewed: Apr 28, 2014)	criteria provided, single submitter	VCV000200423	7	150950303	200423	197222	rs794728386	LOF	0.889	0.022	0.089
KCNH2	416	M	V	NM_000238.4(KCNH2):c.2266A>G (p.Met756Val)	Long QT syndrome 2|Acquired long QT syndrome	Likely pathogenic(Last reviewed: Dec 11, 2019)	criteria provided, single submitter	VCV000067381	7	150950300	67381	78277	rs199473534	LOF	0.824	0.058	0.118
KCNH2	417	K	N	NM_000238.4(KCNH2):c.2271G>C	Congenital long QT syndrome	not provided	no assertion	VCV000067382	7	150950295	67382	78278	rs199472992	LOF	0.908	0.013	0.079

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Lys757Asn)			provided										
KCNH2	419	K	R	NM_000238.4(KCNH2):c.2276A>G (p.Lys759Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 18, 2020)	criteria provided, single submitter	VCV000999182	7	150950290	999182	992191	rs1225877255	LOF	0.891	0.016	0.092
KCNH2	420	Y	C	NM_000238.4(KCNH2):c.1259A>G (p.Tyr420Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067171	7	150952723	67171	78067	rs199473507	LOF	0.904	0.019	0.077
KCNH2	421	T	K	NM_000238.4(KCNH2):c.1262C>A (p.Thr421Lys)	not provided	Pathogenic(Last reviewed: Mar 31, 2013)	criteria provided, single submitter	VCV000200331	7	150952720	200331	197288	rs199472894	LOF	0.893	0.012	0.095
KCNH2	421	T	M	NM_000238.4(KCNH2):c.1262C>T (p.Thr421Met)	Congenital long QT syndrome|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Apr 30, 2021)	criteria provided, conflicting interpretations	VCV000067173	7	150952720	67173	78069	rs199472894	LOF	0.883	0.017	0.1
KCNH2	422	A	T	NM_000238.4(KCNH2):c.1264G>A (p.Ala422Thr)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Jul 31, 2012)	criteria provided, single submitter	VCV000067174	7	150952718	67174	78070	rs199472895	LOF	0.873	0.029	0.099
KCNH2	424	P	L	NM_000238.4(KCNH2):c.2291C>T (p.Pro764Leu)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Dec 23, 2019)	criteria provided, multiple submitters, no conflicts	VCV000237296	7	150950275	237296	239996	rs878853773	LOF	0.886	0.025	0.09
KCNH2	426	G	R	NM_000238.4(KCNH2):c.2296G>A (p.Gly766Arg)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Sep 2, 2017)	criteria provided, single submitter	VCV000519498	7	150950270	519498	509860	rs1057524377	LOF	0.909	0.006	0.085
KCNH2	426	G	W	NM_000238.4(KCNH2):c.2296G>T (p.Gly766Trp)	not provided	Uncertain significance(Last reviewed: Dec 22, 2016)	criteria provided, single submitter	VCV000392164	7	150950270	392164	369591	rs1057524377	LOF	0.913	0.008	0.079
KCNH2	426	P	H	NM_000238.4(KCNH2):c.1277C>A (p.Pro426His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067175	7	150952705	67175	78071	rs199472896	LOF	0.925	0.006	0.069
KCNH2	427	D	Y	NM_000238.4(KCNH2):c.2299G>T (p.Asp767Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067383	7	150950267	67383	78279	rs199472993	LOF	0.899	0.013	0.088
KCNH2	427	Y	C	NM_000238.4(KCNH2):c.1280A>G (p.Tyr427Cys)	Long QT syndrome 1|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Mar 16, 2017)	criteria provided, single submitter	VCV000067179	7	150952702	67179	78075	rs199472897	LOF	0.918	0.017	0.065
KCNH2	427	Y	H	NM_000238.4(KCNH2):c.1279T>C (p.Tyr427His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067176	7	150952703	67176	78072	rs199472898	LOF	0.917	0.015	0.068
KCNH2	427	Y	S	NM_000238.4(KCNH2):c.1280A>C (p.Tyr427Ser)	Congenital long QT syndrome|Cardiovascular phenotype	Likely pathogenic(Last reviewed: Nov 7, 2016)	criteria provided, multiple submitters, no conflicts	VCV000067178	7	150952702	67178	78074	rs199472897	LOF	0.922	0.014	0.063
KCNH2	428	S	L	NM_000238.4(KCNH2):c.1283C>T (p.Ser428Leu)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Jan 19, 2021)	criteria provided, single submitter	VCV000067180	7	150952699	67180	78076	rs199472899	LOF	0.863	0.024	0.113
KCNH2	428	S	P	NM_000238.4(KCNH2):c.1282T>C (p.Ser428Pro)	not provided	Likely pathogenic(Last reviewed: Mar 26, 2014)	criteria provided, single submitter	VCV000200335	7	150952700	200335	197287	rs794728368	LOF	0.898	0.016	0.086
KCNH2	429	A	P	NM_000238.4(KCNH2):c.1285G>C (p.Ala429Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067181	7	150952697	67181	78077	rs199473508	LOF	0.916	0.015	0.069
KCNH2	430	V	A	NM_000238.4(KCNH2):c.2309T>C (p.Val770Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067384	7	150950257	67384	78280	rs199472994	LOF	0.895	0.023	0.082
KCNH2	431	F	L	NM_000238.4(KCNH2):c.1291T>C	Cardiovascular phenotype|Long QT syndrome	Uncertain	criteria	VCV000519399	7	150952691	519399	509868	rs1554426244	LOF	0.881	0.036	0.083

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Phe431Leu)		significance(Last reviewed: Feb 1, 2020)	provided, multiple submitters, no conflicts										
KCNH2	431	F	L	NM_000238.4(KCNH2):c.1291T>C (p.Phe431Leu)	Cardiovascular phenotype|Long QT syndrome	Uncertain significance(Last reviewed: Feb 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000519399	7	150952691	519399	509868	rs1554426244	LOF	0.881	0.036	0.083
KCNH2	431	F	L	NM_000238.4(KCNH2):c.1293C>A (p.Phe431Leu)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: May 10, 2018)	criteria provided, single submitter	VCV000067183	7	150952689	67183	78079	rs199472900	LOF	0.881	0.036	0.083
KCNH2	431	F	L	NM_000238.4(KCNH2):c.1293C>A (p.Phe431Leu)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: May 10, 2018)	criteria provided, single submitter	VCV000067183	7	150952689	67183	78079	rs199472900	LOF	0.881	0.036	0.083
KCNH2	431	H	P	NM_000238.4(KCNH2):c.2312A>C (p.His771Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 8, 2019)	criteria provided, single submitter	VCV000964746	7	150950254	964746	955094	rs1060500659	LOF	0.914	0.015	0.07
KCNH2	431	H	R	NM_000238.4(KCNH2):c.2312A>G (p.His771Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 24, 2018)	criteria provided, single submitter	VCV000405338	7	150950254	405338	395485	rs1060500659	LOF	0.885	0.014	0.101
KCNH2	434	D	Y	NM_000238.4(KCNH2):c.2320G>T (p.Asp774Tyr)	Congenital long QT syndrome|not provided|Long QT syndrome 2	Pathogenic/Likely pathogenic(Last reviewed: May 10, 2017)	criteria provided, multiple submitters, no conflicts	VCV000067385	7	150950246	67385	78281	rs199472995	LOF	0.692	0.118	0.19
KCNH2	436	L	F	NM_000238.4(KCNH2):c.2326C>T (p.Leu776Phe)	not provided	Likely pathogenic(Last reviewed: Sep 17, 2012)	criteria provided, single submitter	VCV000200425	7	150950240	200425	197221	rs771674303	Neutral	0.566	0.313	0.121
KCNH2	436	T	M	NM_000238.4(KCNH2):c.1307C>T (p.Thr436Met)	Arrhythmia|Long QT syndrome|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Aug 26, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067184	7	150952675	67184	78080	rs199472901	Neutral	0.473	0.403	0.124
KCNH2	438	A	T	NM_000238.4(KCNH2):c.2332G>A (p.Ala778Thr)	Long QT syndrome 2	Uncertain significance	criteria provided, single submitter	VCV000979128	7	150950234	979128	967184	rs769076001	GOF	0.524	0.218	0.258
KCNH2	441	F	L	NM_000238.4(KCNH2):c.2343C>G (p.Phe781Leu)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Dec 23, 2015)	criteria provided, single submitter	VCV000264633	7	150950223	264633	258469	rs886039215	Neutral	0.549	0.28	0.171
KCNH2	442	A	V	NM_000238.4(KCNH2):c.1325C>T (p.Ala442Val)	Long QT syndrome|Long QT syndrome 2|Short QT syndrome 1	Uncertain significance(Last reviewed: Aug 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000456885	7	150952657	456885	456848	rs1554426225	Neutral	0.575	0.255	0.17
KCNH2	442	I	T	NM_000238.4(KCNH2):c.2345T>C (p.Ile782Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 23, 2019)	no assertion criteria provided	VCV000977169	7	150950221	977169	965302	rs1801085857	Neutral	0.619	0.226	0.155
KCNH2	444	R	P	NM_000238.4(KCNH2):c.2351_2352inv (p.Arg784Pro)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jun 4, 2021)	criteria provided, multiple submitters, no conflicts	VCV000660517	7	150950214 - 150950215	660517	635897	NA	Neutral	0.551	0.23	0.219
KCNH2	445	G	A	NM_000238.4(KCNH2):c.2354G>C (p.Gly785Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067387	7	150950212	67387	78283	rs199472996	Neutral	0.706	0.153	0.141

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	445	G	V	NM_000238.4(KCNH2):c.2354G>T (p.Gly785Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067388	7	150950212	67388	78284	rs199472996	Neutral	0.688	0.17	0.142
KCNH2	447	I	N	NM_000238.4(KCNH2):c.2360T>A (p.Ile787Asn)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 3, 2019)	criteria provided, conflicting interpretations	VCV000200428	7	150950206	200428	197220	rs794728387	LOF	0.752	0.121	0.126
KCNH2	448	E	D	NM_000238.4(KCNH2):c.2364G>C (p.Glu788Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067390	7	150950202	67390	78286	rs199473535	LOF	0.83	0.026	0.144
KCNH2	448	E	K	NM_000238.4(KCNH2):c.2362G>A (p.Glu788Lys)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Feb 11, 2016)	criteria provided, single submitter	VCV000067389	7	150950204	67389	78285	rs199472997	LOF	0.798	0.024	0.178
KCNH2	449	I	T	NM_000238.4(KCNH2):c.2366T>C (p.Ile789Thr)	Long QT syndrome|Cardiovascular phenotype	Uncertain significance(Last reviewed: Oct 9, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200431	7	150950200	200431	197219	rs794728388	LOF	0.821	0.082	0.098
KCNH2	450	L	P	NM_000238.4(KCNH2):c.2369T>C (p.Leu790Pro)	Long QT syndrome	Uncertain significance(Last reviewed: May 15, 2020)	criteria provided, single submitter	VCV001027323	7	150950197	1027323	1007342	rs1801084514	LOF	0.883	0.031	0.086
KCNH2	451	R	Q	NM_000238.4(KCNH2):c.2372G>A (p.Arg791Gln)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Feb 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000925151	7	150950194	925151	910738	rs770393669	LOF	0.879	0.015	0.106
KCNH2	451	R	W	NM_000238.4(KCNH2):c.2371C>T (p.Arg791Trp)	Long QT syndrome|not provided|Cardiovascular phenotype|Long QT syndrome 2|Arrhythmia|Brugada syndrome 1|Congenital long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Oct 23, 2020)	criteria provided, conflicting interpretations	VCV000067391	7	150950195	67391	78287	rs138498207	LOF	0.858	0.026	0.116
KCNH2	454	V	I	NM_000238.4(KCNH2):c.2380G>A (p.Val794Ile)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Nov 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200434	7	150950186	200434	197218	rs565006344	LOF	0.847	0.044	0.11
KCNH2	455	V	I	NM_000238.4(KCNH2):c.2383G>A (p.Val795Ile)	Arrhythmia	Uncertain significance(Last reviewed: Dec 4, 2020)	criteria provided, single submitter	VCV001171063	7	150950183	1171063	1160694	NA	LOF	0.832	0.047	0.121
KCNH2	456	V	G	NM_000238.4(KCNH2):c.2387T>G (p.Val796Gly)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Nov 20, 2017)	criteria provided, single submitter	VCV000518748	7	150950179	518748	509859	rs1554425163	LOF	0.921	0.013	0.066
KCNH2	456	V	L	NM_000238.4(KCNH2):c.2386G>C (p.Val796Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV000200438	7	150950180	200438	197216	rs143335921	LOF	0.866	0.03	0.104
KCNH2	457	A	D	NM_000238.4(KCNH2):c.2390C>A (p.Ala797Asp)	not provided|not specified	Likely pathogenic(Last reviewed: Jun 12, 2013)	criteria provided, single submitter	VCV000200440	7	150950176	200440	197215	rs794728389	LOF	0.909	0.014	0.077
KCNH2	458	I	F	NM_000238.4(KCNH2):c.1372A>T (p.Ile458Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 21, 2017)	criteria provided, single submitter	VCV000456887	7	150952610	456887	457100	rs374303744	LOF	0.843	0.041	0.116
KCNH2	459	L	P	NM_000238.4(KCNH2):c.2396T>C (p.Leu799Pro)	not provided	Likely pathogenic(Last reviewed: Aug 13, 2019)	criteria provided, single submitter	VCV000200442	7	150950170	200442	197213	rs794728390	LOF	0.912	0.019	0.068

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	460	G	C	NM_000238.4(KCNH2):c.2398G>T (p.Gly800Trp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067393	7	150950168	67393	78289	rs199472998	LOF	0.915	0.013	0.072
KCNH2	460	G	E	NM_000238.4(KCNH2):c.2399G>A (p.Gly800Glu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067394	7	150949049	67394	78290	rs199473536	LOF	0.908	0.009	0.083
KCNH2	462	N	S	NM_000238.4(KCNH2):c.2405A>G (p.Asn802Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 12, 2015)	criteria provided, single submitter	VCV000216326	7	150949043	216326	212568	rs863224632	LOF	0.908	0.015	0.078
KCNH2	465	F	C	NM_000238.4(KCNH2):c.2414T>G (p.Phe805Cys)	Long QT syndrome|not provided|Cardiovascular phenotype|Congenital long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jul 7, 2017)	criteria provided, multiple submitters, no conflicts	VCV000067397	7	150949034	67397	78293	rs199472999	LOF	0.899	0.027	0.074
KCNH2	465	F	S	NM_000238.4(KCNH2):c.2414T>C (p.Phe805Ser)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Feb 2, 2012)	criteria provided, single submitter	VCV000067396	7	150949034	67396	78292	rs199472999	LOF	0.91	0.021	0.069
KCNH2	466	D	G	NM_000238.4(KCNH2):c.1397A>G (p.Asp466Gly)	Long QT syndrome 2	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000909781	7	150952585	909781	897672	rs1584856497	LOF	0.904	0.012	0.084
KCNH2	466	D	V	NM_000238.4(KCNH2):c.1397A>T (p.Asp466Val)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 7, 2018)	criteria provided, single submitter	VCV000646851	7	150952585	646851	635913	rs1584856497	LOF	0.897	0.014	0.089
KCNH2	466	G	A	NM_000238.4(KCNH2):c.2417G>C (p.Gly806Ala)	not provided	Pathogenic(Last reviewed: Aug 16, 2012)	criteria provided, single submitter	VCV000200453	7	150949031	200453	197203	rs199473000	LOF	0.912	0.014	0.074
KCNH2	466	G	E	NM_000238.4(KCNH2):c.2417G>A (p.Gly806Glu)	Congenital long QT syndrome|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Mar 12, 2021)	criteria provided, conflicting interpretations	VCV000067398	7	150949031	67398	78294	rs199473000	LOF	0.912	0.009	0.079
KCNH2	470	N	D	NM_000238.4(KCNH2):c.1408A>G (p.Asn470Asp)	Congenital long QT syndrome|Long QT syndrome 2	Pathogenic(Last reviewed: Feb 17, 2006)	no assertion criteria provided	VCV000014421	7	150952574	14421	29460	rs121912505	LOF	0.891	0.009	0.1
KCNH2	473	A	T	NM_000238.4(KCNH2):c.2437G>A (p.Ala813Thr)	not specified	Uncertain significance(Last reviewed: May 6, 2019)	criteria provided, single submitter	VCV000928527	7	150949011	928527	917004	rs1801032078	LOF	0.871	0.038	0.09
KCNH2	478	S	P	NM_000238.4(KCNH2):c.2452T>C (p.Ser818Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067400	7	150948996	67400	78296	rs199473537	LOF	0.795	0.084	0.121
KCNH2	479	A	G	NM_000238.4(KCNH2):c.2398+58C>G	Arrhythmia	Likely benign(Last reviewed: Sep 4, 2019)	criteria provided, single submitter	VCV000927904	7	150950110	927904	915876	rs774109163	LOF	0.857	0.048	0.096
KCNH2	480	G	R	NM_000238.4(KCNH2):c.2458G>A (p.Gly820Arg)	Arrhythmia|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Jan 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067401	7	150948990	67401	78297	rs199473001	LOF	0.809	0.061	0.13
KCNH2	482	V	L	NM_000238.4(KCNH2):c.2464G>C (p.Val822Leu)	not provided	Likely pathogenic(Last reviewed: Jun 15, 2017)	criteria provided, single submitter	VCV000200456	7	150948984	200456	197199	rs121912506	LOF	0.873	0.047	0.08
KCNH2	483	R	Q	NM_000238.4(KCNH2):c.2468G>A (p.Arg823Gln)	Long QT syndrome|not provided|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Oct 15, 2020)	criteria provided, conflicting interpretations	VCV000418246	7	150948980	418246	407055	rs1064793147	LOF	0.884	0.048	0.068
KCNH2	483	R	W	NM_000238.4(KCNH2):c.2467C>T (p.Arg823Trp)	Long QT syndrome|Congenital long QT syndrome|not provided|Cardiovascular	Pathogenic/Likely pathogenic(Last	criteria provided,	VCV000067402	7	150948981	67402	78298	rs199473538	LOF	0.882	0.047	0.071

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					phenotype	reviewed: Nov 7, 2021)	multiple submitters, no conflicts										
KCNH2	483	V	I	NM_000238.4(KCNH2):c.1447G>A (p.Val483Ile)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Sep 16, 2020)	criteria provided, multiple submitters, no conflicts	VCV000858924	7	150952535	858924	833341	rs755834128	LOF	0.874	0.05	0.076
KCNH2	486	T	I	NM_000238.4(KCNH2):c.2477C>T (p.Thr826Ile)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Jun 12, 2013)	criteria provided, single submitter	VCV000067403	7	150948971	67403	78299	rs199473002	LOF	0.883	0.031	0.086
KCNH2	488	C	R	NM_000238.4(KCNH2):c.2482T>C (p.Cys828Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 15, 2019)	criteria provided, single submitter	VCV000842229	7	150948966	842229	833335	rs1801029729	LOF	0.832	0.027	0.141
KCNH2	489	D	A	NM_000238.4(KCNH2):c.2486A>C (p.Asp829Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 1, 2020)	criteria provided, single submitter	VCV000851362	7	150948962	851362	833334	rs1210175432	LOF	0.914	0.014	0.073
KCNH2	490	A	T	NM_000238.4(KCNH2):c.1468G>A (p.Ala490Thr)	Long QT syndrome 2|not provided|Congenital long QT syndrome|Long QT syndrome, bradycardia-induced|Long QT syndrome	Pathogenic(Last reviewed: Sep 21, 2019)	criteria provided, multiple submitters, no conflicts	VCV000014430	7	150952514	14430	29469	rs28928905	LOF	0.89	0.026	0.084
KCNH2	490	A	V	NM_000238.4(KCNH2):c.1469C>T (p.Ala490Val)	Long QT syndrome 2|Long QT syndrome|Inborn genetic diseases	Conflicting interpretations of pathogenicity(Last reviewed: Aug 20, 2020)	criteria provided, conflicting interpretations	VCV000862292	7	150952513	862292	833340	rs1801214475	LOF	0.88	0.028	0.092
KCNH2	492	H	L	NM_000238.4(KCNH2):c.1475A>T (p.His492Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 14, 2018)	criteria provided, single submitter	VCV000581371	7	150952507	581371	561738	rs1563160015	LOF	0.876	0.027	0.098
KCNH2	493	Y	H	NM_000238.4(KCNH2):c.1477T>C (p.Tyr493His)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 12, 2018)	criteria provided, single submitter	VCV000658590	7	150952505	658590	635912	rs1584856217	LOF	0.923	0.012	0.065
KCNH2	495	L	R	NM_000238.4(KCNH2):c.2398+106T>G	Arrhythmia	Likely benign(Last reviewed: Nov 16, 2018)	criteria provided, single submitter	VCV000919939	7	150950062	919939	916154	rs1489831109	LOF	0.89	0.018	0.092
KCNH2	495	R	Q	NM_000238.4(KCNH2):c.2504G>A (p.Arg835Gln)	not provided	Uncertain significance(Last reviewed: Aug 14, 2019)	criteria provided, single submitter	VCV000200464	7	150948944	200464	197197	rs794728395	LOF	0.893	0.014	0.094
KCNH2	495	R	W	NM_000238.4(KCNH2):c.2503C>T (p.Arg835Trp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067404	7	150948945	67404	78300	rs199473003	LOF	0.885	0.017	0.098
KCNH2	497	D	G	NM_000238.4(KCNH2):c.2510A>G (p.Asp837Gly)	Long QT syndrome|Arrhythmia|Long QT syndrome 2|not provided|Congenital long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jan 25, 2022)	criteria provided, multiple submitters, no conflicts	VCV000067407	7	150948938	67407	78303	rs199473004	LOF	0.918	0.015	0.067
KCNH2	497	D	N	NM_000238.4(KCNH2):c.2509G>A (p.Asp837Asn)	Congenital long QT syndrome|Long QT syndrome|Long QT syndrome 2	Pathogenic/Likely pathogenic(Last reviewed: Feb 7, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067405	7	150948939	67405	78301	rs199473005	LOF	0.911	0.013	0.076
KCNH2	497	D	Y	NM_000238.4(KCNH2):c.2509G>T (p.Asp837Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067406	7	150948939	67406	78302	rs199473005	LOF	0.894	0.014	0.092
KCNH2	497	G	V	NM_000238.4(KCNH2):c.2398+112G>T	Arrhythmia	Likely benign(Last reviewed: Aug 11, 2019)	criteria provided, single submitter	VCV000926469	7	150950056	926469	915868	rs771057684	LOF	0.89	0.024	0.087

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	499	L	M	NM_000238.4(KCNH2):c.2515C>A (p.Leu839Met)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000920490	7	150948933	920490	910732	rs1801028129	LOF	0.835	0.058	0.107
KCNH2	502	L	P	NM_000238.4(KCNH2):c.2525T>C (p.Leu842Pro)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Jun 22, 2017)	criteria provided, single submitter	VCV000519212	7	150948923	519212	509857	rs1554424818	LOF	0.909	0.023	0.068
KCNH2	504	M	V	NM_000238.4(KCNH2):c.2530A>G (p.Met844Val)	not provided|Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 14, 2020)	criteria provided, multiple submitters, no conflicts	VCV000658202	7	150948918	658202	635896	rs754565340	LOF	0.869	0.034	0.096
KCNH2	506	P	A	NM_000238.4(KCNH2):c.2536C>G (p.Pro846Ala)	Long QT syndrome 2	Uncertain significance(Last reviewed: Jun 1, 2014)	no assertion criteria provided	VCV000161254	7	150948912	161254	171107	rs199473006	LOF	0.899	0.026	0.074
KCNH2	506	P	L	NM_000238.4(KCNH2):c.2537C>T (p.Pro846Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 11, 2017)	criteria provided, single submitter	VCV000405353	7	150948911	405353	396141	rs1060500669	LOF	0.878	0.03	0.092
KCNH2	506	P	S	NM_000238.4(KCNH2):c.2536C>T (p.Pro846Ser)	Congenital long QT syndrome|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: May 20, 2019)	criteria provided, conflicting interpretations	VCV000067409	7	150948912	67409	78305	rs199473006	LOF	0.916	0.017	0.067
KCNH2	506	P	T	NM_000238.4(KCNH2):c.2536C>A (p.Pro846Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067408	7	150948912	67408	78304	rs199473006	LOF	0.909	0.02	0.072
KCNH2	507	E	D	NM_000238.4(KCNH2):c.2541G>T (p.Glu847Asp)	Arrhythmia	Uncertain significance(Last reviewed: Apr 19, 2021)	criteria provided, single submitter	VCV001332607	7	150948907	1332607	1323325	NA	LOF	0.908	0.011	0.081
KCNH2	507	P	A	NM_000238.4(KCNH2):c.1519C>G (p.Pro507Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 1, 2018)	criteria provided, single submitter	VCV000656162	7	150952463	656162	635910	rs794728371	LOF	0.918	0.015	0.067
KCNH2	510	D	Y	NM_000238.4(KCNH2):c.2548G>T (p.Asp850Tyr)	Arrhythmia	Uncertain significance(Last reviewed: Jan 28, 2019)	criteria provided, single submitter	VCV000922966	7	150948900	922966	910731	rs1036885088	LOF	0.897	0.015	0.088
KCNH2	514	M	V	NM_000238.4(KCNH2):c.2398+162A>G	Arrhythmia	Likely benign(Last reviewed: Dec 13, 2018)	criteria provided, single submitter	VCV000920091	7	150950006	920091	916153	rs1246310515	LOF	0.836	0.061	0.103
KCNH2	518	I	T	NM_000238.4(KCNH2):c.2573T>C (p.Ile858Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067412	7	150948875	67412	78308	rs199473539	LOF	0.869	0.043	0.087
KCNH2	519	T	I	NM_000238.4(KCNH2):c.2576C>T (p.Thr859Ile)	not provided	Uncertain significance(Last reviewed: Dec 31, 2019)	criteria provided, single submitter	VCV000200469	7	150948872	200469	197196	rs794728396	LOF	0.838	0.056	0.106
KCNH2	521	N	H	NM_000238.4(KCNH2):c.2581A>C (p.Asn861His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067415	7	150948867	67415	78311	rs199473007	LOF	0.89	0.015	0.095
KCNH2	521	N	S	NM_000238.4(KCNH2):c.2582A>G (p.Asn861Ser)	not provided	Pathogenic(Last reviewed: Oct 30, 2013)	criteria provided, single submitter	VCV000200471	7	150948866	200471	197195	rs121912513	LOF	0.895	0.016	0.089
KCNH2	521	N	T	NM_000238.4(KCNH2):c.2582A>C (p.Asn861Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067416	7	150948866	67416	78312	rs121912513	LOF	0.889	0.018	0.093
KCNH2	523	L	M	NM_000238.4(KCNH2):c.1567C>A (p.Leu523Met)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 24,	criteria provided, single submitter	VCV000405357	7	150951826	405357	396158	rs1060500671	LOF	0.866	0.034	0.1

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2016)											
KCNH2	523	R	Q	NM_000238.4(KCNH2):c.2588G>A (p.Arg863Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 11, 2020)	criteria provided, single submitter	VCV001026816	7	150948860	1026816	1007341	rs939435672	LOF	0.887	0.017	0.096
KCNH2	524	D	G	NM_000238.4(KCNH2):c.2591A>G (p.Asp864Gly)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Jan 15, 2020)	criteria provided, single submitter	VCV000067417	7	150948857	67417	78313	rs199473008	LOF	0.91	0.013	0.077
KCNH2	527	M	T	NM_000238.4(KCNH2):c.2600T>C (p.Met867Thr)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Feb 3, 2020)	criteria provided, multiple submitters, no conflicts	VCV000641744	7	150948536	641744	635895	rs752020408	LOF	0.888	0.02	0.092
KCNH2	529	P	L	NM_000238.4(KCNH2):c.2606C>T (p.Pro869Leu)	not provided|Brugada syndrome 1|Arrhythmia	Uncertain significance(Last reviewed: Dec 13, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200473	7	150948530	200473	197194	rs763031434	LOF	0.89	0.02	0.089
KCNH2	531	S	F	NM_000238.4(KCNH2):c.2612C>T (p.Ser871Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 11, 2019)	criteria provided, single submitter	VCV000954476	7	150948524	954476	945519	rs1177213407	LOF	0.876	0.015	0.109
KCNH2	533	G	S	NM_000238.4(KCNH2):c.2617G>A (p.Gly873Ser)	not provided|Brugada syndrome|Long QT syndrome|not specified|Arrhythmia	Benign/Likely benign(Last reviewed: Dec 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067419	7	150948519	67419	78315	rs41314354	LOF	0.893	0.019	0.088
KCNH2	535	T	M	NM_000238.4(KCNH2):c.2624C>T (p.Thr875Met)	Long QT syndrome|not specified|Arrhythmia|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 11, 2021)	criteria provided, conflicting interpretations	VCV000067420	7	150948512	67420	78316	rs140743924	LOF	0.871	0.023	0.105
KCNH2	535	T	R	NM_000238.4(KCNH2):c.2624C>G (p.Thr875Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 26, 2019)	criteria provided, single submitter	VCV000849676	7	150948512	849676	833333	rs140743924	LOF	0.872	0.015	0.112
KCNH2	538	E	D	NM_000238.4(KCNH2):c.2634G>T (p.Glu878Asp)	not provided	Uncertain significance(Last reviewed: Jul 26, 2018)	criteria provided, single submitter	VCV001304037	7	150948502	1304037	1294312	NA	LOF	0.863	0.012	0.125
KCNH2	538	R	H	NM_000238.4(KCNH2):c.2398+235G>A	not provided	Uncertain significance(Last reviewed: Dec 24, 2019)	criteria provided, single submitter	VCV001311311	7	150949933	1311311	1300692	NA	LOF	0.875	0.014	0.11
KCNH2	539	G	R	NM_000238.4(KCNH2):c.2635G>C (p.Gly879Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067421	7	150948501	67421	78317	rs199473040	LOF	0.85	0.017	0.133
KCNH2	539	G	S	NM_000238.4(KCNH2):c.2635G>A (p.Gly879Ser)	Arrhythmia	Uncertain significance(Last reviewed: Nov 29, 2018)	criteria provided, single submitter	VCV000924749	7	150948501	924749	910726	rs199473040	LOF	0.89	0.022	0.088
KCNH2	541	R	C	NM_000238.4(KCNH2):c.1621C>T (p.Arg541Cys)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 7, 2020)	criteria provided, multiple submitters, no conflicts	VCV000937094	7	150951772	937094	933773	rs764666519	LOF	0.853	0.024	0.123
KCNH2	543	R	Q	NM_000238.4(KCNH2):c.2648G>A (p.Arg883Gln)	Arrhythmia	Uncertain significance(Last reviewed: Jan 5, 2021)	criteria provided, single submitter	VCV001171663	7	150948488	1171663	1160692	NA	LOF	0.839	0.019	0.143
KCNH2	544	Q	R	NM_000238.4(KCNH2):c.2651A>G	not specified	Uncertain	criteria	VCV000633279	7	150948485	633279	621262	rs1563149602	LOF	0.743	0.029	0.228

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Gln884Arg)		significance(Last reviewed: Nov 6, 2017)	provided, single submitter										
KCNH2	545	R	C	NM_000238.4(KCNH2):c.2653C>T (p.Arg885Cys)	Long QT syndrome|Congenital long QT syndrome|not specified|Arrhythmia	Uncertain significance(Last reviewed: Oct 21, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067422	7	150948483	67422	78318	rs143512106	LOF	0.819	0.049	0.132
KCNH2	545	R	H	NM_000238.4(KCNH2):c.2654G>A (p.Arg885His)	Long QT syndrome|not specified|Arrhythmia|not provided|Long QT syndrome 2	Uncertain significance(Last reviewed: Nov 26, 2019)	criteria provided, multiple submitters, no conflicts	VCV000402995	7	150948482	402995	389717	rs202194495	LOF	0.842	0.031	0.128
KCNH2	545	R	S	NM_000238.4(KCNH2):c.2653C>A (p.Arg885Ser)	Arrhythmia	Uncertain significance(Last reviewed: Mar 2, 2021)	criteria provided, single submitter	VCV001332377	7	150948483	1332377	1323095	NA	LOF	0.847	0.032	0.121
KCNH2	547	A	T	NM_000238.4(KCNH2):c.1639G>A (p.Ala547Thr)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000663436	7	150951754	663436	635907	rs376021230	LOF	0.864	0.031	0.105
KCNH2	547	R	C	NM_000238.4(KCNH2):c.2659C>T (p.Arg887Cys)	not provided|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Jun 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000193802	7	150948477	193802	190965	rs140279503	LOF	0.868	0.023	0.109
KCNH2	547	R	G	NM_000238.4(KCNH2):c.2659C>G (p.Arg887Gly)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Mar 26, 2021)	criteria provided, multiple submitters, no conflicts	VCV000924438	7	150948477	924438	910724	rs140279503	LOF	0.878	0.018	0.104
KCNH2	547	R	H	NM_000238.4(KCNH2):c.2660G>A (p.Arg887His)	Long QT syndrome|Long QT syndrome 2| Congenital long QT syndrome|not specified|not provided|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Jul 28, 2021)	criteria provided, conflicting interpretations	VCV000067423	7	150948476	67423	78319	rs199473432	LOF	0.873	0.017	0.11
KCNH2	549	L	V	NM_000238.4(KCNH2):c.2665T>G (p.Leu889Val)	Arrhythmia|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jul 29, 2020)	criteria provided, conflicting interpretations	VCV000200475	7	150948471	200475	197192	rs765427343	LOF	0.866	0.029	0.105
KCNH2	550	S	P	NM_000238.4(KCNH2):c.2668T>C (p.Ser890Pro)	Arrhythmia	Uncertain significance(Last reviewed: Nov 1, 2019)	criteria provided, single submitter	VCV000926306	7	150948468	926306	910723	rs1801006262	LOF	0.926	0.012	0.062
KCNH2	551	F	Y	NM_000238.4(KCNH2):c.2672_2673delinsAT (p.Phe891Tyr)	Arrhythmia	Uncertain significance(Last reviewed: Mar 8, 2021)	criteria provided, single submitter	VCV001331999	7	150948463 - 150948464	1331999	1322719	NA	LOF	0.859	0.029	0.112
KCNH2	552	R	C	NM_000238.4(KCNH2):c.2674C>T (p.Arg892Cys)	Cardiovascular phenotype|Arrhythmia|Long QT syndrome|not specified|not provided|Long QT syndrome 2	Conflicting interpretations of pathogenicity(Last reviewed: Nov 11, 2020)	criteria provided, conflicting interpretations	VCV000191470	7	150948462	191470	189257	rs201627778	LOF	0.916	0.017	0.068
KCNH2	552	R	H	NM_000238.4(KCNH2):c.2675G>A (p.Arg892His)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jan 12, 2021)	criteria provided, multiple submitters, no conflicts	VCV001054268	7	150948461	1054268	1044805	NA	LOF	0.916	0.011	0.073
KCNH2	553	R	K	NM_000238.4(KCNH2):c.2678G>A (p.Arg893Lys)	Long QT syndrome	Uncertain significance(Last	criteria provided, single	VCV001025677	7	150948458	1025677	1007340	rs1801005127	LOF	0.91	0.012	0.078

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Mar 2, 2020)	submitter										
KCNH2	554	R	C	NM_000238.4(KCNH2):c.2680C>T (p.Arg894Cys)	not provided|Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Dec 13, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067424	7	150948456	67424	78320	rs199473433	LOF	0.911	0.019	0.071
KCNH2	554	R	H	NM_000238.4(KCNH2):c.2681G>A (p.Arg894His)	Arrhythmia|Long QT syndrome 2|Inborn genetic diseases	Uncertain significance(Last reviewed: Oct 14, 2020)	criteria provided, multiple submitters, no conflicts	VCV000448972	7	150948455	448972	442550	rs199473668	LOF	0.913	0.012	0.076
KCNH2	554	R	L	NM_000238.4(KCNH2):c.2681G>T (p.Arg894Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067425	7	150948455	67425	78321	rs199473668	LOF	0.888	0.023	0.089
KCNH2	555	T	M	NM_000238.4(KCNH2):c.2684C>T (p.Thr895Met)	Arrhythmia|Long QT syndrome 2|Long QT syndrome|not provided|SUDDEN INFANT DEATH SYNDROME|Long QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: May 17, 2020)	criteria provided, conflicting interpretations	VCV000067426	7	150948452	67426	78322	rs199473434	LOF	0.879	0.019	0.103
KCNH2	555	T	R	NM_000238.4(KCNH2):c.2684C>G (p.Thr895Arg)	Arrhythmia	Uncertain significance(Last reviewed: Mar 3, 2021)	criteria provided, single submitter	VCV000919273	7	150948452	919273	910719	rs199473434	LOF	0.9	0.012	0.088
KCNH2	557	K	M	NM_000238.4(KCNH2):c.2690A>T (p.Lys897Met)	not provided|Arrhythmia	Uncertain significance(Last reviewed: Feb 25, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200480	7	150948446	200480	197190	rs1805123	LOF	0.908	0.014	0.078
KCNH2	557	K	T	NM_000238.4(KCNH2):c.2690A>C (p.Lys897Thr)	Arrhythmia|Atrial fibrillation|Cardiovascular phenotype|Long QT syndrome 2|not provided|Long QT syndrome|not specified	Benign(Last reviewed: Sep 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067427	7	150948446	67427	78323	rs1805123	LOF	0.915	0.013	0.072
KCNH2	558	A	P	NM_000238.4(KCNH2):c.1672G>C (p.Ala558Pro)	Long QT syndrome 2|Congenital long QT syndrome	Pathogenic(Last reviewed: Jul 1, 2008)	no assertion criteria provided	VCV000014444	7	150951721	14444	29483	rs121912516	LOF	0.902	0.018	0.08
KCNH2	559	L	P	NM_000238.4(KCNH2):c.1676T>C (p.Leu559Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 28, 2018)	criteria provided, single submitter	VCV000656408	7	150951717	656408	635904	rs199472920	LOF	0.905	0.017	0.078
KCNH2	559	T	K	NM_000238.4(KCNH2):c.2696C>A (p.Thr899Lys)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 12, 2020)	criteria provided, single submitter	VCV001006883	7	150947875	1006883	992190	rs1480554629	LOF	0.907	0.012	0.081
KCNH2	559	T	M	NM_000238.4(KCNH2):c.2696C>T (p.Thr899Met)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Dec 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000862133	7	150947875	862133	833330	rs1480554629	LOF	0.884	0.017	0.099
KCNH2	559	T	R	NM_000238.4(KCNH2):c.2696C>G (p.Thr899Arg)	Long QT syndrome|Cardiovascular phenotype	Uncertain significance(Last reviewed: Jul 17, 2019)	criteria provided, multiple submitters, no conflicts	VCV000456916	7	150947875	456916	456478	rs1480554629	LOF	0.909	0.011	0.08
KCNH2	559	T	S	NM_000238.4(KCNH2):c.2695A>T (p.Thr899Ser)	Arrhythmia	Uncertain significance(Last reviewed: Feb 11, 2019)	criteria provided, single submitter	VCV000919954	7	150947876	919954	910718	rs1800976319	LOF	0.9	0.016	0.084
KCNH2	561	A	V	NM_000238.4(KCNH2):c.1682C>T (p.Ala561Val)	Long QT syndrome 2|not provided|Long QT syndrome|Cardiovascular phenotype|Congenital long QT syndrome|Prolonged QT interval|Obesity	Pathogenic/Likely pathogenic(Last reviewed: Dec 17, 2021)	criteria provided, multiple submitters, no conflicts	VCV000014420	7	150951711	14420	29459	rs121912504	LOF	0.881	0.02	0.099

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	563	G	R	NM_000238.4(KCNH2):c.2707G>A (p.Gly903Arg)	Arrhythmia|Cardiovascular phenotype| Congenital long QT syndrome|not specified|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Feb 12, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067428	7	150947864	67428	78324	rs199473669	LOF	0.917	0.012	0.071
KCNH2	564	E	G	NM_000238.4(KCNH2):c.2711A>G (p.Glu904Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 16, 2018)	criteria provided, single submitter	VCV000642118	7	150947860	642118	635893	rs794727044	LOF	0.911	0.02	0.07
KCNH2	564	E	K	NM_000238.4(KCNH2):c.2710G>A (p.Glu904Lys)	Arrhythmia	Uncertain significance(Last reviewed: Jan 5, 2021)	criteria provided, single submitter	VCV001171262	7	150947861	1171262	1160690	NA	LOF	0.893	0.017	0.091
KCNH2	564	E	V	NM_000238.4(KCNH2):c.2711A>T (p.Glu904Val)	not provided	Uncertain significance(Last reviewed: Feb 2, 2015)	criteria provided, single submitter	VCV000193977	7	150947860	193977	191140	rs794727044	LOF	0.887	0.026	0.086
KCNH2	564	L	Q	NM_000238.4(KCNH2):c.1691T>A (p.Leu564Gln)	Long QT syndrome	Likely pathogenic(Last reviewed: Jul 9, 2019)	criteria provided, single submitter	VCV000572344	7	150951702	572344	561404	rs199472924	LOF	0.887	0.02	0.093
KCNH2	566	S	L	NM_000238.4(KCNH2):c.2717C>T (p.Ser906Leu)	Long QT syndrome|not specified|Congenital long QT syndrome|Long QT syndrome 2|Short QT syndrome 1|Arrhythmia|not provided	Uncertain significance(Last reviewed: May 3, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067429	7	150947854	67429	78325	rs199473435	LOF	0.897	0.016	0.088
KCNH2	567	A	T	NM_000238.4(KCNH2):c.2719G>A (p.Ala907Thr)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: May 8, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200486	7	150947852	200486	197187	rs752686806	LOF	0.893	0.021	0.085
KCNH2	570	P	L	NM_000238.4(KCNH2):c.2729C>T (p.Pro910Leu)	Long QT syndrome|not specified|Cardiovascular phenotype|not provided|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Sep 13, 2021)	criteria provided, conflicting interpretations	VCV000067430	7	150947842	67430	78326	rs199473436	LOF	0.913	0.016	0.071
KCNH2	571	G	R	NM_000238.4(KCNH2):c.2731G>C (p.Gly911Arg)	Arrhythmia	Uncertain significance(Last reviewed: Mar 27, 2019)	criteria provided, single submitter	VCV000918838	7	150947840	918838	910714	rs1800973294	LOF	0.901	0.009	0.09
KCNH2	572	G	R	NM_000238.4(KCNH2):c.1714G>C (p.Gly572Arg)	Long QT syndrome 2|Congenital long QT syndrome	Pathogenic(Last reviewed: Feb 1, 2000)	no assertion criteria provided	VCV000014429	7	150951679	14429	29468	rs9333649	LOF	0.862	0.007	0.131
KCNH2	572	R	Q	NM_000238.4(KCNH2):c.2735G>A (p.Arg912Gln)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Dec 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000527012	7	150947836	527012	522670	rs958442820	LOF	0.897	0.008	0.095
KCNH2	572	R	W	NM_000238.4(KCNH2):c.2734C>T (p.Arg912Trp)	Arrhythmia|Long QT syndrome|not provided	Uncertain significance(Last reviewed: Oct 15, 2020)	criteria provided, multiple submitters, no conflicts	VCV000180384	7	150947837	180384	178573	rs577847157	LOF	0.908	0.011	0.081
KCNH2	573	A	T	NM_000238.4(KCNH2):c.2737G>A (p.Ala913Thr)	not provided	Uncertain significance(Last reviewed: Oct 7, 2020)	criteria provided, single submitter	VCV001188698	7	150947834	1188698	1176883	NA	LOF	0.822	0.043	0.135
KCNH2	573	A	V	NM_000238.4(KCNH2):c.2738C>T (p.Ala913Val)	Long QT syndrome|Cardiovascular phenotype| Long QT syndrome 2|Congenital long QT syndrome|not provided|Arrhythmia|Long QT syndrome 2/9, digenic	Conflicting interpretations of pathogenicity(Last reviewed: Dec 1, 2021)	criteria provided, conflicting interpretations	VCV000014443	7	150947833	14443	29482	rs77331749	LOF	0.833	0.051	0.116
KCNH2	575	A	V	NM_000238.4(KCNH2):c.2744C>T	not provided	not provided	no assertion	VCV000067431	7	150947827	67431	78327	rs199473437	LOF	0.823	0.063	0.114

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Ala915Val)			provided										
KCNH2	577	P	L	NM_000238.4(KCNH2):c.2750C>T (p.Pro917Leu)	Long QT syndrome|Long QT syndrome 2|not specified|not provided|Arrhythmia	Uncertain significance(Last reviewed: Jul 29, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067432	7	150947821	67432	78328	rs76420733	LOF	0.734	0.113	0.153
KCNH2	577	P	Q	NM_000238.4(KCNH2):c.1730C>A (p.Pro577Gln)	not provided	Uncertain significance(Last reviewed: Nov 1, 2019)	criteria provided, single submitter	VCV000872354	7	150951663	872354	859593	rs1801171859	LOF	0.715	0.08	0.205
KCNH2	578	S	N	NM_000238.4(KCNH2):c.2753G>A (p.Ser918Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 15, 2019)	criteria provided, single submitter	VCV000939122	7	150947818	939122	933765	rs1268921735	LOF	0.688	0.092	0.221
KCNH2	580	R	G	NM_000238.4(KCNH2):c.2758C>G (p.Arg920Gly)	not provided|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Apr 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200490	7	150947813	200490	197181	rs199473438	LOF	0.759	0.094	0.146
KCNH2	580	R	Q	NM_000238.4(KCNH2):c.2759G>A (p.Arg920Gln)	Long QT syndrome|Arrhythmia|Cardiovascular phenotype|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Dec 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067434	7	150947812	67434	78330	rs199473670	LOF	0.715	0.089	0.195
KCNH2	580	R	W	NM_000238.4(KCNH2):c.2758C>T (p.Arg920Trp)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome	Uncertain significance(Last reviewed: Dec 23, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067433	7	150947813	67433	78329	rs199473438	Neutral	0.696	0.15	0.154
KCNH2	581	S	L	NM_000238.4(KCNH2):c.1742C>T (p.Ser581Leu)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Dec 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000577995	7	150951651	577995	561727	rs794728483	LOF	0.728	0.128	0.144
KCNH2	582	R	C	NM_000238.4(KCNH2):c.1744C>T (p.Arg582Cys)	Long QT syndrome 2|not provided|Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Nov 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000014428	7	150951649	14428	29467	rs121912508	Neutral	0.648	0.222	0.13
KCNH2	582	R	H	NM_000238.4(KCNH2):c.1745G>A (p.Arg582His)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Feb 19, 2021)	criteria provided, multiple submitters, no conflicts	VCV000926210	7	150951648	926210	910754	rs199473426	Neutral	0.697	0.163	0.139
KCNH2	582	R	L	NM_000238.4(KCNH2):c.2765G>T (p.Arg922Leu)	Arrhythmia	Uncertain significance(Last reviewed: Nov 4, 2019)	criteria provided, single submitter	VCV000924058	7	150947806	924058	910711	rs199473439	Neutral	0.637	0.228	0.135
KCNH2	582	R	Q	NM_000238.4(KCNH2):c.2765G>A (p.Arg922Gln)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Aug 26, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067436	7	150947806	67436	78332	rs199473439	LOF	0.709	0.135	0.155
KCNH2	582	R	W	NM_000238.4(KCNH2):c.2764C>T (p.Arg922Trp)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Mar 18, 2020)	criteria provided, single submitter	VCV000067435	7	150947807	67435	78331	rs199473440	Neutral	0.647	0.22	0.133
KCNH2	583	P	L	NM_000238.4(KCNH2):c.2768C>T (p.Pro923Leu)	Arrhythmia|Long QT syndrome|Long QT syndrome 2	Uncertain significance(Last reviewed: Apr 5, 2021)	criteria provided, multiple submitters, no conflicts	VCV000572278	7	150947803	572278	561705	rs760408746	LOF	0.837	0.067	0.096

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	583	P	Q	NM_000238.4(KCNH2):c.2768C>A (p.Pro923Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 17, 2020)	criteria provided, single submitter	VCV001008348	7	150947803	1008348	992189	rs760408746	LOF	0.835	0.042	0.123
KCNH2	584	G	A	NM_000238.4(KCNH2):c.2771G>C (p.Gly924Ala)	Long QT syndrome|Arrhythmia|not specified|Cardiovascular phenotype|Short QT syndrome 1|Congenital long QT syndrome	Uncertain significance(Last reviewed: Feb 17, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067438	7	150947800	67438	78334	rs199473009	LOF	0.856	0.025	0.119
KCNH2	584	G	E	NM_000238.4(KCNH2):c.2771G>A (p.Gly924Glu)	Cardiovascular phenotype|Congenital long QT syndrome|not provided|Long QT syndrome|Arrhythmia|Long QT syndrome 2	Conflicting interpretations of pathogenicity(Last reviewed: Sep 15, 2020)	criteria provided, conflicting interpretations	VCV000067437	7	150947800	67437	78333	rs199473009	LOF	0.842	0.016	0.142
KCNH2	584	G	R	NM_000238.4(KCNH2):c.2770G>A (p.Gly924Arg)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Sep 26, 2018)	criteria provided, multiple submitters, no conflicts	VCV000200492	7	150947801	200492	197178	rs794728397	LOF	0.795	0.016	0.189
KCNH2	584	G	W	NM_000238.4(KCNH2):c.2770G>T (p.Gly924Trp)	Long QT syndrome|Arrhythmia|Cardiovascular phenotype	Uncertain significance(Last reviewed: Sep 17, 2019)	criteria provided, multiple submitters, no conflicts	VCV000519287	7	150947801	519287	509851	rs794728397	LOF	0.863	0.025	0.113
KCNH2	585	G	A	NM_000238.4(KCNH2):c.2774G>C (p.Gly925Ala)	not provided	Uncertain significance(Last reviewed: Aug 17, 2018)	criteria provided, single submitter	VCV000200495	7	150947797	200495	197177	rs794728398	LOF	0.92	0.013	0.067
KCNH2	585	G	E	NM_000238.4(KCNH2):c.2774G>A (p.Gly925Glu)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jul 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000921867	7	150947797	921867	910709	rs794728398	LOF	0.92	0.008	0.072
KCNH2	585	G	R	NM_000238.4(KCNH2):c.2773G>A (p.Gly925Arg)	not provided	not provided	no assertion provided	VCV000067439	7	150947798	67439	78335	rs199473010	LOF	0.896	0.009	0.096
KCNH2	585	G	V	NM_000238.4(KCNH2):c.2774G>T (p.Gly925Val)	Sudden unexplained death|Long QT syndrome|not provided	Uncertain significance(Last reviewed: Jun 26, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200497	7	150947797	200497	197176	rs794728398	LOF	0.914	0.014	0.072
KCNH2	585	G	V	NM_000238.4(KCNH2):c.2774G>T (p.Gly925Val)	Sudden unexplained death|Long QT syndrome|not provided	Uncertain significance(Last reviewed: Jun 26, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200497	7	150947797	200497	197176	rs794728398	LOF	0.914	0.014	0.072
KCNH2	585	G	V	NM_000238.4(KCNH2):c.2774_2775delinsTT (p.Gly925Val)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 20, 2020)	criteria provided, single submitter	VCV001058588	7	150947796 - 150947797	1058588	1044804	NA	LOF	0.914	0.014	0.072
KCNH2	585	G	V	NM_000238.4(KCNH2):c.2774_2775delinsTT (p.Gly925Val)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 20, 2020)	criteria provided, single submitter	VCV001058588	7	150947796 - 150947797	1058588	1044804	NA	LOF	0.914	0.014	0.072
KCNH2	585	W	C	NM_000238.4(KCNH2):c.1755G>C (p.Trp585Cys)	Long QT syndrome	Likely pathogenic(Last reviewed: Mar 16, 2018)	criteria provided, single submitter	VCV000581669	7	150951638	581669	564156	rs199473430	LOF	0.918	0.022	0.06
KCNH2	586	P	L	NM_000238.4(KCNH2):c.2777C>T (p.Pro926Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 11, 2019)	criteria provided, single submitter	VCV000848808	7	150947794	848808	833327	rs996063617	LOF	0.914	0.017	0.069
KCNH2	587	W	L	NM_000238.4(KCNH2):c.2780G>T	Arrhythmia|Long QT syndrome 2|not provided|	Uncertain	criteria	VCV000200501	7	150947791	200501	197172	rs794728399	LOF	0.882	0.038	0.079

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Trp927Leu)	Long QT syndrome	significance(Last reviewed: Apr 15, 2021)	provided, multiple submitters, no conflicts										
KCNH2	588	G	E	NM_000238.4(KCNH2):c.2783G>A (p.Gly928Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 7, 2020)	criteria provided, single submitter	VCV000850359	7	150947788	850359	833326	rs1339223823	LOF	0.786	0.031	0.183
KCNH2	588	N	K	NM_000238.4(KCNH2):c.1764C>G (p.Asn588Lys)	Short QT syndrome 1|short QT syndrome	Pathogenic(Last reviewed: Apr 1, 2005)	no assertion criteria provided	VCV000014436	7	150951629	14436	29475	rs104894021	GOF	0.706	0.038	0.256
KCNH2	588	N	K	NM_000238.4(KCNH2):c.1764C>G (p.Asn588Lys)	Short QT syndrome 1|short QT syndrome	Pathogenic(Last reviewed: Apr 1, 2005)	no assertion criteria provided	VCV000014436	7	150951629	14436	29475	rs104894021	GOF	0.706	0.038	0.256
KCNH2	588	N	K	NM_000238.4(KCNH2):c.1764C>A (p.Asn588Lys)	Short QT syndrome 1|short QT syndrome	Pathogenic(Last reviewed: Jan 6, 2004)	no assertion criteria provided	VCV000014437	7	150951629	14437	29476	rs104894021	GOF	0.706	0.038	0.256
KCNH2	588	N	K	NM_000238.4(KCNH2):c.1764C>A (p.Asn588Lys)	Short QT syndrome 1|short QT syndrome	Pathogenic(Last reviewed: Jan 6, 2004)	no assertion criteria provided	VCV000014437	7	150951629	14437	29476	rs104894021	GOF	0.706	0.038	0.256
KCNH2	589	E	Q	NM_000238.4(KCNH2):c.2785G>C (p.Glu929Gln)	Arrhythmia	Uncertain significance(Last reviewed: Mar 11, 2020)	criteria provided, single submitter	VCV000926191	7	150947786	926191	910708	rs1800967308	LOF	0.851	0.027	0.122
KCNH2	590	S	G	NM_000238.4(KCNH2):c.2788A>G (p.Ser930Gly)	Arrhythmia	Uncertain significance(Last reviewed: Nov 4, 2019)	criteria provided, single submitter	VCV000924230	7	150947783	924230	910707	rs794728400	LOF	0.86	0.035	0.105
KCNH2	590	S	R	NM_000238.4(KCNH2):c.2788A>C (p.Ser930Arg)	not provided	Uncertain significance(Last reviewed: May 14, 2014)	criteria provided, single submitter	VCV000200503	7	150947783	200503	197170	rs794728400	LOF	0.754	0.03	0.216
KCNH2	591	D	N	NM_000238.4(KCNH2):c.1771G>A (p.Asp591Asn)	Long QT syndrome|Long QT syndrome 2| Arrhythmia	Uncertain significance(Last reviewed: Aug 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000911854	7	150951622	911854	897669	rs376319070	LOF	0.702	0.07	0.227
KCNH2	592	S	P	NM_000238.4(KCNH2):c.2794T>C (p.Ser932Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 13, 2019)	criteria provided, single submitter	VCV000942612	7	150947777	942612	933764	rs1800966532	LOF	0.762	0.079	0.159
KCNH2	593	I	R	NM_000238.4(KCNH2):c.1778T>G (p.Ile593Arg)	not provided|Congenital long QT syndrome|Long QT syndrome 2	Pathogenic(Last reviewed: May 24, 2019)	criteria provided, single submitter	VCV000014423	7	150951615	14423	29462	rs28928904	LOF	0.868	0.033	0.099
KCNH2	597	S	N	NM_000238.4(KCNH2):c.2810G>A (p.Ser937Asn)	Arrhythmia|not provided|Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Dec 16, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067440	7	150947761	67440	78336	rs199473540	LOF	0.753	0.104	0.142
KCNH2	604	E	D	NM_000238.4(KCNH2):c.2832G>T (p.Glu944Asp)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Apr 5, 2021)	criteria provided, multiple submitters, no conflicts	VCV000526953	7	150947739	526953	522770	rs1232364499	LOF	0.833	0.067	0.1
KCNH2	605	G	C	NM_000238.4(KCNH2):c.2833G>T (p.Gly945Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 9, 2020)	criteria provided, single submitter	VCV001063652	7	150947738	1063652	1044803	NA	LOF	0.793	0.084	0.124
KCNH2	605	P	H	NM_000238.4(KCNH2):c.1814C>A (p.Pro605His)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 8,	criteria provided, single submitter	VCV000858913	7	150951579	858913	833337	rs199472938	LOF	0.768	0.075	0.157

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)											
KCNH2	606	P	L	NM_000238.4(KCNH2):c.2837C>T (p.Pro946Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 12, 2020)	criteria provided, single submitter	VCV001008413	7	150947734	1008413	992188	rs1800964240	LOF	0.799	0.083	0.118
KCNH2	608	R	H	NM_000238.4(KCNH2):c.2843G>A (p.Arg948His)	Congenital long QT syndrome|Arrhythmia|not specified|Long QT syndrome 2|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Mar 18, 2021)	criteria provided, conflicting interpretations	VCV000067443	7	150947728	67443	78339	rs199473011	LOF	0.861	0.035	0.105
KCNH2	608	R	S	NM_000238.4(KCNH2):c.2842C>A (p.Arg948Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067442	7	150947729	67442	78338	rs121912514	LOF	0.864	0.037	0.1
KCNH2	611	Y	H	NM_000238.4(KCNH2):c.1831T>C (p.Tyr611His)	Long QT syndrome 2|Congenital long QT syndrome	Pathogenic(Last reviewed: May 13, 2005)	no assertion criteria provided	VCV000029778	7	150951562	29778	38733	rs199472942	LOF	0.926	0.012	0.062
KCNH2	612	P	R	NM_000238.4(KCNH2):c.2855C>G (p.Pro952Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 9, 2020)	criteria provided, single submitter	VCV001036276	7	150947716	1036276	1027883	rs765920319	LOF	0.893	0.011	0.096
KCNH2	613	L	F	NM_000238.4(KCNH2):c.2857C>T (p.Leu953Phe)	Arrhythmia	Uncertain significance(Last reviewed: Dec 19, 2018)	criteria provided, single submitter	VCV000924479	7	150947714	924479	910701	rs1800963184	LOF	0.884	0.022	0.094
KCNH2	614	A	V	NM_000238.4(KCNH2):c.1841C>T (p.Ala614Val)	not provided|Long QT syndrome 2|Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Jan 13, 2022)	criteria provided, multiple submitters, no conflicts	VCV000029777	7	150951552	29777	38732	rs199472944	LOF	0.864	0.02	0.116
KCNH2	614	R	C	NM_000238.4(KCNH2):c.2860C>T (p.Arg954Cys)	not provided|Cardiovascular phenotype|Arrhythmia|SUDDEN INFANT DEATH SYNDROME|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Mar 3, 2021)	criteria provided, conflicting interpretations	VCV000067444	7	150947711	67444	78340	rs141401803	LOF	0.92	0.012	0.068
KCNH2	614	R	H	NM_000238.4(KCNH2):c.2861G>A (p.Arg954His)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Nov 24, 2019)	criteria provided, multiple submitters, no conflicts	VCV000456917	7	150947710	456917	457063	rs772977598	LOF	0.916	0.01	0.074
KCNH2	615	L	V	NM_000238.4(KCNH2):c.2863C>G (p.Leu955Val)	Long QT syndrome|Arrhythmia|not provided|Congenital long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Oct 6, 2020)	criteria provided, conflicting interpretations	VCV000067445	7	150947708	67445	78341	rs199473012	LOF	0.881	0.02	0.099
KCNH2	617	P	L	NM_000238.4(KCNH2):c.2870C>T (p.Pro957Leu)	Long QT syndrome 2	Uncertain significance	criteria provided, single submitter	VCV000979134	7	150947701	979134	967183	rs1800962566	LOF	0.896	0.013	0.09
KCNH2	619	S	C	NM_000238.4(KCNH2):c.2876C>G (p.Ser959Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 2, 2018)	criteria provided, single submitter	VCV000662474	7	150947695	662474	635890	rs1584844763	LOF	0.892	0.018	0.09
KCNH2	620	S	N	NM_000238.4(KCNH2):c.2879G>A (p.Ser960Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067447	7	150947692	67447	78343	rs199473013	LOF	0.886	0.01	0.104
KCNH2	623	P	T	NM_000238.4(KCNH2):c.2887C>A (p.Pro963Thr)	Congenital long QT syndrome|not provided|Arrhythmia	Uncertain significance(Last reviewed: Jan 13, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067449	7	150947684	67449	78345	rs199473014	LOF	0.925	0.009	0.067
KCNH2	624	P	A	NM_000238.4(KCNH2):c.2890C>G (p.Pro964Ala)	Long QT syndrome|Long QT syndrome 2	Uncertain significance(Last reviewed: Jun 16, 2020)	criteria provided, multiple submitters, no	VCV000359307	7	150947681	359307	310195	rs886062087	LOF	0.93	0.012	0.057

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNH2	624	P	L	NM_000238.4(KCNH2):c.2891C>T (p.Pro964Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 18, 2020)	criteria provided, single submitter	VCV001046584	7	150947680	1046584	1027882	rs886427965	LOF	0.923	0.015	0.062
KCNH2	624	P	R	NM_000238.4(KCNH2):c.2891C>G (p.Pro964Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV001021720	7	150947680	1021720	1007337	rs886427965	LOF	0.918	0.011	0.071
KCNH2	624	S	C	NM_000238.4(KCNH2):c.1870A>T (p.Ser624Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 19, 2019)	criteria provided, single submitter	VCV000644725	7	150951523	644725	635902	rs1584853632	LOF	0.921	0.014	0.065
KCNH2	625	G	R	NM_000238.4(KCNH2):c.2893G>A (p.Gly965Arg)	Long QT syndrome|not provided|Long QT syndrome 2	Uncertain significance(Last reviewed: Aug 6, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067450	7	150947678	67450	78346	rs199473015	LOF	0.915	0.014	0.071
KCNH2	627	P	L	NM_000238.4(KCNH2):c.2900C>T (p.Pro967Leu)	Long QT syndrome|Cardiovascular phenotype|not provided|Arrhythmia	Benign/Likely benign(Last reviewed: Nov 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067451	7	150947671	67451	78347	rs199473016	LOF	0.879	0.038	0.082
KCNH2	628	G	S	NM_000238.4(KCNH2):c.1882G>A (p.Gly628Ser)	Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 2|not provided	Pathogenic(Last reviewed: May 12, 2020)	criteria provided, multiple submitters, no conflicts	VCV000014427	7	150951511	14427	29466	rs121912507	LOF	0.906	0.017	0.077
KCNH2	628	P	A	NM_000238.4(KCNH2):c.2902C>G (p.Pro968Ala)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 3, 2020)	criteria provided, multiple submitters, no conflicts	VCV000237298	7	150947669	237298	239994	rs753788508	LOF	0.92	0.019	0.061
KCNH2	628	P	L	NM_000238.4(KCNH2):c.2903C>T (p.Pro968Leu)	Arrhythmia|Long QT syndrome 2|not provided	Uncertain significance(Last reviewed: Dec 19, 2018)	criteria provided, multiple submitters, no conflicts	VCV000067452	7	150947668	67452	78348	rs199473017	LOF	0.912	0.022	0.065
KCNH2	629	G	D	NM_000238.4(KCNH2):c.2906G>A (p.Gly969Asp)	Arrhythmia	Uncertain significance(Last reviewed: Jul 19, 2019)	criteria provided, single submitter	VCV000918705	7	150947665	918705	910697	rs749873669	LOF	0.891	0.017	0.092
KCNH2	629	G	V	NM_000238.4(KCNH2):c.2906G>T (p.Gly969Val)	Arrhythmia	Uncertain significance(Last reviewed: Jan 27, 2020)	criteria provided, single submitter	VCV000926647	7	150947665	926647	910696	rs749873669	LOF	0.888	0.023	0.09
KCNH2	630	G	R	NM_000238.4(KCNH2):c.2908G>A (p.Gly970Arg)	Arrhythmia	Uncertain significance(Last reviewed: Jan 17, 2020)	criteria provided, single submitter	VCV000926177	7	150947663	926177	910695	rs1800959537	LOF	0.821	0.021	0.158
KCNH2	631	E	D	NM_000238.4(KCNH2):c.2913G>T (p.Glu971Asp)	Arrhythmia	Uncertain significance(Last reviewed: Aug 10, 2020)	criteria provided, single submitter	VCV000926285	7	150947658	926285	910694	rs1347067242	LOF	0.844	0.024	0.132
KCNH2	631	S	F	NM_000238.4(KCNH2):c.1892C>T (p.Ser631Phe)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jul 24, 2019)	criteria provided, multiple submitters, no conflicts	VCV000456898	7	150951501	456898	456834	rs1554425720	LOF	0.85	0.049	0.101
KCNH2	637	C	W	NM_000238.4(KCNH2):c.2931C>G (p.Cys977Trp)	Arrhythmia	Uncertain significance(Last reviewed: Jan 15,	criteria provided, single submitter	VCV000919282	7	150947640	919282	910692	rs761585108	LOF	0.898	0.016	0.086

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)											
KCNH2	638	E	K	NM_000238.4(KCNH2):c.2932G>A (p.Glu978Lys)	Long QT syndrome|not specified|not provided|Arrhythmia	Likely benign(Last reviewed: Nov 15, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067453	7	150947639	67453	78349	rs141117135	LOF	0.847	0.013	0.14
KCNH2	638	E	Q	NM_000238.4(KCNH2):c.2932G>C (p.Glu978Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 30, 2020)	criteria provided, single submitter	VCV001042802	7	150947639	1042802	1027881	rs141117135	LOF	0.864	0.014	0.122
KCNH2	640	F	C	NM_000238.4(KCNH2):c.1919T>G (p.Phe640Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV001045742	7	150951474	1045742	1027887	rs1801154920	LOF	0.913	0.019	0.068
KCNH2	641	S	G	NM_000238.4(KCNH2):c.2941A>G (p.Ser981Gly)	Long QT syndrome|Cardiovascular phenotype|not specified|not provided|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Feb 19, 2021)	criteria provided, conflicting interpretations	VCV000067454	7	150947630	67454	78350	rs76649554	LOF	0.914	0.01	0.076
KCNH2	642	D	N	NM_000238.4(KCNH2):c.2944G>A (p.Asp982Asn)	Arrhythmia|Long QT syndrome 2|Long QT syndrome	Uncertain significance(Last reviewed: Apr 20, 2021)	criteria provided, multiple submitters, no conflicts	VCV000237299	7	150947627	237299	239993	rs569452580	LOF	0.923	0.011	0.066
KCNH2	643	T	I	NM_000238.4(KCNH2):c.2948C>T (p.Thr983Ile)	Long QT syndrome|not provided|Congenital long QT syndrome|not specified|Long QT syndrome 2|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Dec 17, 2021)	criteria provided, conflicting interpretations	VCV000067455	7	150947623	67455	78351	rs149955375	LOF	0.861	0.025	0.114
KCNH2	645	N	S	NM_000238.4(KCNH2):c.2954A>G (p.Asn985Ser)	not provided|Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Mar 16, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067456	7	150947617	67456	78352	rs199473541	LOF	0.908	0.009	0.083
KCNH2	646	P	T	NM_000238.4(KCNH2):c.2956C>A (p.Pro986Thr)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Nov 14, 2019)	criteria provided, multiple submitters, no conflicts	VCV000640120	7	150947615	640120	635888	rs1261949255	LOF	0.918	0.016	0.066
KCNH2	650	A	T	NM_000238.4(KCNH2):c.2968G>A (p.Ala990Thr)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Nov 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000968306	7	150947512	968306	955092	rs754362735	LOF	0.874	0.023	0.103
KCNH2	656	N	I	NM_000238.4(KCNH2):c.2987A>T (p.Asn996Ile)	Congenital long QT syndrome|Long QT syndrome 2|not provided	Pathogenic/Likely pathogenic(Last reviewed: Dec 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067458	7	150947493	67458	78354	rs199473018	LOF	0.887	0.015	0.098
KCNH2	657	G	C	NM_000238.4(KCNH2):c.1968_1969delinsTT (p.Gly657Cys)	Long QT syndrome	Likely pathogenic(Last reviewed: Nov 6, 2019)	criteria provided, single submitter	VCV000943187	7	150951097 - 150951098	943187	933771	rs1801136288	LOF	0.931	0.01	0.059
KCNH2	659	S	N	NM_000238.4(KCNH2):c.2996G>A (p.Ser999Asn)	Arrhythmia|not provided	Uncertain significance(Last reviewed: Dec 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000919130	7	150947484	919130	910688	rs974699160	LOF	0.902	0.012	0.086
KCNH2	667	R	C	NM_000238.4(KCNH2):c.3019C>T (p.Arg1007Cys)	Arrhythmia	Uncertain significance(Last reviewed: Jan 9, 2020)	criteria provided, single submitter	VCV000918739	7	150947461	918739	910686	rs1224504959	LOF	0.824	0.088	0.089

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	671	E	G	NM_000238.4(KCNH2):c.3032A>G (p.Glu1011Gly)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Aug 11, 2020)	criteria provided, multiple submitters, no conflicts	VCV000922211	7	150947448	922211	910684	rs1800945318	LOF	0.882	0.028	0.09
KCNH2	679	R	W	NM_000238.4(KCNH2):c.2035C>T (p.Arg679Trp)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Aug 5, 2021)	criteria provided, multiple submitters, no conflicts	VCV000581266	7	150951031	581266	566571	rs79624542	LOF	0.846	0.05	0.104
KCNH2	681	S	G	NM_000238.4(KCNH2):c.3061A>G (p.Ser1021Gly)	Long QT syndrome	Uncertain significance(Last reviewed: May 2, 2018)	criteria provided, single submitter	VCV000572915	7	150947419	572915	566568	rs1563146032	LOF	0.876	0.049	0.075
KCNH2	690	P	L	NM_000238.4(KCNH2):c.3089C>T (p.Pro1030Leu)	Arrhythmia|Long QT syndrome|not provided	Uncertain significance(Last reviewed: Dec 21, 2021)	criteria provided, multiple submitters, no conflicts	VCV000847541	7	150947391	847541	833317	rs904066540	LOF	0.832	0.073	0.094
KCNH2	690	P	R	NM_000238.4(KCNH2):c.3089C>G (p.Pro1030Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 22, 2020)	criteria provided, single submitter	VCV000837323	7	150947391	837323	833318	rs904066540	LOF	0.789	0.041	0.17
KCNH2	691	G	D	NM_000238.4(KCNH2):c.3092G>A (p.Gly1031Asp)	Arrhythmia	Uncertain significance(Last reviewed: Nov 9, 2018)	criteria provided, single submitter	VCV000919480	7	150947388	919480	910679	rs1391531244	LOF	0.847	0.032	0.121
KCNH2	695	R	Q	NM_000238.4(KCNH2):c.3104G>A (p.Arg1035Gln)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jun 7, 2021)	criteria provided, multiple submitters, no conflicts	VCV000456922	7	150947376	456922	456466	rs761933920	LOF	0.852	0.056	0.092
KCNH2	698	E	K	NM_000238.4(KCNH2):c.2092G>A (p.Glu698Lys)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Mar 16, 2021)	criteria provided, multiple submitters, no conflicts	VCV000945957	7	150950974	945957	924754	rs753966347	LOF	0.831	0.026	0.143
KCNH2	706	Q	H	NM_000238.4(KCNH2):c.3138G>T (p.Gln1046His)	Arrhythmia	Uncertain significance(Last reviewed: Jan 27, 2020)	criteria provided, single submitter	VCV000926330	7	150947342	926330	910676	rs1367829679	LOF	0.86	0.032	0.108
KCNH2	706	Q	R	NM_000238.4(KCNH2):c.3137A>G (p.Gln1046Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 21, 2019)	criteria provided, single submitter	VCV000958659	7	150947343	958659	945517	rs1800934521	LOF	0.83	0.027	0.144
KCNH2	710	G	S	NM_000238.4(KCNH2):c.2128G>A (p.Gly710Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 21, 2019)	criteria provided, single submitter	VCV000664978	7	150950938	664978	635899	rs1584851956	LOF	0.797	0.058	0.145
KCNH2	714	T	I	NM_000238.4(KCNH2):c.3161C>T (p.Thr1054Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 27, 2017)	criteria provided, single submitter	VCV000527001	7	150947046	527001	522442	rs1554423904	LOF	0.829	0.078	0.093
KCNH2	715	R	W	NM_000238.4(KCNH2):c.3163C>T (p.Arg1055Trp)	Long QT syndrome|Long QT syndrome 2	Uncertain significance(Last reviewed: Jan 15, 2020)	criteria provided, multiple submitters, no conflicts	VCV000405340	7	150947044	405340	395666	rs541259035	LOF	0.852	0.052	0.095
KCNH2	731	M	V	NM_000238.4(KCNH2):c.3211A>G (p.Met1071Val)	Arrhythmia	Uncertain significance(Last reviewed: Oct 11, 2018)	criteria provided, single submitter	VCV000629473	7	150946996	629473	617337	rs555797304	LOF	0.88	0.035	0.084

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	732	T	M	NM_000238.4(KCNH2):c.3215C>T (p.Thr1072Met)	not provided|Long QT syndrome 2|Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Aug 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000359303	7	150946992	359303	305381	rs781624566	LOF	0.872	0.03	0.098
KCNH2	752	R	Q	NM_000238.4(KCNH2):c.2255G>A (p.Arg752Gln)	Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 2|not provided	Likely pathogenic(Last reviewed: Jul 29, 2021)	criteria provided, multiple submitters, no conflicts	VCV000014435	7	150950311	14435	29474	rs121912512	LOF	0.871	0.031	0.098
KCNH2	754	L	M	NM_000238.4(KCNH2):c.3279_3280delinsAA (p.Leu1094Met)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 11, 2019)	criteria provided, single submitter	VCV000962100	7	150946927 - 150946928	962100	945515	rs1800911944	LOF	0.881	0.035	0.084
KCNH2	761	P	S	NM_000238.4(KCNH2):c.3301C>T (p.Pro1101Ser)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Sep 10, 2018)	criteria provided, multiple submitters, no conflicts	VCV000456928	7	150946906	456928	456799	rs1347039291	LOF	0.857	0.071	0.072
KCNH2	764	P	R	NM_000238.4(KCNH2):c.2291C>G (p.Pro764Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 19, 2019)	criteria provided, single submitter	VCV000955346	7	150950275	955346	945521	rs878853773	LOF	0.704	0.07	0.226
KCNH2	765	L	F	NM_000238.4(KCNH2):c.3315G>C (p.Leu1105Phe)	Arrhythmia	Uncertain significance(Last reviewed: Nov 20, 2018)	criteria provided, single submitter	VCV000926697	7	150946892	926697	910668	rs1800909845	LOF	0.837	0.071	0.091
KCNH2	776	A	T	NM_000238.4(KCNH2):c.3346G>A (p.Ala1116Thr)	Arrhythmia	Uncertain significance(Last reviewed: Jan 14, 2020)	criteria provided, single submitter	VCV000918904	7	150945499	918904	910666	rs1800858723	LOF	0.863	0.051	0.086
KCNH2	777	C	F	NM_000238.4(KCNH2):c.3350G>T (p.Cys1117Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 29, 2018)	criteria provided, single submitter	VCV000582288	7	150945495	582288	564148	rs730880119	LOF	0.786	0.099	0.115
KCNH2	784	R	W	NM_000238.4(KCNH2):c.2350C>T (p.Arg784Trp)	Long QT syndrome 2|Long QT syndrome 2, acquired, susceptibility to|Brugada syndrome| Congenital long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Feb 22, 2021)	criteria provided, conflicting interpretations	VCV000014433	7	150950216	14433	29472	rs12720441	LOF	0.855	0.051	0.094
KCNH2	793	D	N	NM_000238.4(KCNH2):c.2377G>A (p.Asp793Asn)	Arrhythmia	Uncertain significance(Last reviewed: Feb 19, 2020)	criteria provided, single submitter	VCV000921995	7	150950189	921995	910737	rs781352799	LOF	0.802	0.042	0.156
KCNH2	795	R	C	NM_000238.4(KCNH2):c.3403C>T (p.Arg1135Cys)	not provided|Arrhythmia	Uncertain significance(Last reviewed: Oct 18, 2019)	criteria provided, multiple submitters, no conflicts	VCV000920427	7	150945442	920427	910661	rs781369850	LOF	0.872	0.051	0.078
KCNH2	796	V	L	NM_000238.4(KCNH2):c.2386G>T (p.Val796Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 22, 2020)	criteria provided, single submitter	VCV000940061	7	150950180	940061	933767	rs143335921	LOF	0.869	0.049	0.082
KCNH2	797	S	Y	NM_000238.4(KCNH2):c.3410C>A (p.Ser1137Tyr)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 27, 2020)	criteria provided, single submitter	VCV000966027	7	150945435	966027	955090	rs1413026629	LOF	0.878	0.045	0.077
KCNH2	800	G	A	NM_000238.4(KCNH2):c.2398+1G>C	not provided|Long QT syndrome 2	Pathogenic(Last reviewed: Jun 22, 2018)	criteria provided, single submitter	VCV000200444	7	150950167	200444	197212	rs794728391	LOF	0.814	0.078	0.108
KCNH2	800	G	V	NM_000238.4(KCNH2):c.2398+1G>T	not provided|Long QT syndrome	Pathogenic/Likely pathogenic(Last	criteria provided,	VCV000372377	7	150950167	372377	359805	rs794728391	LOF	0.808	0.088	0.105

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Apr 28, 2017)	multiple submitters, no conflicts										
KCNH2	801	M	I	NM_000238.4(KCNH2):c.2398+5G>T	Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jan 7, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200446	7	150950163	200446	197211	rs1554425149	LOF	0.77	0.102	0.128
KCNH2	801	M	T	NM_000238.4(KCNH2):c.2398+4T>C	not specified|Arrhythmia	Likely benign(Last reviewed: Mar 11, 2019)	criteria provided, multiple submitters, no conflicts	VCV000379294	7	150950164	379294	370976	rs1057520559	LOF	0.804	0.069	0.127
KCNH2	801	M	V	NM_000238.4(KCNH2):c.2398+3A>G	Long QT syndrome	Uncertain significance(Last reviewed: Jun 12, 2017)	criteria provided, single submitter	VCV000523701	7	150950165	523701	514350	rs1554425151	LOF	0.781	0.092	0.127
KCNH2	805	A	V	NM_000238.4(KCNH2):c.2398+16C>T	not provided|not specified|Arrhythmia	Benign/Likely benign(Last reviewed: Apr 27, 2019)	criteria provided, multiple submitters, no conflicts	VCV000191543	7	150950152	191543	189260	rs201873431	LOF	0.869	0.051	0.08
KCNH2	808	G	V	NM_000238.4(KCNH2):c.2398+25G>T	Arrhythmia|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Mar 16, 2021)	criteria provided, conflicting interpretations	VCV000627667	7	150950143	627667	619249	rs761167679	LOF	0.818	0.102	0.081
KCNH2	810	L	M	NM_000238.4(KCNH2):c.3448C>A (p.Leu1150Met)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jul 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000456931	7	150945397	456931	456462	rs754883792	LOF	0.795	0.128	0.077
KCNH2	811	H	Q	NM_000238.4(KCNH2):c.3453C>A (p.His1151Gln)	Arrhythmia	Uncertain significance(Last reviewed: Oct 11, 2018)	criteria provided, single submitter	VCV000629472	7	150945392	629472	617334	rs531426751	LOF	0.837	0.065	0.098
KCNH2	815	S	L	NM_000238.4(KCNH2):c.3464C>T (p.Ser1155Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 16, 2018)	criteria provided, single submitter	VCV000580441	7	150945381	580441	566556	rs867402580	Neutral	0.519	0.302	0.179
KCNH2	818	G	A	NM_000238.4(KCNH2):c.3473G>C (p.Gly1158Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 10, 2019)	criteria provided, single submitter	VCV000939464	7	150945372	939464	933762	rs1800852263	LOF	0.772	0.087	0.141
KCNH2	818	S	L	NM_000238.4(KCNH2):c.2453C>T (p.Ser818Leu)	Long QT syndrome 2|Congenital long QT syndrome|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Sep 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000014432	7	150948995	14432	29471	rs121912510	LOF	0.747	0.129	0.123
KCNH2	819	A	E	NM_000238.4(KCNH2):c.2398+58C>A	not specified	Uncertain significance(Last reviewed: Aug 5, 2013)	criteria provided, single submitter	VCV000200233	7	150950110	200233	197209	rs774109163	LOF	0.745	0.091	0.164
KCNH2	819	A	S	NM_000238.4(KCNH2):c.2398+57G>T	not provided	Uncertain significance(Last reviewed: Jun 22, 2012)	criteria provided, single submitter	VCV000200448	7	150950111	200448	197210	rs779355544	LOF	0.729	0.129	0.142
KCNH2	819	A	V	NM_000238.4(KCNH2):c.2398+58C>T	not specified	Likely benign(Last reviewed: Sep 4, 2015)	criteria provided, single submitter	VCV000200235	7	150950110	200235	197208	rs774109163	Neutral	0.712	0.173	0.114
KCNH2	822	V	M	NM_000238.4(KCNH2):c.2464G>A (p.Val822Met)	Long QT syndrome|Long QT syndrome 1| Cardiovascular phenotype|Congenital long QT	Pathogenic(Last reviewed: Jul 23,	criteria provided,	VCV000014424	7	150948984	14424	29463	rs121912506	LOF	0.856	0.05	0.095

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					syndrome|not provided|Long QT syndrome 2	2020)	multiple submitters, no conflicts										
KCNH2	851	S	G	NM_000238.4(KCNH2):c.2398+153A>G	not provided	Uncertain significance(Last reviewed: Dec 19, 2016)	criteria provided, single submitter	VCV000200239	7	150950015	200239	197206	rs794728345	LOF	0.816	0.08	0.104
KCNH2	851	S	R	NM_000238.4(KCNH2):c.2398+155C>A	Arrhythmia	Benign(Last reviewed: Apr 23, 2018)	criteria provided, single submitter	VCV000630018	7	150950013	630018	619281	rs536958259	LOF	0.761	0.06	0.18
KCNH2	852	G	R	NM_000238.4(KCNH2):c.2398+156G>A	Arrhythmia|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Apr 6, 2021)	criteria provided, conflicting interpretations	VCV000191471	7	150950012	191471	189259	rs139247073	LOF	0.865	0.02	0.114
KCNH2	856	L	V	NM_000238.4(KCNH2):c.2566C>G (p.Leu856Val)	Arrhythmia	Uncertain significance(Last reviewed: Oct 5, 2018)	criteria provided, single submitter	VCV000629380	7	150948882	629380	617346	rs1563150352	LOF	0.866	0.05	0.083
KCNH2	859	T	M	NM_000238.4(KCNH2):c.2398+178C>T	Arrhythmia|not provided	Benign/Likely benign(Last reviewed: Oct 16, 2018)	criteria provided, multiple submitters, no conflicts	VCV000191542	7	150949990	191542	189258	rs41314366	LOF	0.843	0.076	0.081
KCNH2	861	N	I	NM_000238.4(KCNH2):c.2582A>T (p.Asn861Ile)	Congenital long QT syndrome|Long QT syndrome 2/5|Long QT syndrome 2	Pathogenic(Last reviewed: Apr 20, 2004)	no assertion criteria provided	VCV000014440	7	150948866	14440	29479	rs121912513	Neutral	0.571	0.282	0.147
KCNH2	865	T	I	NM_000238.4(KCNH2):c.2594C>T (p.Thr865Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 6, 2019)	criteria provided, single submitter	VCV000957208	7	150948542	957208	945520	rs768059097	Neutral	0.303	0.551	0.146
KCNH2	870	H	Y	NM_000238.4(KCNH2):c.2398+210C>T	Arrhythmia	Likely benign(Last reviewed: Jul 2, 2019)	criteria provided, single submitter	VCV000924676	7	150949958	924676	915419	rs1264796799	GOF	0.322	0.296	0.382
KCNH2	871	S	C	NM_000238.4(KCNH2):c.2612C>G (p.Ser871Cys)	Arrhythmia	Uncertain significance(Last reviewed: Nov 20, 2018)	criteria provided, single submitter	VCV000921315	7	150948524	921315	910728	rs1177213407	Neutral	0.347	0.392	0.261
KCNH2	876	E	K	NM_000238.4(KCNH2):c.2626G>A (p.Glu876Lys)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: May 3, 2017)	criteria provided, single submitter	VCV000456914	7	150948510	456914	456490	rs1554424688	GOF	0.36	0.174	0.465
KCNH2	878	R	C	NM_000238.4(KCNH2):c.2398+234C>T	Arrhythmia|not specified	Likely benign(Last reviewed: Apr 15, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200243	7	150949934	200243	197204	rs370393086	Neutral	0.348	0.382	0.27
KCNH2	881	F	I	NM_000238.4(KCNH2):c.2641T>A (p.Phe881Ile)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 12, 2020)	criteria provided, single submitter	VCV000855339	7	150948495	855339	833332	rs1197425750	Neutral	0.408	0.381	0.211
KCNH2	883	R	W	NM_000238.4(KCNH2):c.2647C>T (p.Arg883Trp)	Long QT syndrome|not provided|Arrhythmia	Uncertain significance(Last reviewed: Oct 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000925591	7	150948489	925591	910725	rs201765446	Neutral	0.303	0.439	0.258
KCNH2	887	R	L	NM_000238.4(KCNH2):c.2660G>T (p.Arg887Leu)	Arrhythmia	Uncertain significance(Last reviewed: Aug 8, 2019)	criteria provided, single submitter	VCV000628973	7	150948476	628973	617345	rs199473432	Neutral	0.288	0.47	0.242
KCNH2	895	T	K	NM_000238.4(KCNH2):c.2684C>A (p.Thr895Lys)	Arrhythmia	Uncertain significance(Last	criteria provided, single	VCV000927873	7	150948452	927873	910720	rs199473434	GOF	0.357	0.226	0.417

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Dec 23, 2020)	submitter										
KCNH2	901	Q	R	NM_000238.4(KCNH2):c.2702A>G (p.Gln901Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 13, 2019)	criteria provided, single submitter	VCV000851829	7	150947869	851829	833329	rs1800975753	GOF	0.376	0.173	0.451
KCNH2	903	G	R	NM_000238.4(KCNH2):c.2707G>C (p.Gly903Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV000960253	7	150947864	960253	945518	rs199473669	GOF	0.376	0.176	0.448
KCNH2	907	A	S	NM_000238.4(KCNH2):c.2719G>T (p.Ala907Ser)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jul 14, 2020)	criteria provided, multiple submitters, no conflicts	VCV000834753	7	150947852	834753	833328	rs752686806	GOF	0.401	0.234	0.365
KCNH2	916	G	R	NM_000238.4(KCNH2):c.2746G>A (p.Gly916Arg)	Arrhythmia	Uncertain significance(Last reviewed: Jan 1, 2020)	criteria provided, single submitter	VCV000924836	7	150947825	924836	910713	rs1800972199	GOF	0.386	0.177	0.436
KCNH2	924	G	A	NM_000238.4(KCNH2):c.2769_2771delinsAGC (p.Gly924Ala)	Arrhythmia	Uncertain significance(Last reviewed: Dec 18, 2019)	criteria provided, single submitter	VCV000920801	7	150947800 - 150947802	920801	910710	rs1800969882	GOF	0.347	0.188	0.465
KCNH2	927	W	C	NM_000238.4(KCNH2):c.2781G>T (p.Trp927Cys)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Mar 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV001037100	7	150947790	1037100	1027885	rs866768546	Neutral	0.557	0.307	0.136
KCNH2	931	P	L	NM_000238.4(KCNH2):c.2792C>T (p.Pro931Leu)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jan 5, 2021)	criteria provided, multiple submitters, no conflicts	VCV000654837	7	150947779	654837	635891	rs749759697	Neutral	0.399	0.365	0.236
KCNH2	931	P	R	NM_000238.4(KCNH2):c.2792C>G (p.Pro931Arg)	Arrhythmia	Uncertain significance(Last reviewed: Oct 9, 2019)	criteria provided, single submitter	VCV000928265	7	150947779	928265	910706	rs749759697	GOF	0.426	0.249	0.325
KCNH2	932	S	F	NM_000238.4(KCNH2):c.2795C>T (p.Ser932Phe)	Arrhythmia	Uncertain significance(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV000925842	7	150947776	925842	910705	rs1800966447	Neutral	0.407	0.313	0.28
KCNH2	935	P	A	NM_000238.4(KCNH2):c.2803C>G (p.Pro935Ala)	Arrhythmia	Uncertain significance(Last reviewed: May 23, 2019)	criteria provided, single submitter	VCV000922520	7	150947768	922520	910704	rs1419849010	GOF	0.377	0.233	0.389
KCNH2	940	S	I	NM_000238.4(KCNH2):c.2819G>T (p.Ser940Ile)	Arrhythmia	Uncertain significance(Last reviewed: Oct 18, 2019)	criteria provided, single submitter	VCV000923239	7	150947752	923239	910703	rs1800965243	Neutral	0.366	0.337	0.296
KCNH2	944	E	D	NM_000238.4(KCNH2):c.2832G>C (p.Glu944Asp)	Arrhythmia	Uncertain significance(Last reviewed: Jul 22, 2018)	criteria provided, single submitter	VCV000628923	7	150947739	628923	617342	rs1232364499	GOF	0.433	0.221	0.347
KCNH2	948	R	C	NM_000238.4(KCNH2):c.2842C>T (p.Arg948Cys)	Long QT syndrome|Long QT syndrome 1/2, digenic|Congenital long QT syndrome| Arrhythmia|not provided	Uncertain significance(Last reviewed: Dec 9, 2021)	criteria provided, multiple submitters, no conflicts	VCV000014441	7	150947729	14441	29480	rs121912514	Neutral	0.447	0.342	0.211
KCNH2	951	S	R	NM_000238.4(KCNH2):c.2853C>A (p.Ser951Arg)	Long QT syndrome	Uncertain significance(Last reviewed: May 9, 2019)	criteria provided, single submitter	VCV000840645	7	150947718	840645	833325	rs1443792370	GOF	0.357	0.176	0.467

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH2	952	P	L	NM_000238.4(KCNH2):c.2855C>T (p.Pro952Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 24, 2019)	criteria provided, single submitter	VCV000862901	7	150947716	862901	833324	rs765920319	Neutral	0.39	0.344	0.266
KCNH2	952	P	S	NM_000238.4(KCNH2):c.2854C>T (p.Pro952Ser)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Dec 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000919402	7	150947717	919402	910702	rs753735368	GOF	0.392	0.218	0.39
KCNH2	959	S	F	NM_000238.4(KCNH2):c.2876C>T (p.Ser959Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 14, 2018)	criteria provided, single submitter	VCV000657772	7	150947695	657772	635889	rs1584844763	Neutral	0.383	0.343	0.274
KCNH2	959	S	T	NM_000238.4(KCNH2):c.2875T>A (p.Ser959Thr)	Arrhythmia	Uncertain significance(Last reviewed: Nov 16, 2018)	criteria provided, single submitter	VCV000921152	7	150947696	921152	910700	rs1800962302	GOF	0.382	0.191	0.427
KCNH2	967	P	S	NM_000238.4(KCNH2):c.2899C>T (p.Pro967Ser)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 22, 2019)	criteria provided, multiple submitters, no conflicts	VCV000923330	7	150947672	923330	910698	rs1057450060	GOF	0.389	0.188	0.423
KCNH2	976	D	N	NM_000238.4(KCNH2):c.2926G>A (p.Asp976Asn)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Sep 11, 2019)	criteria provided, multiple submitters, no conflicts	VCV000928384	7	150947645	928384	910693	rs1800958394	GOF	0.412	0.179	0.409
KCNH2	977	C	F	NM_000238.4(KCNH2):c.2930G>T (p.Cys977Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 7, 2016)	criteria provided, single submitter	VCV000405354	7	150947641	405354	395682	rs767252448	Neutral	0.51	0.326	0.164
KCNH2	988	S	A	NM_000238.4(KCNH2):c.2962T>G (p.Ser988Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 13, 2020)	criteria provided, single submitter	VCV000859417	7	150947609	859417	833322	rs747237810	GOF	0.363	0.288	0.348
KCNH2	1001	W	C	NM_000238.4(KCNH2):c.3003G>C (p.Trp1001Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 27, 2019)	criteria provided, single submitter	VCV000857986	7	150947477	857986	833321	rs121912509	Neutral	0.474	0.377	0.15
KCNH2	1003	D	N	NM_000238.4(KCNH2):c.3007G>A (p.Asp1003Asn)	not specified|Long QT syndrome	Uncertain significance(Last reviewed: Mar 11, 2020)	criteria provided, multiple submitters, no conflicts	VCV000373622	7	150947473	373622	359804	rs794728402	GOF	0.376	0.175	0.449
KCNH2	1003	D	Y	NM_000238.4(KCNH2):c.3007G>T (p.Asp1003Tyr)	not provided	Likely pathogenic(Last reviewed: Jul 16, 2014)	criteria provided, single submitter	VCV000200516	7	150947473	200516	197155	rs794728402	GOF	0.312	0.3	0.388
KCNH2	1005	R	Q	NM_000238.4(KCNH2):c.3014G>A (p.Arg1005Gln)	Congenital long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Dec 6, 2019)	criteria provided, single submitter	VCV000067461	7	150947466	67461	78357	rs199473019	GOF	0.369	0.218	0.413
KCNH2	1005	R	W	NM_000238.4(KCNH2):c.3013C>T (p.Arg1005Trp)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 13, 2020)	criteria provided, single submitter	VCV001019746	7	150947467	1019746	1007336	rs1016101226	Neutral	0.323	0.43	0.247
KCNH2	1007	R	H	NM_000238.4(KCNH2):c.3020G>A (p.Arg1007His)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome	Uncertain significance(Last reviewed: Apr 20, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067463	7	150947460	67463	78359	rs199473542	GOF	0.355	0.283	0.361
KCNH2	1014	R	Q	NM_000238.4(KCNH2):c.3041G>A	Arrhythmia|not provided|Long QT syndrome	Uncertain	criteria	VCV000922413	7	150947439	922413	910683	rs932335536	GOF	0.389	0.217	0.393

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Arg1014Gln)		significance(Last reviewed: Apr 8, 2020)	provided, multiple submitters, no conflicts										
KCNH2	1017	A	S	NM_000238.4(KCNH2):c.3049G>T (p.Ala1017Ser)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Dec 8, 2021)	criteria provided, multiple submitters, no conflicts	VCV000565852	7	150947431	565852	561689	rs950543448	GOF	0.378	0.194	0.427
KCNH2	1018	P	A	NM_000238.4(KCNH2):c.3052C>G (p.Pro1018Ala)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Feb 17, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200520	7	150947428	200520	197152	rs41313764	GOF	0.346	0.244	0.41
KCNH2	1018	P	L	NM_000238.4(KCNH2):c.3053C>T (p.Pro1018Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 17, 2018)	criteria provided, single submitter	VCV000651422	7	150947427	651422	635887	rs866657878	Neutral	0.327	0.332	0.341
KCNH2	1020	P	S	NM_000238.4(KCNH2):c.3058C>T (p.Pro1020Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 14, 2016)	criteria provided, single submitter	VCV000359306	7	150947422	359306	302189	rs41307274	GOF	0.35	0.214	0.436
KCNH2	1022	L	P	NM_000238.4(KCNH2):c.3065T>C (p.Leu1022Pro)	Arrhythmia	Uncertain significance(Last reviewed: Mar 9, 2021)	criteria provided, single submitter	VCV001332320	7	150947415	1332320	1323039	NA	Neutral	0.531	0.274	0.195
KCNH2	1024	N	Y	NM_000238.4(KCNH2):c.3070A>T (p.Asn1024Tyr)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV001022039	7	150947410	1022039	1007335	rs1800941962	GOF	0.304	0.282	0.415
KCNH2	1027	L	I	NM_000238.4(KCNH2):c.3079C>A (p.Leu1027Ile)	Arrhythmia|Cardiac arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Oct 8, 2020)	criteria provided, conflicting interpretations	VCV000200254	7	150947401	200254	197150	rs794728346	Neutral	0.498	0.326	0.176
KCNH2	1032	R	P	NM_000238.4(KCNH2):c.3095G>C (p.Arg1032Pro)	Arrhythmia	Uncertain significance(Last reviewed: Nov 2, 2020)	criteria provided, single submitter	VCV001171150	7	150947385	1171150	1160689	NA	GOF	0.361	0.275	0.364
KCNH2	1032	R	Q	NM_000238.4(KCNH2):c.3095G>A (p.Arg1032Gln)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome|not provided|Long QT syndrome 2	Uncertain significance(Last reviewed: Nov 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067466	7	150947385	67466	78362	rs199473020	GOF	0.365	0.221	0.414
KCNH2	1032	R	W	NM_000238.4(KCNH2):c.3094C>T (p.Arg1032Trp)	Arrhythmia|not provided|Long QT syndrome| Long QT syndrome 2	Uncertain significance(Last reviewed: Feb 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200522	7	150947386	200522	197149	rs373394254	Neutral	0.312	0.443	0.246
KCNH2	1033	R	L	NM_000238.4(KCNH2):c.3098G>T (p.Arg1033Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, single submitter	VCV001059933	7	150947382	1059933	1044802	NA	Neutral	0.319	0.377	0.303
KCNH2	1033	R	Q	NM_000238.4(KCNH2):c.3098G>A (p.Arg1033Gln)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Apr 19, 2021)	criteria provided, multiple submitters, no conflicts	VCV000456920	7	150947382	456920	456807	rs750858069	GOF	0.369	0.219	0.411
KCNH2	1033	R	W	NM_000238.4(KCNH2):c.3097C>T (p.Arg1033Trp)	Congenital long QT syndrome|Arrhythmia|not provided|Long QT syndrome|Long QT syndrome 1	Uncertain significance(Last reviewed: Jul 27, 2021)	criteria provided, multiple submitters, no	VCV000067467	7	150947383	67467	78363	rs199473021	Neutral	0.317	0.437	0.247

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNH2	1035	R	W	NM_000238.4(KCNH2):c.3103C>T (p.Arg1035Trp)	Long QT syndrome|not provided|Arrhythmia	Uncertain significance(Last reviewed: Jan 20, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067468	7	150947377	67468	78364	rs199473543	Neutral	0.332	0.433	0.235
KCNH2	1036	G	D	NM_000238.4(KCNH2):c.3107G>A (p.Gly1036Asp)	Long QT syndrome|Congenital long QT syndrome|Arrhythmia|not provided|not specified	Uncertain significance(Last reviewed: Nov 22, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067469	7	150947373	67469	78365	rs199473022	GOF	0.467	0.204	0.329
KCNH2	1037	D	N	NM_000238.4(KCNH2):c.3109G>A (p.Asp1037Asn)	Long QT syndrome|Arrhythmia|not provided	Uncertain significance(Last reviewed: Mar 24, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067470	7	150947371	67470	78366	rs199473023	GOF	0.406	0.171	0.423
KCNH2	1038	V	L	NM_000238.4(KCNH2):c.3112G>T (p.Val1038Leu)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Mar 12, 2019)	criteria provided, multiple submitters, no conflicts	VCV000405360	7	150947368	405360	395853	rs199473544	Neutral	0.455	0.383	0.163
KCNH2	1038	V	M	NM_000238.4(KCNH2):c.3112G>A (p.Val1038Met)	Arrhythmia|not specified|not provided|Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 2	Conflicting interpretations of pathogenicity(Last reviewed: Dec 19, 2021)	criteria provided, conflicting interpretations	VCV000067471	7	150947368	67471	78367	rs199473544	Neutral	0.444	0.394	0.162
KCNH2	1040	S	G	NM_000238.4(KCNH2):c.3118A>G (p.Ser1040Gly)	Long QT syndrome|Arrhythmia|SUDDEN INFANT DEATH SYNDROME	Uncertain significance(Last reviewed: Apr 5, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067472	7	150947362	67472	78368	rs199473024	GOF	0.418	0.259	0.323
KCNH2	1040	S	N	NM_000238.4(KCNH2):c.3119G>A (p.Ser1040Asn)	not provided	Uncertain significance(Last reviewed: Jul 29, 2014)	criteria provided, single submitter	VCV000200526	7	150947361	200526	197141	rs794728404	GOF	0.418	0.203	0.38
KCNH2	1042	L	P	NM_000238.4(KCNH2):c.3125T>C (p.Leu1042Pro)	not provided|Cardiovascular phenotype	Conflicting interpretations of pathogenicity(Last reviewed: Aug 19, 2016)	criteria provided, conflicting interpretations	VCV000264155	7	150947355	264155	258465	rs886039064	Neutral	0.633	0.227	0.14
KCNH2	1042	L	V	NM_000238.4(KCNH2):c.3124C>G (p.Leu1042Val)	Hypertrophic cardiomyopathy	Uncertain significance(Last reviewed: Apr 30, 2017)	criteria provided, single submitter	VCV000427971	7	150947356	427971	419273	rs1346047270	Neutral	0.54	0.319	0.141
KCNH2	1045	L	F	NM_000238.4(KCNH2):c.3133C>T (p.Leu1045Phe)	Arrhythmogenic right ventricular cardiomyopathy|Primary dilated cardiomyopathy|Congenital long QT syndrome|Long QT syndrome|Arrhythmia	Benign(Last reviewed: Jul 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067473	7	150947347	67473	78369	rs199473025	Neutral	0.527	0.351	0.122
KCNH2	1045	L	R	NM_000238.4(KCNH2):c.3134T>G (p.Leu1045Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 7, 2020)	criteria provided, single submitter	VCV001025586	7	150947346	1025586	1007333	rs1800934753	Neutral	0.665	0.17	0.166
KCNH2	1047	R	C	NM_000238.4(KCNH2):c.3139C>T (p.Arg1047Cys)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000161255	7	150947341	161255	171106	rs377095107	Neutral	0.329	0.446	0.224
KCNH2	1047	R	H	NM_000238.4(KCNH2):c.3140G>A (p.Arg1047His)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 5, 2020)	criteria provided, multiple submitters, no	VCV000628410	7	150947340	628410	617339	rs36210421	GOF	0.371	0.313	0.316

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNH2	1047	R	L	NM_000238.4(KCNH2):c.3140G>T (p.Arg1047Leu)	Long QT syndrome|Long QT syndrome 2| Cardiovascular phenotype|Arrhythmia|Sudden unexplained death|not specified|Torsades de pointes|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 5, 2021)	criteria provided, conflicting interpretations	VCV000036429	7	150947340	36429	45092	rs36210421	Neutral	0.319	0.432	0.25
KCNH2	1049	L	P	NM_000238.4(KCNH2):c.3146T>C (p.Leu1049Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067474	7	150947334	67474	78370	rs199473026	Neutral	0.649	0.217	0.134
KCNH2	1051	R	S	NM_000238.4(KCNH2):c.3153G>T (p.Arg1051Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 6, 2019)	criteria provided, single submitter	VCV000952274	7	150947054	952274	924751	rs1800920453	GOF	0.398	0.288	0.314
KCNH2	1055	R	Q	NM_000238.4(KCNH2):c.3164G>A (p.Arg1055Gln)	not provided|not specified|Cardiovascular phenotype|Long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Sep 17, 2020)	criteria provided, conflicting interpretations	VCV000067475	7	150947043	67475	78371	rs41307270	GOF	0.442	0.242	0.316
KCNH2	1057	S	N	NM_000238.4(KCNH2):c.3170G>A (p.Ser1057Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 14, 2020)	criteria provided, single submitter	VCV001022238	7	150947037	1022238	1007332	rs765524161	GOF	0.527	0.187	0.286
KCNH2	1058	A	E	NM_000238.4(KCNH2):c.3173C>A (p.Ala1058Glu)	not provided	not provided	no assertion provided	VCV000067476	7	150947034	67476	78372	rs41313752	GOF	0.528	0.209	0.263
KCNH2	1058	A	T	NM_000238.4(KCNH2):c.3172G>A (p.Ala1058Thr)	Long QT syndrome	Uncertain significance(Last reviewed: May 31, 2017)	criteria provided, single submitter	VCV000456924	7	150947035	456924	457054	rs555269990	Neutral	0.47	0.282	0.248
KCNH2	1060	M	V	NM_000238.4(KCNH2):c.3178A>G (p.Met1060Val)	Arrhythmia	Uncertain significance(Last reviewed: Feb 26, 2019)	criteria provided, single submitter	VCV000924650	7	150947029	924650	910675	rs767156792	Neutral	0.503	0.345	0.152
KCNH2	1062	T	I	NM_000238.4(KCNH2):c.3185C>T (p.Thr1062Ile)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jul 10, 2018)	criteria provided, multiple submitters, no conflicts	VCV000586073	7	150947022	586073	576958	rs761811244	Neutral	0.352	0.472	0.176
KCNH2	1063	V	L	NM_000238.4(KCNH2):c.3187G>C (p.Val1063Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 10, 2020)	criteria provided, single submitter	VCV000845043	7	150947020	845043	833314	rs139722868	Neutral	0.634	0.239	0.127
KCNH2	1064	L	R	NM_000238.4(KCNH2):c.3191T>G (p.Leu1064Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 4, 2019)	criteria provided, single submitter	VCV000845095	7	150947016	845095	833313	rs1800917926	LOF	0.712	0.142	0.145
KCNH2	1066	L	V	NM_000238.4(KCNH2):c.3196C>G (p.Leu1066Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067479	7	150947011	67479	78375	rs199473027	Neutral	0.574	0.293	0.134
KCNH2	1068	Q	R	NM_000238.4(KCNH2):c.3203A>G (p.Gln1068Arg)	Long QT syndrome|not provided|Cardiovascular phenotype|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Mar 16, 2021)	criteria provided, conflicting interpretations	VCV000067480	7	150947004	67480	78376	rs151031345	GOF	0.489	0.187	0.323
KCNH2	1070	Q	L	NM_000238.4(KCNH2):c.3209A>T (p.Gln1070Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 9, 2020)	criteria provided, single submitter	VCV001043117	7	150946998	1043117	1027880	rs1386424341	Neutral	0.353	0.407	0.24
KCNH2	1075	P	L	NM_000238.4(KCNH2):c.3224C>T (p.Pro1075Leu)	Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 2	Uncertain significance(Last reviewed: May 28, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067481	7	150946983	67481	78377	rs199473028	Neutral	0.373	0.352	0.274
KCNH2	1075	P	Q	NM_000238.4(KCNH2):c.3224C>A	Arrhythmia	Uncertain	criteria	VCV000629130	7	150946983	629130	617336	rs199473028	GOF	0.399	0.215	0.387

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Pro1075Gln)		significance(Last reviewed: Aug 28, 2018)	provided, single submitter										
KCNH2	1077	A	T	NM_000238.4(KCNH2):c.3229G>A (p.Ala1077Thr)	not provided|Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jun 10, 2021)	criteria provided, multiple submitters, no conflicts	VCV000526983	7	150946978	526983	522440	rs201382073	GOF	0.399	0.229	0.372
KCNH2	1078	Y	C	NM_000238.4(KCNH2):c.3233A>G (p.Tyr1078Cys)	Long QT syndrome|Congenital long QT syndrome|not specified	Uncertain significance(Last reviewed: Mar 22, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067483	7	150946974	67483	78379	rs199473029	Neutral	0.401	0.423	0.176
KCNH2	1083	T	A	NM_000238.4(KCNH2):c.3247A>G (p.Thr1083Ala)	not provided|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Oct 26, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200529	7	150946960	200529	197137	rs751559990	Neutral	0.358	0.336	0.306
KCNH2	1084	P	L	NM_000238.4(KCNH2):c.3251C>T (p.Pro1084Leu)	Cardiovascular phenotype|Long QT syndrome| Arrhythmia	Uncertain significance(Last reviewed: Oct 9, 2019)	criteria provided, multiple submitters, no conflicts	VCV000456925	7	150946956	456925	456805	rs762510312	Neutral	0.352	0.331	0.316
KCNH2	1088	P	R	NM_000238.4(KCNH2):c.3263C>G (p.Pro1088Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 5, 2019)	criteria provided, single submitter	VCV000954908	7	150946944	954908	945516	rs1800912992	GOF	0.412	0.198	0.391
KCNH2	1093	P	L	NM_000238.4(KCNH2):c.3278C>T (p.Pro1093Leu)	Arrhythmia|Congenital long QT syndrome|not specified|Long QT syndrome|Long QT syndrome 2	Uncertain significance(Last reviewed: Oct 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067485	7	150946929	67485	78381	rs199473545	Neutral	0.344	0.344	0.311
KCNH2	1096	P	S	NM_000238.4(KCNH2):c.3286C>T (p.Pro1096Ser)	Arrhythmia	Uncertain significance(Last reviewed: Sep 23, 2019)	criteria provided, single submitter	VCV000921691	7	150946921	921691	910671	rs1554423737	GOF	0.364	0.202	0.434
KCNH2	1096	P	T	NM_000238.4(KCNH2):c.3286C>A (p.Pro1096Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 9, 2017)	criteria provided, single submitter	VCV000456927	7	150946921	456927	456465	rs1554423737	GOF	0.356	0.21	0.434
KCNH2	1097	V	I	NM_000238.4(KCNH2):c.3289G>A (p.Val1097Ile)	Long QT syndrome|not specified|not provided| Cardiovascular phenotype|Arrhythmia	Uncertain significance(Last reviewed: Sep 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067486	7	150946918	67486	78382	rs199473030	Neutral	0.521	0.31	0.169
KCNH2	1098	S	R	NM_000238.4(KCNH2):c.3292A>C (p.Ser1098Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 12, 2019)	criteria provided, single submitter	VCV000966117	7	150946915	966117	955091	rs1800911203	GOF	0.419	0.196	0.385
KCNH2	1098	S	R	NM_000238.4(KCNH2):c.3292A>C (p.Ser1098Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 12, 2019)	criteria provided, single submitter	VCV000966117	7	150946915	966117	955091	rs1800911203	GOF	0.419	0.196	0.385
KCNH2	1098	S	R	NM_000238.4(KCNH2):c.3294C>G (p.Ser1098Arg)	Arrhythmia	Uncertain significance(Last reviewed: Mar 9, 2019)	criteria provided, single submitter	VCV000928123	7	150946913	928123	910669	rs1203403811	GOF	0.419	0.196	0.385
KCNH2	1098	S	R	NM_000238.4(KCNH2):c.3294C>G (p.Ser1098Arg)	Arrhythmia	Uncertain significance(Last reviewed: Mar 9, 2019)	criteria provided, single submitter	VCV000928123	7	150946913	928123	910669	rs1203403811	GOF	0.419	0.196	0.385
KCNH2	1099	P	L	NM_000238.4(KCNH2):c.3296C>T	Long QT syndrome	Uncertain	criteria	VCV000526951	7	150946911	526951	522764	rs1339742380	Neutral	0.409	0.342	0.249

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Pro1099Leu)		significance(Last reviewed: Apr 4, 2018)	provided, single submitter										
KCNH2	1101	P	L	NM_000238.4(KCNH2):c.3302C>T (p.Pro1101Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067487	7	150946905	67487	78383	rs199473041	Neutral	0.391	0.35	0.259
KCNH2	1102	T	I	NM_000238.4(KCNH2):c.3305C>T (p.Thr1102Ile)	not provided|Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200531	7	150946902	200531	197135	rs794728405	Neutral	0.339	0.333	0.328
KCNH2	1107	S	L	NM_000238.4(KCNH2):c.3320C>T (p.Ser1107Leu)	Long QT syndrome|Short QT syndrome 1	Uncertain significance(Last reviewed: Dec 3, 2019)	criteria provided, multiple submitters, no conflicts	VCV000843735	7	150946887	843735	833312	rs1303580599	Neutral	0.337	0.361	0.302
KCNH2	1108	L	V	NM_000238.4(KCNH2):c.3322C>G (p.Leu1108Val)	Long QT syndrome|Arrhythmia|Long QT syndrome 2|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 6, 2020)	criteria provided, conflicting interpretations	VCV000067488	7	150946885	67488	78384	rs199473031	Neutral	0.528	0.296	0.176
KCNH2	1115	M	V	NM_000238.4(KCNH2):c.3343A>G (p.Met1115Val)	Long QT syndrome|Arrhythmia|not provided| Congenital long QT syndrome	Uncertain significance(Last reviewed: Dec 16, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067490	7	150945502	67490	78386	rs199473546	Neutral	0.504	0.314	0.181
KCNH2	1116	A	V	NM_000238.4(KCNH2):c.3347C>T (p.Ala1116Val)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome	Uncertain significance(Last reviewed: Mar 10, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067491	7	150945498	67491	78387	rs199473032	Neutral	0.445	0.301	0.254
KCNH2	1117	C	Y	NM_000238.4(KCNH2):c.3350G>A (p.Cys1117Tyr)	Ventricular tachycardia	Uncertain significance(Last reviewed: May 7, 2014)	no assertion criteria provided	VCV000180385	7	150945495	180385	178572	rs730880119	Neutral	0.532	0.299	0.169
KCNH2	1118	E	K	NM_000238.4(KCNH2):c.3352G>A (p.Glu1118Lys)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 7, 2020)	criteria provided, single submitter	VCV000855130	7	150945493	855130	833311	rs1800858264	GOF	0.458	0.223	0.32
KCNH2	1119	E	Q	NM_000238.4(KCNH2):c.3355G>C (p.Glu1119Gln)	Long QT syndrome|not provided|Arrhythmia	Uncertain significance(Last reviewed: Jul 28, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067492	7	150945490	67492	78388	rs199473033	GOF	0.472	0.233	0.296
KCNH2	1120	L	V	NM_000238.4(KCNH2):c.3358C>G (p.Leu1120Val)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 8, 2018)	criteria provided, single submitter	VCV000661932	7	150945487	661932	635884	rs1584839287	Neutral	0.558	0.265	0.177
KCNH2	1121	P	S	NM_000238.4(KCNH2):c.3361C>T (p.Pro1121Ser)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Feb 18, 2021)	criteria provided, multiple submitters, no conflicts	VCV000920215	7	150945484	920215	910665	rs1800857722	GOF	0.426	0.209	0.366
KCNH2	1122	P	L	NM_000238.4(KCNH2):c.3365C>T (p.Pro1122Leu)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Feb 15, 2022)	criteria provided, multiple submitters, no conflicts	VCV000200534	7	150945480	200534	197129	rs531460655	Neutral	0.452	0.311	0.237
KCNH2	1122	P	R	NM_000238.4(KCNH2):c.3365C>G (p.Pro1122Arg)	not specified	Uncertain significance(Last reviewed: Jul 3, 2014)	no assertion criteria provided	VCV000234998	7	150945480	234998	236768	rs531460655	GOF	0.462	0.223	0.315
KCNH2	1127	L	F	NM_000238.4(KCNH2):c.3379C>T (p.Leu1127Phe)	Arrhythmia	Uncertain significance(Last	criteria provided, single	VCV000923705	7	150945466	923705	910664	rs1800856518	Neutral	0.518	0.314	0.169

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Apr 14, 2019)	submitter										
KCNH2	1131	G	V	NM_000238.4(KCNH2):c.3392G>T (p.Gly1131Val)	Arrhythmia	Uncertain significance(Last reviewed: Dec 3, 2018)	criteria provided, single submitter	VCV000918364	7	150945453	918364	910663	rs370558996	Neutral	0.466	0.29	0.244
KCNH2	1132	P	A	NM_000238.4(KCNH2):c.3394C>G (p.Pro1132Ala)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jul 28, 2021)	criteria provided, multiple submitters, no conflicts	VCV000191469	7	150945451	191469	189256	rs786205422	Neutral	0.459	0.294	0.247
KCNH2	1135	R	H	NM_000238.4(KCNH2):c.3404G>A (p.Arg1135His)	short QT syndrome	not provided	no assertion provided	VCV000067493	7	150945441	67493	78389	rs199473547	Neutral	0.467	0.281	0.252
KCNH2	1136	L	F	NM_000238.4(KCNH2):c.3406C>T (p.Leu1136Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 30, 2020)	criteria provided, single submitter	VCV001035669	7	150945439	1035669	1027879	rs1344656004	Neutral	0.534	0.312	0.154
KCNH2	1139	P	L	NM_000238.4(KCNH2):c.3416C>T (p.Pro1139Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 14, 2016)	criteria provided, single submitter	VCV000359302	7	150945429	359302	310302	rs886062086	Neutral	0.405	0.34	0.254
KCNH2	1144	A	T	NM_000238.4(KCNH2):c.3430G>A (p.Ala1144Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067495	7	150945415	67495	78391	rs199473034	GOF	0.436	0.204	0.36
KCNH2	1146	T	S	NM_000238.4(KCNH2):c.3436A>T (p.Thr1146Ser)	not provided|Short QT syndrome 1|Long QT syndrome 2|Arrhythmia|Cardiovascular phenotype|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Dec 6, 2020)	criteria provided, conflicting interpretations	VCV000359301	7	150945409	359301	302179	rs778879572	GOF	0.39	0.193	0.417
KCNH2	1153	H	Y	NM_000238.4(KCNH2):c.3457C>T (p.His1153Tyr)	Long QT syndrome|Arrhythmia|Long QT syndrome 2|Long QT syndrome 2|Short QT syndrome 1|not provided|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jun 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067497	7	150945388	67497	78393	rs199473035	Neutral	0.388	0.334	0.278
KCNH2	1154	G	S	NM_000238.4(KCNH2):c.3460G>A (p.Gly1154Ser)	Long QT syndrome|Arrhythmia|not provided	Uncertain significance(Last reviewed: Jan 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067498	7	150945385	67498	78394	rs199473548	GOF	0.434	0.178	0.387
KCNH2	1156	D	G	NM_000238.4(KCNH2):c.3467A>G (p.Asp1156Gly)	Arrhythmia	Uncertain significance(Last reviewed: Dec 11, 2019)	criteria provided, single submitter	VCV000926485	7	150945378	926485	910660	rs1266712553	Neutral	0.475	0.258	0.267
KCNH2	1157	P	L	NM_000238.4(KCNH2):c.3470C>T (p.Pro1157Leu)	Long QT syndrome|not provided|SUDDEN INFANT DEATH SYNDROME|Arrhythmia	Uncertain significance(Last reviewed: Jun 22, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067499	7	150945375	67499	78395	rs143167166	Neutral	0.465	0.31	0.225
KCNH2	1157	P	S	NM_000238.4(KCNH2):c.3469C>T (p.Pro1157Ser)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Mar 19, 2021)	criteria provided, multiple submitters, no conflicts	VCV000456933	7	150945376	456933	457045	rs1449906095	GOF	0.452	0.227	0.321
KCNH5	5	K	N	NM_139318.5(KCNH5):c.15G>C (p.Lys5Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 2, 2020)	criteria provided, single submitter	VCV000949152	14	63045172	949152	927115	rs775719605	Neutral	0.654	0.155	0.191
KCNH5	58	V	I	NM_139318.5(KCNH5):c.172G>A (p.Val58Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV001058253	14	63016856	1058253	1048593	NA	LOF	0.775	0.112	0.112
KCNH5	89	E	Q	NM_139318.5(KCNH5):c.265G>C (p.Glu89Gln)	Early infantile epileptic encephalopathy with	Uncertain	criteria	VCV000582493	14	63006405	582493	568969	rs1162818997	LOF	0.665	0.126	0.209

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					suppression bursts	significance(Last reviewed: Jun 19, 2018)	provided, single submitter										
KCNH5	108	Y	C	NM_139318.5(KCNH5):c.323A>G (p.Tyr108Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 21, 2019)	criteria provided, single submitter	VCV000835675	14	63001441	835675	841578	rs1464940813	LOF	0.848	0.073	0.079
KCNH5	141	D	A	NM_139318.5(KCNH5):c.422A>C (p.Asp141Ala)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 11, 2020)	criteria provided, single submitter	VCV001035483	14	63001342	1035483	1031646	rs1891006879	Neutral	0.568	0.232	0.199
KCNH5	147	T	M	NM_139318.5(KCNH5):c.440C>T (p.Thr147Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 23, 2019)	criteria provided, single submitter	VCV000860425	14	62987181	860425	841577	rs180894715	Neutral	0.619	0.186	0.195
KCNH5	154	R	W	NM_139318.5(KCNH5):c.460C>T (p.Arg154Trp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 18, 2017)	criteria provided, single submitter	VCV000530526	14	62987161	530526	528714	rs745970423	LOF	0.681	0.14	0.179
KCNH5	158	N	K	NM_139318.5(KCNH5):c.474T>G (p.Asn158Lys)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Jun 7, 2020)	criteria provided, single submitter	VCV000848600	14	62987147	848600	841576	rs1890724757	LOF	0.69	0.094	0.216
KCNH5	167	T	M	NM_139318.5(KCNH5):c.500C>T (p.Thr167Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 26, 2018)	criteria provided, single submitter	VCV000569692	14	62987121	569692	568968	rs771253745	Neutral	0.628	0.178	0.194
KCNH5	172	T	R	NM_139318.5(KCNH5):c.515C>G (p.Thr172Arg)	Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Sep 29, 2020)	criteria provided, single submitter	VCV000698214	14	62987106	698214	688322	rs34764419	Neutral	0.64	0.172	0.187
KCNH5	184	V	I	NM_139318.5(KCNH5):c.550G>A (p.Val184Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 30, 2017)	criteria provided, single submitter	VCV000461397	14	62981264	461397	463348	rs752609494	LOF	0.762	0.123	0.115
KCNH5	204	P	L	NM_139318.5(KCNH5):c.611C>T (p.Pro204Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 16, 2017)	criteria provided, single submitter	VCV000530445	14	62981203	530445	528233	rs776842795	LOF	0.691	0.142	0.168
KCNH5	245	I	L	NM_139318.5(KCNH5):c.733A>T (p.Ile245Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 23, 2020)	criteria provided, single submitter	VCV000999581	14	62981081	999581	995892	rs754761406	LOF	0.731	0.153	0.115
KCNH5	245	I	V	NM_139318.5(KCNH5):c.733A>G (p.Ile245Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 16, 2020)	criteria provided, single submitter	VCV000461398	14	62981081	461398	463877	rs754761406	LOF	0.736	0.147	0.116
KCNH5	246	A	S	NM_139318.5(KCNH5):c.736G>T (p.Ala246Ser)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Sep 9, 2020)	criteria provided, single submitter	VCV000574962	14	62981078	574962	572835	rs114447363	LOF	0.788	0.088	0.124
KCNH5	256	V	I	NM_139318.5(KCNH5):c.766G>A (p.Val256Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 16, 2017)	criteria provided, single submitter	VCV000461399	14	62981048	461399	464357	rs756506083	LOF	0.822	0.078	0.1
KCNH5	263	V	I	NM_139318.5(KCNH5):c.787G>A (p.Val263Ile)	Early infantile epileptic encephalopathy with suppression bursts|Seizures	Uncertain significance(Last reviewed: Dec 11, 2017)	criteria provided, single submitter	VCV000530474	14	62981027	530474	528231	rs760127525	LOF	0.82	0.078	0.103
KCNH5	273	P	S	NM_139318.5(KCNH5):c.817C>T (p.Pro273Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 29, 2020)	criteria provided, single submitter	VCV000960733	14	62980997	960733	948599	rs747654948	LOF	0.751	0.1	0.149

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH5	274	G	S	NM_139318.5(KCNH5):c.820G>A (p.Gly274Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 14, 2018)	criteria provided, single submitter	VCV000646789	14	62980994	646789	642535	rs182494425	LOF	0.789	0.073	0.139
KCNH5	310	E	D	NM_139318.5(KCNH5):c.930A>T (p.Glu310Asp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 25, 2020)	criteria provided, single submitter	VCV001040470	14	62980884	1040470	1031645	rs201960719	LOF	0.764	0.103	0.133
KCNH5	327	R	H	NM_139318.5(KCNH5):c.980G>A (p.Arg327His)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Oct 7, 2020)	criteria provided, single submitter	VCV000100784	14	62950522	100784	106649	rs587777164	GOF	0.497	0.044	0.459
KCNH5	333	R	H	NM_139318.5(KCNH5):c.998G>A (p.Arg333His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 12, 2020)	criteria provided, single submitter	VCV001042182	14	62950504	1042182	1031644	rs1383017734	GOF	0.505	0.049	0.446
KCNH5	335	A	T	NM_139318.5(KCNH5):c.1003G>A (p.Ala335Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 17, 2017)	criteria provided, single submitter	VCV000461384	14	62950499	461384	463873	rs376158544	LOF	0.75	0.058	0.193
KCNH5	366	I	M	NM_139318.5(KCNH5):c.1098A>G (p.Ile366Met)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Dec 2, 2020)	criteria provided, single submitter	VCV000416115	14	62950404	416115	399839	rs146883958	LOF	0.819	0.082	0.099
KCNH5	374	E	K	NM_139318.5(KCNH5):c.1120G>A (p.Glu374Lys)	Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Sep 1, 2020)	criteria provided, single submitter	VCV000940401	14	62950382	940401	936650	rs1889977806	LOF	0.828	0.095	0.077
KCNH5	396	I	F	NM_139318.5(KCNH5):c.1186A>T (p.Ile396Phe)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 9, 2019)	criteria provided, single submitter	VCV000938564	14	62950316	938564	936649	rs1889976030	LOF	0.803	0.082	0.115
KCNH5	401	R	C	NM_139318.5(KCNH5):c.1201C>T (p.Arg401Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 15, 2019)	criteria provided, single submitter	VCV000937814	14	62950301	937814	936648	rs753034623	Neutral	0.678	0.171	0.151
KCNH5	401	R	H	NM_139318.5(KCNH5):c.1202G>A (p.Arg401His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 13, 2019)	criteria provided, single submitter	VCV000949515	14	62950300	949515	927114	rs1461051682	LOF	0.677	0.146	0.177
KCNH5	407	G	A	NM_139318.5(KCNH5):c.1220G>C (p.Gly407Ala)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 8, 2019)	criteria provided, single submitter	VCV000950621	14	62950282	950621	927113	rs1889974797	LOF	0.769	0.092	0.139
KCNH5	447	M	V	NM_139318.5(KCNH5):c.1339A>G (p.Met447Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 29, 2017)	criteria provided, single submitter	VCV000461386	14	62950163	461386	463861	rs200506671	LOF	0.772	0.089	0.139
KCNH5	449	S	L	NM_139318.5(KCNH5):c.1346C>T (p.Ser449Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 25, 2018)	criteria provided, single submitter	VCV000572837	14	62950156	572837	568956	rs369774413	LOF	0.835	0.045	0.12
KCNH5	455	V	I	NM_139318.5(KCNH5):c.1363G>A (p.Val455Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 19, 2019)	criteria provided, single submitter	VCV000846406	14	62950139	846406	841574	rs1224129490	LOF	0.794	0.093	0.114
KCNH5	467	V	A	NM_139318.5(KCNH5):c.1400T>C (p.Val467Ala)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 14, 2020)	criteria provided, single submitter	VCV000948390	14	62849822	948390	927112	rs1887769181	LOF	0.826	0.054	0.12
KCNH5	479	N	S	NM_139318.5(KCNH5):c.1436A>G (p.Asn479Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 21, 2020)	criteria provided, single submitter	VCV001001238	14	62849786	1001238	995891	rs1887768573	LOF	0.702	0.107	0.191

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH5	500	G	D	NM_139318.5(KCNH5):c.1499G>A (p.Gly500Asp)	Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Oct 6, 2020)	criteria provided, single submitter	VCV001154530	14	62849723	1154530	1144646	NA	LOF	0.738	0.088	0.174
KCNH5	505	V	I	NM_139318.5(KCNH5):c.1513G>A (p.Val505Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 14, 2019)	criteria provided, single submitter	VCV000858394	14	62849709	858394	841573	rs1480621890	LOF	0.758	0.123	0.119
KCNH5	527	I	F	NM_139318.5(KCNH5):c.1579A>T (p.Ile527Phe)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 6, 2020)	criteria provided, multiple submitters, no conflicts	VCV000658479	14	62802572	658479	642534	rs149561461	LOF	0.732	0.137	0.131
KCNH5	542	R	W	NM_139318.5(KCNH5):c.1624C>T (p.Arg542Trp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 3, 2020)	criteria provided, single submitter	VCV001017233	14	62802527	1017233	1011143	rs543742412	LOF	0.703	0.089	0.208
KCNH5	553	L	S	NM_139318.5(KCNH5):c.1658T>C (p.Leu553Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 21, 2019)	criteria provided, single submitter	VCV000530494	14	62802493	530494	528242	rs1406117275	LOF	0.767	0.113	0.12
KCNH5	556	D	N	NM_139318.5(KCNH5):c.1666G>A (p.Asp556Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 5, 2019)	criteria provided, single submitter	VCV000944554	14	62802485	944554	927111	rs150842380	LOF	0.699	0.082	0.219
KCNH5	598	I	V	NM_139318.5(KCNH5):c.1792A>G (p.Ile598Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 31, 2019)	criteria provided, single submitter	VCV000935009	14	62802359	935009	936647	rs1886679317	LOF	0.838	0.085	0.077
KCNH5	616	I	N	NM_139318.5(KCNH5):c.1847T>A (p.Ile616Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 17, 2018)	criteria provided, single submitter	VCV000581723	14	62779900	581723	568955	rs753938626	LOF	0.821	0.105	0.074
KCNH5	622	T	I	NM_139318.5(KCNH5):c.1865C>T (p.Thr622Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 24, 2020)	criteria provided, single submitter	VCV000838252	14	62779882	838252	841572	rs376649352	Neutral	0.613	0.205	0.181
KCNH5	624	A	G	NM_139318.5(KCNH5):c.1871C>G (p.Ala624Gly)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 6, 2020)	criteria provided, single submitter	VCV001017242	14	62779876	1017242	1011142	rs762614737	LOF	0.724	0.124	0.152
KCNH5	629	N	I	NM_139318.5(KCNH5):c.1886A>T (p.Asn629Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 16, 2020)	criteria provided, single submitter	VCV001012036	14	62779861	1012036	995890	rs1204494580	LOF	0.831	0.092	0.077
KCNH5	643	R	G	NM_139318.5(KCNH5):c.1927C>G (p.Arg643Gly)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 13, 2019)	criteria provided, single submitter	VCV000965550	14	62779820	965550	957241	rs771984476	LOF	0.745	0.077	0.178
KCNH5	654	T	A	NM_139318.5(KCNH5):c.1960A>G (p.Thr654Ala)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Nov 18, 2020)	criteria provided, single submitter	VCV001164896	14	62779787	1164896	1157294	NA	LOF	0.748	0.087	0.165
KCNH5	657	A	G	NM_139318.5(KCNH5):c.1970C>G (p.Ala657Gly)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Aug 10, 2020)	criteria provided, single submitter	VCV001169943	14	62779777	1169943	1157293	NA	LOF	0.787	0.109	0.104
KCNH5	675	I	T	NM_139318.5(KCNH5):c.2024T>C (p.Ile675Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 4, 2019)	criteria provided, single submitter	VCV000943234	14	62708451	943234	936646	rs747312358	Neutral	0.691	0.166	0.142
KCNH5	690	L	R	NM_139318.5(KCNH5):c.2069T>G (p.Leu690Arg)	not provided	Uncertain significance(Last reviewed: May 20,	criteria provided, single submitter	VCV001163361	14	62708406	1163361	1152567	NA	Neutral	0.645	0.214	0.142

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)											
KCNH5	691	R	L	NM_139318.5(KCNH5):c.2072G>T (p.Arg691Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 4, 2020)	criteria provided, single submitter	VCV001002329	14	62708403	1002329	995889	rs200737420	Neutral	0.502	0.328	0.17
KCNH5	691	R	Q	NM_139318.5(KCNH5):c.2072G>A (p.Arg691Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 23, 2019)	criteria provided, single submitter	VCV000845946	14	62708403	845946	841571	rs200737420	Neutral	0.519	0.273	0.208
KCNH5	692	Q	R	NM_139318.5(KCNH5):c.2075A>G (p.Gln692Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 22, 2019)	criteria provided, single submitter	VCV000958018	14	62708400	958018	948598	rs372088477	Neutral	0.523	0.239	0.237
KCNH5	699	S	G	NM_139318.5(KCNH5):c.2095A>G (p.Ser699Gly)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 4, 2018)	criteria provided, single submitter	VCV000569642	14	62708380	569642	568163	rs1360641528	Neutral	0.529	0.307	0.165
KCNH5	699	S	R	NM_139318.5(KCNH5):c.2097C>A (p.Ser699Arg)	Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Nov 10, 2020)	criteria provided, single submitter	VCV000410375	14	62708378	410375	399834	rs148812753	Neutral	0.545	0.253	0.202
KCNH5	705	P	R	NM_139318.5(KCNH5):c.2114C>G (p.Pro705Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 21, 2016)	criteria provided, single submitter	VCV000410376	14	62708361	410376	399689	rs1060502866	Neutral	0.681	0.213	0.106
KCNH5	718	E	Q	NM_139318.5(KCNH5):c.2152G>C (p.Glu718Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV000948838	14	62708323	948838	927110	rs1308657380	Neutral	0.516	0.282	0.202
KCNH5	720	R	Q	NM_139318.5(KCNH5):c.2159G>A (p.Arg720Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 13, 2018)	criteria provided, single submitter	VCV000658599	14	62708316	658599	642533	rs201820912	Neutral	0.509	0.3	0.191
KCNH5	745	A	T	NM_139318.5(KCNH5):c.2233G>A (p.Ala745Thr)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Dec 5, 2020)	criteria provided, single submitter	VCV001166143	14	62708242	1166143	1157292	NA	Neutral	0.526	0.279	0.195
KCNH5	746	S	P	NM_139318.5(KCNH5):c.2236T>C (p.Ser746Pro)	Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Jun 22, 2020)	criteria provided, single submitter	VCV000530451	14	62708239	530451	528234	rs34439924	Neutral	0.508	0.294	0.199
KCNH5	750	T	I	NM_139318.5(KCNH5):c.2249C>T (p.Thr750Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 20, 2020)	criteria provided, single submitter	VCV001055057	14	62708226	1055057	1048592	NA	Neutral	0.494	0.344	0.162
KCNH5	770	T	N	NM_139318.5(KCNH5):c.2309C>A (p.Thr770Asn)	not specified	Uncertain significance(Last reviewed: Oct 8, 2015)	criteria provided, single submitter	VCV000435550	14	62708166	435550	429583	rs1555351942	Neutral	0.511	0.302	0.187
KCNH5	772	E	G	NM_139318.5(KCNH5):c.2315A>G (p.Glu772Gly)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Nov 15, 2020)	criteria provided, single submitter	VCV000416117	14	62708160	416117	400220	rs79913182	Neutral	0.549	0.295	0.157
KCNH5	781	A	T	NM_139318.5(KCNH5):c.2341G>A (p.Ala781Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 28, 2018)	criteria provided, single submitter	VCV000641941	14	62708134	641941	642532	rs1595587605	Neutral	0.532	0.306	0.162
KCNH5	787	N	S	NM_139318.5(KCNH5):c.2360A>G (p.Asn787Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 21, 2019)	criteria provided, single submitter	VCV000849400	14	62708115	849400	841570	rs745444217	Neutral	0.519	0.296	0.185
KCNH5	789	G	C	NM_139318.5(KCNH5):c.2365G>T (p.Gly789Cys)	Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Jul 29, 2020)	criteria provided, single submitter	VCV000530610	14	62708110	530610	528595	rs140205536	Neutral	0.553	0.299	0.148

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNH5	789	G	S	NM_139318.5(KCNH5):c.2365G>A (p.Gly789Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 24, 2018)	criteria provided, single submitter	VCV000649223	14	62708110	649223	642531	rs140205536	Neutral	0.513	0.294	0.193
KCNH5	791	D	G	NM_139318.5(KCNH5):c.2372A>G (p.Asp791Gly)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 12, 2018)	criteria provided, single submitter	VCV000578531	14	62708103	578531	572830	rs548465461	Neutral	0.528	0.304	0.167
KCNH5	794	C	W	NM_139318.5(KCNH5):c.2382T>G (p.Cys794Trp)	Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Feb 27, 2020)	criteria provided, single submitter	VCV000953736	14	62708093	953736	948597	rs1884478264	Neutral	0.632	0.255	0.113
KCNH5	807	N	S	NM_139318.5(KCNH5):c.2420A>G (p.Asn807Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV000950048	14	62708055	950048	927109	rs770323761	Neutral	0.486	0.312	0.203
KCNH5	813	R	Q	NM_139318.5(KCNH5):c.2438G>A (p.Arg813Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 5, 2020)	criteria provided, single submitter	VCV000530516	14	62708037	530516	528232	rs534607642	Neutral	0.528	0.32	0.152
KCNH5	819	G	V	NM_139318.5(KCNH5):c.2456G>T (p.Gly819Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 30, 2019)	criteria provided, single submitter	VCV000962145	14	62708019	962145	948596	rs373106772	Neutral	0.543	0.301	0.156
KCNH5	830	N	S	NM_139318.5(KCNH5):c.2489A>G (p.Asn830Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 23, 2019)	criteria provided, single submitter	VCV000959141	14	62707986	959141	948595	rs151103620	Neutral	0.494	0.311	0.195
KCNH5	845	K	R	NM_139318.5(KCNH5):c.2534A>G (p.Lys845Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 18, 2019)	criteria provided, single submitter	VCV000648052	14	62707941	648052	642530	rs1189342324	Neutral	0.574	0.271	0.155
KCNH5	853	V	M	NM_139318.5(KCNH5):c.2557G>A (p.Val853Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 5, 2018)	criteria provided, single submitter	VCV000665274	14	62707918	665274	642529	rs140331213	Neutral	0.613	0.269	0.118
KCNH5	854	T	I	NM_139318.5(KCNH5):c.2561C>T (p.Thr854Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 22, 2020)	criteria provided, single submitter	VCV001052180	14	62707914	1052180	1048591	NA	Neutral	0.524	0.334	0.142
KCNH5	862	D	G	NM_139318.5(KCNH5):c.2585A>G (p.Asp862Gly)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 16, 2018)	criteria provided, single submitter	VCV000664320	14	62707890	664320	642528	rs1595587388	Neutral	0.504	0.327	0.169
KCNH5	864	C	Y	NM_139318.5(KCNH5):c.2591G>A (p.Cys864Tyr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 20, 2019)	criteria provided, single submitter	VCV000652298	14	62707884	652298	642527	rs1595587379	Neutral	0.611	0.268	0.122
KCNH5	878	A	T	NM_139318.5(KCNH5):c.2632G>A (p.Ala878Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 8, 2020)	criteria provided, single submitter	VCV000461393	14	62707843	461393	464339	rs376837885	Neutral	0.495	0.319	0.186
KCNH5	896	H	Y	NM_139318.5(KCNH5):c.2686C>T (p.His896Tyr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV001014460	14	62707789	1014460	1011141	rs758357872	Neutral	0.516	0.342	0.143
KCNH5	902	P	A	NM_139318.5(KCNH5):c.2704C>G (p.Pro902Ala)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 4, 2020)	criteria provided, single submitter	VCV001000405	14	62707771	1000405	995888	rs757688048	Neutral	0.493	0.336	0.171
KCNH5	928	M	V	NM_139318.5(KCNH5):c.2782A>G (p.Met928Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last	criteria provided, single	VCV000581814	14	62707693	581814	568153	rs775247224	Neutral	0.584	0.279	0.137

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: May 11, 2018)	submitter										
KCNH5	930	A	V	NM_139318.5(KCNH5):c.2789C>T (p.Ala930Val)	not provided	Uncertain significance(Last reviewed: Nov 12, 2019)	no assertion criteria provided	VCV000977409	14	62707686	977409	965537	rs562140660	Neutral	0.558	0.273	0.169
KCNH5	938	I	V	NM_139318.5(KCNH5):c.2812A>G (p.Ile938Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 16, 2017)	criteria provided, single submitter	VCV000530444	14	62707663	530444	528227	rs375347137	Neutral	0.685	0.205	0.11
KCNH5	943	S	L	NM_139318.5(KCNH5):c.2828C>T (p.Ser943Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000530414	14	62707647	530414	528696	rs761523326	Neutral	0.509	0.327	0.164
KCNH5	944	E	K	NM_139318.5(KCNH5):c.2830G>A (p.Glu944Lys)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Apr 14, 2020)	criteria provided, single submitter	VCV001170778	14	62707645	1170778	1157290	NA	Neutral	0.554	0.274	0.172
KCNH5	945	K	E	NM_139318.5(KCNH5):c.2833A>G (p.Lys945Glu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 5, 2018)	criteria provided, single submitter	VCV000575130	14	62707642	575130	568151	rs1566633488	Neutral	0.554	0.285	0.161
KCNH5	962	P	S	NM_139318.5(KCNH5):c.2884C>T (p.Pro962Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 11, 2020)	criteria provided, single submitter	VCV000461394	14	62707591	461394	464333	rs202223175	Neutral	0.528	0.321	0.151
KCNH5	964	Q	E	NM_139318.5(KCNH5):c.2890C>G (p.Gln964Glu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 19, 2018)	criteria provided, single submitter	VCV000568811	14	62707585	568811	568147	rs753525158	Neutral	0.548	0.291	0.161
KCNH5	964	Q	K	NM_139318.5(KCNH5):c.2890C>A (p.Gln964Lys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 29, 2016)	criteria provided, single submitter	VCV000241863	14	62707585	241863	241853	rs753525158	Neutral	0.532	0.288	0.18
KCNH5	976	P	L	NM_139318.5(KCNH5):c.2927C>T (p.Pro976Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 3, 2020)	criteria provided, single submitter	VCV001056451	14	62707548	1056451	1048590	NA	Neutral	0.551	0.311	0.138
KCNH5	983	K	R	NM_139318.5(KCNH5):c.2948A>G (p.Lys983Arg)	Early infantile epileptic encephalopathy with suppression bursts	Benign(Last reviewed: Mar 18, 2020)	criteria provided, single submitter	VCV000962491	14	62707527	962491	948594	rs1271834396	Neutral	0.595	0.261	0.144
KCNH5	984	D	V	NM_139318.5(KCNH5):c.2951A>T (p.Asp984Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 5, 2018)	criteria provided, single submitter	VCV000575955	14	62707524	575955	572829	rs1566633394	Neutral	0.546	0.308	0.146
KCNQ1	1	M	K	NM_000218.3(KCNQ1):c.2T>A (p.Met1Lys)	not provided	Pathogenic(Last reviewed: Feb 3, 2016)	criteria provided, single submitter	VCV000488859	11	2445100	488859	481985	rs199473485	Neutral	0.661	0.179	0.159
KCNQ1	1	M	L	NM_000218.3(KCNQ1):c.1A>T (p.Met1Leu)	Long QT syndrome	Pathogenic(Last reviewed: Aug 2, 2016)	criteria provided, single submitter	VCV000220309	11	2445099	220309	222138	rs199473441	Neutral	0.635	0.224	0.141
KCNQ1	1	M	T	NM_000218.3(KCNQ1):c.2T>C (p.Met1Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053033	11	2445100	53033	67701	rs199473485	Neutral	0.628	0.212	0.16
KCNQ1	1	M	V	NM_000218.3(KCNQ1):c.1A>G (p.Met1Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067064	11	2445099	67064	77960	rs199473441	Neutral	0.627	0.226	0.148
KCNQ1	2	A	T	NM_000218.3(KCNQ1):c.4G>A (p.Ala2Thr)	not provided	Uncertain significance	no assertion criteria provided	VCV001050694	11	2445102	1050694	1038052	NA	Neutral	0.536	0.273	0.191
KCNQ1	2	A	V	NM_000218.3(KCNQ1):c.5C>T (p.Ala2Val)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: May 27,	criteria provided, single submitter	VCV000067091	11	2445103	67091	77987	rs199473442	Neutral	0.525	0.299	0.177

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2014)											
KCNQ1	3	A	G	NM_000218.3(KCNQ1):c.8C>G (p.Ala3Gly)	not provided	Uncertain significance(Last reviewed: Dec 31, 2019)	criteria provided, single submitter	VCV000200866	11	2445106	200866	197406	rs794728543	Neutral	0.616	0.215	0.169
KCNQ1	7	P	S	NM_000218.3(KCNQ1):c.19C>T (p.Pro7Ser)	Congenital long QT syndrome|not specified|Long QT syndrome 1|Long QT syndrome	Uncertain significance(Last reviewed: Sep 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053028	11	2445117	53028	67696	rs199473443	Neutral	0.56	0.218	0.221
KCNQ1	9	R	T	NM_000218.3(KCNQ1):c.26G>C (p.Arg9Thr)	Long QT syndrome 1|Jervell and Lange-Nielsen syndrome 1	Uncertain significance	criteria provided, single submitter	VCV000979113	11	2445124	979113	967192	rs1469698360	Neutral	0.556	0.263	0.181
KCNQ1	10	A	D	NM_000218.3(KCNQ1):c.29C>A (p.Ala10Asp)	short QT syndrome|Long QT syndrome|Familial atrial fibrillation|Congenital long QT syndrome|Jervell and Lange-Nielsen syndrome	Uncertain significance(Last reviewed: Jun 14, 2016)	criteria provided, single submitter	VCV000304214	11	2445127	304214	326169	rs886048161	Neutral	0.612	0.191	0.197
KCNQ1	11	E	K	NM_000218.3(KCNQ1):c.31G>A (p.Glu11Lys)	not provided|Cardiovascular phenotype	Conflicting interpretations of pathogenicity(Last reviewed: Aug 12, 2019)	criteria provided, conflicting interpretations	VCV000418271	11	2445129	418271	408324	rs959449103	Neutral	0.559	0.227	0.213
KCNQ1	13	K	R	NM_000218.3(KCNQ1):c.38A>G (p.Lys13Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 13, 2018)	criteria provided, single submitter	VCV000405256	11	2445136	405256	398459	rs1060500622	Neutral	0.56	0.233	0.207
KCNQ1	14	R	C	NM_000218.3(KCNQ1):c.40C>T (p.Arg14Cys)	not provided	not provided	no assertion provided	VCV000067071	11	2445138	67071	77967	rs199473444	Neutral	0.59	0.263	0.147
KCNQ1	14	R	L	NM_000218.3(KCNQ1):c.41G>T (p.Arg14Leu)	Long QT syndrome|not provided|Long QT syndrome 1|Jervell and Lange-Nielsen syndrome 1|Atrial fibrillation, familial, 3|Short QT syndrome 2	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000804969	11	2445139	804969	793392	rs1424013094	Neutral	0.507	0.328	0.165
KCNQ1	22	G	R	NM_000218.3(KCNQ1):c.64G>C (p.Gly22Arg)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Aug 8, 2018)	criteria provided, multiple submitters, no conflicts	VCV000200868	11	2445162	200868	197408	rs794728545	GOF	0.611	0.148	0.241
KCNQ1	24	R	W	NM_000218.3(KCNQ1):c.70C>T (p.Arg24Trp)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 24, 2019)	criteria provided, single submitter	VCV000946105	11	2445168	946105	926181	rs990778345	Neutral	0.579	0.266	0.155
KCNQ1	25	R	P	NM_000218.3(KCNQ1):c.74G>C (p.Arg25Pro)	Conduction disorder of the heart	Uncertain significance	criteria provided, single submitter	VCV000684795	11	2445172	684795	672417	rs1589884210	Neutral	0.578	0.246	0.176
KCNQ1	27	S	R	NM_000218.3(KCNQ1):c.81C>A (p.Ser27Arg)	not provided	Uncertain significance(Last reviewed: May 7, 2014)	criteria provided, single submitter	VCV000200869	11	2445179	200869	197409	rs794728546	Neutral	0.558	0.189	0.254
KCNQ1	28	A	G	NM_000218.3(KCNQ1):c.83C>G (p.Ala28Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 18, 2020)	criteria provided, single submitter	VCV001036317	11	2445181	1036317	1030169	rs767089696	Neutral	0.559	0.247	0.194
KCNQ1	35	P	L	NM_000218.3(KCNQ1):c.104C>T (p.Pro35Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 10, 2020)	criteria provided, single submitter	VCV000840373	11	2445202	840373	838232	rs1373379406	Neutral	0.506	0.303	0.191
KCNQ1	39	F	V	NM_000218.3(KCNQ1):c.496T>G (p.Phe166Val)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 3, 2019)	criteria provided, single submitter	VCV000846376	11	2570646	846376	838233	rs768419064	Neutral	0.487	0.346	0.167

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	45	P	R	NM_000218.3(KCNQ1):c.134C>G (p.Pro45Arg)	not provided	Uncertain significance(Last reviewed: Jul 2, 2020)	criteria provided, single submitter	VCV001312976	11	2445232	1312976	1303237	NA	Neutral	0.609	0.168	0.224
KCNQ1	45	V	L	NM_000218.3(KCNQ1):c.514G>C (p.Val172Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 6, 2018)	criteria provided, single submitter	VCV000653086	11	2570664	653086	639902	rs199472694	Neutral	0.569	0.266	0.165
KCNQ1	46	A	T	NM_000218.3(KCNQ1):c.136G>A (p.Ala46Thr)	not provided|Cardiovascular phenotype|not specified|Congenital long QT syndrome|Long QT syndrome 1	Uncertain significance(Last reviewed: Sep 6, 2018)	criteria provided, multiple submitters, no conflicts	VCV000052982	11	2445234	52982	67650	rs199473671	Neutral	0.432	0.395	0.173
KCNQ1	48	L	P	NM_000218.3(KCNQ1):c.524T>C (p.Leu175Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 18, 2018)	criteria provided, single submitter	VCV000644915	11	2570674	644915	639903	rs1589956588	Neutral	0.545	0.272	0.183
KCNQ1	57	G	D	NM_000218.3(KCNQ1):c.170G>A (p.Gly57Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 31, 2020)	criteria provided, single submitter	VCV001057004	11	2445268	1057004	1047158	NA	Neutral	0.63	0.154	0.216
KCNQ1	57	G	V	NM_000218.3(KCNQ1):c.170G>T (p.Gly57Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067051	11	2445268	67051	77947	rs199473445	Neutral	0.578	0.214	0.208
KCNQ1	58	V	L	NM_000218.3(KCNQ1):c.553G>C (p.Val185Leu)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Apr 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV001044083	11	2570703	1044083	1030173	rs749351255	Neutral	0.548	0.277	0.175
KCNQ1	59	P	A	NM_000218.3(KCNQ1):c.175C>G (p.Pro59Ala)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Nov 27, 2017)	criteria provided, single submitter	VCV000518877	11	2445273	518877	510270	rs1554958059	Neutral	0.565	0.233	0.202
KCNQ1	61	P	T	NM_000218.3(KCNQ1):c.181C>A (p.Pro61Thr)	not specified|not provided	Uncertain significance(Last reviewed: Aug 16, 2019)	criteria provided, multiple submitters, no conflicts	VCV000517627	11	2445279	517627	497616	rs1273257287	Neutral	0.54	0.231	0.229
KCNQ1	62	A	T	NM_000218.3(KCNQ1):c.184G>A (p.Ala62Thr)	not specified	Likely benign(Last reviewed: Dec 17, 2013)	criteria provided, single submitter	VCV000200880	11	2445282	200880	197411	rs794728554	Neutral	0.516	0.244	0.239
KCNQ1	63	P	S	NM_000218.3(KCNQ1):c.187C>T (p.Pro63Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 21, 2020)	criteria provided, single submitter	VCV001058075	11	2445285	1058075	1047159	NA	Neutral	0.555	0.222	0.223
KCNQ1	63	P	T	NM_000218.3(KCNQ1):c.187C>A (p.Pro63Thr)	Atrial fibrillation, familial, 3|Short QT syndrome 2|Long QT syndrome 1|Jervell and Lange-Nielsen syndrome 1	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000879571	11	2445285	879571	867802	rs1280717988	Neutral	0.54	0.238	0.222
KCNQ1	64	P	S	NM_000218.3(KCNQ1):c.190C>T (p.Pro64Ser)	not provided	Uncertain significance(Last reviewed: Jan 31, 2020)	criteria provided, multiple submitters, no conflicts	VCV000502107	11	2445288	502107	493531	rs1554958062	Neutral	0.551	0.214	0.235
KCNQ1	65	R	C	NM_000218.3(KCNQ1):c.574C>T (p.Arg192Cys)	Arrhythmia|not specified|Long QT syndrome	Uncertain significance(Last reviewed: Nov 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000578178	11	2570724	578178	567156	rs775059928	Neutral	0.607	0.238	0.154
KCNQ1	66	S	F	NM_000218.3(KCNQ1):c.197C>T (p.Ser66Phe)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Nov 7, 2019)	criteria provided, single submitter	VCV000067063	11	2445295	67063	77959	rs199473446	Neutral	0.514	0.291	0.195

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	66	S	Y	NM_000218.3(KCNQ1):c.197C>A (p.Ser66Tyr)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 16, 2019)	criteria provided, single submitter	VCV000933445	11	2445295	933445	935468	rs199473446	Neutral	0.507	0.274	0.219
KCNQ1	70	P	T	NM_000218.3(KCNQ1):c.208C>A (p.Pro70Thr)	not provided	Uncertain significance(Last reviewed: Jan 29, 2013)	criteria provided, single submitter	VCV000200871	11	2445306	200871	197413	rs794728548	Neutral	0.489	0.3	0.211
KCNQ1	73	P	T	NM_000218.3(KCNQ1):c.217C>A (p.Pro73Thr)	Short QT syndrome 2|not provided|Cardiovascular phenotype|Congenital long QT syndrome|Long QT syndrome 1|Long QT syndrome	Uncertain significance(Last reviewed: Jun 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053031	11	2445315	53031	67699	rs199472676	Neutral	0.536	0.24	0.224
KCNQ1	77	S	F	NM_000218.3(KCNQ1):c.230C>T (p.Ser77Phe)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Mar 24, 2021)	criteria provided, multiple submitters, no conflicts	VCV000451983	11	2445328	451983	444781	rs774349962	Neutral	0.484	0.317	0.199
KCNQ1	78	D	A	NM_000218.3(KCNQ1):c.233A>C (p.Asp78Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 19, 2019)	criteria provided, single submitter	VCV000943522	11	2445331	943522	935469	rs1846021008	Neutral	0.488	0.314	0.199
KCNQ1	78	D	H	NM_000218.3(KCNQ1):c.232G>C (p.Asp78His)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 3, 2020)	criteria provided, single submitter	VCV001060673	11	2445330	1060673	1047160	NA	Neutral	0.494	0.305	0.201
KCNQ1	80	G	A	NM_000218.3(KCNQ1):c.239G>C (p.Gly80Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 27, 2019)	criteria provided, single submitter	VCV000960512	11	2445337	960512	947393	rs1846021110	Neutral	0.593	0.192	0.215
KCNQ1	81	P	L	NM_000218.3(KCNQ1):c.242C>T (p.Pro81Leu)	not provided	Uncertain significance(Last reviewed: Oct 15, 2018)	criteria provided, single submitter	VCV000200875	11	2445340	200875	197414	rs771921468	Neutral	0.566	0.253	0.182
KCNQ1	83	M	I	NM_000218.3(KCNQ1):c.630G>A (p.Met210Ile)	Arrhythmia	Uncertain significance(Last reviewed: Oct 18, 2019)	criteria provided, single submitter	VCV000923241	11	2571350	923241	911559	rs776811872	Neutral	0.363	0.495	0.141
KCNQ1	83	P	S	NM_000218.3(KCNQ1):c.247C>T (p.Pro83Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV001040957	11	2445345	1040957	1030170	rs772821418	Neutral	0.514	0.316	0.17
KCNQ1	84	P	R	NM_000218.3(KCNQ1):c.251C>G (p.Pro84Arg)	not provided	Uncertain significance(Last reviewed: Jan 11, 2013)	criteria provided, single submitter	VCV000200876	11	2445349	200876	197415	rs794728551	Neutral	0.582	0.184	0.234
KCNQ1	84	P	S	NM_000218.3(KCNQ1):c.250C>T (p.Pro84Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 7, 2020)	criteria provided, single submitter	VCV000948475	11	2445348	948475	926182	rs1846021360	Neutral	0.535	0.277	0.188
KCNQ1	92	S	C	NM_000218.3(KCNQ1):c.275C>G (p.Ser92Cys)	not provided	Uncertain significance(Last reviewed: Jan 16, 2019)	criteria provided, single submitter	VCV001211571	11	2445373	1211571	1201568	NA	Neutral	0.506	0.326	0.168
KCNQ1	92	S	P	NM_000218.3(KCNQ1):c.274T>C (p.Ser92Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 2, 2018)	criteria provided, single submitter	VCV000577361	11	2445372	577361	565633	rs764455359	Neutral	0.415	0.407	0.179
KCNQ1	95	S	R	NM_000218.3(KCNQ1):c.285C>A (p.Ser95Arg)	not specified	Uncertain significance(Last reviewed: Jul 3, 2018)	criteria provided, single submitter	VCV000666842	11	2445383	666842	654676	rs1589884451	Neutral	0.565	0.22	0.215

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	96	T	R	NM_000218.3(KCNQ1):c.287C>G (p.Thr96Arg)	Long QT syndrome 1	Likely pathogenic(Last reviewed: Jan 1, 2019)	criteria provided, single submitter	VCV000983021	11	2445385	983021	970932	rs1337409061	Neutral	0.461	0.33	0.209
KCNQ1	99	P	R	NM_000218.3(KCNQ1):c.296C>G (p.Pro99Arg)	not provided|Long QT syndrome 1|Beckwith-Wiedemann syndrome|Jervell and Lange-Nielsen syndrome 1|Atrial fibrillation, familial, 3|Short QT syndrome 2|Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200877	11	2445394	200877	197416	rs370435862	LOF	0.684	0.137	0.179
KCNQ1	100	I	L	NM_000218.3(KCNQ1):c.679A>C (p.Ile227Leu)	not provided	Likely pathogenic(Last reviewed: Aug 10, 2012)	criteria provided, single submitter	VCV000200819	11	2571399	200819	197439	rs794728511	Neutral	0.368	0.479	0.153
KCNQ1	104	R	L	NM_000218.3(KCNQ1):c.692G>T (p.Arg231Leu)	Cardiovascular phenotype	Likely pathogenic(Last reviewed: Jun 15, 2016)	criteria provided, single submitter	VCV000519257	11	2572021	519257	510274	rs199472709	Neutral	0.543	0.211	0.246
KCNQ1	104	R	S	NM_000218.3(KCNQ1):c.691C>A (p.Arg231Ser)	not provided	Pathogenic(Last reviewed: Aug 13, 2019)	criteria provided, single submitter	VCV000280173	11	2572020	280173	264532	rs199473457	Neutral	0.559	0.25	0.191
KCNQ1	105	H	L	NM_000218.3(KCNQ1):c.314A>T (p.His105Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067065	11	2445412	67065	77961	rs199473447	Neutral	0.474	0.257	0.27
KCNQ1	106	L	P	NM_000218.3(KCNQ1):c.698T>C (p.Leu233Pro)	not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jun 15, 2020)	criteria provided, conflicting interpretations	VCV000993829	11	2572027	993829	981737	rs780236727	Neutral	0.554	0.329	0.117
KCNQ1	106	V	D	NM_000218.3(KCNQ1):c.317T>A (p.Val106Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 12, 2018)	criteria provided, single submitter	VCV000573930	11	2445415	573930	570472	rs1565023268	Neutral	0.689	0.189	0.123
KCNQ1	107	Q	H	NM_000218.3(KCNQ1):c.321G>T (p.Gln107His)	Long QT syndrome 1	Likely pathogenic(Last reviewed: Jan 9, 2018)	criteria provided, single submitter	VCV000200878	11	2445419	200878	197417	rs1554958092	LOF	0.81	0.081	0.109
KCNQ1	107	Q	L	NM_000218.3(KCNQ1):c.701A>T (p.Gln234Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 17, 2016)	criteria provided, single submitter	VCV000405267	11	2572030	405267	398593	rs794728570	GOF	0.672	0.082	0.246
KCNQ1	107	Q	P	NM_000218.3(KCNQ1):c.701A>C (p.Gln234Pro)	not provided	Likely pathogenic(Last reviewed: Aug 14, 2019)	criteria provided, single submitter	VCV000200896	11	2572030	200896	197440	rs794728570	LOF	0.773	0.105	0.121
KCNQ1	107	Q	R	NM_000218.3(KCNQ1):c.701A>G (p.Gln234Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 31, 2015)	criteria provided, single submitter	VCV000220083	11	2572030	220083	222139	rs794728570	LOF	0.828	0.072	0.1
KCNQ1	109	L	P	NM_000218.3(KCNQ1):c.707T>C (p.Leu236Pro)	not provided|Long QT syndrome 1	Pathogenic/Likely pathogenic(Last reviewed: Apr 17, 2019)	criteria provided, multiple submitters, no conflicts	VCV000200820	11	2572036	200820	197441	rs794728512	Neutral	0.605	0.283	0.112
KCNQ1	109	L	R	NM_000218.3(KCNQ1):c.707T>G (p.Leu236Arg)	not provided	Likely pathogenic(Last reviewed: Mar 27, 2015)	criteria provided, single submitter	VCV000200897	11	2572036	200897	197442	rs794728512	Neutral	0.734	0.162	0.104
KCNQ1	110	V	E	NM_000218.3(KCNQ1):c.329_330delinsAG (p.Val110Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 21, 2020)	criteria provided, single submitter	VCV001020240	11	2445427 - 2445428	1020240	1009621	rs1846023075	LOF	0.76	0.121	0.119
KCNQ1	110	V	I	NM_000218.3(KCNQ1):c.328G>A (p.Val110Ile)	not provided|Long QT syndrome|not specified	Conflicting	criteria	VCV000053034	11	2445426	53034	67702	rs199472677	Neutral	0.626	0.25	0.124

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						interpretations of pathogenicity(Last reviewed: Sep 17, 2020)	provided, conflicting interpretations										
KCNQ1	111	Y	C	NM_000218.3(KCNQ1):c.332A>G (p.Tyr111Cys)	not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: Jul 26, 2017)	criteria provided, single submitter	VCV000053035	11	2445430	53035	67703	rs199472678	LOF	0.854	0.042	0.104
KCNQ1	113	H	R	NM_000218.3(KCNQ1):c.719A>G (p.His240Arg)	Arrhythmia	Uncertain significance(Last reviewed: Feb 19, 2021)	criteria provided, single submitter	VCV001171337	11	2572048	1171337	1160869	NA	LOF	0.792	0.09	0.118
KCNQ1	114	L	P	NM_000218.3(KCNQ1):c.341T>C (p.Leu114Pro)	Congenital long QT syndrome|Long QT syndrome	Likely pathogenic(Last reviewed: May 15, 2017)	criteria provided, single submitter	VCV000053036	11	2445439	53036	67704	rs199473448	Neutral	0.729	0.169	0.103
KCNQ1	114	V	F	NM_000218.3(KCNQ1):c.721G>T (p.Val241Phe)	Long QT syndrome	Uncertain significance(Last reviewed: May 15, 2016)	criteria provided, single submitter	VCV000405255	11	2572050	405255	398149	rs199956744	LOF	0.728	0.14	0.133
KCNQ1	114	V	I	NM_000218.3(KCNQ1):c.721G>A (p.Val241Ile)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 3, 2019)	criteria provided, multiple submitters, no conflicts	VCV000191477	11	2572050	191477	189329	rs199956744	Neutral	0.679	0.208	0.114
KCNQ1	115	E	G	NM_000218.3(KCNQ1):c.344A>G (p.Glu115Gly)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jul 30, 2020)	criteria provided, single submitter	VCV000053037	11	2445442	53037	67705	rs199472679	LOF	0.816	0.087	0.097
KCNQ1	115	E	K	NM_000218.3(KCNQ1):c.343G>A (p.Glu115Lys)	not provided	Pathogenic(Last reviewed: Sep 10, 2013)	criteria provided, single submitter	VCV000200879	11	2445441	200879	197418	rs794728553	LOF	0.815	0.088	0.097
KCNQ1	116	R	H	NM_000218.3(KCNQ1):c.347G>A (p.Arg116His)	not specified	Uncertain significance(Last reviewed: Feb 26, 2015)	criteria provided, single submitter	VCV000178873	11	2445445	178873	175698	rs727504506	LOF	0.768	0.111	0.121
KCNQ1	117	P	L	NM_000218.3(KCNQ1):c.350C>T (p.Pro117Leu)	Long QT syndrome|Long QT syndrome 1| Congenital long QT syndrome	Uncertain significance(Last reviewed: Sep 25, 2020)	criteria provided, single submitter	VCV000003141	11	2445448	3141	18180	rs120074191	LOF	0.855	0.036	0.109
KCNQ1	117	P	S	NM_000218.3(KCNQ1):c.349C>T (p.Pro117Ser)	Long QT syndrome 1	Likely pathogenic(Last reviewed: Jan 1, 2016)	criteria provided, single submitter	VCV000254159	11	2445447	254159	248631	rs886037906	LOF	0.894	0.025	0.08
KCNQ1	119	G	D	NM_000218.3(KCNQ1):c.356G>A (p.Gly119Asp)	not provided	not provided	no assertion provided	VCV000067066	11	2445454	67066	77962	rs199472680	LOF	0.875	0.044	0.081
KCNQ1	119	G	R	NM_000218.3(KCNQ1):c.355G>C (p.Gly119Arg)	Long QT syndrome 1|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Apr 12, 2018)	criteria provided, conflicting interpretations	VCV000560702	11	2445453	560702	551782	rs1325525794	LOF	0.857	0.045	0.097
KCNQ1	119	G	V	NM_000218.3(KCNQ1):c.356G>T (p.Gly119Val)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 8, 2014)	criteria provided, single submitter	VCV001037209	11	2445454	1037209	1030171	rs199472680	LOF	0.857	0.051	0.092
KCNQ1	121	W	F	NM_000218.3(KCNQ1):c.743_744delinsTC (p.Trp248Phe)	Jervell and Lange-Nielsen syndrome 1	not provided	no assertion provided	VCV000053093	11	2572072 - 2572073	53093	67761	rs397508123	LOF	0.73	0.145	0.125
KCNQ1	122	C	Y	NM_000218.3(KCNQ1):c.365G>A (p.Cys122Tyr)	not provided|Congenital long QT syndrome|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Apr 25, 2019)	criteria provided, conflicting interpretations	VCV000053039	11	2445463	53039	67707	rs199472681	LOF	0.76	0.037	0.202

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	124	L	Q	NM_000218.3(KCNQ1):c.752T>A (p.Leu251Gln)	Long QT syndrome	Uncertain significance(Last reviewed: May 12, 2020)	criteria provided, single submitter	VCV001004846	11	2572081	1004846	994445	rs199472716	LOF	0.803	0.114	0.083
KCNQ1	125	Y	C	NM_000218.3(KCNQ1):c.374A>G (p.Tyr125Cys)	not provided	Likely pathogenic(Last reviewed: Sep 2, 2015)	criteria provided, single submitter	VCV000429656	11	2445472	429656	421849	rs1131691513	LOF	0.835	0.034	0.132
KCNQ1	125	Y	D	NM_000218.3(KCNQ1):c.373T>G (p.Tyr125Asp)	not provided	Likely pathogenic(Last reviewed: Jan 12, 2012)	criteria provided, single submitter	VCV000200910	11	2445471	200910	197420	rs794728578	LOF	0.873	0.023	0.105
KCNQ1	125	Y	N	NM_000218.3(KCNQ1):c.373T>A (p.Tyr125Asn)	Long QT syndrome	Likely pathogenic	criteria provided, single submitter	VCV000684802	11	2445471	684802	672418	rs794728578	LOF	0.853	0.034	0.112
KCNQ1	126	H	L	NM_000218.3(KCNQ1):c.377A>T (p.His126Leu)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 6, 2017)	criteria provided, conflicting interpretations	VCV000200911	11	2445475	200911	197421	rs794728579	GOF	0.611	0.122	0.267
KCNQ1	126	S	A	NM_000218.3(KCNQ1):c.757T>G (p.Ser253Ala)	Arrhythmia	Uncertain significance(Last reviewed: Apr 20, 2021)	criteria provided, single submitter	VCV000924383	11	2572086	924383	911565	rs764781840	GOF	0.5	0.236	0.264
KCNQ1	126	S	C	NM_000218.3(KCNQ1):c.758C>G (p.Ser253Cys)	Long QT syndrome 1	Pathogenic(Last reviewed: Jan 1, 2014)	criteria provided, single submitter	VCV000207971	11	2572087	207971	204199	rs794728513	LOF	0.66	0.107	0.233
KCNQ1	126	S	F	NM_000218.3(KCNQ1):c.758C>T (p.Ser253Phe)	not provided	Likely pathogenic(Last reviewed: Feb 21, 2013)	criteria provided, single submitter	VCV000200822	11	2572087	200822	197444	rs794728513	GOF	0.562	0.098	0.34
KCNQ1	126	S	P	NM_000218.3(KCNQ1):c.757T>C (p.Ser253Pro)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jun 26, 2019)	criteria provided, conflicting interpretations	VCV000200821	11	2572086	200821	197443	rs764781840	LOF	0.665	0.112	0.223
KCNQ1	127	F	L	NM_000218.3(KCNQ1):c.381C>A (p.Phe127Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067067	11	2445479	67067	77963	rs199472682	Neutral	0.63	0.256	0.114
KCNQ1	129	V	A	NM_000218.3(KCNQ1):c.386T>C (p.Val129Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 30, 2019)	criteria provided, single submitter	VCV000945727	11	2445484	945727	926183	rs1846024488	Neutral	0.551	0.306	0.143
KCNQ1	129	V	G	NM_000218.3(KCNQ1):c.386T>G (p.Val129Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 24, 2020)	no assertion criteria provided	VCV000977167	11	2445484	977167	965307	rs1846024488	LOF	0.812	0.062	0.126
KCNQ1	129	V	I	NM_000218.3(KCNQ1):c.385G>A (p.Val129Ile)	not provided	not provided	no assertion provided	VCV000067068	11	2445483	67068	77964	rs199472683	Neutral	0.455	0.411	0.134
KCNQ1	131	L	P	NM_000218.3(KCNQ1):c.392T>C (p.Leu131Pro)	not provided	Uncertain significance(Last reviewed: Jan 12, 2018)	criteria provided, single submitter	VCV000200873	11	2527933	200873	197423	rs794728550	LOF	0.826	0.086	0.088
KCNQ1	132	I	L	NM_000218.3(KCNQ1):c.394A>C (p.Ile132Leu)	Congenital long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jul 16, 2020)	criteria provided, single submitter	VCV000053040	11	2527935	53040	67708	rs199472684	Neutral	0.468	0.412	0.12
KCNQ1	133	V	I	NM_000218.3(KCNQ1):c.397G>A (p.Val133Ile)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jan 23, 2020)	criteria provided, single submitter	VCV000067069	11	2527938	67069	77965	rs199473449	Neutral	0.457	0.404	0.14

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	134	L	P	NM_000218.3(KCNQ1):c.401T>C (p.Leu134Pro)	Long QT syndrome|Congenital long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 13, 2019)	criteria provided, conflicting interpretations	VCV000067070	11	2527942	67070	77966	rs199472685	LOF	0.817	0.091	0.091
KCNQ1	135	L	P	NM_000218.3(KCNQ1):c.785T>C (p.Leu262Pro)	Long QT syndrome 1|Prolonged QT interval	Pathogenic(Last reviewed: Jul 10, 2020)	criteria provided, single submitter	VCV000560359	11	2572850	560359	551407	rs1564821090	LOF	0.785	0.118	0.097
KCNQ1	136	C	F	NM_000218.3(KCNQ1):c.407G>T (p.Cys136Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053041	11	2527948	53041	67709	rs199472686	GOF	0.661	0.061	0.278
KCNQ1	137	L	F	NM_000218.3(KCNQ1):c.409C>T (p.Leu137Phe)	not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: Aug 19, 2014)	criteria provided, single submitter	VCV000053042	11	2527950	53042	67710	rs199473450	Neutral	0.612	0.27	0.119
KCNQ1	139	L	M	NM_000218.3(KCNQ1):c.796C>A (p.Leu266Met)	Arrhythmia|Short QT syndrome 2	Uncertain significance(Last reviewed: Oct 5, 2018)	criteria provided, multiple submitters, no conflicts	VCV000598782	11	2572861	598782	589841	rs1263583359	Neutral	0.541	0.356	0.103
KCNQ1	139	L	R	NM_000218.3(KCNQ1):c.797T>G (p.Leu266Arg)	not provided|Long QT syndrome	Likely pathogenic(Last reviewed: Sep 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000393006	11	2572862	393006	372345	rs199473460	LOF	0.832	0.079	0.089
KCNQ1	140	S	G	NM_000218.3(KCNQ1):c.418A>G (p.Ser140Gly)	Atrial fibrillation, familial, 3|Atrial fibrillation	Pathogenic(Last reviewed: Jan 10, 2003)	no assertion criteria provided	VCV000003143	11	2527959	3143	18182	rs120074192	LOF	0.743	0.066	0.19
KCNQ1	140	Y	F	NM_000218.3(KCNQ1):c.800A>T (p.Tyr267Phe)	Cardiovascular phenotype|Long QT syndrome	Uncertain significance(Last reviewed: Aug 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000264075	11	2572865	264075	258693	rs886039033	LOF	0.706	0.124	0.17
KCNQ1	141	I	V	NM_000218.3(KCNQ1):c.802A>G (p.Ile268Val)	not provided|Arrhythmia	Uncertain significance(Last reviewed: Nov 4, 2019)	criteria provided, multiple submitters, no conflicts	VCV000378922	11	2572867	378922	372346	rs757421492	Neutral	0.692	0.184	0.124
KCNQ1	141	V	M	NM_000218.3(KCNQ1):c.421G>A (p.Val141Met)	Short QT syndrome 2|Long QT syndrome|Atrial fibrillation|not provided|Cardiovascular phenotype	Pathogenic(Last reviewed: Jan 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067072	11	2527962	67072	77968	rs199472687	Neutral	0.691	0.16	0.149
KCNQ1	142	G	R	NM_000218.3(KCNQ1):c.805G>C (p.Gly269Arg)	not provided	Pathogenic(Last reviewed: Dec 1, 2011)	criteria provided, single submitter	VCV000200898	11	2572870	200898	197445	rs120074193	LOF	0.882	0.02	0.097
KCNQ1	142	G	V	NM_000218.3(KCNQ1):c.806G>T (p.Gly269Val)	not provided	Pathogenic(Last reviewed: Oct 14, 2014)	criteria provided, single submitter	VCV000200899	11	2572871	200899	197446	rs120074194	LOF	0.795	0.06	0.145
KCNQ1	143	S	F	NM_000218.3(KCNQ1):c.428C>T (p.Ser143Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 27, 2018)	criteria provided, single submitter	VCV000188120	11	2527969	188120	186152	rs786204083	GOF	0.719	0.032	0.249
KCNQ1	144	T	A	NM_000218.3(KCNQ1):c.430A>G (p.Thr144Ala)	Congenital long QT syndrome|Long QT syndrome|not provided|Cardiovascular phenotype	Conflicting interpretations of pathogenicity(Last reviewed: Jan 13, 2022)	criteria provided, conflicting interpretations	VCV000067073	11	2527971	67073	77969	rs199473451	LOF	0.758	0.058	0.184
KCNQ1	146	E	G	NM_000218.3(KCNQ1):c.437A>G (p.Glu146Gly)	not specified|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jan 22, 2020)	criteria provided, multiple submitters, no conflicts	VCV000418272	11	2527978	418272	408327	rs914460959	LOF	0.848	0.061	0.091

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	146	E	K	NM_000218.3(KCNQ1):c.436G>A (p.Glu146Lys)	Congenital long QT syndrome|Long QT syndrome|not provided|Arrhythmia	Uncertain significance(Last reviewed: Oct 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053043	11	2527977	53043	67711	rs199472688	LOF	0.847	0.055	0.097
KCNQ1	147	Q	L	NM_000218.3(KCNQ1):c.440A>T (p.Gln147Leu)	not provided	Uncertain significance(Last reviewed: Jun 1, 2020)	criteria provided, single submitter	VCV001312672	11	2527981	1312672	1302933	NA	GOF	0.585	0.135	0.28
KCNQ1	147	Q	R	NM_000218.3(KCNQ1):c.440A>G (p.Gln147Arg)	Atrial fibrillation	not provided	no assertion provided	VCV000067074	11	2527981	67074	77970	rs199472689	LOF	0.756	0.101	0.143
KCNQ1	149	A	T	NM_000218.3(KCNQ1):c.445G>A (p.Ala149Thr)	Arrhythmia	Uncertain significance(Last reviewed: Jul 15, 2021)	criteria provided, single submitter	VCV001331890	11	2527986	1331890	1322610	NA	Neutral	0.531	0.369	0.1
KCNQ1	149	S	F	NM_000218.3(KCNQ1):c.827C>T (p.Ser276Phe)	not provided	Likely pathogenic(Last reviewed: May 25, 2012)	criteria provided, single submitter	VCV000200823	11	2572892	200823	197447	rs794728514	LOF	0.686	0.135	0.179
KCNQ1	150	A	G	NM_000218.3(KCNQ1):c.449C>G (p.Ala150Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053044	11	2527990	53044	67712	rs199473452	Neutral	0.729	0.186	0.084
KCNQ1	150	A	T	NM_000218.3(KCNQ1):c.448G>A (p.Ala150Thr)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: May 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200909	11	2527989	200909	197426	rs794728577	Neutral	0.555	0.358	0.087
KCNQ1	152	F	C	NM_000218.3(KCNQ1):c.836T>G (p.Phe279Cys)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 19, 2018)	criteria provided, single submitter	VCV000663237	11	2572901	663237	639907	rs1589958473	LOF	0.902	0.033	0.065
KCNQ1	153	T	M	NM_000218.3(KCNQ1):c.458C>T (p.Thr153Met)	Long QT syndrome|Hypertrophic cardiomyopathy|Arrhythmia|Cardiovascular phenotype|not specified|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 3, 2021)	criteria provided, conflicting interpretations	VCV000067075	11	2527999	67075	77971	rs143709408	LOF	0.739	0.135	0.127
KCNQ1	155	L	V	NM_000218.3(KCNQ1):c.844C>G (p.Leu282Val)	Arrhythmia|not provided	Uncertain significance(Last reviewed: Jan 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000919046	11	2572909	919046	911569	rs979241598	Neutral	0.662	0.262	0.076
KCNQ1	156	A	P	NM_000218.3(KCNQ1):c.847G>C (p.Ala283Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 8, 2016)	criteria provided, single submitter	VCV000405263	11	2572912	405263	398469	rs1060500627	LOF	0.872	0.054	0.074
KCNQ1	156	L	P	NM_000218.3(KCNQ1):c.467T>C (p.Leu156Pro)	not provided	Likely pathogenic	criteria provided, single submitter	VCV000684787	11	2528008	684787	672419	rs1589931156	LOF	0.906	0.036	0.058
KCNQ1	157	F	C	NM_000218.3(KCNQ1):c.470T>G (p.Phe157Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053046	11	2528011	53046	67714	rs199472690	LOF	0.898	0.032	0.07
KCNQ1	159	M	L	NM_000218.3(KCNQ1):c.475A>T (p.Met159Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 12, 2018)	criteria provided, single submitter	VCV000664331	11	2528016	664331	639900	rs1589931165	Neutral	0.658	0.241	0.101
KCNQ1	160	A	S	NM_000218.3(KCNQ1):c.859G>T (p.Ala287Ser)	Arrhythmia	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV000926686	11	2572924	926686	911570	rs765665086	LOF	0.779	0.123	0.098
KCNQ1	160	A	T	NM_000218.3(KCNQ1):c.859G>A (p.Ala287Thr)	not provided|Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Sep 18, 2020)	criteria provided, multiple submitters, no	VCV000657731	11	2572924	657731	639908	rs765665086	Neutral	0.729	0.168	0.103

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNQ1	160	A	V	NM_000218.3(KCNQ1):c.860C>T (p.Ala287Val)	Arrhythmia	Uncertain significance(Last reviewed: Nov 16, 2018)	criteria provided, single submitter	VCV000923836	11	2572925	923836	911571	rs199472735	Neutral	0.654	0.223	0.123
KCNQ1	160	E	K	NM_000218.3(KCNQ1):c.478G>A (p.Glu160Lys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053049	11	2570628	53049	67717	rs199473453	LOF	0.867	0.044	0.088
KCNQ1	160	E	V	NM_000218.3(KCNQ1):c.479A>T (p.Glu160Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067076	11	2570629	67076	77972	rs199472691	LOF	0.785	0.086	0.129
KCNQ1	161	V	M	NM_000218.3(KCNQ1):c.862G>A (p.Val288Met)	not provided|Arrhythmia	Uncertain significance(Last reviewed: Dec 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000452888	11	2572927	452888	444784	rs946704016	Neutral	0.712	0.196	0.093
KCNQ1	162	V	M	NM_000218.3(KCNQ1):c.484G>A (p.Val162Met)	Arrhythmia|not specified|not provided|Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Jun 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067077	11	2570634	67077	77973	rs199472692	Neutral	0.719	0.2	0.082
KCNQ1	164	V	A	NM_000218.3(KCNQ1):c.491T>C (p.Val164Ala)	Arrhythmia	Uncertain significance(Last reviewed: Apr 2, 2019)	criteria provided, single submitter	VCV000922815	11	2570641	922815	911552	rs1848316801	LOF	0.774	0.136	0.09
KCNQ1	165	V	A	NM_000218.3(KCNQ1):c.494T>C (p.Val165Ala)	Arrhythmia	Uncertain significance(Last reviewed: Nov 28, 2018)	criteria provided, single submitter	VCV000922039	11	2570644	922039	911553	rs1848317004	Neutral	0.729	0.192	0.079
KCNQ1	165	V	M	NM_000218.3(KCNQ1):c.493G>A (p.Val165Met)	Arrhythmia|not specified	Uncertain significance(Last reviewed: Jul 31, 2019)	criteria provided, multiple submitters, no conflicts	VCV000426347	11	2570643	426347	415264	rs1085307580	Neutral	0.719	0.197	0.084
KCNQ1	167	V	M	NM_000218.3(KCNQ1):c.880G>A (p.Val294Met)	Arrhythmia	Uncertain significance(Last reviewed: Oct 30, 2018)	criteria provided, single submitter	VCV000629619	11	2572945	629619	617853	rs749073770	Neutral	0.744	0.159	0.097
KCNQ1	168	G	R	NM_000218.3(KCNQ1):c.502G>C (p.Gly168Arg)	Cardiovascular phenotype|not provided| Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Aug 11, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053053	11	2570652	53053	67721	rs179489	LOF	0.909	0.017	0.074
KCNQ1	168	G	R	NM_000218.3(KCNQ1):c.502G>C (p.Gly168Arg)	Cardiovascular phenotype|not provided| Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Aug 11, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053053	11	2570652	53053	67721	rs179489	LOF	0.909	0.017	0.074
KCNQ1	168	G	R	NM_000218.3(KCNQ1):c.502G>A (p.Gly168Arg)	Long QT syndrome 1|Long QT syndrome|not provided|Congenital long QT syndrome| Arrhythmia|Cardiovascular phenotype	Pathogenic(Last reviewed: Oct 28, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053052	11	2570652	53052	67720	rs179489	LOF	0.909	0.017	0.074
KCNQ1	168	G	R	NM_000218.3(KCNQ1):c.502G>A (p.Gly168Arg)	Long QT syndrome 1|Long QT syndrome|not provided|Congenital long QT syndrome| Arrhythmia|Cardiovascular phenotype	Pathogenic(Last reviewed: Oct 28, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053052	11	2570652	53052	67720	rs179489	LOF	0.909	0.017	0.074
KCNQ1	169	T	M	NM_000218.3(KCNQ1):c.506C>T (p.Thr169Met)	Long QT syndrome 1|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Jan 4, 2021)	criteria provided, multiple submitters, no conflicts	VCV000405270	11	2570656	405270	398114	rs199472693	LOF	0.73	0.11	0.161

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	169	T	R	NM_000218.3(KCNQ1):c.506C>G (p.Thr169Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053055	11	2570656	53055	67723	rs199472693	LOF	0.842	0.065	0.093
KCNQ1	171	Y	H	NM_000218.3(KCNQ1):c.511T>C (p.Tyr171His)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV001034572	11	2570661	1034572	1030172	rs1848317948	LOF	0.869	0.044	0.087
KCNQ1	172	V	M	NM_000218.3(KCNQ1):c.514G>A (p.Val172Met)	Arrhythmia|Cardiovascular phenotype|Short QT syndrome 2|Atrial fibrillation, familial, 3|Jervell and Lange-Nielsen syndrome 1|Congenital long QT syndrome|not provided|Long QT syndrome 1|Long QT syndrome|Atrial fibrillation, familial, 3|Beckwith-Wiedemann syndrome|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome 1|Short QT syndrome 2|not specified	Conflicting interpretations of pathogenicity(Last reviewed: Jul 26, 2021)	criteria provided, conflicting interpretations	VCV000067078	11	2570664	67078	77974	rs199472694	Neutral	0.626	0.286	0.088
KCNQ1	173	A	S	NM_000218.3(KCNQ1):c.898G>T (p.Ala300Ser)	Arrhythmia	Uncertain significance(Last reviewed: Nov 20, 2018)	criteria provided, single submitter	VCV000919940	11	2572963	919940	911578	rs120074187	Neutral	0.659	0.228	0.113
KCNQ1	173	V	D	NM_000218.3(KCNQ1):c.518T>A (p.Val173Asp)	Congenital long QT syndrome|Recurrent spontaneous abortion	Pathogenic(Last reviewed: Mar 1, 2021)	criteria provided, single submitter	VCV000053057	11	2570668	53057	67725	rs199472695	LOF	0.858	0.058	0.085
KCNQ1	174	D	V	NM_000218.3(KCNQ1):c.902A>T (p.Asp301Val)	Cardiovascular phenotype|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Sep 9, 2019)	criteria provided, conflicting interpretations	VCV000519367	11	2572967	519367	510276	rs1554893260	LOF	0.801	0.035	0.164
KCNQ1	174	R	C	NM_000218.3(KCNQ1):c.520C>T (p.Arg174Cys)	Long QT syndrome|not provided|Congenital long QT syndrome|Arrhythmia|Long QT syndrome 1	Pathogenic/Likely pathogenic(Last reviewed: Dec 14, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053058	11	2570670	53058	67726	rs199472696	LOF	0.887	0.013	0.1
KCNQ1	174	R	H	NM_000218.3(KCNQ1):c.521G>A (p.Arg174His)	Congenital long QT syndrome|not provided|Long QT syndrome|Long QT syndrome 1	Pathogenic/Likely pathogenic(Last reviewed: Oct 7, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053059	11	2570671	53059	67727	rs199472697	LOF	0.862	0.031	0.106
KCNQ1	174	R	L	NM_000218.3(KCNQ1):c.521G>T (p.Arg174Leu)	not provided	Likely pathogenic(Last reviewed: Jun 17, 2014)	criteria provided, single submitter	VCV000200814	11	2570671	200814	197429	rs199472697	LOF	0.767	0.032	0.201
KCNQ1	174	R	P	NM_000218.3(KCNQ1):c.521G>C (p.Arg174Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053060	11	2570671	53060	67728	rs199472697	LOF	0.845	0.039	0.116
KCNQ1	175	A	V	NM_000218.3(KCNQ1):c.905C>T (p.Ala302Val)	not provided|Long QT syndrome 1|Cardiovascular phenotype|Long QT syndrome|Congenital long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Dec 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000036439	11	2572970	36439	45102	rs193922365	Neutral	0.698	0.175	0.127
KCNQ1	175	L	F	NM_000218.3(KCNQ1):c.523C>T (p.Leu175Phe)	Long QT syndrome 1	Uncertain significance(Last reviewed: Nov 16, 2016)	criteria provided, single submitter	VCV000431030	11	2570673	431030	424564	rs1131692322	LOF	0.772	0.134	0.094
KCNQ1	176	W	R	NM_000218.3(KCNQ1):c.526T>C (p.Trp176Arg)	not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 21, 2019)	criteria provided, conflicting interpretations	VCV000449041	11	2570676	449041	444782	rs1554892900	LOF	0.857	0.046	0.097
KCNQ1	177	W	S	NM_000218.3(KCNQ1):c.911G>C (p.Trp304Ser)	not provided	Likely pathogenic(Last reviewed: Apr 12, 2021)	criteria provided, single submitter	VCV001212402	11	2572976	1212402	1202396	NA	LOF	0.897	0.026	0.078
KCNQ1	178	A	P	NM_000218.3(KCNQ1):c.532G>C (p.Ala178Pro)	Congenital long QT syndrome|Long QT syndrome 1	Pathogenic(Last reviewed: Jan 1,	no assertion criteria provided	VCV000003113	11	2570682	3113	18152	rs120074177	LOF	0.876	0.037	0.086

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						1996)											
KCNQ1	178	A	T	NM_000218.3(KCNQ1):c.532G>A (p.Ala178Thr)	Cardiovascular phenotype|Long QT syndrome| not provided|Congenital long QT syndrome|Long QT syndrome 1|Long QT syndrome 1|Jervell and Lange-Nielsen syndrome 1	Pathogenic/Likely pathogenic(Last reviewed: May 4, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053061	11	2570682	53061	67729	rs120074177	LOF	0.856	0.048	0.096
KCNQ1	178	W	L	NM_000218.3(KCNQ1):c.914G>T (p.Trp305Leu)	not provided|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome 1	Pathogenic(Last reviewed: Jan 3, 2022)	criteria provided, multiple submitters, no conflicts	VCV000200824	11	2572979	200824	197448	rs120074186	LOF	0.874	0.03	0.096
KCNQ1	179	G	A	NM_000218.3(KCNQ1):c.536G>C (p.Gly179Ala)	not provided	Likely pathogenic(Last reviewed: Jan 11, 2018)	criteria provided, single submitter	VCV000378906	11	2570686	378906	372137	rs76737438	LOF	0.894	0.024	0.081
KCNQ1	179	G	E	NM_000218.3(KCNQ1):c.917G>A (p.Gly306Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 30, 2020)	criteria provided, single submitter	VCV000998695	11	2572982	998695	994446	rs199472742	LOF	0.915	0.014	0.071
KCNQ1	179	G	S	NM_000218.3(KCNQ1):c.535G>A (p.Gly179Ser)	Congenital long QT syndrome|Long QT syndrome 1|not provided|Arrhythmia| Cardiovascular phenotype|Long QT syndrome	Pathogenic(Last reviewed: Aug 30, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053063	11	2570685	53063	67731	rs199473394	LOF	0.902	0.023	0.075
KCNQ1	180	V	E	NM_000218.3(KCNQ1):c.920T>A (p.Val307Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 1, 2019)	criteria provided, single submitter	VCV000849152	11	2572985	849152	838239	rs1589958619	LOF	0.791	0.067	0.142
KCNQ1	181	R	C	NM_000218.3(KCNQ1):c.541C>T (p.Arg181Cys)	Arrhythmia|not provided	Uncertain significance(Last reviewed: Oct 30, 2018)	criteria provided, single submitter	VCV000067079	11	2570691	67079	77975	rs199473395	LOF	0.835	0.068	0.097
KCNQ1	183	K	R	NM_000218.3(KCNQ1):c.548A>G (p.Lys183Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067080	11	2570698	67080	77976	rs199473396	LOF	0.84	0.073	0.087
KCNQ1	184	Y	D	NM_000218.3(KCNQ1):c.550T>G (p.Tyr184Asp)	not provided	Likely pathogenic(Last reviewed: May 2, 2016)	criteria provided, single submitter	VCV000200815	11	2570700	200815	197430	rs199473661	LOF	0.752	0.073	0.175
KCNQ1	184	Y	H	NM_000218.3(KCNQ1):c.550T>C (p.Tyr184His)	Congenital long QT syndrome|not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jun 22, 2020)	criteria provided, conflicting interpretations	VCV000067081	11	2570700	67081	77977	rs199473661	LOF	0.685	0.136	0.18
KCNQ1	184	Y	S	NM_000218.3(KCNQ1):c.551A>C (p.Tyr184Ser)	Long QT syndrome 1|Congenital long QT syndrome	Pathogenic(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000053064	11	2570701	53064	67732	rs199473397	LOF	0.667	0.122	0.211
KCNQ1	185	T	S	NM_000218.3(KCNQ1):c.934A>T (p.Thr312Ser)	Long QT syndrome 1	Likely pathogenic	no assertion criteria provided	VCV000522585	11	2583447	522585	513199	rs1554894445	Neutral	0.498	0.304	0.199
KCNQ1	185	V	M	NM_000218.3(KCNQ1):c.553G>A (p.Val185Met)	Arrhythmia	Uncertain significance(Last reviewed: Jun 19, 2019)	criteria provided, single submitter	VCV000919842	11	2570703	919842	911555	rs749351255	Neutral	0.572	0.256	0.173
KCNQ1	186	G	A	NM_000218.3(KCNQ1):c.557G>C (p.Gly186Ala)	not specified	Uncertain significance(Last reviewed: Oct 19, 2021)	criteria provided, single submitter	VCV001321388	11	2570707	1321388	1311672	NA	LOF	0.784	0.074	0.142
KCNQ1	186	G	C	NM_000218.3(KCNQ1):c.556G>T (p.Gly186Cys)	not provided	Likely pathogenic(Last reviewed: May 2, 2016)	criteria provided, single submitter	VCV000432149	11	2570706	432149	425920	rs199473398	LOF	0.845	0.045	0.11

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	186	G	D	NM_000218.3(KCNQ1):c.557G>A (p.Gly186Asp)	Arrhythmia|not provided|Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 1	Likely pathogenic(Last reviewed: Apr 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000207967	11	2570707	207967	204198	rs794728568	LOF	0.85	0.04	0.11
KCNQ1	186	G	R	NM_000218.3(KCNQ1):c.556G>C (p.Gly186Arg)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Oct 31, 2019)	criteria provided, single submitter	VCV000053065	11	2570706	53065	67733	rs199473398	LOF	0.83	0.034	0.136
KCNQ1	186	G	S	NM_000218.3(KCNQ1):c.556G>A (p.Gly186Ser)	Congenital long QT syndrome|Long QT syndrome 1	Uncertain significance(Last reviewed: Dec 17, 2018)	criteria provided, single submitter	VCV000067082	11	2570706	67082	77978	rs199473398	LOF	0.795	0.076	0.129
KCNQ1	186	G	V	NM_000218.3(KCNQ1):c.557G>T (p.Gly186Val)	not provided	Pathogenic(Last reviewed: Nov 28, 2011)	criteria provided, single submitter	VCV000200894	11	2570707	200894	197432	rs794728568	LOF	0.782	0.07	0.148
KCNQ1	187	L	F	NM_000218.3(KCNQ1):c.559C>T (p.Leu187Phe)	not provided	Uncertain significance(Last reviewed: Oct 3, 2019)	criteria provided, single submitter	VCV001309157	11	2570709	1309157	1299857	NA	Neutral	0.488	0.401	0.111
KCNQ1	187	L	P	NM_000218.3(KCNQ1):c.560T>C (p.Leu187Pro)	not provided|Congenital long QT syndrome|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Sep 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053066	11	2570710	53066	67734	rs199473399	Neutral	0.484	0.403	0.113
KCNQ1	187	L	R	NM_000218.3(KCNQ1):c.560T>G (p.Leu187Arg)	Long QT syndrome	Uncertain significance(Last reviewed: May 30, 2019)	criteria provided, single submitter	VCV000843851	11	2570710	843851	838234	rs199473399	Neutral	0.657	0.233	0.11
KCNQ1	188	W	S	NM_000218.3(KCNQ1):c.563G>C (p.Trp188Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 30, 2018)	criteria provided, single submitter	VCV000643351	11	2570713	643351	639904	rs1240024663	LOF	0.77	0.114	0.116
KCNQ1	188	Y	H	NM_000218.3(KCNQ1):c.943T>C (p.Tyr315His)	not provided	Pathogenic(Last reviewed: Sep 11, 2017)	criteria provided, single submitter	VCV000449302	11	2583456	449302	444785	rs1554894448	Neutral	0.61	0.263	0.126
KCNQ1	189	G	E	NM_000218.3(KCNQ1):c.566G>A (p.Gly189Glu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053068	11	2570716	53068	67736	rs199473400	LOF	0.908	0.018	0.074
KCNQ1	189	G	R	NM_000218.3(KCNQ1):c.565G>C (p.Gly189Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067083	11	2570715	67083	77979	rs104894252	LOF	0.903	0.017	0.08
KCNQ1	189	G	R	NM_000218.3(KCNQ1):c.565G>C (p.Gly189Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067083	11	2570715	67083	77979	rs104894252	LOF	0.903	0.017	0.08
KCNQ1	189	G	R	NM_000218.3(KCNQ1):c.565G>A (p.Gly189Arg)	not provided|Congenital long QT syndrome|Long QT syndrome 1|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jun 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003114	11	2570715	3114	18153	rs104894252	LOF	0.903	0.017	0.08
KCNQ1	189	G	R	NM_000218.3(KCNQ1):c.565G>A (p.Gly189Arg)	not provided|Congenital long QT syndrome|Long QT syndrome 1|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jun 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003114	11	2570715	3114	18153	rs104894252	LOF	0.903	0.017	0.08
KCNQ1	189	G	W	NM_000218.3(KCNQ1):c.946G>T (p.Gly316Trp)	not provided	Pathogenic(Last reviewed: Nov 18, 2013)	criteria provided, single submitter	VCV000200826	11	2583459	200826	197451	rs104894255	LOF	0.834	0.021	0.145
KCNQ1	190	D	A	NM_000218.3(KCNQ1):c.950A>C (p.Asp317Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 8, 2020)	criteria provided, single submitter	VCV001056776	11	2583463	1056776	1047163	NA	LOF	0.823	0.049	0.128
KCNQ1	190	R	L	NM_000218.3(KCNQ1):c.569G>T (p.Arg190Leu)	Long QT syndrome|Arrhythmia|not provided|Long QT syndrome 1|Congenital long QT	Pathogenic/Likely pathogenic(Last	criteria provided,	VCV000067084	11	2570719	67084	77980	rs120074178	LOF	0.747	0.043	0.209

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					syndrome	reviewed: Aug 27, 2019)	multiple submitters, no conflicts										
KCNQ1	190	R	P	NM_000218.3(KCNQ1):c.569G>C (p.Arg190Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 11, 2016)	criteria provided, single submitter	VCV000237228	11	2570719	237228	241085	rs120074178	LOF	0.842	0.05	0.108
KCNQ1	190	R	Q	NM_000218.3(KCNQ1):c.569G>A (p.Arg190Gln)	not provided|Long QT syndrome|Cardiovascular phenotype|Long QT syndrome 1|Arrhythmia|Congenital long QT syndrome	Pathogenic(Last reviewed: Nov 29, 2021)	criteria provided, multiple submitters, no conflicts	VCV000003117	11	2570719	3117	18156	rs120074178	LOF	0.855	0.048	0.097
KCNQ1	190	R	W	NM_000218.3(KCNQ1):c.568C>T (p.Arg190Trp)	Long QT syndrome|Cardiovascular phenotype|not provided|Congenital long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Aug 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053070	11	2570718	53070	67738	rs199473662	LOF	0.791	0.036	0.173
KCNQ1	191	K	T	NM_000218.3(KCNQ1):c.953A>C (p.Lys318Thr)	not provided	Uncertain significance(Last reviewed: Feb 3, 2017)	criteria provided, single submitter	VCV000200827	11	2583466	200827	197452	rs794728516	Neutral	0.724	0.148	0.128
KCNQ1	191	L	P	NM_000218.3(KCNQ1):c.572T>C (p.Leu191Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053071	11	2570722	53071	67739	rs199473401	Neutral	0.7	0.202	0.098
KCNQ1	192	R	H	NM_000218.3(KCNQ1):c.575G>A (p.Arg192His)	Arrhythmia|Congenital long QT syndrome|Long QT syndrome|not provided|Long QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Apr 15, 2021)	criteria provided, conflicting interpretations	VCV000067085	11	2570725	67085	77981	rs199472698	LOF	0.778	0.094	0.128
KCNQ1	192	R	P	NM_000218.3(KCNQ1):c.575G>C (p.Arg192Pro)	Long QT syndrome|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Nov 8, 2019)	criteria provided, multiple submitters, no conflicts	VCV000053073	11	2570725	53073	67741	rs199472698	LOF	0.75	0.114	0.136
KCNQ1	193	F	L	NM_000218.3(KCNQ1):c.579T>A (p.Phe193Leu)	not provided	not provided	no assertion provided	VCV000067086	11	2570729	67086	77982	rs199473454	LOF	0.778	0.105	0.117
KCNQ1	193	F	L	NM_000218.3(KCNQ1):c.579T>A (p.Phe193Leu)	not provided	not provided	no assertion provided	VCV000067086	11	2570729	67086	77982	rs199473454	LOF	0.778	0.105	0.117
KCNQ1	193	F	L	NM_000218.3(KCNQ1):c.577T>C (p.Phe193Leu)	Long QT syndrome 1	not provided	no assertion provided	VCV000053074	11	2570727	53074	67742	rs397508119	LOF	0.778	0.105	0.117
KCNQ1	193	F	L	NM_000218.3(KCNQ1):c.577T>C (p.Phe193Leu)	Long QT syndrome 1	not provided	no assertion provided	VCV000053074	11	2570727	53074	67742	rs397508119	LOF	0.778	0.105	0.117
KCNQ1	194	A	P	NM_000218.3(KCNQ1):c.580G>C (p.Ala194Pro)	Congenital long QT syndrome|Long QT syndrome|not provided	Uncertain significance(Last reviewed: Aug 22, 2019)	criteria provided, multiple submitters, no conflicts	VCV000053075	11	2570730	53075	67743	rs199472699	Neutral	0.729	0.168	0.103
KCNQ1	194	Q	P	NM_000218.3(KCNQ1):c.962A>C (p.Gln321Pro)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200829	11	2583475	200829	197454	rs794728518	LOF	0.797	0.087	0.116
KCNQ1	195	R	P	NM_000218.3(KCNQ1):c.584G>C (p.Arg195Pro)	Long QT syndrome	Uncertain significance	criteria provided, single submitter	VCV000684829	11	2570734	684829	672420	rs138362632	LOF	0.874	0.035	0.092
KCNQ1	195	R	Q	NM_000218.3(KCNQ1):c.584G>A (p.Arg195Gln)	Cardiovascular phenotype|not specified|not provided|Long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Apr 20, 2020)	criteria provided, conflicting interpretations	VCV000067088	11	2570734	67088	77984	rs138362632	LOF	0.884	0.031	0.085
KCNQ1	195	R	W	NM_000218.3(KCNQ1):c.583C>T (p.Arg195Trp)	Arrhythmia|Congenital long QT syndrome|Long	Uncertain	criteria	VCV000067087	11	2570733	67087	77983	rs150172393	LOF	0.821	0.04	0.139

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					QT syndrome	significance(Last reviewed: Oct 14, 2020)	provided, multiple submitters, no conflicts										
KCNQ1	195	T	R	NM_000218.3(KCNQ1):c.965C>G (p.Thr322Arg)	not provided	Pathogenic(Last reviewed: May 17, 2013)	criteria provided, single submitter	VCV000200830	11	2583478	200830	197455	rs199472755	LOF	0.864	0.047	0.089
KCNQ1	196	K	Q	NM_000218.3(KCNQ1):c.586A>C (p.Lys196Gln)	Autosomal dominant KCNQ1-related disease	Uncertain significance(Last reviewed: Aug 25, 2019)	no assertion criteria provided	VCV000800853	11	2570736	800853	788850	rs1589956747	LOF	0.864	0.049	0.087
KCNQ1	196	K	T	NM_000218.3(KCNQ1):c.587A>C (p.Lys196Thr)	not provided	Uncertain significance(Last reviewed: Jul 9, 2020)	criteria provided, single submitter	VCV000200816	11	2570737	200816	197434	rs794728509	LOF	0.862	0.048	0.089
KCNQ1	197	P	H	NM_000218.3(KCNQ1):c.590C>A (p.Pro197His)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 12, 2017)	criteria provided, single submitter	VCV000527016	11	2570740	527016	526103	rs200108320	LOF	0.876	0.021	0.104
KCNQ1	197	P	L	NM_000218.3(KCNQ1):c.590C>T (p.Pro197Leu)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Feb 19, 2021)	criteria provided, multiple submitters, no conflicts	VCV000191476	11	2570740	191476	189328	rs200108320	LOF	0.738	0.043	0.219
KCNQ1	197	P	S	NM_000218.3(KCNQ1):c.589C>T (p.Pro197Ser)	not provided	Likely pathogenic(Last reviewed: Aug 2, 2019)	criteria provided, single submitter	VCV000200817	11	2570739	200817	197435	rs794728510	LOF	0.868	0.03	0.102
KCNQ1	197	V	F	NM_000218.3(KCNQ1):c.970G>T (p.Val324Phe)	Arrhythmia	Uncertain significance(Last reviewed: Nov 25, 2019)	criteria provided, single submitter	VCV000924962	11	2583483	924962	911584	rs1848536066	LOF	0.734	0.11	0.156
KCNQ1	198	G	E	NM_000218.3(KCNQ1):c.974G>A (p.Gly325Glu)	not provided	Likely pathogenic(Last reviewed: Aug 14, 2019)	criteria provided, single submitter	VCV000200831	11	2583487	200831	197457	rs794728519	LOF	0.862	0.024	0.113
KCNQ1	198	G	R	NM_000218.3(KCNQ1):c.973G>C (p.Gly325Arg)	Long QT syndrome	Pathogenic(Last reviewed: Aug 20, 2018)	criteria provided, single submitter	VCV000660595	11	2583486	660595	639910	rs199472756	LOF	0.867	0.02	0.113
KCNQ1	198	I	V	NM_000218.3(KCNQ1):c.592A>G (p.Ile198Val)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Feb 5, 2021)	criteria provided, single submitter	VCV000067089	11	2570742	67089	77985	rs199472700	Neutral	0.7	0.208	0.092
KCNQ1	199	S	A	NM_000218.3(KCNQ1):c.595T>G (p.Ser199Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067090	11	2570745	67090	77986	rs199472701	GOF	0.557	0.115	0.328
KCNQ1	202	A	T	NM_000218.3(KCNQ1):c.985G>A (p.Ala329Thr)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Jul 19, 2017)	criteria provided, single submitter	VCV000518688	11	2583498	518688	510277	rs543428644	Neutral	0.496	0.279	0.224
KCNQ1	202	A	V	NM_000218.3(KCNQ1):c.986C>T (p.Ala329Val)	Arrhythmia	Uncertain significance(Last reviewed: Nov 24, 2020)	criteria provided, single submitter	VCV001171771	11	2583499	1171771	1160872	NA	Neutral	0.423	0.337	0.24
KCNQ1	202	D	G	NM_000218.3(KCNQ1):c.605A>G (p.Asp202Gly)	not provided	Pathogenic(Last reviewed: Oct 25, 2011)	criteria provided, single submitter	VCV000200895	11	2571325	200895	197437	rs794728569	LOF	0.769	0.054	0.177
KCNQ1	202	D	H	NM_000218.3(KCNQ1):c.604G>C (p.Asp202His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067092	11	2570754	67092	77988	rs199472702	LOF	0.707	0.058	0.235
KCNQ1	202	D	N	NM_000218.3(KCNQ1):c.604G>A (p.Asp202Asn)	Long QT syndrome|not provided|Cardiovascular phenotype|Congenital long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Dec 14,	criteria provided, multiple	VCV000053077	11	2570754	53077	67745	rs199472702	LOF	0.779	0.057	0.164

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)	submitters, no conflicts										
KCNQ1	203	L	P	NM_000218.3(KCNQ1):c.608T>C (p.Leu203Pro)	Congenital long QT syndrome|Marfan syndrome	Likely pathogenic(Last reviewed: Sep 30, 2016)	criteria provided, single submitter	VCV000067093	11	2571328	67093	77989	rs199472823	LOF	0.837	0.076	0.087
KCNQ1	204	I	F	NM_000218.3(KCNQ1):c.610A>T (p.Ile204Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053078	11	2571330	53078	67746	rs199472703	Neutral	0.647	0.258	0.094
KCNQ1	204	I	M	NM_000218.3(KCNQ1):c.612C>G (p.Ile204Met)	not provided|Long QT syndrome|Congenital long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Mar 19, 2020)	criteria provided, conflicting interpretations	VCV000053079	11	2571332	53079	67747	rs199473455	Neutral	0.616	0.3	0.084
KCNQ1	205	V	M	NM_000218.3(KCNQ1):c.613G>A (p.Val205Met)	Long QT syndrome|Congenital long QT syndrome|not provided|Long QT syndrome 1|Cardiovascular phenotype	Pathogenic(Last reviewed: Oct 27, 2021)	criteria provided, multiple submitters, no conflicts	VCV000037255	11	2571333	37255	45847	rs151344631	Neutral	0.516	0.34	0.144
KCNQ1	207	V	M	NM_000218.3(KCNQ1):c.619G>A (p.Val207Met)	not specified|Arrhythmia|Long QT syndrome|not provided	Likely benign(Last reviewed: May 12, 2021)	criteria provided, multiple submitters, no conflicts	VCV000042492	11	2571339	42492	51662	rs75813654	Neutral	0.475	0.414	0.111
KCNQ1	209	S	F	NM_000218.3(KCNQ1):c.626C>T (p.Ser209Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053080	11	2571346	53080	67748	rs199472704	GOF	0.438	0.093	0.469
KCNQ1	209	S	P	NM_000218.3(KCNQ1):c.625T>C (p.Ser209Pro)	Atrial fibrillation, familial, 3|Atrial fibrillation|Long QT syndrome	Pathogenic(Last reviewed: Jun 24, 2016)	criteria provided, single submitter	VCV000067094	11	2571345	67094	77990	rs199472705	GOF	0.604	0.096	0.3
KCNQ1	210	I	F	NM_000218.3(KCNQ1):c.1009A>T (p.Ile337Phe)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Apr 17, 2019)	criteria provided, conflicting interpretations	VCV000200832	11	2583522	200832	197459	rs794728520	Neutral	0.584	0.301	0.115
KCNQ1	210	I	M	NM_000218.3(KCNQ1):c.1011C>G (p.Ile337Met)	not provided	Uncertain significance(Last reviewed: Feb 3, 2013)	criteria provided, single submitter	VCV000200833	11	2583524	200833	197460	rs794728521	Neutral	0.55	0.349	0.102
KCNQ1	210	M	T	NM_000218.3(KCNQ1):c.629T>C (p.Met210Thr)	Cardiovascular phenotype	Uncertain significance(Last reviewed: May 6, 2015)	criteria provided, single submitter	VCV000263972	11	2571349	263972	258691	rs886038994	Neutral	0.616	0.247	0.137
KCNQ1	210	M	V	NM_000218.3(KCNQ1):c.628A>G (p.Met210Val)	Arrhythmia	Uncertain significance(Last reviewed: Jan 8, 2020)	criteria provided, single submitter	VCV000922727	11	2571348	922727	911558	rs1848330783	Neutral	0.485	0.391	0.124
KCNQ1	214	C	W	NM_000218.3(KCNQ1):c.642C>G (p.Cys214Trp)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 24, 2019)	criteria provided, single submitter	VCV000942858	11	2571362	942858	935470	rs775479779	LOF	0.773	0.046	0.181
KCNQ1	215	V	G	NM_000218.3(KCNQ1):c.644T>G (p.Val215Gly)	Long QT syndrome 1|Atrial fibrillation, familial, 3|Beckwith-Wiedemann syndrome|Short QT syndrome 2|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome	Uncertain significance(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000456868	11	2571364	456868	461128	rs368011737	LOF	0.796	0.098	0.106
KCNQ1	215	V	M	NM_000218.3(KCNQ1):c.643G>A (p.Val215Met)	Long QT syndrome|not provided|Congenital long QT syndrome	Uncertain significance(Last reviewed: May 18, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053081	11	2571363	53081	67749	rs17215479	Neutral	0.476	0.405	0.119
KCNQ1	217	A	G	NM_000218.3(KCNQ1):c.1031C>G (p.Ala344Gly)	not provided	Likely pathogenic(Last	criteria provided, single	VCV000200900	11	2583544	200900	197463	rs199472763	LOF	0.827	0.074	0.1

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Jul 5, 2016)	submitter										
KCNQ1	217	A	T	NM_000218.3(KCNQ1):c.1030G>A (p.Ala344Thr)	not provided	Likely pathogenic(Last reviewed: Apr 8, 2019)	criteria provided, single submitter	VCV000449042	11	2583543	449042	444786	rs1554894481	LOF	0.751	0.124	0.125
KCNQ1	217	S	F	NM_000218.3(KCNQ1):c.650C>T (p.Ser217Phe)	short QT syndrome|Jervell and Lange-Nielsen syndrome|Familial atrial fibrillation|Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Jun 14, 2016)	criteria provided, single submitter	VCV000304217	11	2571370	304217	327120	rs886048163	LOF	0.727	0.088	0.184
KCNQ1	218	G	R	NM_000218.3(KCNQ1):c.1033G>A (p.Gly345Arg)	Long QT syndrome|not provided	Pathogenic(Last reviewed: Jun 16, 2020)	criteria provided, single submitter	VCV000200837	11	2585212	200837	197468	rs199473471	LOF	0.877	0.026	0.097
KCNQ1	218	K	E	NM_000218.3(KCNQ1):c.652A>G (p.Lys218Glu)	Torsades de pointes|not provided	Uncertain significance(Last reviewed: Nov 6, 2017)	criteria provided, single submitter	VCV000067095	11	2571372	67095	77991	rs36210419	LOF	0.883	0.03	0.087
KCNQ1	218	K	T	NM_000218.3(KCNQ1):c.653A>C (p.Lys218Thr)	Arrhythmia	Uncertain significance(Last reviewed: Jul 20, 2019)	criteria provided, single submitter	VCV000925518	11	2571373	925518	911560	rs1848331549	LOF	0.874	0.038	0.088
KCNQ1	220	Q	K	NM_000218.3(KCNQ1):c.658C>A (p.Gln220Lys)	Arrhythmia	Uncertain significance(Last reviewed: Oct 17, 2019)	criteria provided, single submitter	VCV000927874	11	2571378	927874	911561	rs755789171	LOF	0.807	0.054	0.139
KCNQ1	223	G	V	NM_000218.3(KCNQ1):c.1049G>T (p.Gly350Val)	Long QT syndrome|not provided	Likely pathogenic(Last reviewed: Dec 9, 2016)	criteria provided, multiple submitters, no conflicts	VCV000200838	11	2585228	200838	197469	rs794728524	LOF	0.81	0.032	0.158
KCNQ1	224	F	L	NM_000218.3(KCNQ1):c.1051T>C (p.Phe351Leu)	Long QT syndrome 1|not provided	Uncertain significance(Last reviewed: Jan 9, 2020)	criteria provided, single submitter	VCV000207968	11	2585230	207968	204200	rs779383393	LOF	0.787	0.082	0.131
KCNQ1	224	T	M	NM_000218.3(KCNQ1):c.671C>T (p.Thr224Met)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Feb 24, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067096	11	2571391	67096	77992	rs199472706	LOF	0.719	0.059	0.221
KCNQ1	225	S	L	NM_000218.3(KCNQ1):c.674C>T (p.Ser225Leu)	not provided|Long QT syndrome 1|Long QT syndrome|Congenital long QT syndrome| Arrhythmia	Pathogenic/Likely pathogenic(Last reviewed: Oct 28, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053083	11	2571394	53083	67751	rs199473456	GOF	0.649	0.059	0.292
KCNQ1	226	A	V	NM_000218.3(KCNQ1):c.677C>T (p.Ala226Val)	Congenital long QT syndrome|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Oct 29, 2018)	criteria provided, multiple submitters, no conflicts	VCV000067097	11	2571397	67097	77993	rs199472707	LOF	0.774	0.115	0.111
KCNQ1	229	G	D	NM_000218.3(KCNQ1):c.686G>A (p.Gly229Asp)	Atrial fibrillation, familial, 3|not provided| Congenital long QT syndrome|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: May 26, 2020)	criteria provided, conflicting interpretations	VCV000053085	11	2572015	53085	67753	rs199472708	LOF	0.92	0.015	0.065
KCNQ1	230	Q	E	NM_000218.3(KCNQ1):c.1069C>G (p.Gln357Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 29, 2017)	criteria provided, single submitter	VCV000526978	11	2585248	526978	526549	rs1554894743	LOF	0.903	0.018	0.079
KCNQ1	230	Q	H	NM_000218.2(KCNQ1):c.1071G>Y (p.Gln357His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000132649	11	2585250	132649	136397	rs758766691	LOF	0.888	0.021	0.091
KCNQ1	231	R	C	NM_000218.3(KCNQ1):c.691C>T (p.Arg231Cys)	Congenital long QT syndrome|Long QT syndrome 1|Long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last	criteria provided,	VCV000053086	11	2572020	53086	67754	rs199473457	LOF	0.905	0.011	0.084

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Oct 1, 2021)	multiple submitters, no conflicts										
KCNQ1	231	R	H	NM_000218.3(KCNQ1):c.692G>A (p.Arg231His)	Atrial fibrillation, familial, 3|Long QT syndrome| Atrial fibrillation, familial, 3|Beckwith-Wiedemann syndrome|Long QT syndrome 1| Jervell and Lange-Nielsen syndrome 1|Short QT syndrome 2|not provided|Long QT syndrome 1| Congenital long QT syndrome	Pathogenic(Last reviewed: Dec 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053087	11	2572021	53087	67755	rs199472709	LOF	0.889	0.011	0.1
KCNQ1	232	Q	P	NM_000218.3(KCNQ1):c.1076A>C (p.Gln359Pro)	not provided	Likely pathogenic(Last reviewed: Jun 3, 2015)	criteria provided, single submitter	VCV000427114	11	2585255	427114	415265	rs1085307965	LOF	0.898	0.022	0.08
KCNQ1	233	R	K	NM_000218.3(KCNQ1):c.1079G>A (p.Arg360Lys)	Arrhythmia	Uncertain significance(Last reviewed: Oct 14, 2020)	criteria provided, single submitter	VCV001172261	11	2585258	1172261	1160873	NA	LOF	0.87	0.033	0.098
KCNQ1	233	R	W	NM_000218.3(KCNQ1):c.1078A>T (p.Arg360Trp)	not provided	Likely pathogenic(Last reviewed: Feb 26, 2018)	criteria provided, single submitter	VCV000200839	11	2585257	200839	197470	rs199473406	LOF	0.817	0.025	0.158
KCNQ1	235	I	N	NM_000218.3(KCNQ1):c.704T>A (p.Ile235Asn)	Congenital long QT syndrome|Long QT syndrome|Arrhythmia	Pathogenic(Last reviewed: Aug 31, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053088	11	2572033	53088	67756	rs199472710	LOF	0.909	0.014	0.077
KCNQ1	239	L	P	NM_000218.3(KCNQ1):c.716T>C (p.Leu239Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053089	11	2572045	53089	67757	rs199473458	LOF	0.836	0.082	0.082
KCNQ1	239	R	L	NM_000218.3(KCNQ1):c.1097G>T (p.Arg366Leu)	not provided	Likely pathogenic(Last reviewed: Jan 13, 2020)	criteria provided, single submitter	VCV001185948	11	2585276	1185948	1177444	NA	LOF	0.792	0.043	0.164
KCNQ1	240	Q	H	NM_000218.3(KCNQ1):c.1101G>C (p.Gln367His)	not provided	Uncertain significance(Last reviewed: Apr 27, 2021)	criteria provided, single submitter	VCV001203515	11	2585280	1203515	1194487	NA	LOF	0.878	0.016	0.106
KCNQ1	240	Q	R	NM_000218.3(KCNQ1):c.1100A>G (p.Gln367Arg)	not provided	Uncertain significance(Last reviewed: Oct 7, 2020)	criteria provided, single submitter	VCV001215084	11	2585279	1215084	1205067	NA	LOF	0.882	0.016	0.102
KCNQ1	241	V	G	NM_000218.3(KCNQ1):c.722T>G (p.Val241Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067098	11	2572051	67098	77994	rs199472711	LOF	0.876	0.04	0.083
KCNQ1	242	D	E	NM_000218.3(KCNQ1):c.726C>A (p.Asp242Glu)	not provided|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Sep 28, 2021)	criteria provided, conflicting interpretations	VCV000665471	11	2572055	665471	639906	rs1589957756	LOF	0.913	0.02	0.066
KCNQ1	242	D	N	NM_000218.3(KCNQ1):c.724G>A (p.Asp242Asn)	Wolff-Parkinson-White pattern|Congenital long QT syndrome|Long QT syndrome|not provided	Pathogenic(Last reviewed: Jul 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053090	11	2572053	53090	67758	rs199472712	LOF	0.907	0.028	0.065
KCNQ1	242	D	Y	NM_000218.3(KCNQ1):c.724G>T (p.Asp242Tyr)	Congenital long QT syndrome|not provided|Long QT syndrome	Likely pathogenic(Last reviewed: Jun 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067099	11	2572053	67099	77995	rs199472712	LOF	0.846	0.034	0.12
KCNQ1	242	P	A	NM_000218.3(KCNQ1):c.1105C>G (p.Pro369Ala)	Inborn genetic diseases|not provided|not specified	Conflicting interpretations of pathogenicity(Last reviewed: Dec 6, 2017)	criteria provided, conflicting interpretations	VCV000200840	11	2585284	200840	197472	rs794728525	LOF	0.871	0.038	0.091

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	242	P	L	NM_000218.3(KCNQ1):c.1106C>T (p.Pro369Leu)	not provided	Uncertain significance(Last reviewed: Feb 3, 2022)	criteria provided, single submitter	VCV001339593	11	2585285	1339593	1330759	NA	LOF	0.819	0.042	0.139
KCNQ1	243	A	E	NM_000218.3(KCNQ1):c.1109C>A (p.Ala370Glu)	Long QT syndrome 1	Likely pathogenic(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV000869435	11	2585288	869435	857652	rs775362401	LOF	0.916	0.017	0.068
KCNQ1	243	A	V	NM_000218.3(KCNQ1):c.1109C>T (p.Ala370Val)	not provided|Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Mar 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000629754	11	2585288	629754	617855	rs775362401	LOF	0.88	0.04	0.08
KCNQ1	243	R	C	NM_000218.3(KCNQ1):c.727C>T (p.Arg243Cys)	not provided|Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Oct 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053091	11	2572056	53091	67759	rs199472713	LOF	0.899	0.02	0.081
KCNQ1	243	R	H	NM_000218.3(KCNQ1):c.728G>A (p.Arg243His)	Congenital long QT syndrome|Arrhythmia| Jervell and Lange-Nielsen syndrome 1|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jan 3, 2022)	criteria provided, conflicting interpretations	VCV000053092	11	2572057	53092	67760	rs120074196	LOF	0.902	0.014	0.084
KCNQ1	243	R	P	NM_000218.3(KCNQ1):c.728G>C (p.Arg243Pro)	Long QT syndrome 1/2, digenic|Congenital long QT syndrome	Pathogenic(Last reviewed: Sep 1, 2006)	no assertion criteria provided	VCV000003150	11	2572057	3150	18189	rs120074196	LOF	0.907	0.016	0.077
KCNQ1	243	R	S	NM_000218.3(KCNQ1):c.727C>A (p.Arg243Ser)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Oct 26, 2017)	criteria provided, single submitter	VCV000067100	11	2572056	67100	77996	rs199472713	LOF	0.911	0.013	0.075
KCNQ1	244	A	P	NM_000218.3(KCNQ1):c.1111G>C (p.Ala371Pro)	Long QT syndrome	Likely pathogenic(Last reviewed: Sep 13, 2019)	criteria provided, single submitter	VCV000237221	11	2585290	237221	241086	rs199473412	LOF	0.876	0.051	0.073
KCNQ1	245	G	R	NM_000218.3(KCNQ1):c.733G>A (p.Gly245Arg)	Long QT syndrome	Uncertain significance(Last reviewed: May 12, 2019)	criteria provided, single submitter	VCV000950580	11	2572062	950580	926184	rs1060500628	LOF	0.88	0.022	0.098
KCNQ1	248	I	F	NM_000218.3(KCNQ1):c.1123A>T (p.Ile375Phe)	not provided	Uncertain significance(Last reviewed: Apr 22, 2020)	criteria provided, single submitter	VCV000200841	11	2585302	200841	197474	rs794728526	LOF	0.828	0.084	0.089
KCNQ1	248	W	C	NM_000218.3(KCNQ1):c.744G>C (p.Trp248Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067102	11	2572073	67102	77998	rs199472714	LOF	0.917	0.017	0.066
KCNQ1	248	W	R	NM_000218.3(KCNQ1):c.742T>C (p.Trp248Arg)	Congenital long QT syndrome|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 24, 2017)	criteria provided, conflicting interpretations	VCV000067101	11	2572071	67101	77997	rs199473459	LOF	0.89	0.019	0.09
KCNQ1	249	Q	E	NM_000218.3(KCNQ1):c.1126C>G (p.Gln376Glu)	not specified|Long QT syndrome	Uncertain significance(Last reviewed: Sep 11, 2019)	criteria provided, multiple submitters, no conflicts	VCV000811417	11	2585305	811417	799643	rs1564825414	LOF	0.808	0.043	0.148
KCNQ1	249	Q	H	NM_000218.3(KCNQ1):c.1128G>T (p.Gln376His)	not specified	Uncertain significance(Last reviewed: Sep 3, 2019)	criteria provided, single submitter	VCV000928773	11	2585307	928773	917084	rs1848577607	LOF	0.773	0.055	0.172
KCNQ1	250	L	H	NM_000218.3(KCNQ1):c.749T>A (p.Leu250His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053094	11	2572078	53094	67762	rs199472715	LOF	0.879	0.028	0.093
KCNQ1	250	L	P	NM_000218.3(KCNQ1):c.749T>C	Congenital long QT syndrome|Long QT	Uncertain	criteria	VCV000067103	11	2572078	67103	77999	rs199472715	LOF	0.868	0.044	0.088

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Leu250Pro)	syndrome	significance(Last reviewed: Nov 29, 2019)	provided, single submitter										
KCNQ1	251	A	S	NM_000218.3(KCNQ1):c.1132G>T (p.Ala378Ser)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 10, 2019)	criteria provided, single submitter	VCV000948540	11	2587573	948540	926185	rs756571615	LOF	0.757	0.113	0.13
KCNQ1	251	L	P	NM_000218.3(KCNQ1):c.752T>C (p.Leu251Pro)	not provided|Congenital long QT syndrome|Long QT syndrome 1	Pathogenic(Last reviewed: Jun 26, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053095	11	2572081	53095	67763	rs199472716	LOF	0.838	0.063	0.099
KCNQ1	253	R	K	NM_000218.3(KCNQ1):c.1139G>A (p.Arg380Lys)	Arrhythmia	Uncertain significance(Last reviewed: Nov 17, 2020)	criteria provided, single submitter	VCV001171179	11	2587580	1171179	1160874	NA	LOF	0.85	0.021	0.129
KCNQ1	254	C	Y	NM_000218.3(KCNQ1):c.1142G>A (p.Cys381Tyr)	not provided	Likely pathogenic(Last reviewed: Apr 22, 2013)	criteria provided, single submitter	VCV000200844	11	2587583	200844	197476	rs368507376	LOF	0.822	0.027	0.151
KCNQ1	254	V	L	NM_000218.3(KCNQ1):c.760G>T (p.Val254Leu)	Cardiovascular phenotype|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Jan 4, 2017)	criteria provided, single submitter	VCV000053096	11	2572089	53096	67764	rs120074179	LOF	0.779	0.102	0.119
KCNQ1	254	V	L	NM_000218.3(KCNQ1):c.760G>T (p.Val254Leu)	Cardiovascular phenotype|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Jan 4, 2017)	criteria provided, single submitter	VCV000053096	11	2572089	53096	67764	rs120074179	LOF	0.779	0.102	0.119
KCNQ1	254	V	L	NM_000218.3(KCNQ1):c.760G>C (p.Val254Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067104	11	2572089	67104	78000	rs120074179	LOF	0.779	0.102	0.119
KCNQ1	254	V	L	NM_000218.3(KCNQ1):c.760G>C (p.Val254Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067104	11	2572089	67104	78000	rs120074179	LOF	0.779	0.102	0.119
KCNQ1	254	V	M	NM_000218.2(KCNQ1):c.[1249G>A;760G>A]	Long QT syndrome 1	Pathogenic(Last reviewed: Mar 1, 2004)	no assertion criteria provided	VCV000444879	11|11	2572089	444879	18157|38428	rs120074179|rs267607197	LOF	0.82	0.071	0.109
KCNQ1	254	V	M	NM_000218.2(KCNQ1):c.[1249G>A;760G>A]	Long QT syndrome 1	Pathogenic(Last reviewed: Mar 1, 2004)	no assertion criteria provided	VCV000444879	11|11	2572089	444879	18157|38428	rs120074179|rs267607197	LOF	0.82	0.071	0.109
KCNQ1	254	V	M	NM_000218.3(KCNQ1):c.760G>A (p.Val254Met)	Long QT syndrome|Cardiovascular phenotype|Long QT syndrome 1|Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Oct 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003118	11	2572089	3118	18157	rs120074179	LOF	0.82	0.071	0.109
KCNQ1	254	V	M	NM_000218.3(KCNQ1):c.760G>A (p.Val254Met)	Long QT syndrome|Cardiovascular phenotype|Long QT syndrome 1|Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Oct 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003118	11	2572089	3118	18157	rs120074179	LOF	0.82	0.071	0.109
KCNQ1	258	H	N	NM_000218.3(KCNQ1):c.772C>A (p.His258Asn)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053098	11	2572101	53098	67766	rs199472717	LOF	0.799	0.04	0.161
KCNQ1	258	H	P	NM_000218.3(KCNQ1):c.773A>C (p.His258Pro)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Sep 6, 2019)	criteria provided, single submitter	VCV000067105	11	2572102	67105	78001	rs199472718	LOF	0.801	0.046	0.153
KCNQ1	258	H	R	NM_000218.3(KCNQ1):c.773A>G (p.His258Arg)	not provided|Congenital long QT syndrome	Likely pathogenic(Last reviewed: May 1, 2019)	criteria provided, single submitter	VCV000053099	11	2572102	53099	67767	rs199472718	LOF	0.779	0.033	0.188
KCNQ1	259	R	C	NM_000218.3(KCNQ1):c.775C>T (p.Arg259Cys)	Cardiovascular phenotype|not provided|Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 1	Pathogenic(Last reviewed: Dec 23, 2020)	criteria provided, multiple	VCV000053100	11	2572104	53100	67768	rs199472719	LOF	0.871	0.022	0.107

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							submitters, no conflicts										
KCNQ1	259	R	H	NM_000218.3(KCNQ1):c.776G>A (p.Arg259His)	Congenital long QT syndrome|Long QT syndrome 1|Long QT syndrome|Arrhythmia| Atrial fibrillation, familial, 3|Long QT syndrome 1|Short QT syndrome 2|KCNQ1-Related Disorders|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jan 5, 2022)	criteria provided, conflicting interpretations	VCV000053101	11	2572105	53101	67769	rs199472720	LOF	0.808	0.032	0.16
KCNQ1	259	R	L	NM_000218.3(KCNQ1):c.776G>T (p.Arg259Leu)	Congenital long QT syndrome|Long QT syndrome|not provided	Pathogenic(Last reviewed: Dec 28, 2018)	criteria provided, multiple submitters, no conflicts	VCV000053102	11	2572105	53102	67770	rs199472720	LOF	0.766	0.047	0.188
KCNQ1	259	R	P	NM_000218.3(KCNQ1):c.776G>C (p.Arg259Pro)	Long QT syndrome 1	Likely pathogenic(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV000869436	11	2572105	869436	857651	rs199472720	LOF	0.827	0.036	0.137
KCNQ1	261	E	D	NM_000218.3(KCNQ1):c.783G>C (p.Glu261Asp)	Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Sep 19, 2017)	criteria provided, single submitter	VCV000053104	11	2572848	53104	67772	rs199472721	LOF	0.877	0.024	0.099
KCNQ1	261	E	K	NM_000218.3(KCNQ1):c.781G>A (p.Glu261Lys)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Dec 10, 2018)	criteria provided, single submitter	VCV000053103	11	2572846	53103	67771	rs199472722	LOF	0.853	0.027	0.121
KCNQ1	261	E	L	NM_000218.3(KCNQ1):c.781_782delinsTT (p.Glu261Leu)	Long QT syndrome	Pathogenic(Last reviewed: Sep 27, 2017)	criteria provided, single submitter	VCV000405253	11	2572846 - 2572847	405253	398119	rs1060500621	LOF	0.792	0.048	0.16
KCNQ1	261	E	Q	NM_000218.3(KCNQ1):c.781G>C (p.Glu261Gln)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: Sep 19, 2016)	criteria provided, single submitter	VCV000067106	11	2572846	67106	78002	rs199472722	LOF	0.855	0.03	0.115
KCNQ1	262	L	V	NM_000218.3(KCNQ1):c.784C>G (p.Leu262Val)	not provided|Congenital long QT syndrome	Uncertain significance(Last reviewed: Oct 17, 2019)	criteria provided, single submitter	VCV000053105	11	2572849	53105	67773	rs199472723	LOF	0.822	0.108	0.07
KCNQ1	262	S	A	NM_000218.3(KCNQ1):c.1165T>G (p.Ser389Ala)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 16, 2018)	criteria provided, single submitter	VCV000574906	11	2587606	574906	570495	rs199472772	LOF	0.792	0.074	0.134
KCNQ1	262	S	D	NM_000218.3(KCNQ1):c.1165_1166inv (p.Ser389Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 21, 2017)	criteria provided, single submitter	VCV000526914	11	2587606 - 2587607	526914	526107	NA	LOF	0.87	0.041	0.089
KCNQ1	265	T	I	NM_000218.3(KCNQ1):c.794C>T (p.Thr265Ile)	not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: Jun 28, 2012)	criteria provided, single submitter	VCV000053106	11	2572859	53106	67774	rs199472724	LOF	0.809	0.06	0.13
KCNQ1	266	K	E	NM_000218.3(KCNQ1):c.1177A>G (p.Lys393Glu)	Arrhythmia	Uncertain significance(Last reviewed: Apr 14, 2019)	criteria provided, single submitter	VCV000919675	11	2587618	919675	911592	rs1848611273	LOF	0.925	0.014	0.061
KCNQ1	266	L	P	NM_000218.3(KCNQ1):c.797T>C (p.Leu266Pro)	Cardiovascular phenotype|not provided| Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Jun 15, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053108	11	2572862	53108	67776	rs199473460	LOF	0.921	0.017	0.061
KCNQ1	267	I	T	NM_000218.3(KCNQ1):c.1181T>C (p.Ile394Thr)	Arrhythmia	Uncertain significance(Last reviewed: Dec 2, 2019)	criteria provided, single submitter	VCV000923338	11	2587622	923338	911593	rs1437113154	LOF	0.904	0.027	0.069
KCNQ1	268	I	S	NM_000218.3(KCNQ1):c.803T>G (p.Ile268Ser)	Congenital long QT syndrome|not provided|Long QT syndrome	Uncertain significance(Last reviewed: May 17,	criteria provided, multiple	VCV000067107	11	2572868	67107	78003	rs199472725	LOF	0.897	0.041	0.062

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2017)	submitters, no conflicts										
KCNQ1	269	G	D	NM_000218.3(KCNQ1):c.806G>A (p.Gly269Asp)	Long QT syndrome 1|not provided|Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Jul 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000003145	11	2572871	3145	18184	rs120074194	LOF	0.922	0.012	0.066
KCNQ1	269	G	S	NM_000218.3(KCNQ1):c.805G>A (p.Gly269Ser)	not provided|Long QT syndrome|Atrial fibrillation, familial, 3|Beckwith-Wiedemann syndrome|Short QT syndrome 2|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome 1|not specified|Congenital long QT syndrome|Long QT syndrome 1	Pathogenic(Last reviewed: Oct 28, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003144	11	2572870	3144	18183	rs120074193	LOF	0.898	0.027	0.075
KCNQ1	270	R	Q	NM_000218.3(KCNQ1):c.1190G>A (p.Arg397Gln)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Dec 21, 2021)	criteria provided, multiple submitters, no conflicts	VCV000372649	11	2587631	372649	359874	rs374090960	LOF	0.918	0.015	0.067
KCNQ1	272	A	G	NM_000218.3(KCNQ1):c.1196C>G (p.Ala399Gly)	not provided|not specified|Arrhythmia	Uncertain significance(Last reviewed: Aug 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000228760	11	2587637	228760	230194	rs876657836	LOF	0.88	0.044	0.076
KCNQ1	272	A	S	NM_000218.3(KCNQ1):c.1195G>T (p.Ala399Ser)	Arrhythmia|Long QT syndrome|Inborn genetic diseases	Uncertain significance(Last reviewed: Nov 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000985535	11	2587636	985535	973791	rs571899254	LOF	0.857	0.066	0.077
KCNQ1	272	A	V	NM_000218.3(KCNQ1):c.1196C>T (p.Ala399Val)	Arrhythmia|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Jun 29, 2021)	criteria provided, multiple submitters, no conflicts	VCV000918468	11	2587637	918468	911595	rs876657836	LOF	0.811	0.099	0.09
KCNQ1	272	G	D	NM_000218.3(KCNQ1):c.815G>A (p.Gly272Asp)	Long QT syndrome|Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Aug 20, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053109	11	2572880	53109	67777	rs199472726	LOF	0.924	0.012	0.064
KCNQ1	272	G	V	NM_000218.3(KCNQ1):c.815G>T (p.Gly272Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067108	11	2572880	67108	78004	rs199472726	LOF	0.871	0.03	0.099
KCNQ1	273	L	F	NM_000218.3(KCNQ1):c.817C>T (p.Leu273Phe)	Cardiovascular phenotype|Congenital long QT syndrome|not provided|Long QT syndrome 1|Long QT syndrome	Pathogenic(Last reviewed: Oct 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003119	11	2572882	3119	18158	rs120074180	LOF	0.841	0.083	0.076
KCNQ1	273	L	P	NM_000218.3(KCNQ1):c.818T>C (p.Leu273Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 20, 2016)	criteria provided, single submitter	VCV000405254	11	2572883	405254	398124	rs199472727	LOF	0.903	0.035	0.061
KCNQ1	273	L	R	NM_000218.3(KCNQ1):c.818T>G (p.Leu273Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053110	11	2572883	53110	67778	rs199472727	LOF	0.922	0.024	0.054
KCNQ1	273	P	A	NM_000218.3(KCNQ1):c.1198C>G (p.Pro400Ala)	Arrhythmia|Long QT syndrome|Cardiovascular phenotype	Conflicting interpretations of pathogenicity(Last reviewed: Nov 16, 2018)	criteria provided, conflicting interpretations	VCV000518535	11	2587639	518535	510278	rs1489132337	LOF	0.858	0.047	0.095
KCNQ1	274	I	V	NM_000218.3(KCNQ1):c.820A>G (p.Ile274Val)	Long QT syndrome|Short QT syndrome 2|Atrial fibrillation, familial, 3|Jervell and Lange-Nielsen syndrome 1|Congenital long QT syndrome|not specified|Arrhythmia|SUDDEN INFANT DEATH SYNDROME|not provided|Long QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Jul 7, 2021)	criteria provided, conflicting interpretations	VCV000067109	11	2572885	67109	78005	rs199472728	LOF	0.856	0.061	0.083
KCNQ1	274	R	Q	NM_000218.3(KCNQ1):c.1202G>A	not specified|Long QT syndrome	Uncertain	criteria	VCV000228765	11	2587643	228765	230195	rs542628042	LOF	0.92	0.014	0.067

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Arg401Gln)		significance(Last reviewed: Sep 24, 2019)	provided, multiple submitters, no conflicts										
KCNQ1	274	R	W	NM_000218.3(KCNQ1):c.1201C>T (p.Arg401Trp)	not provided|not specified|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Jun 3, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200903	11	2587642	200903	197478	rs766616232	LOF	0.883	0.014	0.103
KCNQ1	275	F	S	NM_000218.3(KCNQ1):c.824T>C (p.Phe275Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053111	11	2572889	53111	67779	rs199472729	LOF	0.925	0.014	0.061
KCNQ1	275	S	N	NM_000218.3(KCNQ1):c.1205G>A (p.Ser402Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Jan 24, 2016)	criteria provided, single submitter	VCV000237222	11	2587646	237222	241087	rs878853753	LOF	0.915	0.018	0.067
KCNQ1	277	S	L	NM_000218.3(KCNQ1):c.830C>T (p.Ser277Leu)	Congenital long QT syndrome|Cardiovascular phenotype|not provided|Long QT syndrome	Pathogenic(Last reviewed: Sep 7, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053116	11	2572895	53116	67784	rs199472730	LOF	0.779	0.035	0.186
KCNQ1	277	S	P	NM_000218.3(KCNQ1):c.829T>C (p.Ser277Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067110	11	2572894	67110	78006	rs199473461	LOF	0.856	0.025	0.119
KCNQ1	277	S	W	NM_000218.3(KCNQ1):c.830C>G (p.Ser277Trp)	Long QT syndrome|Congenital long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last reviewed: Oct 2, 2019)	criteria provided, multiple submitters, no conflicts	VCV000053115	11	2572895	53115	67783	rs199472730	LOF	0.817	0.015	0.168
KCNQ1	277	T	S	NM_000218.3(KCNQ1):c.1211C>G (p.Thr404Ser)	Arrhythmia	Uncertain significance(Last reviewed: Jan 10, 2019)	criteria provided, single submitter	VCV000918527	11	2587652	918527	911597	rs1490239200	LOF	0.849	0.045	0.105
KCNQ1	278	Y	H	NM_000218.3(KCNQ1):c.832T>C (p.Tyr278His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067111	11	2572897	67111	78007	rs199472731	LOF	0.909	0.023	0.067
KCNQ1	279	F	I	NM_000218.3(KCNQ1):c.835T>A (p.Phe279Ile)	Short QT syndrome 2	Pathogenic(Last reviewed: Feb 9, 2017)	no assertion criteria provided	VCV000375594	11	2572900	375594	362405	rs1057519584	LOF	0.847	0.083	0.07
KCNQ1	280	V	E	NM_000218.3(KCNQ1):c.839T>A (p.Val280Glu)	Congenital long QT syndrome|Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: Jul 29, 2019)	criteria provided, multiple submitters, no conflicts	VCV000053117	11	2572904	53117	67785	rs199473462	LOF	0.904	0.027	0.069
KCNQ1	281	Y	C	NM_000218.3(KCNQ1):c.842A>G (p.Tyr281Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067112	11	2572907	67112	78008	rs199472732	LOF	0.924	0.009	0.067
KCNQ1	282	L	P	NM_000218.3(KCNQ1):c.845T>C (p.Leu282Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067113	11	2572910	67113	78009	rs199472733	LOF	0.874	0.057	0.069
KCNQ1	283	A	G	NM_000218.3(KCNQ1):c.848C>G (p.Ala283Gly)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Aug 7, 2017)	criteria provided, single submitter	VCV000067114	11	2572913	67114	78010	rs199473463	LOF	0.813	0.079	0.108
KCNQ1	284	E	G	NM_000218.3(KCNQ1):c.851A>G (p.Glu284Gly)	Long QT syndrome	Uncertain significance(Last reviewed: Nov 11, 2019)	criteria provided, single submitter	VCV000965412	11	2572916	965412	956453	rs1848362105	LOF	0.861	0.036	0.103
KCNQ1	284	E	K	NM_000218.3(KCNQ1):c.850G>A (p.Glu284Lys)	Long QT syndrome|Congenital long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 6, 2020)	criteria provided, conflicting interpretations	VCV000067115	11	2572915	67115	78011	rs199472734	LOF	0.852	0.038	0.11
KCNQ1	287	A	E	NM_000218.3(KCNQ1):c.860C>A (p.Ala287Glu)	Congenital long QT syndrome|Arrhythmia|not specified|Cardiovascular phenotype|Long QT	Uncertain significance(Last	criteria provided,	VCV000053118	11	2572925	53118	67786	rs199472735	LOF	0.801	0.054	0.146

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					syndrome|not provided	reviewed: Oct 9, 2020)	multiple submitters, no conflicts										
KCNQ1	289	V	M	NM_000218.3(KCNQ1):c.1246G>A (p.Val416Met)	Arrhythmia	Uncertain significance(Last reviewed: Apr 2, 2019)	criteria provided, single submitter	VCV000926680	11	2587687	926680	911601	rs777546418	Neutral	0.689	0.169	0.142
KCNQ1	290	E	K	NM_000218.3(KCNQ1):c.868G>A (p.Glu290Lys)	Congenital long QT syndrome|not specified|Long QT syndrome 1|Arrhythmia|Jervell and Lange-Nielsen syndrome 1|Atrial fibrillation, familial, 3|Short QT syndrome 2	Uncertain significance(Last reviewed: May 19, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053120	11	2572933	53120	67788	rs199473464	LOF	0.843	0.052	0.105
KCNQ1	291	V	I	NM_000218.3(KCNQ1):c.1252G>A (p.Val418Ile)	not provided|Arrhythmia	Uncertain significance(Last reviewed: Sep 16, 2020)	criteria provided, multiple submitters, no conflicts	VCV000927875	11	2588713	927875	911603	rs777347006	LOF	0.807	0.09	0.104
KCNQ1	292	G	D	NM_000218.3(KCNQ1):c.875G>A (p.Gly292Asp)	Arrhythmia|Cardiovascular phenotype|Congenital long QT syndrome|not provided|Long QT syndrome	Uncertain significance(Last reviewed: May 13, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067116	11	2572940	67116	78012	rs199472736	LOF	0.875	0.045	0.079
KCNQ1	292	K	R	NM_000218.3(KCNQ1):c.1256A>G (p.Lys419Arg)	Arrhythmia	Uncertain significance(Last reviewed: Oct 26, 2018)	criteria provided, single submitter	VCV000629983	11	2588717	629983	617856	rs757328965	LOF	0.857	0.053	0.089
KCNQ1	292	K	T	NM_000218.3(KCNQ1):c.1256A>C (p.Lys419Thr)	not provided	Uncertain significance(Last reviewed: May 1, 2017)	criteria provided, single submitter	VCV000501370	11	2588717	501370	492794	rs757328965	LOF	0.861	0.05	0.089
KCNQ1	293	R	C	NM_000218.3(KCNQ1):c.877C>T (p.Arg293Cys)	Arrhythmia|Cardiovascular phenotype|Atrial fibrillation, familial, 3|Congenital long QT syndrome|Long QT syndrome|not provided|Long QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Jul 26, 2021)	criteria provided, conflicting interpretations	VCV000067117	11	2572942	67117	78013	rs199472737	LOF	0.745	0.139	0.116
KCNQ1	293	R	H	NM_000218.3(KCNQ1):c.878G>A (p.Arg293His)	not specified|Arrhythmia|not provided|Long QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Nov 4, 2020)	criteria provided, conflicting interpretations	VCV000067118	11	2572943	67118	78014	rs199473465	LOF	0.722	0.152	0.126
KCNQ1	296	F	S	NM_000218.3(KCNQ1):c.887T>C (p.Phe296Ser)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Aug 26, 2020)	criteria provided, single submitter	VCV000067119	11	2572952	67119	78015	rs199472738	LOF	0.722	0.041	0.237
KCNQ1	297	G	S	NM_000218.3(KCNQ1):c.889G>A (p.Gly297Ser)	Long QT syndrome|not specified|not provided	Uncertain significance(Last reviewed: Jul 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067120	11	2572954	67120	78016	rs34320941	LOF	0.749	0.057	0.194
KCNQ1	300	A	E	NM_000218.3(KCNQ1):c.899C>A (p.Ala300Glu)	Long QT syndrome|not provided	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000405257	11	2572964	405257	398596	rs1001293702	LOF	0.908	0.017	0.074
KCNQ1	300	A	T	NM_000218.3(KCNQ1):c.898G>A (p.Ala300Thr)	Long QT syndrome|Cardiovascular phenotype|Atrial fibrillation, familial, 3|Short QT syndrome 2|Long QT syndrome 1|Jervell and Lange-Nielsen syndrome 1|not specified|not provided|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Nov 4, 2020)	criteria provided, conflicting interpretations	VCV000003128	11	2572963	3128	18167	rs120074187	LOF	0.874	0.041	0.085
KCNQ1	302	A	E	NM_000218.3(KCNQ1):c.905C>A (p.Ala302Glu)	Long QT syndrome|Congenital long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last	criteria provided, conflicting	VCV000067121	11	2572970	67121	78017	rs193922365	LOF	0.918	0.014	0.068

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: May 30, 2018)	interpretations										
KCNQ1	302	A	T	NM_000218.3(KCNQ1):c.904G>A (p.Ala302Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053121	11	2572969	53121	67789	rs199472739	LOF	0.886	0.033	0.081
KCNQ1	302	N	S	NM_000218.3(KCNQ1):c.1286A>G (p.Asn429Ser)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Nov 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000579711	11	2588747	579711	565639	rs1464968488	LOF	0.911	0.014	0.075
KCNQ1	303	L	P	NM_000218.3(KCNQ1):c.908T>C (p.Leu303Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067122	11	2572973	67122	78018	rs199472740	LOF	0.856	0.06	0.084
KCNQ1	304	W	R	NM_000218.3(KCNQ1):c.910T>C (p.Trp304Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053122	11	2572975	53122	67790	rs199473466	LOF	0.892	0.016	0.092
KCNQ1	305	T	I	NM_000218.3(KCNQ1):c.1295C>T (p.Thr432Ile)	not specified	Uncertain significance(Last reviewed: Jun 21, 2016)	criteria provided, single submitter	VCV000505109	11	2588756	505109	496996	rs751644427	LOF	0.791	0.037	0.173
KCNQ1	305	W	R	NM_000218.3(KCNQ1):c.913T>C (p.Trp305Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053123	11	2572978	53123	67791	rs199472741	LOF	0.904	0.011	0.085
KCNQ1	305	W	S	NM_000218.3(KCNQ1):c.914G>C (p.Trp305Ser)	Long QT syndrome|Jervell and Lange-Nielsen syndrome 1|Cardiovascular phenotype| Congenital long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last reviewed: Dec 20, 2018)	criteria provided, multiple submitters, no conflicts	VCV000003127	11	2572979	3127	18166	rs120074186	LOF	0.908	0.012	0.08
KCNQ1	306	G	R	NM_000218.3(KCNQ1):c.916G>A (p.Gly306Arg)	not provided|Long QT syndrome 1|Congenital long QT syndrome	Pathogenic(Last reviewed: Aug 3, 2021)	criteria provided, single submitter	VCV000003115	11	2572981	3115	18154	rs120074181	LOF	0.92	0.008	0.072
KCNQ1	306	G	R	NM_000218.3(KCNQ1):c.916G>A (p.Gly306Arg)	not provided|Long QT syndrome 1|Congenital long QT syndrome	Pathogenic(Last reviewed: Aug 3, 2021)	criteria provided, single submitter	VCV000003115	11	2572981	3115	18154	rs120074181	LOF	0.92	0.008	0.072
KCNQ1	306	G	R	NM_000218.3(KCNQ1):c.916G>C (p.Gly306Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067123	11	2572981	67123	78019	rs120074181	LOF	0.92	0.008	0.072
KCNQ1	306	G	R	NM_000218.3(KCNQ1):c.916G>C (p.Gly306Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067123	11	2572981	67123	78019	rs120074181	LOF	0.92	0.008	0.072
KCNQ1	306	G	V	NM_000218.3(KCNQ1):c.917G>T (p.Gly306Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053125	11	2572982	53125	67793	rs199472742	LOF	0.876	0.015	0.109
KCNQ1	307	V	L	NM_000218.3(KCNQ1):c.919G>T (p.Val307Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 1, 2020)	criteria provided, single submitter	VCV000859740	11	2572984	859740	838238	rs120074195	LOF	0.789	0.078	0.133
KCNQ1	307	V	L	NM_000218.3(KCNQ1):c.919G>T (p.Val307Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Jul 1, 2020)	criteria provided, single submitter	VCV000859740	11	2572984	859740	838238	rs120074195	LOF	0.789	0.078	0.133
KCNQ1	307	V	L	NM_000218.3(KCNQ1):c.919G>C (p.Val307Leu)	short QT syndrome|Short QT syndrome 2	Pathogenic(Last reviewed: May 25, 2004)	no assertion criteria provided	VCV000003148	11	2572984	3148	18187	rs120074195	LOF	0.789	0.078	0.133
KCNQ1	307	V	L	NM_000218.3(KCNQ1):c.919G>C (p.Val307Leu)	short QT syndrome|Short QT syndrome 2	Pathogenic(Last reviewed: May 25, 2004)	no assertion criteria provided	VCV000003148	11	2572984	3148	18187	rs120074195	LOF	0.789	0.078	0.133
KCNQ1	307	V	M	NM_000218.3(KCNQ1):c.919G>A (p.Val307Met)	Arrhythmia	Uncertain significance(Last reviewed: Mar 16, 2019)	criteria provided, single submitter	VCV000922206	11	2572984	922206	911581	rs120074195	LOF	0.827	0.061	0.112
KCNQ1	308	V	D	NM_000218.3(KCNQ1):c.923T>A (p.Val308Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053130	11	2583436	53130	67798	rs199473467	LOF	0.913	0.014	0.073
KCNQ1	309	T	I	NM_000218.3(KCNQ1):c.926C>T (p.Thr309Ile)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Jul 29, 2020)	criteria provided, single submitter	VCV000053132	11	2583439	53132	67800	rs199472743	LOF	0.803	0.032	0.165

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	309	T	R	NM_000218.3(KCNQ1):c.926C>G (p.Thr309Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053131	11	2583439	53131	67799	rs199472743	LOF	0.875	0.019	0.106
KCNQ1	309	T	S	NM_000218.3(KCNQ1):c.925A>T (p.Thr309Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000068483	11	2583438	68483	79375	rs199473468	LOF	0.848	0.039	0.113
KCNQ1	310	V	D	NM_000218.3(KCNQ1):c.929T>A (p.Val310Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067124	11	2583442	67124	78020	rs199472744	LOF	0.896	0.023	0.08
KCNQ1	310	V	I	NM_000218.3(KCNQ1):c.928G>A (p.Val310Ile)	Congenital long QT syndrome|not provided	Likely pathogenic(Last reviewed: Jul 28, 2017)	criteria provided, single submitter	VCV000053133	11	2583441	53133	67801	rs199472745	LOF	0.818	0.076	0.106
KCNQ1	311	T	A	NM_000218.3(KCNQ1):c.931A>G (p.Thr311Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067125	11	2583444	67125	78021	rs199473469	LOF	0.793	0.05	0.157
KCNQ1	311	T	I	NM_000218.3(KCNQ1):c.932C>T (p.Thr311Ile)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053134	11	2583445	53134	67802	rs199472746	LOF	0.779	0.044	0.177
KCNQ1	312	T	I	NM_000218.3(KCNQ1):c.935C>T (p.Thr312Ile)	Long QT syndrome|Long QT syndrome 1| Congenital long QT syndrome|not provided| Arrhythmia	Pathogenic/Likely pathogenic(Last reviewed: Jun 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000003116	11	2583448	3116	18155	rs120074182	LOF	0.906	0.017	0.077
KCNQ1	313	I	F	NM_000218.3(KCNQ1):c.937A>T (p.Ile313Phe)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 11, 2018)	criteria provided, single submitter	VCV000664353	11	2583450	664353	639909	rs1589965249	LOF	0.903	0.022	0.075
KCNQ1	313	I	M	NM_000218.3(KCNQ1):c.939C>G (p.Ile313Met)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053135	11	2583452	53135	67803	rs199472747	LOF	0.906	0.02	0.074
KCNQ1	314	G	A	NM_000218.3(KCNQ1):c.941G>C (p.Gly314Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067126	11	2583454	67126	78022	rs199472748	LOF	0.916	0.014	0.07
KCNQ1	314	G	C	NM_000218.3(KCNQ1):c.940G>T (p.Gly314Cys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053137	11	2583453	53137	67805	rs120074184	LOF	0.923	0.017	0.061
KCNQ1	314	G	D	NM_000218.3(KCNQ1):c.941G>A (p.Gly314Asp)	not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: Jul 9, 2021)	criteria provided, single submitter	VCV000053138	11	2583454	53138	67806	rs199472748	LOF	0.918	0.013	0.069
KCNQ1	314	G	R	NM_000218.3(KCNQ1):c.940G>C (p.Gly314Arg)	Cardiovascular phenotype|Congenital long QT syndrome	Pathogenic(Last reviewed: May 12, 2017)	criteria provided, single submitter	VCV000053136	11	2583453	53136	67804	rs120074184	LOF	0.912	0.013	0.075
KCNQ1	314	G	S	NM_000218.3(KCNQ1):c.940G>A (p.Gly314Ser)	Long QT syndrome|Congenital long QT syndrome|not provided|Long QT syndrome 1	Pathogenic(Last reviewed: Aug 26, 2021)	criteria provided, multiple submitters, no conflicts	VCV000003123	11	2583453	3123	18162	rs120074184	LOF	0.914	0.013	0.073
KCNQ1	314	P	L	NM_000218.3(KCNQ1):c.1322C>T (p.Pro441Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Apr 2, 2019)	no assertion criteria provided	VCV000977165	11	2588783	977165	965308	rs1848630761	LOF	0.915	0.019	0.067
KCNQ1	315	Y	C	NM_000218.3(KCNQ1):c.944A>G (p.Tyr315Cys)	Long QT syndrome|Cardiovascular phenotype| not provided|Congenital long QT syndrome|Long QT syndrome 1	Pathogenic/Likely pathogenic(Last reviewed: Aug 14, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053140	11	2583457	53140	67808	rs74462309	LOF	0.896	0.02	0.084
KCNQ1	315	Y	F	NM_000218.3(KCNQ1):c.944A>T (p.Tyr315Phe)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053141	11	2583457	53141	67809	rs74462309	LOF	0.881	0.03	0.089
KCNQ1	315	Y	S	NM_000218.3(KCNQ1):c.944A>C (p.Tyr315Ser)	Long QT syndrome|Congenital long QT syndrome	Pathogenic(Last reviewed: Mar 9, 2020)	criteria provided, single submitter	VCV000053139	11	2583457	53139	67807	rs74462309	LOF	0.884	0.016	0.1
KCNQ1	316	G	E	NM_000218.3(KCNQ1):c.947G>A (p.Gly316Glu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053144	11	2583460	53144	67812	rs199472749	LOF	0.917	0.013	0.069
KCNQ1	316	G	R	NM_000218.3(KCNQ1):c.946G>A (p.Gly316Arg)	not provided|Congenital long QT syndrome|Long QT syndrome	Conflicting interpretations of	criteria provided,	VCV000053142	11	2583459	53142	67810	rs104894255	LOF	0.914	0.013	0.073

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						pathogenicity(Last reviewed: Oct 5, 2020)	conflicting interpretations										
KCNQ1	316	G	R	NM_000218.3(KCNQ1):c.946G>A (p.Gly316Arg)	not provided|Congenital long QT syndrome|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Oct 5, 2020)	criteria provided, conflicting interpretations	VCV000053142	11	2583459	53142	67810	rs104894255	LOF	0.914	0.013	0.073
KCNQ1	316	G	R	NM_000218.3(KCNQ1):c.946G>C (p.Gly316Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053143	11	2583459	53143	67811	rs104894255	LOF	0.914	0.013	0.073
KCNQ1	316	G	R	NM_000218.3(KCNQ1):c.946G>C (p.Gly316Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053143	11	2583459	53143	67811	rs104894255	LOF	0.914	0.013	0.073
KCNQ1	316	G	V	NM_000218.3(KCNQ1):c.947G>T (p.Gly316Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067127	11	2583460	67127	78023	rs199472749	LOF	0.92	0.015	0.064
KCNQ1	316	I	V	NM_000218.3(KCNQ1):c.1327A>G (p.Ile443Val)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 24, 2020)	criteria provided, single submitter	VCV001018531	11	2588788	1018531	1009623	rs1848631038	LOF	0.906	0.023	0.071
KCNQ1	317	D	G	NM_000218.3(KCNQ1):c.950A>G (p.Asp317Gly)	Long QT syndrome|Congenital long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last reviewed: Aug 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067129	11	2583463	67129	78025	rs199472750	LOF	0.875	0.052	0.074
KCNQ1	317	D	N	NM_000218.3(KCNQ1):c.949G>A (p.Asp317Asn)	Long QT syndrome|Cardiovascular phenotype| not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: Jun 27, 2019)	criteria provided, multiple submitters, no conflicts	VCV000053145	11	2583462	53145	67813	rs199472751	LOF	0.879	0.044	0.077
KCNQ1	317	D	Y	NM_000218.3(KCNQ1):c.949G>T (p.Asp317Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067128	11	2583462	67128	78024	rs199472751	LOF	0.865	0.053	0.082
KCNQ1	318	K	N	NM_000218.3(KCNQ1):c.954G>C (p.Lys318Asn)	not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: Jun 21, 2013)	criteria provided, single submitter	VCV000053146	11	2583467	53146	67814	rs199472752	LOF	0.875	0.039	0.086
KCNQ1	319	D	E	NM_000218.3(KCNQ1):c.1338C>A (p.Asp446Glu)	Arrhythmia	Likely benign(Last reviewed: Mar 19, 2019)	criteria provided, single submitter	VCV000926541	11	2588799	926541	911612	rs199472780	LOF	0.861	0.029	0.11
KCNQ1	319	D	N	NM_000218.3(KCNQ1):c.1336G>A (p.Asp446Asn)	Arrhythmia|not provided	Uncertain significance(Last reviewed: Apr 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000200846	11	2588797	200846	197481	rs149089817	LOF	0.844	0.039	0.117
KCNQ1	319	V	M	NM_000218.3(KCNQ1):c.955G>A (p.Val319Met)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, single submitter	VCV001044172	11	2583468	1044172	1030174	rs1310931869	LOF	0.757	0.086	0.157
KCNQ1	320	P	A	NM_000218.3(KCNQ1):c.958C>G (p.Pro320Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053147	11	2583471	53147	67815	rs199472753	LOF	0.876	0.026	0.099
KCNQ1	320	P	H	NM_000218.3(KCNQ1):c.1340C>A (p.Pro447His)	Cardiovascular phenotype|Long QT syndrome	Uncertain significance(Last reviewed: Sep 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000264157	11	2588801	264157	258695	rs143149582	LOF	0.888	0.021	0.091
KCNQ1	320	P	H	NM_000218.3(KCNQ1):c.1340C>A (p.Pro447His)	Cardiovascular phenotype|Long QT syndrome	Uncertain significance(Last reviewed: Sep 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000264157	11	2588801	264157	258695	rs143149582	LOF	0.888	0.021	0.091
KCNQ1	320	P	H	NM_000218.3(KCNQ1):c.959C>A (p.Pro320His)	Long QT syndrome|Congenital long QT syndrome	Pathogenic(Last reviewed: Aug 26, 2018)	criteria provided, single submitter	VCV000053148	11	2583472	53148	67816	rs199473470	LOF	0.888	0.021	0.091

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	320	P	H	NM_000218.3(KCNQ1):c.959C>A (p.Pro320His)	Long QT syndrome|Congenital long QT syndrome	Pathogenic(Last reviewed: Aug 26, 2018)	criteria provided, single submitter	VCV000053148	11	2583472	53148	67816	rs199473470	LOF	0.888	0.021	0.091
KCNQ1	320	P	S	NM_000218.3(KCNQ1):c.958C>T (p.Pro320Ser)	Long QT syndrome|not specified|Congenital long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Nov 15, 2021)	criteria provided, conflicting interpretations	VCV000067130	11	2583471	67130	78026	rs199472753	LOF	0.881	0.024	0.095
KCNQ1	321	P	Q	NM_000218.3(KCNQ1):c.1343C>A (p.Pro448Gln)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 25, 2015)	criteria provided, single submitter	VCV000219923	11	2588804	219923	222140	rs12720449	LOF	0.809	0.054	0.137
KCNQ1	322	T	A	NM_000218.3(KCNQ1):c.964A>G (p.Thr322Ala)	Long QT syndrome|Long QT syndrome 1|not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: Dec 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053149	11	2583477	53149	67817	rs199472754	LOF	0.873	0.032	0.095
KCNQ1	322	T	K	NM_000218.3(KCNQ1):c.965C>A (p.Thr322Lys)	Long QT syndrome 1|Congenital long QT syndrome	Likely pathogenic(Last reviewed: May 27, 2019)	criteria provided, single submitter	VCV000053150	11	2583478	53150	67818	rs199472755	LOF	0.882	0.028	0.089
KCNQ1	322	T	M	NM_000218.3(KCNQ1):c.965C>T (p.Thr322Met)	not provided|Cardiovascular phenotype|Long QT syndrome|Congenital long QT syndrome	Pathogenic(Last reviewed: Nov 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053151	11	2583478	53151	67819	rs199472755	LOF	0.865	0.037	0.098
KCNQ1	322	T	P	NM_000218.3(KCNQ1):c.964A>C (p.Thr322Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 22, 2019)	criteria provided, single submitter	VCV000960695	11	2583477	960695	947394	rs199472754	LOF	0.886	0.033	0.081
KCNQ1	323	E	G	NM_000218.3(KCNQ1):c.1349A>G (p.Glu450Gly)	Arrhythmia	Uncertain significance(Last reviewed: Feb 17, 2014)	criteria provided, single submitter	VCV000374106	11	2588810	374106	360930	rs1057518902	LOF	0.833	0.072	0.095
KCNQ1	323	E	K	NM_000218.3(KCNQ1):c.1348G>A (p.Glu450Lys)	Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 20, 2020)	criteria provided, conflicting interpretations	VCV001002115	11	2588809	1002115	994447	rs148266527	LOF	0.818	0.074	0.108
KCNQ1	325	G	R	NM_000218.3(KCNQ1):c.973G>A (p.Gly325Arg)	not provided|Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 1	Pathogenic(Last reviewed: Aug 28, 2020)	criteria provided, multiple submitters, no conflicts	VCV000053152	11	2583486	53152	67820	rs199472756	LOF	0.89	0.01	0.1
KCNQ1	325	R	L	NM_000218.3(KCNQ1):c.1355G>T (p.Arg452Leu)	not provided	Pathogenic(Last reviewed: Jan 9, 2012)	criteria provided, single submitter	VCV000200904	11	2588816	200904	197482	rs145229963	LOF	0.794	0.022	0.184
KCNQ1	330	S	F	NM_000218.3(KCNQ1):c.1370C>T (p.Ser457Phe)	not provided	Uncertain significance(Last reviewed: Nov 25, 2020)	criteria provided, single submitter	VCV001190302	11	2588831	1190302	1180816	NA	LOF	0.787	0.034	0.179
KCNQ1	331	C	Y	NM_000218.3(KCNQ1):c.992G>A (p.Cys331Tyr)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 22, 2019)	criteria provided, single submitter	VCV000835945	11	2583505	835945	838240	rs1848536656	LOF	0.881	0.018	0.1
KCNQ1	332	D	N	NM_000218.3(KCNQ1):c.1375G>A (p.Asp459Asn)	Long QT syndrome|Atrial fibrillation, familial, 3|Short QT syndrome 2|not provided|Long QT syndrome 1|Jervell and Lange-Nielsen syndrome 1|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Mar 9, 2021)	criteria provided, conflicting interpretations	VCV000628813	11	2588836	628813	617858	rs747704276	LOF	0.92	0.016	0.064
KCNQ1	333	G	D	NM_000218.3(KCNQ1):c.1379G>A (p.Gly460Asp)	not provided|Arrhythmia	Uncertain significance(Last	criteria provided,	VCV000919916	11	2588840	919916	911615	rs770410327	LOF	0.917	0.012	0.07

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Mar 1, 2021)	multiple submitters, no conflicts										
KCNQ1	335	F	L	NM_000218.3(KCNQ1):c.1003T>C (p.Phe335Leu)	not provided	not provided	no assertion provided	VCV000067003	11	2583516	67003	77899	rs199472757	LOF	0.854	0.076	0.07
KCNQ1	336	S	T	NM_000218.3(KCNQ1):c.1388G>C (p.Ser463Thr)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000526963	11	2588849	526963	526552	rs184636161	LOF	0.869	0.031	0.099
KCNQ1	338	S	F	NM_000218.3(KCNQ1):c.1013C>T (p.Ser338Phe)	Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: Sep 26, 2016)	criteria provided, single submitter	VCV000052928	11	2583526	52928	67596	rs199472758	LOF	0.826	0.027	0.148
KCNQ1	339	F	S	NM_000218.3(KCNQ1):c.1016T>C (p.Phe339Ser)	not provided|Congenital long QT syndrome|Long QT syndrome 1	Pathogenic/Likely pathogenic(Last reviewed: Jan 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV000052929	11	2583529	52929	67597	rs199472759	LOF	0.926	0.014	0.06
KCNQ1	339	F	Y	NM_000218.3(KCNQ1):c.1016T>A (p.Phe339Tyr)	Long QT syndrome 1|Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Oct 15, 2018)	criteria provided, multiple submitters, no conflicts	VCV000067004	11	2583529	67004	77900	rs199472759	LOF	0.902	0.029	0.069
KCNQ1	341	A	E	NM_000218.3(KCNQ1):c.1022C>A (p.Ala341Glu)	Cardiovascular phenotype|Long QT syndrome| Long QT syndrome 1|Long QT syndrome 1/2, digenic|Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Jun 10, 2021)	criteria provided, multiple submitters, no conflicts	VCV000003120	11	2583535	3120	18159	rs12720459	LOF	0.909	0.024	0.067
KCNQ1	341	A	G	NM_000218.3(KCNQ1):c.1022C>G (p.Ala341Gly)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: May 30, 2013)	criteria provided, single submitter	VCV000067005	11	2583535	67005	77901	rs12720459	LOF	0.886	0.041	0.073
KCNQ1	341	A	V	NM_000218.3(KCNQ1):c.1022C>T (p.Ala341Val)	Long QT syndrome 1|Long QT syndrome| Cardiovascular phenotype|not provided	Pathogenic(Last reviewed: Oct 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003121	11	2583535	3121	18160	rs12720459	LOF	0.825	0.088	0.087
KCNQ1	342	L	F	NM_000218.3(KCNQ1):c.1024C>T (p.Leu342Phe)	not provided|Cardiovascular phenotype| Congenital long QT syndrome|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jan 31, 2020)	criteria provided, multiple submitters, no conflicts	VCV000052932	11	2583537	52932	67600	rs199472760	LOF	0.851	0.081	0.068
KCNQ1	342	L	H	NM_000218.3(KCNQ1):c.1025T>A (p.Leu342His)	Long QT syndrome	Uncertain significance(Last reviewed: Jun 12, 2017)	criteria provided, single submitter	VCV000405269	11	2583538	405269	398476	rs794728522	LOF	0.918	0.029	0.053
KCNQ1	343	P	L	NM_000218.3(KCNQ1):c.1028C>T (p.Pro343Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052935	11	2583541	52935	67603	rs199472761	LOF	0.847	0.032	0.121
KCNQ1	343	P	R	NM_000218.3(KCNQ1):c.1028C>G (p.Pro343Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052934	11	2583541	52934	67602	rs199472761	LOF	0.928	0.012	0.06
KCNQ1	343	P	S	NM_000218.3(KCNQ1):c.1027C>T (p.Pro343Ser)	Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Sep 8, 2020)	criteria provided, single submitter	VCV000052933	11	2583540	52933	67601	rs199472762	LOF	0.907	0.024	0.069
KCNQ1	344	A	E	NM_000218.3(KCNQ1):c.1031C>A (p.Ala344Glu)	not specified|Long QT syndrome|Congenital long QT syndrome|Cardiovascular phenotype	Pathogenic/Likely pathogenic(Last reviewed: Sep 9, 2019)	criteria provided, multiple submitters, no conflicts	VCV000067006	11	2583544	67006	77902	rs199472763	LOF	0.9	0.028	0.072
KCNQ1	344	A	V	NM_000218.3(KCNQ1):c.1031C>T (p.Ala344Val)	not provided|Long QT syndrome|Congenital long QT syndrome	Pathogenic(Last reviewed: Oct 5, 2020)	criteria provided, multiple	VCV000052936	11	2583544	52936	67604	rs199472763	LOF	0.808	0.099	0.093

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							submitters, no conflicts										
KCNQ1	345	G	E	NM_000218.3(KCNQ1):c.1034G>A (p.Gly345Glu)	Long QT syndrome 1|Congenital long QT syndrome	Pathogenic(Last reviewed: Jan 1, 1996)	no assertion criteria provided	VCV000003122	11	2585213	3122	18161	rs120074183	LOF	0.923	0.011	0.066
KCNQ1	345	G	R	NM_000218.3(KCNQ1):c.1033G>C (p.Gly345Arg)	Long QT syndrome|Congenital long QT syndrome	Pathogenic(Last reviewed: Aug 6, 2015)	criteria provided, single submitter	VCV000052940	11	2585212	52940	67608	rs199473471	LOF	0.925	0.011	0.065
KCNQ1	349	S	P	NM_000218.3(KCNQ1):c.1045T>C (p.Ser349Pro)	Congenital long QT syndrome|Long QT syndrome	Likely pathogenic(Last reviewed: Oct 30, 2017)	criteria provided, single submitter	VCV000052941	11	2585224	52941	67609	rs199472764	LOF	0.81	0.086	0.104
KCNQ1	349	S	W	NM_000218.3(KCNQ1):c.1046C>G (p.Ser349Trp)	Long QT syndrome|not provided|Congenital long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Apr 3, 2019)	criteria provided, multiple submitters, no conflicts	VCV000052942	11	2585225	52942	67610	rs199472765	LOF	0.789	0.045	0.165
KCNQ1	350	G	R	NM_000218.3(KCNQ1):c.1048G>C (p.Gly350Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052943	11	2585227	52943	67611	rs199472824	LOF	0.912	0.021	0.067
KCNQ1	350	G	R	NM_000218.3(KCNQ1):c.1048G>C (p.Gly350Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052943	11	2585227	52943	67611	rs199472824	LOF	0.912	0.021	0.067
KCNQ1	350	G	R	NM_000218.3(KCNQ1):c.1048G>A (p.Gly350Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067007	11	2585227	67007	77903	rs199472824	LOF	0.912	0.021	0.067
KCNQ1	350	G	R	NM_000218.3(KCNQ1):c.1048G>A (p.Gly350Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067007	11	2585227	67007	77903	rs199472824	LOF	0.912	0.021	0.067
KCNQ1	351	F	S	NM_000218.3(KCNQ1):c.1052T>C (p.Phe351Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052944	11	2585231	52944	67612	rs199473402	LOF	0.76	0.149	0.092
KCNQ1	353	L	P	NM_000218.3(KCNQ1):c.1058T>C (p.Leu353Pro)	Congenital long QT syndrome|not provided|Long QT syndrome	Uncertain significance(Last reviewed: Dec 3, 2019)	criteria provided, multiple submitters, no conflicts	VCV000052945	11	2585237	52945	67613	rs199473403	Neutral	0.625	0.281	0.094
KCNQ1	354	K	R	NM_000218.3(KCNQ1):c.1061A>G (p.Lys354Arg)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067008	11	2585240	67008	77904	rs199473404	Neutral	0.71	0.195	0.095
KCNQ1	357	Q	R	NM_000218.3(KCNQ1):c.1070A>G (p.Gln357Arg)	Long QT syndrome|Congenital long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Jul 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000052948	11	2585249	52948	67616	rs199473405	LOF	0.774	0.143	0.083
KCNQ1	360	R	G	NM_000218.3(KCNQ1):c.1078A>G (p.Arg360Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067009	11	2585257	67009	77905	rs199473406	LOF	0.767	0.146	0.087
KCNQ1	360	R	M	NM_000218.3(KCNQ1):c.1079G>T (p.Arg360Met)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067010	11	2585258	67010	77906	rs199473407	Neutral	0.618	0.278	0.104
KCNQ1	360	R	T	NM_000218.3(KCNQ1):c.1079G>C (p.Arg360Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052951	11	2585258	52951	67619	rs199473407	Neutral	0.589	0.315	0.096
KCNQ1	362	K	R	NM_000218.3(KCNQ1):c.1085A>G (p.Lys362Arg)	Long QT syndrome 1|Decreased fetal movement|Abnormality of the nares|Generalized hypotonia|Diffuse white matter abnormalities|Enlarged naris|Abnormal cerebral white matter morphology|Polyhydramnios|High palate|Primary microcephaly|Toe clinodactyly|Generalized neonatal hypotonia|Neonatal hypotonia|Secondary microcephaly|not provided|Congenital long QT syndrome|Arrhythmia|Long QT syndrome|Atrial fibrillation, familial, 3|Long QT syndrome 1|Short QT syndrome 2|Beckwith-Wiedemann syndrome|Jervell and Lange-Nielsen syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Jun 3, 2021)	criteria provided, conflicting interpretations	VCV000052953	11	2585264	52953	67621	rs12720458	LOF	0.771	0.15	0.079
KCNQ1	363	H	N	NM_000218.3(KCNQ1):c.1087C>A	Long QT syndrome|Congenital long QT	Conflicting	criteria	VCV000052954	11	2585266	52954	67622	rs199473408	Neutral	0.728	0.199	0.074

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.His363Asn)	syndrome|not provided	interpretations of pathogenicity(Last reviewed: May 15, 2019)	provided, conflicting interpretations										
KCNQ1	365	N	H	NM_000218.3(KCNQ1):c.1093A>C (p.Asn365His)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067011	11	2585272	67011	77907	rs199473409	Neutral	0.704	0.222	0.074
KCNQ1	366	R	P	NM_000218.3(KCNQ1):c.1097G>C (p.Arg366Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052957	11	2585276	52957	67625	rs199473410	LOF	0.778	0.146	0.075
KCNQ1	366	R	Q	NM_000218.3(KCNQ1):c.1097G>A (p.Arg366Gln)	not provided|Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Sep 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000052956	11	2585276	52956	67624	rs199473410	LOF	0.8	0.13	0.07
KCNQ1	366	R	W	NM_000218.3(KCNQ1):c.1096C>T (p.Arg366Trp)	Cardiovascular phenotype|not provided|Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 1	Pathogenic/Likely pathogenic(Last reviewed: Jan 10, 2022)	criteria provided, multiple submitters, no conflicts	VCV000052955	11	2585275	52955	67623	rs199473411	Neutral	0.659	0.242	0.099
KCNQ1	367	Q	H	NM_000218.3(KCNQ1):c.1101G>T (p.Gln367His)	Long QT syndrome|Congenital long QT syndrome	Uncertain significance(Last reviewed: Mar 25, 2018)	criteria provided, single submitter	VCV000052958	11	2585280	52958	67626	rs199473663	Neutral	0.753	0.172	0.076
KCNQ1	371	A	E	NM_000218.3(KCNQ1):c.1112C>A (p.Ala371Glu)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV001038475	11	2585291	1038475	1030175	rs1848577150	LOF	0.903	0.028	0.069
KCNQ1	371	A	T	NM_000218.3(KCNQ1):c.1111G>A (p.Ala371Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052959	11	2585290	52959	67627	rs199473412	LOF	0.858	0.072	0.07
KCNQ1	372	A	D	NM_000218.3(KCNQ1):c.1115C>A (p.Ala372Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052960	11	2585294	52960	67628	rs199473472	LOF	0.848	0.075	0.077
KCNQ1	373	S	P	NM_000218.3(KCNQ1):c.1117T>C (p.Ser373Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067012	11	2585296	67012	77908	rs199472766	Neutral	0.739	0.17	0.091
KCNQ1	374	L	H	NM_000218.3(KCNQ1):c.1121T>A (p.Leu374His)	Long QT syndrome|not provided|Congenital long QT syndrome|Arrhythmia|Cardiovascular phenotype	Conflicting interpretations of pathogenicity(Last reviewed: Feb 10, 2021)	criteria provided, conflicting interpretations	VCV000052961	11	2585300	52961	67629	rs199472767	LOF	0.878	0.059	0.064
KCNQ1	379	W	G	NM_000218.3(KCNQ1):c.1135T>G (p.Trp379Gly)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Jan 17, 2020)	criteria provided, single submitter	VCV000067014	11	2587576	67014	77910	rs199472768	LOF	0.917	0.023	0.06
KCNQ1	379	W	R	NM_000218.3(KCNQ1):c.1135T>C (p.Trp379Arg)	SUDDEN INFANT DEATH SYNDROME|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: May 26, 2020)	criteria provided, conflicting interpretations	VCV000067013	11	2587576	67013	77909	rs199472768	LOF	0.891	0.025	0.084
KCNQ1	379	W	S	NM_000218.3(KCNQ1):c.1136G>C (p.Trp379Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067015	11	2587577	67015	77911	rs199472769	LOF	0.903	0.028	0.07
KCNQ1	380	R	G	NM_000218.3(KCNQ1):c.1138A>G (p.Arg380Gly)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067016	11	2587579	67016	77912	rs199472770	LOF	0.862	0.073	0.065
KCNQ1	380	R	Q	NM_000218.3(KCNQ1):c.1520G>A (p.Arg507Gln)	Long QT syndrome|Arrhythmia|Cardiovascular phenotype	Uncertain significance(Last reviewed: Apr 20, 2021)	criteria provided, multiple submitters, no conflicts	VCV000519296	11	2768849	519296	510280	rs369571296	LOF	0.821	0.107	0.072
KCNQ1	380	R	S	NM_000218.3(KCNQ1):c.1140G>T (p.Arg380Ser)	Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Aug 22, 2019)	criteria provided, single submitter	VCV000052964	11	2587581	52964	67632	rs199472771	LOF	0.806	0.12	0.074
KCNQ1	380	R	W	NM_000218.3(KCNQ1):c.1138A>T	Long QT syndrome	Uncertain	criteria	VCV000581004	11	2587579	581004	564562	rs199472770	LOF	0.761	0.134	0.104

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Arg380Trp)		significance(Last reviewed: Jun 21, 2019)	provided, single submitter										
KCNQ1	381	C	W	NM_000218.3(KCNQ1):c.1143C>G (p.Cys381Trp)	Arrhythmia	Uncertain significance(Last reviewed: Jan 28, 2019)	criteria provided, single submitter	VCV000928367	11	2587584	928367	911588	rs1848610060	LOF	0.835	0.055	0.11
KCNQ1	383	H	P	NM_000218.3(KCNQ1):c.1529A>C (p.His510Pro)	not provided	Uncertain significance(Last reviewed: Mar 15, 2019)	criteria provided, single submitter	VCV001308222	11	2768858	1308222	1296843	NA	LOF	0.797	0.124	0.079
KCNQ1	384	A	T	NM_000218.3(KCNQ1):c.1150G>A (p.Ala384Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Dec 18, 2019)	criteria provided, single submitter	VCV000842623	11	2587591	842623	838242	rs1848610150	Neutral	0.445	0.486	0.069
KCNQ1	384	R	Q	NM_000218.3(KCNQ1):c.1532G>A (p.Arg511Gln)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 23, 2019)	criteria provided, multiple submitters, no conflicts	VCV000526906	11	2768861	526906	526237	rs753962384	LOF	0.774	0.152	0.074
KCNQ1	385	E	K	NM_000218.3(KCNQ1):c.1153G>A (p.Glu385Lys)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: May 7, 2019)	criteria provided, single submitter	VCV000067017	11	2587594	67017	77913	rs199473473	LOF	0.781	0.145	0.073
KCNQ1	386	T	S	NM_000218.3(KCNQ1):c.1537A>T (p.Thr513Ser)	not specified	Uncertain significance(Last reviewed: Mar 5, 2013)	no assertion criteria provided	VCV000235000	11	2768866	235000	236781	rs778975231	Neutral	0.522	0.383	0.094
KCNQ1	388	D	N	NM_000218.3(KCNQ1):c.1162G>A (p.Asp388Asn)	Arrhythmia|not specified	Uncertain significance(Last reviewed: Apr 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000928161	11	2587603	928161	911590	rs537637760	LOF	0.764	0.12	0.115
KCNQ1	388	K	N	NM_000218.3(KCNQ1):c.1545G>T (p.Lys515Asn)	not provided	Likely pathogenic(Last reviewed: Oct 25, 2013)	criteria provided, single submitter	VCV000200852	11	2768874	200852	197491	rs794728532	LOF	0.749	0.145	0.106
KCNQ1	389	S	P	NM_000218.3(KCNQ1):c.1165T>C (p.Ser389Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067018	11	2587606	67018	77914	rs199472772	Neutral	0.632	0.261	0.106
KCNQ1	389	S	Y	NM_000218.3(KCNQ1):c.1166C>A (p.Ser389Tyr)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Nov 6, 2019)	criteria provided, single submitter	VCV000052966	11	2587607	52966	67634	rs199472773	Neutral	0.488	0.406	0.106
KCNQ1	391	T	I	NM_000218.3(KCNQ1):c.1172C>T (p.Thr391Ile)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052967	11	2587613	52967	67635	rs199473474	Neutral	0.494	0.397	0.109
KCNQ1	392	W	R	NM_000218.3(KCNQ1):c.1174T>C (p.Trp392Arg)	Congenital long QT syndrome|not provided	Pathogenic/Likely pathogenic	no assertion criteria provided	VCV000067019	11	2587615	67019	77915	rs199472774	LOF	0.87	0.036	0.095
KCNQ1	393	K	M	NM_000218.3(KCNQ1):c.1178A>T (p.Lys393Met)	Long QT syndrome|not provided|Congenital long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Oct 11, 2021)	criteria provided, conflicting interpretations	VCV000052969	11	2587619	52969	67637	rs199472775	Neutral	0.475	0.426	0.1
KCNQ1	393	K	N	NM_000218.3(KCNQ1):c.1179G>T (p.Lys393Asn)	Cardiomyopathy|Arrhythmia|not specified|Short QT syndrome 2|Atrial fibrillation, familial, 3| Jervell and Lange-Nielsen syndrome 1|Long QT syndrome|not provided|Long QT syndrome 1| Congenital long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Mar 25, 2021)	criteria provided, conflicting interpretations	VCV000067020	11	2587620	67020	77916	rs12720457	Neutral	0.491	0.415	0.094
KCNQ1	393	M	I	NM_000218.3(KCNQ1):c.1560G>A (p.Met520Ile)	not provided|Long QT syndrome	Uncertain significance(Last reviewed: Nov 5, 2020)	criteria provided, multiple submitters, no	VCV001044140	11	2768889	1044140	1030181	rs1846543163	Neutral	0.339	0.588	0.073

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
							conflicts										
KCNQ1	393	M	T	NM_000218.3(KCNQ1):c.1559T>C (p.Met520Thr)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 14, 2020)	criteria provided, single submitter	VCV001018501	11	2768888	1018501	1009624	rs199473479	Neutral	0.309	0.614	0.077
KCNQ1	397	R	W	NM_000218.3(KCNQ1):c.1189C>T (p.Arg397Trp)	Long QT syndrome 1|Congenital long QT syndrome|not specified|Long QT syndrome|not provided|Arrhythmia|Familial atrial fibrillation| Jervell and Lange-Nielsen syndrome| Cardiovascular phenotype|Wolff-Parkinson-White pattern|short QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Apr 29, 2021)	criteria provided, conflicting interpretations	VCV000052970	11	2587630	52970	67638	rs199472776	Neutral	0.344	0.555	0.101
KCNQ1	398	K	R	NM_000218.3(KCNQ1):c.1193A>G (p.Lys398Arg)	Arrhythmia|not provided|Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Mar 9, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067021	11	2587634	67021	77917	rs199472777	Neutral	0.587	0.298	0.115
KCNQ1	399	K	N	NM_000218.3(KCNQ1):c.1578G>T (p.Lys526Asn)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Oct 25, 2016)	criteria provided, single submitter	VCV000519343	11	2768907	519343	510281	rs1554919483	Neutral	0.475	0.394	0.131
KCNQ1	401	K	R	NM_000218.3(KCNQ1):c.1583A>G (p.Lys528Arg)	Arrhythmia	Uncertain significance(Last reviewed: Aug 20, 2019)	criteria provided, single submitter	VCV000922455	11	2768912	922455	911636	rs1846543781	Neutral	0.52	0.304	0.175
KCNQ1	403	Q	P	NM_000218.3(KCNQ1):c.1589A>C (p.Gln530Pro)	not provided	Uncertain significance(Last reviewed: Apr 23, 2021)	criteria provided, single submitter	VCV000392431	11	2768918	392431	372149	rs1057524487	Neutral	0.358	0.482	0.16
KCNQ1	405	A	V	NM_000218.3(KCNQ1):c.1595C>T (p.Ala532Val)	Long QT syndrome	Uncertain significance(Last reviewed: Mar 28, 2019)	criteria provided, single submitter	VCV000841742	11	2775964	841742	838248	rs867549263	Neutral	0.345	0.52	0.135
KCNQ1	406	R	Q	NM_000218.3(KCNQ1):c.1598G>A (p.Arg533Gln)	not provided|Arrhythmia	Uncertain significance(Last reviewed: Dec 13, 2019)	criteria provided, multiple submitters, no conflicts	VCV000444243	11	2775967	444243	437883	rs574321120	Neutral	0.478	0.361	0.16
KCNQ1	408	P	A	NM_000218.3(KCNQ1):c.1222C>G (p.Pro408Ala)	Long QT syndrome|not specified|Arrhythmia|not provided|Atrial fibrillation, familial, 3|Jervell and Lange-Nielsen syndrome 1|Cardiovascular phenotype|Short QT syndrome 2|Long QT syndrome 1	Benign/Likely benign(Last reviewed: Dec 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000042484	11	2587663	42484	51654	rs28730756	Neutral	0.48	0.308	0.212
KCNQ1	413	K	R	NM_000218.3(KCNQ1):c.1238A>G (p.Lys413Arg)	Arrhythmia	Uncertain significance(Last reviewed: Nov 12, 2020)	criteria provided, single submitter	VCV000919123	11	2587679	919123	911600	rs1351149628	Neutral	0.532	0.282	0.186
KCNQ1	417	Q	L	NM_000218.3(KCNQ1):c.1631A>T (p.Gln544Leu)	not provided	Uncertain significance(Last reviewed: Jul 23, 2018)	criteria provided, single submitter	VCV000200906	11	2776000	200906	197492	rs794728574	Neutral	0.318	0.558	0.125
KCNQ1	417	V	M	NM_000218.3(KCNQ1):c.1249G>A (p.Val417Met)	Arrhythmia	Likely benign(Last reviewed: May 24, 2019)	criteria provided, single submitter	VCV000446189	11	2587690	446189	38428	rs267607197	Neutral	0.478	0.405	0.117
KCNQ1	422	K	E	NM_000218.3(KCNQ1):c.1264A>G (p.Lys422Glu)	Long QT syndrome 1	Uncertain significance(Last reviewed: Feb 3, 2020)	criteria provided, single submitter	VCV000812542	11	2588725	812542	800784	rs1589969375	Neutral	0.524	0.318	0.159
KCNQ1	422	K	T	NM_000218.3(KCNQ1):c.1265A>C (p.Lys422Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067022	11	2588726	67022	77918	rs199472778	Neutral	0.384	0.467	0.15
KCNQ1	425	L	I	NM_000218.3(KCNQ1):c.1654C>A (p.Leu552Ile)	Arrhythmia	Uncertain significance(Last	criteria provided, single	VCV001331959	11	2776023	1331959	1322679	NA	Neutral	0.519	0.344	0.137

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Apr 13, 2021)	submitter										
KCNQ1	427	V	M	NM_000218.3(KCNQ1):c.1660G>A (p.Val554Met)	not provided	Pathogenic(Last reviewed: Apr 16, 2013)	criteria provided, single submitter	VCV000200853	11	2776029	200853	197493	rs794728533	Neutral	0.457	0.411	0.133
KCNQ1	428	D	G	NM_000218.3(KCNQ1):c.1283A>G (p.Asp428Gly)	not provided	not provided	no assertion provided	VCV000067023	11	2588744	67023	77919	rs199472779	Neutral	0.517	0.304	0.179
KCNQ1	428	R	L	NM_000218.3(KCNQ1):c.1664G>T (p.Arg555Leu)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2019)	criteria provided, single submitter	VCV000940744	11	2776033	940744	935472	rs199472800	Neutral	0.33	0.539	0.132
KCNQ1	434	R	K	NM_000218.3(KCNQ1):c.1682G>A (p.Arg561Lys)	not provided	Likely pathogenic(Last reviewed: Nov 19, 2013)	criteria provided, single submitter	VCV000200854	11	2776051	200854	197495	rs794728534	Neutral	0.522	0.293	0.185
KCNQ1	435	E	K	NM_000218.3(KCNQ1):c.1303G>A (p.Glu435Lys)	Arrhythmia	Uncertain significance(Last reviewed: Sep 27, 2019)	criteria provided, single submitter	VCV000923088	11	2588764	923088	911608	rs1848630440	Neutral	0.529	0.241	0.23
KCNQ1	435	R	S	NM_000218.3(KCNQ1):c.1686G>T (p.Arg562Ser)	not provided	Pathogenic(Last reviewed: Dec 3, 2012)	criteria provided, single submitter	VCV000200855	11	2776986	200855	197497	rs794728535	Neutral	0.451	0.385	0.165
KCNQ1	437	D	G	NM_000218.3(KCNQ1):c.1691A>G (p.Asp564Gly)	Long QT syndrome 1	Likely pathogenic(Last reviewed: Feb 18, 2017)	no assertion criteria provided	VCV000403694	11	2776991	403694	390668	rs1554920808	Neutral	0.534	0.282	0.184
KCNQ1	440	I	F	NM_000218.3(KCNQ1):c.1699A>T (p.Ile567Phe)	not provided	Likely pathogenic(Last reviewed: Oct 1, 2020)	criteria provided, single submitter	VCV001012792	11	2776999	1012792	1000727	rs1846717292	Neutral	0.523	0.349	0.128
KCNQ1	441	P	S	NM_000218.3(KCNQ1):c.1321C>T (p.Pro441Ser)	not specified|Long QT syndrome|not provided	Uncertain significance(Last reviewed: Jul 2, 2018)	criteria provided, multiple submitters, no conflicts	VCV000067024	11	2588782	67024	77920	rs199473475	Neutral	0.547	0.278	0.175
KCNQ1	442	H	D	NM_000218.3(KCNQ1):c.1324C>G (p.His442Asp)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 31, 2019)	criteria provided, single submitter	VCV000963906	11	2588785	963906	947395	rs72549409	Neutral	0.552	0.272	0.176
KCNQ1	443	P	L	NM_000218.3(KCNQ1):c.1709C>T (p.Pro570Leu)	Long QT syndrome 1	Uncertain significance(Last reviewed: Nov 30, 2018)	criteria provided, single submitter	VCV000637976	11	2777009	637976	625809	rs1590082242	Neutral	0.456	0.396	0.148
KCNQ1	446	D	E	NM_000218.3(KCNQ1):c.1338C>G (p.Asp446Glu)	Long QT syndrome|Congenital long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Aug 26, 2020)	criteria provided, conflicting interpretations	VCV000067025	11	2588799	67025	77921	rs199472780	Neutral	0.594	0.231	0.176
KCNQ1	448	P	L	NM_000218.3(KCNQ1):c.1343C>T (p.Pro448Leu)	Long QT syndrome|not specified|Short QT syndrome 2|Jervell and Lange-Nielsen syndrome 1|Atrial fibrillation, familial, 3|not provided|Long QT syndrome 1|Congenital long QT syndrome|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Mar 4, 2020)	criteria provided, conflicting interpretations	VCV000067027	11	2588804	67027	77923	rs12720449	Neutral	0.389	0.406	0.205
KCNQ1	448	P	R	NM_000218.3(KCNQ1):c.1343C>G (p.Pro448Arg)	Cardiovascular phenotype|Short QT syndrome 2|Atrial fibrillation, familial, 3|Jervell and Lange-Nielsen syndrome 1|not specified|not provided|Long QT syndrome|Arrhythmia|Long QT syndrome 1	Benign/Likely benign(Last reviewed: Dec 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067026	11	2588804	67026	77922	rs12720449	GOF	0.489	0.17	0.341
KCNQ1	449	E	K	NM_000218.3(KCNQ1):c.1345G>A (p.Glu449Lys)	Arrhythmia|Long QT syndrome	Uncertain significance(Last reviewed: May 20,	criteria provided, multiple	VCV000924512	11	2588806	924512	911613	rs372583676	Neutral	0.527	0.266	0.207

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)	submitters, no conflicts										
KCNQ1	449	V	I	NM_000218.3(KCNQ1):c.1726G>A (p.Val576Ile)	Arrhythmia|not provided|Long QT syndrome|Long QT syndrome 1|not specified	Conflicting interpretations of pathogenicity(Last reviewed: Jan 23, 2020)	criteria provided, conflicting interpretations	VCV000200813	11	2777026	200813	197500	rs750409379	Neutral	0.431	0.447	0.122
KCNQ1	451	E	K	NM_000218.3(KCNQ1):c.1732G>A (p.Glu578Lys)	not specified	Uncertain significance(Last reviewed: Mar 18, 2016)	criteria provided, single submitter	VCV000372394	11	2777032	372394	359875	rs1057517750	Neutral	0.508	0.297	0.195
KCNQ1	451	R	Q	NM_000218.3(KCNQ1):c.1352G>A (p.Arg451Gln)	Long QT syndrome|Atrial fibrillation, familial, 3|Short QT syndrome 2|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome 1|not provided|Arrhythmia	Conflicting interpretations of pathogenicity(Last reviewed: Apr 6, 2021)	criteria provided, conflicting interpretations	VCV000067029	11	2588813	67029	77925	rs199472781	Neutral	0.438	0.423	0.139
KCNQ1	451	R	W	NM_000218.3(KCNQ1):c.1351C>T (p.Arg451Trp)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Oct 6, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067028	11	2588812	67028	77924	rs199472782	Neutral	0.272	0.618	0.11
KCNQ1	452	R	Q	NM_000218.3(KCNQ1):c.1355G>A (p.Arg452Gln)	Long QT syndrome|Arrhythmia|short QT syndrome|Cardiovascular phenotype|not provided|not specified|Jervell and Lange-Nielsen syndrome|Familial atrial fibrillation|Congenital long QT syndrome|Atrial fibrillation, familial, 3|Beckwith-Wiedemann syndrome|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome 1|Short QT syndrome 2	Uncertain significance(Last reviewed: Apr 6, 2021)	criteria provided, multiple submitters, no conflicts	VCV000067030	11	2588816	67030	77926	rs145229963	Neutral	0.472	0.391	0.137
KCNQ1	452	R	W	NM_000218.3(KCNQ1):c.1354C>T (p.Arg452Trp)	Long QT syndrome|Long QT syndrome 1|not provided|Congenital long QT syndrome|Arrhythmia|Cardiovascular phenotype|Atrial fibrillation, familial, 3|Long QT syndrome 1|Short QT syndrome 2|Beckwith-Wiedemann syndrome|Jervell and Lange-Nielsen syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Jun 2, 2021)	criteria provided, conflicting interpretations	VCV000052980	11	2588815	52980	67648	rs140452381	Neutral	0.253	0.638	0.109
KCNQ1	455	H	Y	NM_000218.3(KCNQ1):c.1363C>T (p.His455Tyr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000052981	11	2588824	52981	67649	rs199473476	Neutral	0.352	0.527	0.121
KCNQ1	460	G	S	NM_000218.3(KCNQ1):c.1378G>A (p.Gly460Ser)	Long QT syndrome|not provided|SUDDEN INFANT DEATH SYNDROME|Arrhythmia	Uncertain significance(Last reviewed: Dec 11, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067031	11	2588839	67031	77927	rs199472783	GOF	0.571	0.138	0.291
KCNQ1	461	I	F	NM_000218.3(KCNQ1):c.1762A>T (p.Ile588Phe)	not provided|Long QT syndrome 1	Pathogenic/Likely pathogenic(Last reviewed: May 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000200856	11	2778005	200856	197501	rs794728536	Neutral	0.462	0.404	0.134
KCNQ1	461	I	T	NM_000218.3(KCNQ1):c.1763T>C (p.Ile588Thr)	not provided	Likely pathogenic(Last reviewed: Aug 25, 2011)	criteria provided, single submitter	VCV000200908	11	2778006	200908	197502	rs794728576	Neutral	0.518	0.309	0.173
KCNQ1	461	I	V	NM_000218.3(KCNQ1):c.1762A>G (p.Ile588Val)	Arrhythmia	Uncertain significance(Last reviewed: Sep 13, 2019)	criteria provided, single submitter	VCV000918864	11	2778005	918864	911641	rs794728536	Neutral	0.523	0.338	0.139
KCNQ1	462	D	N	NM_000218.3(KCNQ1):c.1384G>A (p.Asp462Asn)	Long QT syndrome	Uncertain significance(Last reviewed: Aug 14, 2020)	criteria provided, single submitter	VCV000940662	11	2588845	940662	935471	rs776205380	Neutral	0.551	0.266	0.183
KCNQ1	462	G	S	NM_000218.3(KCNQ1):c.1765G>A (p.Gly589Ser)	Arrhythmia|Long QT syndrome	Uncertain significance(Last	criteria provided,	VCV000923600	11	2778008	923600	911643	rs780676796	Neutral	0.552	0.249	0.199

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Oct 8, 2020)	multiple submitters, no conflicts										
KCNQ1	463	A	P	NM_000218.3(KCNQ1):c.1768G>C (p.Ala590Pro)	Cardiovascular phenotype	Uncertain significance(Last reviewed: Jul 6, 2016)	criteria provided, single submitter	VCV000519284	11	2778011	519284	510282	rs199472813	Neutral	0.522	0.308	0.17
KCNQ1	463	S	G	NM_000218.3(KCNQ1):c.1387A>G (p.Ser463Gly)	Arrhythmia	Uncertain significance(Last reviewed: Jul 25, 2019)	criteria provided, single submitter	VCV000923366	11	2588848	923366	911616	rs1848632977	Neutral	0.504	0.34	0.155
KCNQ1	464	R	L	NM_000218.3(KCNQ1):c.1772G>T (p.Arg591Leu)	not provided	Pathogenic(Last reviewed: Nov 4, 2013)	criteria provided, single submitter	VCV000200857	11	2778015	200857	197503	rs199472814	Neutral	0.315	0.565	0.119
KCNQ1	464	R	P	NM_000218.3(KCNQ1):c.1772G>C (p.Arg591Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 17, 2019)	criteria provided, single submitter	VCV000854899	11	2778015	854899	838249	rs199472814	Neutral	0.464	0.39	0.146
KCNQ1	466	N	K	NM_000218.3(KCNQ1):c.1779C>G (p.Asn593Lys)	Long QT syndrome	Uncertain significance(Last reviewed: Oct 27, 2020)	criteria provided, single submitter	VCV001044816	11	2778022	1044816	1030183	rs1421636319	Neutral	0.513	0.273	0.214
KCNQ1	469	E	G	NM_000218.3(KCNQ1):c.1787A>G (p.Glu596Gly)	not provided	Pathogenic(Last reviewed: Apr 12, 2014)	criteria provided, single submitter	VCV000200859	11	2778030	200859	197505	rs794728538	Neutral	0.509	0.314	0.177
KCNQ1	511	R	W	NM_000218.3(KCNQ1):c.1531C>T (p.Arg511Trp)	Long QT syndrome|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Oct 22, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067034	11	2768860	67034	77930	rs199472785	Neutral	0.707	0.207	0.086
KCNQ1	514	I	T	NM_000218.3(KCNQ1):c.1541T>C (p.Ile514Thr)	Congenital long QT syndrome|Long QT syndrome 1	Uncertain significance(Last reviewed: Nov 16, 2018)	criteria provided, single submitter	VCV000052990	11	2768870	52990	67658	rs199472786	LOF	0.91	0.03	0.06
KCNQ1	517	I	T	NM_000218.3(KCNQ1):c.1550T>C (p.Ile517Thr)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067035	11	2768879	67035	77931	rs199473478	LOF	0.908	0.03	0.061
KCNQ1	518	R	G	NM_000218.3(KCNQ1):c.1552C>G (p.Arg518Gly)	Long QT syndrome 1|Long QT syndrome|Congenital long QT syndrome	Likely pathogenic(Last reviewed: May 15, 2019)	criteria provided, multiple submitters, no conflicts	VCV000052991	11	2768881	52991	67659	rs17215500	LOF	0.901	0.045	0.054
KCNQ1	518	R	P	NM_000218.3(KCNQ1):c.1553G>C (p.Arg518Pro)	Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Apr 6, 2018)	criteria provided, single submitter	VCV000052992	11	2768882	52992	67660	rs145974930	LOF	0.873	0.065	0.062
KCNQ1	518	R	Q	NM_000218.3(KCNQ1):c.1553G>A (p.Arg518Gln)	Long QT syndrome|not specified|Congenital long QT syndrome|not provided|Arrhythmia	Uncertain significance(Last reviewed: Jan 24, 2022)	criteria provided, multiple submitters, no conflicts	VCV000067036	11	2768882	67036	77932	rs145974930	LOF	0.881	0.061	0.059
KCNQ1	519	R	C	NM_000218.3(KCNQ1):c.1555C>T (p.Arg519Cys)	Congenital long QT syndrome|Arrhythmia|Cardiovascular phenotype|not specified|Atrial fibrillation, familial, 3|Beckwith-Wiedemann syndrome|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome 1|Short QT syndrome 2	Uncertain significance(Last reviewed: Mar 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067037	11	2768884	67037	77933	rs199472787	LOF	0.883	0.058	0.06
KCNQ1	519	R	H	NM_000218.3(KCNQ1):c.1556G>A (p.Arg519His)	Short QT syndrome 2|Atrial fibrillation, familial, 3|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome|not provided|Arrhythmia|Long QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Jun 4, 2020)	criteria provided, conflicting interpretations	VCV000067038	11	2768885	67038	77934	rs199472788	LOF	0.863	0.075	0.062
KCNQ1	520	M	R	NM_000218.3(KCNQ1):c.1559T>G (p.Met520Arg)	Cardiovascular phenotype|not provided|Congenital long QT syndrome|Long QT	Pathogenic/Likely pathogenic(Last	criteria provided,	VCV000052993	11	2768888	52993	67661	rs199473479	LOF	0.902	0.02	0.077

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					syndrome	reviewed: Dec 9, 2019)	multiple submitters, no conflicts										
KCNQ1	522	Y	S	NM_000218.3(KCNQ1):c.1565A>C (p.Tyr522Ser)	Congenital long QT syndrome|not provided	Pathogenic(Last reviewed: Aug 14, 2012)	criteria provided, single submitter	VCV000067039	11	2768894	67039	77935	rs199472789	LOF	0.9	0.047	0.054
KCNQ1	524	V	G	NM_000218.3(KCNQ1):c.1571T>G (p.Val524Gly)	Congenital long QT syndrome|not provided|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Nov 25, 2020)	criteria provided, multiple submitters, no conflicts	VCV000052994	11	2768900	52994	67662	rs199472790	LOF	0.884	0.056	0.06
KCNQ1	525	A	T	NM_000218.3(KCNQ1):c.1573G>A (p.Ala525Thr)	not provided|Long QT syndrome 1, recessive| Cardiovascular phenotype|Congenital long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jun 1, 2016)	criteria provided, multiple submitters, no conflicts	VCV000003132	11	2768902	3132	18171	rs120074188	LOF	0.891	0.051	0.058
KCNQ1	525	A	V	NM_000218.3(KCNQ1):c.1574C>T (p.Ala525Val)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067040	11	2768903	67040	77936	rs199472791	LOF	0.863	0.073	0.064
KCNQ1	526	K	E	NM_000218.3(KCNQ1):c.1576A>G (p.Lys526Glu)	Long QT syndrome|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Oct 7, 2018)	criteria provided, multiple submitters, no conflicts	VCV000052995	11	2768905	52995	67663	rs199472792	LOF	0.807	0.122	0.071
KCNQ1	526	K	Q	NM_000218.3(KCNQ1):c.1576A>C (p.Lys526Gln)	Long QT syndrome|Long QT syndrome 1	Uncertain significance(Last reviewed: Jan 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000853542	11	2768905	853542	838247	rs199472792	Neutral	0.725	0.198	0.077
KCNQ1	533	R	W	NM_000218.3(KCNQ1):c.1597C>T (p.Arg533Trp)	Arrhythmia|Long QT syndrome|Congenital long QT syndrome|Long QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Jan 24, 2020)	criteria provided, conflicting interpretations	VCV000067041	11	2775966	67041	77937	rs199472793	Neutral	0.493	0.408	0.099
KCNQ1	539	R	Q	NM_000218.3(KCNQ1):c.1616G>A (p.Arg539Gln)	Long QT syndrome|Cardiovascular phenotype| not provided|Arrhythmia|Long QT syndrome 1| Congenital long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: May 26, 2021)	criteria provided, conflicting interpretations	VCV000067042	11	2775985	67042	77938	rs199472794	LOF	0.811	0.129	0.06
KCNQ1	539	R	W	NM_000218.3(KCNQ1):c.1615C>T (p.Arg539Trp)	Long QT syndrome 1|Atrial fibrillation, familial, 3|Beckwith-Wiedemann syndrome|Long QT syndrome 1|Jervell and Lange-Nielsen syndrome 1|Short QT syndrome 2|Long QT syndrome|not provided|Congenital long QT syndrome	Pathogenic(Last reviewed: Mar 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000052998	11	2775984	52998	67666	rs199472795	Neutral	0.671	0.245	0.085
KCNQ1	541	V	I	NM_000218.3(KCNQ1):c.1621G>A (p.Val541Ile)	Long QT syndrome|not specified|Congenital long QT syndrome|Wolff-Parkinson-White pattern|not provided|Arrhythmia|Long QT syndrome 1	Conflicting interpretations of pathogenicity(Last reviewed: Nov 25, 2020)	criteria provided, conflicting interpretations	VCV000067043	11	2775990	67043	77939	rs199472796	LOF	0.851	0.082	0.067
KCNQ1	543	E	K	NM_000218.3(KCNQ1):c.1627G>A (p.Glu543Lys)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067044	11	2775996	67044	77940	rs199472797	LOF	0.845	0.096	0.059
KCNQ1	546	S	L	NM_000218.3(KCNQ1):c.1637C>T (p.Ser546Leu)	Long QT syndrome 1|Long QT syndrome|not provided|Congenital long QT syndrome| Arrhythmia|Cardiovascular phenotype	Pathogenic/Likely pathogenic(Last reviewed: Jan 3, 2022)	criteria provided, multiple submitters, no conflicts	VCV000053000	11	2776006	53000	67668	rs199473480	Neutral	0.712	0.187	0.101
KCNQ1	547	Q	R	NM_000218.3(KCNQ1):c.1640A>G (p.Gln547Arg)	Congenital long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Apr 13, 2020)	criteria provided, single submitter	VCV000067045	11	2776009	67045	77941	rs199472798	LOF	0.856	0.083	0.061
KCNQ1	548	G	D	NM_000218.3(KCNQ1):c.1643G>A (p.Gly548Asp)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last	criteria provided, single	VCV000053001	11	2776012	53001	67669	rs199472799	LOF	0.913	0.036	0.051

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Oct 20, 2020)	submitter										
KCNQ1	554	V	A	NM_000218.3(KCNQ1):c.1661T>C (p.Val554Ala)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053002	11	2776030	53002	67670	rs199473481	Neutral	0.733	0.194	0.073
KCNQ1	554	V	L	NM_000218.3(KCNQ1):c.1660G>T (p.Val554Leu)	Arrhythmia	Uncertain significance(Last reviewed: Dec 10, 2020)	criteria provided, single submitter	VCV000923332	11	2776029	923332	911638	rs794728533	Neutral	0.709	0.214	0.077
KCNQ1	555	R	C	NM_000218.3(KCNQ1):c.1663C>T (p.Arg555Cys)	Long QT syndrome|Cardiovascular phenotype|Prolonged QT interval|not provided|Long QT syndrome 1|Congenital long QT syndrome|Arrhythmia	Pathogenic(Last reviewed: Mar 26, 2021)	criteria provided, multiple submitters, no conflicts	VCV000003126	11	2776032	3126	18165	rs120074185	LOF	0.862	0.078	0.061
KCNQ1	555	R	H	NM_000218.3(KCNQ1):c.1664G>A (p.Arg555His)	Congenital long QT syndrome|Cardiovascular phenotype|Long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last reviewed: Jul 10, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053003	11	2776033	53003	67671	rs199472800	LOF	0.795	0.137	0.068
KCNQ1	555	R	S	NM_000218.3(KCNQ1):c.1663C>A (p.Arg555Ser)	Congenital long QT syndrome|Long QT syndrome|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Mar 11, 2020)	criteria provided, conflicting interpretations	VCV000067046	11	2776032	67046	77942	rs120074185	LOF	0.788	0.143	0.069
KCNQ1	557	K	E	NM_000218.3(KCNQ1):c.1669A>G (p.Lys557Glu)	Congenital long QT syndrome|Cardiovascular phenotype|Long QT syndrome|not provided	Pathogenic/Likely pathogenic(Last reviewed: Oct 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067047	11	2776038	67047	77943	rs199472801	LOF	0.867	0.079	0.055
KCNQ1	562	R	M	NM_000218.3(KCNQ1):c.1685G>T (p.Arg562Met)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067048	11	2776054	67048	77944	rs199472802	Neutral	0.758	0.176	0.067
KCNQ1	565	Q	P	NM_000218.3(KCNQ1):c.1694A>C (p.Gln565Pro)	Long QT syndrome	Uncertain significance(Last reviewed: Sep 24, 2019)	criteria provided, single submitter	VCV000940743	11	2776994	940743	935473	rs1846716972	Neutral	0.692	0.234	0.074
KCNQ1	566	S	F	NM_000218.3(KCNQ1):c.1697C>T (p.Ser566Phe)	Long QT syndrome 1|Congenital long QT syndrome|Long QT syndrome|not specified|not provided	Likely pathogenic(Last reviewed: Jan 18, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053006	11	2776997	53006	67674	rs199472804	Neutral	0.539	0.373	0.088
KCNQ1	566	S	P	NM_000218.3(KCNQ1):c.1696T>C (p.Ser566Pro)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067049	11	2776996	67049	77945	rs199472803	Neutral	0.694	0.234	0.072
KCNQ1	566	S	Y	NM_000218.3(KCNQ1):c.1697C>A (p.Ser566Tyr)	not provided|Congenital long QT syndrome|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jul 10, 2020)	criteria provided, conflicting interpretations	VCV000053005	11	2776997	53005	67673	rs199472804	Neutral	0.571	0.35	0.079
KCNQ1	567	I	S	NM_000218.3(KCNQ1):c.1700T>G (p.Ile567Ser)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053008	11	2777000	53008	67676	rs199472805	Neutral	0.722	0.182	0.095
KCNQ1	567	I	T	NM_000218.3(KCNQ1):c.1700T>C (p.Ile567Thr)	Long QT syndrome 1|Long QT syndrome|Congenital long QT syndrome|Arrhythmia|not provided	Pathogenic/Likely pathogenic(Last reviewed: Sep 28, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053007	11	2777000	53007	67675	rs199472805	Neutral	0.652	0.257	0.091
KCNQ1	568	G	A	NM_000218.3(KCNQ1):c.1703G>C (p.Gly568Ala)	Long QT syndrome 1|Congenital long QT syndrome	Pathogenic(Last reviewed: Jan 1, 2012)	no assertion criteria provided	VCV000053010	11	2777003	53010	67678	rs199472806	Neutral	0.713	0.188	0.099
KCNQ1	568	G	R	NM_000218.3(KCNQ1):c.1702G>A (p.Gly568Arg)	Congenital long QT syndrome|Cardiovascular phenotype|not provided|not specified|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jun 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053009	11	2777002	53009	67677	rs199472807	LOF	0.76	0.087	0.153

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ1	568	G	R	NM_000218.3(KCNQ1):c.1702G>A (p.Gly568Arg)	Congenital long QT syndrome|Cardiovascular phenotype|not provided|not specified|Long QT syndrome	Pathogenic/Likely pathogenic(Last reviewed: Jun 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053009	11	2777002	53009	67677	rs199472807	LOF	0.76	0.087	0.153
KCNQ1	568	G	R	NM_000218.3(KCNQ1):c.1702G>C (p.Gly568Arg)	Long QT syndrome	Likely pathogenic(Last reviewed: Mar 8, 2018)	criteria provided, single submitter	VCV000570319	11	2777002	570319	565649	rs199472807	LOF	0.76	0.087	0.153
KCNQ1	568	G	R	NM_000218.3(KCNQ1):c.1702G>C (p.Gly568Arg)	Long QT syndrome	Likely pathogenic(Last reviewed: Mar 8, 2018)	criteria provided, single submitter	VCV000570319	11	2777002	570319	565649	rs199472807	LOF	0.76	0.087	0.153
KCNQ1	569	K	E	NM_000218.3(KCNQ1):c.1705A>G (p.Lys569Glu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000067050	11	2777005	67050	77946	rs199472808	Neutral	0.695	0.213	0.092
KCNQ1	571	S	L	NM_000218.3(KCNQ1):c.1712C>T (p.Ser571Leu)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Sep 3, 2020)	criteria provided, single submitter	VCV000067052	11	2777012	67052	77948	rs199472809	Neutral	0.365	0.534	0.101
KCNQ1	573	F	L	NM_000218.3(KCNQ1):c.1719C>A (p.Phe573Leu)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053012	11	2777019	53012	67680	rs199472810	Neutral	0.554	0.337	0.109
KCNQ1	574	I	V	NM_000218.3(KCNQ1):c.1720A>G (p.Ile574Val)	Long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Oct 16, 2019)	criteria provided, multiple submitters, no conflicts	VCV000630538	11	2777020	630538	617861	rs761947465	Neutral	0.573	0.322	0.105
KCNQ1	583	R	C	NM_000218.3(KCNQ1):c.1747C>T (p.Arg583Cys)	not provided|Atrial fibrillation, familial, 3| Beckwith-Wiedemann syndrome|Short QT syndrome 2|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome 1|Acquired susceptibility to long QT syndrome 1|KCNQ1-Related Disorders|Congenital long QT syndrome|Long QT syndrome 1	Likely pathogenic(Last reviewed: Nov 11, 2021)	criteria provided, multiple submitters, no conflicts	VCV000003142	11	2777990	3142	18181	rs17221854	Neutral	0.454	0.433	0.113
KCNQ1	583	R	H	NM_000218.3(KCNQ1):c.1748G>A (p.Arg583His)	Long QT syndrome|Arrhythmia|Congenital long QT syndrome|not provided	Uncertain significance(Last reviewed: Oct 21, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067053	11	2777991	67053	77949	rs199473482	Neutral	0.474	0.403	0.123
KCNQ1	584	G	S	NM_000218.3(KCNQ1):c.1750G>A (p.Gly584Ser)	Long QT syndrome|Arrhythmia|SUDDEN INFANT DEATH SYNDROME	Uncertain significance(Last reviewed: Sep 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000067054	11	2777993	67054	77950	rs199472811	Neutral	0.629	0.186	0.185
KCNQ1	586	N	D	NM_000218.3(KCNQ1):c.1756A>G (p.Asn586Asp)	Congenital long QT syndrome	not provided	no assertion provided	VCV000053014	11	2777999	53014	67682	rs199472812	Neutral	0.664	0.216	0.119
KCNQ1	587	T	M	NM_000218.3(KCNQ1):c.1760C>T (p.Thr587Met)	Jervell and Lange-Nielsen syndrome 1| Cardiovascular phenotype|not provided| Congenital long QT syndrome|Long QT syndrome	Pathogenic(Last reviewed: Oct 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003138	11	2778003	3138	18177	rs120074189	Neutral	0.469	0.454	0.077
KCNQ1	587	T	R	NM_000218.3(KCNQ1):c.1760C>G (p.Thr587Arg)	Long QT syndrome	Uncertain significance(Last reviewed: Feb 1, 2020)	criteria provided, single submitter	VCV001037124	11	2778003	1037124	1030182	rs120074189	Neutral	0.675	0.198	0.127
KCNQ1	589	G	D	NM_000218.3(KCNQ1):c.1766G>A (p.Gly589Asp)	Long QT syndrome|Jervell and Lange-Nielsen syndrome 1|Long QT syndrome 1| Cardiovascular phenotype|Long QT syndrome 1| Jervell and Lange-Nielsen syndrome 1| Congenital long QT syndrome|Arrhythmia|not provided	Pathogenic(Last reviewed: Feb 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000003140	11	2778009	3140	18179	rs120074190	LOF	0.812	0.08	0.107
KCNQ1	590	A	T	NM_000218.3(KCNQ1):c.1768G>A (p.Ala590Thr)	not provided|Long QT syndrome 1|Congenital long QT syndrome|Arrhythmia|not specified	Conflicting interpretations of	criteria provided,	VCV000053015	11	2778011	53015	67683	rs199472813	Neutral	0.628	0.278	0.093

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						pathogenicity(Last reviewed: May 4, 2021)	conflicting interpretations										
KCNQ1	591	R	C	NM_000218.3(KCNQ1):c.1771C>T (p.Arg591Cys)	Congenital long QT syndrome|Arrhythmia	Uncertain significance(Last reviewed: Feb 18, 2020)	criteria provided, single submitter	VCV000053016	11	2778014	53016	67684	rs199473483	Neutral	0.673	0.249	0.079
KCNQ1	591	R	H	NM_000218.3(KCNQ1):c.1772G>A (p.Arg591His)	Long QT syndrome|not provided|Long QT syndrome 1|Congenital long QT syndrome	Pathogenic(Last reviewed: Jan 3, 2022)	criteria provided, multiple submitters, no conflicts	VCV000053017	11	2778015	53017	67685	rs199472814	Neutral	0.641	0.28	0.078
KCNQ1	594	R	P	NM_000218.3(KCNQ1):c.1781G>C (p.Arg594Pro)	not provided|Cardiovascular phenotype|Congenital long QT syndrome|Long QT syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Jan 13, 2022)	criteria provided, conflicting interpretations	VCV000053019	11	2778024	53019	67687	rs199472815	Neutral	0.623	0.289	0.088
KCNQ1	594	R	Q	NM_000218.3(KCNQ1):c.1781G>A (p.Arg594Gln)	Long QT syndrome|Cardiovascular phenotype|Long QT syndrome 1|Congenital long QT syndrome|Long QT syndrome 1|Jervell and Lange-Nielsen syndrome 1|not provided	Pathogenic/Likely pathogenic(Last reviewed: Nov 18, 2021)	criteria provided, multiple submitters, no conflicts	VCV000053018	11	2778024	53018	67686	rs199472815	Neutral	0.663	0.254	0.083
KCNQ1	596	E	K	NM_000218.3(KCNQ1):c.1786G>A (p.Glu596Lys)	Congenital long QT syndrome|Long QT syndrome	Uncertain significance(Last reviewed: Apr 20, 2020)	criteria provided, single submitter	VCV000067055	11	2778029	67055	77951	rs199472816	LOF	0.749	0.154	0.097
KCNQ1	598	K	R	NM_000218.3(KCNQ1):c.1793A>G (p.Lys598Arg)	SUDDEN INFANT DEATH SYNDROME|Long QT syndrome 1	Uncertain significance(Last reviewed: Nov 3, 2021)	criteria provided, single submitter	VCV000067056	11	2778036	67056	77952	rs199472817	Neutral	0.697	0.198	0.105
KCNQ2	1	M	K	NM_172107.4(KCNQ2):c.2T>A (p.Met1Lys)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic/Likely pathogenic(Last reviewed: Dec 2, 2018)	criteria provided, multiple submitters, no conflicts	VCV000497690	20	63472462	497690	489114	rs118192186	LOF	0.8	0.095	0.105
KCNQ2	1	M	T	NM_172107.4(KCNQ2):c.2T>C (p.Met1Thr)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Jan 15, 2020)	criteria provided, single submitter	VCV000021783	20	63472462	21783	34635	rs118192186	LOF	0.8	0.092	0.108
KCNQ2	1	M	V	NM_172107.4(KCNQ2):c.1A>G (p.Met1Val)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Benign familial neonatal seizures 1	Pathogenic/Likely pathogenic(Last reviewed: Jun 6, 2020)	criteria provided, multiple submitters, no conflicts	VCV000021774	20	63472463	21774	34626	rs118192185	LOF	0.795	0.094	0.111
KCNQ2	4	K	N	NM_172107.4(KCNQ2):c.12G>C (p.Lys4Asn)	not provided|not specified	Uncertain significance(Last reviewed: Feb 26, 2020)	criteria provided, single submitter	VCV000373753	20	63472452	373753	360440	rs776223064	LOF	0.832	0.065	0.103
KCNQ2	6	R	P	NM_172107.4(KCNQ2):c.17G>C (p.Arg6Pro)	not provided	Uncertain significance(Last reviewed: Jun 11, 2019)	criteria provided, single submitter	VCV000429227	20	63472447	429227	422347	rs866843916	LOF	0.838	0.064	0.098
KCNQ2	8	G	C	NM_172107.4(KCNQ2):c.22G>T (p.Gly8Cys)	not provided	Uncertain significance(Last reviewed: Feb 1, 2018)	criteria provided, single submitter	VCV000450703	20	63472442	450703	446297	rs769045070	LOF	0.813	0.078	0.11
KCNQ2	8	G	S	NM_172107.4(KCNQ2):c.22G>A (p.Gly8Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 4, 2019)	criteria provided, single submitter	VCV000939478	20	63472442	939478	939037	rs769045070	LOF	0.817	0.079	0.104
KCNQ2	9	G	D	NM_172107.4(KCNQ2):c.26G>A (p.Gly9Asp)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Jan 1,	criteria provided, single submitter	VCV000562034	20	63472438	562034	553187	NA	LOF	0.815	0.082	0.103

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2017)											
KCNQ2	11	Y	H	NM_172107.4(KCNQ2):c.31T>C (p.Tyr11His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 7, 2017)	criteria provided, single submitter	VCV000461420	20	63472433	461420	471005	rs1270851643	LOF	0.816	0.07	0.114
KCNQ2	17	E	D	NM_172107.4(KCNQ2):c.51G>T (p.Glu17Asp)	Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: Jun 28, 2019)	criteria provided, single submitter	VCV001029261	20	63472413	1029261	1018721	rs2082239531	LOF	0.825	0.073	0.102
KCNQ2	21	K	E	NM_172107.4(KCNQ2):c.61A>G (p.Lys21Glu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 26, 2020)	criteria provided, single submitter	VCV001051012	20	63472403	1051012	1051788	NA	LOF	0.848	0.053	0.099
KCNQ2	22	V	M	NM_172107.4(KCNQ2):c.64G>A (p.Val22Met)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 7, 2020)	criteria provided, multiple submitters, no conflicts	VCV001023279	20	63472400	1023279	1014211	rs2082239153	LOF	0.825	0.087	0.088
KCNQ2	35	T	S	NM_172107.4(KCNQ2):c.104C>G (p.Thr35Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 30, 2020)	criteria provided, single submitter	VCV001063140	20	63472360	1063140	1051787	NA	LOF	0.827	0.063	0.11
KCNQ2	40	L	P	NM_172107.4(KCNQ2):c.119T>C (p.Leu40Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 4, 2019)	criteria provided, single submitter	VCV000852195	20	63472345	852195	848549	rs2082237607	LOF	0.847	0.062	0.091
KCNQ2	43	A	V	NM_172107.4(KCNQ2):c.128C>T (p.Ala43Val)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Seizures	Conflicting interpretations of pathogenicity(Last reviewed: Sep 1, 2020)	criteria provided, conflicting interpretations	VCV000205939	20	63472336	205939	203807	rs749554385	LOF	0.818	0.06	0.122
KCNQ2	50	R	H	NM_172107.4(KCNQ2):c.149G>A (p.Arg50His)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Sep 24, 2019)	criteria provided, multiple submitters, no conflicts	VCV000425273	20	63472315	425273	413607	rs1064797286	LOF	0.852	0.049	0.099
KCNQ2	53	I	T	NM_172107.4(KCNQ2):c.158T>C (p.Ile53Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 30, 2018)	criteria provided, single submitter	VCV000639171	20	63472306	639171	650470	rs1038663676	LOF	0.841	0.056	0.104
KCNQ2	57	P	S	NM_172107.4(KCNQ2):c.169C>T (p.Pro57Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 26, 2020)	criteria provided, single submitter	VCV001057327	20	63472295	1057327	1051785	NA	LOF	0.826	0.071	0.103
KCNQ2	58	R	S	NM_172107.4(KCNQ2):c.171_172delinsAA (p.Arg58Ser)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001326316	20	63472292 - 63472293	1326316	1316718	NA	LOF	0.839	0.057	0.104
KCNQ2	62	A	V	NM_172107.4(KCNQ2):c.185C>T (p.Ala62Val)	Early infantile epileptic encephalopathy with suppression bursts|not specified	Likely benign(Last reviewed: Jul 1, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205857	20	63472279	205857	203806	rs796052612	LOF	0.798	0.073	0.129
KCNQ2	64	A	S	NM_172107.4(KCNQ2):c.190G>T (p.Ala64Ser)	Early infantile epileptic encephalopathy with suppression bursts|not specified	Conflicting interpretations of pathogenicity(Last reviewed: Jul 9, 2020)	criteria provided, conflicting interpretations	VCV000205858	20	63472274	205858	203805	rs780110473	LOF	0.796	0.086	0.118
KCNQ2	67	P	A	NM_172107.4(KCNQ2):c.199C>G (p.Pro67Ala)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 23, 2019)	criteria provided, single submitter	VCV000842102	20	63472265	842102	848548	rs972841085	LOF	0.843	0.059	0.098
KCNQ2	67	P	S	NM_172107.4(KCNQ2):c.199C>T (p.Pro67Ser)	Early infantile epileptic encephalopathy 7	Uncertain	criteria	VCV000931897	20	63472265	931897	919926	rs972841085	LOF	0.839	0.06	0.101

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						significance(Last reviewed: Apr 2, 2020)	provided, single submitter										
KCNQ2	80	F	S	NM_172107.4(KCNQ2):c.239T>C (p.Phe80Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 30, 2019)	criteria provided, single submitter	VCV000963165	20	63472225	963165	951151	rs2082234077	LOF	0.854	0.053	0.094
KCNQ2	82	Y	N	NM_172107.4(KCNQ2):c.244T>A (p.Tyr82Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 13, 2018)	criteria provided, single submitter	VCV000649095	20	63472220	649095	650471	rs1600885336	LOF	0.818	0.05	0.132
KCNQ2	85	L	P	NM_172107.4(KCNQ2):c.254T>C (p.Leu85Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV000962209	20	63472210	962209	951150	rs2082233689	LOF	0.846	0.066	0.088
KCNQ2	88	P	Q	NM_172107.4(KCNQ2):c.263C>A (p.Pro88Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 16, 2020)	criteria provided, single submitter	VCV001058247	20	63472201	1058247	1051784	NA	LOF	0.865	0.04	0.095
KCNQ2	96	H	N	NM_172107.4(KCNQ2):c.286C>A (p.His96Asn)	Epileptic encephalopathy, early infantile, 1	Pathogenic(Last reviewed: Jan 1, 2019)	no assertion criteria provided	VCV000975416	20	63472178	975416	963918	rs2082232988	LOF	0.852	0.052	0.096
KCNQ2	96	H	P	NM_172107.4(KCNQ2):c.287A>C (p.His96Pro)	not provided	Likely pathogenic(Last reviewed: Jun 22, 2015)	criteria provided, single submitter	VCV000372576	20	63472177	372576	360479	rs868055567	LOF	0.859	0.054	0.088
KCNQ2	96	H	Y	NM_172107.4(KCNQ2):c.286C>T (p.His96Tyr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 9, 2019)	criteria provided, single submitter	VCV000856094	20	63472178	856094	848547	rs2082232988	LOF	0.865	0.037	0.099
KCNQ2	97	A	V	NM_172107.4(KCNQ2):c.290C>T (p.Ala97Val)	not provided	Uncertain significance(Last reviewed: Oct 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000430277	20	63472174	430277	422346	rs1131691879	LOF	0.848	0.05	0.102
KCNQ2	101	L	H	NM_172107.4(KCNQ2):c.302T>A (p.Leu101His)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Feb 1, 2020)	criteria provided, single submitter	VCV000944053	20	63446832	944053	929253	rs2081442073	LOF	0.853	0.048	0.099
KCNQ2	103	V	I	NM_172107.4(KCNQ2):c.307G>A (p.Val103Ile)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000450514	20	63446827	450514	446295	rs1555874588	LOF	0.816	0.042	0.143
KCNQ2	104	F	S	NM_172107.4(KCNQ2):c.311T>C (p.Phe104Ser)	not provided	Likely pathogenic(Last reviewed: Mar 22, 2017)	criteria provided, single submitter	VCV000424370	20	63446823	424370	410835	rs1064796940	LOF	0.858	0.05	0.092
KCNQ2	106	C	G	NM_172107.4(KCNQ2):c.316T>G (p.Cys106Gly)	Early infantile epileptic encephalopathy 7	Likely pathogenic	criteria provided, single submitter	VCV001332854	20	63446818	1332854	1323572	NA	LOF	0.852	0.064	0.083
KCNQ2	107	L	F	NM_172107.4(KCNQ2):c.319C>T (p.Leu107Phe)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Oct 8, 2018)	criteria provided, single submitter	VCV000219241	20	63446815	219241	217263	rs864321712	LOF	0.825	0.042	0.133
KCNQ2	107	L	P	NM_172107.4(KCNQ2):c.320T>C (p.Leu107Pro)	not provided	Uncertain significance(Last reviewed: Feb 1, 2017)	criteria provided, single submitter	VCV000444571	20	63446814	444571	438211	rs1555874569	LOF	0.846	0.059	0.094
KCNQ2	108	V	M	NM_172107.4(KCNQ2):c.322G>A (p.Val108Met)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 8,	criteria provided, multiple	VCV000391566	20	63446812	391566	378224	rs749164961	LOF	0.829	0.04	0.132

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)	submitters, no conflicts										
KCNQ2	112	F	L	NM_172107.4(KCNQ2):c.336T>G (p.Phe112Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 10, 2020)	criteria provided, single submitter	VCV000846358	20	63446798	846358	848546	rs2081440418	LOF	0.859	0.036	0.105
KCNQ2	113	S	F	NM_172107.4(KCNQ2):c.338C>T (p.Ser113Phe)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Dec 4, 2019)	criteria provided, conflicting interpretations	VCV000205862	20	63446796	205862	203800	rs796052616	LOF	0.869	0.036	0.094
KCNQ2	113	S	Y	NM_172107.4(KCNQ2):c.338_339delinsAT (p.Ser113Tyr)	not provided	Likely pathogenic(Last reviewed: Apr 27, 2017)	criteria provided, single submitter	VCV000449878	20	63446795 - 63446796	449878	446294	rs1555874555	LOF	0.864	0.038	0.098
KCNQ2	114	T	A	NM_172107.4(KCNQ2):c.340A>G (p.Thr114Ala)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369741	20	63446794	369741	354032	rs1057516076	LOF	0.853	0.044	0.103
KCNQ2	114	T	I	NM_172107.4(KCNQ2):c.341C>T (p.Thr114Ile)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369742	20	63446793	369742	354031	rs1057516077	LOF	0.879	0.031	0.089
KCNQ2	114	T	P	NM_172107.4(KCNQ2):c.340A>C (p.Thr114Pro)	not provided	Likely pathogenic(Last reviewed: Jul 7, 2020)	criteria provided, single submitter	VCV001197529	20	63446794	1197529	1188893	NA	LOF	0.851	0.057	0.092
KCNQ2	114	T	S	NM_172107.4(KCNQ2):c.341C>G (p.Thr114Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 25, 2019)	criteria provided, single submitter	VCV000952581	20	63446793	952581	929252	rs1057516077	LOF	0.841	0.053	0.106
KCNQ2	119	E	G	NM_172107.4(KCNQ2):c.356A>G (p.Glu119Gly)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021786	20	63446778	21786	34638	rs118192193	LOF	0.866	0.048	0.086
KCNQ2	122	S	L	NM_172107.4(KCNQ2):c.365C>T (p.Ser122Leu)	Early infantile epileptic encephalopathy with suppression bursts|KCNQ2-Related Disorders| not provided|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7| Benign familial neonatal seizures 1	Pathogenic/Likely pathogenic(Last reviewed: Jun 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000021787	20	63446769	21787	34639	rs118192194	LOF	0.886	0.028	0.086
KCNQ2	127	Y	C	NM_172107.4(KCNQ2):c.380A>G (p.Tyr127Cys)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic/Likely pathogenic(Last reviewed: Dec 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205863	20	63446754	205863	203799	rs796052617	LOF	0.856	0.041	0.104
KCNQ2	129	L	V	NM_172107.4(KCNQ2):c.385C>G (p.Leu129Val)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001326323	20	63446749	1326323	1316725	NA	LOF	0.846	0.041	0.112
KCNQ2	130	E	K	NM_172107.4(KCNQ2):c.388G>A (p.Glu130Lys)	Early infantile epileptic encephalopathy 7	Pathogenic	no assertion criteria provided	VCV000219238	20	63445364	219238	217262	rs864321710	LOF	0.865	0.038	0.097
KCNQ2	132	V	L	NM_172107.4(KCNQ2):c.394G>T (p.Val132Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 7, 2018)	criteria provided, single submitter	VCV000656207	20	63445358	656207	650473	rs1600789325	LOF	0.836	0.04	0.124
KCNQ2	132	V	M	NM_172107.4(KCNQ2):c.394G>A (p.Val132Met)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Nov 1, 2021)	criteria provided, conflicting interpretations	VCV000872263	20	63445358	872263	860674	rs1600789325	LOF	0.843	0.04	0.117
KCNQ2	134	I	V	NM_172107.4(KCNQ2):c.400A>G (p.Ile134Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 13, 2020)	criteria provided, single submitter	VCV001009644	20	63445352	1009644	999090	rs2081381281	LOF	0.837	0.042	0.121

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	135	V	L	NM_172107.4(KCNQ2):c.403G>T (p.Val135Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 22, 2020)	criteria provided, single submitter	VCV000937749	20	63445349	937749	939035	rs775649577	LOF	0.836	0.04	0.124
KCNQ2	139	V	M	NM_172107.4(KCNQ2):c.415G>A (p.Val139Met)	not provided	Uncertain significance(Last reviewed: Jul 19, 2017)	criteria provided, single submitter	VCV000450575	20	63445337	450575	446293	rs773083197	LOF	0.844	0.04	0.117
KCNQ2	144	R	G	NM_172107.4(KCNQ2):c.430C>G (p.Arg144Gly)	Early infantile epileptic encephalopathy 7|KCNQ2-Related Disorders|Inborn genetic diseases	Likely pathogenic(Last reviewed: Dec 6, 2019)	criteria provided, multiple submitters, no conflicts	VCV000520399	20	63445322	520399	511019	rs1555873985	LOF	0.773	0.077	0.15
KCNQ2	144	R	Q	NM_172107.4(KCNQ2):c.431G>A (p.Arg144Gln)	Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7|Intellectual disability|not provided	Pathogenic/Likely pathogenic(Last reviewed: Oct 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205864	20	63445321	205864	203798	rs796052618	LOF	0.724	0.047	0.229
KCNQ2	144	R	W	NM_172107.4(KCNQ2):c.430C>T (p.Arg144Trp)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy 7|Intellectual disability|Inborn genetic diseases|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 1, 2020)	criteria provided, conflicting interpretations	VCV000452487	20	63445322	452487	446292	NA	LOF	0.825	0.039	0.136
KCNQ2	146	W	R	NM_172107.4(KCNQ2):c.436T>A (p.Trp146Arg)	not specified	Uncertain significance(Last reviewed: Jun 26, 2017)	criteria provided, single submitter	VCV000432987	20	63445316	432987	426355	rs1555873982	LOF	0.876	0.039	0.084
KCNQ2	147	A	P	NM_172107.4(KCNQ2):c.439G>C (p.Ala147Pro)	Seizures	Uncertain significance(Last reviewed: Jan 30, 2017)	criteria provided, single submitter	VCV000590026	20	63445313	590026	580568	rs1568941739	LOF	0.829	0.089	0.082
KCNQ2	148	A	T	NM_172107.4(KCNQ2):c.442G>A (p.Ala148Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 16, 2019)	criteria provided, single submitter	VCV000967422	20	63445310	967422	958870	rs2081379814	LOF	0.847	0.066	0.088
KCNQ2	154	Y	D	NM_172107.4(KCNQ2):c.460T>G (p.Tyr154Asp)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369743	20	63445292	369743	354030	rs1057516078	LOF	0.871	0.043	0.086
KCNQ2	155	R	C	NM_172107.4(KCNQ2):c.463C>T (p.Arg155Cys)	not provided	Uncertain significance(Last reviewed: Oct 2, 2015)	criteria provided, single submitter	VCV000283735	20	63445289	283735	267972	rs886042698	LOF	0.859	0.042	0.098
KCNQ2	155	R	H	NM_172107.4(KCNQ2):c.464G>A (p.Arg155His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 4, 2019)	criteria provided, single submitter	VCV000965462	20	63445288	965462	958869	rs779076192	LOF	0.867	0.036	0.097
KCNQ2	158	R	K	NM_172107.4(KCNQ2):c.473G>A (p.Arg158Lys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, single submitter	VCV001053573	20	63445279	1053573	1051783	NA	LOF	0.827	0.09	0.083
KCNQ2	159	G	E	NM_172107.4(KCNQ2):c.476G>A (p.Gly159Glu)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369747	20	63445276	369747	354027	rs1057516081	LOF	0.852	0.056	0.093
KCNQ2	159	G	R	NM_172107.4(KCNQ2):c.475G>A (p.Gly159Arg)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369746	20	63445277	369746	354028	rs1057516080	LOF	0.867	0.045	0.088
KCNQ2	159	G	V	NM_172107.4(KCNQ2):c.476G>T (p.Gly159Val)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Mar 27, 2017)	no assertion criteria provided	VCV000438297	20	63445276	438297	431924	rs1057516081	LOF	0.881	0.042	0.076
KCNQ2	160	R	P	NM_172107.4(KCNQ2):c.479G>C	not specified	Uncertain	criteria	VCV000373279	20	63445273	373279	360501	rs1057518323	LOF	0.824	0.05	0.126

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Arg160Pro)		significance(Last reviewed: Nov 16, 2016)	provided, single submitter										
KCNQ2	160	R	W	NM_172107.4(KCNQ2):c.478C>T (p.Arg160Trp)	not provided	Likely pathogenic(Last reviewed: May 29, 2019)	criteria provided, single submitter	VCV001215427	20	63445274	1215427	1205409	NA	LOF	0.851	0.033	0.116
KCNQ2	161	L	P	NM_172107.4(KCNQ2):c.482T>C (p.Leu161Pro)	not provided	Uncertain significance(Last reviewed: Mar 4, 2019)	criteria provided, single submitter	VCV001307528	20	63445270	1307528	1298798	NA	LOF	0.849	0.065	0.086
KCNQ2	162	K	R	NM_172107.4(KCNQ2):c.485A>G (p.Lys162Arg)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 21, 2019)	criteria provided, multiple submitters, no conflicts	VCV000392510	20	63445267	392510	378416	rs754250122	LOF	0.804	0.04	0.156
KCNQ2	169	C	F	NM_172107.4(KCNQ2):c.506G>T (p.Cys169Phe)	not provided|KCNQ2-Related Disorders	Likely pathogenic(Last reviewed: Mar 11, 2021)	criteria provided, multiple submitters, no conflicts	VCV000418726	20	63445246	418726	410832	rs1064793392	LOF	0.845	0.039	0.116
KCNQ2	169	C	R	NM_172107.4(KCNQ2):c.505T>C (p.Cys169Arg)	not provided	Likely pathogenic(Last reviewed: Mar 5, 2020)	criteria provided, single submitter	VCV000450447	20	63445247	450447	446291	rs1555873939	LOF	0.864	0.042	0.094
KCNQ2	170	V	L	NM_172107.4(KCNQ2):c.508G>C (p.Val170Leu)	not provided	Uncertain significance(Last reviewed: Mar 1, 2021)	criteria provided, single submitter	VCV001175999	20	63445244	1175999	1166355	NA	LOF	0.76	0.037	0.203
KCNQ2	172	D	G	NM_172107.4(KCNQ2):c.515A>G (p.Asp172Gly)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Dec 14, 2017)	criteria provided, single submitter	VCV000461422	20	63444834	461422	470490	rs1555873823	LOF	0.843	0.065	0.092
KCNQ2	175	V	L	NM_172107.4(KCNQ2):c.523G>C (p.Val175Leu)	not provided|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: May 29, 2018)	criteria provided, single submitter	VCV000369748	20	63444826	369748	354026	rs1057516082	LOF	0.75	0.036	0.213
KCNQ2	177	I	V	NM_172107.4(KCNQ2):c.529A>G (p.Ile177Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 4, 2019)	criteria provided, single submitter	VCV000934162	20	63444820	934162	939034	rs542553752	LOF	0.797	0.047	0.156
KCNQ2	178	A	T	NM_172107.4(KCNQ2):c.532G>A (p.Ala178Thr)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 28, 2019)	criteria provided, multiple submitters, no conflicts	VCV000444570	20	63444817	444570	438210	rs1555873790	LOF	0.834	0.055	0.111
KCNQ2	178	A	V	NM_172107.4(KCNQ2):c.533C>T (p.Ala178Val)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001326315	20	63444816	1326315	1316717	NA	LOF	0.824	0.047	0.129
KCNQ2	182	V	M	NM_172107.4(KCNQ2):c.544G>A (p.Val182Met)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Apr 8, 2020)	criteria provided, conflicting interpretations	VCV000427046	20	63444805	427046	415697	rs1085307920	LOF	0.77	0.037	0.193
KCNQ2	185	A	S	NM_172107.4(KCNQ2):c.553G>T (p.Ala185Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 14, 2019)	criteria provided, single submitter	VCV000952406	20	63444796	952406	929251	rs1600786349	LOF	0.828	0.068	0.105
KCNQ2	185	A	T	NM_172107.4(KCNQ2):c.553G>A (p.Ala185Thr)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|not provided|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Nov 1,	criteria provided, conflicting interpretations	VCV000809271	20	63444796	809271	798007	rs1600786349	LOF	0.834	0.058	0.108

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2021)											
KCNQ2	185	A	V	NM_172107.4(KCNQ2):c.554C>T (p.Ala185Val)	not provided	Uncertain significance(Last reviewed: May 11, 2017)	criteria provided, single submitter	VCV000429684	20	63444795	429684	422345	rs1131691529	LOF	0.839	0.045	0.116
KCNQ2	186	G	S	NM_172107.4(KCNQ2):c.556G>A (p.Gly186Ser)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Jan 6, 2022)	criteria provided, conflicting interpretations	VCV000958083	20	63444793	958083	951148	rs2081363570	LOF	0.86	0.054	0.085
KCNQ2	187	S	F	NM_172107.4(KCNQ2):c.560C>T (p.Ser187Phe)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 7, 2020)	criteria provided, single submitter	VCV001020770	20	63444789	1020770	1014210	rs2081363375	LOF	0.877	0.034	0.089
KCNQ2	188	Q	K	NM_172107.4(KCNQ2):c.562C>A (p.Gln188Lys)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Jul 7, 2020)	no assertion criteria provided	VCV000978712	20	63444787	978712	966804	rs2081363302	LOF	0.885	0.038	0.077
KCNQ2	188	Q	P	NM_172107.4(KCNQ2):c.563A>C (p.Gln188Pro)	not provided	Uncertain significance(Last reviewed: Apr 1, 2019)	criteria provided, single submitter	VCV000425272	20	63444786	425272	413606	rs1064797285	LOF	0.882	0.045	0.074
KCNQ2	189	G	V	NM_172107.4(KCNQ2):c.566G>T (p.Gly189Val)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369751	20	63444783	369751	354025	rs1057516083	LOF	0.891	0.037	0.073
KCNQ2	190	N	K	NM_172107.4(KCNQ2):c.570C>A (p.Asn190Lys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 23, 2020)	criteria provided, single submitter	VCV001058699	20	63444779	1058699	1051782	NA	LOF	0.874	0.032	0.094
KCNQ2	191	V	I	NM_172107.4(KCNQ2):c.571G>A (p.Val191Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 5, 2017)	criteria provided, single submitter	VCV000530415	20	63444778	530415	534212	rs745564113	LOF	0.871	0.033	0.096
KCNQ2	193	A	D	NM_172107.4(KCNQ2):c.578C>A (p.Ala193Asp)	not provided	Pathogenic(Last reviewed: May 3, 2012)	criteria provided, single submitter	VCV000205865	20	63444771	205865	203796	rs796052619	LOF	0.857	0.066	0.078
KCNQ2	193	A	V	NM_172107.4(KCNQ2):c.578C>T (p.Ala193Val)	Early infantile epileptic encephalopathy 7| Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Sep 15, 2020)	criteria provided, conflicting interpretations	VCV000977299	20	63444771	977299	965428	rs796052619	LOF	0.877	0.044	0.079
KCNQ2	194	T	I	NM_172107.4(KCNQ2):c.581C>T (p.Thr194Ile)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Feb 14, 2020)	criteria provided, single submitter	VCV000870206	20	63444768	870206	858357	rs2081362775	LOF	0.879	0.031	0.09
KCNQ2	195	S	C	NM_172107.4(KCNQ2):c.584C>G (p.Ser195Cys)	Early infantile epileptic encephalopathy 7| Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000982654	20	63444765	982654	971152	rs1568940442	LOF	0.866	0.041	0.094
KCNQ2	195	S	F	NM_172107.4(KCNQ2):c.584C>T (p.Ser195Phe)	Seizures|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Jan 15, 2020)	criteria provided, conflicting interpretations	VCV000590184	20	63444765	590184	580684	rs1568940442	LOF	0.881	0.032	0.087
KCNQ2	195	S	P	NM_172107.4(KCNQ2):c.583T>C (p.Ser195Pro)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile epileptic encephalopathy 7	Pathogenic/Likely pathogenic(Last reviewed: Feb 2, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205866	20	63444766	205866	203795	rs796052620	LOF	0.851	0.057	0.091
KCNQ2	196	A	V	NM_172107.4(KCNQ2):c.587C>T (p.Ala196Val)	Early infantile epileptic encephalopathy 7| Benign familial neonatal seizures 1|Early	Pathogenic(Last reviewed: Sep 16,	criteria provided,	VCV000021792	20	63444762	21792	34644	rs118192199	LOF	0.857	0.045	0.098

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					infantile epileptic encephalopathy with suppression bursts|not provided	2021)	multiple submitters, no conflicts										
KCNQ2	197	L	P	NM_172107.4(KCNQ2):c.590T>C (p.Leu197Pro)	Epileptic encephalopathy	Likely pathogenic	no assertion criteria provided	VCV000812790	20	63444759	812790	801203	rs1600786071	LOF	0.851	0.052	0.097
KCNQ2	198	R	P	NM_172107.4(KCNQ2):c.593G>C (p.Arg198Pro)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV001066808	20	63444756	1066808	1056624	NA	GOF	0.564	0.044	0.392
KCNQ2	198	R	Q	NM_172107.4(KCNQ2):c.593G>A (p.Arg198Gln)	Early infantile epileptic encephalopathy with suppression bursts|See cases|Inborn genetic diseases|not provided|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Apr 26, 2021)	criteria provided, multiple submitters, no conflicts	VCV000205867	20	63444756	205867	203794	rs796052621	GOF	0.478	0.035	0.486
KCNQ2	200	L	P	NM_172107.4(KCNQ2):c.599T>C (p.Leu200Pro)	not provided	Likely pathogenic(Last reviewed: Jul 23, 2013)	criteria provided, single submitter	VCV000205868	20	63444750	205868	203793	rs796052622	LOF	0.855	0.05	0.095
KCNQ2	201	R	C	NM_172107.4(KCNQ2):c.601C>T (p.Arg201Cys)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts|not provided|Benign familial neonatal seizures 1	Conflicting interpretations of pathogenicity(Last reviewed: Nov 16, 2021)	criteria provided, conflicting interpretations	VCV000205869	20	63444748	205869	203792	rs796052623	GOF	0.572	0.043	0.385
KCNQ2	201	R	H	NM_172107.4(KCNQ2):c.602G>A (p.Arg201His)	not provided|Severe intellectual deficiency|Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Apr 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000369753	20	63444747	369753	354023	rs1057516085	GOF	0.467	0.037	0.496
KCNQ2	203	L	P	NM_172107.4(KCNQ2):c.608T>C (p.Leu203Pro)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369754	20	63444741	369754	354022	rs1057516086	LOF	0.856	0.049	0.095
KCNQ2	204	Q	H	NM_172107.4(KCNQ2):c.612G>T (p.Gln204His)	not provided	Likely pathogenic(Last reviewed: Nov 23, 2016)	criteria provided, single submitter	VCV000205872	20	63444737	205872	203789	rs796052625	GOF	0.539	0.04	0.421
KCNQ2	204	Q	H	NM_172107.4(KCNQ2):c.612G>T (p.Gln204His)	not provided	Likely pathogenic(Last reviewed: Nov 23, 2016)	criteria provided, single submitter	VCV000205872	20	63444737	205872	203789	rs796052625	GOF	0.539	0.04	0.421
KCNQ2	204	Q	H	NM_172107.4(KCNQ2):c.612G>C (p.Gln204His)	not provided	Likely pathogenic(Last reviewed: Sep 2, 2020)	criteria provided, single submitter	VCV000205871	20	63444737	205871	203790	rs796052625	GOF	0.539	0.04	0.421
KCNQ2	204	Q	H	NM_172107.4(KCNQ2):c.612G>C (p.Gln204His)	not provided	Likely pathogenic(Last reviewed: Sep 2, 2020)	criteria provided, single submitter	VCV000205871	20	63444737	205871	203790	rs796052625	GOF	0.539	0.04	0.421
KCNQ2	204	Q	R	NM_172107.4(KCNQ2):c.611A>G (p.Gln204Arg)	not provided	Pathogenic(Last reviewed: Sep 11, 2012)	criteria provided, single submitter	VCV000205870	20	63444738	205870	203791	rs796052624	GOF	0.629	0.036	0.335
KCNQ2	205	I	M	NM_172107.4(KCNQ2):c.615T>G (p.Ile205Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 8, 2020)	criteria provided, single submitter	VCV000851348	20	63444734	851348	848544	rs971211988	LOF	0.861	0.032	0.106
KCNQ2	205	I	V	NM_172107.4(KCNQ2):c.613A>G (p.Ile205Val)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369755	20	63444736	369755	354021	rs1057516087	LOF	0.863	0.034	0.103
KCNQ2	206	L	P	NM_172107.4(KCNQ2):c.617T>C (p.Leu206Pro)	Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000803624	20	63444732	803624	792001	rs1339542565	LOF	0.858	0.048	0.094

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	206	L	R	NM_172107.4(KCNQ2):c.617T>G (p.Leu206Arg)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001326322	20	63444732	1326322	1316724	NA	LOF	0.868	0.035	0.097
KCNQ2	207	R	Q	NM_172107.4(KCNQ2):c.620G>A (p.Arg207Gln)	Early infantile epileptic encephalopathy with suppression bursts|Seizures, benign familial neonatal, 1, and/or myokymia|Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 1|not provided|Early infantile epileptic encephalopathy 7	Pathogenic/Likely pathogenic(Last reviewed: Mar 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000007391	20	63444729	7391	22430	rs118192200	GOF	0.474	0.036	0.49
KCNQ2	207	R	W	NM_172107.4(KCNQ2):c.619C>T (p.Arg207Trp)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|Seizures, benign familial neonatal, 1, and/or myokymia|Inborn genetic diseases|not provided	Pathogenic(Last reviewed: Sep 15, 2021)	criteria provided, multiple submitters, no conflicts	VCV000007386	20	63444730	7386	22425	rs74315391	GOF	0.629	0.036	0.336
KCNQ2	208	M	V	NM_172107.4(KCNQ2):c.622A>G (p.Met208Val)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Aug 15, 2019)	criteria provided, single submitter	VCV000021793	20	63444727	21793	34645	rs118192201	LOF	0.868	0.034	0.098
KCNQ2	209	I	T	NM_172107.4(KCNQ2):c.626T>C (p.Ile209Thr)	Seizures	Pathogenic(Last reviewed: Jan 28, 2020)	no assertion criteria provided	VCV000812512	20	63444723	812512	800755	rs1600785769	LOF	0.861	0.037	0.102
KCNQ2	210	R	C	NM_172107.4(KCNQ2):c.628C>T (p.Arg210Cys)	Intellectual disability|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Aug 29, 2019)	criteria provided, single submitter	VCV000205873	20	63444721	205873	203788	rs796052626	GOF	0.583	0.042	0.375
KCNQ2	210	R	H	NM_172107.4(KCNQ2):c.629G>A (p.Arg210His)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic(Last reviewed: Sep 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000279931	20	63444720	279931	264902	rs886041262	GOF	0.485	0.036	0.479
KCNQ2	210	R	P	NM_172107.4(KCNQ2):c.629G>C (p.Arg210Pro)	Early infantile epileptic encephalopathy 7|not provided	Pathogenic(Last reviewed: May 25, 2018)	criteria provided, multiple submitters, no conflicts	VCV000280614	20	63444720	280614	265110	rs886041262	GOF	0.575	0.043	0.382
KCNQ2	211	M	V	NM_172107.4(KCNQ2):c.631A>G (p.Met211Val)	not provided	Likely pathogenic(Last reviewed: Jan 11, 2017)	criteria provided, single submitter	VCV000392705	20	63444718	392705	378211	rs1057524599	LOF	0.869	0.033	0.098
KCNQ2	212	D	A	NM_172107.4(KCNQ2):c.635A>C (p.Asp212Ala)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Jun 17, 2019)	criteria provided, single submitter	VCV000944385	20	63444714	944385	929250	rs118192202	LOF	0.822	0.04	0.137
KCNQ2	212	D	E	NM_172107.4(KCNQ2):c.636C>A (p.Asp212Glu)	not provided	Pathogenic(Last reviewed: Apr 10, 2014)	criteria provided, single submitter	VCV000205874	20	63444713	205874	203787	rs796052627	LOF	0.797	0.041	0.162
KCNQ2	212	D	G	NM_172107.4(KCNQ2):c.635A>G (p.Asp212Gly)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021794	20	63444714	21794	34646	rs118192202	LOF	0.809	0.061	0.131
KCNQ2	212	D	V	NM_172107.4(KCNQ2):c.635A>T (p.Asp212Val)	not provided	Likely pathogenic(Last reviewed: Apr 11, 2016)	criteria provided, single submitter	VCV000420930	20	63444714	420930	410831	rs118192202	LOF	0.856	0.032	0.112
KCNQ2	213	R	Q	NM_172107.4(KCNQ2):c.638G>A (p.Arg213Gln)	not provided|Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy 7	Pathogenic/Likely pathogenic(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000039760	20	63444711	39760	48359	NA	GOF	0.574	0.033	0.393
KCNQ2	213	R	W	NM_172107.4(KCNQ2):c.637C>T (p.Arg213Trp)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Oct 19, 2020)	criteria provided, multiple submitters, no conflicts	VCV000021795	20	63444712	21795	34647	rs118192203	GOF	0.696	0.034	0.27

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	214	R	P	NM_172107.4(KCNQ2):c.641G>C (p.Arg214Pro)	not provided	Likely pathogenic(Last reviewed: Dec 6, 2016)	criteria provided, single submitter	VCV000373698	20	63444708	373698	360499	rs1057518555	LOF	0.869	0.035	0.097
KCNQ2	214	R	Q	NM_172107.4(KCNQ2):c.641G>A (p.Arg214Gln)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Sep 9, 2020)	criteria provided, single submitter	VCV000578032	20	63444708	578032	575133	rs1057518555	LOF	0.86	0.029	0.11
KCNQ2	214	R	W	NM_172107.4(KCNQ2):c.640C>T (p.Arg214Trp)	Benign familial neonatal seizures 1|not provided	Pathogenic/Likely pathogenic(Last reviewed: Nov 1, 2019)	criteria provided, multiple submitters, no conflicts	VCV000007385	20	63444709	7385	22424	rs28939684	LOF	0.868	0.032	0.099
KCNQ2	215	G	R	NM_172107.4(KCNQ2):c.643G>A (p.Gly215Arg)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369756	20	63444706	369756	354020	rs1057516088	LOF	0.878	0.034	0.088
KCNQ2	217	T	A	NM_172107.4(KCNQ2):c.649A>G (p.Thr217Ala)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 30, 2020)	criteria provided, single submitter	VCV000369757	20	63444700	369757	354019	rs1057516089	LOF	0.807	0.041	0.153
KCNQ2	217	T	I	NM_172107.4(KCNQ2):c.650C>T (p.Thr217Ile)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy 7	Conflicting interpretations of pathogenicity(Last reviewed: Nov 1, 2021)	criteria provided, conflicting interpretations	VCV000813779	20	63444699	813779	802038	NA	LOF	0.855	0.03	0.115
KCNQ2	217	T	N	NM_172107.4(KCNQ2):c.650C>A (p.Thr217Asn)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369758	20	63444699	369758	354018	rs1057516090	LOF	0.765	0.049	0.186
KCNQ2	217	T	P	NM_172107.4(KCNQ2):c.649A>C (p.Thr217Pro)	Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000803623	20	63444700	803623	792000	rs1057516089	LOF	0.813	0.051	0.136
KCNQ2	219	K	N	NM_172107.4(KCNQ2):c.657G>C (p.Lys219Asn)	not provided	Likely pathogenic(Last reviewed: Jan 10, 2017)	criteria provided, single submitter	VCV000391958	20	63444692	391958	377148	rs770290221	LOF	0.802	0.041	0.157
KCNQ2	223	S	F	NM_172107.4(KCNQ2):c.668C>T (p.Ser223Phe)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001326326	20	63444681	1326326	1316728	NA	LOF	0.864	0.033	0.103
KCNQ2	223	S	P	NM_172107.4(KCNQ2):c.667T>C (p.Ser223Pro)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Dec 20, 2019)	criteria provided, single submitter	VCV000841056	20	63444682	841056	848543	rs2081358991	LOF	0.851	0.05	0.099
KCNQ2	224	V	L	NM_172107.4(KCNQ2):c.670G>C (p.Val224Leu)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Feb 24, 2021)	criteria provided, single submitter	VCV001048634	20	63444679	1048634	1036749	NA	LOF	0.848	0.034	0.118
KCNQ2	228	H	Q	NM_172107.4(KCNQ2):c.684C>A (p.His228Gln)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Mar 4, 2020)	criteria provided, single submitter	VCV000021797	20	63444665	21797	34649	rs118192204	LOF	0.848	0.035	0.118
KCNQ2	228	H	R	NM_172107.4(KCNQ2):c.683A>G (p.His228Arg)	KCNQ2-Related Disorders	not provided	no assertion provided	VCV000972975	20	63444666	972975	961353	rs2081358611	LOF	0.863	0.032	0.105
KCNQ2	228	H	Y	NM_172107.4(KCNQ2):c.682C>T (p.His228Tyr)	not provided	Likely pathogenic(Last reviewed: Aug 31, 2020)	criteria provided, single submitter	VCV000432161	20	63444667	432161	426354	rs1555873665	LOF	0.866	0.031	0.103
KCNQ2	230	K	M	NM_172107.4(KCNQ2):c.689A>T (p.Lys230Met)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Jun 19, 2019)	criteria provided, single submitter	VCV000933711	20	63444660	933711	939033	rs2081358317	LOF	0.83	0.03	0.14

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	231	E	D	NM_172107.4(KCNQ2):c.693G>T (p.Glu231Asp)	Seizures	Pathogenic/Likely pathogenic(Last reviewed: Dec 13, 2016)	criteria provided, multiple submitters, no conflicts	VCV000211234	20	63442529	211234	208706	rs797044938	LOF	0.833	0.049	0.118
KCNQ2	231	E	D	NM_172107.4(KCNQ2):c.693G>T (p.Glu231Asp)	Seizures	Pathogenic/Likely pathogenic(Last reviewed: Dec 13, 2016)	criteria provided, multiple submitters, no conflicts	VCV000211234	20	63442529	211234	208706	rs797044938	LOF	0.833	0.049	0.118
KCNQ2	231	E	D	NM_172107.4(KCNQ2):c.693G>C (p.Glu231Asp)	Inborn genetic diseases	Pathogenic(Last reviewed: Apr 8, 2015)	criteria provided, single submitter	VCV000208761	20	63442529	208761	205322	rs797044938	LOF	0.833	0.049	0.118
KCNQ2	231	E	D	NM_172107.4(KCNQ2):c.693G>C (p.Glu231Asp)	Inborn genetic diseases	Pathogenic(Last reviewed: Apr 8, 2015)	criteria provided, single submitter	VCV000208761	20	63442529	208761	205322	rs797044938	LOF	0.833	0.049	0.118
KCNQ2	231	E	G	NM_172107.4(KCNQ2):c.692A>G (p.Glu231Gly)	not provided	Pathogenic(Last reviewed: May 9, 2013)	criteria provided, single submitter	VCV000205876	20	63442530	205876	203784	rs796052629	LOF	0.838	0.058	0.103
KCNQ2	231	E	Q	NM_172107.4(KCNQ2):c.691G>C (p.Glu231Gln)	not provided	Pathogenic(Last reviewed: Aug 18, 2015)	criteria provided, single submitter	VCV000419718	20	63442531	419718	410830	rs1064794063	LOF	0.839	0.039	0.122
KCNQ2	234	T	I	NM_172107.4(KCNQ2):c.701C>T (p.Thr234Ile)	Early infantile epileptic encephalopathy with suppression bursts|Seizures|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 22, 2019)	criteria provided, conflicting interpretations	VCV000197892	20	63442521	197892	195053	rs794727741	LOF	0.864	0.033	0.103
KCNQ2	234	T	P	NM_172107.4(KCNQ2):c.700A>C (p.Thr234Pro)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369759	20	63442522	369759	354017	rs1057516091	LOF	0.839	0.062	0.098
KCNQ2	235	A	T	NM_172107.4(KCNQ2):c.703G>A (p.Ala235Thr)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Oct 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000872261	20	63442519	872261	860672	rs2081192615	LOF	0.845	0.062	0.093
KCNQ2	235	A	V	NM_172107.4(KCNQ2):c.704C>T (p.Ala235Val)	Seizures|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Aug 16, 2018)	criteria provided, multiple submitters, no conflicts	VCV000211235	20	63442518	211235	208705	rs797045638	LOF	0.861	0.045	0.094
KCNQ2	236	W	R	NM_172107.4(KCNQ2):c.706T>C (p.Trp236Arg)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Oct 2, 2021)	criteria provided, single submitter	VCV001320124	20	63442516	1320124	1310369	NA	LOF	0.875	0.036	0.089
KCNQ2	237	Y	C	NM_172107.4(KCNQ2):c.710A>G (p.Tyr237Cys)	Inborn genetic diseases	Pathogenic(Last reviewed: Sep 24, 2013)	criteria provided, single submitter	VCV000208758	20	63442512	208758	205321	rs797044935	LOF	0.848	0.042	0.11
KCNQ2	238	I	L	NM_172107.4(KCNQ2):c.712A>C (p.Ile238Leu)	not provided	Likely pathogenic(Last reviewed: Jan 30, 2018)	criteria provided, single submitter	VCV000421609	20	63442510	421609	410829	rs747050726	LOF	0.809	0.036	0.155
KCNQ2	238	I	V	NM_172107.4(KCNQ2):c.712A>G (p.Ile238Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 3, 2017)	criteria provided, single submitter	VCV000530433	20	63442510	530433	533636	rs747050726	LOF	0.822	0.039	0.139
KCNQ2	239	G	R	NM_172107.4(KCNQ2):c.715G>C (p.Gly239Arg)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369760	20	63442507	369760	354016	rs1057516092	LOF	0.879	0.041	0.08
KCNQ2	239	G	V	NM_172107.4(KCNQ2):c.716G>T (p.Gly239Val)	Benign familial neonatal seizures 1|not provided	Conflicting interpretations of pathogenicity(Last	criteria provided, conflicting	VCV000205877	20	63442506	205877	203783	rs796052630	LOF	0.88	0.037	0.082

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: May 28, 2019)	interpretations										
KCNQ2	242	C	G	NM_172107.4(KCNQ2):c.724T>G (p.Cys242Gly)	KCNQ2-Related Disorders	Likely pathogenic(Last reviewed: Mar 26, 2018)	no assertion criteria provided	VCV000545120	20	63442498	545120	535384	rs1555871915	LOF	0.848	0.069	0.083
KCNQ2	243	L	F	NM_172107.4(KCNQ2):c.727C>T (p.Leu243Phe)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021798	20	63442495	21798	34650	rs118192205	LOF	0.836	0.041	0.123
KCNQ2	243	L	V	NM_172107.4(KCNQ2):c.727C>G (p.Leu243Val)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Jan 6, 2018)	criteria provided, single submitter	VCV000692002	20	63442495	692002	679811	rs118192205	LOF	0.838	0.043	0.119
KCNQ2	245	L	P	NM_172107.4(KCNQ2):c.734T>C (p.Leu245Pro)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Sep 8, 2020)	criteria provided, single submitter	VCV001074752	20	63442488	1074752	1064859	NA	LOF	0.847	0.059	0.094
KCNQ2	247	S	L	NM_172107.4(KCNQ2):c.740C>T (p.Ser247Leu)	Inborn genetic diseases|not provided	Pathogenic/Likely pathogenic(Last reviewed: Dec 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205878	20	63442482	205878	203782	rs74315392	LOF	0.868	0.033	0.099
KCNQ2	247	S	P	NM_172107.4(KCNQ2):c.739T>C (p.Ser247Pro)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Aug 22, 2018)	criteria provided, single submitter	VCV000813771	20	63442483	813771	802036	NA	LOF	0.847	0.062	0.091
KCNQ2	247	S	W	NM_172107.4(KCNQ2):c.740C>G (p.Ser247Trp)	Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 2	Pathogenic(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV000007389	20	63442482	7389	22428	rs74315392	LOF	0.869	0.036	0.094
KCNQ2	248	F	S	NM_172107.4(KCNQ2):c.743T>C (p.Phe248Ser)	not provided	Likely pathogenic(Last reviewed: Feb 10, 2016)	criteria provided, single submitter	VCV000372909	20	63442479	372909	360484	rs1057518068	LOF	0.858	0.049	0.093
KCNQ2	249	L	P	NM_172107.4(KCNQ2):c.746T>C (p.Leu249Pro)	Intellectual disability	Likely pathogenic(Last reviewed: Dec 1, 2019)	no assertion criteria provided	VCV000870318	20	63442476	870318	858396	rs2081190512	LOF	0.851	0.056	0.092
KCNQ2	250	V	G	NM_172107.4(KCNQ2):c.749T>G (p.Val250Gly)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Jan 8, 2020)	criteria provided, single submitter	VCV000021800	20	63442473	21800	34652	rs118192206	LOF	0.832	0.076	0.092
KCNQ2	250	V	L	NM_172107.4(KCNQ2):c.748G>T (p.Val250Leu)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic(Last reviewed: Jul 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000872260	20	63442474	872260	860671	rs2081190344	LOF	0.837	0.036	0.127
KCNQ2	252	L	F	NM_172107.4(KCNQ2):c.756G>T (p.Leu252Phe)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 6, 2020)	criteria provided, single submitter	VCV001001558	20	63442466	1001558	999088	rs2081190032	LOF	0.854	0.043	0.103
KCNQ2	253	A	S	NM_172107.4(KCNQ2):c.757G>T (p.Ala253Ser)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Sep 29, 2020)	criteria provided, single submitter	VCV000975904	20	63442465	975904	964543	rs1057516093	LOF	0.829	0.088	0.084
KCNQ2	253	A	T	NM_172107.4(KCNQ2):c.757G>A (p.Ala253Thr)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369761	20	63442465	369761	354015	rs1057516093	LOF	0.843	0.075	0.083
KCNQ2	254	E	D	NM_172107.4(KCNQ2):c.762G>C (p.Glu254Asp)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Sep 7, 2016)	criteria provided, single submitter	VCV000976223	20	63442460	976223	964542	rs2081189736	LOF	0.816	0.069	0.115

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	254	E	G	NM_172107.4(KCNQ2):c.761A>G (p.Glu254Gly)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 14, 2019)	criteria provided, multiple submitters, no conflicts	VCV000197889	20	63442461	197889	195050	rs794727739	LOF	0.823	0.083	0.094
KCNQ2	255	K	R	NM_172107.4(KCNQ2):c.764A>G (p.Lys255Arg)	not provided	Likely pathogenic(Last reviewed: Jun 19, 2017)	criteria provided, single submitter	VCV000432713	20	63442458	432713	426353	rs1555871862	LOF	0.868	0.035	0.098
KCNQ2	256	G	R	NM_172107.4(KCNQ2):c.766G>A (p.Gly256Arg)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Dec 23, 2019)	criteria provided, single submitter	VCV000975979	20	63442456	975979	964541	rs1057518500	LOF	0.865	0.052	0.082
KCNQ2	256	G	R	NM_172107.4(KCNQ2):c.766G>A (p.Gly256Arg)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Dec 23, 2019)	criteria provided, single submitter	VCV000975979	20	63442456	975979	964541	rs1057518500	LOF	0.865	0.052	0.082
KCNQ2	256	G	R	NM_172107.4(KCNQ2):c.766G>C (p.Gly256Arg)	not specified	Uncertain significance(Last reviewed: Nov 28, 2016)	criteria provided, single submitter	VCV000373592	20	63442456	373592	360589	rs1057518500	LOF	0.865	0.052	0.082
KCNQ2	256	G	R	NM_172107.4(KCNQ2):c.766G>C (p.Gly256Arg)	not specified	Uncertain significance(Last reviewed: Nov 28, 2016)	criteria provided, single submitter	VCV000373592	20	63442456	373592	360589	rs1057518500	LOF	0.865	0.052	0.082
KCNQ2	256	G	V	NM_172107.4(KCNQ2):c.767G>T (p.Gly256Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV000945463	20	63442455	945463	929249	rs1371722284	LOF	0.871	0.048	0.081
KCNQ2	258	N	K	NM_172107.4(KCNQ2):c.774C>G (p.Asn258Lys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 18, 2018)	criteria provided, single submitter	VCV000566350	20	63442448	566350	572968	rs770187706	LOF	0.875	0.039	0.086
KCNQ2	258	N	S	NM_172107.4(KCNQ2):c.773A>G (p.Asn258Ser)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021801	20	63442449	21801	34653	rs118192207	LOF	0.868	0.044	0.088
KCNQ2	259	D	E	NM_172107.4(KCNQ2):c.777C>A (p.Asp259Glu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 20, 2019)	criteria provided, single submitter	VCV000852143	20	63442445	852143	848541	rs2081188808	LOF	0.818	0.084	0.098
KCNQ2	259	D	G	NM_172107.4(KCNQ2):c.776A>G (p.Asp259Gly)	not provided	Likely pathogenic(Last reviewed: Nov 28, 2016)	criteria provided, single submitter	VCV000373573	20	63442446	373573	360437	rs1057518489	LOF	0.816	0.087	0.097
KCNQ2	259	D	N	NM_172107.4(KCNQ2):c.775G>A (p.Asp259Asn)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Oct 4, 2019)	criteria provided, conflicting interpretations	VCV000692196	20	63442447	692196	679939	rs777257591	LOF	0.817	0.084	0.099
KCNQ2	259	D	Y	NM_172107.4(KCNQ2):c.775G>T (p.Asp259Tyr)	Benign familial neonatal seizures 1	Pathogenic	no assertion criteria provided	VCV000219240	20	63442447	219240	217261	rs777257591	LOF	0.816	0.075	0.108
KCNQ2	260	H	Y	NM_172107.4(KCNQ2):c.778C>T (p.His260Tyr)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Jan 1, 2018)	criteria provided, single submitter	VCV000495248	20	63442444	495248	486774	rs1555871832	LOF	0.872	0.04	0.088
KCNQ2	261	F	C	NM_172107.4(KCNQ2):c.782T>G (p.Phe261Cys)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Jun 7, 2019)	criteria provided, multiple submitters, no conflicts	VCV000946410	20	63442440	946410	929248	rs796052631	LOF	0.87	0.042	0.088
KCNQ2	261	F	S	NM_172107.4(KCNQ2):c.782T>C	Seizures|not provided	Conflicting	criteria	VCV000205879	20	63442440	205879	203781	rs796052631	LOF	0.883	0.044	0.073

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Phe261Ser)		interpretations of pathogenicity(Last reviewed: Jun 27, 2016)	provided, conflicting interpretations										
KCNQ2	263	T	A	NM_172107.4(KCNQ2):c.787A>G (p.Thr263Ala)	Epileptic encephalopathy, early infantile, 1	Likely pathogenic(Last reviewed: Jan 1, 2019)	no assertion criteria provided	VCV000975414	20	63442435	975414	963917	rs2081188385	LOF	0.853	0.059	0.088
KCNQ2	263	T	I	NM_172107.4(KCNQ2):c.788C>T (p.Thr263Ile)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: May 8, 2019)	criteria provided, single submitter	VCV000571819	20	63442434	571819	573613	rs1568932462	LOF	0.848	0.056	0.096
KCNQ2	264	Y	N	NM_172107.4(KCNQ2):c.790T>A (p.Tyr264Asn)	not provided	Likely pathogenic(Last reviewed: Sep 27, 2015)	criteria provided, single submitter	VCV000205880	20	63442432	205880	203780	rs796052632	LOF	0.854	0.048	0.098
KCNQ2	265	A	P	NM_172107.4(KCNQ2):c.793G>C (p.Ala265Pro)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369762	20	63442429	369762	354014	rs794727740	LOF	0.837	0.086	0.077
KCNQ2	265	A	T	NM_172107.4(KCNQ2):c.793G>A (p.Ala265Thr)	Seizures|Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile epileptic encephalopathy 7	Pathogenic/Likely pathogenic(Last reviewed: Oct 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000197891	20	63442429	197891	195052	rs794727740	LOF	0.85	0.069	0.082
KCNQ2	265	A	V	NM_172107.4(KCNQ2):c.794C>T (p.Ala265Val)	Intellectual disability|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 1|KCNQ2-Related Disorders|Seizures|Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic/Likely pathogenic(Last reviewed: Aug 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000120176	20	63442428	120176	125784	rs587777219	LOF	0.873	0.05	0.076
KCNQ2	266	D	E	NM_172107.4(KCNQ2):c.798T>A (p.Asp266Glu)	Epileptic encephalopathy	Pathogenic(Last reviewed: Nov 16, 2016)	criteria provided, single submitter	VCV000375518	20	63442424	375518	362349	rs1057519536	LOF	0.835	0.056	0.109
KCNQ2	266	D	G	NM_172107.4(KCNQ2):c.797A>G (p.Asp266Gly)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Sep 1, 2020)	criteria provided, single submitter	VCV000852899	20	63442425	852899	848540	rs2081187692	LOF	0.824	0.087	0.089
KCNQ2	267	A	T	NM_172107.4(KCNQ2):c.799G>A (p.Ala267Thr)	not provided	Likely pathogenic(Last reviewed: May 2, 2019)	criteria provided, single submitter	VCV001210663	20	63442423	1210663	1200662	NA	LOF	0.827	0.084	0.089
KCNQ2	267	A	V	NM_172107.4(KCNQ2):c.800C>T (p.Ala267Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 9, 2019)	criteria provided, single submitter	VCV000951223	20	63442422	951223	929247	rs2081187554	LOF	0.861	0.059	0.079
KCNQ2	268	L	F	NM_172107.4(KCNQ2):c.802C>T (p.Leu268Phe)	Epileptic encephalopathy|not provided|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Dec 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000369763	20	63442420	369763	354013	rs1057516094	LOF	0.866	0.042	0.093
KCNQ2	268	L	P	NM_172107.4(KCNQ2):c.803T>C (p.Leu268Pro)	Early infantile epileptic encephalopathy 7	Pathogenic	no assertion criteria provided	VCV000219236	20	63442419	219236	217260	rs864321708	LOF	0.86	0.063	0.078
KCNQ2	269	W	L	NM_172107.4(KCNQ2):c.806G>T (p.Trp269Leu)	not provided	Likely pathogenic(Last reviewed: Jan 9, 2013)	criteria provided, single submitter	VCV000205881	20	63442416	205881	203778	rs796052633	LOF	0.879	0.036	0.085
KCNQ2	269	W	S	NM_172107.4(KCNQ2):c.806G>C (p.Trp269Ser)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Mar 29, 2021)	criteria provided, single submitter	VCV001285561	20	63442416	1285561	1275404	NA	LOF	0.879	0.045	0.076
KCNQ2	270	W	C	NM_172107.4(KCNQ2):c.810G>T (p.Trp270Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 27,	criteria provided, single submitter	VCV000851253	20	63442412	851253	848539	rs2081187047	LOF	0.87	0.041	0.089

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)											
KCNQ2	271	G	D	NM_172107.4(KCNQ2):c.812G>A (p.Gly271Asp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 22, 2018)	criteria provided, single submitter	VCV000565381	20	63442410	565381	573608	rs118192209	LOF	0.834	0.077	0.089
KCNQ2	271	G	R	NM_172107.4(KCNQ2):c.811G>C (p.Gly271Arg)	not provided	Likely pathogenic(Last reviewed: Oct 1, 2016)	criteria provided, single submitter	VCV000425271	20	63442411	425271	413605	rs1064797284	LOF	0.868	0.047	0.084
KCNQ2	271	G	V	NM_172107.4(KCNQ2):c.812G>T (p.Gly271Val)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021803	20	63442410	21803	34655	rs118192209	LOF	0.881	0.045	0.074
KCNQ2	272	L	P	NM_172107.4(KCNQ2):c.815T>C (p.Leu272Pro)	not provided	Uncertain significance(Last reviewed: Jul 1, 2016)	criteria provided, single submitter	VCV000374744	20	63442407	374744	361630	rs1057519227	LOF	0.861	0.063	0.076
KCNQ2	273	I	M	NM_172107.4(KCNQ2):c.819C>G (p.Ile273Met)	not provided	Likely pathogenic(Last reviewed: Oct 20, 2016)	criteria provided, single submitter	VCV000422481	20	63439706	422481	410828	rs771282785	LOF	0.86	0.04	0.1
KCNQ2	273	I	N	NM_172107.4(KCNQ2):c.818T>A (p.Ile273Asn)	not provided	Likely pathogenic(Last reviewed: Jun 17, 2013)	criteria provided, single submitter	VCV000205883	20	63439707	205883	203776	rs796052635	LOF	0.859	0.057	0.084
KCNQ2	274	T	M	NM_172107.4(KCNQ2):c.821C>T (p.Thr274Met)	Early infantile epileptic encephalopathy 7|not provided|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Jan 3, 2022)	criteria provided, multiple submitters, no conflicts	VCV000167208	20	63439704	167208	177432	rs727503974	LOF	0.853	0.043	0.103
KCNQ2	275	L	V	NM_172107.4(KCNQ2):c.823C>G (p.Leu275Val)	not provided	Likely pathogenic(Last reviewed: Jul 17, 2019)	criteria provided, single submitter	VCV001177922	20	63439702	1177922	1167758	NA	LOF	0.865	0.044	0.091
KCNQ2	276	T	I	NM_172107.4(KCNQ2):c.827C>T (p.Thr276Ile)	not provided|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Sep 8, 2020)	criteria provided, single submitter	VCV000369764	20	63439698	369764	354012	rs1057516095	LOF	0.881	0.039	0.08
KCNQ2	276	T	N	NM_172107.4(KCNQ2):c.827C>A (p.Thr276Asn)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Nov 11, 2019)	criteria provided, single submitter	VCV000941582	20	63439698	941582	939031	rs1057516095	LOF	0.863	0.058	0.079
KCNQ2	277	T	N	NM_172107.4(KCNQ2):c.830C>A (p.Thr277Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 11, 2018)	criteria provided, single submitter	VCV000665007	20	63439695	665007	650413	rs1600755607	LOF	0.868	0.047	0.085
KCNQ2	277	T	P	NM_172107.4(KCNQ2):c.829A>C (p.Thr277Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 11, 2019)	criteria provided, single submitter	VCV000643905	20	63439696	643905	650412	rs1555870554	LOF	0.869	0.05	0.08
KCNQ2	277	T	S	NM_172107.4(KCNQ2):c.829A>T (p.Thr277Ser)	Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Aug 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000495250	20	63439696	495250	486773	rs1555870554	LOF	0.868	0.047	0.086
KCNQ2	278	I	N	NM_172107.4(KCNQ2):c.833T>A (p.Ile278Asn)	not provided	Likely pathogenic(Last reviewed: Oct 20, 2016)	criteria provided, single submitter	VCV000390326	20	63439692	390326	379751	rs1057523728	LOF	0.872	0.05	0.079
KCNQ2	278	I	T	NM_172107.4(KCNQ2):c.833T>C (p.Ile278Thr)	not provided|Early infantile epileptic encephalopathy 7	Pathogenic/Likely pathogenic(Last reviewed: Dec 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000429212	20	63439692	429212	422344	rs1057523728	LOF	0.87	0.045	0.084

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	279	G	C	NM_172107.4(KCNQ2):c.835G>T (p.Gly279Cys)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369765	20	63439690	369765	354011	rs1057516096	LOF	0.864	0.05	0.086
KCNQ2	280	Y	C	NM_172107.4(KCNQ2):c.839A>G (p.Tyr280Cys)	Seizures	Likely pathogenic(Last reviewed: Oct 12, 2016)	criteria provided, single submitter	VCV000589856	20	63439686	589856	580436	rs1568927820	LOF	0.86	0.05	0.09
KCNQ2	280	Y	H	NM_172107.4(KCNQ2):c.838T>C (p.Tyr280His)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Sep 25, 2020)	no assertion criteria provided	VCV000982412	20	63439687	982412	970531	rs2081102604	LOF	0.874	0.045	0.081
KCNQ2	281	G	E	NM_172107.4(KCNQ2):c.842G>A (p.Gly281Glu)	not provided	Likely pathogenic(Last reviewed: Aug 11, 2016)	criteria provided, single submitter	VCV000421991	20	63439683	421991	410827	rs1064795489	LOF	0.863	0.049	0.088
KCNQ2	281	G	R	NM_172107.4(KCNQ2):c.841G>C (p.Gly281Arg)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 8, 2017)	criteria provided, single submitter	VCV000488538	20	63439684	488538	481406	rs794727813	LOF	0.857	0.046	0.097
KCNQ2	281	G	R	NM_172107.4(KCNQ2):c.841G>C (p.Gly281Arg)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 8, 2017)	criteria provided, single submitter	VCV000488538	20	63439684	488538	481406	rs794727813	LOF	0.857	0.046	0.097
KCNQ2	281	G	R	NM_172107.4(KCNQ2):c.841G>A (p.Gly281Arg)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile epileptic encephalopathy 7	Conflicting interpretations of pathogenicity(Last reviewed: Aug 2, 2020)	criteria provided, conflicting interpretations	VCV000198284	20	63439684	198284	195445	rs794727813	LOF	0.857	0.046	0.097
KCNQ2	281	G	R	NM_172107.4(KCNQ2):c.841G>A (p.Gly281Arg)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile epileptic encephalopathy 7	Conflicting interpretations of pathogenicity(Last reviewed: Aug 2, 2020)	criteria provided, conflicting interpretations	VCV000198284	20	63439684	198284	195445	rs794727813	LOF	0.857	0.046	0.097
KCNQ2	281	G	W	NM_172107.4(KCNQ2):c.841G>T (p.Gly281Trp)	not provided|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Oct 3, 2017)	criteria provided, multiple submitters, no conflicts	VCV000216949	20	63439684	216949	213659	rs794727813	LOF	0.863	0.052	0.086
KCNQ2	282	D	H	NM_172107.4(KCNQ2):c.844G>C (p.Asp282His)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 31, 2020)	criteria provided, conflicting interpretations	VCV000430396	20	63439681	430396	422343	rs796052636	LOF	0.878	0.044	0.078
KCNQ2	282	D	N	NM_172107.4(KCNQ2):c.844G>A (p.Asp282Asn)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic/Likely pathogenic(Last reviewed: Jan 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205884	20	63439681	205884	203775	rs796052636	LOF	0.873	0.049	0.079
KCNQ2	282	D	V	NM_172107.4(KCNQ2):c.845A>T (p.Asp282Val)	Epileptic encephalopathy	Pathogenic	no assertion criteria provided	VCV000812789	20	63439680	812789	801202	rs1600755440	LOF	0.879	0.043	0.077
KCNQ2	283	K	E	NM_172107.4(KCNQ2):c.847A>G (p.Lys283Glu)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Feb 7, 2019)	criteria provided, single submitter	VCV000648864	20	63439678	648864	650414	rs1600755429	LOF	0.872	0.038	0.089
KCNQ2	283	K	R	NM_172107.4(KCNQ2):c.848A>G (p.Lys283Arg)	not provided	Pathogenic(Last reviewed: May 20, 2021)	criteria provided, single submitter	VCV000205885	20	63439677	205885	203774	rs796052637	LOF	0.88	0.035	0.085
KCNQ2	284	Y	C	NM_172107.4(KCNQ2):c.851A>G (p.Tyr284Cys)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000007381	20	63439674	7381	22420	rs28939683	LOF	0.863	0.04	0.098
KCNQ2	284	Y	D	NM_172107.4(KCNQ2):c.850T>G (p.Tyr284Asp)	Early infantile epileptic encephalopathy 7	Pathogenic	no assertion criteria provided	VCV000219234	20	63439675	219234	217259	rs864321706	LOF	0.876	0.043	0.081

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	285	P	H	NM_172107.4(KCNQ2):c.854C>A (p.Pro285His)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369766	20	63439671	369766	354010	rs1057516097	LOF	0.883	0.041	0.076
KCNQ2	285	P	L	NM_172107.4(KCNQ2):c.854C>T (p.Pro285Leu)	not provided	Likely pathogenic(Last reviewed: Sep 1, 2019)	criteria provided, single submitter	VCV000871424	20	63439671	871424	860670	rs1057516097	LOF	0.894	0.034	0.072
KCNQ2	285	P	S	NM_172107.4(KCNQ2):c.853C>T (p.Pro285Ser)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Jan 11, 2019)	criteria provided, single submitter	VCV000975869	20	63439672	975869	964540	rs2081101244	LOF	0.879	0.046	0.075
KCNQ2	287	T	I	NM_172107.4(KCNQ2):c.860C>T (p.Thr287Ile)	not provided	Uncertain significance(Last reviewed: Apr 7, 2014)	criteria provided, single submitter	VCV000167207	20	63439665	167207	177168	rs727503973	LOF	0.85	0.055	0.095
KCNQ2	287	T	N	NM_172107.4(KCNQ2):c.860C>A (p.Thr287Asn)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369767	20	63439665	369767	354009	rs727503973	LOF	0.853	0.065	0.082
KCNQ2	288	W	C	NM_172107.4(KCNQ2):c.864G>C (p.Trp288Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 2, 2019)	criteria provided, single submitter	VCV000838229	20	63439661	838229	848538	rs2081100544	LOF	0.865	0.044	0.091
KCNQ2	290	G	D	NM_172107.4(KCNQ2):c.869G>A (p.Gly290Asp)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|not provided	Pathogenic(Last reviewed: Oct 11, 2018)	criteria provided, single submitter	VCV000039762	20	63439656	39762	48361	rs397514582	LOF	0.826	0.083	0.091
KCNQ2	290	G	S	NM_172107.4(KCNQ2):c.868G>A (p.Gly290Ser)	not provided|Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Oct 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000369768	20	63439657	369768	354008	rs1057516098	LOF	0.833	0.081	0.086
KCNQ2	290	G	V	NM_172107.4(KCNQ2):c.869G>T (p.Gly290Val)	Generalized hypotonia|Seizures	Pathogenic(Last reviewed: Jan 1, 2017)	criteria provided, single submitter	VCV000523563	20	63439656	523563	514107	rs397514582	LOF	0.879	0.048	0.073
KCNQ2	291	R	S	NM_172107.4(KCNQ2):c.873G>C (p.Arg291Ser)	Seizures	Uncertain significance(Last reviewed: Apr 20, 2016)	criteria provided, single submitter	VCV000587825	20	63439652	587825	580433	rs1057519535	LOF	0.808	0.047	0.145
KCNQ2	291	R	S	NM_172107.4(KCNQ2):c.873G>C (p.Arg291Ser)	Seizures	Uncertain significance(Last reviewed: Apr 20, 2016)	criteria provided, single submitter	VCV000587825	20	63439652	587825	580433	rs1057519535	LOF	0.808	0.047	0.145
KCNQ2	291	R	S	NM_172107.4(KCNQ2):c.873G>T (p.Arg291Ser)	Epileptic encephalopathy	Likely pathogenic(Last reviewed: Nov 16, 2016)	criteria provided, single submitter	VCV000375517	20	63439652	375517	362348	rs1057519535	LOF	0.808	0.047	0.145
KCNQ2	291	R	S	NM_172107.4(KCNQ2):c.873G>T (p.Arg291Ser)	Epileptic encephalopathy	Likely pathogenic(Last reviewed: Nov 16, 2016)	criteria provided, single submitter	VCV000375517	20	63439652	375517	362348	rs1057519535	LOF	0.808	0.047	0.145
KCNQ2	292	L	P	NM_172107.4(KCNQ2):c.875T>C (p.Leu292Pro)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Jun 7, 2019)	criteria provided, multiple submitters, no conflicts	VCV000405208	20	63439650	405208	404202	rs1060500602	LOF	0.835	0.083	0.082
KCNQ2	293	L	F	NM_172107.4(KCNQ2):c.877C>T (p.Leu293Phe)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 7, 2019)	criteria provided, single submitter	VCV000948377	20	63439648	948377	929246	rs2081099943	LOF	0.838	0.057	0.105
KCNQ2	293	L	P	NM_172107.4(KCNQ2):c.878T>C (p.Leu293Pro)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: May 13, 2021)	no assertion criteria provided	VCV001164059	20	63439647	1164059	1153056	NA	LOF	0.824	0.092	0.084

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	294	A	G	NM_172107.4(KCNQ2):c.881C>G (p.Ala294Gly)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021805	20	63439644	21805	34657	rs118192211	Neutral	0.651	0.26	0.089
KCNQ2	294	A	S	NM_172107.4(KCNQ2):c.880G>T (p.Ala294Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 10, 2020)	criteria provided, single submitter	VCV001002473	20	63439645	1002473	999087	rs2081099858	Neutral	0.724	0.178	0.098
KCNQ2	294	A	V	NM_172107.4(KCNQ2):c.881C>T (p.Ala294Val)	Early infantile epileptic encephalopathy 7|Epileptic encephalopathy|Early infantile epileptic encephalopathy with suppression bursts|KCNQ2-Related Disorders|not provided	Pathogenic(Last reviewed: Dec 20, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205886	20	63439644	205886	203772	NA	LOF	0.815	0.095	0.09
KCNQ2	296	T	N	NM_172107.4(KCNQ2):c.887C>A (p.Thr296Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 6, 2019)	criteria provided, single submitter	VCV000940822	20	63439638	940822	939030	rs2081099327	LOF	0.837	0.073	0.091
KCNQ2	296	T	P	NM_172107.4(KCNQ2):c.886A>C (p.Thr296Pro)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369769	20	63439639	369769	354007	rs756921902	LOF	0.837	0.075	0.088
KCNQ2	301	G	D	NM_172107.4(KCNQ2):c.902G>A (p.Gly301Asp)	Seizures|Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Aug 1, 2017)	criteria provided, multiple submitters, no conflicts	VCV000431095	20	63439623	431095	424618	rs1131691936	LOF	0.831	0.085	0.084
KCNQ2	301	G	S	NM_172107.4(KCNQ2):c.901G>A (p.Gly301Ser)	Epileptic encephalopathy|not provided|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Oct 2, 2021)	criteria provided, conflicting interpretations	VCV000369770	20	63439624	369770	354006	rs1057516099	LOF	0.832	0.083	0.085
KCNQ2	301	G	V	NM_172107.4(KCNQ2):c.902G>T (p.Gly301Val)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Feb 21, 2020)	criteria provided, conflicting interpretations	VCV000430386	20	63439623	430386	422342	rs1131691936	LOF	0.869	0.048	0.083
KCNQ2	303	S	C	NM_172107.4(KCNQ2):c.908C>G (p.Ser303Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 28, 2020)	criteria provided, single submitter	VCV000942443	20	63439617	942443	939029	rs2081098548	LOF	0.85	0.053	0.096
KCNQ2	304	F	C	NM_172107.4(KCNQ2):c.911T>G (p.Phe304Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 26, 2020)	criteria provided, single submitter	VCV001060320	20	63439614	1060320	1051781	NA	LOF	0.841	0.053	0.106
KCNQ2	304	F	S	NM_172107.4(KCNQ2):c.911T>C (p.Phe304Ser)	not provided|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Sep 9, 2019)	criteria provided, single submitter	VCV000369771	20	63439614	369771	354005	rs1057516100	LOF	0.852	0.058	0.089
KCNQ2	305	F	L	NM_172107.4(KCNQ2):c.915C>G (p.Phe305Leu)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Mar 7, 2020)	criteria provided, single submitter	VCV000461423	20	63439610	461423	470995	rs775918190	LOF	0.826	0.041	0.133
KCNQ2	305	F	L	NM_172107.4(KCNQ2):c.915C>G (p.Phe305Leu)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Mar 7, 2020)	criteria provided, single submitter	VCV000461423	20	63439610	461423	470995	rs775918190	LOF	0.826	0.041	0.133
KCNQ2	305	F	L	NM_172107.4(KCNQ2):c.915C>A (p.Phe305Leu)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic/Likely pathogenic(Last reviewed: Mar 18, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205887	20	63439610	205887	203771	rs775918190	LOF	0.826	0.041	0.133
KCNQ2	305	F	L	NM_172107.4(KCNQ2):c.915C>A (p.Phe305Leu)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic/Likely pathogenic(Last reviewed: Mar 18, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205887	20	63439610	205887	203771	rs775918190	LOF	0.826	0.041	0.133

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	306	A	E	NM_172107.4(KCNQ2):c.917C>A (p.Ala306Glu)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Oct 5, 2020)	criteria provided, single submitter	VCV001066055	20	63439608	1066055	1056622	NA	LOF	0.817	0.087	0.096
KCNQ2	306	A	P	NM_172107.4(KCNQ2):c.916G>C (p.Ala306Pro)	not provided	Pathogenic(Last reviewed: Sep 2, 2020)	criteria provided, single submitter	VCV000205888	20	63439609	205888	203770	rs74315390	LOF	0.796	0.116	0.088
KCNQ2	306	A	T	NM_172107.4(KCNQ2):c.916G>A (p.Ala306Thr)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|Seizures|not provided	Pathogenic(Last reviewed: Oct 26, 2020)	criteria provided, multiple submitters, no conflicts	VCV000007382	20	63439609	7382	22421	rs74315390	LOF	0.813	0.09	0.097
KCNQ2	306	A	V	NM_172107.4(KCNQ2):c.917C>T (p.Ala306Val)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Apr 6, 2021)	criteria provided, multiple submitters, no conflicts	VCV000219235	20	63439608	219235	217258	rs864321707	LOF	0.84	0.063	0.098
KCNQ2	309	A	V	NM_172107.4(KCNQ2):c.926C>T (p.Ala309Val)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Mar 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000369772	20	63439599	369772	354004	NA	LOF	0.843	0.062	0.095
KCNQ2	310	G	S	NM_172107.4(KCNQ2):c.928G>A (p.Gly310Ser)	Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7	Conflicting interpretations of pathogenicity(Last reviewed: Mar 6, 2018)	criteria provided, conflicting interpretations	VCV000576538	20	63438720	576538	572967	rs1568925719	LOF	0.837	0.079	0.084
KCNQ2	311	I	N	NM_172107.4(KCNQ2):c.932T>A (p.Ile311Asn)	not provided	Likely pathogenic(Last reviewed: May 5, 2017)	criteria provided, single submitter	VCV000429385	20	63438716	429385	422341	rs1131691356	LOF	0.842	0.064	0.093
KCNQ2	312	L	F	NM_172107.4(KCNQ2):c.936G>T (p.Leu312Phe)	Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Dec 7, 2021)	criteria provided, single submitter	VCV001329944	20	63438712	1329944	1320628	NA	LOF	0.824	0.047	0.129
KCNQ2	312	L	S	NM_172107.4(KCNQ2):c.935T>C (p.Leu312Ser)	not provided	Likely pathogenic(Last reviewed: Dec 15, 2016)	criteria provided, single submitter	VCV000373802	20	63438713	373802	360476	rs1057518619	LOF	0.84	0.066	0.094
KCNQ2	313	G	W	NM_172107.4(KCNQ2):c.937G>T (p.Gly313Trp)	not provided	Uncertain significance(Last reviewed: Nov 4, 2013)	criteria provided, single submitter	VCV000205890	20	63438711	205890	203768	rs796052639	LOF	0.878	0.041	0.081
KCNQ2	314	S	P	NM_172107.4(KCNQ2):c.940T>C (p.Ser314Pro)	not provided	Likely pathogenic(Last reviewed: May 25, 2016)	criteria provided, single submitter	VCV000421323	20	63438708	421323	410826	rs1064795058	LOF	0.817	0.073	0.11
KCNQ2	315	G	R	NM_172107.4(KCNQ2):c.943G>C (p.Gly315Arg)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000219237	20	63438705	219237	217257	rs864321709	LOF	0.865	0.043	0.092
KCNQ2	317	A	T	NM_172107.4(KCNQ2):c.949G>A (p.Ala317Thr)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Sep 12, 2019)	criteria provided, single submitter	VCV000812166	20	63438699	812166	800409	rs1600751236	LOF	0.827	0.035	0.138
KCNQ2	319	K	E	NM_172107.4(KCNQ2):c.955A>G (p.Lys319Glu)	Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Jul 16, 2021)	no assertion criteria provided	VCV001077102	20	63438693	1077102	1065742	NA	LOF	0.883	0.031	0.085
KCNQ2	319	K	R	NM_172107.4(KCNQ2):c.956A>G (p.Lys319Arg)	not provided	Likely pathogenic(Last reviewed: Feb 7, 2017)	criteria provided, single submitter	VCV000393264	20	63438692	393264	377145	rs1057524860	LOF	0.885	0.031	0.085

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	324	H	Y	NM_172107.4(KCNQ2):c.970C>T (p.His324Tyr)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Aug 16, 2020)	criteria provided, single submitter	VCV001066649	20	63438678	1066649	1056621	NA	LOF	0.832	0.033	0.136
KCNQ2	325	R	G	NM_172107.4(KCNQ2):c.973A>G (p.Arg325Gly)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369775	20	63438675	369775	354001	rs1057516103	LOF	0.86	0.04	0.1
KCNQ2	327	K	N	NM_172107.4(KCNQ2):c.981G>T (p.Lys327Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 15, 2020)	criteria provided, single submitter	VCV001059815	20	63438667	1059815	1051780	NA	LOF	0.886	0.036	0.079
KCNQ2	328	H	N	NM_172107.4(KCNQ2):c.982C>A (p.His328Asn)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Aug 20, 2020)	criteria provided, conflicting interpretations	VCV000431951	20	63438666	431951	426352	rs1555869803	LOF	0.866	0.037	0.097
KCNQ2	329	F	S	NM_172107.4(KCNQ2):c.986T>C (p.Phe329Ser)	not provided	Likely pathogenic(Last reviewed: Feb 16, 2017)	criteria provided, single submitter	VCV000423623	20	63438662	423623	410825	rs1064796535	LOF	0.797	0.039	0.164
KCNQ2	331	K	T	NM_172107.4(KCNQ2):c.992A>C (p.Lys331Thr)	not provided	Likely pathogenic(Last reviewed: May 18, 2017)	criteria provided, single submitter	VCV000618186	20	63438656	618186	610161	rs1568925523	LOF	0.879	0.033	0.087
KCNQ2	332	R	G	NM_172107.4(KCNQ2):c.994A>G (p.Arg332Gly)	Seizures|Inborn genetic diseases|Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Apr 23, 2017)	criteria provided, multiple submitters, no conflicts	VCV000461424	20	63438654	461424	470488	rs1555869758	LOF	0.872	0.038	0.089
KCNQ2	332	R	K	NM_172107.4(KCNQ2):c.995G>A (p.Arg332Lys)	not provided	Uncertain significance(Last reviewed: Oct 31, 2012)	criteria provided, single submitter	VCV000205891	20	63438653	205891	203767	rs796052640	LOF	0.894	0.031	0.074
KCNQ2	333	R	Q	NM_172107.4(KCNQ2):c.998G>A (p.Arg333Gln)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Nov 29, 2021)	criteria provided, multiple submitters, no conflicts	VCV000021810	20	63438650	21810	34662	rs118192216	LOF	0.878	0.032	0.09
KCNQ2	333	R	W	NM_172107.4(KCNQ2):c.997C>T (p.Arg333Trp)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Aug 27, 2021)	criteria provided, multiple submitters, no conflicts	VCV000021809	20	63438651	21809	34661	rs118192215	LOF	0.865	0.034	0.101
KCNQ2	335	P	A	NM_172107.4(KCNQ2):c.1003C>G (p.Pro335Ala)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Nov 25, 2018)	criteria provided, single submitter	VCV000646824	20	63438645	646824	650418	rs1555869700	LOF	0.864	0.035	0.101
KCNQ2	335	P	L	NM_172107.4(KCNQ2):c.1004C>T (p.Pro335Leu)	not provided|Seizures|Epicanthus|Autistic behavior|Intellectual disability, moderate| Abnormal facial shape|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic/Likely pathogenic(Last reviewed: Jul 14, 2021)	criteria provided, multiple submitters, no conflicts	VCV000205892	20	63438644	205892	203766	rs796052641	LOF	0.848	0.034	0.118
KCNQ2	335	P	S	NM_172107.4(KCNQ2):c.1003C>T (p.Pro335Ser)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Feb 10, 2018)	criteria provided, single submitter	VCV000530528	20	63438645	530528	533690	rs1555869700	LOF	0.869	0.035	0.096
KCNQ2	336	A	V	NM_172107.4(KCNQ2):c.1007C>T (p.Ala336Val)	not provided	Likely pathogenic(Last reviewed: Mar 7, 2019)	criteria provided, single submitter	VCV001202917	20	63438641	1202917	1195548	NA	LOF	0.82	0.035	0.145
KCNQ2	337	A	G	NM_172107.4(KCNQ2):c.1010C>G (p.Ala337Gly)	Early infantile epileptic encephalopathy 7|not provided	Pathogenic(Last reviewed: Feb 6, 2014)	criteria provided, single submitter	VCV000205894	20	63438638	205894	203764	rs796052643	LOF	0.839	0.044	0.117

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	337	A	T	NM_172107.4(KCNQ2):c.1009G>A (p.Ala337Thr)	not provided	Pathogenic(Last reviewed: Aug 30, 2012)	criteria provided, single submitter	VCV000205893	20	63438639	205893	203765	rs796052642	LOF	0.821	0.035	0.144
KCNQ2	339	L	Q	NM_172107.4(KCNQ2):c.1016T>A (p.Leu339Gln)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Jan 1, 2018)	criteria provided, single submitter	VCV000495272	20	63438632	495272	486772	rs118192217	LOF	0.807	0.035	0.158
KCNQ2	339	L	R	NM_172107.4(KCNQ2):c.1016T>G (p.Leu339Arg)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021755	20	63438632	21755	34607	rs118192217	LOF	0.83	0.035	0.135
KCNQ2	340	I	M	NM_172107.4(KCNQ2):c.1020C>G (p.Ile340Met)	not provided	Likely pathogenic(Last reviewed: Jul 17, 2014)	criteria provided, single submitter	VCV000205896	20	63438628	205896	203763	rs752073942	LOF	0.73	0.036	0.233
KCNQ2	341	Q	H	NM_172107.4(KCNQ2):c.1023G>C (p.Gln341His)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Apr 15, 2020)	criteria provided, single submitter	VCV000856627	20	63438625	856627	848537	rs2081071680	LOF	0.872	0.031	0.097
KCNQ2	341	Q	R	NM_172107.4(KCNQ2):c.1022A>G (p.Gln341Arg)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Dec 19, 2018)	criteria provided, single submitter	VCV000976423	20	63438626	976423	964539	rs2081071796	LOF	0.886	0.031	0.083
KCNQ2	342	S	L	NM_172107.4(KCNQ2):c.1025C>T (p.Ser342Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 28, 2020)	criteria provided, single submitter	VCV001044294	20	63433902	1044294	1034795	rs2080907267	LOF	0.799	0.035	0.165
KCNQ2	344	W	R	NM_172107.4(KCNQ2):c.1030T>C (p.Trp344Arg)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369777	20	63433897	369777	353999	rs1057516105	LOF	0.826	0.036	0.139
KCNQ2	348	A	T	NM_172107.4(KCNQ2):c.1042G>A (p.Ala348Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 5, 2019)	criteria provided, single submitter	VCV000950074	20	63433885	950074	929245	rs2080906700	LOF	0.837	0.036	0.127
KCNQ2	348	A	V	NM_172107.4(KCNQ2):c.1043C>T (p.Ala348Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 11, 2019)	criteria provided, single submitter	VCV000567177	20	63433884	567177	572963	rs1568916986	LOF	0.804	0.036	0.16
KCNQ2	349	T	A	NM_172107.4(KCNQ2):c.1045A>G (p.Thr349Ala)	not provided	Uncertain significance(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001335466	20	63433882	1335466	1326437	NA	LOF	0.842	0.041	0.118
KCNQ2	349	T	I	NM_172107.4(KCNQ2):c.1046C>T (p.Thr349Ile)	Benign familial neonatal seizures 1|Epileptic encephalopathy, early infantile, 1|Early infantile epileptic encephalopathy 7	Conflicting interpretations of pathogenicity(Last reviewed: Jan 3, 2022)	criteria provided, conflicting interpretations	VCV000975417	20	63433881	975417	963916	rs2080906453	LOF	0.81	0.041	0.149
KCNQ2	349	T	P	NM_172107.4(KCNQ2):c.1045A>C (p.Thr349Pro)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001326317	20	63433882	1326317	1316719	NA	LOF	0.86	0.046	0.094
KCNQ2	350	N	I	NM_172107.4(KCNQ2):c.1049A>T (p.Asn350Ile)	not provided	Likely pathogenic(Last reviewed: Mar 5, 2014)	criteria provided, single submitter	VCV000205897	20	63433878	205897	203761	rs772800738	LOF	0.823	0.039	0.138
KCNQ2	350	N	K	NM_172107.4(KCNQ2):c.1050C>A (p.Asn350Lys)	Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000803621	20	63433877	803621	791998	rs1371059392	LOF	0.875	0.036	0.088
KCNQ2	350	N	S	NM_172107.4(KCNQ2):c.1049A>G (p.Asn350Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 8, 2020)	criteria provided, single submitter	VCV001053550	20	63433878	1053550	1051779	NA	LOF	0.857	0.038	0.104

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	351	L	F	NM_172107.4(KCNQ2):c.1051C>T (p.Leu351Phe)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369779	20	63433876	369779	353997	rs1057516106	GOF	0.514	0.047	0.439
KCNQ2	351	L	V	NM_172107.4(KCNQ2):c.1051C>G (p.Leu351Val)	Benign familial neonatal seizures 1|Epilepsy, benign neonatal, 1, and/or myokymia	Likely pathogenic(Last reviewed: Sep 28, 2018)	criteria provided, single submitter	VCV000369778	20	63433876	369778	353998	rs1057516106	GOF	0.475	0.048	0.476
KCNQ2	352	S	P	NM_172107.4(KCNQ2):c.1054T>C (p.Ser352Pro)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369781	20	63433873	369781	353995	rs1057516108	LOF	0.833	0.045	0.122
KCNQ2	353	R	C	NM_172107.4(KCNQ2):c.1057C>T (p.Arg353Cys)	Early infantile epileptic encephalopathy with suppression bursts|not specified|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Dec 23, 2020)	criteria provided, conflicting interpretations	VCV000205898	20	63433870	205898	203760	rs118192218	LOF	0.815	0.047	0.139
KCNQ2	353	R	G	NM_172107.4(KCNQ2):c.1056_1057delinsCG (p.Arg353Gly)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Oct 19, 2020)	criteria provided, single submitter	VCV001074881	20	63433870 - 63433871	1074881	1064856	NA	LOF	0.864	0.046	0.09
KCNQ2	353	R	G	NM_172107.4(KCNQ2):c.1056_1057delinsCG (p.Arg353Gly)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Oct 19, 2020)	criteria provided, single submitter	VCV001074881	20	63433870 - 63433871	1074881	1064856	NA	LOF	0.864	0.046	0.09
KCNQ2	353	R	G	NM_172107.4(KCNQ2):c.1057C>G (p.Arg353Gly)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021756	20	63433870	21756	34608	rs118192218	LOF	0.864	0.046	0.09
KCNQ2	353	R	G	NM_172107.4(KCNQ2):c.1057C>G (p.Arg353Gly)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021756	20	63433870	21756	34608	rs118192218	LOF	0.864	0.046	0.09
KCNQ2	353	R	H	NM_172107.4(KCNQ2):c.1058G>A (p.Arg353His)	Early infantile epileptic encephalopathy 7|not provided|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic/Likely pathogenic(Last reviewed: Oct 6, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205899	20	63433869	205899	203759	rs796052645	LOF	0.875	0.037	0.088
KCNQ2	355	D	E	NM_172107.4(KCNQ2):c.1065C>A (p.Asp355Glu)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Jan 8, 2020)	criteria provided, single submitter	VCV000963965	20	63433862	963965	951147	rs200395340	LOF	0.879	0.041	0.079
KCNQ2	355	D	E	NM_172107.4(KCNQ2):c.1065C>A (p.Asp355Glu)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Jan 8, 2020)	criteria provided, single submitter	VCV000963965	20	63433862	963965	951147	rs200395340	LOF	0.879	0.041	0.079
KCNQ2	355	D	E	NM_172107.4(KCNQ2):c.1065C>G (p.Asp355Glu)	not provided	Uncertain significance(Last reviewed: Mar 30, 2017)	criteria provided, single submitter	VCV000445403	20	63433862	445403	438675	rs200395340	LOF	0.879	0.041	0.079
KCNQ2	355	D	E	NM_172107.4(KCNQ2):c.1065C>G (p.Asp355Glu)	not provided	Uncertain significance(Last reviewed: Mar 30, 2017)	criteria provided, single submitter	VCV000445403	20	63433862	445403	438675	rs200395340	LOF	0.879	0.041	0.079
KCNQ2	355	D	G	NM_172107.4(KCNQ2):c.1064A>G (p.Asp355Gly)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Jun 26, 2020)	criteria provided, single submitter	VCV001074630	20	63433863	1074630	1064855	NA	LOF	0.864	0.047	0.09
KCNQ2	355	D	V	NM_172107.4(KCNQ2):c.1064A>T (p.Asp355Val)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 26, 2018)	criteria provided, single submitter	VCV000976159	20	63433863	976159	964538	rs2080905274	LOF	0.838	0.039	0.123
KCNQ2	355	D	Y	NM_172107.4(KCNQ2):c.1063G>T (p.Asp355Tyr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 9, 2020)	criteria provided, single submitter	VCV001003511	20	63433864	1003511	999086	rs2080905348	LOF	0.858	0.038	0.104
KCNQ2	356	L	P	NM_172107.4(KCNQ2):c.1067T>C (p.Leu356Pro)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of	criteria provided,	VCV000422045	20	63433860	422045	410824	rs1057518772	LOF	0.807	0.039	0.154

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						pathogenicity(Last reviewed: Dec 5, 2018)	conflicting interpretations										
KCNQ2	356	L	R	NM_172107.4(KCNQ2):c.1067T>G (p.Leu356Arg)	Intellectual disability|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Jan 1, 2016)	criteria provided, single submitter	VCV000373925	20	63433860	373925	361066	rs1057518772	LOF	0.821	0.034	0.144
KCNQ2	356	L	V	NM_172107.4(KCNQ2):c.1066C>G (p.Leu356Val)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369782	20	63433861	369782	353994	rs1057516109	GOF	0.669	0.038	0.293
KCNQ2	358	S	F	NM_172107.4(KCNQ2):c.1073C>T (p.Ser358Phe)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369783	20	63433854	369783	353993	rs1057516110	LOF	0.799	0.035	0.166
KCNQ2	358	S	P	NM_172107.4(KCNQ2):c.1072T>C (p.Ser358Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 26, 2019)	criteria provided, single submitter	VCV000844119	20	63433855	844119	848536	rs2080904614	LOF	0.858	0.038	0.104
KCNQ2	359	T	A	NM_172107.4(KCNQ2):c.1075A>G (p.Thr359Ala)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jun 25, 2019)	criteria provided, conflicting interpretations	VCV000418633	20	63433852	418633	410823	rs1064793334	LOF	0.821	0.033	0.146
KCNQ2	359	T	K	NM_172107.4(KCNQ2):c.1076C>A (p.Thr359Lys)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Sep 24, 2018)	criteria provided, single submitter	VCV000021757	20	63433851	21757	34609	rs118192219	LOF	0.869	0.031	0.1
KCNQ2	359	T	M	NM_172107.4(KCNQ2):c.1076C>T (p.Thr359Met)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Benign familial neonatal seizures 1	Conflicting interpretations of pathogenicity(Last reviewed: Aug 19, 2021)	criteria provided, conflicting interpretations	VCV000976330	20	63433851	976330	964537	rs118192219	LOF	0.789	0.032	0.18
KCNQ2	360	W	C	NM_172107.4(KCNQ2):c.1080G>T (p.Trp360Cys)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Jan 18, 2018)	criteria provided, single submitter	VCV000637022	20	63433847	637022	624870	rs1600732174	GOF	0.642	0.05	0.308
KCNQ2	360	W	R	NM_172107.4(KCNQ2):c.1078T>C (p.Trp360Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 21, 2019)	criteria provided, single submitter	VCV000848129	20	63433849	848129	848535	rs2080904141	LOF	0.825	0.037	0.138
KCNQ2	362	Y	C	NM_172107.4(KCNQ2):c.1085A>G (p.Tyr362Cys)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369784	20	63433842	369784	353992	rs1057516111	GOF	0.475	0.049	0.476
KCNQ2	363	Y	C	NM_172107.4(KCNQ2):c.1088A>G (p.Tyr363Cys)	not provided	Uncertain significance(Last reviewed: Jun 1, 2016)	criteria provided, multiple submitters, no conflicts	VCV000198922	20	63433839	198922	196082	rs794727935	GOF	0.551	0.045	0.404
KCNQ2	367	V	I	NM_172107.4(KCNQ2):c.1099G>A (p.Val367Ile)	not provided	Uncertain significance(Last reviewed: Apr 15, 2020)	criteria provided, single submitter	VCV001202324	20	63433828	1202324	1192273	NA	GOF	0.613	0.043	0.344
KCNQ2	368	T	N	NM_172107.4(KCNQ2):c.1103C>A (p.Thr368Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 17, 2019)	criteria provided, single submitter	VCV000946732	20	63433824	946732	929244	rs900170305	LOF	0.81	0.041	0.149
KCNQ2	368	T	S	NM_172107.4(KCNQ2):c.1103C>G (p.Thr368Ser)	not provided	Uncertain significance(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV001311180	20	63433824	1311180	1300561	NA	LOF	0.775	0.041	0.184
KCNQ2	370	P	R	NM_172107.4(KCNQ2):c.1109C>G (p.Pro370Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 19,	criteria provided, single submitter	VCV000405211	20	63433818	405211	403698	rs765394232	LOF	0.837	0.042	0.121

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2018)											
KCNQ2	370	P	T	NM_172107.4(KCNQ2):c.1108C>A (p.Pro370Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 17, 2020)	criteria provided, single submitter	VCV001024136	20	63433819	1024136	1014209	rs2080902130	LOF	0.789	0.041	0.17
KCNQ2	371	M	L	NM_172107.4(KCNQ2):c.1111A>C (p.Met371Leu)	not provided	Uncertain significance(Last reviewed: Nov 1, 2019)	criteria provided, single submitter	VCV001309884	20	63433816	1309884	1299259	NA	GOF	0.624	0.045	0.331
KCNQ2	373	R	I	NM_172107.4(KCNQ2):c.1118G>T (p.Ser373Ile)	Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: Jan 3, 2022)	criteria provided, single submitter	VCV001333324	20	63433809	1333324	1324099	NA	LOF	0.745	0.051	0.204
KCNQ2	374	S	L	NM_172107.4(KCNQ2):c.1121C>T (p.Ser374Leu)	Seizures|Early infantile epileptic encephalopathy with suppression bursts|not specified	Conflicting interpretations of pathogenicity(Last reviewed: Sep 16, 2020)	criteria provided, conflicting interpretations	VCV000513677	20	63431367	513677	507877	rs777940990	LOF	0.751	0.06	0.188
KCNQ2	374	Y	C	NM_172107.4(KCNQ2):c.1118+3A>G	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369785	20	63433806	369785	353991	rs1057516112	GOF	0.686	0.066	0.249
KCNQ2	374	Y	D	NM_172107.4(KCNQ2):c.1118+2T>G	Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic(Last reviewed: Feb 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV001073519	20	63433807	1073519	1064852	NA	LOF	0.787	0.064	0.149
KCNQ2	374	Y	H	NM_172107.4(KCNQ2):c.1118+2T>C	Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic/Likely pathogenic(Last reviewed: Mar 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000283350	20	63433807	283350	267587	rs886042605	LOF	0.756	0.058	0.186
KCNQ2	375	R	C	NM_172107.4(KCNQ2):c.1118+5C>T	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 25, 2019)	criteria provided, single submitter	VCV000840062	20	63433804	840062	852381	rs903746022	LOF	0.76	0.062	0.177
KCNQ2	376	R	C	NM_172107.4(KCNQ2):c.1118+8C>T	not specified	Likely benign(Last reviewed: Mar 16, 2017)	criteria provided, single submitter	VCV000516129	20	63433801	516129	507301	rs754114486	LOF	0.75	0.07	0.179
KCNQ2	377	R	W	NM_172107.4(KCNQ2):c.1118+11C>T	not specified	Likely benign(Last reviewed: Jan 4, 2018)	criteria provided, single submitter	VCV000514453	20	63433798	514453	508271	rs138176417	LOF	0.799	0.055	0.146
KCNQ2	378	T	S	NM_172107.4(KCNQ2):c.1133C>G (p.Thr378Ser)	not provided	Uncertain significance(Last reviewed: Feb 20, 2020)	criteria provided, single submitter	VCV001304076	20	63431355	1304076	1294351	NA	LOF	0.752	0.054	0.194
KCNQ2	379	P	L	NM_172107.4(KCNQ2):c.1118+18C>T	Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: Jan 17, 2020)	criteria provided, single submitter	VCV001029262	20	63433791	1029262	1018720	rs2080900462	GOF	0.705	0.061	0.234
KCNQ2	379	Y	F	NM_172107.4(KCNQ2):c.1136A>T (p.Tyr379Phe)	not provided	Uncertain significance(Last reviewed: May 26, 2017)	criteria provided, single submitter	VCV000430416	20	63431352	430416	422340	rs752773828	GOF	0.628	0.056	0.317
KCNQ2	380	G	R	NM_172107.4(KCNQ2):c.1138G>A (p.Gly380Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 4, 2020)	criteria provided, single submitter	VCV001016895	20	63431350	1016895	1014208	rs764614789	LOF	0.796	0.069	0.135
KCNQ2	381	A	D	NM_172107.4(KCNQ2):c.1142C>A (p.Ala381Asp)	not provided	Uncertain significance(Last reviewed: Oct 14, 2019)	criteria provided, single submitter	VCV001308969	20	63431346	1308969	1300997	NA	LOF	0.763	0.058	0.179

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	385	I	N	NM_172107.4(KCNQ2):c.1154T>A (p.Ile385Asn)	not provided	Uncertain significance(Last reviewed: Aug 1, 2016)	criteria provided, single submitter	VCV000374487	20	63428430	374487	361373	rs1057519123	GOF	0.709	0.054	0.237
KCNQ2	391	I	V	NM_172107.4(KCNQ2):c.1118+53A>G	not specified	Likely benign(Last reviewed: Dec 27, 2012)	criteria provided, single submitter	VCV000205834	20	63433756	205834	203756	rs780492963	GOF	0.527	0.054	0.42
KCNQ2	391	L	P	NM_172107.4(KCNQ2):c.1172T>C (p.Leu391Pro)	not provided	Uncertain significance(Last reviewed: Mar 8, 2012)	criteria provided, single submitter	VCV000205900	20	63428412	205900	203752	rs796052646	GOF	0.688	0.056	0.256
KCNQ2	393	L	V	NM_172107.4(KCNQ2):c.1207C>G (p.Leu403Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 10, 2020)	criteria provided, single submitter	VCV000998578	20	63428377	998578	999085	rs1166282198	GOF	0.53	0.053	0.417
KCNQ2	394	L	P	NM_172107.4(KCNQ2):c.1181T>C (p.Leu394Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 27, 2019)	criteria provided, single submitter	VCV000842767	20	63428403	842767	848534	rs2080695319	LOF	0.738	0.067	0.195
KCNQ2	398	D	H	NM_172107.4(KCNQ2):c.1222G>C (p.Asp408His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 17, 2019)	criteria provided, single submitter	VCV000951470	20	63424202	951470	929243	rs2080560881	LOF	0.822	0.06	0.117
KCNQ2	400	K	E	NM_172107.4(KCNQ2):c.1198A>G (p.Lys400Glu)	not provided	Uncertain significance(Last reviewed: Jul 23, 2018)	criteria provided, single submitter	VCV000205901	20	63428386	205901	203751	rs796052647	LOF	0.834	0.062	0.104
KCNQ2	401	P	Q	NM_172107.4(KCNQ2):c.1232C>A (p.Pro411Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 10, 2020)	criteria provided, single submitter	VCV001025896	20	63424192	1025896	1014207	rs754682495	LOF	0.811	0.063	0.125
KCNQ2	401	P	R	NM_172107.4(KCNQ2):c.1232C>G (p.Pro411Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 7, 2020)	criteria provided, single submitter	VCV001057136	20	63424192	1057136	1051778	NA	LOF	0.814	0.068	0.118
KCNQ2	404	A	D	NM_172107.4(KCNQ2):c.1211C>A (p.Ala404Asp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 25, 2016)	criteria provided, single submitter	VCV000405207	20	63428373	405207	403693	rs1060500601	LOF	0.783	0.068	0.149
KCNQ2	406	R	G	NM_172107.4(KCNQ2):c.1216A>G (p.Arg406Gly)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Feb 19, 2018)	criteria provided, multiple submitters, no conflicts	VCV000452880	20	63428368	452880	446288	rs1240338507	LOF	0.831	0.061	0.108
KCNQ2	410	P	L	NM_172107.4(KCNQ2):c.1229C>T (p.Pro410Leu)	not provided|Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 1	Uncertain significance(Last reviewed: Oct 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000461403	20	63424195	461403	470987	rs752579642	LOF	0.791	0.077	0.131
KCNQ2	411	P	L	NM_172107.4(KCNQ2):c.1232C>T (p.Pro411Leu)	Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Jul 3, 2017)	criteria provided, single submitter	VCV000461404	20	63424192	461404	469445	rs754682495	LOF	0.785	0.081	0.134
KCNQ2	413	P	R	NM_172107.4(KCNQ2):c.1238C>G (p.Pro413Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 31, 2017)	criteria provided, single submitter	VCV000461405	20	63424186	461405	470986	rs1270343938	LOF	0.814	0.079	0.108
KCNQ2	414	S	C	NM_172107.4(KCNQ2):c.1241C>G (p.Ser414Cys)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 27, 2019)	criteria provided, multiple submitters, no conflicts	VCV000449179	20	63424183	449179	446286	rs1555859111	LOF	0.804	0.066	0.13

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	417	K	E	NM_172107.4(KCNQ2):c.1249A>G (p.Lys417Glu)	not provided	Uncertain significance(Last reviewed: Jun 20, 2019)	criteria provided, single submitter	VCV001306474	20	63419671	1306474	1296417	NA	LOF	0.801	0.092	0.107
KCNQ2	417	S	P	NM_172107.4(KCNQ2):c.1333T>C (p.Ser445Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 29, 2020)	criteria provided, single submitter	VCV001009330	20	63415095	1009330	999084	rs2080246296	LOF	0.789	0.096	0.115
KCNQ2	418	G	V	NM_172107.4(KCNQ2):c.1253G>T (p.Gly418Val)	not specified|Early infantile epileptic encephalopathy with suppression bursts	Likely benign(Last reviewed: Oct 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205902	20	63419667	205902	203750	rs201750561	LOF	0.763	0.09	0.147
KCNQ2	420	P	L	NM_172107.4(KCNQ2):c.1259C>T (p.Pro420Leu)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: Mar 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000287222	20	63419661	287222	271459	rs139164500	LOF	0.756	0.075	0.169
KCNQ2	420	P	Q	NM_172107.4(KCNQ2):c.1259C>A (p.Pro420Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV000855526	20	63419661	855526	848532	rs139164500	LOF	0.785	0.075	0.14
KCNQ2	420	P	R	NM_172107.4(KCNQ2):c.1259C>G (p.Pro420Arg)	not provided	Uncertain significance(Last reviewed: Jul 23, 2014)	criteria provided, single submitter	VCV000205903	20	63419661	205903	203749	rs139164500	LOF	0.789	0.08	0.132
KCNQ2	420	P	S	NM_172107.4(KCNQ2):c.1258C>T (p.Pro420Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 5, 2019)	criteria provided, single submitter	VCV000837816	20	63419662	837816	848533	rs2080406431	LOF	0.775	0.076	0.148
KCNQ2	421	A	V	NM_172107.4(KCNQ2):c.1352C>T (p.Ala451Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 13, 2020)	criteria provided, single submitter	VCV000999695	20	63415076	999695	999083	rs774027338	GOF	0.656	0.059	0.285
KCNQ2	422	A	T	NM_172107.4(KCNQ2):c.1354G>A (p.Ala452Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 8, 2020)	criteria provided, single submitter	VCV001011047	20	63415074	1011047	999082	rs2080245324	LOF	0.752	0.072	0.176
KCNQ2	424	P	S	NM_172107.4(KCNQ2):c.1270C>T (p.Pro424Ser)	Seizures|not provided|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Oct 16, 2020)	criteria provided, conflicting interpretations	VCV000405210	20	63419650	405210	404201	rs748120886	LOF	0.771	0.083	0.146
KCNQ2	427	G	E	NM_172107.4(KCNQ2):c.1280G>A (p.Gly427Glu)	Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: Aug 25, 2019)	no assertion criteria provided	VCV000800807	20	63419640	800807	788941	rs1601590796	LOF	0.79	0.081	0.129
KCNQ2	428	P	L	NM_172107.4(KCNQ2):c.1373C>T (p.Pro458Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 4, 2020)	criteria provided, single submitter	VCV001002529	20	63415055	1002529	999081	rs1206195455	LOF	0.734	0.075	0.191
KCNQ2	429	C	F	NM_172107.4(KCNQ2):c.1286G>T (p.Cys429Phe)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 5, 2019)	criteria provided, single submitter	VCV000665620	20	63419634	665620	650426	rs1601590761	LOF	0.703	0.068	0.23
KCNQ2	429	C	Y	NM_172107.4(KCNQ2):c.1286G>A (p.Cys429Tyr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 8, 2019)	criteria provided, single submitter	VCV000844133	20	63419634	844133	848531	rs1601590761	LOF	0.719	0.067	0.214
KCNQ2	430	P	S	NM_172107.4(KCNQ2):c.1288C>T (p.Pro430Ser)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021761	20	63419632	21761	34613	rs118192224	LOF	0.781	0.084	0.135

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	432	A	T	NM_172107.4(KCNQ2):c.1378G>A (p.Ala460Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 3, 2020)	criteria provided, single submitter	VCV000937790	20	63415050	937790	939028	rs1284566908	LOF	0.767	0.077	0.156
KCNQ2	432	R	C	NM_172107.4(KCNQ2):c.1294C>T (p.Arg432Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 24, 2020)	criteria provided, single submitter	VCV000575399	20	63419626	575399	572959	rs368720575	LOF	0.776	0.085	0.138
KCNQ2	432	R	H	NM_172107.4(KCNQ2):c.1295G>A (p.Arg432His)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Sep 24, 2019)	criteria provided, conflicting interpretations	VCV000424483	20	63419625	424483	410820	rs758074713	LOF	0.802	0.078	0.119
KCNQ2	434	S	N	NM_172107.4(KCNQ2):c.1301G>A (p.Ser434Asn)	not provided	Uncertain significance(Last reviewed: Jan 23, 2020)	criteria provided, single submitter	VCV001312018	20	63419619	1312018	1302549	NA	LOF	0.859	0.05	0.092
KCNQ2	435	R	Q	NM_172107.4(KCNQ2):c.1394G>A (p.Arg465Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 22, 2019)	criteria provided, single submitter	VCV000861986	20	63415034	861986	848529	rs1459078042	LOF	0.865	0.044	0.09
KCNQ2	440	D	H	NM_172107.4(KCNQ2):c.1408G>C (p.Asp470His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 8, 2019)	criteria provided, single submitter	VCV000936153	20	63415020	936153	939027	rs751907059	LOF	0.834	0.065	0.101
KCNQ2	440	D	N	NM_172107.4(KCNQ2):c.1408G>A (p.Asp470Asn)	not provided	Uncertain significance(Last reviewed: Aug 22, 2019)	criteria provided, single submitter	VCV001212149	20	63415020	1212149	1202144	NA	LOF	0.833	0.07	0.097
KCNQ2	442	R	C	NM_172107.4(KCNQ2):c.1324C>T (p.Arg442Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 30, 2017)	criteria provided, single submitter	VCV000461407	20	63415104	461407	470485	rs1354411692	LOF	0.826	0.055	0.119
KCNQ2	443	Q	H	NM_172107.4(KCNQ2):c.1383G>T (p.Gln461His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 25, 2020)	criteria provided, single submitter	VCV000842741	20	63415045	842741	848530	rs1349137821	LOF	0.853	0.046	0.101
KCNQ2	450	V	M	NM_172107.4(KCNQ2):c.1348G>A (p.Val450Met)	not provided|Seizures|Early infantile epileptic encephalopathy with suppression bursts|not specified	Benign/Likely benign(Last reviewed: Oct 11, 2021)	criteria provided, multiple submitters, no conflicts	VCV000129332	20	63415080	129332	134778	rs146492238	GOF	0.698	0.056	0.246
KCNQ2	459	R	G	NM_172107.4(KCNQ2):c.1465C>G (p.Arg489Gly)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV001020383	20	63414963	1020383	1014206	rs775387876	LOF	0.816	0.076	0.107
KCNQ2	461	Q	P	NM_172107.4(KCNQ2):c.1382A>C (p.Gln461Pro)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369794	20	63415046	369794	353986	rs1057516116	LOF	0.799	0.074	0.127
KCNQ2	465	A	P	NM_172107.4(KCNQ2):c.1483G>C (p.Ala495Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 22, 2020)	criteria provided, single submitter	VCV001015967	20	63414945	1015967	1014205	rs200251507	LOF	0.798	0.07	0.133
KCNQ2	469	A	T	NM_172107.4(KCNQ2):c.1405G>A (p.Ala469Thr)	not provided	Conflicting interpretations of pathogenicity(Last reviewed: Mar 1, 2020)	criteria provided, conflicting interpretations	VCV000205905	20	63415023	205905	203737	rs781778855	LOF	0.756	0.06	0.185
KCNQ2	469	R	C	NM_172107.4(KCNQ2):c.1489C>T (p.Arg497Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 15, 2019)	criteria provided, single submitter	VCV000840173	20	63414939	840173	848527	rs540461827	LOF	0.772	0.069	0.159

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	470	I	V	NM_172107.4(KCNQ2):c.1492A>G (p.Ile498Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 24, 2018)	criteria provided, single submitter	VCV000644498	20	63414936	644498	650430	rs1601572937	LOF	0.745	0.063	0.192
KCNQ2	471	A	V	NM_172107.4(KCNQ2):c.1502_1503delinsTG (p.Ala501Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 22, 2019)	criteria provided, single submitter	VCV000851545	20	63414925 - 63414926	851545	848526	rs2080237716	LOF	0.727	0.061	0.212
KCNQ2	471	A	V	NM_172107.4(KCNQ2):c.1502_1503delinsTG (p.Ala501Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 22, 2019)	criteria provided, single submitter	VCV000851545	20	63414925 - 63414926	851545	848526	rs2080237716	LOF	0.727	0.061	0.212
KCNQ2	471	A	V	NM_172107.4(KCNQ2):c.1502C>T (p.Ala501Val)	not provided	Uncertain significance	no assertion criteria provided	VCV001049677	20	63414926	1049677	1038782	NA	LOF	0.727	0.061	0.212
KCNQ2	471	A	V	NM_172107.4(KCNQ2):c.1502C>T (p.Ala501Val)	not provided	Uncertain significance	no assertion criteria provided	VCV001049677	20	63414926	1049677	1038782	NA	LOF	0.727	0.061	0.212
KCNQ2	472	A	T	NM_172107.4(KCNQ2):c.1504G>A (p.Ala502Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 17, 2019)	criteria provided, single submitter	VCV000855445	20	63414924	855445	848525	rs540103798	LOF	0.781	0.058	0.16
KCNQ2	473	R	W	NM_172107.4(KCNQ2):c.1471C>T (p.Arg491Trp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 19, 2019)	criteria provided, single submitter	VCV000847091	20	63414957	847091	848528	rs1251584590	LOF	0.801	0.064	0.135
KCNQ2	493	R	C	NM_172107.4(KCNQ2):c.1477C>T (p.Arg493Cys)	not provided	Uncertain significance(Last reviewed: Mar 20, 2017)	criteria provided, single submitter	VCV000424395	20	63414951	424395	410819	rs759338332	LOF	0.83	0.047	0.123
KCNQ2	497	R	H	NM_172107.4(KCNQ2):c.1490G>A (p.Arg497His)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 23, 2019)	criteria provided, multiple submitters, no conflicts	VCV000530434	20	63414938	530434	533616	rs769414365	LOF	0.867	0.042	0.091
KCNQ2	501	A	P	NM_172107.4(KCNQ2):c.1501G>C (p.Ala501Pro)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369797	20	63414927	369797	353984	rs1057516118	LOF	0.781	0.056	0.163
KCNQ2	502	A	S	NM_172107.4(KCNQ2):c.1504G>T (p.Ala502Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 19, 2017)	criteria provided, single submitter	VCV000530467	20	63414924	530467	533677	rs540103798	LOF	0.763	0.047	0.189
KCNQ2	502	A	V	NM_172107.4(KCNQ2):c.1505C>T (p.Ala502Val)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Conflicting interpretations of pathogenicity(Last reviewed: Aug 31, 2020)	criteria provided, conflicting interpretations	VCV000205906	20	63414923	205906	203736	rs375264483	GOF	0.684	0.047	0.269
KCNQ2	507	V	I	NM_172107.4(KCNQ2):c.1612G>A (p.Val538Ile)	not provided	Uncertain significance(Last reviewed: Oct 2, 2019)	criteria provided, single submitter	VCV001218158	20	63414107	1218158	1208140	NA	LOF	0.726	0.055	0.219
KCNQ2	509	I	V	NM_172107.4(KCNQ2):c.1618A>G (p.Ile540Val)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV001067283	20	63414101	1067283	1056619	NA	LOF	0.751	0.057	0.192
KCNQ2	512	I	S	NM_172107.4(KCNQ2):c.1619T>G (p.Ile540Ser)	Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Jul 5, 2019)	criteria provided, single submitter	VCV000947411	20	63414100	947411	929237	rs2080210194	LOF	0.713	0.063	0.224
KCNQ2	513	C	R	NM_172107.4(KCNQ2):c.1630T>C (p.Cys544Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV001036550	20	63414089	1036550	1034794	rs118192232	LOF	0.81	0.073	0.117

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	514	G	E	NM_172107.4(KCNQ2):c.1541G>A (p.Gly514Glu)	not provided	Uncertain significance(Last reviewed: Aug 1, 2017)	criteria provided, single submitter	VCV000450825	20	63414178	450825	446284	rs1358495877	LOF	0.822	0.073	0.104
KCNQ2	514	G	R	NM_172107.4(KCNQ2):c.1540G>A (p.Gly514Arg)	not provided	Uncertain significance(Last reviewed: Nov 15, 2018)	criteria provided, single submitter	VCV000205948	20	63414179	205948	203735	rs772669887	LOF	0.817	0.074	0.108
KCNQ2	515	E	D	NM_172107.4(KCNQ2):c.1545G>T (p.Glu515Asp)	not specified|Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Feb 6, 2020)	criteria provided, conflicting interpretations	VCV000445484	20	63414174	445484	438756	rs117067974	LOF	0.85	0.062	0.088
KCNQ2	515	E	D	NM_172107.4(KCNQ2):c.1545G>T (p.Glu515Asp)	not specified|Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Feb 6, 2020)	criteria provided, conflicting interpretations	VCV000445484	20	63414174	445484	438756	rs117067974	LOF	0.85	0.062	0.088
KCNQ2	515	E	D	NM_172107.4(KCNQ2):c.1545G>C (p.Glu515Asp)	Early infantile epileptic encephalopathy with suppression bursts|Seizures|not specified|not provided|Benign familial neonatal seizures 1	Conflicting interpretations of pathogenicity(Last reviewed: Aug 30, 2021)	criteria provided, conflicting interpretations	VCV000021764	20	63414174	21764	34616	rs117067974	LOF	0.85	0.062	0.088
KCNQ2	515	E	D	NM_172107.4(KCNQ2):c.1545G>C (p.Glu515Asp)	Early infantile epileptic encephalopathy with suppression bursts|Seizures|not specified|not provided|Benign familial neonatal seizures 1	Conflicting interpretations of pathogenicity(Last reviewed: Aug 30, 2021)	criteria provided, conflicting interpretations	VCV000021764	20	63414174	21764	34616	rs117067974	LOF	0.85	0.062	0.088
KCNQ2	518	M	I	NM_172107.4(KCNQ2):c.1638G>A (p.Met546Ile)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 15, 2019)	criteria provided, single submitter	VCV000934692	20	63413575	934692	939025	rs2080191127	GOF	0.66	0.052	0.288
KCNQ2	520	S	P	NM_172107.4(KCNQ2):c.1651T>C (p.Ser551Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 25, 2020)	criteria provided, single submitter	VCV001034586	20	63413562	1034586	1034793	rs2080190604	LOF	0.855	0.052	0.093
KCNQ2	521	K	R	NM_172107.4(KCNQ2):c.1655A>G (p.Lys552Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 6, 2018)	criteria provided, single submitter	VCV000642572	20	63413558	642572	650433	rs1555853613	LOF	0.864	0.053	0.084
KCNQ2	524	F	I	NM_172107.4(KCNQ2):c.1663T>A (p.Phe555Ile)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001326319	20	63413550	1326319	1316721	NA	GOF	0.623	0.047	0.33
KCNQ2	525	C	S	NM_172107.4(KCNQ2):c.1573T>A (p.Cys525Ser)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Dec 3, 2018)	criteria provided, multiple submitters, no conflicts	VCV000205907	20	63414146	205907	203734	rs371162714	LOF	0.77	0.05	0.18
KCNQ2	526	K	N	NM_172107.4(KCNQ2):c.1662G>T (p.Lys554Asn)	Early infantile epileptic encephalopathy 7| Benign familial neonatal seizures 2|Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000007388	20	63413551	7388	22427	rs267607198	LOF	0.861	0.039	0.099
KCNQ2	530	E	K	NM_172107.4(KCNQ2):c.1588G>A (p.Glu530Lys)	Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7	Conflicting interpretations of pathogenicity(Last reviewed: Aug 20, 2020)	criteria provided, conflicting interpretations	VCV000495266	20	63414131	495266	486771	rs897976020	LOF	0.816	0.095	0.089
KCNQ2	530	P	S	NM_172107.4(KCNQ2):c.1681C>T (p.Pro561Ser)	KCNQ2-Related Disorders	Pathogenic(Last reviewed: May 14, 2020)	criteria provided, single submitter	VCV001301899	20	63413532	1301899	1292181	NA	LOF	0.813	0.094	0.094
KCNQ2	530	S	N	NM_172107.4(KCNQ2):c.1673G>A (p.Ser558Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last	criteria provided, single	VCV000660856	20	63413540	660856	650434	rs1601566660	LOF	0.819	0.092	0.089

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Sep 13, 2018)	submitter										
KCNQ2	531	D	V	NM_172107.4(KCNQ2):c.1592A>T (p.Asp531Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 10, 2017)	criteria provided, single submitter	VCV000530537	20	63414127	530537	534194	rs1555854018	LOF	0.81	0.093	0.097
KCNQ2	532	D	G	NM_172107.4(KCNQ2):c.1688A>G (p.Asp563Gly)	Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Dec 10, 2019)	criteria provided, single submitter	VCV000836158	20	63413525	836158	848523	rs2080189052	LOF	0.866	0.052	0.082
KCNQ2	533	P	R	NM_172107.4(KCNQ2):c.1682C>G (p.Pro561Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 28, 2019)	criteria provided, single submitter	VCV000943803	20	63413531	943803	939024	rs796052652	LOF	0.86	0.043	0.097
KCNQ2	534	M	I	NM_172107.4(KCNQ2):c.1695G>A (p.Met565Ile)	not provided	Uncertain significance(Last reviewed: Dec 1, 2020)	criteria provided, single submitter	VCV001013460	20	63413518	1013460	1001175	rs2080188612	LOF	0.8	0.04	0.16
KCNQ2	536	V	I	NM_172107.4(KCNQ2):c.1699G>A (p.Val567Ile)	not provided	Uncertain significance(Last reviewed: Mar 1, 2021)	criteria provided, single submitter	VCV001175997	20	63413514	1175997	1166353	NA	GOF	0.716	0.038	0.246
KCNQ2	541	R	G	NM_172107.4(KCNQ2):c.1621A>G (p.Arg541Gly)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369800	20	63414098	369800	353981	rs1555853971	LOF	0.848	0.052	0.1
KCNQ2	541	R	T	NM_172107.4(KCNQ2):c.1622G>C (p.Arg541Thr)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Mar 11, 2020)	criteria provided, conflicting interpretations	VCV000488537	20	63414097	488537	481405	rs1555853970	LOF	0.849	0.036	0.115
KCNQ2	542	A	V	NM_172107.4(KCNQ2):c.1625C>T (p.Ala542Val)	not provided	Uncertain significance(Last reviewed: Jan 17, 2015)	criteria provided, single submitter	VCV000205949	20	63414094	205949	203732	rs796052648	LOF	0.871	0.046	0.082
KCNQ2	543	G	S	NM_172107.4(KCNQ2):c.1720G>A (p.Gly574Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 14, 2020)	criteria provided, single submitter	VCV001022468	20	63413493	1022468	1014204	rs2080187424	LOF	0.86	0.06	0.08
KCNQ2	543	V	M	NM_172107.4(KCNQ2):c.1627G>A (p.Val543Met)	Early infantile epileptic encephalopathy 7|not provided|Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 1|Seizures	Conflicting interpretations of pathogenicity(Last reviewed: Sep 1, 2020)	criteria provided, conflicting interpretations	VCV000194446	20	63414092	194446	191609	rs794727134	LOF	0.859	0.037	0.104
KCNQ2	544	C	Y	NM_172107.4(KCNQ2):c.1631G>A (p.Cys544Tyr)	not provided	Likely pathogenic(Last reviewed: Nov 28, 2016)	criteria provided, single submitter	VCV000373576	20	63414088	373576	360583	rs1057518492	LOF	0.867	0.041	0.092
KCNQ2	545	V	I	NM_172107.4(KCNQ2):c.1633G>A (p.Val545Ile)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 25, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205911	20	63413580	205911	203730	rs757608181	LOF	0.857	0.038	0.105
KCNQ2	546	M	T	NM_172107.4(KCNQ2):c.1637T>C (p.Met546Thr)	not provided	Pathogenic(Last reviewed: Aug 16, 2016)	criteria provided, single submitter	VCV000280703	20	63413576	280703	265109	rs886041860	LOF	0.865	0.049	0.086
KCNQ2	546	M	V	NM_172107.4(KCNQ2):c.1636A>G (p.Met546Val)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000039761	20	63413577	39761	48360	rs397515420	LOF	0.87	0.04	0.09
KCNQ2	546	V	M	NM_172107.4(KCNQ2):c.1690G>A (p.Val564Met)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Aug 11,	criteria provided, single submitter	VCV000813775	20	63413523	813775	802033	NA	LOF	0.86	0.036	0.104

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2017)											
KCNQ2	547	R	G	NM_172107.4(KCNQ2):c.1639C>G (p.Arg547Gly)	Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Apr 12, 2016)	criteria provided, single submitter	VCV000226112	20	63413574	226112	227922	rs796052650	LOF	0.849	0.053	0.098
KCNQ2	547	R	Q	NM_172107.4(KCNQ2):c.1640G>A (p.Arg547Gln)	not provided	Uncertain significance(Last reviewed: May 20, 2021)	criteria provided, single submitter	VCV000390674	20	63413573	390674	379746	rs760984494	LOF	0.846	0.035	0.119
KCNQ2	547	R	W	NM_172107.4(KCNQ2):c.1639C>T (p.Arg547Trp)	Benign familial neonatal seizures 1|not provided|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic/Likely pathogenic(Last reviewed: Apr 9, 2021)	criteria provided, multiple submitters, no conflicts	VCV000205912	20	63413574	205912	203729	rs796052650	LOF	0.866	0.033	0.101
KCNQ2	552	K	N	NM_172107.4(KCNQ2):c.1749G>C (p.Lys583Asn)	Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: May 13, 2021)	criteria provided, multiple submitters, no conflicts	VCV001068430	20	63413464	1068430	1056618	NA	LOF	0.849	0.042	0.109
KCNQ2	552	K	T	NM_172107.4(KCNQ2):c.1655A>C (p.Lys552Thr)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369804	20	63413558	369804	353978	rs1555853613	LOF	0.863	0.037	0.1
KCNQ2	553	R	L	NM_172107.4(KCNQ2):c.1658G>T (p.Arg553Leu)	not provided|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Nov 8, 2017)	criteria provided, single submitter	VCV000369805	20	63413555	369805	353977	rs118192234	LOF	0.884	0.029	0.087
KCNQ2	553	R	P	NM_172107.4(KCNQ2):c.1658G>C (p.Arg553Pro)	Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Jun 20, 2017)	criteria provided, single submitter	VCV000560650	20	63413555	560650	551748	rs118192234	LOF	0.876	0.039	0.084
KCNQ2	553	R	Q	NM_172107.4(KCNQ2):c.1658G>A (p.Arg553Gln)	not provided|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic(Last reviewed: Feb 18, 2021)	criteria provided, multiple submitters, no conflicts	VCV000021767	20	63413555	21767	34619	rs118192234	LOF	0.867	0.032	0.101
KCNQ2	553	R	W	NM_172107.4(KCNQ2):c.1657C>T (p.Arg553Trp)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|Seizures|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 1, 2021)	criteria provided, conflicting interpretations	VCV000205913	20	63413556	205913	203728	rs759584387	LOF	0.881	0.032	0.087
KCNQ2	555	F	L	NM_172107.4(KCNQ2):c.1663T>C (p.Phe555Leu)	Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Feb 22, 2018)	criteria provided, single submitter	VCV000813758	20	63413550	813758	802034	NA	LOF	0.875	0.035	0.09
KCNQ2	556	K	E	NM_172107.4(KCNQ2):c.1666A>G (p.Lys556Glu)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000205914	20	63413547	205914	203727	rs1555853593	LOF	0.88	0.039	0.081
KCNQ2	557	L	P	NM_172107.4(KCNQ2):c.1754T>C (p.Leu585Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 18, 2019)	criteria provided, single submitter	VCV000843962	20	63413459	843962	848521	rs2080185796	LOF	0.877	0.053	0.07
KCNQ2	559	D	A	NM_172107.4(KCNQ2):c.1769A>C (p.Asp590Ala)	not provided	Uncertain significance(Last reviewed: May 31, 2019)	criteria provided, single submitter	VCV001306160	20	63408531	1306160	1297431	NA	LOF	0.886	0.041	0.073
KCNQ2	560	R	Q	NM_172107.4(KCNQ2):c.1679G>A (p.Arg560Gln)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Likely pathogenic(Last reviewed: Aug 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000372677	20	63413534	372677	360435	rs1057517919	LOF	0.845	0.071	0.084
KCNQ2	560	R	W	NM_172107.4(KCNQ2):c.1678C>T (p.Arg560Trp)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile	Pathogenic/Likely pathogenic(Last	criteria provided,	VCV000205915	20	63413535	205915	203726	rs773171451	LOF	0.835	0.076	0.089

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					epileptic encephalopathy 7|Absent speech|Limb dystonia|Global developmental delay| Neurodevelopmental disorder	reviewed: Oct 26, 2021)	multiple submitters, no conflicts									
KCNQ2	561	P	L	NM_172107.4(KCNQ2):c.1682C>T (p.Pro561Leu)	not provided|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Dec 18, 2018)	criteria provided, single submitter	VCV000205916	20	63413531	205916	203725	rs796052652	LOF	0.882	0.038	0.079
KCNQ2	563	D	E	NM_172107.4(KCNQ2):c.1689C>G (p.Asp563Glu)	Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369806	20	63413524	369806	353976	rs35450031	LOF	0.869	0.047	0.084
KCNQ2	563	D	N	NM_172107.4(KCNQ2):c.1687G>A (p.Asp563Asn)	Seizures|Early infantile epileptic encephalopathy with suppression bursts|not provided|Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Pathogenic(Last reviewed: Sep 15, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205917	20	63413526	205917	203724	rs796052653	LOF	0.863	0.051	0.086
KCNQ2	563	D	Y	NM_172107.4(KCNQ2):c.1687G>T (p.Asp563Tyr)	not provided	Pathogenic(Last reviewed: Jan 20, 2021)	criteria provided, single submitter	VCV000429424	20	63413526	429424	422338	rs796052653	LOF	0.881	0.04	0.08
KCNQ2	563	R	P	NM_172107.4(KCNQ2):c.1742G>C (p.Arg581Pro)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Jun 15, 2020)	criteria provided, conflicting interpretations	VCV000804957	20	63413471	804957	793850	rs118192235	LOF	0.882	0.041	0.077
KCNQ2	565	V	M	NM_172107.4(KCNQ2):c.1777G>A (p.Val593Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV000934644	20	63408523	934644	939023	rs372976131	LOF	0.863	0.036	0.1
KCNQ2	567	V	D	NM_172107.4(KCNQ2):c.1700T>A (p.Val567Asp)	not provided	Pathogenic(Last reviewed: Sep 14, 2020)	criteria provided, single submitter	VCV000429666	20	63413513	429666	422337	rs1131691518	LOF	0.868	0.052	0.08
KCNQ2	573	M	R	NM_172107.4(KCNQ2):c.1763+1576T>G	Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: May 12, 2020)	criteria provided, single submitter	VCV001098634	20	63411874	1098634	1087405	NA	LOF	0.88	0.041	0.079
KCNQ2	574	G	D	NM_172107.4(KCNQ2):c.1721G>A (p.Gly574Asp)	Early infantile epileptic encephalopathy 7	Likely pathogenic(Last reviewed: Feb 19, 2020)	no assertion criteria provided	VCV000205918	20	63413492	205918	203723	rs796052654	LOF	0.849	0.066	0.085
KCNQ2	578	M	I	NM_172107.4(KCNQ2):c.1734G>C (p.Met578Ile)	Early infantile epileptic encephalopathy 7|not provided	Pathogenic(Last reviewed: Jul 30, 2020)	criteria provided, single submitter	VCV000205919	20	63413479	205919	203722	rs796052655	LOF	0.867	0.042	0.091
KCNQ2	578	M	T	NM_172107.4(KCNQ2):c.1733T>C (p.Met578Thr)	not provided	Likely pathogenic(Last reviewed: Sep 19, 2017)	criteria provided, single submitter	VCV000452187	20	63413480	452187	446282	rs1555853524	LOF	0.876	0.046	0.078
KCNQ2	578	M	V	NM_172107.4(KCNQ2):c.1732A>G (p.Met578Val)	not provided|Benign familial neonatal seizures 1	Likely pathogenic(Last reviewed: Jan 15, 2019)	criteria provided, single submitter	VCV000369807	20	63413481	369807	353975	rs1057516123	LOF	0.87	0.042	0.088
KCNQ2	579	E	K	NM_172107.4(KCNQ2):c.1828G>A (p.Glu610Lys)	Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: Apr 30, 2019)	criteria provided, single submitter	VCV000930863	20	63408472	930863	919925	rs1350422852	LOF	0.876	0.042	0.082
KCNQ2	579	E	Q	NM_172107.4(KCNQ2):c.1828G>C (p.Glu610Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 25, 2020)	criteria provided, single submitter	VCV001006547	20	63408472	1006547	999079	rs1350422852	LOF	0.87	0.045	0.086
KCNQ2	580	P	L	NM_172107.4(KCNQ2):c.1823C>T (p.Pro608Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 30, 2019)	criteria provided, single submitter	VCV000844345	20	63408477	844345	848520	rs140217688	LOF	0.876	0.039	0.085

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	581	R	G	NM_172107.4(KCNQ2):c.1741C>G (p.Arg581Gly)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts|not provided	Pathogenic(Last reviewed: Jul 26, 2019)	criteria provided, multiple submitters, no conflicts	VCV000265380	20	63413472	265380	260235	rs118192236	LOF	0.861	0.049	0.09
KCNQ2	581	R	L	NM_172107.4(KCNQ2):c.1742G>T (p.Arg581Leu)	not provided	Likely pathogenic(Last reviewed: Aug 8, 2012)	criteria provided, single submitter	VCV000205920	20	63413471	205920	203720	rs118192235	LOF	0.881	0.029	0.09
KCNQ2	581	R	Q	NM_172107.4(KCNQ2):c.1742G>A (p.Arg581Gln)	not provided|Benign familial neonatal seizures 1|Epileptic encephalopathy|Early infantile epileptic encephalopathy with suppression bursts	Pathogenic/Likely pathogenic(Last reviewed: Sep 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000021769	20	63413471	21769	34621	rs118192235	LOF	0.858	0.033	0.109
KCNQ2	586	Q	P	NM_172107.4(KCNQ2):c.1757A>C (p.Gln586Pro)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jul 31, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205921	20	63413456	205921	203719	rs796052656	LOF	0.864	0.05	0.086
KCNQ2	588	R	S	NM_172107.4(KCNQ2):c.1764A>T (p.Arg588Ser)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021770	20	63408536	21770	34622	rs118192237	LOF	0.862	0.039	0.099
KCNQ2	592	I	M	NM_172107.4(KCNQ2):c.1776C>G (p.Ile592Met)	Benign Rolandic epilepsy	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000369810	20	63408524	369810	353974	rs201868078	LOF	0.865	0.038	0.096
KCNQ2	595	R	W	NM_172107.4(KCNQ2):c.1783C>T (p.Arg595Trp)	Benign familial neonatal seizures 1|not provided	Uncertain significance(Last reviewed: Jun 24, 2019)	criteria provided, single submitter	VCV000369811	20	63408517	369811	353973	rs1555851550	LOF	0.849	0.06	0.091
KCNQ2	598	A	V	NM_172107.4(KCNQ2):c.1793C>T (p.Ala598Val)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Jul 14, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205922	20	63408507	205922	203717	rs746079131	LOF	0.792	0.079	0.129
KCNQ2	600	T	M	NM_172107.4(KCNQ2):c.1799C>T (p.Thr600Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, single submitter	VCV000570302	20	63408501	570302	571351	rs1203090297	LOF	0.778	0.102	0.12
KCNQ2	602	M	T	NM_172107.4(KCNQ2):c.1898T>C (p.Met633Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 14, 2018)	criteria provided, single submitter	VCV000646001	20	63407365	646001	650443	rs777332271	LOF	0.821	0.08	0.098
KCNQ2	604	R	C	NM_172107.4(KCNQ2):c.1810C>T (p.Arg604Cys)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Sep 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205923	20	63408490	205923	203716	rs780942181	LOF	0.809	0.086	0.105
KCNQ2	604	R	H	NM_172107.4(KCNQ2):c.1811G>A (p.Arg604His)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV000568566	20	63408489	568566	573602	rs1203495492	LOF	0.827	0.079	0.094
KCNQ2	605	T	I	NM_172107.4(KCNQ2):c.1814C>T (p.Thr605Ile)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 27, 2017)	criteria provided, single submitter	VCV000405209	20	63408486	405209	404199	rs751334184	LOF	0.797	0.086	0.117
KCNQ2	605	T	S	NM_172107.4(KCNQ2):c.1814C>G (p.Thr605Ser)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Sep 19, 2020)	criteria provided, conflicting interpretations	VCV000205924	20	63408486	205924	203715	rs751334184	LOF	0.798	0.094	0.109
KCNQ2	611	A	V	NM_172107.4(KCNQ2):c.1832C>T (p.Ala611Val)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Benign/Likely benign(Last reviewed:	criteria provided,	VCV000205847	20	63408468	205847	203714	rs199644682	LOF	0.768	0.096	0.135

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						Oct 6, 2020)	multiple submitters, no conflicts										
KCNQ2	612	E	K	NM_172107.4(KCNQ2):c.1834G>A (p.Glu612Lys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 23, 2018)	criteria provided, single submitter	VCV000575263	20	63408466	575263	573600	rs1180808546	LOF	0.792	0.1	0.108
KCNQ2	612	I	L	NM_172107.4(KCNQ2):c.1927A>C (p.Ile643Leu)	KCNQ2-Related Disorders|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 12, 2021)	criteria provided, multiple submitters, no conflicts	VCV000639127	20	63407336	639127	650444	rs1021032210	LOF	0.789	0.09	0.121
KCNQ2	622	R	W	NM_172107.4(KCNQ2):c.1864C>T (p.Arg622Trp)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Aug 27, 2020)	criteria provided, conflicting interpretations	VCV000420848	20	63408436	420848	410816	rs772405187	LOF	0.864	0.054	0.082
KCNQ2	624	G	R	NM_172107.4(KCNQ2):c.1870G>A (p.Gly624Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 9, 2018)	criteria provided, single submitter	VCV000580776	20	63408430	580776	571349	rs771211103	LOF	0.84	0.064	0.096
KCNQ2	624	G	R	NM_172107.4(KCNQ2):c.1870G>A (p.Gly624Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 9, 2018)	criteria provided, single submitter	VCV000580776	20	63408430	580776	571349	rs771211103	LOF	0.84	0.064	0.096
KCNQ2	624	G	R	NM_172107.4(KCNQ2):c.1870G>C (p.Gly624Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 5, 2017)	criteria provided, single submitter	VCV000405213	20	63408430	405213	404203	rs771211103	LOF	0.84	0.064	0.096
KCNQ2	624	G	R	NM_172107.4(KCNQ2):c.1870G>C (p.Gly624Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 5, 2017)	criteria provided, single submitter	VCV000405213	20	63408430	405213	404203	rs771211103	LOF	0.84	0.064	0.096
KCNQ2	625	E	K	NM_172107.4(KCNQ2):c.1966G>A (p.Glu656Lys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 21, 2020)	criteria provided, single submitter	VCV000961623	20	63407297	961623	951146	rs545544936	LOF	0.87	0.051	0.079
KCNQ2	626	A	T	NM_172107.4(KCNQ2):c.1969G>A (p.Ala657Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 12, 2020)	criteria provided, single submitter	VCV001026116	20	63407294	1026116	1014202	rs780679486	LOF	0.867	0.053	0.08
KCNQ2	636	P	L	NM_172107.4(KCNQ2):c.1991C>T (p.Pro664Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV000936289	20	63407272	936289	939022	rs765493484	LOF	0.885	0.038	0.077
KCNQ2	637	L	R	NM_172107.4(KCNQ2):c.1910T>G (p.Leu637Arg)	Benign familial neonatal seizures 1	Pathogenic(Last reviewed: Mar 31, 2016)	no assertion criteria provided	VCV000021771	20	63407353	21771	34623	rs118192240	LOF	0.88	0.042	0.078
KCNQ2	637	T	N	NM_172107.4(KCNQ2):c.1964C>A (p.Thr655Asn)	not provided	Uncertain significance(Last reviewed: Mar 1, 2018)	criteria provided, single submitter	VCV000809268	20	63407299	809268	798004	rs1459962137	LOF	0.869	0.052	0.079
KCNQ2	638	P	L	NM_172107.4(KCNQ2):c.1997C>T (p.Pro666Leu)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Conflicting interpretations of pathogenicity(Last reviewed: Mar 17, 2020)	criteria provided, conflicting interpretations	VCV000803618	20	63407266	803618	791995	rs762130930	LOF	0.883	0.039	0.078
KCNQ2	640	L	P	NM_172107.4(KCNQ2):c.1919T>C (p.Leu640Pro)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 28, 2018)	criteria provided, single submitter	VCV000581083	20	63407344	581083	571347	rs1568862485	LOF	0.863	0.059	0.078

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	646	Q	R	NM_172107.4(KCNQ2):c.1937A>G (p.Gln646Arg)	not provided	Likely pathogenic(Last reviewed: Aug 8, 2013)	criteria provided, single submitter	VCV000205925	20	63407326	205925	203710	rs766755499	LOF	0.876	0.045	0.079
KCNQ2	647	R	Q	NM_172107.4(KCNQ2):c.1940G>A (p.Arg647Gln)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Jun 6, 2020)	criteria provided, multiple submitters, no conflicts	VCV000501056	20	63407323	501056	492480	rs765583552	LOF	0.868	0.046	0.086
KCNQ2	647	R	W	NM_172107.4(KCNQ2):c.1939C>T (p.Arg647Trp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 10, 2018)	criteria provided, single submitter	VCV000579920	20	63407324	579920	573598	rs773450069	LOF	0.858	0.05	0.092
KCNQ2	650	D	N	NM_172107.4(KCNQ2):c.2041G>A (p.Asp681Asn)	not provided	Uncertain significance(Last reviewed: Dec 31, 2018)	criteria provided, single submitter	VCV000804958	20	63407222	804958	793849	rs1380546879	LOF	0.833	0.076	0.091
KCNQ2	652	P	L	NM_172107.4(KCNQ2):c.1955C>T (p.Pro652Leu)	Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7|not provided	Conflicting interpretations of pathogenicity(Last reviewed: May 14, 2021)	criteria provided, conflicting interpretations	VCV000205951	20	63407308	205951	203708	rs770730662	LOF	0.822	0.071	0.107
KCNQ2	652	P	R	NM_172107.4(KCNQ2):c.1955C>G (p.Pro652Arg)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Sep 19, 2018)	criteria provided, multiple submitters, no conflicts	VCV000430045	20	63407308	430045	422336	rs770730662	LOF	0.813	0.079	0.107
KCNQ2	662	S	R	NM_172107.4(KCNQ2):c.2079C>G (p.Ser693Arg)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Jul 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV001007306	20	63407184	1007306	999078	rs2079971597	LOF	0.788	0.103	0.11
KCNQ2	667	A	V	NM_172107.4(KCNQ2):c.2000C>T (p.Ala667Val)	Early infantile epileptic encephalopathy with suppression bursts|not specified	Uncertain significance(Last reviewed: Feb 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000373184	20	63407263	373184	360475	rs563291132	LOF	0.805	0.083	0.112
KCNQ2	668	K	E	NM_172107.4(KCNQ2):c.2095A>G (p.Lys699Glu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 6, 2020)	criteria provided, single submitter	VCV001051022	20	63407168	1051022	1051777	NA	LOF	0.826	0.082	0.092
KCNQ2	669	P	L	NM_172107.4(KCNQ2):c.2006C>T (p.Pro669Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV000651375	20	63407257	651375	650447	rs774306858	LOF	0.784	0.106	0.11
KCNQ2	670	F	L	NM_172107.4(KCNQ2):c.2103C>G (p.Phe701Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 23, 2020)	criteria provided, single submitter	VCV001052247	20	63407160	1052247	1051776	NA	LOF	0.797	0.095	0.108
KCNQ2	673	P	S	NM_172107.4(KCNQ2):c.2110C>T (p.Pro704Ser)	not provided	Uncertain significance(Last reviewed: Dec 31, 2019)	criteria provided, single submitter	VCV001311778	20	63407153	1311778	1299835	NA	LOF	0.779	0.112	0.109
KCNQ2	674	S	L	NM_172107.4(KCNQ2):c.2105C>T (p.Ser702Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 4, 2018)	criteria provided, single submitter	VCV000658483	20	63407158	658483	650450	rs1555850884	LOF	0.784	0.099	0.117
KCNQ2	675	D	E	NM_172107.4(KCNQ2):c.2025C>G (p.Asp675Glu)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Aug 1, 2020)	criteria provided, multiple submitters, no conflicts	VCV000530521	20	63407238	530521	533682	rs553337046	LOF	0.787	0.11	0.103

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	677	P	H	NM_172107.4(KCNQ2):c.2123C>A (p.Pro708His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 4, 2019)	criteria provided, single submitter	VCV000848817	20	63407140	848817	848518	rs1360560623	LOF	0.792	0.105	0.103
KCNQ2	677	R	P	NM_172107.4(KCNQ2):c.2030G>C (p.Arg677Pro)	Early infantile epileptic encephalopathy 7|Benign familial neonatal seizures 1	Uncertain significance(Last reviewed: May 22, 2018)	criteria provided, single submitter	VCV000626125	20	63407233	626125	614476	rs1326189284	LOF	0.823	0.088	0.089
KCNQ2	677	R	W	NM_172107.4(KCNQ2):c.2029C>T (p.Arg677Trp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 3, 2018)	criteria provided, single submitter	VCV000582756	20	63407234	582756	575131	rs371851085	LOF	0.819	0.086	0.095
KCNQ2	679	A	V	NM_172107.4(KCNQ2):c.2120C>T (p.Ala707Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, single submitter	VCV001039210	20	63407143	1039210	1034791	rs753033741	LOF	0.765	0.093	0.141
KCNQ2	681	P	L	NM_172107.4(KCNQ2):c.2126C>T (p.Pro709Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Dec 27, 2019)	criteria provided, single submitter	VCV000858105	20	63407137	858105	848517	rs2079968880	LOF	0.773	0.105	0.122
KCNQ2	684	G	S	NM_172107.4(KCNQ2):c.2050G>A (p.Gly684Ser)	not provided	Uncertain significance(Last reviewed: Nov 26, 2013)	criteria provided, single submitter	VCV000205928	20	63407213	205928	203706	rs577281854	LOF	0.776	0.108	0.115
KCNQ2	686	I	V	NM_172107.4(KCNQ2):c.2056A>G (p.Ile686Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 25, 2020)	criteria provided, single submitter	VCV001041204	20	63407207	1041204	1034792	rs558926802	LOF	0.799	0.073	0.128
KCNQ2	689	I	L	NM_172107.4(KCNQ2):c.2065A>C (p.Ile689Leu)	Seizures|Early infantile epileptic encephalopathy with suppression bursts|not specified	Benign(Last reviewed: Nov 13, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205848	20	63407198	205848	203705	rs201701585	LOF	0.779	0.08	0.141
KCNQ2	690	V	M	NM_172107.4(KCNQ2):c.2068G>A (p.Val690Met)	not provided	Uncertain significance(Last reviewed: Nov 1, 2017)	criteria provided, single submitter	VCV000546903	20	63407195	546903	537392	rs373902907	LOF	0.787	0.08	0.133
KCNQ2	691	R	H	NM_172107.4(KCNQ2):c.2072G>A (p.Arg691His)	not specified|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Aug 23, 2018)	criteria provided, conflicting interpretations	VCV000205930	20	63407191	205930	203704	rs773194884	LOF	0.817	0.084	0.099
KCNQ2	694	R	H	NM_172107.4(KCNQ2):c.2174G>A (p.Arg725His)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, single submitter	VCV001022214	20	63407089	1022214	1014201	rs775271635	LOF	0.818	0.084	0.098
KCNQ2	694	S	T	NM_172107.4(KCNQ2):c.2081G>C (p.Ser694Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 21, 2020)	criteria provided, single submitter	VCV000405206	20	63407182	405206	404190	rs769558746	LOF	0.796	0.091	0.113
KCNQ2	696	E	K	NM_172107.4(KCNQ2):c.2032G>A (p.Glu678Lys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV000964951	20	63407231	964951	958866	rs2079974270	LOF	0.762	0.125	0.113
KCNQ2	696	G	S	NM_172107.4(KCNQ2):c.2179G>A (p.Gly727Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 19, 2019)	criteria provided, single submitter	VCV000840292	20	63407084	840292	848516	rs368549458	LOF	0.742	0.137	0.121
KCNQ2	696	P	L	NM_172107.4(KCNQ2):c.2171C>T (p.Pro724Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 9, 2018)	criteria provided, single submitter	VCV000642202	20	63407092	642202	650452	rs761544880	LOF	0.757	0.124	0.119

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	696	T	M	NM_172107.4(KCNQ2):c.2087C>T (p.Thr696Met)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jun 4, 2021)	criteria provided, conflicting interpretations	VCV000461412	20	63407176	461412	471465	rs570139975	LOF	0.764	0.114	0.123
KCNQ2	699	T	A	NM_172107.4(KCNQ2):c.2188A>G (p.Thr730Ala)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 5, 2020)	criteria provided, single submitter	VCV001010969	20	63407075	1010969	999077	rs1441250745	LOF	0.772	0.113	0.116
KCNQ2	705	V	M	NM_172107.4(KCNQ2):c.2197G>A (p.Val733Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, single submitter	VCV000850259	20	63407066	850259	848515	rs756563523	LOF	0.782	0.099	0.119
KCNQ2	706	G	D	NM_172107.4(KCNQ2):c.2210G>A (p.Gly737Asp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 7, 2018)	criteria provided, single submitter	VCV000657511	20	63407053	657511	650453	rs751657917	LOF	0.736	0.146	0.118
KCNQ2	707	A	T	NM_172107.4(KCNQ2):c.2119G>A (p.Ala707Thr)	Early infantile epileptic encephalopathy with suppression bursts|Seizures	Conflicting interpretations of pathogenicity(Last reviewed: Aug 17, 2020)	criteria provided, conflicting interpretations	VCV000577370	20	63407144	577370	571344	rs543477138	LOF	0.756	0.119	0.125
KCNQ2	707	D	N	NM_172107.4(KCNQ2):c.2203G>A (p.Asp735Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 10, 2019)	criteria provided, single submitter	VCV000937305	20	63407060	937305	939021	rs1333247951	LOF	0.752	0.135	0.113
KCNQ2	708	L	Q	NM_172107.4(KCNQ2):c.2216T>A (p.Leu739Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV001006011	20	63407047	1006011	999076	rs2079965385	LOF	0.778	0.107	0.114
KCNQ2	713	R	C	NM_172107.4(KCNQ2):c.2221C>T (p.Arg741Cys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 22, 2019)	criteria provided, single submitter	VCV000846206	20	63407042	846206	848514	rs2079965070	LOF	0.79	0.094	0.116
KCNQ2	716	A	V	NM_172107.4(KCNQ2):c.2240C>T (p.Ala747Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 24, 2019)	criteria provided, single submitter	VCV000937276	20	63407023	937276	939020	rs1244838198	LOF	0.792	0.088	0.12
KCNQ2	716	T	I	NM_172107.4(KCNQ2):c.2147C>T (p.Thr716Ile)	Early infantile epileptic encephalopathy 7	Pathogenic	no assertion criteria provided	VCV000219239	20	63407116	219239	217256	rs864321711	LOF	0.798	0.084	0.118
KCNQ2	719	R	W	NM_172107.4(KCNQ2):c.2248C>T (p.Arg750Trp)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 6, 2020)	criteria provided, single submitter	VCV001040518	20	63407015	1040518	1034790	rs759060719	LOF	0.799	0.097	0.104
KCNQ2	722	S	N	NM_172107.4(KCNQ2):c.2165G>A (p.Ser722Asn)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 18, 2017)	criteria provided, single submitter	VCV000461413	20	63407098	461413	470984	rs980574000	LOF	0.752	0.135	0.114
KCNQ2	725	P	L	NM_172107.4(KCNQ2):c.2228C>T (p.Pro743Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Aug 21, 2020)	criteria provided, single submitter	VCV000971350	20	63407035	971350	958865	rs757187432	LOF	0.762	0.126	0.113
KCNQ2	725	R	C	NM_172107.4(KCNQ2):c.2173C>T (p.Arg725Cys)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Mar 31, 2021)	criteria provided, conflicting interpretations	VCV000461414	20	63407090	461414	469438	rs768284828	LOF	0.779	0.115	0.106
KCNQ2	726	A	T	NM_172107.4(KCNQ2):c.2260G>A (p.Ala754Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Sep 15, 2020)	criteria provided, single submitter	VCV001040253	20	63407003	1040253	1034789	rs773822234	LOF	0.749	0.128	0.123

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ2	726	P	A	NM_172107.4(KCNQ2):c.2122C>G (p.Pro708Ala)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Nov 10, 2019)	criteria provided, single submitter	VCV000958346	20	63407141	958346	951144	rs767485841	LOF	0.746	0.138	0.115
KCNQ2	730	A	S	NM_172107.4(KCNQ2):c.2281G>T (p.Ala761Ser)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Conflicting interpretations of pathogenicity(Last reviewed: Mar 12, 2021)	criteria provided, conflicting interpretations	VCV001041104	20	63406982	1041104	1034788	rs587780366	LOF	0.774	0.113	0.113
KCNQ2	731	E	K	NM_172107.4(KCNQ2):c.2245G>A (p.Glu749Lys)	Early infantile epileptic encephalopathy 7|not provided	Uncertain significance(Last reviewed: May 1, 2016)	criteria provided, multiple submitters, no conflicts	VCV000809267	20	63407018	809267	798003	rs796052658	LOF	0.793	0.104	0.103
KCNQ2	736	R	Q	NM_172107.4(KCNQ2):c.2300G>A (p.Arg767Gln)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV000842415	20	63406963	842415	848512	rs752551225	LOF	0.784	0.111	0.105
KCNQ2	737	G	S	NM_172107.4(KCNQ2):c.2209G>A (p.Gly737Ser)	not provided	Likely benign(Last reviewed: Sep 3, 2020)	criteria provided, single submitter	VCV000339330	20	63407054	339330	346010	rs755013341	LOF	0.749	0.13	0.121
KCNQ2	740	V	A	NM_172107.4(KCNQ2):c.2219T>C (p.Val740Ala)	not provided	Uncertain significance(Last reviewed: Mar 30, 2017)	criteria provided, single submitter	VCV000424526	20	63407044	424526	410814	rs1064797020	LOF	0.791	0.094	0.115
KCNQ2	743	C	Y	NM_172107.4(KCNQ2):c.2321_2322delinsAT (p.Cys774Tyr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jun 14, 2019)	criteria provided, single submitter	VCV000855374	20	63406941 - 63406942	855374	848511	rs2079958591	LOF	0.789	0.104	0.107
KCNQ2	745	P	L	NM_172107.4(KCNQ2):c.2234C>T (p.Pro745Leu)	Intellectual disability|not specified	Uncertain significance(Last reviewed: Nov 29, 2016)	criteria provided, single submitter	VCV000373718	20	63407029	373718	360471	rs1057518569	LOF	0.786	0.104	0.11
KCNQ2	751	R	W	NM_172107.4(KCNQ2):c.2344C>T (p.Arg782Trp)	not provided	Uncertain significance(Last reviewed: Jun 14, 2019)	criteria provided, single submitter	VCV001302247	20	63406919	1302247	1292529	NA	LOF	0.792	0.103	0.105
KCNQ2	751	S	L	NM_172107.4(KCNQ2):c.2252C>T (p.Ser751Leu)	Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: Aug 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000339329	20	63407011	339329	346004	rs774002673	LOF	0.781	0.101	0.117
KCNQ2	755	Y	C	NM_172107.4(KCNQ2):c.2264A>G (p.Tyr755Cys)	Early infantile epileptic encephalopathy with suppression bursts|not specified|not provided| Seizures	Benign/Likely benign(Last reviewed: Dec 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000129333	20	63406999	129333	134779	rs3746366	LOF	0.76	0.119	0.121
KCNQ2	756	G	S	NM_172107.4(KCNQ2):c.2266G>A (p.Gly756Ser)	Early infantile epileptic encephalopathy with suppression bursts|Seizures|not provided	Benign/Likely benign(Last reviewed: Nov 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205849	20	63406997	205849	203700	rs200909197	LOF	0.738	0.14	0.122
KCNQ2	759	I	T	NM_172107.4(KCNQ2):c.2369T>C (p.Ile790Thr)	not provided	Uncertain significance(Last reviewed: Apr 8, 2019)	criteria provided, single submitter	VCV001304987	20	63406894	1304987	1295262	NA	LOF	0.758	0.122	0.12
KCNQ2	760	R	C	NM_172107.4(KCNQ2):c.2278C>T (p.Arg760Cys)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy 7	Uncertain significance(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000530437	20	63406985	530437	533610	rs1449467609	LOF	0.778	0.111	0.111
KCNQ2	760	R	H	NM_172107.4(KCNQ2):c.2279G>A	Early infantile epileptic encephalopathy with	Uncertain	criteria	VCV000216827	20	63406984	216827	213472	rs758530960	LOF	0.788	0.109	0.103

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Arg760His)	suppression bursts	significance(Last reviewed: Apr 17, 2020)	provided, single submitter										
KCNQ2	761	A	T	NM_172107.4(KCNQ2):c.2281G>A (p.Ala761Thr)	not provided	Uncertain significance(Last reviewed: Aug 8, 2013)	criteria provided, single submitter	VCV000129334	20	63406982	129334	134780	rs587780366	LOF	0.771	0.104	0.125
KCNQ2	762	V	M	NM_172107.4(KCNQ2):c.2377G>A (p.Val793Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 9, 2020)	criteria provided, single submitter	VCV001025174	20	63406886	1025174	1014200	rs377227909	LOF	0.769	0.113	0.118
KCNQ2	767	R	W	NM_172107.4(KCNQ2):c.2299C>T (p.Arg767Trp)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Dec 26, 2018)	criteria provided, multiple submitters, no conflicts	VCV000432454	20	63406964	432454	426350	rs755823361	LOF	0.781	0.115	0.104
KCNQ2	768	T	M	NM_172107.4(KCNQ2):c.2357C>T (p.Thr786Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 23, 2019)	criteria provided, single submitter	VCV000852085	20	63406906	852085	848510	rs754872408	LOF	0.757	0.123	0.12
KCNQ2	771	T	I	NM_172107.4(KCNQ2):c.2312C>T (p.Thr771Ile)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Sep 14, 2020)	criteria provided, conflicting interpretations	VCV000461417	20	63406951	461417	471463	rs759258191	LOF	0.764	0.114	0.121
KCNQ2	772	P	L	NM_172107.4(KCNQ2):c.2315C>T (p.Pro772Leu)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 7, 2019)	criteria provided, single submitter	VCV000530457	20	63406948	530457	533670	rs774206764	LOF	0.753	0.129	0.117
KCNQ2	774	C	F	NM_172107.4(KCNQ2):c.2321G>T (p.Cys774Phe)	not provided	Uncertain significance(Last reviewed: Apr 19, 2017)	criteria provided, single submitter	VCV000426357	20	63406942	426357	415695	rs762596565	LOF	0.766	0.114	0.12
KCNQ2	777	P	R	NM_172107.4(KCNQ2):c.2330C>G (p.Pro777Arg)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Oct 11, 2020)	criteria provided, conflicting interpretations	VCV000205851	20	63406933	205851	203698	rs142729516	LOF	0.753	0.132	0.114
KCNQ2	777	P	S	NM_172107.4(KCNQ2):c.2329C>T (p.Pro777Ser)	Early infantile epileptic encephalopathy with suppression bursts|not provided|Continuous spike and waves during slow-wave sleep syndrome	Conflicting interpretations of pathogenicity(Last reviewed: Aug 1, 2021)	criteria provided, conflicting interpretations	VCV000205850	20	63406934	205850	203699	rs748400155	LOF	0.753	0.133	0.114
KCNQ2	780	N	T	NM_172107.4(KCNQ2):c.2339A>C (p.Asn780Thr)	not provided|Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy with suppression bursts|Seizures|Early infantile epileptic encephalopathy 7|not specified	Benign(Last reviewed: Aug 19, 2021)	criteria provided, multiple submitters, no conflicts	VCV000021779	20	63406924	21779	34631	rs1801475	LOF	0.773	0.11	0.116
KCNQ2	785	A	S	NM_172107.4(KCNQ2):c.2446G>T (p.Ala816Ser)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Mar 10, 2020)	criteria provided, single submitter	VCV001023639	20	63406817	1023639	1014199	rs796052660	LOF	0.799	0.101	0.1
KCNQ2	785	D	E	NM_172107.4(KCNQ2):c.2355C>A (p.Asp785Glu)	not provided	Uncertain significance(Last reviewed: Jun 1, 2017)	criteria provided, single submitter	VCV000444569	20	63406908	444569	438209	rs781121496	LOF	0.796	0.107	0.097
KCNQ2	793	V	L	NM_172107.4(KCNQ2):c.2377G>T (p.Val793Leu)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Aug 28, 2020)	criteria provided, conflicting interpretations	VCV000279817	20	63406886	279817	264882	rs377227909	LOF	0.811	0.073	0.116
KCNQ2	795	P	R	NM_172107.4(KCNQ2):c.2477C>G	not provided	Uncertain	criteria	VCV001314255	20	63406786	1314255	1304516	NA	LOF	0.795	0.1	0.106

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Pro826Arg)		significance(Last reviewed: Apr 6, 2021)	provided, single submitter										
KCNQ2	802	F	L	NM_172107.4(KCNQ2):c.2404T>C (p.Phe802Leu)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205932	20	63406859	205932	203697	rs796052659	LOF	0.805	0.07	0.125
KCNQ2	816	A	T	NM_172107.4(KCNQ2):c.2446G>A (p.Ala816Thr)	not provided	Uncertain significance(Last reviewed: Jul 23, 2018)	criteria provided, single submitter	VCV000205933	20	63406817	205933	203696	rs796052660	LOF	0.773	0.103	0.124
KCNQ2	822	A	T	NM_172107.4(KCNQ2):c.2464G>A (p.Ala822Thr)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 27, 2020)	criteria provided, single submitter	VCV000461419	20	63406799	461419	470980	rs751425807	LOF	0.794	0.089	0.118
KCNQ2	822	A	V	NM_172107.4(KCNQ2):c.2465C>T (p.Ala822Val)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Jul 18, 2018)	criteria provided, multiple submitters, no conflicts	VCV000205934	20	63406798	205934	203695	rs796052661	LOF	0.789	0.084	0.126
KCNQ2	822	G	R	NM_172107.4(KCNQ2):c.2548G>A (p.Gly850Arg)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 9, 2020)	criteria provided, single submitter	VCV000855255	20	63406715	855255	848508	rs779986547	LOF	0.787	0.101	0.112
KCNQ2	825	A	G	NM_172107.4(KCNQ2):c.2474C>G (p.Ala825Gly)	Early infantile epileptic encephalopathy 7|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 9, 2019)	criteria provided, multiple submitters, no conflicts	VCV000582445	20	63406789	582445	571338	rs1568858969	LOF	0.83	0.083	0.087
KCNQ2	828	C	G	NM_172107.4(KCNQ2):c.2536T>G (p.Cys846Gly)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: May 21, 2020)	criteria provided, single submitter	VCV001046700	20	63406727	1046700	1034784	rs2079947062	LOF	0.831	0.081	0.087
KCNQ2	834	I	V	NM_172107.4(KCNQ2):c.2500A>G (p.Ile834Val)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Feb 4, 2019)	criteria provided, multiple submitters, no conflicts	VCV000502110	20	63406763	502110	493534	rs1555850289	LOF	0.827	0.061	0.111
KCNQ2	835	A	V	NM_172107.4(KCNQ2):c.2504C>T (p.Ala835Val)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Apr 6, 2018)	criteria provided, single submitter	VCV000578192	20	63406759	578192	575130	rs754310883	LOF	0.836	0.061	0.103
KCNQ2	842	V	M	NM_172107.4(KCNQ2):c.2470G>A (p.Val824Met)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, single submitter	VCV001038229	20	63406793	1038229	1034786	rs750108040	LOF	0.823	0.076	0.101
KCNQ2	843	R	K	NM_172107.4(KCNQ2):c.2612G>A (p.Arg871Lys)	Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Oct 28, 2020)	criteria provided, single submitter	VCV001039409	20	63406651	1039409	1034782	rs2079942562	LOF	0.849	0.069	0.082
KCNQ2	848	P	L	NM_172107.4(KCNQ2):c.2543C>T (p.Pro848Leu)	not provided|Early infantile epileptic encephalopathy with suppression bursts	Uncertain significance(Last reviewed: Jan 3, 2022)	criteria provided, multiple submitters, no conflicts	VCV000205935	20	63406720	205935	203693	rs377633507	LOF	0.814	0.086	0.1
KCNQ2	852	P	L	NM_172107.4(KCNQ2):c.2555C>T (p.Pro852Leu)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Uncertain significance(Last reviewed: Sep 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000205936	20	63406708	205936	203692	rs745508762	LOF	0.807	0.088	0.105
KCNQ2	854	R	C	NM_172107.4(KCNQ2):c.2560C>T	not provided|Early infantile epileptic	Benign(Last	criteria	VCV000379065	20	63406703	379065	378378	rs373536274	LOF	0.808	0.091	0.1

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Arg854Cys)	encephalopathy with suppression bursts	reviewed: Sep 30, 2020)	provided, multiple submitters, no conflicts										
KCNQ2	855	S	L	NM_172107.4(KCNQ2):c.2564C>T (p.Ser855Leu)	Early infantile epileptic encephalopathy with suppression bursts|Benign familial neonatal seizures 1|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Dec 18, 2020)	criteria provided, conflicting interpretations	VCV000021780	20	63406699	21780	34632	rs12481082	LOF	0.832	0.07	0.098
KCNQ2	858	G	S	NM_172107.4(KCNQ2):c.2572G>A (p.Gly858Ser)	Early infantile epileptic encephalopathy with suppression bursts|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Nov 1, 2021)	criteria provided, conflicting interpretations	VCV000205937	20	63406691	205937	203690	rs756609768	LOF	0.79	0.106	0.104
KCNQ2	862	F	L	NM_172107.4(KCNQ2):c.2584T>C (p.Phe862Leu)	not provided	Uncertain significance(Last reviewed: Jan 17, 2017)	criteria provided, single submitter	VCV000205938	20	63406679	205938	203689	rs796052662	LOF	0.781	0.094	0.125
KCNQ2	868	A	P	NM_172107.4(KCNQ2):c.2602G>C (p.Ala868Pro)	not provided	Uncertain significance(Last reviewed: Oct 6, 2017)	criteria provided, single submitter	VCV000452658	20	63406661	452658	446281	rs774595024	LOF	0.788	0.108	0.104
KCNQ2	871	R	S	NM_172107.4(KCNQ2):c.2613G>T (p.Arg871Ser)	Benign familial neonatal seizures 1|Early infantile epileptic encephalopathy with suppression bursts|Early infantile epileptic encephalopathy 7|not specified	Benign/Likely benign(Last reviewed: Oct 9, 2020)	criteria provided, multiple submitters, no conflicts	VCV000129343	20	63406650	129343	134789	rs587780369	LOF	0.815	0.097	0.089
KCNQ3	1	M	T	NM_004519.4(KCNQ3):c.2T>C (p.Met1Thr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 3, 2020)	criteria provided, single submitter	VCV001059498	8	132480531	1059498	1045256	NA	Neutral	0.685	0.207	0.108
KCNQ3	9	M	I	NM_004519.4(KCNQ3):c.387G>C (p.Val129=)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Feb 1, 2017)	criteria provided, single submitter	VCV000405214	8	132186181	405214	396571	rs1060500604	Neutral	0.707	0.188	0.106
KCNQ3	11	A	V	NM_004519.4(KCNQ3):c.32C>T (p.Ala11Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Feb 2, 2019)	criteria provided, single submitter	VCV000840823	8	132480501	840823	834141	rs1822534206	Neutral	0.672	0.204	0.124
KCNQ3	12	A	V	NM_004519.4(KCNQ3):c.35C>T (p.Ala12Val)	not specified|not provided|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Jan 4, 2019)	criteria provided, conflicting interpretations	VCV000205956	8	132480498	205956	202192	rs796052672	Neutral	0.71	0.17	0.12
KCNQ3	15	G	R	NM_004519.4(KCNQ3):c.403G>C (p.Gly135Arg)	Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Mar 5, 2019)	criteria provided, single submitter	VCV000931102	8	132186165	931102	919140	rs1826949712	Neutral	0.702	0.181	0.118
KCNQ3	15	G	V	NM_004519.4(KCNQ3):c.404G>T (p.Gly135Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 23, 2020)	criteria provided, single submitter	VCV001040583	8	132186164	1040583	1028294	rs1826949634	Neutral	0.681	0.199	0.12
KCNQ3	17	D	N	NM_004519.4(KCNQ3):c.49G>A (p.Asp17Asn)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Dec 18, 2017)	criteria provided, single submitter	VCV000538548	8	132480484	538548	523494	rs1355500787	Neutral	0.653	0.227	0.12
KCNQ3	21	G	R	NM_004519.4(KCNQ3):c.61G>A (p.Gly21Arg)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 4, 2020)	criteria provided, single submitter	VCV000838374	8	132480472	838374	834140	rs1822526871	Neutral	0.716	0.169	0.115
KCNQ3	23	G	S	NM_004519.4(KCNQ3):c.67G>A (p.Gly23Ser)	not provided	Uncertain significance(Last	criteria provided, single	VCV001163893	8	132480466	1163893	1152291	NA	Neutral	0.694	0.19	0.116

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Jul 16, 2020)	submitter										
KCNQ3	24	G	R	NM_004519.4(KCNQ3):c.70G>A (p.Gly24Arg)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 10, 2020)	criteria provided, single submitter	VCV001062443	8	132480463	1062443	1045254	NA	Neutral	0.693	0.19	0.116
KCNQ3	25	A	T	NM_004519.4(KCNQ3):c.73G>A (p.Ala25Thr)	Seizures	Uncertain significance(Last reviewed: Jun 15, 2016)	criteria provided, single submitter	VCV000587943	8	132480460	587943	579508	rs1280461599	Neutral	0.696	0.196	0.107
KCNQ3	31	G	R	NM_004519.4(KCNQ3):c.91G>A (p.Gly31Arg)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 3, 2019)	criteria provided, single submitter	VCV000954939	8	132480442	954939	945855	rs1433483426	Neutral	0.712	0.172	0.116
KCNQ3	33	A	V	NM_004519.4(KCNQ3):c.98C>T (p.Ala33Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 24, 2019)	criteria provided, single submitter	VCV000835443	8	132480435	835443	834138	rs761196042	Neutral	0.683	0.205	0.112
KCNQ3	34	A	E	NM_004519.4(KCNQ3):c.101C>A (p.Ala34Glu)	not provided	Uncertain significance(Last reviewed: Dec 1, 2019)	criteria provided, single submitter	VCV000871939	8	132480432	871939	859650	rs1412895282	Neutral	0.718	0.173	0.109
KCNQ3	36	A	P	NM_004519.4(KCNQ3):c.105_106inv (p.Ala36Pro)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 24, 2016)	criteria provided, single submitter	VCV000405215	8	132480427 - 132480428	405215	396572	NA	Neutral	0.695	0.194	0.111
KCNQ3	38	D	E	NM_004519.4(KCNQ3):c.114C>G (p.Asp38Glu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 20, 2020)	criteria provided, single submitter	VCV001045214	8	132480419	1045214	1028296	rs1477607835	Neutral	0.667	0.224	0.109
KCNQ3	39	E	G	NM_004519.4(KCNQ3):c.116A>G (p.Glu39Gly)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 30, 2020)	criteria provided, single submitter	VCV001040473	8	132480417	1040473	1028295	rs1822522895	Neutral	0.66	0.226	0.113
KCNQ3	39	E	Q	NM_004519.4(KCNQ3):c.115G>C (p.Glu39Gln)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Apr 23, 2019)	criteria provided, single submitter	VCV000645492	8	132480418	645492	636603	rs1448580874	Neutral	0.656	0.226	0.118
KCNQ3	43	V	M	NM_004519.4(KCNQ3):c.127G>A (p.Val43Met)	not provided	Uncertain significance(Last reviewed: May 13, 2014)	criteria provided, single submitter	VCV000193284	8	132480406	193284	190448	rs794726918	Neutral	0.699	0.201	0.101
KCNQ3	49	D	N	NM_004519.4(KCNQ3):c.145G>A (p.Asp49Asn)	Benign familial neonatal seizures 2|Benign Neonatal Epilepsy	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000361882	8	132480388	361882	307891	rs886062693	Neutral	0.688	0.184	0.128
KCNQ3	50	E	K	NM_004519.4(KCNQ3):c.508G>A (p.Glu170Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 4, 2020)	criteria provided, single submitter	VCV001036952	8	132184337	1036952	1028292	rs1826889658	Neutral	0.706	0.179	0.115
KCNQ3	51	E	D	NM_004519.4(KCNQ3):c.153G>C (p.Glu51Asp)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 4, 2019)	criteria provided, single submitter	VCV000834158	8	132480380	834158	834136	rs1405186695	Neutral	0.696	0.181	0.124
KCNQ3	52	Q	E	NM_004519.4(KCNQ3):c.154C>G (p.Gln52Glu)	not provided	Uncertain significance(Last reviewed: Nov 30, 2016)	criteria provided, single submitter	VCV000498242	8	132480379	498242	489666	rs1327292650	LOF	0.725	0.146	0.129
KCNQ3	57	L	P	NM_004519.4(KCNQ3):c.170T>C (p.Leu57Pro)	Benign familial neonatal seizures|Benign Neonatal Epilepsy	Uncertain significance(Last reviewed: Apr 11,	criteria provided, multiple	VCV000361881	8	132480363	361881	304271	rs886062692	LOF	0.811	0.111	0.079

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2019)	submitters, no conflicts										
KCNQ3	58	G	E	NM_004519.4(KCNQ3):c.173G>A (p.Gly58Glu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 8, 2019)	criteria provided, single submitter	VCV000958186	8	132480360	958186	945854	rs1822520136	LOF	0.784	0.103	0.113
KCNQ3	58	G	V	NM_004519.4(KCNQ3):c.173G>T (p.Gly58Val)	not provided	Uncertain significance(Last reviewed: May 20, 2020)	criteria provided, single submitter	VCV001196914	8	132480360	1196914	1187314	NA	LOF	0.768	0.11	0.122
KCNQ3	60	G	R	NM_004519.4(KCNQ3):c.178G>A (p.Gly60Arg)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 9, 2019)	criteria provided, single submitter	VCV000941767	8	132480355	941767	934097	rs1283325203	LOF	0.769	0.11	0.121
KCNQ3	65	G	A	NM_004519.4(KCNQ3):c.194G>C (p.Gly65Ala)	not provided	Likely pathogenic(Last reviewed: Apr 1, 2020)	criteria provided, single submitter	VCV000932600	8	132480339	932600	920788	rs1822519573	LOF	0.765	0.123	0.112
KCNQ3	65	G	R	NM_004519.4(KCNQ3):c.193G>C (p.Gly65Arg)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 18, 2020)	criteria provided, single submitter	VCV001051202	8	132480340	1051202	1045253	NA	LOF	0.781	0.107	0.112
KCNQ3	69	G	D	NM_004519.4(KCNQ3):c.566G>A (p.Gly189Asp)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 27, 2020)	criteria provided, single submitter	VCV001037603	8	132184279	1037603	1028291	rs1826887156	LOF	0.826	0.078	0.096
KCNQ3	72	G	S	NM_004519.4(KCNQ3):c.214G>A (p.Gly72Ser)	See cases|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 4, 2020)	criteria provided, multiple submitters, no conflicts	VCV000664040	8	132480319	664040	636602	rs759310149	Neutral	0.701	0.179	0.12
KCNQ3	74	R	H	NM_004519.4(KCNQ3):c.221G>A (p.Arg74His)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Feb 18, 2020)	criteria provided, single submitter	VCV000962495	8	132480312	962495	945853	rs1163071564	Neutral	0.682	0.195	0.124
KCNQ3	75	D	E	NM_004519.4(KCNQ3):c.225C>G (p.Asp75Glu)	Seizures|Benign familial neonatal seizures|not specified	Likely benign(Last reviewed: Nov 11, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205957	8	132480308	205957	202187	rs138254004	Neutral	0.679	0.2	0.121
KCNQ3	84	G	V	NM_004519.4(KCNQ3):c.251G>T (p.Gly84Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 26, 2020)	criteria provided, single submitter	VCV001015105	8	132480282	1015105	1007770	rs781753428	LOF	0.786	0.092	0.122
KCNQ3	85	I	L	NM_004519.4(KCNQ3):c.253A>C (p.Ile85Leu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jul 30, 2019)	criteria provided, single submitter	VCV000978547	8	132480280	978547	945852	rs969479579	LOF	0.802	0.101	0.097
KCNQ3	87	L	F	NM_004519.4(KCNQ3):c.259C>T (p.Leu87Phe)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 3, 2018)	criteria provided, single submitter	VCV000640814	8	132480274	640814	636601	rs752131052	LOF	0.827	0.087	0.086
KCNQ3	94	S	R	NM_004519.4(KCNQ3):c.282C>G (p.Ser94Arg)	Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Jan 13, 2018)	criteria provided, single submitter	VCV000910649	8	132480251	910649	898896	rs200519334	LOF	0.769	0.093	0.138
KCNQ3	98	K	R	NM_004519.4(KCNQ3):c.293A>G (p.Lys98Arg)	not provided|Seizures	Uncertain significance(Last reviewed: Mar 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000431878	8	132480240	431878	425778	rs143194379	LOF	0.75	0.11	0.14

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ3	107	R	Q	NM_004519.4(KCNQ3):c.680G>A (p.Arg227Gln)	Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: May 6, 2019)	criteria provided, single submitter	VCV000975970	8	132180254	975970	964298	rs1826712021	LOF	0.838	0.033	0.129
KCNQ3	110	L	F	NM_004519.4(KCNQ3):c.330G>C (p.Leu110Phe)	not provided	Uncertain significance(Last reviewed: Jul 19, 2013)	criteria provided, single submitter	VCV000205958	8	132480203	205958	202186	rs796052673	LOF	0.847	0.064	0.089
KCNQ3	110	R	S	NM_004519.4(KCNQ3):c.688C>A (p.Arg230Ser)	Intellectual disability	Pathogenic(Last reviewed: Apr 20, 2020)	criteria provided, single submitter	VCV000978934	8	132180246	978934	966957	rs796052676	LOF	0.819	0.055	0.126
KCNQ3	113	D	N	NM_004519.4(KCNQ3):c.337G>A (p.Asp113Asn)	Benign familial neonatal seizures|Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Oct 14, 2018)	criteria provided, multiple submitters, no conflicts	VCV000665904	8	132480196	665904	636600	rs1299548932	LOF	0.845	0.017	0.137
KCNQ3	116	R	C	NM_004519.4(KCNQ3):c.706C>T (p.Arg236Cys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 30, 2020)	criteria provided, single submitter	VCV001038319	8	132180228	1038319	1028290	rs1180768843	LOF	0.767	0.042	0.191
KCNQ3	116	R	H	NM_004519.4(KCNQ3):c.707G>A (p.Arg236His)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 26, 2020)	criteria provided, single submitter	VCV001026294	8	132180227	1026294	1007767	rs750016305	LOF	0.781	0.027	0.191
KCNQ3	118	P	L	NM_004519.4(KCNQ3):c.353C>T (p.Pro118Leu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 21, 2020)	criteria provided, single submitter	VCV001003299	8	132480180	1003299	992558	rs1822515868	LOF	0.846	0.038	0.116
KCNQ3	120	G	C	NM_004519.4(KCNQ3):c.358G>T (p.Gly120Cys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Nov 7, 2019)	criteria provided, single submitter	VCV000961761	8	132480175	961761	945851	rs1822515624	LOF	0.856	0.028	0.115
KCNQ3	122	R	Q	NM_004519.4(KCNQ3):c.725G>A (p.Arg242Gln)	not provided|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 8, 2021)	criteria provided, multiple submitters, no conflicts	VCV000934080	8	132180209	934080	934096	rs1826708929	LOF	0.818	0.02	0.161
KCNQ3	124	L	V	NM_004519.4(KCNQ3):c.370C>G (p.Leu124Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 20, 2020)	criteria provided, single submitter	VCV001007285	8	132480163	1007285	992557	rs371579261	LOF	0.838	0.071	0.091
KCNQ3	125	Y	H	NM_004519.4(KCNQ3):c.373T>C (p.Tyr125His)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 27, 2019)	criteria provided, single submitter	VCV000852826	8	132480160	852826	834135	rs1416256789	LOF	0.866	0.043	0.092
KCNQ3	129	L	F	NM_004519.4(KCNQ3):c.745C>T (p.Leu249Phe)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Feb 6, 2020)	criteria provided, single submitter	VCV001055480	8	132180189	1055480	1045249	NA	LOF	0.85	0.066	0.084
KCNQ3	135	G	E	NM_004519.4(KCNQ3):c.404G>A (p.Gly135Glu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV001062958	8	132186164	1062958	1045252	NA	LOF	0.874	0.035	0.091
KCNQ3	137	H	Y	NM_004519.4(KCNQ3):c.769C>T (p.His257Tyr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 25, 2019)	criteria provided, single submitter	VCV000937869	8	132180165	937869	934095	rs1826707510	LOF	0.863	0.04	0.097
KCNQ3	138	I	T	NM_004519.4(KCNQ3):c.413T>C (p.Ile138Thr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 15, 2020)	criteria provided, single submitter	VCV001042285	8	132186155	1042285	1028293	rs367706720	LOF	0.862	0.059	0.08

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ3	141	L	F	NM_004519.4(KCNQ3):c.781C>T (p.Leu261Phe)	not provided	Uncertain significance(Last reviewed: Jun 11, 2019)	criteria provided, single submitter	VCV001303275	8	132175605	1303275	1293553	NA	LOF	0.848	0.064	0.087
KCNQ3	141	V	I	NM_004519.4(KCNQ3):c.421G>A (p.Val141Ile)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV000952875	8	132186147	952875	925018	rs767730041	LOF	0.836	0.068	0.096
KCNQ3	150	T	N	NM_004519.4(KCNQ3):c.449C>A (p.Thr150Asn)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jul 21, 2020)	criteria provided, single submitter	VCV000579229	8	132186119	579229	567370	rs974274243	LOF	0.862	0.033	0.105
KCNQ3	159	V	F	NM_004519.4(KCNQ3):c.835G>T (p.Val279Phe)	Benign familial neonatal seizures 2	not provided	no assertion provided	VCV001334438	8	132175551	1334438	1325333	NA	LOF	0.857	0.053	0.089
KCNQ3	163	E	K	NM_004519.4(KCNQ3):c.847G>A (p.Glu283Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 5, 2020)	criteria provided, single submitter	VCV000663040	8	132175539	663040	636598	rs1586801275	LOF	0.862	0.034	0.104
KCNQ3	163	E	Q	NM_004519.4(KCNQ3):c.847G>C (p.Glu283Gln)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 15, 2019)	criteria provided, single submitter	VCV000971003	8	132175539	971003	955290	rs1586801275	LOF	0.861	0.033	0.106
KCNQ3	163	E	V	NM_004519.4(KCNQ3):c.848A>T (p.Glu283Val)	Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Jan 12, 2018)	criteria provided, single submitter	VCV000909732	8	132175538	909732	898893	rs1826520634	LOF	0.861	0.038	0.101
KCNQ3	164	I	V	NM_004519.4(KCNQ3):c.490A>G (p.Ile164Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 29, 2019)	criteria provided, single submitter	VCV000839067	8	132184355	839067	834134	rs1826890238	LOF	0.853	0.056	0.092
KCNQ3	166	V	F	NM_004519.4(KCNQ3):c.856G>T (p.Val286Phe)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 25, 2019)	criteria provided, single submitter	VCV000965065	8	132175530	965065	955289	rs549372035	LOF	0.855	0.06	0.085
KCNQ3	173	G	V	NM_004519.4(KCNQ3):c.878G>T (p.Gly293Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 23, 2020)	criteria provided, single submitter	VCV000967531	8	132175508	967531	955288	rs1064795142	LOF	0.866	0.047	0.087
KCNQ3	180	F	C	NM_004519.4(KCNQ3):c.899T>G (p.Phe300Cys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 23, 2020)	criteria provided, single submitter	VCV000999329	8	132175487	999329	992556	rs1554627439	LOF	0.867	0.034	0.099
KCNQ3	183	R	Q	NM_004519.4(KCNQ3):c.548G>A (p.Arg183Gln)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 24, 2020)	criteria provided, single submitter	VCV001062685	8	132184297	1062685	1045251	NA	LOF	0.816	0.042	0.143
KCNQ3	185	D	Y	NM_004519.4(KCNQ3):c.913G>T (p.Asp305Tyr)	not provided	Likely pathogenic(Last reviewed: May 11, 2015)	criteria provided, single submitter	VCV000427162	8	132175473	427162	415123	rs1085307996	LOF	0.831	0.055	0.113
KCNQ3	188	R	W	NM_004519.4(KCNQ3):c.562C>T (p.Arg188Trp)	not provided	Uncertain significance(Last reviewed: Jan 12, 2016)	criteria provided, single submitter	VCV000206002	8	132184283	206002	202184	rs754551218	LOF	0.79	0.059	0.15
KCNQ3	188	W	S	NM_004519.4(KCNQ3):c.923G>C (p.Trp308Ser)	Benign familial neonatal seizures 2|not provided	Likely pathogenic(Last reviewed: Mar 16, 2016)	criteria provided, single submitter	VCV000420680	8	132175463	420680	407312	rs1064794632	LOF	0.806	0.075	0.119
KCNQ3	190	G	D	NM_004519.4(KCNQ3):c.929G>A (p.Gly310Asp)	Benign familial neonatal seizures	Likely pathogenic(Last reviewed: Sep 29,	criteria provided, single submitter	VCV001067197	8	132175457	1067197	1055704	NA	LOF	0.879	0.021	0.1

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)											
KCNQ3	190	R	Q	NM_004519.4(KCNQ3):c.569G>A (p.Arg190Gln)	not provided	Uncertain significance(Last reviewed: Jan 29, 2019)	criteria provided, single submitter	VCV000205960	8	132184276	205960	202183	rs796052674	LOF	0.813	0.02	0.166
KCNQ3	193	T	I	NM_004519.4(KCNQ3):c.938C>T (p.Thr313Ile)	not provided	Likely pathogenic(Last reviewed: Aug 1, 2020)	criteria provided, single submitter	VCV001012603	8	132174345	1012603	1000614	rs1826478037	LOF	0.869	0.02	0.111
KCNQ3	196	T	N	NM_004519.4(KCNQ3):c.947C>A (p.Thr316Asn)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jul 7, 2020)	criteria provided, single submitter	VCV000999109	8	132174336	999109	992555	rs1826477775	LOF	0.802	0.096	0.102
KCNQ3	202	K	Q	NM_004519.4(KCNQ3):c.964A>C (p.Lys322Gln)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jul 25, 2020)	criteria provided, single submitter	VCV001003471	8	132174319	1003471	992554	rs1826477076	LOF	0.84	0.029	0.131
KCNQ3	203	I	T	NM_004519.4(KCNQ3):c.608T>C (p.Ile203Thr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 12, 2017)	criteria provided, single submitter	VCV000538552	8	132180326	538552	523627	rs1554628237	LOF	0.858	0.063	0.08
KCNQ3	203	I	V	NM_004519.4(KCNQ3):c.607A>G (p.Ile203Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 4, 2020)	criteria provided, single submitter	VCV001015294	8	132180327	1015294	1007768	rs370369681	LOF	0.817	0.088	0.095
KCNQ3	210	R	H	NM_004519.4(KCNQ3):c.989G>A (p.Arg330His)	Benign familial neonatal seizures|Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Jul 29, 2020)	criteria provided, single submitter	VCV000852086	8	132174294	852086	834131	rs1162306056	LOF	0.846	0.04	0.114
KCNQ3	210	R	L	NM_004519.4(KCNQ3):c.989G>T (p.Arg330Leu)	Benign familial neonatal seizures 2	not provided	no assertion provided	VCV001334436	8	132174294	1334436	1325331	NA	LOF	0.828	0.059	0.113
KCNQ3	214	A	T	NM_004519.4(KCNQ3):c.640G>A (p.Ala214Thr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Apr 6, 2018)	criteria provided, single submitter	VCV000581829	8	132180294	581829	562365	rs1563791055	LOF	0.871	0.043	0.085
KCNQ3	219	G	V	NM_004519.4(KCNQ3):c.656G>T (p.Gly219Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV000863246	8	132180278	863246	834133	rs1826713982	LOF	0.888	0.026	0.086
KCNQ3	220	G	V	NM_004519.4(KCNQ3):c.1019G>T (p.Gly340Val)	Benign familial neonatal seizures 2	not provided	no assertion provided	VCV001334437	8	132174264	1334437	1325332	NA	LOF	0.871	0.023	0.106
KCNQ3	223	F	S	NM_004519.4(KCNQ3):c.1028T>C (p.Phe343Ser)	not provided	Uncertain significance(Last reviewed: Feb 21, 2020)	criteria provided, single submitter	VCV001315235	8	132174255	1315235	1305496	NA	LOF	0.898	0.024	0.077
KCNQ3	230	R	C	NM_004519.4(KCNQ3):c.688C>T (p.Arg230Cys)	Intellectual disability|Benign familial neonatal seizures|Severe neurodevelopmental delay|Benign familial neonatal seizures|Lennox-Gastaut syndrome|Intellectual disability|Seizures, benign familial infantile, 5|Autistic behavior|Benign familial neonatal seizures 2|not provided	Pathogenic/Likely pathogenic(Last reviewed: Jan 21, 2022)	criteria provided, multiple submitters, no conflicts	VCV000205963	8	132180246	205963	202181	rs796052676	GOF	0.478	0.044	0.478
KCNQ3	230	R	H	NM_004519.4(KCNQ3):c.689G>A (p.Arg230His)	Benign familial neonatal seizures 2|Inborn genetic diseases|Benign familial neonatal seizures|not provided	Pathogenic/Likely pathogenic(Last reviewed: Jun 19, 2020)	criteria provided, multiple submitters, no conflicts	VCV000424397	8	132180245	424397	407314	NA	GOF	0.429	0.037	0.534
KCNQ3	236	A	T	NM_004519.4(KCNQ3):c.1066G>A (p.Ala356Thr)	Benign familial neonatal seizures 2	Likely pathogenic(Last reviewed: Jun 10, 2021)	no assertion criteria provided	VCV001319941	8	132172672	1319941	1310189	NA	GOF	0.667	0.027	0.305

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ3	244	R	C	NM_004519.4(KCNQ3):c.1090C>T (p.Arg364Cys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Dec 10, 2018)	criteria provided, single submitter	VCV000656938	8	132172648	656938	636595	rs1459374430	GOF	0.716	0.042	0.241
KCNQ3	244	R	H	NM_004519.4(KCNQ3):c.1091G>A (p.Arg364His)	Benign familial neonatal seizures|not provided| Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Mar 17, 2020)	criteria provided, conflicting interpretations	VCV000934997	8	132172647	934997	934093	rs1204519015	GOF	0.726	0.027	0.247
KCNQ3	247	H	N	NM_004519.4(KCNQ3):c.1099C>A (p.His367Asn)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 1, 2020)	criteria provided, single submitter	VCV001015596	8	132172639	1015596	1007765	rs1826414946	LOF	0.826	0.026	0.148
KCNQ3	253	A	G	NM_004519.4(KCNQ3):c.758C>G (p.Ala253Gly)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Nov 25, 2019)	criteria provided, single submitter	VCV000860349	8	132180176	860349	834132	rs1826707798	LOF	0.83	0.035	0.135
KCNQ3	253	K	T	NM_004519.4(KCNQ3):c.1118A>C (p.Lys373Thr)	not provided	Uncertain significance(Last reviewed: Feb 3, 2020)	criteria provided, single submitter	VCV001315047	8	132172620	1315047	1305308	NA	LOF	0.776	0.027	0.198
KCNQ3	255	A	T	NM_004519.4(KCNQ3):c.1123G>A (p.Ala375Thr)	Benign familial neonatal seizures	Likely pathogenic(Last reviewed: Jun 12, 2020)	criteria provided, single submitter	VCV001067624	8	132172615	1067624	1055703	NA	LOF	0.811	0.024	0.165
KCNQ3	258	L	V	NM_004519.4(KCNQ3):c.1132C>G (p.Leu378Val)	not provided	Uncertain significance(Last reviewed: Oct 27, 2021)	criteria provided, single submitter	VCV001254648	8	132172606	1254648	1244581	NA	LOF	0.832	0.044	0.124
KCNQ3	262	A	T	NM_004519.4(KCNQ3):c.1144G>A (p.Ala382Thr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 4, 2020)	criteria provided, single submitter	VCV001058084	8	132170425	1058084	1045248	NA	LOF	0.88	0.036	0.083
KCNQ3	263	T	M	NM_004519.4(KCNQ3):c.788C>T (p.Thr263Met)	Benign familial neonatal seizures|not provided	Uncertain significance(Last reviewed: May 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000498225	8	132175598	498225	489649	rs1479652323	LOF	0.862	0.024	0.114
KCNQ3	266	Y	C	NM_004519.4(KCNQ3):c.797A>G (p.Tyr266Cys)	not provided	Likely pathogenic(Last reviewed: Jan 24, 2013)	criteria provided, single submitter	VCV000205964	8	132175589	205964	202180	rs796052677	LOF	0.855	0.057	0.088
KCNQ3	286	V	I	NM_004519.4(KCNQ3):c.856G>A (p.Val286Ile)	not provided|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jul 29, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205965	8	132175530	205965	202179	rs549372035	LOF	0.847	0.064	0.09
KCNQ3	287	P	L	NM_004519.4(KCNQ3):c.860C>T (p.Pro287Leu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 3, 2019)	criteria provided, single submitter	VCV000405220	8	132175526	405220	395912	rs531151809	LOF	0.83	0.08	0.09
KCNQ3	293	G	E	NM_004519.4(KCNQ3):c.878G>A (p.Gly293Glu)	not provided	Likely pathogenic(Last reviewed: Mar 9, 2017)	criteria provided, single submitter	VCV000421443	8	132175508	421443	407313	rs1064795142	LOF	0.794	0.124	0.082
KCNQ3	299	E	K	NM_004519.4(KCNQ3):c.895G>A (p.Glu299Lys)	Benign familial neonatal seizures 2	not provided	no assertion provided	VCV000021413	8	132175491	21413	34265	rs118192247	LOF	0.888	0.027	0.085
KCNQ3	300	F	S	NM_004519.4(KCNQ3):c.899T>C (p.Phe300Ser)	Benign familial neonatal seizures 2	Likely pathogenic(Last reviewed: Jan 1, 2018)	criteria provided, single submitter	VCV000495233	8	132175487	495233	486747	rs1554627439	LOF	0.91	0.03	0.06

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ3	305	D	G	NM_004519.4(KCNQ3):c.914A>G (p.Asp305Gly)	Benign familial neonatal seizures 2	not provided	no assertion provided	VCV000021414	8	132175472	21414	34266	rs118192248	LOF	0.884	0.017	0.098
KCNQ3	305	D	V	NM_004519.4(KCNQ3):c.914A>T (p.Asp305Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 18, 2018)	criteria provided, single submitter	VCV000571923	8	132175472	571923	561955	rs118192248	LOF	0.881	0.015	0.104
KCNQ3	306	A	T	NM_004519.4(KCNQ3):c.916G>A (p.Ala306Thr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 15, 2018)	criteria provided, single submitter	VCV000659620	8	132175470	659620	636597	rs1586801127	LOF	0.908	0.013	0.079
KCNQ3	306	A	V	NM_004519.4(KCNQ3):c.917C>T (p.Ala306Val)	not provided	Likely pathogenic(Last reviewed: Aug 1, 2016)	criteria provided, single submitter	VCV000205966	8	132175469	205966	202178	rs796052678	LOF	0.901	0.016	0.083
KCNQ3	307	L	S	NM_004519.4(KCNQ3):c.1280T>C (p.Leu427Ser)	not provided|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 27, 2020)	criteria provided, multiple submitters, no conflicts	VCV001015608	8	132141314	1015608	1007764	rs1825289329	LOF	0.917	0.02	0.063
KCNQ3	309	W	R	NM_004519.4(KCNQ3):c.925T>C (p.Trp309Arg)	Benign familial neonatal seizures 2	Pathogenic(Last reviewed: Jun 1, 2000)	no assertion criteria provided	VCV000021415	8	132175461	21415	34267	rs118192249	LOF	0.901	0.023	0.076
KCNQ3	310	G	V	NM_004519.4(KCNQ3):c.929G>T (p.Gly310Val)	Benign familial neonatal seizures 2	Pathogenic(Last reviewed: Jan 1, 1998)	no assertion criteria provided	VCV000007392	8	132175457	7392	22431	rs118192250	LOF	0.897	0.016	0.087
KCNQ3	311	R	H	NM_004519.4(KCNQ3):c.1292G>A (p.Arg431His)	not provided	Uncertain significance(Last reviewed: May 24, 2021)	criteria provided, single submitter	VCV001326421	8	132141302	1326421	1316781	NA	LOF	0.878	0.022	0.1
KCNQ3	317	I	T	NM_004519.4(KCNQ3):c.950T>C (p.Ile317Thr)	Benign familial neonatal seizures|Benign familial neonatal seizures 2	Pathogenic(Last reviewed: Jul 14, 2020)	criteria provided, multiple submitters, no conflicts	VCV000661030	8	132174333	661030	636596	rs1586800133	LOF	0.899	0.034	0.067
KCNQ3	319	Y	C	NM_004519.4(KCNQ3):c.956A>G (p.Tyr319Cys)	not provided	Uncertain significance(Last reviewed: Oct 1, 2017)	criteria provided, single submitter	VCV000444762	8	132174327	444762	438402	rs1554627218	LOF	0.872	0.055	0.073
KCNQ3	320	G	E	NM_004519.4(KCNQ3):c.959G>A (p.Gly320Glu)	Benign familial neonatal seizures|Seizures	Uncertain significance(Last reviewed: Mar 6, 2018)	criteria provided, multiple submitters, no conflicts	VCV000577120	8	132174324	577120	561951	rs1563787894	LOF	0.894	0.039	0.067
KCNQ3	324	P	A	NM_004519.4(KCNQ3):c.970C>G (p.Pro324Ala)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 8, 2019)	criteria provided, single submitter	VCV000941355	8	132174313	941355	934094	rs1826476981	LOF	0.905	0.025	0.07
KCNQ3	327	P	T	NM_004519.4(KCNQ3):c.1339C>A (p.Pro447Thr)	not provided	Uncertain significance(Last reviewed: Aug 29, 2019)	criteria provided, single submitter	VCV001305660	8	132141255	1305660	1298253	NA	LOF	0.901	0.027	0.071
KCNQ3	329	N	I	NM_004519.4(KCNQ3):c.1346A>T (p.Asn449Ile)	Malignant tumor of prostate	Uncertain significance	no assertion criteria provided	VCV000161570	8	132141248	161570	171446	rs193920887	LOF	0.892	0.016	0.093
KCNQ3	330	R	C	NM_004519.4(KCNQ3):c.988C>T (p.Arg330Cys)	Benign familial neonatal seizures 2|Benign familial neonatal seizures	Pathogenic/Likely pathogenic(Last reviewed: Oct 2, 2021)	criteria provided, multiple submitters, no conflicts	VCV000021417	8	132174295	21417	34269	rs118192251	LOF	0.841	0.03	0.128
KCNQ3	332	I	F	NM_004519.4(KCNQ3):c.994A>T (p.Ile332Phe)	not provided	Likely benign(Last reviewed: Jan 18, 2018)	criteria provided, single submitter	VCV000633509	8	132174289	633509	621959	rs1563787859	LOF	0.88	0.038	0.082

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ3	333	A	P	NM_004519.4(KCNQ3):c.997G>C (p.Ala333Pro)	not provided	Uncertain significance(Last reviewed: Jan 1, 2017)	criteria provided, single submitter	VCV000425449	8	132174286	425449	413783	rs1064797349	LOF	0.906	0.019	0.074
KCNQ3	334	A	T	NM_004519.4(KCNQ3):c.1000G>A (p.Ala334Thr)	Rolandic epilepsy|Inborn genetic diseases| Benign familial neonatal seizures	Uncertain significance(Last reviewed: Apr 9, 2019)	criteria provided, multiple submitters, no conflicts	VCV000433111	8	132174283	433111	426672	rs1381851622	LOF	0.881	0.036	0.083
KCNQ3	347	N	D	NM_004519.4(KCNQ3):c.1399A>G (p.Asn467Asp)	not provided	Uncertain significance(Last reviewed: Feb 3, 2021)	criteria provided, single submitter	VCV001181815	8	132141195	1181815	1171813	NA	LOF	0.874	0.024	0.102
KCNQ3	348	A	V	NM_004519.4(KCNQ3):c.1043C>T (p.Ala348Val)	not provided|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205967	8	132174240	205967	202177	rs796052679	LOF	0.877	0.031	0.092
KCNQ3	348	N	K	NM_004519.4(KCNQ3):c.1404T>G (p.Asn468Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 16, 2019)	criteria provided, single submitter	VCV000946362	8	132141190	946362	925017	rs754462752	LOF	0.87	0.019	0.11
KCNQ3	348	N	K	NM_004519.4(KCNQ3):c.1404T>G (p.Asn468Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 16, 2019)	criteria provided, single submitter	VCV000946362	8	132141190	946362	925017	rs754462752	LOF	0.87	0.019	0.11
KCNQ3	348	N	K	NM_004519.4(KCNQ3):c.1404T>A (p.Asn468Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Dec 11, 2019)	criteria provided, single submitter	VCV000860868	8	132141190	860868	834128	rs754462752	LOF	0.87	0.019	0.11
KCNQ3	348	N	K	NM_004519.4(KCNQ3):c.1404T>A (p.Asn468Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Dec 11, 2019)	criteria provided, single submitter	VCV000860868	8	132141190	860868	834128	rs754462752	LOF	0.87	0.019	0.11
KCNQ3	351	R	C	NM_004519.4(KCNQ3):c.1411C>T (p.Arg471Cys)	not provided	Uncertain significance(Last reviewed: Feb 1, 2020)	criteria provided, single submitter	VCV001315176	8	132141183	1315176	1305437	NA	LOF	0.834	0.061	0.104
KCNQ3	354	G	R	NM_004519.4(KCNQ3):c.1060G>A (p.Gly354Arg)	not provided	Pathogenic(Last reviewed: Oct 13, 2021)	criteria provided, single submitter	VCV000449733	8	132172678	449733	444214	rs796052680	LOF	0.861	0.021	0.118
KCNQ3	354	G	W	NM_004519.4(KCNQ3):c.1060G>T (p.Gly354Trp)	not provided	Uncertain significance(Last reviewed: Jul 28, 2014)	criteria provided, single submitter	VCV000205968	8	132172678	205968	202176	rs796052680	LOF	0.865	0.031	0.104
KCNQ3	356	A	V	NM_004519.4(KCNQ3):c.1067C>T (p.Ala356Val)	not provided	Likely pathogenic(Last reviewed: Jun 15, 2017)	criteria provided, single submitter	VCV000432744	8	132172671	432744	425777	rs1554627025	LOF	0.842	0.066	0.091
KCNQ3	371	G	E	NM_004519.4(KCNQ3):c.1472G>A (p.Gly491Glu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Dec 21, 2018)	criteria provided, single submitter	VCV000658569	8	132140172	658569	636594	rs753920912	Neutral	0.732	0.181	0.086
KCNQ3	377	A	E	NM_004519.4(KCNQ3):c.1490C>A (p.Ala497Glu)	not provided	Uncertain significance(Last reviewed: Dec 9, 2019)	criteria provided, single submitter	VCV001211402	8	132140154	1211402	1201399	NA	LOF	0.774	0.137	0.089
KCNQ3	381	A	V	NM_004519.4(KCNQ3):c.1142C>T (p.Ala381Val)	Benign familial neonatal seizures 2|Rolandic epilepsy	Pathogenic(Last reviewed: Jan 1, 2017)	no assertion criteria provided	VCV000433110	8	132170427	433110	426671	rs1554626549	LOF	0.812	0.097	0.091
KCNQ3	387	P	R	NM_004519.4(KCNQ3):c.1520C>G	Benign familial neonatal seizures	Uncertain	criteria	VCV001002077	8	132140124	1002077	992553	rs370333805	LOF	0.773	0.137	0.09

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Pro507Arg)		significance(Last reviewed: Oct 6, 2020)	provided, single submitter										
KCNQ3	393	I	T	NM_004519.4(KCNQ3):c.1178T>C (p.Ile393Thr)	Benign familial neonatal seizures|Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000570705	8	132170391	570705	564638	rs201814804	LOF	0.756	0.15	0.093
KCNQ3	400	R	K	NM_004519.4(KCNQ3):c.1199G>A (p.Arg400Lys)	Benign familial neonatal seizures|Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Jan 8, 2020)	criteria provided, multiple submitters, no conflicts	VCV000471214	8	132170370	471214	457193	rs943073757	Neutral	0.726	0.174	0.1
KCNQ3	403	E	K	NM_004519.4(KCNQ3):c.1207G>A (p.Glu403Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 16, 2019)	criteria provided, single submitter	VCV000070789	8	132170362	70789	81680	rs142445773	Neutral	0.741	0.164	0.095
KCNQ3	403	R	G	NM_004519.4(KCNQ3):c.1567A>G (p.Arg523Gly)	not provided	Uncertain significance(Last reviewed: Oct 15, 2019)	criteria provided, single submitter	VCV001309235	8	132140077	1309235	1299934	NA	Neutral	0.733	0.17	0.097
KCNQ3	406	V	I	NM_004519.4(KCNQ3):c.1216G>A (p.Val406Ile)	not provided|Benign familial neonatal seizures| Benign familial neonatal seizures 2|Seizures	Conflicting interpretations of pathogenicity(Last reviewed: Nov 8, 2018)	criteria provided, conflicting interpretations	VCV000205969	8	132170353	205969	202174	rs144474368	LOF	0.84	0.067	0.093
KCNQ3	408	R	C	NM_004519.4(KCNQ3):c.1582C>T (p.Arg528Cys)	Benign familial neonatal seizures 2|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 13, 2020)	criteria provided, multiple submitters, no conflicts	VCV001034514	8	132138003	1034514	1028288	rs1393085163	Neutral	0.694	0.208	0.099
KCNQ3	409	P	R	NM_004519.4(KCNQ3):c.1226C>G (p.Pro409Arg)	not provided|Benign familial neonatal seizures| Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Oct 9, 2020)	criteria provided, conflicting interpretations	VCV000205970	8	132170343	205970	202173	rs149272208	Neutral	0.666	0.24	0.095
KCNQ3	410	Y	H	NM_004519.4(KCNQ3):c.1588T>C (p.Tyr530His)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 30, 2018)	criteria provided, single submitter	VCV000644020	8	132137997	644020	636592	rs781350596	Neutral	0.699	0.21	0.091
KCNQ3	414	E	G	NM_004519.4(KCNQ3):c.1241A>G (p.Glu414Gly)	Benign familial neonatal seizures 2|Benign familial neonatal seizures|Benign Neonatal Epilepsy|Seizures|not specified	Benign(Last reviewed: Dec 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000021409	8	132163489	21409	34261	rs2303995	Neutral	0.678	0.233	0.089
KCNQ3	417	E	M	NM_004519.4(KCNQ3):c.1249_1250delinsAT (p.Glu417Met)	Benign familial neonatal seizures 2|not specified	Uncertain significance(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000450849	8	132163480 - 132163481	450849	444213	rs1554625699	Neutral	0.61	0.292	0.098
KCNQ3	422	D	G	NM_004519.4(KCNQ3):c.1625A>G (p.Asp542Gly)	not provided	Uncertain significance(Last reviewed: May 6, 2019)	criteria provided, single submitter	VCV001305364	8	132137960	1305364	1295639	NA	Neutral	0.531	0.367	0.101
KCNQ3	422	D	N	NM_004519.4(KCNQ3):c.1624G>A (p.Asp542Asn)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jul 14, 2019)	criteria provided, single submitter	VCV000940181	8	132137961	940181	934091	rs1005759975	Neutral	0.511	0.383	0.107
KCNQ3	428	E	D	NM_004519.4(KCNQ3):c.1644G>T (p.Glu548Asp)	not provided	Uncertain significance(Last reviewed: Apr 9, 2020)	criteria provided, single submitter	VCV001191192	8	132137941	1191192	1180410	NA	Neutral	0.56	0.34	0.101

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ3	433	G	R	NM_004519.4(KCNQ3):c.1657G>A (p.Gly553Arg)	Benign familial neonatal seizures 2|Intellectual disability	Uncertain significance(Last reviewed: Apr 20, 2020)	criteria provided, single submitter	VCV000978935	8	132137928	978935	966956	rs1825159081	Neutral	0.615	0.288	0.097
KCNQ3	443	Y	F	NM_004519.4(KCNQ3):c.1688A>T (p.Tyr563Phe)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 6, 2020)	criteria provided, single submitter	VCV001054424	8	132137897	1054424	1045247	NA	Neutral	0.651	0.259	0.09
KCNQ3	446	T	M	NM_004519.4(KCNQ3):c.1697C>T (p.Thr566Met)	not provided|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000853636	8	132137888	853636	834124	rs746403693	Neutral	0.631	0.267	0.102
KCNQ3	449	D	G	NM_004519.4(KCNQ3):c.1706A>G (p.Asp569Gly)	not provided	Uncertain significance(Last reviewed: Jul 8, 2021)	criteria provided, single submitter	VCV001313762	8	132134383	1313762	1304023	NA	Neutral	0.543	0.361	0.096
KCNQ3	450	M	I	NM_004519.4(KCNQ3):c.1710G>A (p.Met570Ile)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 12, 2020)	criteria provided, single submitter	VCV001016296	8	132134379	1016296	1007763	rs375264509	Neutral	0.666	0.236	0.098
KCNQ3	454	P	R	NM_004519.4(KCNQ3):c.1721C>G (p.Pro574Arg)	not provided	Uncertain significance(Last reviewed: Oct 10, 2019)	criteria provided, single submitter	VCV001311782	8	132134368	1311782	1302313	NA	Neutral	0.575	0.329	0.096
KCNQ3	455	E	Q	NM_004519.4(KCNQ3):c.1363G>C (p.Glu455Gln)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 27, 2020)	criteria provided, single submitter	VCV000852958	8	132141231	852958	834129	rs1338855011	Neutral	0.524	0.376	0.1
KCNQ3	455	G	R	NM_004519.4(KCNQ3):c.1723G>A (p.Gly575Arg)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 30, 2019)	criteria provided, single submitter	VCV000951413	8	132134366	951413	925016	rs755560218	Neutral	0.589	0.315	0.096
KCNQ3	456	P	R	NM_004519.4(KCNQ3):c.1727C>G (p.Pro576Arg)	not provided	Uncertain significance(Last reviewed: Jan 24, 2020)	criteria provided, single submitter	VCV001312683	8	132134362	1312683	1302944	NA	Neutral	0.625	0.282	0.093
KCNQ3	459	T	M	NM_004519.4(KCNQ3):c.1736C>T (p.Thr579Met)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 19, 2018)	criteria provided, single submitter	VCV000660000	8	132134353	660000	636591	rs761487326	Neutral	0.624	0.28	0.096
KCNQ3	464	V	A	NM_004519.4(KCNQ3):c.1391T>C (p.Val464Ala)	Intellectual disability|not provided|Benign familial neonatal seizures	Likely benign(Last reviewed: Jan 12, 2021)	criteria provided, multiple submitters, no conflicts	VCV000205952	8	132141203	205952	202172	rs143664009	Neutral	0.655	0.253	0.092
KCNQ3	468	G	A	NM_004519.4(KCNQ3):c.1763G>C (p.Gly588Ala)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 30, 2020)	criteria provided, single submitter	VCV001056432	8	132134326	1056432	1045246	NA	Neutral	0.543	0.351	0.106
KCNQ3	468	N	S	NM_004519.4(KCNQ3):c.1403A>G (p.Asn468Ser)	Benign familial neonatal seizures|not provided| Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Oct 1, 2020)	criteria provided, conflicting interpretations	VCV000021410	8	132141191	21410	34262	rs118192252	Neutral	0.477	0.404	0.119
KCNQ3	469	S	L	NM_004519.4(KCNQ3):c.1766C>T (p.Ser589Leu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 6, 2020)	criteria provided, single submitter	VCV001024659	8	132134323	1024659	1007762	rs1824994797	Neutral	0.612	0.289	0.099
KCNQ3	470	A	V	NM_004519.4(KCNQ3):c.1769C>T (p.Ala590Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Nov 1, 2019)	criteria provided, single submitter	VCV000960588	8	132134320	960588	945850	rs1824994259	Neutral	0.711	0.195	0.094

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ3	471	R	P	NM_004519.4(KCNQ3):c.1412G>C (p.Arg471Pro)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Feb 24, 2020)	criteria provided, single submitter	VCV000853692	8	132141182	853692	834127	rs1198584017	Neutral	0.675	0.233	0.092
KCNQ3	473	R	H	NM_004519.4(KCNQ3):c.1418G>A (p.Arg473His)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 20, 2020)	criteria provided, single submitter	VCV001042515	8	132141176	1042515	1028289	rs138181943	Neutral	0.726	0.184	0.09
KCNQ3	474	T	M	NM_004519.4(KCNQ3):c.1421C>T (p.Thr474Met)	not provided|Benign familial neonatal seizures| Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Oct 24, 2020)	criteria provided, conflicting interpretations	VCV000205971	8	132141173	205971	202171	rs757583944	LOF	0.785	0.122	0.093
KCNQ3	477	R	H	NM_004519.4(KCNQ3):c.1430G>A (p.Arg477His)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Nov 2, 2018)	criteria provided, single submitter	VCV000538550	8	132141164	538550	523492	rs141821338	LOF	0.746	0.156	0.098
KCNQ3	481	N	K	NM_004519.4(KCNQ3):c.1803T>G (p.Asn601Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 9, 2020)	criteria provided, single submitter	VCV001014603	8	132132261	1014603	1007761	rs1469007568	Neutral	0.587	0.314	0.099
KCNQ3	486	S	R	NM_004519.4(KCNQ3):c.1458T>A (p.Ser486Arg)	not provided	Uncertain significance(Last reviewed: Jan 26, 2016)	criteria provided, single submitter	VCV000285490	8	132141136	285490	269727	rs886043116	Neutral	0.625	0.282	0.093
KCNQ3	491	G	R	NM_004519.4(KCNQ3):c.1471G>A (p.Gly491Arg)	Benign familial neonatal seizures|not provided	Uncertain significance(Last reviewed: Nov 11, 2019)	criteria provided, multiple submitters, no conflicts	VCV000279818	8	132140173	279818	264363	rs201552546	Neutral	0.609	0.296	0.095
KCNQ3	493	I	T	NM_004519.4(KCNQ3):c.1838T>C (p.Ile613Thr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 24, 2020)	criteria provided, single submitter	VCV001001990	8	132132226	1001990	992552	rs1488846886	Neutral	0.645	0.262	0.093
KCNQ3	497	A	T	NM_004519.4(KCNQ3):c.1489G>A (p.Ala497Thr)	Benign familial neonatal seizures|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jan 19, 2021)	criteria provided, conflicting interpretations	VCV000661154	8	132140155	661154	636593	rs368279666	Neutral	0.602	0.304	0.094
KCNQ3	497	A	V	NM_004519.4(KCNQ3):c.1490C>T (p.Ala497Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Dec 13, 2019)	criteria provided, single submitter	VCV000863207	8	132140154	863207	834126	rs1048969639	Neutral	0.647	0.26	0.094
KCNQ3	503	G	R	NM_004519.4(KCNQ3):c.1507G>A (p.Gly503Arg)	not provided|Benign familial neonatal seizures| Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Aug 4, 2020)	criteria provided, conflicting interpretations	VCV000205972	8	132140137	205972	202170	rs773584143	Neutral	0.6	0.299	0.1
KCNQ3	507	P	A	NM_004519.4(KCNQ3):c.1519C>G (p.Pro507Ala)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 29, 2019)	criteria provided, single submitter	VCV000839066	8	132140125	839066	834125	rs1480684032	Neutral	0.724	0.192	0.084
KCNQ3	509	E	K	NM_004519.4(KCNQ3):c.1525G>A (p.Glu509Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Aug 1, 2018)	criteria provided, single submitter	VCV000570800	8	132140119	570800	564636	rs774311301	Neutral	0.741	0.174	0.085
KCNQ3	510	D	A	NM_004519.4(KCNQ3):c.1529A>C (p.Asp510Ala)	not provided	Uncertain significance(Last reviewed: Apr 7, 2017)	criteria provided, single submitter	VCV000501097	8	132140115	501097	492521	rs1554622841	Neutral	0.735	0.18	0.084
KCNQ3	510	Q	K	NM_004519.4(KCNQ3):c.1888C>A (p.Gln630Lys)	Benign familial neonatal seizures	Uncertain significance(Last	criteria provided, single	VCV000838296	8	132129993	838296	834122	rs1466449283	Neutral	0.74	0.176	0.084

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						reviewed: Dec 19, 2019)	submitter										
KCNQ3	511	M	V	NM_004519.4(KCNQ3):c.1531A>G (p.Met511Val)	not specified	Uncertain significance(Last reviewed: May 20, 2016)	criteria provided, single submitter	VCV000279819	8	132140113	279819	264260	rs200219106	LOF	0.766	0.147	0.088
KCNQ3	513	P	L	NM_004519.4(KCNQ3):c.1538C>T (p.Pro513Leu)	not provided	Uncertain significance(Last reviewed: Feb 5, 2013)	criteria provided, single submitter	VCV000205973	8	132140106	205973	202169	rs768520561	LOF	0.757	0.156	0.087
KCNQ3	515	L	V	NM_004519.4(KCNQ3):c.1543C>G (p.Leu515Val)	Benign familial neonatal seizures|not provided	Uncertain significance(Last reviewed: Aug 27, 2019)	criteria provided, multiple submitters, no conflicts	VCV000288032	8	132140101	288032	272269	rs368013249	LOF	0.842	0.077	0.082
KCNQ3	517	A	T	NM_004519.4(KCNQ3):c.1549G>A (p.Ala517Thr)	Benign familial neonatal seizures	Likely benign(Last reviewed: Dec 21, 2017)	criteria provided, single submitter	VCV000538558	8	132140095	538558	523257	rs1554622834	LOF	0.817	0.1	0.084
KCNQ3	518	A	T	NM_004519.4(KCNQ3):c.1552G>A (p.Ala518Thr)	Benign familial neonatal seizures|not specified	Uncertain significance(Last reviewed: Aug 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000447643	8	132140092	447643	441170	rs745463637	LOF	0.82	0.102	0.079
KCNQ3	521	A	G	NM_004519.4(KCNQ3):c.1562C>G (p.Ala521Gly)	not specified	Uncertain significance(Last reviewed: Dec 7, 2016)	criteria provided, single submitter	VCV000373612	8	132140082	373612	359694	rs1057518505	LOF	0.837	0.09	0.072
KCNQ3	522	V	I	NM_004519.4(KCNQ3):c.1564G>A (p.Val522Ile)	not provided|Benign familial neonatal seizures| Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Nov 2, 2020)	criteria provided, conflicting interpretations	VCV000193878	8	132140080	193878	191041	rs143683496	LOF	0.848	0.064	0.088
KCNQ3	531	K	E	NM_004519.4(KCNQ3):c.1591A>G (p.Lys531Glu)	not provided|Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Sep 30, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205974	8	132137994	205974	202167	rs796052681	LOF	0.779	0.133	0.088
KCNQ3	538	Y	C	NM_004519.4(KCNQ3):c.1973A>G (p.Tyr658Cys)	Intellectual disability	Likely benign(Last reviewed: Jan 1, 2019)	no assertion criteria provided	VCV000975565	8	132129908	975565	963645	rs1423597114	LOF	0.804	0.108	0.088
KCNQ3	538	Y	N	NM_004519.4(KCNQ3):c.1972T>A (p.Tyr658Asn)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 30, 2020)	criteria provided, single submitter	VCV001040197	8	132129909	1040197	1028285	rs1824811259	LOF	0.842	0.08	0.078
KCNQ3	539	R	S	NM_004519.4(KCNQ3):c.1617G>C (p.Arg539Ser)	not specified	Uncertain significance(Last reviewed: Sep 19, 2016)	criteria provided, single submitter	VCV000447644	8	132137968	447644	441169	rs140607300	LOF	0.744	0.159	0.097
KCNQ3	552	K	E	NM_004519.4(KCNQ3):c.2014A>G (p.Lys672Glu)	Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Jan 12, 2018)	criteria provided, single submitter	VCV000909669	8	132129867	909669	898886	rs554613662	LOF	0.824	0.029	0.147
KCNQ3	555	N	S	NM_004519.4(KCNQ3):c.2024A>G (p.Asn675Ser)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 15, 2019)	criteria provided, single submitter	VCV000654846	8	132129857	654846	636588	rs1586750924	LOF	0.863	0.027	0.11
KCNQ3	556	R	G	NM_004519.4(KCNQ3):c.2026A>G (p.Arg676Gly)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 2, 2020)	criteria provided, single submitter	VCV001006098	8	132129855	1006098	992551	rs1307554348	LOF	0.805	0.045	0.15
KCNQ3	562	T	A	NM_004519.4(KCNQ3):c.2044A>G	Benign familial neonatal seizures	Uncertain	criteria	VCV000660237	8	132129837	660237	636587	rs768163501	LOF	0.842	0.026	0.132

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
				(p.Thr682Ala)		significance(Last reviewed: Oct 9, 2018)	provided, single submitter										
KCNQ3	565	Q	R	NM_004519.4(KCNQ3):c.1694A>G (p.Gln565Arg)	not provided	Uncertain significance(Last reviewed: Jul 17, 2017)	criteria provided, single submitter	VCV000450476	8	132137891	450476	444212	rs1003860988	LOF	0.837	0.023	0.139
KCNQ3	570	M	T	NM_004519.4(KCNQ3):c.1709T>C (p.Met570Thr)	not provided|Benign familial neonatal seizures 2	Uncertain significance(Last reviewed: Jun 1, 2021)	criteria provided, multiple submitters, no conflicts	VCV000205975	8	132134380	205975	202165	rs199999939	LOF	0.847	0.069	0.084
KCNQ3	573	P	L	NM_004519.4(KCNQ3):c.2078C>T (p.Pro693Leu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 24, 2020)	criteria provided, single submitter	VCV000937870	8	132129803	937870	934090	rs755177673	LOF	0.762	0.141	0.097
KCNQ3	574	P	S	NM_004519.4(KCNQ3):c.1720C>T (p.Pro574Ser)	Benign familial neonatal seizures 2|Seizures| Benign Neonatal Epilepsy|Intellectual disability| Benign familial neonatal seizures|not provided| not specified	Benign/Likely benign(Last reviewed: Dec 5, 2020)	criteria provided, multiple submitters, no conflicts	VCV000021411	8	132134369	21411	34263	rs74582884	Neutral	0.726	0.141	0.133
KCNQ3	574	P	T	NM_004519.4(KCNQ3):c.1720C>A (p.Pro574Thr)	not provided|Inborn genetic diseases|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 28, 2021)	criteria provided, multiple submitters, no conflicts	VCV000205976	8	132134369	205976	202163	rs74582884	Neutral	0.724	0.143	0.133
KCNQ3	578	S	N	NM_004519.4(KCNQ3):c.2093G>A (p.Ser698Asn)	not provided	Uncertain significance(Last reviewed: Apr 2, 2019)	criteria provided, single submitter	VCV001308484	8	132129788	1308484	1297105	NA	Neutral	0.626	0.234	0.14
KCNQ3	592	T	S	NM_004519.4(KCNQ3):c.1775C>G (p.Thr592Ser)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 28, 2019)	criteria provided, single submitter	VCV000471218	8	132134314	471218	457798	rs556421495	Neutral	0.574	0.29	0.136
KCNQ3	594	P	S	NM_004519.4(KCNQ3):c.1780C>T (p.Pro594Ser)	Benign familial neonatal seizures|not provided	Uncertain significance(Last reviewed: Aug 30, 2019)	criteria provided, multiple submitters, no conflicts	VCV000646535	8	132134309	646535	636590	rs868191966	Neutral	0.615	0.257	0.129
KCNQ3	595	S	P	NM_004519.4(KCNQ3):c.1783T>C (p.Ser595Pro)	not provided	Uncertain significance(Last reviewed: Jun 3, 2019)	criteria provided, single submitter	VCV000424010	8	132134306	424010	407311	rs1064796743	Neutral	0.576	0.269	0.154
KCNQ3	600	R	K	NM_004519.4(KCNQ3):c.1799G>A (p.Arg600Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 20, 2018)	criteria provided, single submitter	VCV000471219	8	132134290	471219	457187	rs1554622023	Neutral	0.61	0.261	0.129
KCNQ3	604	Y	N	NM_004519.4(KCNQ3):c.1810T>A (p.Tyr604Asn)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 16, 2018)	criteria provided, single submitter	VCV000405219	8	132132254	405219	395900	rs1060500606	Neutral	0.577	0.317	0.105
KCNQ3	608	G	A	NM_004519.4(KCNQ3):c.2183G>C (p.Gly728Ala)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Nov 26, 2019)	criteria provided, single submitter	VCV000860624	8	132129698	860624	834120	rs1824797413	Neutral	0.56	0.279	0.161
KCNQ3	615	D	G	NM_004519.4(KCNQ3):c.1844A>G (p.Asp615Gly)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 20, 2020)	criteria provided, single submitter	VCV000205977	8	132132220	205977	202162	rs777180732	Neutral	0.686	0.21	0.104
KCNQ3	617	S	T	NM_004519.4(KCNQ3):c.1850G>C (p.Ser617Thr)	not provided	Uncertain significance(Last reviewed: Jan 9,	criteria provided, single submitter	VCV000205979	8	132132214	205979	202161	rs758002609	LOF	0.776	0.094	0.13

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2013)											
KCNQ3	629	V	F	NM_004519.4(KCNQ3):c.1885G>T (p.Val629Phe)	not provided	Uncertain significance(Last reviewed: Nov 1, 2013)	criteria provided, single submitter	VCV000205980	8	132129996	205980	202159	rs185511111	LOF	0.857	0.068	0.074
KCNQ3	629	V	I	NM_004519.4(KCNQ3):c.1885G>A (p.Val629Ile)	Benign familial neonatal seizures|not provided	Conflicting interpretations of pathogenicity(Last reviewed: Jun 22, 2021)	criteria provided, conflicting interpretations	VCV000449145	8	132129996	449145	444211	rs185511111	LOF	0.849	0.073	0.078
KCNQ3	629	V	L	NM_004519.4(KCNQ3):c.1885G>C (p.Val629Leu)	Benign familial neonatal seizures|not provided| Benign familial neonatal seizures 2|not specified|Rolandic epilepsy	Uncertain significance(Last reviewed: Sep 18, 2020)	criteria provided, multiple submitters, no conflicts	VCV000206003	8	132129996	206003	202160	rs185511111	LOF	0.852	0.07	0.078
KCNQ3	630	I	V	NM_004519.4(KCNQ3):c.2248A>G (p.Ile750Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Apr 16, 2019)	criteria provided, single submitter	VCV000852113	8	132129633	852113	834119	rs776413259	LOF	0.808	0.1	0.092
KCNQ3	640	V	M	NM_004519.4(KCNQ3):c.1918G>A (p.Val640Met)	not provided|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 22, 2017)	criteria provided, multiple submitters, no conflicts	VCV000405217	8	132129963	405217	396288	rs767903815	LOF	0.866	0.055	0.079
KCNQ3	647	M	I	NM_004519.4(KCNQ3):c.1941G>A (p.Met647Ile)	Benign familial neonatal seizures 2|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 28, 2019)	criteria provided, multiple submitters, no conflicts	VCV000571968	8	132129940	571968	562352	rs1227129126	LOF	0.815	0.098	0.087
KCNQ3	647	M	L	NM_004519.4(KCNQ3):c.1939A>T (p.Met647Leu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 3, 2019)	criteria provided, single submitter	VCV000952842	8	132129942	952842	925015	rs1274926731	LOF	0.812	0.101	0.087
KCNQ3	649	R	W	NM_004519.4(KCNQ3):c.1945C>T (p.Arg649Trp)	not provided	Uncertain significance(Last reviewed: Jan 1, 2018)	criteria provided, single submitter	VCV000547040	8	132129936	547040	537529	rs770863845	LOF	0.733	0.149	0.118
KCNQ3	653	Q	R	NM_004519.4(KCNQ3):c.1958A>G (p.Gln653Arg)	Benign familial neonatal seizures|Benign familial neonatal seizures 2|Benign Neonatal Epilepsy|not specified|Seizures	Benign/Likely benign(Last reviewed: Nov 16, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205981	8	132129923	205981	202158	rs554833870	LOF	0.787	0.099	0.114
KCNQ3	654	I	V	NM_004519.4(KCNQ3):c.2320A>G (p.Ile774Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 14, 2018)	criteria provided, single submitter	VCV000663775	8	132129561	663775	636586	rs1284135753	LOF	0.821	0.096	0.084
KCNQ3	655	T	M	NM_004519.4(KCNQ3):c.1964C>T (p.Thr655Met)	Benign familial neonatal seizures|not provided	Likely benign(Last reviewed: Nov 10, 2020)	criteria provided, multiple submitters, no conflicts	VCV000378036	8	132129917	378036	371511	rs199942237	Neutral	0.704	0.189	0.107
KCNQ3	659	R	K	NM_004519.4(KCNQ3):c.2336G>A (p.Arg779Lys)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jul 7, 2020)	criteria provided, single submitter	VCV001017095	8	132129545	1017095	1007760	rs746006725	Neutral	0.619	0.233	0.148
KCNQ3	662	I	V	NM_004519.4(KCNQ3):c.2344A>G (p.Ile782Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Mar 6, 2020)	criteria provided, single submitter	VCV000863332	8	132129537	863332	834117	rs1391097567	Neutral	0.625	0.258	0.118
KCNQ3	665	S	L	NM_004519.4(KCNQ3):c.1994C>T (p.Ser665Leu)	Seizures|Benign familial neonatal seizures|not provided|Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Nov 13,	criteria provided, conflicting interpretations	VCV000194513	8	132129887	194513	191676	rs147173555	Neutral	0.56	0.307	0.133

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)											
KCNQ3	666	S	T	NM_004519.4(KCNQ3):c.2357G>C (p.Ser786Thr)	not provided	Uncertain significance(Last reviewed: Jan 23, 2020)	criteria provided, single submitter	VCV001311477	8	132129524	1311477	1299534	NA	Neutral	0.554	0.284	0.162
KCNQ3	669	A	T	NM_004519.4(KCNQ3):c.2005G>A (p.Ala669Thr)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 22, 2018)	criteria provided, single submitter	VCV000205982	8	132129876	205982	202157	rs201812160	Neutral	0.586	0.292	0.121
KCNQ3	679	D	N	NM_004519.4(KCNQ3):c.2035G>A (p.Asp679Asn)	not provided	Uncertain significance(Last reviewed: Jan 24, 2017)	criteria provided, single submitter	VCV000393167	8	132129846	393167	371509	rs773672399	Neutral	0.618	0.238	0.144
KCNQ3	680	L	V	NM_004519.4(KCNQ3):c.2038T>G (p.Leu680Val)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 1, 2018)	criteria provided, single submitter	VCV000478607	8	132129843	478607	458186	rs1223253841	Neutral	0.715	0.187	0.098
KCNQ3	686	N	S	NM_004519.4(KCNQ3):c.2057A>G (p.Asn686Ser)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Apr 3, 2019)	criteria provided, single submitter	VCV000837953	8	132129824	837953	834121	rs779744869	LOF	0.744	0.133	0.123
KCNQ3	687	Y	F	NM_004519.4(KCNQ3):c.2060A>T (p.Tyr687Phe)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 6, 2019)	criteria provided, single submitter	VCV000948423	8	132129821	948423	925014	rs757584140	Neutral	0.672	0.229	0.099
KCNQ3	691	G	D	NM_004519.4(KCNQ3):c.2072G>A (p.Gly691Asp)	Benign familial neonatal seizures|not provided	Uncertain significance(Last reviewed: Nov 14, 2019)	criteria provided, multiple submitters, no conflicts	VCV000570094	8	132129809	570094	561949	rs758790946	Neutral	0.653	0.209	0.138
KCNQ3	691	G	S	NM_004519.4(KCNQ3):c.2071G>A (p.Gly691Ser)	Benign familial neonatal seizures|not provided| Benign familial neonatal seizures 2	Benign/Likely benign(Last reviewed: Mar 20, 2020)	criteria provided, multiple submitters, no conflicts	VCV000205983	8	132129810	205983	202156	rs747379988	Neutral	0.642	0.205	0.153
KCNQ3	692	D	N	NM_004519.4(KCNQ3):c.2434G>A (p.Asp812Asn)	not provided	Uncertain significance(Last reviewed: Dec 16, 2020)	criteria provided, single submitter	VCV001205319	8	132129447	1205319	1194013	NA	Neutral	0.584	0.264	0.152
KCNQ3	692	P	A	NM_004519.4(KCNQ3):c.2074C>G (p.Pro692Ala)	Benign familial neonatal seizures 2|not provided	Uncertain significance(Last reviewed: Jul 2, 2018)	criteria provided, multiple submitters, no conflicts	VCV000429488	8	132129807	429488	421658	rs1131691408	Neutral	0.571	0.291	0.138
KCNQ3	695	P	L	NM_004519.4(KCNQ3):c.2084C>T (p.Pro695Leu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Feb 24, 2020)	criteria provided, single submitter	VCV000572531	8	132129797	572531	561947	rs760983146	Neutral	0.57	0.307	0.123
KCNQ3	696	P	T	NM_004519.4(KCNQ3):c.2086C>A (p.Pro696Thr)	Benign familial neonatal seizures|not provided	Uncertain significance(Last reviewed: Apr 14, 2020)	criteria provided, multiple submitters, no conflicts	VCV000810313	8	132129795	810313	796120	rs1055327554	Neutral	0.621	0.233	0.146
KCNQ3	708	S	I	NM_004519.4(KCNQ3):c.2123G>T (p.Ser708Ile)	not provided|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Jun 6, 2020)	criteria provided, conflicting interpretations	VCV000413257	8	132129758	413257	396570	rs977989588	Neutral	0.66	0.201	0.139
KCNQ3	710	Y	C	NM_004519.4(KCNQ3):c.2129A>G (p.Tyr710Cys)	Benign familial neonatal seizures 2|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Sep 17,	criteria provided, conflicting interpretations	VCV000405216	8	132129752	405216	396563	rs1060500605	Neutral	0.605	0.288	0.107

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2020)											
KCNQ3	710	Y	H	NM_004519.4(KCNQ3):c.2128T>C (p.Tyr710His)	Benign familial neonatal seizures|not provided	Likely benign(Last reviewed: Dec 31, 2019)	criteria provided, multiple submitters, no conflicts	VCV000205984	8	132129753	205984	202155	rs181746838	Neutral	0.598	0.286	0.117
KCNQ3	715	H	R	NM_004519.4(KCNQ3):c.2144A>G (p.His715Arg)	not provided|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Oct 21, 2020)	criteria provided, conflicting interpretations	VCV000205985	8	132129737	205985	202154	rs112314858	Neutral	0.61	0.27	0.121
KCNQ3	716	D	H	NM_004519.4(KCNQ3):c.2146G>C (p.Asp716His)	not provided|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Jul 16, 2020)	criteria provided, conflicting interpretations	VCV000205986	8	132129735	205986	202153	rs149324120	Neutral	0.624	0.234	0.142
KCNQ3	716	G	D	NM_004519.4(KCNQ3):c.2507G>A (p.Gly836Asp)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Dec 16, 2019)	criteria provided, single submitter	VCV000835624	8	132129374	835624	834115	rs894796810	Neutral	0.647	0.22	0.133
KCNQ3	722	R	Q	NM_004519.4(KCNQ3):c.2165G>A (p.Arg722Gln)	Benign familial neonatal seizures|Benign familial neonatal seizures 2|Benign Neonatal Epilepsy	Uncertain significance(Last reviewed: May 29, 2019)	criteria provided, multiple submitters, no conflicts	VCV000361874	8	132129716	361874	313013	rs377725346	Neutral	0.574	0.294	0.132
KCNQ3	723	G	E	NM_004519.4(KCNQ3):c.2168G>A (p.Gly723Glu)	Benign familial neonatal seizures|Seizures|not specified|Benign familial neonatal seizures 2	Benign(Last reviewed: Nov 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000138050	8	132129713	138050	141753	rs142149782	Neutral	0.598	0.274	0.128
KCNQ3	732	A	T	NM_004519.4(KCNQ3):c.2194G>A (p.Ala732Thr)	not provided	Uncertain significance(Last reviewed: Jan 28, 2014)	criteria provided, single submitter	VCV000205987	8	132129687	205987	202152	rs796052682	Neutral	0.589	0.299	0.112
KCNQ3	740	T	M	NM_004519.4(KCNQ3):c.2219C>T (p.Thr740Met)	Benign familial neonatal seizures 2|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Feb 24, 2020)	criteria provided, multiple submitters, no conflicts	VCV000579852	8	132129662	579852	564635	rs764065145	Neutral	0.573	0.309	0.119
KCNQ3	742	V	G	NM_004519.4(KCNQ3):c.2225T>G (p.Val742Gly)	not specified	Uncertain significance(Last reviewed: Dec 1, 2016)	criteria provided, single submitter	VCV000372609	8	132129656	372609	359887	rs1057517883	Neutral	0.649	0.259	0.092
KCNQ3	743	S	A	NM_004519.4(KCNQ3):c.2587T>G (p.Ser863Ala)	not provided	Uncertain significance(Last reviewed: Jun 10, 2019)	criteria provided, single submitter	VCV001306085	8	132129294	1306085	1297356	NA	Neutral	0.613	0.242	0.144
KCNQ3	744	V	I	NM_004519.4(KCNQ3):c.2590G>A (p.Val864Ile)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 8, 2020)	criteria provided, single submitter	VCV001058710	8	132129291	1058710	1045241	NA	Neutral	0.666	0.226	0.108
KCNQ3	746	T	M	NM_004519.4(KCNQ3):c.2237C>T (p.Thr746Met)	not provided|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Apr 7, 2021)	criteria provided, conflicting interpretations	VCV000430518	8	132129644	430518	421657	rs375833070	Neutral	0.645	0.224	0.131
KCNQ3	747	V	F	NM_004519.4(KCNQ3):c.2239G>T (p.Val747Phe)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 14, 2020)	criteria provided, single submitter	VCV000570729	8	132129642	570729	567365	rs1563761711	Neutral	0.693	0.205	0.101
KCNQ3	755	D	N	NM_004519.4(KCNQ3):c.2263G>A (p.Asp755Asn)	Benign familial neonatal seizures|not provided|Rolandic epilepsy|Seizures|Benign familial	Benign/Likely benign(Last reviewed:	criteria provided,	VCV000205988	8	132129618	205988	202151	rs150821246	Neutral	0.591	0.281	0.128

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					neonatal seizures 2	Jun 4, 2021)	multiple submitters, no conflicts										
KCNQ3	757	R	Q	NM_004519.4(KCNQ3):c.2270G>A (p.Arg757Gln)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 2, 2020)	criteria provided, single submitter	VCV000852618	8	132129611	852618	834118	rs769248820	Neutral	0.601	0.282	0.117
KCNQ3	765	D	E	NM_004519.4(KCNQ3):c.2295C>A (p.Asp765Glu)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 5, 2020)	criteria provided, single submitter	VCV000935235	8	132129586	935235	934089	rs201183533	Neutral	0.554	0.311	0.134
KCNQ3	769	P	H	NM_004519.4(KCNQ3):c.2306C>A (p.Pro769His)	Benign Neonatal Epilepsy|Benign familial neonatal seizures|not specified|Benign familial neonatal seizures 2|not provided|Seizures	Benign/Likely benign(Last reviewed: Dec 2, 2020)	criteria provided, multiple submitters, no conflicts	VCV000138051	8	132129575	138051	141754	rs114095081	Neutral	0.564	0.304	0.132
KCNQ3	773	R	Q	NM_004519.4(KCNQ3):c.2318G>A (p.Arg773Gln)	not provided|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Jun 7, 2021)	criteria provided, conflicting interpretations	VCV000205989	8	132129563	205989	202150	rs769160647	Neutral	0.554	0.312	0.134
KCNQ3	777	R	Q	NM_004519.4(KCNQ3):c.2330G>A (p.Arg777Gln)	Benign familial neonatal seizures|Seizures|not provided|Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Nov 22, 2020)	criteria provided, conflicting interpretations	VCV000205990	8	132129551	205990	202149	rs201328910	Neutral	0.553	0.31	0.137
KCNQ3	777	R	W	NM_004519.4(KCNQ3):c.2329C>T (p.Arg777Trp)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 12, 2018)	criteria provided, single submitter	VCV000647590	8	132129552	647590	636585	rs776128068	Neutral	0.582	0.317	0.101
KCNQ3	780	R	C	NM_004519.4(KCNQ3):c.2338C>T (p.Arg780Cys)	not provided|Benign familial neonatal seizures 2|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Jul 12, 2020)	criteria provided, conflicting interpretations	VCV000405218	8	132129543	405218	395898	rs138852641	Neutral	0.584	0.317	0.099
KCNQ3	780	R	H	NM_004519.4(KCNQ3):c.2339G>A (p.Arg780His)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 25, 2020)	criteria provided, single submitter	VCV000205991	8	132129542	205991	202148	rs747235713	Neutral	0.548	0.341	0.11
KCNQ3	784	R	Q	NM_004519.4(KCNQ3):c.2351G>A (p.Arg784Gln)	not provided	Likely benign(Last reviewed: Feb 28, 2019)	criteria provided, single submitter	VCV000205992	8	132129530	205992	202147	rs754896169	Neutral	0.567	0.293	0.14
KCNQ3	794	S	L	NM_004519.4(KCNQ3):c.2381C>T (p.Ser794Leu)	Seizures|Benign familial neonatal seizures	Uncertain significance(Last reviewed: May 30, 2020)	criteria provided, multiple submitters, no conflicts	VCV000077175	8	132129500	77175	88066	rs267601778	Neutral	0.638	0.247	0.115
KCNQ3	795	V	I	NM_004519.4(KCNQ3):c.2383G>A (p.Val795Ile)	not provided|Benign familial neonatal seizures	Uncertain significance(Last reviewed: Nov 10, 2017)	criteria provided, multiple submitters, no conflicts	VCV000205993	8	132129498	205993	202146	rs764544537	Neutral	0.676	0.231	0.093
KCNQ3	796	N	D	NM_004519.4(KCNQ3):c.2386A>G (p.Asn796Asp)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jun 4, 2020)	criteria provided, single submitter	VCV000644399	8	132129495	644399	636584	rs1289523204	Neutral	0.629	0.245	0.126
KCNQ3	798	E	A	NM_004519.4(KCNQ3):c.2393A>C (p.Glu798Ala)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jul 2, 2018)	criteria provided, single submitter	VCV000568547	8	132129488	568547	564632	rs1563761409	Neutral	0.595	0.281	0.124
KCNQ3	808	S	N	NM_004519.4(KCNQ3):c.2423G>A (p.Ser808Asn)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Jan 4,	criteria provided, single submitter	VCV000538553	8	132129458	538553	523623	rs1554621319	Neutral	0.652	0.22	0.128

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2018)											
KCNQ3	815	D	Y	NM_004519.4(KCNQ3):c.2443G>T (p.Asp815Tyr)	Benign familial neonatal seizures|not provided	Uncertain significance(Last reviewed: Nov 29, 2018)	criteria provided, multiple submitters, no conflicts	VCV000205994	8	132129438	205994	202145	rs530506549	Neutral	0.567	0.314	0.118
KCNQ3	819	G	S	NM_004519.4(KCNQ3):c.2455G>A (p.Gly819Ser)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Oct 25, 2018)	criteria provided, single submitter	VCV000471224	8	132129426	471224	457816	rs540574784	Neutral	0.6	0.281	0.119
KCNQ3	821	N	S	NM_004519.4(KCNQ3):c.2462A>G (p.Asn821Ser)	not provided|Benign familial neonatal seizures 2|Benign Neonatal Epilepsy|Seizures|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Mar 24, 2021)	criteria provided, conflicting interpretations	VCV000021412	8	132129419	21412	34264	rs118192254	Neutral	0.555	0.312	0.132
KCNQ3	823	G	R	NM_004519.4(KCNQ3):c.2467G>A (p.Gly823Arg)	Seizures	Uncertain significance(Last reviewed: Sep 5, 2017)	no assertion criteria provided	VCV000633484	8	132129414	633484	621909	rs1563761293	Neutral	0.645	0.221	0.134
KCNQ3	831	R	Q	NM_004519.4(KCNQ3):c.2492G>A (p.Arg831Gln)	Benign familial neonatal seizures 2|not specified|Benign familial neonatal seizures	Conflicting interpretations of pathogenicity(Last reviewed: Jan 12, 2018)	criteria provided, conflicting interpretations	VCV000211238	8	132129389	211238	207503	rs149004528	Neutral	0.587	0.299	0.114
KCNQ3	831	R	W	NM_004519.4(KCNQ3):c.2491C>T (p.Arg831Trp)	Benign familial neonatal seizures|not specified| not provided|Seizures	Conflicting interpretations of pathogenicity(Last reviewed: Oct 20, 2020)	criteria provided, conflicting interpretations	VCV000205954	8	132129390	205954	202144	rs185628977	Neutral	0.595	0.319	0.087
KCNQ3	835	E	D	NM_004519.4(KCNQ3):c.2505G>C (p.Glu835Asp)	Benign familial neonatal seizures	Uncertain significance(Last reviewed: Sep 2, 2020)	criteria provided, single submitter	VCV000538554	8	132129376	538554	523620	rs748320350	Neutral	0.643	0.24	0.117
KCNQ3	845	F	S	NM_004519.4(KCNQ3):c.2534T>C (p.Phe845Ser)	Benign familial neonatal seizures|not provided	Uncertain significance(Last reviewed: Oct 25, 2021)	criteria provided, multiple submitters, no conflicts	VCV000850290	8	132129347	850290	834114	rs751186425	Neutral	0.632	0.277	0.091
KCNQ3	846	T	M	NM_004519.4(KCNQ3):c.2537C>T (p.Thr846Met)	not provided	Uncertain significance(Last reviewed: Jun 2, 2017)	criteria provided, single submitter	VCV000432492	8	132129344	432492	425776	rs765623435	Neutral	0.584	0.31	0.107
KCNQ3	849	G	S	NM_004519.4(KCNQ3):c.2545G>A (p.Gly849Ser)	not provided	Uncertain significance(Last reviewed: Sep 6, 2017)	criteria provided, single submitter	VCV000449652	8	132129336	449652	444210	rs761201259	Neutral	0.593	0.267	0.14
KCNQ3	856	T	I	NM_004519.4(KCNQ3):c.2567C>T (p.Thr856Ile)	not specified	Uncertain significance(Last reviewed: Dec 12, 2016)	criteria provided, single submitter	VCV000373751	8	132129314	373751	359827	rs762078830	Neutral	0.612	0.278	0.111
KCNQ3	871	P	A	NM_004519.4(KCNQ3):c.2611C>G (p.Pro871Ala)	Benign familial neonatal seizures 2|Benign familial neonatal seizures|not specified	Uncertain significance(Last reviewed: Apr 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000372395	8	132129270	372395	359883	rs200647826	Neutral	0.582	0.297	0.121
KCNQ3	872	I	V	NM_004519.4(KCNQ3):c.2614A>G (p.Ile872Val)	not provided|Benign familial neonatal seizures| Benign familial neonatal seizures 2	Conflicting interpretations of pathogenicity(Last reviewed: Jul 21, 2020)	criteria provided, conflicting interpretations	VCV000205995	8	132129267	205995	202143	rs199682667	Neutral	0.642	0.258	0.1
KCNQ4	47	L	P	NM_004700.4(KCNQ4):c.140T>C (p.Leu47Pro)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Oct 20,	no assertion criteria provided	VCV000585006	1	40784233	585006	576062	rs1271250198	LOF	0.766	0.15	0.084

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2017)											
KCNQ4	50	S	N	NM_004700.4(KCNQ4):c.149G>A (p.Ser50Asn)	not specified	Likely benign(Last reviewed: Dec 6, 2017)	criteria provided, single submitter	VCV000513457	1	40784242	513457	498496	rs1553165192	Neutral	0.716	0.15	0.134
KCNQ4	52	L	Q	NM_004700.4(KCNQ4):c.155T>A (p.Leu52Gln)	not provided	Uncertain significance(Last reviewed: Apr 2, 2020)	criteria provided, single submitter	VCV001218070	1	40784248	1218070	1208052	NA	Neutral	0.731	0.174	0.095
KCNQ4	59	P	L	NM_004700.4(KCNQ4):c.176C>T (p.Pro59Leu)	not provided	Benign/Likely benign(Last reviewed: Apr 20, 2021)	criteria provided, multiple submitters, no conflicts	VCV000730845	1	40784269	730845	732448	rs775420649	Neutral	0.575	0.304	0.122
KCNQ4	92	E	K	NM_004700.4(KCNQ4):c.274G>A (p.Glu92Lys)	not provided	Uncertain significance(Last reviewed: Dec 3, 2019)	criteria provided, single submitter	VCV001310832	1	40784367	1310832	1301536	NA	LOF	0.84	0.032	0.128
KCNQ4	129	N	S	NM_004700.4(KCNQ4):c.386A>G (p.Asn129Ser)	not specified	Uncertain significance(Last reviewed: Jan 27, 2021)	criteria provided, single submitter	VCV000505394	1	40817336	505394	496204	rs761034165	LOF	0.9	0.017	0.082
KCNQ4	140	I	T	NM_004700.4(KCNQ4):c.419T>C (p.Ile140Thr)	Hearing impairment|not provided	Uncertain significance(Last reviewed: Apr 12, 2021)	criteria provided, single submitter	VCV001049775	1	40818177	1049775	1037030	NA	LOF	0.795	0.146	0.059
KCNQ4	182	F	L	NM_004700.4(KCNQ4):c.546C>G (p.Phe182Leu)	Autosomal dominant nonsyndromic hearing loss 2A|not provided	Benign/Likely benign(Last reviewed: Sep 23, 2020)	criteria provided, multiple submitters, no conflicts	VCV000021424	1	40818518	21424	34276	rs80358273	Neutral	0.494	0.408	0.098
KCNQ4	222	G	D	NM_004700.4(KCNQ4):c.665G>A (p.Gly222Asp)	not provided	Uncertain significance	no assertion criteria provided	VCV001297718	1	40818637	1297718	1287505	NA	LOF	0.877	0.037	0.086
KCNQ4	227	L	P	NM_004700.4(KCNQ4):c.680T>C (p.Leu227Pro)	not provided	Uncertain significance(Last reviewed: Oct 29, 2020)	criteria provided, single submitter	VCV001313583	1	40818652	1313583	1303844	NA	LOF	0.863	0.067	0.07
KCNQ4	228	G	C	NM_004700.4(KCNQ4):c.682G>T (p.Gly228Cys)	Autosomal dominant nonsyndromic hearing loss 2A|not specified	Uncertain significance(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000504603	1	40818654	504603	496165	rs367890569	LOF	0.835	0.08	0.084
KCNQ4	229	S	L	NM_004700.4(KCNQ4):c.686C>T (p.Ser229Leu)	not provided	Uncertain significance(Last reviewed: Sep 18, 2019)	criteria provided, single submitter	VCV001312296	1	40818658	1312296	1301678	NA	LOF	0.825	0.055	0.12
KCNQ4	230	V	E	NM_004700.4(KCNQ4):c.689T>A (p.Val230Glu)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000208365	1	40818661	208365	204594	rs797044965	LOF	0.855	0.054	0.091
KCNQ4	234	H	L	NM_004700.4(KCNQ4):c.701A>T (p.His234Leu)	Bilateral sensorineural hearing impairment	Pathogenic	criteria provided, single submitter	VCV001185583	1	40818673	1185583	1175544	NA	LOF	0.771	0.104	0.125
KCNQ4	234	H	R	NM_004700.4(KCNQ4):c.701A>G (p.His234Arg)	not provided	Likely pathogenic(Last reviewed: Feb 16, 2017)	criteria provided, single submitter	VCV000423333	1	40818673	423333	405169	rs1064796365	LOF	0.858	0.054	0.088
KCNQ4	251	F	L	NM_004700.4(KCNQ4):c.751T>C (p.Phe251Leu)	not specified	Uncertain significance(Last reviewed: Nov 14, 2018)	criteria provided, single submitter	VCV000666843	1	40819389	666843	654209	rs1000705787	Neutral	0.63	0.292	0.077
KCNQ4	260	E	K	NM_004700.4(KCNQ4):c.778G>A (p.Glu260Lys)	Autosomal dominant nonsyndromic hearing	Uncertain	criteria	VCV000021426	1	40819416	21426	34278	rs80358274	LOF	0.903	0.018	0.079

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					loss 2A|not provided	significance(Last reviewed: Jun 7, 2021)	provided, single submitter										
KCNQ4	262	D	G	NM_004700.4(KCNQ4):c.785A>G (p.Asp262Gly)	not provided	Uncertain significance(Last reviewed: Sep 30, 2019)	criteria provided, single submitter	VCV001308735	1	40819423	1308735	1300763	NA	LOF	0.846	0.041	0.113
KCNQ4	262	D	V	NM_004700.4(KCNQ4):c.785A>T (p.Asp262Val)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000021427	1	40819423	21427	34279	rs80358275	LOF	0.829	0.034	0.137
KCNQ4	266	D	Y	NM_004700.4(KCNQ4):c.796G>T (p.Asp266Tyr)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Dec 7, 2017)	no assertion criteria provided	VCV000591002	1	40819434	591002	581958	rs1558014576	LOF	0.856	0.045	0.099
KCNQ4	270	Y	H	NM_004700.4(KCNQ4):c.808T>C (p.Tyr270His)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000208367	1	40819446	208367	204596	rs797044967	LOF	0.889	0.04	0.072
KCNQ4	274	L	H	NM_004700.4(KCNQ4):c.821T>A (p.Leu274His)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000006247	1	40819459	6247	21286	rs80358276	LOF	0.91	0.034	0.057
KCNQ4	275	W	C	NM_004700.4(KCNQ4):c.825G>C (p.Trp275Cys)	not specified|Nonsyndromic hearing loss and deafness	Likely pathogenic(Last reviewed: Sep 10, 2018)	reviewed by expert panel	VCV000505302	1	40819463	505302	496205	rs956666801	LOF	0.802	0.113	0.085
KCNQ4	275	W	R	NM_004700.4(KCNQ4):c.823T>C (p.Trp275Arg)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000208368	1	40819461	208368	204597	rs797044968	LOF	0.881	0.052	0.067
KCNQ4	275	W	S	NM_004700.4(KCNQ4):c.824G>C (p.Trp275Ser)	not provided	Likely pathogenic(Last reviewed: Jun 10, 2016)	criteria provided, single submitter	VCV000372951	1	40819462	372951	359318	rs1057518095	LOF	0.849	0.066	0.084
KCNQ4	276	W	S	NM_004700.4(KCNQ4):c.827G>C (p.Trp276Ser)	not provided|Autosomal dominant nonsyndromic hearing loss 2A|Rare genetic deafness	Pathogenic(Last reviewed: Jun 24, 2014)	criteria provided, single submitter	VCV000006242	1	40819465	6242	21281	rs80358277	LOF	0.867	0.056	0.077
KCNQ4	277	G	R	NM_004700.4(KCNQ4):c.829G>A (p.Gly277Arg)	Autosomal dominant nonsyndromic hearing loss 2A|not specified	Uncertain significance(Last reviewed: Oct 31, 2018)	criteria provided, multiple submitters, no conflicts	VCV000178690	1	40819467	178690	172446	rs727504459	LOF	0.925	0.021	0.054
KCNQ4	278	T	A	NM_004700.4(KCNQ4):c.832A>G (p.Thr278Ala)	not specified	Uncertain significance(Last reviewed: Jul 11, 2017)	criteria provided, single submitter	VCV000517414	1	40819470	517414	496662	rs763326539	LOF	0.877	0.026	0.098
KCNQ4	280	T	K	NM_004700.4(KCNQ4):c.839C>A (p.Thr280Lys)	not provided	Likely pathogenic(Last reviewed: Jan 20, 2022)	criteria provided, single submitter	VCV001201725	1	40819879	1201725	1189636	NA	LOF	0.92	0.019	0.061
KCNQ4	281	L	S	NM_004700.4(KCNQ4):c.842T>C (p.Leu281Ser)	Autosomal dominant nonsyndromic hearing loss 2A|not provided	Pathogenic(Last reviewed: Jan 17, 2020)	criteria provided, multiple submitters, no conflicts	VCV000006246	1	40819882	6246	21285	rs80358278	LOF	0.9	0.035	0.065
KCNQ4	284	I	N	NM_004700.4(KCNQ4):c.851T>A (p.Ile284Asn)	not provided	Uncertain significance(Last reviewed: Sep 19, 2017)	criteria provided, single submitter	VCV000593866	1	40819891	593866	584929	rs1558015500	LOF	0.882	0.06	0.058
KCNQ4	285	G	C	NM_004700.4(KCNQ4):c.853G>T (p.Gly285Cys)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Feb 17, 2011)	no assertion criteria provided	VCV000006244	1	40819893	6244	21283	rs28937588	LOF	0.867	0.05	0.083
KCNQ4	285	G	S	NM_004700.4(KCNQ4):c.853G>A (p.Gly285Ser)	Rare genetic deafness|Nonsyndromic hearing loss and deafness|Autosomal dominant	Pathogenic(Last reviewed: Sep 11,	reviewed by expert panel	VCV000006241	1	40819893	6241	21280	rs28937588	LOF	0.885	0.038	0.077

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
					nonsyndromic hearing loss 2A	2018)											
KCNQ4	286	Y	C	NM_004700.4(KCNQ4):c.857A>G (p.Tyr286Cys)	not specified	Uncertain significance(Last reviewed: Mar 3, 2016)	criteria provided, single submitter	VCV000228776	1	40819897	228776	228471	rs876657841	LOF	0.84	0.089	0.071
KCNQ4	286	Y	S	NM_004700.4(KCNQ4):c.857A>C (p.Tyr286Ser)	Autosomal dominant nonsyndromic hearing loss 2A	Likely pathogenic	no assertion criteria provided	VCV001184480	1	40819897	1184480	1174252	NA	LOF	0.864	0.071	0.065
KCNQ4	287	G	R	NM_004700.4(KCNQ4):c.859G>C (p.Gly287Arg)	Autosomal dominant nonsyndromic hearing loss 2A|not provided	Pathogenic(Last reviewed: Jun 20, 2013)	no assertion criteria provided	VCV000156337	1	40819899	156337	166117	rs137853969	LOF	0.883	0.041	0.076
KCNQ4	287	G	S	NM_004700.4(KCNQ4):c.859G>A (p.Gly287Ser)	Autosomal dominant nonsyndromic hearing loss 2A	Likely pathogenic(Last reviewed: Aug 3, 2020)	criteria provided, single submitter	VCV000979187	1	40819899	979187	967260	rs137853969	LOF	0.891	0.036	0.073
KCNQ4	287	G	V	NM_004700.4(KCNQ4):c.860G>T (p.Gly287Val)	not provided	Uncertain significance(Last reviewed: Jun 29, 2020)	criteria provided, single submitter	VCV001304285	1	40819900	1304285	1294560	NA	LOF	0.887	0.041	0.073
KCNQ4	288	D	N	NM_004700.4(KCNQ4):c.862G>A (p.Asp288Asn)	not provided	Uncertain significance(Last reviewed: Sep 19, 2019)	criteria provided, single submitter	VCV001304257	1	40819902	1304257	1294532	NA	LOF	0.876	0.052	0.072
KCNQ4	291	P	L	NM_004700.4(KCNQ4):c.872C>T (p.Pro291Leu)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000208370	1	40819912	208370	204599	rs797044970	LOF	0.869	0.033	0.099
KCNQ4	291	P	S	NM_004700.4(KCNQ4):c.871C>T (p.Pro291Ser)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000208369	1	40819911	208369	204598	rs797044969	LOF	0.891	0.019	0.09
KCNQ4	296	G	S	NM_004700.4(KCNQ4):c.886G>A (p.Gly296Ser)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000006249	1	40819926	6249	21288	rs80358279	LOF	0.923	0.02	0.057
KCNQ4	297	R	S	NM_004700.4(KCNQ4):c.891G>T (p.Arg297Ser)	not provided|Autosomal dominant nonsyndromic hearing loss 2A	Uncertain significance(Last reviewed: Oct 16, 2020)	criteria provided, single submitter	VCV000208371	1	40819931	208371	204600	rs797044971	LOF	0.814	0.032	0.153
KCNQ4	321	G	S	NM_004700.4(KCNQ4):c.961G>A (p.Gly321Ser)	Autosomal dominant nonsyndromic hearing loss 2A|Rare genetic deafness	Likely pathogenic(Last reviewed: Feb 19, 2020)	criteria provided, single submitter	VCV000006243	1	40820180	6243	21282	rs28939710	LOF	0.914	0.021	0.065
KCNQ4	338	R	W	NM_004700.4(KCNQ4):c.1012C>T (p.Arg338Trp)	not provided	Uncertain significance(Last reviewed: Apr 23, 2019)	criteria provided, single submitter	VCV001202991	1	40820231	1202991	1192869	NA	LOF	0.802	0.098	0.1
KCNQ4	339	R	S	NM_004700.4(KCNQ4):c.1017G>T (p.Arg339Ser)	not specified	Uncertain significance(Last reviewed: Jun 12, 2017)	criteria provided, single submitter	VCV000505853	1	40820236	505853	496663	rs769086004	LOF	0.805	0.048	0.147
KCNQ4	351	R	C	NM_004700.4(KCNQ4):c.1051C>T (p.Arg351Cys)	not specified	Uncertain significance(Last reviewed: Feb 11, 2020)	criteria provided, single submitter	VCV000930077	1	40822323	930077	918202	rs1167593525	LOF	0.774	0.086	0.139
KCNQ4	386	V	M	NM_004700.4(KCNQ4):c.1156G>A (p.Val386Met)	not provided	Uncertain significance(Last reviewed: Jun 30, 2021)	criteria provided, single submitter	VCV001331134	1	40824122	1331134	1321809	NA	Neutral	0.733	0.197	0.071
KCNQ4	388	R	W	NM_004700.4(KCNQ4):c.1162C>T (p.Arg388Trp)	not specified	Uncertain significance(Last reviewed: Aug 19, 2014)	criteria provided, single submitter	VCV000163751	1	40824128	163751	172447	rs371079509	Neutral	0.703	0.203	0.093

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ4	398	E	G	NM_004700.4(KCNQ4):c.1193A>G (p.Glu398Gly)	not specified	Uncertain significance(Last reviewed: Apr 5, 2016)	criteria provided, single submitter	VCV000228769	1	40824159	228769	228475	rs773975779	Neutral	0.584	0.309	0.107
KCNQ4	400	R	W	NM_004700.4(KCNQ4):c.1198C>T (p.Arg400Trp)	not provided	Uncertain significance(Last reviewed: Jul 23, 2018)	criteria provided, single submitter	VCV000651900	1	40824164	651900	627832	rs1239252987	Neutral	0.53	0.378	0.092
KCNQ4	401	R	W	NM_004700.4(KCNQ4):c.1201C>T (p.Arg401Trp)	not specified	Uncertain significance(Last reviewed: Mar 5, 2015)	criteria provided, single submitter	VCV000228767	1	40824167	228767	228476	rs754165873	Neutral	0.522	0.389	0.089
KCNQ4	403	P	L	NM_004700.4(KCNQ4):c.1208C>T (p.Pro403Leu)	not provided	Uncertain significance(Last reviewed: Jan 21, 2020)	criteria provided, single submitter	VCV001187613	1	40824174	1187613	1175905	NA	Neutral	0.58	0.306	0.114
KCNQ4	406	D	N	NM_004700.4(KCNQ4):c.1216G>A (p.Asp406Asn)	not specified	Uncertain significance(Last reviewed: Aug 10, 2017)	criteria provided, single submitter	VCV000517519	1	40824182	517519	496626	rs1264123218	Neutral	0.66	0.249	0.091
KCNQ4	411	R	C	NM_004700.4(KCNQ4):c.1231C>T (p.Arg411Cys)	not specified|not provided	Benign/Likely benign(Last reviewed: Jun 14, 2021)	criteria provided, multiple submitters, no conflicts	VCV000666707	1	40824197	666707	654211	rs199809248	Neutral	0.524	0.382	0.094
KCNQ4	420	R	Q	NM_004700.4(KCNQ4):c.1259G>A (p.Arg420Gln)	not provided	Uncertain significance(Last reviewed: Sep 19, 2019)	criteria provided, multiple submitters, no conflicts	VCV000593390	1	40824225	593390	584454	rs374078257	Neutral	0.554	0.312	0.134
KCNQ4	423	S	G	NM_004700.4(KCNQ4):c.1267A>G (p.Ser423Gly)	not provided	Uncertain significance(Last reviewed: May 24, 2021)	criteria provided, single submitter	VCV001199646	1	40824233	1199646	1189637	NA	Neutral	0.509	0.395	0.096
KCNQ4	423	S	R	NM_004700.4(KCNQ4):c.1267A>C (p.Ser423Arg)	not specified	Uncertain significance(Last reviewed: Apr 12, 2014)	criteria provided, single submitter	VCV000163753	1	40824233	163753	172448	rs368294870	Neutral	0.529	0.371	0.1
KCNQ4	428	P	L	NM_004700.4(KCNQ4):c.1283C>T (p.Pro428Leu)	not specified	Uncertain significance(Last reviewed: Mar 11, 2020)	criteria provided, single submitter	VCV000930078	1	40824249	930078	918203	rs932108929	Neutral	0.481	0.435	0.084
KCNQ4	433	R	W	NM_004700.4(KCNQ4):c.1297C>T (p.Arg433Trp)	not provided	Uncertain significance(Last reviewed: Jul 13, 2017)	criteria provided, single submitter	VCV000450535	1	40831088	450535	442839	rs760023398	Neutral	0.608	0.306	0.087
KCNQ4	434	R	H	NM_004700.4(KCNQ4):c.1463G>A (p.Arg488His)	not provided	Uncertain significance(Last reviewed: Sep 29, 2020)	criteria provided, single submitter	VCV001201537	1	40831254	1201537	1189638	NA	Neutral	0.507	0.397	0.096
KCNQ4	442	M	T	NM_004700.4(KCNQ4):c.1325T>C (p.Met442Thr)	not specified	Benign(Last reviewed: Oct 8, 2019)	criteria provided, single submitter	VCV000163754	1	40831116	163754	172587	rs142453905	Neutral	0.452	0.45	0.098
KCNQ4	446	R	H	NM_004700.4(KCNQ4):c.1499G>A (p.Arg500His)	not provided	Uncertain significance(Last reviewed: Jan 23, 2019)	criteria provided, single submitter	VCV001304466	1	40831290	1304466	1294741	NA	Neutral	0.542	0.347	0.11
KCNQ4	447	T	I	NM_004700.4(KCNQ4):c.1502C>T (p.Thr501Ile)	Autosomal dominant nonsyndromic hearing loss 2A	not provided	no assertion provided	VCV001339804	1	40831293	1339804	1331014	NA	Neutral	0.476	0.425	0.099
KCNQ4	455	H	Q	NM_004700.4(KCNQ4):c.1365T>G (p.His455Gln)	not specified|Autosomal dominant nonsyndromic hearing loss 2A	Benign(Last reviewed: Nov 30,	criteria provided,	VCV000045101	1	40831156	45101	54268	rs34287852	Neutral	0.484	0.403	0.113

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
						2021)	multiple submitters, no conflicts										
KCNQ4	458	P	T	NM_004700.4(KCNQ4):c.1372C>A (p.Pro458Thr)	not provided	Uncertain significance(Last reviewed: May 2, 2017)	criteria provided, single submitter	VCV000429247	1	40831163	429247	421242	rs1010168019	Neutral	0.466	0.419	0.116
KCNQ4	469	Q	R	NM_004700.4(KCNQ4):c.1406A>G (p.Gln469Arg)	not specified	Uncertain significance(Last reviewed: Sep 14, 2017)	criteria provided, single submitter	VCV000506132	1	40831197	506132	496166	rs1553168273	Neutral	0.522	0.363	0.116
KCNQ4	473	M	L	NM_004700.4(KCNQ4):c.1579A>T (p.Met527Leu)	Inborn genetic diseases|not provided	Uncertain significance(Last reviewed: Jan 2, 2018)	criteria provided, single submitter	VCV000985992	1	40833079	985992	973209	rs759364617	Neutral	0.475	0.432	0.093
KCNQ4	492	R	Q	NM_004700.4(KCNQ4):c.1475G>A (p.Arg492Gln)	not provided	Uncertain significance(Last reviewed: Jul 24, 2015)	criteria provided, single submitter	VCV000282147	1	40831266	282147	266384	rs775830606	Neutral	0.552	0.328	0.119
KCNQ4	501	P	L	NM_004700.4(KCNQ4):c.1664C>T (p.Pro555Leu)	not provided	Uncertain significance	no assertion criteria provided	VCV001050307	1	40835017	1050307	1037031	NA	Neutral	0.501	0.4	0.099
KCNQ4	506	A	V	NM_004700.4(KCNQ4):c.1517C>T (p.Ala506Val)	not specified	Uncertain significance(Last reviewed: Aug 10, 2017)	criteria provided, single submitter	VCV000504605	1	40833017	504605	496167	rs145732892	Neutral	0.465	0.44	0.095
KCNQ4	511	V	I	NM_004700.4(KCNQ4):c.1531G>A (p.Val511Ile)	not specified	Uncertain significance(Last reviewed: Jan 15, 2016)	criteria provided, single submitter	VCV000228770	1	40833031	228770	228479	rs778538229	Neutral	0.492	0.429	0.079
KCNQ4	522	T	M	NM_004700.4(KCNQ4):c.1565C>T (p.Thr522Met)	Autosomal dominant nonsyndromic hearing loss 2A	Uncertain significance(Last reviewed: May 25, 2018)	criteria provided, single submitter	VCV001032259	1	40833065	1032259	1019377	rs748693577	Neutral	0.658	0.199	0.142
KCNQ4	537	I	M	NM_004700.4(KCNQ4):c.1611C>G (p.Ile537Met)	not specified	Uncertain significance(Last reviewed: Aug 21, 2015)	criteria provided, single submitter	VCV000228771	1	40833111	228771	228480	rs876657837	LOF	0.816	0.122	0.062
KCNQ4	569	E	Q	NM_004700.4(KCNQ4):c.1867G>C (p.Glu623Gln)	Inborn genetic diseases	Uncertain significance(Last reviewed: Mar 15, 2017)	criteria provided, single submitter	VCV000985100	1	40837786	985100	973210	rs1648848951	LOF	0.888	0.045	0.067
KCNQ4	601	D	Y	NM_004700.4(KCNQ4):c.1801G>T (p.Asp601Tyr)	not specified	Uncertain significance(Last reviewed: Jul 5, 2015)	criteria provided, single submitter	VCV000228772	1	40837720	228772	228482	rs876657838	Neutral	0.542	0.364	0.094
KCNQ4	606	D	E	NM_004700.4(KCNQ4):c.1818C>G (p.Asp606Glu)	not provided|not specified	Benign(Last reviewed: Dec 31, 2019)	criteria provided, multiple submitters, no conflicts	VCV000163756	1	40837737	163756	172588	rs139835231	Neutral	0.515	0.373	0.112
KCNQ4	652	L	V	NM_004700.4(KCNQ4):c.1954C>G (p.Leu652Val)	not specified	Uncertain significance(Last reviewed: Jun 19, 2014)	criteria provided, single submitter	VCV000179794	1	40838389	179794	172450	rs727505133	Neutral	0.499	0.411	0.09
KCNQ4	653	G	S	NM_004700.4(KCNQ4):c.1957G>A (p.Gly653Ser)	not specified	Uncertain significance(Last reviewed: Jan 27, 2016)	criteria provided, single submitter	VCV000228773	1	40838392	228773	228483	rs876657839	Neutral	0.644	0.242	0.114
KCNQ4	669	H	R	NM_004700.4(KCNQ4):c.2006A>G (p.His669Arg)	not specified	Uncertain significance(Last reviewed: Sep 8, 2015)	criteria provided, single submitter	VCV000228775	1	40838441	228775	228485	rs749565877	Neutral	0.522	0.376	0.102

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ4	680	S	F	NM_004700.4(KCNQ4):c.2039C>T (p.Ser680Phe)	Autosomal dominant nonsyndromic hearing loss 2A	Pathogenic(Last reviewed: Aug 20, 2015)	no assertion criteria provided	VCV000208373	1	40838474	208373	204602	rs772135867	Neutral	0.486	0.404	0.11
KCNQ5	3	R	S	NM_019842.4(KCNQ5):c.7C>A (p.Arg3Ser)	not provided|Intellectual disability|Intellectual disability, autosomal dominant 46	Conflicting interpretations of pathogenicity(Last reviewed: Dec 1, 2021)	criteria provided, conflicting interpretations	VCV000932587	6	72622196	932587	920763	rs549110435	LOF	0.678	0.123	0.199
KCNQ5	5	H	Q	NM_019842.4(KCNQ5):c.15C>G (p.His5Gln)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Nov 3, 2021)	criteria provided, single submitter	VCV001319294	6	72622204	1319294	1309547	NA	GOF	0.665	0.114	0.221
KCNQ5	8	G	R	NM_019842.4(KCNQ5):c.22G>A (p.Gly8Arg)	Intellectual disability	Likely benign(Last reviewed: Jan 1, 2019)	no assertion criteria provided	VCV000975566	6	72622211	975566	963615	rs191331629	GOF	0.645	0.094	0.261
KCNQ5	17	W	R	NM_019842.4(KCNQ5):c.49T>C (p.Trp17Arg)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Jun 21, 2019)	criteria provided, single submitter	VCV000932094	6	72622238	932094	919051	rs971617867	LOF	0.779	0.104	0.116
KCNQ5	29	G	W	NM_019842.4(KCNQ5):c.85G>T (p.Gly29Trp)	not provided	Uncertain significance(Last reviewed: Nov 1, 2021)	criteria provided, single submitter	VCV001335629	6	72622274	1335629	1326600	NA	LOF	0.7	0.123	0.177
KCNQ5	39	V	A	NM_019842.4(KCNQ5):c.116T>C (p.Val39Ala)	not provided	Uncertain significance(Last reviewed: Nov 20, 2019)	criteria provided, single submitter	VCV001310055	6	72622305	1310055	1301906	NA	LOF	0.754	0.099	0.147
KCNQ5	80	R	W	NM_019842.4(KCNQ5):c.238C>T (p.Arg80Trp)	not provided	Likely benign(Last reviewed: Nov 1, 2020)	criteria provided, single submitter	VCV001013510	6	72622427	1013510	1000544	rs750955277	LOF	0.675	0.128	0.197
KCNQ5	98	S	G	NM_019842.4(KCNQ5):c.292A>G (p.Ser98Gly)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Feb 24, 2021)	criteria provided, single submitter	VCV001342329	6	72622481	1342329	1333905	NA	LOF	0.721	0.085	0.194
KCNQ5	109	R	L	NM_019842.4(KCNQ5):c.326G>T (p.Arg109Leu)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Oct 25, 2021)	criteria provided, single submitter	VCV001321257	6	72622515	1321257	1311512	NA	LOF	0.756	0.052	0.192
KCNQ5	145	V	G	NM_019842.4(KCNQ5):c.434T>G (p.Val145Gly)	Intellectual disability, autosomal dominant 46	Pathogenic(Last reviewed: Aug 3, 2017)	no assertion criteria provided	VCV000431385	6	73003943	431385	424908	rs1135401955	LOF	0.871	0.032	0.097
KCNQ5	161	L	S	NM_019842.4(KCNQ5):c.482T>C (p.Leu161Ser)	not provided	Uncertain significance(Last reviewed: May 1, 2020)	criteria provided, single submitter	VCV000932588	6	73003991	932588	920764	rs1342144805	LOF	0.877	0.031	0.091
KCNQ5	170	V	I	NM_019842.4(KCNQ5):c.508G>A (p.Val170Ile)	Intellectual disability	Uncertain significance(Last reviewed: Apr 20, 2020)	criteria provided, single submitter	VCV000978947	6	73041954	978947	966954	rs771263285	LOF	0.859	0.045	0.095
KCNQ5	174	E	D	NM_019842.4(KCNQ5):c.522G>C (p.Glu174Asp)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Apr 17, 2019)	criteria provided, single submitter	VCV000931402	6	73041968	931402	919052	rs1771727703	LOF	0.838	0.035	0.127
KCNQ5	187	R	Q	NM_019842.4(KCNQ5):c.560G>A (p.Arg187Gln)	not provided	Pathogenic(Last reviewed: Jan 12, 2022)	criteria provided, single submitter	VCV001334356	6	73042006	1334356	1325228	NA	GOF	0.703	0.045	0.252
KCNQ5	256	G	A	NM_019842.4(KCNQ5):c.767G>C (p.Gly256Ala)	not provided	Uncertain significance(Last reviewed: Sep 9, 2021)	criteria provided, single submitter	VCV001253854	6	73077472	1253854	1243791	NA	LOF	0.832	0.03	0.137

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ5	292	N	S	NM_019842.4(KCNQ5):c.875A>G (p.Asn292Ser)	Intellectual disability, autosomal dominant 46	Likely pathogenic(Last reviewed: Mar 18, 2020)	no assertion criteria provided	VCV000988749	6	73077844	988749	976653	NA	GOF	0.696	0.055	0.249
KCNQ5	341	L	I	NM_019842.4(KCNQ5):c.1021C>A (p.Leu341Ile)	Intellectual disability, autosomal dominant 46	Pathogenic(Last reviewed: Aug 3, 2017)	no assertion criteria provided	VCV000431386	6	73105359	431386	424909	rs1135401956	LOF	0.857	0.046	0.097
KCNQ5	347	G	A	NM_019842.4(KCNQ5):c.1040G>C (p.Gly347Ala)	Intellectual disability, autosomal dominant 46	Likely pathogenic(Last reviewed: Jan 1, 2019)	no assertion criteria provided	VCV000975567	6	73111318	975567	963616	rs1775232605	LOF	0.817	0.023	0.16
KCNQ5	347	G	S	NM_019842.4(KCNQ5):c.1039G>A (p.Gly347Ser)	Global developmental delay|Inborn genetic diseases	Likely pathogenic(Last reviewed: Nov 1, 2019)	criteria provided, multiple submitters, no conflicts	VCV000521883	6	73111317	521883	511686	rs1554210415	LOF	0.797	0.022	0.181
KCNQ5	369	P	L	NM_019842.4(KCNQ5):c.1106C>T (p.Pro369Leu)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Nov 17, 2017)	criteria provided, single submitter	VCV000976198	6	73111384	976198	964281	rs1135401958	GOF	0.639	0.044	0.317
KCNQ5	369	P	Q	NM_019842.4(KCNQ5):c.1106C>A (p.Pro369Gln)	Intellectual disability, autosomal dominant 46	Pathogenic(Last reviewed: May 28, 2019)	criteria provided, single submitter	VCV000802242	6	73111384	802242	790657	rs1135401958	GOF	0.536	0.041	0.424
KCNQ5	369	P	R	NM_019842.4(KCNQ5):c.1106C>G (p.Pro369Arg)	Intellectual disability, autosomal dominant 46	Pathogenic(Last reviewed: Aug 3, 2017)	no assertion criteria provided	VCV000431388	6	73111384	431388	424911	rs1135401958	GOF	0.478	0.043	0.479
KCNQ5	369	P	S	NM_019842.4(KCNQ5):c.1105C>T (p.Pro369Ser)	not provided	Pathogenic(Last reviewed: Dec 22, 2017)	criteria provided, single submitter	VCV001072107	6	73111383	1072107	1060844	NA	GOF	0.603	0.04	0.356
KCNQ5	369	P	T	NM_019842.4(KCNQ5):c.1105C>A (p.Pro369Thr)	Inborn genetic diseases	Likely pathogenic(Last reviewed: Dec 14, 2017)	criteria provided, single submitter	VCV000521362	6	73111383	521362	511687	rs1554210418	GOF	0.599	0.041	0.36
KCNQ5	408	Q	K	NM_019842.4(KCNQ5):c.1249C>A (p.Gln417Lys)	not provided	Uncertain significance(Last reviewed: Apr 19, 2021)	criteria provided, single submitter	VCV001314753	6	73133422	1314753	1305014	NA	GOF	0.616	0.064	0.32
KCNQ5	422	R	W	NM_019842.4(KCNQ5):c.1291A>T (p.Arg431Trp)	Intellectual disability, autosomal dominant 46	Likely pathogenic(Last reviewed: Apr 3, 2020)	no assertion criteria provided	VCV000984630	6	73133464	984630	972719	rs1776322078	GOF	0.653	0.099	0.248
KCNQ5	429	R	G	NM_019842.4(KCNQ5):c.1312C>G (p.Arg438Gly)	Intellectual disability, mild	Uncertain significance(Last reviewed: Nov 14, 2019)	no assertion criteria provided	VCV000977441	6	73133485	977441	965500	rs750114221	LOF	0.717	0.085	0.197
KCNQ5	448	S	I	NM_019842.4(KCNQ5):c.1286G>T (p.Ser429Ile)	not provided|Intellectual disability, autosomal dominant 46	Pathogenic(Last reviewed: Dec 13, 2017)	criteria provided, single submitter	VCV000431387	6	73133459	431387	424910	rs1135401957	GOF	0.47	0.057	0.473
KCNQ5	485	E	V	NM_019842.4(KCNQ5):c.1784A>T (p.Glu595Val)	not provided	Uncertain significance(Last reviewed: Nov 12, 2020)	criteria provided, single submitter	VCV001305324	6	73192639	1305324	1295599	NA	GOF	0.558	0.067	0.375
KCNQ5	493	L	F	NM_019842.4(KCNQ5):c.1807C>T (p.Leu603Phe)	not provided	Uncertain significance(Last reviewed: Aug 5, 2019)	criteria provided, single submitter	VCV001307615	6	73192662	1307615	1298885	NA	LOF	0.724	0.061	0.215
KCNQ5	501	D	N	NM_019842.4(KCNQ5):c.1501G>A (p.Asp501Asn)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Apr 27, 2019)	criteria provided, single submitter	VCV000638520	6	73169778	638520	626166	rs1582480404	GOF	0.612	0.057	0.331

gene	pos	aa1	aa2	Name	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession	GRCh38Chromosome	GRCh38Location	VariationID	AlleleID(s)	dbSNP ID	prediction	prob.LOF	prob.Neutral	prob.GOF
KCNQ5	518	Q	H	NM_019842.4(KCNQ5):c.1884A>T (p.Gln628His)	not provided	Uncertain significance(Last reviewed: Jul 20, 2019)	criteria provided, single submitter	VCV001306814	6	73194499	1306814	1295764	NA	LOF	0.731	0.038	0.231
KCNQ5	526	A	P	NM_019842.4(KCNQ5):c.1906G>C (p.Ala636Pro)	not provided	Uncertain significance(Last reviewed: Aug 1, 2020)	criteria provided, single submitter	VCV001013513	6	73194521	1013513	1000547	rs774223061	LOF	0.79	0.033	0.176
KCNQ5	675	V	A	NM_019842.4(KCNQ5):c.2354T>C (p.Val785Ala)	Seizures	Uncertain significance(Last reviewed: Jan 6, 2020)	no assertion criteria provided	VCV000977399	6	73194969	977399	965501	rs747610703	LOF	0.816	0.044	0.14
KCNQ5	692	R	H	NM_019842.4(KCNQ5):c.2405G>A (p.Arg802His)	not provided	Likely benign	no assertion criteria provided	VCV001299845	6	73195020	1299845	1289897	NA	LOF	0.761	0.059	0.18
KCNQ5	704	A	T	NM_019842.4(KCNQ5):c.2110G>A (p.Ala704Thr)	Malignant tumor of prostate	Uncertain significance	no assertion criteria provided	VCV000161727	6	73194725	161727	171425	rs148543637	LOF	0.725	0.044	0.231
KCNQ5	726	I	N	NM_019842.4(KCNQ5):c.2507T>A (p.Ile836Asn)	not provided	Uncertain significance(Last reviewed: Aug 1, 2021)	criteria provided, single submitter	VCV001299035	6	73195122	1299035	1288897	NA	LOF	0.841	0.057	0.101
KCNQ5	780	E	V	NM_019842.4(KCNQ5):c.2366A>T (p.Glu789Val)	Intellectual disability, autosomal dominant 46| not provided	Uncertain significance(Last reviewed: May 14, 2021)	criteria provided, multiple submitters, no conflicts	VCV000809966	6	73194981	809966	795893	rs771383058	LOF	0.765	0.069	0.166
KCNQ5	799	R	Q	NM_019842.4(KCNQ5):c.2726G>A (p.Arg909Gln)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Jan 29, 2021)	criteria provided, single submitter	VCV001341834	6	73195341	1341834	1333081	NA	LOF	0.734	0.06	0.206
KCNQ5	817	L	F	NM_019842.4(KCNQ5):c.2478G>C (p.Leu826Phe)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Mar 17, 2020)	criteria provided, single submitter	VCV000931884	6	73195093	931884	919053	rs1765738668	LOF	0.842	0.049	0.109
KCNQ5	820	K	Q	NM_019842.4(KCNQ5):c.2788A>C (p.Lys930Gln)	Intellectual disability, autosomal dominant 46	Uncertain significance(Last reviewed: Feb 17, 2020)	criteria provided, single submitter	VCV001027752	6	73195403	1027752	1016776	rs1765753921	GOF	0.694	0.048	0.258
KCNQ5	928	H	Q	NM_019842.4(KCNQ5):c.2784T>A (p.His928Gln)	not provided	Benign(Last reviewed: Jun 28, 2018)	criteria provided, single submitter	VCV000750703	6	73195399	750703	750213	rs117938048	LOF	0.737	0.058	0.205

Supplementary Table 3. Predicted functional effect of 3760 known voltage-gated potassium channels missense variants. Known variants were extracted from ClinVar (https://www.ncbi.nlm.nih.gov/clinvar) with the search query "KCNA1[gene] OR KCNA2[gene] OR (…) KCNQ5[gene]" for all channels in the training dataset and filtered for missense variants. This yielded 3769 records, last accessed 7/MAR/2022 1:59 pm. Of these, 9 records were excluded due to missing data. Protein changes listed in ClinVar were matched to their corresponding canonical sequences (UniProt). Functional effects were predicted with our pre-trained MTL-SVM model (column 'prediction') with class probabilities estimated by Platt scaling (columns 'prob.LOF', 'prob.Neutral', 'prob.GOF'). Interestingly, variants with a predicted neutral effect were significantly more likely to be benign or likely benign (n = 127/1202) than variants with a predicted non-neutral effect (n = 87/2344, Welch's two-sample t-test $p<0.001$) Abbreviations: aa1 – wild-type amino acid; aa2 – mutated amino acid; LOF – loss-of-function; GOF – gain-of-function; pos – missense variant position on the amino acid sequence.