# Supplementary Online Content

SoRelle JA, Pascual JM, Gotway G, Park JY. Assessment of interlaboratory variation in the interpretation of genomic test results in patients with epilepsy. *JAMA Netw Open.* 2020;3(4):e203812. doi:10.1001/jamanetworkopen.2020.3812



This supplementary material has been provided by the authors to give readers additional information about their work.

**eTable 1.** Genes Associated With Epilepsy

| | | | | | |
|---|---|---|---|---|---|
| *ADSL* | *CLN5* | *GABRG2* | *LIAS* | *PPT1* | *SRPX2* |
| *ALDH7A1*[a] | *CLN6* | *GAMT* | *MAGI2* | *PRICKLE1* | *STXBP1* |
| *ARX* | *CLN8* | *GATM* | *MBD5* | *PRRT2*[a] | *SYN1* |
| *ATP1A2* | *CNTNAP2* | *GRIN2A*[a] | *MECP2* | *SCARB2* | *TBC1D24* |
| *ATP6AP2* | *CSTB* | *GRIN2B* | *MEF2C* | *SCN1A*[a] | *TCF4* |
| *CACNB4* | *CTSD* | *GOSR2* | *MFSD8* | *SCN1B* | *TPP1* |
| *CDKL5* | *DNAJC5* | *KANSL1* | *NHLRC1* | *SCN2A*[a] | *TSC1*[a] |
| *CHRNA2*[a] | *EFHC1* | *KCNJ10* | *NRXN1* | *SCN8A*[a] | *TSC2*[a] |
| *CHRNA4*[a] | *EPM2A*[a] | *KCNQ2*[a] | *PCDH19*[a] | *SLC25A22* | *UBE3A* |
| *CHRNA7* | *FOLR1* | *KCNQ3*[a] | *PNKP* | *SLC2A1*[a] | *ZEB2* |
| *CHRNB2*[a] | *FOXG1* | *KCTD7* | *PNPO*[a] | *SLC9A6* | |
| *CLN3* | *GABRA1* | *LGI1* | *POLG*[a] | *SPTAN1* | |

[a]Denotes genes with therapeutic significance

**eTable 2.** Variants With Clinically Substantial Discrepancies

| Gene | Nucleotide change | Amino acid change | Transcript | dbSNP |
|---|---|---|---|---|
| ADSL | c.153+1G>T | | NM_000026.2 | rs1555903969 |
| ADSL | c.1342T>C | p.Ser448Pro | NM_000026.2 | rs771121666 |
| ADSL | c.1400C>G | p.Pro467Arg | NM_000026.2 | rs1057521071 |
| ADSL | c.953C>T | p.Pro318Leu | NM_000026.2 | rs202064195 |
| ALDH7A1 | c.34delG | p.Ala12Leufs | NM_001182.4 | rs750693623 |
| ALDH7A1 | c.575C>T | p.Thr192Met | NM_001182.4 | rs376917645 |
| ALDH7A1 | c.664A>G | p.Thr222Ala | NM_001182.4 | rs777829351 |
| ALDH7A1 | c.1232C>T | p.Pro411Leu | NM_001182.4 | rs780233639 |
| ALDH7A1 | c.1301A>G | p.Tyr434Cys | NM_001182.4 | rs747597620 |
| ARX | c.428_451dup24 | p.Ala150_Ala151insGlyAlaAlaAlaAlaAlaAlaAla | NM_139058.2 | rs387906493 |
| ARX | c.453_458dupGGCCGC | p.Ala155_Trp156insAlaAla | NM_139058.2 | rs398124512 |
| ATP1A2 | c.1777C>T | p.Arg593Trp | NM_000702.3 | rs886039530 |
| ATP1A2 | c.2438T>A | p.Met813Lys | NM_000702.3 | rs796052277 |
| CDKL5 | c.2842C>T | p.Arg948Ter | NM_001323289.2 | rs1555955296 |
| CHRNB2 | c.1291G>C | p.Val431Leu | NM_000748.2 | rs1064796396 |
| CLN3 | c.1213C>T | p.Arg405Trp | NM_001042432.1 | rs139842473 |
| CLN3 | c.790+3A>C | | NM_001042432.1 | rs386833738 |
| CLN5 | c.1121A>G | p.Tyr374Cys | NM_006493.2 | rs148862100 |
| CLN5 | c.334C>T | p.Arg112Cys | NM_006493.2 | rs786205211 |
| CLN6 | c.794_796delCCT | p.Ser265del | NM_017882.2 | rs768422260 |
| CLN6 | c.898T>C | p.Trp300Arg | NM_017882.2 | rs750937323 |
| CLN8 | c.209G>A | p.Arg70His | NM_018941.3 | rs386834124 |
| CLN8 | c.703delC | p.Val236Serfs | NM_018941.3 | rs761621368 |
| CSTB | c.1_2insAT | p.Met1Asnfs | NM_000100.3 | rs1555955296 |
| CTSD | c.751G>A | p.Asp251Asn | NM_001909.4 | rs1064796396 |
| FOLR1 | c.493+2T>C | | NM_016725.2 | rs1044894207 |
| FOXG1 | c.670G>A | p.Gly224Ser | NM_005249.4 | rs727503935 |
| FOXG1 | c.685A>C | p.Ile229Leu | NM_005249.4 | rs1064797186 |
| GABRA1 | c.335G>A | p.Arg112Gln | NM_000806.5 | rs587777308 |
| GABRA1 | c.640C>T | p.Arg214Cys | NM_000806.5 | rs727503940 |
| GABRA1 | c.851T>C | p.Val284Ala | NM_000806.5 | rs794727962 |
| GABRG2 | c.269C>T | p.Thr90Met | NM000816.3 | rs1057520498 |
| GABRG2 | c.549-3T>G | | NM198903.2 | rs750459631 |
| GABRG2 | c.1000G>A | p.Ala334Thr | NM000816.3 | rs398123523 |
| GABRG2 | c.1336C>T | p.Arg446Trp | NM000816.3 | rs796052515 |
| GAMT | c.507_521dup | p.Ser173_Trp174insCysAsnLeuThrSer | NM_000156.5 | rs763407972 |
| GRIN2A | c.2069C>T | p.Thr690Met | NM_000833.4 | rs1445802934 |
| GRIN2A | c.2197G>A | p.Ala733Thr | NM_000833.4 | rs796052550 |
| GRIN2A | c.2452G>A | p.Ala818Thr | NM_000833.4 | rs1555483699 |
| GRIN2B | c.1547A>G | p.Asn516Ser | NM_000834.4 | rs886041295 |
| GRIN2B | c.1672G>A | p.Val558Ile | NM_000834.4 | rs1057519004 |
| GRIN2B | c.1727_1732delTCTTTG | p.Val576_Phe577del | NM_000834.4 | rs1555111511 |
| GRIN2B | c.2002G>A | p.Asp558Asn | NM_000834.4 | rs876661151 |
| GRIN2B | c.2087G>A | p.Arg696His | NM_000834.4 | rs1555103971 |
| GRIN2B | c.2450A>G | p.Asn817Ser | NM_000834.4 | rs1555103159 |
| GRIN2B | c.2459G>C | p.Gly820Ala | NM_000834.3 | rs797044849 |
| KANSL1 | c.868C>T | p.Arg290Ter | NM_001193466.1 | rs144637717 |
| KCNJ10 | c.76C>T | p.Arg26Ter | NM_002241.4 | rs138943405 |
| KCNJ10 | c.221C>T | p.Thr74Ile | NM_002241.4 | rs796052604 |
| KCNQ2 | c.593G>A | p.Arg198Gln | NM_172107.3 | rs796052621 |
| KCNQ2 | c.601C>T | p.Arg201Cys | NM_172107.3 | rs796052623 |
| KCNQ2 | c.637C>T | p.Arg213Trp | NM_172107.3 | rs118192203 |
| KCNQ2 | c.701C>T | p.Thr234Ile | NM_172107.3 | rs794727741 |
| KCNQ2 | c.704C>T | p.Ala235Val | NM_172107.3 | rs797045638 |
| KCNQ2 | c.782T>C | p.Phe261Ser | NM_172107.3 | rs796052631 |
| KCNQ2 | c.841G>A | p.Gly281Arg | NM_172107.3 | rs794727813 |
| KCNQ2 | c.901G>A | p.Gly301Ser | NM_172107.3 | rs1057516099 |
| KCNQ2 | c.1057C>T | p.Arg353Cys | NM_172107.3 | rs118192218 |
| KCNQ2 | c.1627G>A | p.Val543Met | NM_172107.3 | rs794727134 |
| KCNQ2 | c.1639C>T | p.Arg547Trp | NM_172107.3 | rs796052650 |
| KCNQ2 | c.1887+5G>A | | NM_172107.3 | rs777916008 |
| KCNQ2 | c.2245G>T | p.Glu749Ter | NM_172107.3 | rs796052658 |
| KCTD7 | c.172G>A | p.Gly58Arg | NM_153033.4 | rs750033880 |
| KCTD7 | c.190A>G | p.thr64Ala | NM_153033.4 | rs201296399 |
| KCTD7 | c.456G>A | p.Val152= | NM_153033.4 | rs796052686 |
| LIAS | c.983T>A | p.Phe328Tyr | NM_006859.3 | rs779931959 |

| | | | | |
|---|---|---|---|---|
| MECP2 | c.343C>T | p.Arg115Cys | NM_004992.3 | rs267608388 |
| MECP2 | c.397C>T | p.Arg133Cys | NM_004992.3 | rs28934904 |
| MECP2 | c.455C>G | p.Pro152Arg | NM_004992.3 | rs61748404 |
| MECP2 | c.568C>T | p.Arg190Cys | NM_004992.3 | rs587783137 |
| MECP2 | c.602C>T | p.Ala201Val | NM_004992.3 | rs61748381 |
| MECP2 | c.1129_1198del70 | p.Lys377Profs | NM_004992.3 | rs1557135353 |
| MECP2 | c.1200A>C | p.Pro400= | NM_001110792.1 | rs797044732 |
| MFSD8 | c.362A>G | p.Tyr121Cys | NM_152778.2 | rs118203978 |
| MFSD8 | c.1006G>C | p.Glu336Gln | NM_152778.2 | rs150418024 |
| NHLRC1 | c.386C>A | p.Pro129His | NM_198586.2 | rs750465793 |
| NHLRC1 | c.1142A>G | p.Asp381Gly | NM_198586.2 | rs200201752 |
| NRXN1 | c.3619C>T | p.Arg1207Ter | NM_001135659.2 | rs149830411 |
| PCDH19 | c.697G>T | p.Asp233Tyr | NM_001184880.1 | rs1555985482 |
| PCDH19 | c.707C>G | Pro236Arg | NM_001184880.1 | rs1060502176 |
| PCDH19 | c.790G>T | p.Asp264Tyr | NM_001105243.1 | rs587784300 |
| PCDH19 | c.1114C>T | p.Arg372Trp | NM_001184880.1 | rs796052812 |
| PCDH19 | c.1682C>G | p.Pro561Arg | NM_001184880.1 | rs796052819 |
| PNKP | c.1029+2T>C | | NM_007254.3 | rs199919568 |
| PNKP | c.1123G>T | p.Gly375Trp | NM_007254.3 | rs786203983 |
| PNKP | c.1385G>C | p.Arg462Pro | NM_007254.3 | rs376854895 |
| PNKP | c.968C>T | p.Thr323Met | NM_007254.3 | rs372148913 |
| PNPO | c.686G>A | p.Arg229Gln | NM_018129.3 | rs773450573 |
| POLG | c.488C>T | p.Pro163Leu | NM_002693.2 | rs752892262 |
| POLG | c.752C>T | p.Thr251Ile | NM_002693.2 | rs113994094 |
| POLG | c.830A>T | p.His277Leu | NM_002693.2 | rs138929605 |
| POLG | c.855G>C | p.Gln285His | NM_002693.2 | rs141367015 |
| POLG | c.1156C>T | p.Arg386Cys | NM_002693.2 | rs199759055 |
| POLG | c.1276G>A | p.Gly426Ser | NM_002693.2 | rs775576189 |
| POLG | c.1402A>G | p.Asn468Asp | NM_002693.2 | rs145843073 |
| POLG | c.1760C>T | p.Pro587Leu | NM_002693.2 | rs113994096 |
| POLG | c.1763G>A | p.Gly588Asp | NM_002693.2 | rs371334941 |
| POLG | c.1790G>A | p.Arg597Gln | NM_002693.2 | Chr15(GRCh38):g.89325609G>A |
| POLG | c.2209G>C | p.Gly737Arg | NM_002693.2 | rs121918054 |
| POLG | c.2243G>C | p.Trp748Ser | NM_002693.2 | rs113994097 |
| POLG | c.2246T>C | p.Phe749Ser | NM_002693.2 | rs202037973 |
| POLG | c.2264A>C | p.Lys755Thr | NM_002693.2 | rs770438363 |
| POLG | c.2293C>A | p.Pro765Thr | NM_002693.2 | rs1003442806 |
| POLG | c.2419C>T | p.Arg807Cys | NM_002693.2 | rs769827124 |
| POLG | c.2606G>A | p.Arg869Gln | NM_002693.2 | rs1356604153 |
| POLG | c.2620T>A | p.Leu874Met | NM_002693.2 | rs758402960 |
| POLG | c.2636A>G | p.Gln879Arg | NM_002693.2 | rs368587966 |
| POLG | c.2642C>T | p.Pro881Leu | NM_002693.2 | rs375935084 |
| POLG | c.264C>G | p.Phe88Leu | NM_002693.2 | rs144439703 |
| POLG | c.2663G>A | p.Gly888Asp | NM_002693.2 | rs878854560 |
| POLG | c.2665G>A | p.Ala889Thr | NM_002693.2 | rs763393580 |
| POLG | c.2740A>C | p.Thr914Pro | NM_002693.2 | rs139590686 |
| POLG | c.2857C>T | p.Arg953Cys | NM_002693.2 | rs11546842 |
| POLG | c.2890C>T | p.Arg964Cys | NM_002693.2 | rs201477273 |
| POLG | c.3139C>T | p.Arg1047Trp | NM_002693.2 | rs181860632 |
| POLG | c.3151G>A | p.Gly1051Arg | NM_002693.2 | Chr15(GRCh38):g.89319053G>A |
| POLG | c.3151G>C | p.Gly1051Arg | NM_002693.2 | rs121918049 |
| POLG | c.3287G>A | p.Arg1096His | NM_002693.2 | rs368435864 |
| POLG | c.3383G>A | p.Arg1128His | NM_002693.2 | Chr15(GRCh28):g.89318640G>A |
| POLG | c.3412C>T | p.Arg1138Cys | NM_002693.2 | rs767138032 |
| POLG | c.3527C>T | p.Ser1176Leu | NM_002693.2 | rs776031396 |
| POLG | c.3573G>T | p.Lys1191Asn | NM_002693.2 | rs1085307741 |
| POLG | c.3640C>T | p.Gln1214Ter | NM_002693.2 | rs781256643 |
| PRICKLE1 | c.824C>T | p.Thr275Met | NM_153026.2 | rs199546979 |
| PRRT2 | c.922C>T | p.Arg308Cys | NM_145239.2 | rs932713001 |
| SCN1A | c.602+1G>A | | NM_001165963.2 | rs794726827 |
| SCN1A | c.602+2dupT | | NM_001165963.2 | rs796053054 |
| SCN1A | c.791T>C | p.Ile264Thr | NM_001165963.2 | rs745664511 |
| SCN1A | *c.986G>T* | p.Gly329Val | NM_001165963.2 | rs779184118 |
| SCN1A | c.1216G>T | p.Val406Phe | NM_001165963.2 | rs121918768 |
| SCN1A | c.1264G>A | p.Val422Met | NM_001165963.2 | rs886042528 |
| SCN1A | c.2594G>A | p.Arg865Gln | NM_001165963.2 | rs1057517862 |
| SCN1A | c.2665G>A | p.Ala889Thr | NM_001165963.2 | rs1266877537 |
| SCN1A | c.2729A>G | p.Gln910Arg | NM_001165963.2 | rs1064795735 |
| SCN1A | c.2839G>A | p.Val947Met | NM_001165963.2 | rs796052986 |

| | | | | |
|---|---|---|---|---|
| SCN1A | c.2941C>A | p.Leu981Ile | NM_001165963.2 | rs1057518112 |
| SCN1A | c.3698G>A | p.Gly1233Asp | NM_001165963.2 | rs1553532470 |
| SCN1A | c.3776T>C | p.Phe1259Ser | NM_001165963.2 | rs398123591 |
| SCN1A | c.3879+5G>A | | NM_001165963.2 | rs796052999 |
| SCN1A | c.4171A>C | p.Asn1391His | NM_001165963.2 | rs1295072436 |
| SCN1A | c.4321G>A | p.Ala1441Thr | NM_001165963.2 | rs121917974 |
| SCN1A | c.4547C>T | p.Ser1516Leu | NM_001165963.2 | rs139300715 |
| SCN1A | c.4556C>T | p.Pro1519Leu | NM_001165963.2 | rs372425457 |
| SCN1A | c.472G>C | p.Glu158Gln | NM_001165963.2 | rs796053090 |
| SCN1A | c.4787G>A | p.Arg1596His | NM_001165963.2 | rs575368466 |
| SCN1A | c.4793A>T | p.Tyr1598Phe | NM_001165963.2 | rs377325221 |
| SCN1A | c.4973C>T | p.Thr1658Met | NM_001165963.2 | rs121917922 |
| SCN1A | c.5306A>G | p.Tyr1769Cys | NM_001165963.2 | rs886039460 |
| SCN1A | c.5563C>T | p.Pro1855Ser | NM_001165963.2 | rs794727415 |
| SCN1A | c.5797delC | p.Arg1933Glufs | NM_001165963.2 | rs587780446 |
| SCN1B | c.457G>A | p.Asp153Asn | NM_001037.4 | rs72550247 |
| SCN2A | c.605C>T | p.Ala202Val | NM_020822.2 | rs1553567409 |
| SCN2A | c.1267G>C | p.Val423Leu | NM_020822.2 | rs796053180 |
| SCN2A | c.2657T>C | p.Leu886Ser | NM_020822.2 | rs796053118 |
| SCN2A | c.4782G>C | p.Trp1594Cys | NM_020822.2 | rs1057521747 |
| SCN2A | c.4877G>A | p.Arg1626Gln | NM_020822.2 | rs796053155 |
| SCN2A | c.4879G>A | p.Val1627Met | NM_020822.2 | rs796053156 |
| SCN2A | c.4886G>A | p.Arg1629His | NM_020822.2 | rs796053157 |
| SCN2A | c.5645G>A | p.Arg1882Gln | NM_020822.2 | rs794727444 |
| SCN8A | c.632T>C | p.Val211Ala | NM_014191.3 | rs1057518487 |
| SCN8A | c.1250A>C | p.Gln417Pro | NM_014191.3 | rs878854973 |
| SCN8A | c.1445A>G | p.Lys482Arg | NM_014191.3 | rs769520392 |
| SCN8A | c.2549G>A | p.Arg850Gln | NM_014191.3 | rs587780586 |
| SCN8A | c.3955G>T | p.Ala1319Ser | NM_014191.3 | rs796053214 |
| SCN8A | c.4235T>C | p.Phe1412Ser | NM_014191.3 | rs1555228665 |
| SCN8A | c.4441A>G | p.Met1481Val | NM_014191.3 | rs886041670 |
| SCN8A | c.4840A>G | p.Thr1614Ala | NM_014191.3 | rs1555230909 |
| SCN8A | c.4850G>A | p.Arg1617Gln | NM_014191.3 | rs587777721 |
| SCN8A | c.4877G>A | p.Arg1626His | NM_014191.3 | rs886044328 |
| SCN8A | c.5630A>G | p.Asn1877Ser | NM_014191.3 | rs587780455 |
| SLC2A1 | c.400G>A | p.Gly134Ser | NM_006516.3 | rs1057518953 |
| SLC9A6 | c.430-9_430-5delTTTTA | | NM_006359.2 | rs796053290 |
| SPTAN1 | c.6908_6916dupACCAGCTGG | p.Leu2305_Gly2306insAspGlnLeu | NM_001130438.2 | rs587784440 |
| SRPX2 | c.1030C>A | p.Leu3444Ile | NM_014467.2 | rs149051060 |
| STXBP1 | c.734A>G | p.His245Arg | NM_001032221.3 | rs587784453 |
| STXBP1 | c.874C>T | p.Arg292Cys | NM_003165.3 | rs786205598 |
| SYN1 | c.1297C>T | p.His433Tyr | NM_133499.2 | rs41298474 |
| TBC1D24 | c.241_252delATCGTGGGCAAG | p.Ile81_Lys84del | NM_001199107.1 | rs761918906 |
| TBC1D24 | c.328G>A | p.Gly110Ser | NM_001199107.1 | rs747821285 |
| TBC1D24 | c.457G>A | p.Glu153Lys | NM_001199107.1 | rs376712059 |
| TBC1D24 | c.679C>T | p.Arg227Trp | NM_001199107.1 | rs748302886 |
| TBC1D24 | c.680G>T | p.Arg227Leu | NM_001199107.1 | rs756181906 |
| TBC1D24 | c.845C>G | p.Pro282Arg | NM_020705.2 | rs747538224 |
| TCF4 | c.1741G>T | p.Val581Phe | NM_001083962.1 | rs587784460 |
| TPP1 | c.1016G>A | p.Arg339Gln | NM_000391.3 | rs765380155 |
| TPP1 | c.1058C>A | p.Thr353Asn | NM_000391.3 | rs145966505 |
| TPP1 | c.381-10dupT | | NM_000391.3 | rs146315473 |
| TPP1 | c.887-18A>G | | NM_000391.3 | rs935526225 |
| TSC1 | c.737+3A>G | | NM_000368.4 | rs118203439 |
| TSC1 | c.737G>A | p.Arg246Lys | NM_000368.4 | rs118203436 |
| TSC2 | c.886G>A | p.Val296Met | NM_000548.4 | rs747237113 |
| TSC2 | c.1864C>T | p.Arg622Trp | NM_000548.4 | rs397514914 |
| TSC2 | c.2666C>T | p.Ala889Val | NM_000548.4 | rs137854155 |
| TSC2 | c.3599G>A | p.Arg1200Gln | NM_000548.3 | rs397515225 |
| TSC2 | c.4747G>A | p.Glu1583Lys | NM_000548.4 | rs886039446 |
| TSC2 | c.5260-1G>C | | NM_000548.3 | rs1057518103 |
| UBE3A | c.349T>C | p.Cys117Arg | NM_130838.1 | rs587782907 |
| UBE3A | c.947T>C | p.Met316Thr | NM_130838.1 | rs863225071 |
| UBE3A | c.1805A>G | p.Asn602Ser | NM_130838.1 | rs587784521 |
| UBE3A | c.2503_2508delCTTAAA | p.Leu835_Lys836del | NM_130838.1 | rs863225070 |
| UBE3A | c.2503C>T | p.Leu835Phe | NM_130838.1 | rs587783097 |

**eTable 3.** *SCN1A* Variant Discordance, Not Clinically Substantial

| Nucleotide change | Amino acid change | Transcript | Lab A | Lab B | Lab C | Lab D | Others |
|---|---|---|---|---|---|---|---|
| c.2T>C | p.Met1Thr | NM_001165963.2 | P | LP | | | |
| c.68C>T | p.Ala23Val | NM_006920.6 | B | LB | | | |
| c.90A>G | p.Ala30= | NM_001165963.1 | LB | | | VUS | |
| c.144C>T | p.Gly48= | NM_001165963.1 | LB | | | VUS | |
| c.251A>G | p.Tyr84Cys | NM_006920.4 | P | LP | | | |
| c.265-4A>G | | NM_001165963.1 | B | LB | | | |
| c.333A>G | p.Leu111= | NM_001165963.1 | B | | | | VUS |
| c.345T>C | p.Asn115= | NM_001165963.1 | | B | B | B | LB |
| c.579C>T | p.Leu193= | NM_001165963.1 | B | | | VUS | |
| c.694+10A>G | | NM_001165963.1 | B | LB | | VUS | |
| c.852T>C | p.Asn284= | NM_001165963.1 | LB | | | VUS | |
| c.965-1G>A | | NM_001165963.1 | P | LP | | | |
| c.1000C>G | p.Leu334Val | NM_001165963.1 | LB | VUS | LB | | VUS |
| c.1065T>C | p.Gly355= | NM_001165963.1 | B | | | | LB |
| c.1131A>C | p.Arg377= | NM_001165963.1 | B | B | LB | B | LB, VUS |
| c.1150T>A | p.Trp384Arg | NM_001165963.1 | LP | P | | | |
| c.1171-10_1171-9delTT | | NM_001165963.1 | B | B | | B | LB |
| c.1212A>G | p.Val404= | NM_001165963.1 | | | B | B | B |
| c.1329A>G | p.Glu443= | NM_001165963.2 | | LB | | | |
| c.1378-3T>C | | NM_001165963.1 | B | LB | LB | | |
| c.1410C>T | p.Ser470= | NM_001165963.1 | B | | LB | | VUS |
| c.1499G>A | p.Arg500Gln | NM_001165963.1 | VUS | LB | | VUS | |
| c.1625G>A | p.Arg542Gln | NM_006920.4 | B | B | LB | LB | VUS |
| c.1662+9C>A | | NM_001165963.1 | B | B | | B | LB |
| c.1680T>C | p.Arg560= | NM_001165963.2 | | LB | LB | VUS | |
| c.1739G>A | p.Arg580Gln | NM_001165963.1 | VUS | VUS | | VUS | LB |
| c.1797G>A | p.Glu599= | NM_001165963.2 | | LB | LB | VUS | |
| c.1803C>T | p.Asn601= | NM_001165963.2 | | LB | | | VUS |
| c.1811G>A | p.Arg604His | NM_006920.4 | B | B | B | B | LB |
| c.1818T>C | p.Asp606= | NM_001165963.1 | | LB | | VUS | |
| c.2044-5delT | | NM_001165963.1 | LB | B | | | LB |
| c.2044-5dupT | | NM_001165963.1 | B | B | | VUS | |
| c.2176+11A>C | | NM_001165963.1 | LB | | | | VUS |
| c.2292T>C | p.Val764= | NM_001165963.1 | | | B | B | LB, B |
| c.2378C>T | p.Thr793Met | NM_001165963.1 | VUS | LB | | | |
| c.2421C>T | p.Phe807= | NM_001165963.1 | B | B | LB | LB | LB |
| c.2590C>T | p.Leu864= | NM_001165963.2 | LB | VUS | | | |
| c.2591C>T | p.Thr864Met | NM_006920.4 | | P | LP | | |
| c.2758C>T | p.Arg920Cys | NM_006920.4 | P | P | | LP | |
| c.2889T>C | p.Ala963= | NM_001165963.1 | | B | LB | B | LB |
| c.3039A>G | p.Gln1013= | NM_001165963.2 | | VUS | LB | | VUS |
| c.3060C>A | p.His1020Gln | NM_001165963.1 | VUS | LB | | VUS | |
| c.3199G>A | p.Ala1067Thr | NM_001165963.1 | B | | B | B | B, LB |
| c.3481G>A | p.Ala1161Thr | NM_001165963.1 | VUS | LB | | VUS | |
| c.3488C>G | P.Thr1163Ser | NM_001165963.1 | LB | B | LB | VUS | LB |
| c.3573T>C | p.Cys1191= | NM_001165963.1 | LB | LB | | | VUS |
| c.3585T>C | p.As1195= | NM_001165963.1 | LB | | | VUS | |
| c.3591A>G | p.Glu1197= | NM_001165963.2 | LB | | | VUS | |
| c.3681A>C | p.Glu1227Asp | NM_006920.4 | VUS | LB | | VUS | |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| c.3690T>C | p.Tyr1230= | NM_006920.4 | B | LB | B | B | LB |
| c.3701G>A | p.Arg1234Gln | NM_006920.4 | P | LP | | | |
| c.3705+10delA | | NM_001165963.1 | B | | | | VUS |
| c.3749C>T | p.Thr1250Met | NM_001165963.1 | VUS | LB | | | |
| c.3886T>C | p.Leu1296= | NM_001165963.1 | B | B | LB | | |
| c.3891A>T | p.Glu1297Asp | NM_006920.4 | LB | LB | LB | VUS | |
| c.3899C>T | p.Thr1300Ile | NM_001165963.1 | VUS | VUS | LB | VUS | |
| c.3948G>A | p.Arg1316= | NM_001165963.1 | B | B | LB | LB | |
| c.4129A>G | p.Il31377Val | NM_001165963.1 | VUS | | LB | VUS | |
| c.4167C>T | p.Asp1389= | NM_001165963.1 | LB | LB | | | VUS |
| c.4339-5G>A | | NM_001165963.1 | | LB | | VUS | |
| c.4393A>G | p.Ile1465Val | NM_001165963.1 | B | LB | LB | B | |
| c.4548G>A | p.Ser1516= | NM_001165963.1 | B | B | LB | B | |
| c.4551A>G | p.Ly1517= | NM_001165963.1 | LB | B | LB | | LB |
| c.4557G>A | p.Pro1519= | NM_001165963.1 | B | LB | LB | | |
| c.4581+12C>T | | NM_001165963.1 | LB | | | | VUS |
| c.4724G>A | p.Arg1575His | NM_001165963.1 | VUS | LB | | VUS | |
| c.4729T>C | p.Cys1577Arg | NM_006920.4 | LP | P | | | |
| c.4731T>C | p.Asn1577= | NM_001165963.1 | B | B | LB | LB | VUS |
| c.4789G>T | p.Asp1597Tyr | NM_006920.4 | | LP | | | P |
| c.4855A>G | p.Met1619Val | NM_001165963.1 | VUS | B | | | |
| c.4872G>A | p.Leu1624= | NM_001165963.1 | B | B | LB | VUS | |
| c.4887C>T | p.Phe1629= | NM_001165963.1 | LB | | | | VUS, LB |
| c.4905C>T | p.Phe1635= | NM_001165963.2 | LB | | | | B |
| c.4945C>T | p.Leu1649= | NM_001165963.1 | B | B | LB | B | LB |
| c.5217C>T | p.Pro1739= | NM_001165963.2 | | LB | | | VUS |
| c.5286A>G | p.Gly1762= | NM_001165963.1 | | B | LB | LB | |
| c.5346C>T | p.Ile1782= | NM_001165963.1 | B | | | | VUS |
| c.5418G>A | p.Glu1806= | NM_001165963.1 | B | B | B | B | LB |
| c.5532A>G | p.Pro1844= | NM_001165963.1 | LB | | | VUS | |
| c.5568G>A | p.Met1856Ile | NM_001165963.1 | VUS | LB | VUS | VUS | |
| c.5693C>T | p.Thr1898Ile | NM_006920.4 | LP | P | | | |
| c.5749C>G | p.Arg1917Gly | NM_006920.4 | LB | B | LB | LB | LB |
| c.5809C>G | p.Gln1937Glu | NM_001165963.2 | VUS | LB | | | |
| c.5864T>C | p.Ile1955Thr | NM_001165963.1 | B | LB | B | B | LB |

*SCN1A* Transcript NM_001165963.2. [a] An empty cell is present for splice site variants as no amino acid is changed. Empty cells under the "Lab" categories are present when no interpretation was submitted from the corresponding laboratory. P, pathogenic; LP, likely pathogenic; VUS, variant of uncertain significance; LB, likely benign; B, benign.