The role of calsequestrin-2 in calcium-mediated arrhythmias

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Supplementary Table S1: CSQ2 variants associated with CPVT

Variant	Mutation Type	Domain	Phenotype	References
T4I	Missense	N-terminus	CPVT	(Landstrom et al. 2017)
S15C	Missense	N-terminus	CPVT	(Landstrom et al. 2017)
A19E	Missense	N-terminus	CPVT	(Landstrom et al. 2017)
E21+14X	Deletion		CPVT	(Hayashi et al. 2009)
L37I	Missense	I	CPVT	(Landstrom et al. 2017)
E39K	Missense	I	CPVT	(Ng et al. 2020)
E39X	Non-sense	I	CPVT	(Ng et al. 2020)
D50N	Missense	I	CPVT	(Landstrom et al. 2017)
C53F	Missense	I	CPVT	(Ng et al. 2020)
H57R	Missense	I	CPVT	(Landstrom et al. 2017)
E58V	Missense	I	CPVT	(Landstrom et al. 2017)
T66A	SNP	I	CPVT, SD	(Laitinen et al. 2003, Wong et al. 2009)
Q67X	Non-sense	I	CPVT	(Josephs et al. 2017)
K68Nfs*5	Frameshift		PVCs	(Ng et al. 2020)
Q69E	Missense	I	CPVT	(Landstrom et al. 2017)
Q71Hfs*2	Frameshift		CPVT	(Ng et al. 2020)
V76M	SNP	I	SD	(Laitinen et al. 2003, Wong et al. 2009)
L77P	Missense	I	CPVT	(Al-Hassnan et al. 2013)
234+2T>C	Splicing		CPVT	(Ng et al. 2020)
Q82H	Missense	I	CPVT	(Landstrom et al. 2017)
K87X	Non-sense	I	CPVT	(Kawamura et al. 2013)
I89V	Missense	I	CPVT	(Landstrom et al. 2017)
A96G	Missense	I	CPVT	(Landstrom et al. 2017)
320-2A>G	Splicing		CPVT	(Roux-Buisson et al. 2011)
S113N	Missense	I	CPVT	(Landstrom et al. 2017)
S113Rfs*6	Frameshift		CPVT	(di Barletta et al. 2006, Ng et al. 2020)
R121C	Missense	I	CPVT	(Landstrom et al. 2017)
G127G	Splicing		CPVT	(Roux-Buisson et al. 2011)
V135M	Missense	I	CPVT	(Landstrom et al. 2017)
F137L	Missense	I	CPVT	(Landstrom et al. 2017)
L138P	Missense	I	CPVT	(Landstrom et al. 2017)
421-2A>T	Splicing		CPVT	(Ng et al. 2020)
E159K	Missense	II	CPVT	(Landstrom et al. 2017)
R160H	Missense	II	CPVT	(Landstrom et al. 2017)
I161V	Missense	II	CPVT	(Landstrom et al. 2017)
I161L	Missense	II	CPVT	(Landstrom et al. 2017)
I161T	Missense	II	CPVT	(Landstrom et al. 2017)
Y164C	Missense	II	CPVT	(Landstrom et al. 2017)
S173I	Missense	II	CPVT, SD	(Ng et al. 2020, Titus et al. 2020)

E177Q	Missense	II	SD	(Wong et al. 2009)
F182S	Missense	II	CPVT	(Ng et al. 2020)
F182Lfs*28	Frameshift		CPVT	(Hayashi et al. 2009, Roux-Buisson et al. 2011)
P191L	Missense	II	CPVT	(Nouira et al. 2019)
V203Lfs*7	Frameshift	II	CPVT	(Ng et al. 2020)
P231S	Missense	II	CPVT	(Ng et al. 2020)
H244Y	Missense	II	CPVT	(Landstrom et al. 2017)
Q245X	Non-sense	II	CPVT	(Ng et al. 2020)
737+1G>A	Splicing		CPVT	(Roux-Buisson et al. 2011)
737+2T>A	Splicing		CPVT	(Ng et al. 2020)
738-3C>A	Splicing		CPVT	(Ng et al. 2020)
738-2A>G	Splicing		CPVT	(Proost et al. 2017)
R250C	Missense	III	CPVT	(Landstrom et al. 2017, Gao et al. 2018, Li et al. 2019)
R250G	Missense	III	CPVT	(Landstrom et al. 2017)
R251H	Missense	III	CA	(Landstrom et al. 2017, Ng et al. 2020)
R253H	Missense	III	CPVT	(Landstrom et al. 2017)
W261X	Non-sense	III	CPVT	(Ng et al. 2020)
I270T	Missense	III	CA	(Ng et al. 2020)
838+1G>A	Splicing		CPVT	(Li et al. 2019)
383+3A>G	Splicing		CPVT	(Ng et al. 2020)
L288V	Missense	III	CPVT	(Landstrom et al. 2017)
P308L	Missense	III	CPVT	(Hong et al. 2012, Ng et al. 2020)
P308Q	Missense	III	Syncope	(Ng et al. 2020)
D310N	Missense	III	CPVT	(Landstrom et al. 2017, Ng et al. 2020)
939+5G>C	Splicing		CPVT	(Roux-Buisson et al. 2011)
V315I	Missense	III	CPVT	(Landstrom et al. 2017)
D325E	Missense	III	CPVT	(Ng et al. 2020)
P329S	Missense	III	CPVT	(Landstrom et al. 2017, Wang et al. 2020)
Q330X	Non-sense		CPVT	(Hong et al. 2012)
G332R	Missense	III	CPVT	(Landstrom et al. 2017, Wang et al. 2020)
1014+1G>A	Splicing		CPVT	(Landstrom et al. 2017, Ng et al. 2020)
1014+3G>A	Splicing		CPVT	(Landstrom et al. 2017)
A339Vfs*2	Frameshift		CPVT	(Ng et al. 2020)
D340X	Non-sense		CPVT	(Ng et al. 2020)
D351G	Missense	III	SIDS	(Landstrom et al. 2017, Neubauer et al. 2017, Wang et al. 2020)
E359Rfs*12	Frameshift		CPVT	(Ng et al. 2020)
G359Afs*12	Frameshift		CPVT	(Li et al. 2019)
W361R	Missense	III	CA	(Ng et al. 2020)
L366P	Missense	III	SD	(Ng et al. 2020)
E377D	Missense	C-terminus	CPVT	(Landstrom et al. 2017)
D379V	Missense	C-terminus	CPVT	(Landstrom et al. 2017)
383_383del	Deletion		CPVT	(Landstrom et al. 2017)
N384H	Missense	C-terminus	CPVT	(Landstrom et al. 2017)
D392Vfs*84	Frameshift		CPVT	(Li et al. 2019)
D395D	SNP	C-terminus	SD	(Wong et al. 2009)
395_396del	Deletion		CPVT	(Landstrom et al. 2017)
D396N	Missense	C-terminus	CPVT	(Landstrom et al. 2017)
E399A	Missense	C-terminus	CPVT	(Landstrom et al. 2017)

CA: cardiac arrest; CPVT: catecholaminergic polymorphic ventricular tachycardiac; CSQ2: calsequestrin-2; PVCs: premature ventricular contractions; SD: sudden death; SIDS: sudden infant death syndrome; SNP: single-nucleotide polymorphism

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