| Gene | Exon | Nucleotide change | Amino acid change | rs-number (dbSNP) | Variation type | Index cases (n) | Minor allele frequency (MAF) dbSNP |
|---|---|---|---|---|---|---|---|
| ***KCNQ1*** | 1 | c.1-5T>C | - | unknown | Intron | 1 | NA |
| | 5 | c.720C>T | p.H240H | rs28730754 | Silent | 1 | T=0.005/11 |
| | 10 | c.1179G>T | p.K393N | rs12720457 | Missense | 1 | T=0.001/1 |
| | 10 | c.1343C>G | p.P448R | rs8179001 | Missense | 3 | NA |
| | 16 | c.1926C>T | p.C642C | rs12720454 | Silent | 1 | NA |
| | 16 | c.1942G>A | p.V648I | rs34150427 | Missense | 1 | A=0.005/12 |
| ***KCNH2*** | 4 | c.638A>G | p.D213G | unknown | Missense | 1 | NA |
| | 7 | c.1812C>T | p.G604G | unknown | Silent | 1 | NA |
| | 11 | c.2690A>C | p.K897T | rs1805123 | Missense | 56 | G=0.129/283 |
| | 12 | c.2948C>T | p.T983I | rs149955375 | Missense | 1 | NA |
| | 13 | c.3140G>T | p.R1047L | rs36210421 | Missense | 14 | A=0.017/36 |
| ***SCN5A*** | 3 | c.354C>T | p.H118H | rs45533640 | Silent | 2 | A=0.001/1 |
| | 4 | c.462A>T | p.P154P | unknown | Silent | 1 | NA |
| | 5 | c.553G>A | p.A185T | rs192113333 | Missense | 1 | NA |
| | 6 | c.630G>A | p.V210V | unknown | Silent | 1 | NA |
| | 7 | c.717C>T | p.I239I | rs41285129 | Silent | 1 | NA |
| | 8 | c.993C>T | p.D331D | unknown | Silent | 1 | NA |
| | 10 | c.1141-3C>A | - | rs41312433 | Intron | 54 | T=0.145/318 |
| | 12 | c.1587T>C | p.I529I | rs45624133 | Silent | 1 | G=0.003/7 |
| | 12 | c.1653G>A | p.A551A | unknown | Silent | 1 | NA |
| | 12 | c.1673A>G | p.H558R | rs1805124 | Missense | 66 | C=0.206/451 |
| | 12 | c.1681C>T | p.L561L | rs45522138 | Silent | 2 | A=0.001/2 |
| | 12 | c.1715C>A | p.A572D | rs36210423 | Missense | 3 | A=0.001/1 |
| | 17 | c.3093C>T | p.G1031G | unknown | Silent | 1 | NA |
| | 18 | c.3308C>A | p.S1103Y | rs7626962 | Missense | 1 | T=0.012/26 |
| | 20 | c.3578G>A | p.R1193Q | rs41261344 | Missense | 1 | T=0.011/25 |
| | 23 | c.4218G>A | p.G1406G | rs41311123 | Missense | 5 | A=0.001/1 |
| | 26 | c.4509C>T | p.S1503S | rs45548237 | Silent | 1 | A=0.002/5 |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | 28 | c.4824C>T | p.L1608L | rs45437099 | Silent | 1 | NA |
| | 28 | c.4848C>T | p.F1616F | rs41315495 | Silent | 1 | A=0.034/75 |
| | 28 | c.5851G>T | p.V1951L | rs41315493 | Missense | 1 | A=0.004/9 |
| | 28 | c.6010T>C | p.F2004L | rs41311117 | Missense | 1 | G=0.001/3 |
| ***KCNE1*** | 4 | c.29C>T | p.T10M | rs144917638 | Missense | 1 | NA |
| | 4 | c.112A>G | p.S38G | rs1805127 | Missense | 51 | NA |
| | 4 | c.253G>A | p.D85N | rs1805128 | Missense | 12 | NA |
| | 4 | c.293G>A | p.R98Q | rs150454912 | Missense | 1 | NA |
| ***KCNE2*** | 2 | c.22A>G | p.T8A | rs2234916 | Missense | 2 | NA |

**Additional file 2.** Non-pathogenic variants in the *KCNQ1, KCNH2, SCN5A, KCNE1,* and *KCNE2*-genes. Variants within *RYR2* are not reported due to small sample size. All missense substitutions, rare silent substitutions (MAF less than 5%) and variants within -5 or +5 from the exon boundary are reported. Common silent substitutions (MAF more than 5%) and intronic variants more than 5 bp from an exon/intron boundary are not reported. NA = available.