<b>序号</b> 1 2	突变基因 KCNQ1 KCNQ1	B 临床表型 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	mRNNA参考模板 NM_000218.3 NM_000218.3	碱基改变 Phe340Leu Ala341Glu	<b>氨基酸改变</b> 1020T>G 1022C>A	<b>突变类型</b> 错义突变 错义突变
3 4 5 6	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Ala341Val Ser373Trpfs*10 Gly345Glu Gly345Ala	1022C>T 1033-1—1117dup 1034G>A 1034G>C	错义突变       移码突变       错义突变
7 8 9	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	Leu347Pro Gln357Arg 360_361dupKQ	1040T>C 1070A>G 1071_1076dupGAAGCA	错义突变       错义突变       移码突变
10 11 12 13	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	ins 6 bp codon 359 Arg366Trp Leu374Arg Arg380Gly	1073_1078dupAGCAGA 1096C>T 1121T>G 1138A>G	插入突变       错义突变       错义突变       错义突变
14 15 16 17	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	ins 1 bp codon 383 ins 1 bp codon 384 T391I 356-357delGln	1148dupC 1149dupT 1172C>T 1176-1181del	移码突变 移码突变 错义突变 错义突变
17 18 19 20	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	del 1 bp codon 396 Val440AlafsX26 Gly45Ser	1170-1181del 1189delC 1319delT 133G>A	超入光度       缺失突变       错义突变       错义突变
21 22 23 24	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	ins 1 bp codon 447 His455Tyr Pro448Arg	1343dupC 1363C>T 1394-1G>T 1453C>G	移码突变       错义突变       剪接突变       错义突变
25 26 27	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	Val516Ile Arg518X) Arg519His	1546G>A 1552C>T 1556G>A	错义突变       错义突变       错义突变
28 29 30 31	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Arg533Trp Arg539Trp Gln560Lys Arg562Ser	1597C>T 1615C>T 1678C>A 1686G>T	错义突变       错义突变       错义突变       错义突变
32 33 34	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	Gly568Ala Lys581Term Arg583His	1703G>C 1741A>T 1748G>A	错义突变       无义突变       错义突变
35 36 37 38	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Thr587Met Gly589Ser R623Qfs*20 Pro630Ala	1760C>T 1765G>A 1887dupC 1888C>G	错义突变       错义突变       移码突变       错义突变
39 40 41 42	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	del626-631GSGGPP Met1Thr Thr104Ile His105ThrfsX133	1986-2003del 2T>C 311C>T 313delC	缺失突变       错义突变       错义突变       缺失突变
43 44 45	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	del 1 bp codon 105 Gln107His Arg109Leu	313delC 321G>T 326G>T	缺失突变错义突变错义突变
46 47 48 49	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	His126Leu Leu131Pro Ser140Arg del 1 bp codon 144	377A>T 392T>C 420C>A 432delC	错义突变       错义突变       错义突变       缺失突变
50 51 52 53	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Ala150Thr del 2 bp codon 151 - Arg16Arg	448G>A 451_452delCT 478-2A>T 47G>A	错义突变       缺失突变       剪接突变       错义突变
54 55 56	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	del 3 bp codon 167 del 3 bp codon 167 Gly168Arg	499_501delTTC 500_502delTCG 502G>C	缺失突变错义突变
57 58 59 60	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	del 1 bp codon 168 Thr169Met L175Rfs*106 Trp176Arg	504delG 506C>T 515-525del11 526T>C	缺失突变       错义突变       缺失突变       错义突变
61 62 63	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	W176X Ala178Pro Ser182Arg	528G>A 532G>C 546C>A	错义突变       错义突变       错义突变
64 65 66 67	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Tyr184Cys Leu187Pro Gły189Arg Gły189Ala	551A>G 560T>C 565G>A 566G>C	错义突变       错义突变       错义突变       错义突变
68 69 70 71	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Arg190W Arg190Gln Arg192Cys Arg195Pro	568C>T 569G>A 574C>T 584G>C	错义突变       错义突变       错义突变       错义突变
72 73 74	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	del 1 bp codon 195 Lys196Thr Pro197Ser	585delG 587A>C 589C>T	缺失突变 错义突变 错义突变
75 76 77 78	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Asp202Asn Lys218Glu Gly219Glu Ser245Leu	604G>A 652A>G 656G>A 674C>T	错义突变       错义突变       错义突变
79 80 81	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	Ile227Leu Arg231Cys Leu236Gh	679A>C 691C>T 707T>A	错义突变 错义突变 错义突变
82 83 84 85	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Leu236Pro Arg243Cys Leu251Gln Val254Met	707T>C 727C>T 752T>A 760G>A	错义突变       错义突变       错义突变       错义突变
86 87 88	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	His258Leu Arg259Cys Thr265Ile	773A>T 775C>T 794C>T	<ul><li>错义突变</li><li>错义突变</li><li>错义突变</li></ul>
90 91 92	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Gly272Asp Leu273Phe del 3 bp codon 276 Ser277Leu	815G>A 817C>T 828_830delCTC 830C>G	错义突变       错义突变       缺失突变       错义突变
93 94 95 96	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	Ala302Val Trp305Term Trp305Leu	905C>T 914G>A 914G>T 916G>A	错义突变       错义突变       错义突变
96 97 98 99	KCNQ1 KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Gly306Arg Gly306Val - del 3 bp codon 312	917G>A 917G>T 921+1G>T 935_937delCCA	错义突变       错义突变       剪接突变       缺失突变
100 101 102 103	KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3 NM_000218.3	Thr312Ile Gly314Ser Gly314Cys Pro320Ser	935C>T 940G>A 940G>T 954G>C	错义突变       错义突变       错义突变       错义突变
104 105 106	KCNQ1 KCNQ1 KCNQ1	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2	NM_000218.3 NM_000218.3 NM_000218.3	Thr322Met Gly325Arg del 2 bp codon 333	965C>T 973G>A 998_999delCT	错义突变       错义突变       缺失突变
107 108 109 110	KCNQ1 SCN4B AKAP9	LQT1(RWS)/SIDS/JLN2 LQT1(RWS)/SIDS/JLN2 LQT10 LQT11	NM_000218.3 NM_000218.3 NM_005751	- del.exon3-10 Cys189Met Gln1300Argfs*12	c.477+5G>A del.exon3-10 566C>T 3897delA	剪接突变       缺失突变       错义突变       缺失突变
111 112 113 114	KCNJ5 KCNH2 KCNH2	LQT13 LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000890 NM_000238.4 NM_000238.4 NM_000238.4	Gly387Arg Arg366X Val374Phe Trp410Arg	1473G>C 1096C>T 1120G>T 1228T>C	错义突变       无义突变       错义突变       错义突变
115 116 117	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4	Leu413Pro del 14 bp codon 415 Ile42Asn	1238T>C 1245_1258delGCTGCTGGTC 125T>A	错义突变 移码突变 错义突变
118 119 120 121	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Thr443ProfsX78  del 2 bp / ins 1 bp codon 44  Glu444Asp  Leu457Pro	1332G>T 1370T>C	移码突变 移码突变 错义突变 错义突变
122 123 124 125	KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Phe463Leu Tyr475Cys Glu480Val 494delPhe	1387T>C 1424A>G 1439A>T 1480_1482delTTC	错义突变 错义突变 错义突变 缺失突变
126 127 128	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4	- Arg534Cys Val535Met	1557+1G>A 1600C>T 1603G>A	剪接突变 错义突变 错义突变
129 130 131 132	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	del 19 bp codon 540 Glu544Ala Leu559His Ala561Val	1619_1637delATCGCTACTC 1631A>C 1676T>A 1681C>T	错义突变       错义突变       错义突变
133 134 135 136	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Ala561Thr Gly572Ser Glu575Lys Pro596Ala	1681G>A 1714G>A 1723G>A 1786C>G	错义突变       错义突变       错义突变       错义突变
137 138 139	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4	Pro596Ser Gly601Ser Gly604Ser	1786C>T 1801G>A 1810G>A	错义突变       错义突变       错义突变
140 141 142 143	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Gly604Cys Asp609Gly Thr613Met Ala614Val	1810G>T 1826A>G 1838C>T 1841C>T	错义突变       错义突变       错义突变       错义突变
144 145 146 147	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Tyr616Term Ser620Ile ins 11 bp codon 628 Gly626Ser	1848C>A 1859G>T 1872_1882dupTGTGGGCTTC 1876G>A	无义突变 错义突变
148 149 150	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4	Phe627Leu Gly628Ser Asn629Thr	1879T>C 1882G>A 1886A>C	错义突变       错义突变       错义突变
151 152 153 154	KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Asn633Asp Asn633Ser Thr634Pro	1897A>G 1898A>G 1900A>C 1945+6T>C	错义突变       错义突变       错义突变       剪接突变
155 156 157	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4	Tyr652Term ins 2 bp codon 674 ins 1 bp codon 703 Glv71Trp	1956C>A 2021_2022insGA 2108dupA 211G>T	无义突变 移码突变 移码突变
158 159 160 161	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Pro72Thr Ile783Asn	2146-12_2146-2delCCCTGCC 214C>A 2344T>A	错义突变 错义突变
162 163 164 165	KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Arg784Trp Arg797Thr Phe805Ser Ser818Leu	2350C>T 2389G>A 2414T>C 2453C>T	错义突变       错义突变       错义突变       错义突变
166 167 168	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4	Arg823Trp Asp837Tyr Arg863X	2467C>T 2509G>T 2587C>T	错义突变       错义突变       错义突变
169 170 171 172	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Gły873AlafsX5 Asp896ArgfsX77 Lys897Thr Arg912Trp	2616delC 2682_2685delCACG 2690A>C 2734C>T	移码突变 缺失突变 错义突变 错义突变
173 174 175 176	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	del 1 bp codon 923 Pro926Ser Pro926AlaX14 Cys984ThrfsX75	2768delC 2776C>T 2776insG 2944_2948dupGACAC	移码突变       错义突变       插入突变       移码突变
177 178 179	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4	Leu987Valfs131X Ala1017ProfsX40 Asp102His	2959-2960delCT 3048delC 304G>C	缺失突变缺失突变错义突变
180 181 182 183	KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	1031Gly+87X del 14 bp codon 1032 Arg1035TrpfsX20 Glu1039Term	3094insC 3096_3109delGCGGCCCCGC 3102_3111delCCGGGGCGAC 3115G>T	缺失突变 无义突变
184 185 186	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4	Gln1068Term Pro1075Leu deletion 28 bp, c.381_408	3202C>T 3224C>T 381_408del28	无义突变 错义突变 缺失突变
187 188 189 190	KCNH2 KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Trp154X del 1 bp codon 16 Ala169Gly ins 6 bp codon 172	461G>A 46delG 506C>G 508_513dupCTGCTG	无义突变       移码突变       错义突变       移码突变
191 192 193 194	KCNH2 KCNH2	LQT2/SIDS LQT2/SIDS LQT2/SIDS LQT2/SIDS	NM_000238.4 NM_000238.4 NM_000238.4 NM_000238.4	Pro191ArgX10 del 1 bp codon 191 Glu229X Gln247Term	572delC 572delC 685G>T 739C>T	缺失突变       移码突变       错义突变       无义突变
195 196 197	KCNH2 SCN5A SCN5A	LQT2/SIDS LQT3/SIDS LQT3/SIDS	NM_000238.4 NM_198056.3 NM_198056.3	Ala32Thr - Val411Met	94G>A 1141-3C > A 1231C>T	<ul><li>错义突变</li><li>剪接突变</li><li>错义突变</li></ul>
198 199 200 201	SCN5A SCN5A	LQT3/SIDS LQT3/SIDS LQT3/SIDS LQT3/SIDS	NM_198056.3 NM_198056.3 NM_198056.3 NM_198056.3	Gly615Glu Leu618Phe Phe1250Leu Asn1325Ser	1844G>A 1852C>T 3748T>C 3974A>G	错义突变       错义突变       错义突变       错义突变
202 203 204	SCN5A SCN5A SCN5A	LQT3/SIDS LQT3/SIDS LQT3/SIDS	NM_198056.3 NM_198056.3 NM_198056.3	Phe1473Ser Gln1475Pro del 9 bp codon 1504	4418T>C 4424A>C 4519_4527delCAGAAGCCC	错义突变       错义突变       缺失突变
205 206 207 208	SCN5A SCN5A SCN5A	LQT3/SIDS LQT3/SIDS LQT3/SIDS LQT3/SIDS	NM_198056.3 NM_198056.3 NM_198056.3 NM_198056.3	Pro155Thr Leu1608Pro Gly1631Asp Arg1644His	463G>T 4823T>C 4892G>A 4931G>A	错义突变       错义突变       错义突变       错义突变
209 210 211	SCN5A SCN5A SCN5A	LQT3/SIDS LQT3/SIDS LQT3/SIDS	NM_198056.3 NM_198056.3 NM_198056.3	Met1652Arg Val1763Met Glu1784Lys	4955T>G 5287G>A 5350G>A	错义突变       错义突变       错义突变
212 213 214 215	SCN5A SCN5A ANK2	LQT3/SIDS LQT3/SIDS LQT4	NM_198056.3 NM_198056.3 NM_198056.3 NM_001148.6	del 3 bp codon 1790 1790delAsp Gly213Val Gly1439Cys	5368_5370delGAC 5468_5470delGAC 638G>T 4315G>T	缺失突变       缺失突变       错义突变
216 217 218 219	KCNE1 KCNE1 KCNE1	LQT5/JLN2 LQT5/JLN2 LQT5/JLN2 LQT7 (ATS)	NM_000219.6 NM_000219.6 NM_000219.6 NM_000891.3	Gly52Arg Lys70Asn Tyr81Cys Val77Glu	154G>A 210G>C 242A>G 230T>A	错义突变       错义突变       错义突变
220 221 222	KCNJ2 KCNJ2 KCNJ2	LQT7 (ATS) LQT7 (ATS) LQT7 (ATS)	NM_000891.3 NM_000891.3 NM_000891.3	Pro186Thr Arg189Ser Thr192Ile	556C>A 567A>T 575C>T	错义突变       错义突变       错义突变
223 224 225 226	KCNJ2 KCNJ2 KCNJ2	LQT7 (ATS) LQT7 (ATS) LQT7 (ATS) LQT7 (ATS)	NM_000891.3 NM_000891.3 NM_000891.3 NM_000891.3	G1y215Asp Glu299Gly Met307Val Met307Ile	644G>A 896A>G 919A>G 921G>C	错义突变       错义突变       错义突变       错义突变
226 227 228 229	CACNA1C	LQT8 (TS ) LQT8 (TS )	NM_000891.3 NM_000719.7 NM_000719.7 NM_000719.7	Met307lle Gly406Arg Gly419Arg Met456Ile	921G>C 1216G>A 1255G>A 1368G>A	错义突变       错义突变       错义突变