

序号	突变基因	临床表型	mRNA参考模板	碱基改变	氨基酸改变	突变类型
1	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Phe340Leu	1020T>G	错义突变
2	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ala341Glu	1022C>A	错义突变
3	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ala341Val	1022C>T	错义突变
4	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ser373Trpfs*10	1033-1-1117dup	移码突变
5	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly345Glu	1034G>A	错义突变
6	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly345Ala	1034G>C	错义突变
7	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Leu347Pro	1040T>C	错义突变
8	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gln357Arg	1070A>G	错义突变
9	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	360_361dupKQ	1071_1076dupGAAGCA	移码突变
10	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	ins 6 bp codon 359	1073_1078dupAGCAGA	插入突变
11	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg366Trp	1096C>T	错义突变
12	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Leu374Arg	1121T>G	错义突变
13	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg380Gly	1138A>G	错义突变
14	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	ins 1 bp codon 383	1148dupC	移码突变
15	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	ins 1 bp codon 384	1149dupT	移码突变
16	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	T391I	1172C>T	错义突变
17	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	356-357delGln	1176-1181del	错义突变
18	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 1 bp codon 396	1189delC	缺失突变
19	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Val440AlafsX26	1319delT	错义突变
20	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly45Ser	133G>A	错义突变
21	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	ins 1 bp codon 447	1343dupC	移码突变
22	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	His455Tyr	1363C>T	错义突变
23	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3		1394-1G>T	剪接突变
24	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Pro448Arg	1453C>G	错义突变
25	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Val518Leu	1546G>A	错义突变
26	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg518X	1552C>T	错义突变
27	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg519His	1556G>A	错义突变
28	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg533Trp	1597C>T	错义突变
29	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg539Trp	1615C>T	错义突变
30	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gln560Lys	1678C>A	错义突变
31	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg562Ser	1686G>T	错义突变
32	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly568Ala	1703G>C	错义突变
33	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Lys581Term	1741A>T	无义突变
34	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg583His	1748G>A	错义突变
35	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Thr587Met	1760C>T	错义突变
36	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly589Ser	1765G>A	错义突变
37	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	R623Qfs*20	1887dupC	移码突变
38	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Pro630Ala	1888C>G	错义突变
39	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del626-631GSGGPP	1986-2003del	缺失突变
40	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Met1Thr	2T>C	错义突变
41	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Thr104Ile	311C>T	错义突变
42	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	His105ThrfsX133	313delC	缺失突变
43	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 1 bp codon 105	313delC	缺失突变
44	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gln107His	321G>T	错义突变
45	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg109Leu	326G>T	错义突变
46	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	His126Leu	377A>T	错义突变
47	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Leu131Pro	392T>C	错义突变
48	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ser140Arg	420C>A	错义突变
49	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 1 bp codon 144	432delC	缺失突变
50	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ala150Thr	448G>A	错义突变
51	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 2 bp codon 151	451_452delCT	缺失突变
52	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	-	478-2A>T	剪接突变
53	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg16Arg	47G>A	错义突变
54	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 3 bp codon 167	499_501delITTC	缺失突变
55	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 3 bp codon 167	500_502delITTC	缺失突变
56	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly168Arg	502G>C	错义突变
57	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 1 bp codon 168	504delG	缺失突变
58	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Thr169Met	506C>T	错义突变
59	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	L175Rfs*106	515-525del11	缺失突变
60	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Trp176Arg	526T>C	错义突变
61	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	W176X	528G>A	错义突变
62	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ala178Pro	532G>C	错义突变
63	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ser182Arg	546C>A	错义突变
64	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Thr184Cys	551A>G	错义突变
65	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Leu187Pro	560T>C	错义突变
66	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly189Arg	565G>A	错义突变
67	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly189Ala	566G>C	错义突变
68	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg190W	568C>T	错义突变
69	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg190Gln	569G>A	错义突变
70	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg192Cys	574C>T	错义突变
71	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg195Pro	584G>C	错义突变
72	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 1 bp codon 195	585delG	缺失突变
73	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Lys196Thr	587A>C	错义突变
74	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Pro197Ser	589C>T	错义突变
75	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Asp202Asn	604G>A	错义突变
76	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Lys218Glu	652A>G	错义突变
77	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly219Glu	656G>A	错义突变
78	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ser245Leu	674C>T	错义突变
79	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ile227Leu	679A>C	错义突变
80	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg231Cys	691C>T	错义突变
81	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Leu236Gln	707T>A	错义突变
82	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Leu236Pro	707T>C	错义突变
83	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg243Cys	727C>T	错义突变
84	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Leu251Gln	752T>A	错义突变
85	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Val254Met	760G>A	错义突变
86	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	His258Leu	775A>T	错义突变
87	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Arg259Cys	773C>T	错义突变
88	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Thr265Ile	794C>T	错义突变
89	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly272Asp	815G>A	错义突变
90	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Leu273Phe	817C>T	错义突变
91	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 3 bp codon 276	828_830delCTC	缺失突变
92	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ser277Leu	830C>G	错义突变
93	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Ala302Val	905C>T	错义突变
94	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Trp305Term	914G>A	错义突变
95	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Trp305Leu	914G>T	错义突变
96	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly306Arg	916G>A	错义突变
97	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly306Val	917G>T	错义突变
98	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	-	921+1G>T	剪接突变
99	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 3 bp codon 312	935_937delCCA	缺失突变
100	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Thr312Ile	935C>T	错义突变
101	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly314Ser	940G>A	错义突变
102	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly314Cys	940G>T	错义突变
103	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Pro320Ser	954G>C	错义突变
104	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Thr322Met	965C>T	错义突变
105	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	Gly325Arg	973G>A	错义突变
106	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del 2 bp codon 333	998_999delCT	缺失突变
107	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	-	c.477+5G>A	剪接突变
108	KCNQ1	LQT1(RWS)/SIDS/JLN2	NM_000218.3	del.exon3-10	del.exon3-10	缺失突变
109	SCN4B	LQT10		Cys189Met	566C>T	错义突变
110	AKAP9	LQT11	NM_005751	Gln1300Argfs*12	3897delA	缺失突变
111	KCNJ5	LQT13	NM_000890	Gly387Arg	1473G>C	错义突变
112	KCNH2	LQT2/SIDS	NM_000238.4	Arg366X	1096C>T	无义突变
113	KCNH2	LQT2/SIDS	NM_000238.4	Val374Phe	1120G>T	错义突变
114	KCNH2	LQT2/SIDS	NM_000238.4	Trp410Arg	1228T>C	错义突变
115	KCNH2	LQT2/SIDS	NM_000238.4	Leu413Pro	1238T>C	错义突变
116	KCNH2	LQT2/SIDS	NM_000238.4	del 14 bp codon 415	1245_1258delGCTGCTGGTC	移码突变
117	KCNH2	LQT2/SIDS	NM_000238.4	Ile42Asn	125T>A	错义突变
118	KCNH2	LQT2/SIDS	NM_000238.4	Thr443ProfsX78	1326delT	移码突变
119	KCNH2	LQT2/SIDS	NM_000238.4	del 2 bp / ins 1 bp codon 441	1328_1329delCCinsG	移码突变
120	KCNH2	LQT2/SIDS	NM_000238.4	Glu444Asp	1332G>T	错义突变
121	KCNH2	LQT2/SIDS	NM_000238.4	Leu457Pro	1370T>C	错义突变
122	KCNH2	LQT2/SIDS	NM_000238.4	Phe463Leu	1387T>C	错义突变
123	KCNH2	LQT2/SIDS	NM_000238.4	Tyr475Cys	1424A>G	错义突变
124	KCNH2	LQT2/SIDS	NM_000238.4	Glu480Val	1439A>T	错义突变
125	KCNH2	LQT2/SIDS	NM_000238.4	494delPhe	1480_1482delITTC	缺失突变
126	KCNH2	LQT2/SIDS	NM_000238.4	-	1557+1G>A	剪接突变
127	KCNH2	LQT2/SIDS	NM_000238.4	Arg534Cys	1600C>T	错义突变
128	KCNH2	LQT2/SIDS	NM_000238.4	Val535Met	1603G>A	错义突变
129	KCNH2	LQT2/SIDS	NM_000238.4	del 19 bp codon 540	1619_1637delATCGCTACTC	移码突变
130	KCNH2	LQT2/SIDS	NM_000238.4	Glu544Ala	1631A>C	错义突变
131	KCNH2	LQT2/SIDS	NM_000238.4	Leu559His	1676T>A	错义突变
132	KCNH2	LQT2/SIDS	NM_000238.4	Ala561Val	1681C>T	错义突变
133	KCNH2	LQT2/SIDS	NM_000238.4	Ala561Thr	1681G>A	错义突变
134	KCNH2	LQT2/SIDS	NM_000238.4	Gly572Ser	1714G>A	错义突变
135	KCNH2	LQT2/SIDS	NM_000238.4	Glu575Lys	1723G>A	错义突变
136	KCNH2	LQT2/SIDS	NM_000238.4	Pro596Ala	1786C>G	错义突变
137	KCNH2	LQT2/SIDS	NM_000238.4	Pro596Ser	1786C>T	错义突变
138	KCNH2	LQT2/SIDS	NM_000238.4	Gly601Ser	1801G>A	错义突变
139	KCNH2	LQT2/SIDS	NM_000238.4	Gly604Ser	1810G>A	错义突变
140	KCNH2	LQT2/SIDS	NM_000238.4	Gly604Cys	1810C>T	错义突变
141	KCNH2	LQT2/SIDS	NM_000238.4	Asp609Gly	1826A>G	错义突变
142	KCNH2	LQT2/SIDS	NM_000238.4	Thr613Met	1838C>T	错义突变
143	KCNH2	LQT2/SIDS	NM_000238.4	Ala614Val	1841C>T	错义突变
144	KCNH2	LQT2/SIDS	NM_000238.4	Tyr616Term	1848C>A	无义突变
145	KCNH2	LQT2/SIDS	NM_000238.4	Ser620Ile	1859G>T	错义突变
146	KCNH2	LQT2/SIDS	NM_000238.4	ins 11 bp codon 628	1872_1882dupTGTGGGCTTC	移码突变
147	KCNH2	LQT2/SIDS	NM_000238.4	Gly626Ser	1876G>A	错义突变
148	KCNH2	LQT2/SIDS	NM_000238.4	Phe627Leu	1879T>C	错义突变
149	KCNH2	LQT2/SIDS	NM_000238.4	Gly628Ser	1882G>A	错义突变
150	KCNH2	LQT2/SIDS	NM_000238.4	Asn629Thr	1886A>C	错义突变
151	KCNH2	LQT2/SIDS	NM_000238.4	Asn633Asp	1897A>G	错义突变
152	KCNH2	LQT2/SIDS	NM_000238.4	Asn633Ser	1898A>G	错义突变
153	KCNH2	LQT2/SIDS	NM_000238.			