S1 Table. SLC12A3 mutations identified in patients with Gitelman syndrome.

Patient No.	Nationality	Nucleotide change	Predicted protein change	Reference
	Homo (n=212): Europe (n=122) + North America (n=3) + Asia (n=87)			
1	Japan	c. 804_805ins ATTGGCGTGGTCTCGGTC	NA	[1]
2	Japan	c. 2573T>A	p. Leu858His	[2]
3	Japan	c. 2891G>A	p. Arg964Gln	[2]
4	Japan	c. 179C>T	p. Thr60Met	[2]
5	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
6	Japan	c. 2573T>A	p. Leu858His	[2]
7	Japan	c. 1262G>T	p. Cys421Phe	[2]
8	Japan	c. 539C>A	p. Thr180Lys	[2]
9	Japan	c. 539C>A	p. Thr180Lys	[2]
10	Japan	c. 539C>A	p. Thr180Lys	[2]
11	Japan	c. 2573T>A	p. Leu858His	[2]
12	Japan	c. 3052C>T	p. Arg1018*	[2]
13	Japan	c. 1868T>C	p. Leu623Pro	[2]
14	Japan	c. 539C>A	p. Thr180Lys	[2]
15	Japan	c. 1664C>T	p. Ser555Leu	[2]
16	Japan	c. 1844C>T	p. Ser615Leu	[2]
17	Japan	c. 2891G>A	p. Arg964Gln	[2]
18	Japan	c. 2573T>A	p. Leu858His	[2]
19	Japan	c. 2573T>A	p. Leu858His	[2]
20	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
21	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
22	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
23	Japan	c. 3052C>T	p. Arg1018*	[2]
24	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
25	Japan	c. 1271G>A	p. Gly424Asp	[2]
26	Japan	c. 1930delC	p. Gln644Serfs*28	[2]
27	Japan	c. 2191G>A	p. Gly731Arg	[2]
28	Japan	c. 2573T>A	p. Leu858His	[2]
29	Japan	c. 2573T>A	p. Leu858His	[2]
30	Japan	c. 2573T>A	p. Leu858His	[2]
31	Japan	c. 2927G>T	p. Ser976Phe	[2]
32	Japan	c. 2573T>A	p. Leu858His	[2]

33	Japan	c. 3052C>T	p. Arg1018*	[2]
34	Japan	c. 2573T>A	p. Leu858His	[2]
35	Japan	c. 1336-2A > T	p. T446LfsX6	[3]
36	Japan	c. 2573T>A	p. Leu858His	[4]
37	Japan	NA	p. Thr180Lys	[5]
38	Japan	c. 788 ins18bp	In frame insertion	[6]
39	Japan	c. 2552T>A	p. Leu849His	[7]
40	Japan	c. CGG>CAG	p. Arg955Gln	[8]
41	Japan	c. CGG>CAG	p. Arg955Gln	[8]
42	Japan	c. CGG>CAG	p. Arg955Gln	[8]
43	China	c. 2039delG	p. Gly680Asp	[9]
44	China	c. 1562_1564delTCA	p. 522delIle	[10]
45	China	c. 841T>C	p. Trp281Arg	[11]
46	China	c. 1196G>C	p. Arg399Pro	[12]
47	China	c. 2099T>C	p. Leu700Pro	[13]
48	China	c. 2099T>C	p. Leu700Pro	[13]
49	China	c. 2687 G>A	p. Arg896Gln	[14]
50	China	NA	NA	[15]
51	China	NA	p. Asp486Asn	[16]
52	China	c. 179C>T	p. Thr60Met	[16]
53	China	c. 179C>T	p. Thr60Met	[16]
54	China	c. 179C>T	p. Thr60Met	[17]
55	China	c. 488C>T	p. Thr163Met	[18]
56	China	c. 488C>T	p. Thr163Met	[18]
57	China	c. 488C>T	p. Thr163Met	[18]
58	China	c. C2129A	p. Ser710Term	[19]
59	China	c. C2129A	p. Ser710Term	[19]
60	China	NA	p. Arg655His	[20]
61	China	c. 179C>T	p. Thr60Met	[20]
62	China	c. 179C>T	p. Thr60Met	[20]
63	China	c. 185C>T	p. Thr60Met	[21]
64	China	c. 1970G>A	p. Arg655His	[21]
65	China	c. 1163A>G	p. Tyr386Cys	[21]
66	China	c. 185C>T	p. Thr60Met	[22]
67	China	c. 185C>T	p. Thr60Met	[22]
68	China	c. 185C>T	p. Thr60Met	[22]
69	China	c. 1456G>A	p. Asp486Asn	[23]
70	China	c. 179C>T	p. Thr60Met	[23]
71	China	c. 179C>T	p. Thr60Met	[23]
72	China	c. 48C>A	p. Cys16X	[23]
		c. 602G>A	p. Gly201Asp	
		c. 391G>A	p. Glu131Lys	
73	Korea	c. 179C>T	p. Thr60Met	[24]

74	Korea	c. 1216A>C	p. Asn406His	[25]
75	Korea	c. 1216A>C	p. Asn406His	[25]
76	Korea	c. 1706C>T	p. Ala569Val	[25]
77	Korea	c. 2099T>C	p. Leu700Pro	[25]
78	Korea	c. 2359C>T	p. Gln787*	[25]
79	Korea	c. 2738G>A	p. Arg913Gln	[25]
80	Korea	c. 2927C>T	p. Ser976Phe	[25]
81	Korea	c. 2927C>T	p. Ser976Phe	[26]
82	Korea	NA	p. Gln95del	[26]
83	Korea	NA	p. Ser967Phe	[27]
84	Romania	c. 1805_1806delAT	p. Tyr602Cysfs*31	[28]
85	Romania	c. 2660+1G>A	NA	[28]
86	UK	c. 2878_2879ins AGGGGTGCACCCTG	p. Val960Glufs*12	[29]
87	UK	c. 2221G>A	p. Gly741Arg	[29]
88	UK	c. 1195C>T	p. Arg399Cys	[30]
89	UK	c. 2221G>A	p. Gly741Arg	[30]
90	UK	c. 2221G>A	p. Gly741Arg	[30]
91	UK	c. 2687G>A	p. Arg896Gln	[31]
92	UK	c. 2221G>A	p. Gly741Arg	[32]
93	UK	c. CCC>CTC	p. Pro349Leu	[8]
94	UK	c. CCC>CTC	p. Pro349Leu	[8]
95	CA	c. 533C>T	p. Ser178Leu	[33]
96	CA	c. 533C>T	p. Ser178Leu	[33]
97	CA	c. 1704C>A	p. Pro560His	[34]
98	Sri Lanka	c. 1276A>T	p. Asn426Tyr	[35]
99	Turkey	c. 1145C>T	p. Thr382Met	[36]
100	Turkey	c. 602-16G>A	SP	[37]
101	France	c. 2221G>A	p. Gly741Arg	[38]
102	France	c. 2221G>A	p. Gly741Arg	[38]
103	France	c. 1180+1G>T	SP	[38]
104	France	c. 1180+1G>T	SP	[38]
105	France	c. 1180+1G>T	SP	[39]
106	France	c. 1180+1G>T	SP	[39]
107	France	c. 1925G>A	p. Arg642His	[39]
108	Italy	c. 1095+1G>A	NA	[40]
109	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
110	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
111	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
112	Italy	c. 1180+1G>T	SP	[42]
113	Italy	c. 531-2A>G	SP	[43]
114	Italy	c. 2897A>G	p. Arg958Gly	[43]
115	Italy	c. 539T>C	p. Trp172Arg	[43]

116	Italy	c. 1949C>G	p. Arg642Gly	[43]
117	Italy	c. 2736G>A	p. Arg904Gln	[44]
118	Italy	c. 45_46del2-bp	Complete loss of TSC-protein	[45]
119	Italy	c. 45_46del2-bp	Complete loss of TSC-protein	[45]
120	Italy	NA	p. Asp62Asn	[45]
121	Greece	c. 2538G>A	p. W844stop	[46]
122	Spain	c. 361G>T	p. Glu121Ter	[47]
123	Spain	c. 1939G>A	p. Val647Met	[48]
124	Spain	c. 1939G>A	p. Val647Met	[48]
125	Spain	c. 1939G>A	p. Val647Met	[48]
126	Spain	c. 1939G>A	p. Val647Met	[48]
127	Spain	c. 1939G>A	p. Val647Met	[48]
128	Spain	c. 1939G>A	p. Val647Met	[48]
129	Spain	c. 1925G>A	p. Arg642His	[49]
130	Spain	c. 1925G>A	p. Arg642His	[49]
131	Spain	G to A transition at position +1 of the donor splice site	NA	[50]
132	Spain	intron 9 +1 G>T	NA	[51]
133	Spain	intron 9 +1 G>T	NA	[51]
134	Spain	intron 9 +1 G>T	NA	[51]
135	Spain	intron 9 +1 G>T	NA	[51]
136	Spain	intron 9 +1 G>T	NA	[51]
137	Spain	intron 9 +1 G>T	NA	[51]
138	Spain	intron 9 +1 G>T	NA	[51]
139	Spain	intron 9 +1 G>T	NA	[51]
140	Spain	intron 9 +1 G>T	NA	[51]
141	Spain	intron 9 +1 G>T	NA	[51]
142	Spain	intron 9 +1 G>T	NA	[51]
143	Spain	intron 9 +1 G>T	NA	[51]
144	Spain	intron 9 +1 G>T	NA	[51]
145	Spain	intron 9 +1 G>T	NA	[51]
146	Spain	intron 9 +1 G>T	NA	[51]
147	Spain	intron 9 +1 G>T	NA	[51]
148	Spain	intron 9 +1 G>T	NA	[51]
149	Spain	intron 9 +1 G>T	NA	[51]
150	Spain	intron 9 +1 G>T	NA	[51]
151	Spain	intron 9 +1 G>T	NA	[51]
152	Spain	c. CTT>CCT	p. Leu850Pro	[8]
153	Spain	c. CTT>CCT	p. Leu850Pro	[8]
154	Spain	c. CTT>CCT	p. Leu850Pro	[8]
155	Netherlands	c. 602-16G>A	SP	[52]
156	Netherlands	c. 1420C>T	p. Thr465Thr	[34]
		c. 669T>C	p. Leu215Pro	

		c. 2736G>A	p. Arg904Gln	
		c. 2881+1G>T	SP	
157	Netherlands	c. 1971C>G	p. Thr649Arg	[34]
		c. 701G>A	p. Ala226Thr	
158	Netherlands	c. 2927C >T	p. Arg968Stop	[34]
159	Netherlands	c. 2927C>T	p. Arg968Stop	[34]
		c. 2780C>T	p. Arg919Cys	
160	Belgium	NA	p. Arg399Cys	[53]
161	India	c. 2879_2883+9 ins14bp	p. Val 960 Glu fsx12	[54]
162	Germany	NA	p. Val647Met	[55]
163	Germany	NA	p. Val647Met	[55]
164	Germany	c. 260_263insCC	frameshift Pro79 introducing stop 35 codons downstream	[34]
165	Germany	c. 1988C>T	p. Arg655Cys	[34]
166	Sweden	c. 2859+1G>T	SP	[56]
167	Sweden	c.2667+1T>G	SP	[56]
168	Sweden	c.2859+1G>T	SP	[56]
169	Sweden	c. 1569-1G>A	SP	[56]
170	Sweden	c. 695G>A	p. Gly230Asp	[56]
171	Sweden	c.2859+1G>T	SP	[56]
172	Czech Republic	c. 1315G>A	p. Gly439Ser	[57]
173	Czech Republic	c. 790G>C	p. Gly264Arg	[57]
174	Portugal	c. 650C>T	p. Arg209Trp	[34]
175	Europe[1]	c. 1925G>A	p. Arg642His	[58]
176	Europe[1]	c. 514T>C	p. Trp172Arg	[58]
177	Europe[1]	c. 2883+1G>T	SP	[58]
178	Europe[1]	c. 2089_2095del	p. Thr697fs	[58]
179	Europe[1]	c. 3053G>A	p. Arg1018Gln	[58]
180	Europe[1]	c. 2965G>A	p. Gly989Arg	[58]
181	Europe[1]	c. 2981G>A	p. Cys994Tyr	[58]
182	Europe[1]	c. 1195C>T	p. Arg399Cys	[58]
183	Europe[1]	c. 2089_2095del	p. Thr697fs	[58]
184	Europe[1]	c. 1196_1202dup	p. Ser402X	[58]
185	Europe[1]	c. 1180+1G>T	SP	[58]
186	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
187	Europe[1]	c. 1387G>A	p. Gly463Arg	[58]
188	Europe[1]	c. 237_238dup	p. Arg80fs	[58]
189	Europe[1]	c. 506-1G>A	SP	[58]
190	Europe[1]	c. 602-?_852+?dell	p. Gly502fs	[58]

191	Europe[1]	c. 488C>T	p. Thr163Met	[58]
192	Europe[1]	c. 1928C>T	p. Pro643Leu	[58]
193	Europe[1]	c. 2572C>T	p. Leu858Phe	[58]
194	Europe[1]	c. 1096-1G>A	SP	[58]
195	Europe[1]	c. 2089_2095del	p. Thr697fs	[58]
196	Europe[1]	c. 2883+1G>T	SP	[58]
197	Europe[1]	c. 910A>C	p. Thr304Pro	[58]
198	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
199	Europe[1]	c. 2883+1G>T	SP	[58]
200	Europe[1]	c. 2548G>C	p. Gly850Arg	[58]
201	Europe[1]	c. 247C>T	p. Arg83Trp	[58]
202	Europe[1]	c. 1928C>T	p. Pro643Leu	[58]
203	Europe[1]	c. 2981G>A	p. Cys994Tyr	[58]
204	Europe[1]	c. 1315G>A	p. Gly439Ser	[58]
205	Europe[1]	c. 1939G>A	p. Val647Met	[58]
206	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
207	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
208	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
209	Europe[1]	c. 1679C>G	p. Pro560Arg	[58]
210	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
211	Europe[1]	c. 3053G>A	p. Arg1018Cys	[58]
212	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
Co-homo (n=6): Europe (n=3) + Asia (n=3)				
213	China	c. C2782T	p. Arg928Cys	[19]
		c. C1395T	p. Thr465Thr	
		c. A366G	p. Ala122Ala	
214	China	NA	p. Thr60Met	[20]
		NA	p. Arg655His	
215	China	c. 179C>T	p. Thr60Met	[23]
		c. 1964G>A	p. Arg655His	
216	Czech Republic	c. 1315G>A	p. Gly439Ser	[57]
		c. 2711G>A	p. Arg904Gln	
217	Czech Republic	c. 791G>C	p. Gly264Ala	[57]
		c. 2711G>A	p. Arg904Gln	
218	Europe[1]	c. 575T>C	p. Ile192Thr	[58]
		c. 1698_1700del	p. Asn566del	
Hetero (n=131): Europe (n=57) + North America (n=11) + Asia (n=63)				
219	China	c. 2582G>A	p. Arg861His	[59]
220	China	NA	p. Asn359Lys	[60, 61]
221	China	c. 2816G>A	p. Trp939X	[62]
222	China	c.486_490delinsA	p. Thr163fs	[62]
223	China	c. 836T>G	p. Met279Arg	[63]
224	China	c. 545C>A	p. Thr180Lys	[64]

225	China	c. 1844C>T	p. Ser615Leu	[16]
226	China	NA	p. Asp486Asn	[16]
227	China	NA	p. Gly196Val R959frameshift	[16]
-----228	China	c. 486-490del TACGGinsA	p. 162frameshift	[65]
229	China	NA	p. Leu858His	[17]
230	China	NA	p. Leu671Pro	[17]
231	China	c. C2782T	p. Arg928Cys	[19]
232	China	c. C2782T	p. Arg928Cys	[19]
233	China	c. C2782T	p. Arg928Cys	[19]
234	China	c. C2782T	p. Arg928Cys	[19]
235	China	c. C2782T	p. Arg928Cys	[19]
236	China	c. C2782T	p. Arg928Cys	[19]
237	China	c. C2782T	p. Arg928Cys	[19]
238	China	c. C2782T	p. Arg928Cys	[19]
239	China	c. C2129A	p. Ser710Term	[19]
240	China	c. C2129A	p. Ser710Term	[19]
241	China	c. C2129A	p. Ser710Term	[19]
242	China	c. C2129A	p. Ser710Term	[19]
243	China	c. C2129A	p. Ser710Term	[19]
244	China	c. C2129A	p. Ser710Term	[19]
245	China	c. C2129A	p. Ser710Term	[19]
246	China	NA	p. Asn566Lys	[20]
247	China	c. 185C>T	p. Thr60Met	[21]
248	China	c. 2761C>T	p. Arg919Cys	[21]
249	China	c. 1462G>A	p. Asp486Asn	[21]
250	China	IVS16-2A>G	SP	[21]
251	China	c. 185C>T	p. Thr60Met	[22]
252	China	c. 1844C>T	p. Ser615Leu	[23]
253	China	c.1456G>A	p. Asp486Asn	[23]
254	China	c. 1698C>A	p. Asn566Lys	[23]
255	China	c. 1456G>A	p. Asp486Asn	[23]
256	China	c. 1964G>T	p. Arg655Leu	[23]
257	China	c. 2738G>A	p. Arg913Gln	[23]
258	China	c. 2738G>A	p. Arg913Gln	[23]
259	China	c. 2782C>T	p. Arg928Cys	[23]
260	China	c. 1456G>A	p. Asp486Asn	[23]
261	China	c. 2782C>T	p. Arg928Cys	[23]
262	China	c. 1456G>A	p. Asp486Asn	[23]
263	China	c. 806 insTTGGCGT GGTCTCGGTCA	p. 269 ins IGVVSV	[23]
264	Korea	c. 2716C>T	p. Pro906Ser	[66]

265	Korea	c. 961C>T	p. Arg321Trp	[25]
266	Korea	c. 964+1G>T	NA	[25]
267	Korea	c. 1077C>G	p. Asn359Lys	[25]
268	Korea	c. 1667C>T	p. Pro556Leu	[25]
269	Korea	c. 1732G > A	p. Val578Met	[25]
270	Korea	c. 2369-4G>A	NA	[25]
271	Korea	c. 2660+1delG	NA	[25]
272	Korea	c. 539C>A	p. Thr180Lys	[25]
273	Korea	c. 781C>T	p. Arg261Cys	[67]
274	Korea	c. 781C>T	p. Arg261Cys	[67]
275	Korea	NA	p. Thr180Lys	[26]
276	Korea	c. 2666+1delG	NA	[26]
277	Korea	NA	p. Gln95del	[26]
278	Japan	c. 3052C>T	p. Arg1018X	[68]
279	Japan	c. 545C>A	p. Thr180Lys	[69]
280	Japan	c. 1712C>T	p. Ala569Val	[70]
281	Japan	c. 1A>T	p. Met1Leu	[71]
282	Italy	c. 1415G>A	p. Ala464Thr	[43]
283	Italy	c. 2579C>T	p. Arg852Cys	[43]
284	Italy	c. 2979G>A	p. Cys985Tyr	[43]
285	Italy	c. 2144_2158delGCC TTCTACTCGGATinsTG	p. Ala707fsX729	[43]
286	Italy	c. 1413G>A	p. Gly463Glu	[43]
287	Italy	c. 1413G>A	p. Gly463Glu	[43]
288	Italy	c. 513C>T	p. Thr163Met	[43]
289	Italy	c. 513C>T	p. Thr163Met	[43]
290	Italy	c. 1869C>G	p. Ser615Trp	[43]
291	Italy	NA	p. Cys13Thr	[45]
292	Italy	c. 263insCC	NA	[45]
293	Italy	c. 263insCC	NA	[45]
294	Italy	NA	p. Arg209Trp	[45]
295	Italy	NA	p. Leu542Pro	[45]
296	Italy	c. 2320del1bp	NA	[45]
297	Germany	NA	p. Asp486Asn	[55]
298	Germany	c. 807G>A	p. Arg261His	[34]
299	Germany	c. 1950G>A	p. Arg642His or SP	[34]
300	Germany	c. 2574>C	p. Leu850Pro	[34]
301	France	c. 1390G>A	p. Ala464Thr	[39]
302	France	c. 2782C>T	p. Arg928Cys	[39]
303	France	c. 791G>C	p. Gly264Ala	[39]
304	France	c. 791G>C	p. Gly264Ala	[39]
305	France	c. 791G>C	p. Gly264Ala	[39]

306	France	c. 791G>C	p. Gly264Ala	[39]
307	France	c. 2327/2328delA	truncated SLC12A3 protein, frameshift Phe767 introducing stop at 6 codons downstream	[34]
308	Sweden	c. 2987G>A	p. Cys994Tyr	[56]
309	Czech Republic	c. 1315G>A	p. Gly439Ser	[57]
310	Czech Republic	c. 238ins.CC	Pro79fsX35	[57]
311	Czech Republic	c. 238ins.CC	Pro79fsX35	[57]
312	Czech Republic	c. 2711G>A	p. Arg904Gln	[57]
313	Ca	c. 260_263insCC	frameshift Pro79 introducing stop 35 codons downstream	[34]
314	Ca	c. 1862G>A	p. Gly613Ser	[34]
315	Ca	c. GAC>AAC	p. Asp486Asn	[8]
316	United States	c. CGC>CAC	p. Arg655His	[8]
317	United States	c. CGC>CTC	p. Arg653Leu	[8]
318	United States	c. CGC>CTC	p. Arg653Leu	[8]
319	United States	c. GGT>GTT	5′ SP	[8]
320	United States	c. GGT>GTT	5′ SP	[8]
321	United States	c. GGT>GTT	5′ SP	[8]
322	United States	c. GGT>GTT	5′ SP	[8]
323	United States	c. GGT>GTT	5′ SP	[8]
324	Spain	c. GCG>GTG	p. Ala588Val	[8]
325	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
326	Europe[1]	c. 2883+1G>T	SP	[58]
327	Europe[1]	c. 2883+1G>T	SP	[58]
328	Europe[1]	c. 775G>A	p. Asp259Asn	[58]
329	Europe[1]	c. 1387G>A	p. Gly463Arg	[58]
330	Europe[1]	c. 1024G>T	p. Gly342X	[58]
331	Europe[1]	c. 2186G>T	p. Gly729Val	[58]
332	Europe[1]	c. 2883+1G>T	SP	[58]
333	Europe[1]	c. 1335+1G>C	SP	[58]
334	Europe[1]	c. 1196_1202dup	p. Ser402X	[58]
335	Europe[1]	c. 911C>T	p. Thr304Met	[58]
336	Europe[1]	c. 2531G>A	p. Trp844X	[58]
337	Europe[1]	c. 2883+1G>T	SP	[58]
338	Europe[1]	c. 2252C>T	p. Pro751Leu	[58]
339	Europe[1]	c. 237_238dup	p. Arg80fs	[58]

340	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
341	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
342	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
343	Europe[1]	c. 2164C>T	p. Gln722X	[58]
344	Europe[1]	c. 1196_1202dup	p. Ser402X	[58]
345	Europe[1]	c. 2830C>T	p. Arg944Trp	[58]
346	Europe[1]	c. 2884-6G>A	SP	[58]
347	Europe[1]	c. 2497T>A	p. Ser833Thr	[58]
348	Europe[1]	c. 1568-4G>A	SP	[58]
349	Europe[1]	c. 403C>T	p. Arg135Cys	[58]
Co-hetero (n=590):				
Europe (n=261) + North America (n=7) + Asia (n=316) + Oceania (n=1) + Africa (n=5)				
350	China	c. 473G>A	p. Arg158Gln	[72]
		c. 602-16G>A	SP	
351	China	c. 1456G>A	p. Asp486Asn	[72]
		c. 965-1G>A	SP	
352	China	c. 911C>T	p. Thr304Met	[72]
		c. 506-1G>A	SP	
353	China	c. 1567G>A	p. Ala523Thr	[73]
		c. 2542G>A	p. Asp523Asn	
354	China	c. 911C > T	p. Thr304Met	[62]
		c. 2099T > C	p. Leu700Pro	
355	China	c. 1456G > A	p. Asp486Asn	[62]
		c. 2633+1G>A	SP	
356	China	c. 1456G>A	p. Asp486Asn	[62]
		c. 2633+1G > A	SP	
357	China	c. 486_490delinsA	p. Thr163fs	[62]
		c. 1456G>A	p. Asp486Asn	
358	China	c. 805_806ins TTGGCGTG GTCTCGGTCA	p. Thr269fs	[62]
		c. 1288T>G	p. Cys430Gly	
359	China	c.473G>A	p. Arg158Gln	[62]
		c.634G > A	p. Gly212Ser	
360	China	c. 234delG	p. E78fs	[62]
		c. 179C > T	p. Thr60Met	
361	China	c. 234delG	p. E78fs	[62]
		c. 179C>T	p. Thr60Met	
362	China	c. 486_490delinsA	p. Thr163fs	[62]
		c. 1925G>A	p. Arg642His	
363	China	c. 486_490delinsA	p. Thr163fs	[62]
		c. 1925G>A	p. Arg642His	
364	China	c. 179C>T	p. Thr60Met	[62]

		c. 506-1G>A	SP	
365	China	c. 179C>T	p. Thr60Met	[62]
		c. 506-1G>A	SP	
366	China	c. 506-1G>A	SP	[62]
		c. 2129C>T	p. Ser710T	
367	China	c. 506-1G> A	SP	[62]
		c. 1077C>G	p. Asn359Lys	
368	China	c. 486_490delinsA	p. Thr163fs	[62]
		c. 965-1 _969 delinsACCGAAA	p. fs	
		c. 976_977delGT	p. Val326fs	
369	China	c. 1919A>G	p. Asn640Ser	[74]
		c. 2522A>G	p. Asp841Gly	
370	China	c. 2842delT	NA	[75]
		c. 1569_1586del	NA	
371	China	NA	p. Thr60Met	[76]
		NA	p. Thr163Met	
		NA	p. Arg871His	
372	China	NA	p. Arg83Gln	[76]
		NA	p. Thr163Met	
		NA	p. Arg871His	
373	China	NA	p. Arg83Gln	[76]
		NA	p. Thr163Met	
		NA	p. Arg871His	
374	China	NA	p. Thr60Met	[76]
		NA	p. Arg83Gln	
375	China	NA	p. Thr60Met	[76]
		NA	p. Thr163Met	
		NA	p. Arg871His	
376	China	NA	p. Arg83Gln	[76]
		NA	p. Gly362Ser	
377	China	NA	p. Gly729Val	[76]
		NA	p. Gly439Ser	
378	China	NA	p. Thr60Met	[76]
		NA	p. Arg83Gln	
379	China	c. 1456G>A	p. Asp486Asn	[77]
		c. 2102_2107 (delACAAGA)	p. 701_702 delAsnLys	
380	China	c. 179C>T	p. Thr60Met	[78]
		c. 1456G>A	p. Asp486Asn	
381	China	NA	p. Gly439Ser	[16]
		NA	p. Ser615Leu	
382	China	NA	p. Arg399Cys	[16]

		NA	p. Asp486Asn	
383	China	NA	p. Trp151Ter	[16]
		NA	p. Ala370Pro	
		NA	p. Gly800Arg	
384	China	NA	p. Gln131Lys	[16]
		NA	p. Gly201Asp	
385	China	NA	p. Val169Ile	[16]
		NA	p. Leu170Gln	
386	China	NA	p. Tyr70Cys	[16]
		NA	p. Arg861Cys	
387	China	NA	p. Leu215Pro	[16]
		NA	p. Trp844Ter	
388	China	NA	p. 809frameshift	[16]
		NA	p. Arg913Gln	
389	China	NA	p. Val677Met	[16]
		NA	p. Ser976Phe	
390	China	NA	p. Leu700Val	[16]
		NA	p. Arg913Gln	
391	China	NA	p. Thr428Ile	[16]
		NA	p. Asp486Asn	
392	China	c. 179C>T	p. Thr60Met	[65]
		c. 506-1G>A	SP	
393	China	c. 179C>T	p. Thr60Met	[65]
		c. 506-1G>A	SP	
394	China	c. 506-1G>A	SP	[65]
		c. 2129C>T	NA	
395	China	c. 506-1G>A	SP	[65]
		c. 1077C>G	p. Asn359Lys	
396	China	c. 486-490del TACGGinsA	p. 162frameshift	[65]
		c. 965-1_969 delgCGGAC insACCGA and c. 976-977delGT	NA	
397	China	NA	p. Asn359Lys	[17]
		NA	p. Asp486Asn	
398	China	NA	p. Asn359Lys	[17]
		c. 493-496delACGG	NA	
399	China	NA	p. Asp486Asn	[17]
		NA	p. Arg928Cys	
400	China	c. 179C>T	p. Thr60Met	[79]
		c. 366A>G	p. Ala122Ala	
		965-1_976g_I7E8_ cggacatttttg	p. 965-1_976 del13ins12	

		to a_I7E8_ccgaaaatttt		
401	China	c. 179C>T	p. Thr60Met	[79]
		c. 366A>G	p. Ala122Ala	
		965-1_976g_I7E8_ cggacatttttg to a_I7E8_ccgaaaatttt	p. 965-1_976 del13ins12	
402	China	c. 366A>G	p. Ala122Ala	[79]
		965-1_976g_I7E8_ cggacatttttg to a_I7E8_ccgaaaatttt	p. 965-1_976 del13ins12	
403	China	c. 179C>T	p. Thr60Met	[79]
		c. 366A>G	p. Ala122Ala	
404	China	c. 179C>T	p. Thr60Met	[79]
		c. 366A>G	p. Ala122Ala	
405	China	c. 179C>T	p. Thr60Met	[79]
		c. 366A>G	p. Ala122Ala	
406	China	c. 1077C>G	p. Asn359Lys	[79]
		c. 1145C>T	p. Thr382Met	
		c. 2738G>A	p. Arg913Gln	
407	China	c. 1077C>G	p. Asn359Lys	[79]
		c. 2738G>A	p. Arg913Gln	
408	China	c. 1077C>G	p. Asn359Lys	[79]
		c. 2738G>A	p. Arg913Gln	
409	China	c. 1077C>G	p. Asn359Lys	[79]
		c. 2738G>A	p. Arg913Gln	
410	China	c. 179C>T	p. Thr60Met	[80]
		c. 234delG	p. E78fs	
411	China	c. 486-490del TACGGinsA	p. 162frameshift	[80]
		c. 1925G>A	p. Arg642His	
412	China	c. 486-490del TACGGinsA	p. 162frameshift	[80]
		c. 1925G>A	p. Arg642His	
413	China	NA	p. Thr163Met	[81]
		NA	p. Thr649Met	
414	China	NA	p. Thr649Met	[81]
		c. 2069delA	NA	
415	China	NA	p. Asn442Lys	[82]
		c. IVS6-1G>A	NA	
416	China	c. 2135C>A	NA	[83]
		c. 2881-2delAG	NA	
417	China	c. 2135C>A	NA	[83]
		c. 2881-2delAG	NA	

418	China	c. 2135C>A	NA	[83]
		c. 2881-2delAG	NA	
419	China	c. 2135C>A	NA	[83]
		c. 2881-2delAG	NA	
420	China	c. 2135C>A	NA	[83]
		c. 2881-2delAG	NA	
421	China	c. C494T	p. Thr163Met	[84]
		c. C1284G	p. Asn426Lys	
422	China	NA	p. Arg913Gln	[20]
		c. 1670-8C>T	NA	
423	China	NA	p. Arg913Gln	[20]
		c. 1670-8C>T	NA	
424	China	c. 185C>T	p. Thr60Met	[21]
		c. 492_496del TACGGinsA	p. 162Frameshift	
425	China	c. 1022C>T	p. Thr339Ile	[21]
		c. 1083C>G	p. Asn359Lys	
426	China	c. 1083C>G	p. Asn359Lys	[21]
		c. 1322G>T	p. Gly439Val	
427	China	c. 185C>T	p. Thr60Met	[21]
		c. 2717G>A	p. Arg904Gln	
428	China	IVS7-1G>A g. 7427_7438delins CCGAAAATTTT	SP	[21]
		c. 2717G>A	p. Arg904Gln	
429	China	IVS7-1G>A g. 7427_7438delins CCGAAAATTTT	SP	[21]
		c. 1268G>T	p. Cys421Phe	
430	China	c. 593G>T	p. Gly196Val	[22]
		c. 1322G>T	p. Gly439Val	
431	China	c. 1294T>G	p. Cys430Gly	[22]
		c. 346_353del ACTGATGG	114 frameshift	
432	China	c. 185C>T	p. Thr60Met	[22]
		c. 346_353del ACTGATGG	114 frameshift	
433	China	c. 185C>T	p. Thr60Met	[22]
		c. 1294T>G	p. Cys430Gly	
434	China	c. 185C>T	p. Thr60Met	[22]
		c. 1384delG	460 frameshift	
435	China	c. 1322G>T	p. Gly439Val	[22]
		c. 2883_2884delAG	959 frameshift	

436	China	c. 1718T>G	p. Leu571Pro	[22]
		c. 2969insGCT	p. 997insCys	
437	China	c. 1718T>G	p. Leu571Pro	[22]
		c. 2969insGCT	p. 997insCys	
438	China	c. 185C>T	p. Thr60Met	[22]
		c. 1462G>A	p. Asp486Asn	
439	China	c. 644T>C	p. Leu215Pro	[23]
		c. 2532G>A	p. Trp844X	
440	China	c. 1315G>A	p. Gly439Ser	[23]
		c. 1844C>T	p. Ser615Leu	
441	China	c. 452G>A	p. Trp151X	[23]
		c. 2398G>A	p. Gly800Arg	
		c. 1108G>C	p. Ala370Pro	
442	China	c. 2454_2461del CAAGGCCC	p. 819frameshift	[23]
		c. 2738G>A	Arg913Gln	
443	China	c. 486-490del TACGGinsA	p. 162frameshift	[23]
		c. 1288T>G	p. Cys430Gly	
		c. 1975G>A	p. Val659Met	
444	China	c. 1283C>T	p. Thr428Ile	[23]
		c. 1456G>A	p. Asp486Asn	
445	China	c. 2877_2878delAG	p. Arg959fs	[23]
		c. 587G>T	p. Gly196Val	
446	China	c. 179C>T	p. Thr60Met	[23]
		c. 1602A>C	p. Asn534Lys	
447	China	c. 248G>A	p. Arg83Gln	[23]
		c. 2782C>T	p. Arg928Cys	
448	China	c. 1456G>A	p. Asp486Asn	[23]
		c. 806 insTTGGCGT GGTCTCGGTCA	p. 269 ins IGVVSV	
449	China	c. 1456G>A	p. Asp486Asn	[23]
		c. 1195C>T	p. Arg399Cys	
450	China	c. 2927C>T	p. Ser976Phe	[23]
		c. 2029G>A	p. Val677Met	
451	China	c. 506-1G>A	SP	[23]
		c. 496G>A	p. Leu170Gln	
452	China	c. 209A>G	p. Tyr70Cys	[23]
		c. 2581C>T	p. Arg861Cys	
453	China	c. 2098C>G	p. Leu700Val	[23]
		c. 2738G>A	p. Arg913Gln	
454	China	c. 1946C>T	p. Thr649Met	[23]
		c. 1909C>T	p. His637Tyr	

455	China	c. 2782C>T	p. Arg928Cys	[23]
		c. 1924C>T	p. Arg642Cys	
456	China	c. 1077C>G	p. Asn359Lys	[23]
		c. 1850A>G	p. Gln617Arg	
457	China	c. 1315G>A	p. Gly439Ser	[23]
		c. 1924C>T	p. Arg642Cys	
458	China	c. 2582G>A	p. Arg861His	[23]
		c. 1698C>A	p. Asn566Lys	
459	China	c. 539C>A	p. Thr180Lys	[23]
		c. 179C>T	p. Thr60Met	
460	China	c. 815T>C	p. Leu272Pro	[23]
		c. 965-1_976delins ACCGAAAATTTT	SP	
461	China	Intron 7 as –1 G>A and exon 8 nt +1 to +12 delCGGACATTT TTGinsCCGAAAATTTT	NA	[85]
		c. 185C>T	p. Thr60Met	
462	China	c. 676G>A	p. Ala226Thr	Our study
		c. 179C>A	p. Thr60Met	
463	China	c. 509T>A	p. Leu170Gln	Our study
		c. 179C>T	p. Thr60Met	
		c. 421G>A	p. Gly141Arg	
464	Japan	c. 1732G>A	p. Val578Met	[86]
		c. 2537_38delTT	p. 846fs	
465	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
466	Japan	c. 1195C>T	p. Arg399Cys	[2]
		c. 2573T>A	p. Leu858His	
467	Japan	c. 1257_1259dup	p. Ala420dup	[2]
		c. 1868T>C	p. Leu623Pro	
468	Japan	c. 2029G>A	p. Val677Met	[2]
		c. 2573T>A	p. Leu858His	
469	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 1196_1202dup GTGATGC	p. Ser402*	
470	Japan	c. 1195C>T	p. Arg399Cys	[2]
		c. 1670-1G>T	IVS13 as G-T -1	
471	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 3052C>T	p. Arg1018*	
472	Japan	c. 1868T>C	p. Leu623Pro	[2]
		c. 2503C>T	p. Gln835*	

473	Japan	c. 179C>T	p. Thr60Met	[2]
		c. 2029G>A	p. Val677Met	
474	Japan	c. 139delC	p. His47Thrfs*67	[2]
		c. 1924C>T	p. Arg642Cys	
475	Japan	c. 2221G>A	p. Gly741Arg	[2]
		c. 2573T>A	p. Leu858His	
476	Japan	c. 1195C>T	p. Arg399Cys	[2]
		c. 1924C>T	p. Arg642Cys	
477	Japan	c. 1195C>T	p. Arg399Cys	[2]
		c. 1924C>T	p. Arg642Cys	
478	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 863T>C	p. Leu288Pro	
479	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 1732G>A	p. Val578Met	
480	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2891G>A	p. Arg964Gln	
481	Japan	c. 1706C>T	p. Ala569Val	[2]
		c. 2573T>A	p. Leu858His	
482	Japan	c. 139delC	p. His47Thrfs*67	[2]
		c. 2573T>A	p. Leu858His	
483	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 1868T>C	p. Leu623Pro	
484	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2747+2T>A	IVS23+2T>A	
485	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2747+2T>A	IVS23+2T>A	
486	Japan	c. 817dupG	p. Ala273Glyfs*38	[2]
		c. 1670-191C>T	Cryptic exon insertion	
487	Japan	c. 1868T>C	p. Leu623Pro	[2]
		c. 2548+253C>T	Cryptic exon insertion	
488	Japan	c. 179C>T	p. Thr60Met	[2]
		c. 2573T>A	p. Leu858His	
489	Japan	c. 1045C>T	p. Pro349Ser	[2]
		c. 1706C>T	p. Ala569Val	
490	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 3053G>A	p. Arg1018Gln	
491	Japan	c. 960C>G	p. Tyr320*	[2]
		c. 2573T>A	p. Leu858His	
492	Japan	c. 1100C>T	p. Pro367Leu	[2]
		c. 2573T>A	p. Leu858His	
493	Japan	c. 1732G>A	p. Val578Met	[2]
		c. 2573T>A	p. Leu858His	

494	Japan	c. 1844C>T	p. Ser615Leu	[2]
		c. 2537_2538delTT	p. Phe846*	
495	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 3052C>T	p. Arg1018*	
496	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2877_2878delAG	p. Arg959Serfs*11	
497	Japan	c. 488C>T	p. Thr163Met	[2]
		c. 1939delG	p. Val647Cysfs*25	
498	Japan	c. 1195C>T	p. Arg399Cys	[2]
		c. 2891G>A	p. Arg964Gln	
499	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 1195C>T	p. Arg399Cys	
500	Japan	c. 179C>T	p. Thr60Met	[2]
		c. 1670-1G> T	IVS13 as G-T -1	
501	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	
502	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 1924C>T	p. Arg642Cys	
503	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 2573T>A	p. Leu858His	
504	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 2573T>A	p. Leu858His	
505	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2029G>A	p. Val677Met	
506	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2927C>T	p. Ser976Phe	
507	Japan	c. 179C>T	p. Thr60Met	[2]
		c. 1670-1G> T	IVS13 as G-T -1	
508	Japan	c. 488C>T	p. Thr163Met	[2]
		c. 2573T>A	p. Leu858His	
509	Japan	c. 179C>T	p. Thr60Met	[2]
		c. 3052C>T	p. Arg1018*	
510	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
511	Japan	c. 1706C>T	p. Ala569Val	[2]
		c. 2927C>T	p. Ser976Phe	
512	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
513	Japan	c. 1868T>C	p. Leu623Pro	[2]
		c. 1963C>T	p. Arg655Cys	

514	Japan	c. 506-1G>A	SP	[2]
		c. 2927C>T	p. Ser976Phe	
515	Japan	c. 539C>A	p. Thr180Lys	[2]
		c.1924C>T	p. Arg642Cys	
516	Japan	c. 1216A>C	p. Asn406His	[2]
		c. 2927C>T	p. Ser976Phe	
517	Japan	c. 817dupG	p. Ala273Glyfs*38	[2]
		c. 1924C>T	p. Arg642Cys	
518	Japan	c. 1868T>C	p. Leu623Pro	[2]
		c. 1930delC	p. Gln644Serfs*28	
519	Japan	c. 1868T>C	p. Leu623Pro	[2]
		c. 1930delC	p. Gln644Serfs*28	
520	Japan	c. 488C>T	p. Thr163Met	[2]
		c. 1963C>T	p. Arg655Cys	
521	Japan	c. 505+5g>c	IVS3 ds G-A +5	[2]
		c. 1868T>C	p. Leu623Pro	
522	Japan	c. 178A>G	p. Thr60Ala	[2]
		c. 2573T>A	p. Leu858His	
523	Japan	c. 178A>G	p. Thr60Ala	[2]
		c. 2573T>A	p. Leu858His	
524	Japan	c. 664_666delATT	p. Ile222del	[2]
		c. 2573T>A	p. Leu858His	
525	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
526	Japan	c. 2537_2538delTT	p. Phe846*	[2]
		c. 2573T>A	p. Leu858His	
527	Japan	c. 2537_2538delTT	p. Phe846*	[2]
		c. 2573T>A	p. Leu858His	
528	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 2573T>A	p. Leu858His	
529	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 1963C>T	p. Arg655Cys	
530	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 1963C>T	p. Arg655Cys	
531	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
532	Japan	c. 1930delC	p. Gln644Serfs*28	[2]
		c. 2573T>A	p. Leu858His	
533	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 1930delC	p. Gln644Serfs*28	
534	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 1049C>T	p. Ser350Leu	
535	Japan	c. 1963C>T	p. Arg655Cys	[2]

		c. 2927C>T	p. Ser976Phe	
536	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 668delT	p. Phe223Serfs*79	
537	Japan	c. 1670-1G>T	IVS13G>T	[2]
		c. 2573T>A	p. Leu858His	
538	Japan	c. 179C>T	p. Thr60Met	[2]
		c. 2927C>T	p. Ser976Phe	
539	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 3052C>T	p. Arg1018*	
540	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 3052C>T	p. Arg1018*	
541	Japan	c. 817dupG	p. Ala273Glyfs*38	[2]
		c. 3052C>T	p. Arg1018*	
542	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 3052C>T	p. Arg1018*	
543	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 3053G>A	p. Arg1018Gln	
544	Japan	c. 664_666delATT*	p. Ile222del	[2]
		c. 2573T>A	p. Leu858His	
545	Japan	c. 1732G>A	p. Val578Met	[2]
		c. 2573T>A	p. Leu858His	
546	Japan	c. 1A>T	p. Met1Leu	[2]
		c. 1868T>C	p. Leu623Pro	
547	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 3052C>T	p. Arg1018*	
548	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 1930delC	p. Gln644Serfs*28	
549	Japan	c. 1278C>A*	p. Asn426Lys	[2]
		c. 1930delC	p. Gln644Serfs*28	
550	Japan	c. 488C>T	p. Thr163Met	[2]
		c. 2573T>A	p. Leu858His	
551	Japan	c. 1844C>T	p. Ser615Leu	[2]
		c. 1930delC	p. Gln644Serfs*28	
552	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 1868T>C	p. Leu623Pro	
553	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2891G>A	p. Arg964Gln	
554	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 2573T>A	p. Leu858His	
555	Japan	c. 1289G>A	p. Cys430Tyr	[2]

		c. 2573T>A	p. Leu858His	
556	Japan	c. 247C>T	p. Arg83Trp	[2]
		c. 2573T>A	p. Leu858His	
557	Japan	c. 2537_2538delTT	p. Phe846*	[2]
		c. 2573T>A	p. Leu858His	
558	Japan	c. 2029G>A	p. Val677Met	[2]
		c. 2573T>A	p. Leu858His	
559	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 3052C>T	p. Arg1018*	
560	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2686C>T*	p. Arg896*	
561	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
562	Japan	c. 626G>C*	p. Arg209Pro	[2]
		c. 2029G>A	p. Val677Met	
563	Japan	c. 1925G>A	p. Arg642His	[2]
		c. 3052C>T	p. Arg1018*	
564	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2891G>A	p. Arg964Gln	
565	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 2573T>A	p. Leu858His	
566	Japan	c. 1732G>A	p. Val578Met	[2]
		c. 2573T>A	p. Leu858His	
567	Japan	c. 704C>T*	p. Thr235Met	[2]
		c. 1709C>T	p. Ala570Val	
568	Japan	c. 1201_1210del CCTCTGGGG	p. Ala401_Gly 403del	[2]
		c. 2891G>A	p. Arg964Gln	
569	Japan	c. 911C>T	p. Thr304Met	[2]
		c. 2573T>A	p. Leu858His	
570	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 1289G>A	p. Cys430Tyr	
571	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 1289G>A	p. Cys430Tyr	
572	Japan	c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	[2]
		c. 1826-1G>A	IVS14-1G>A	
573	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 3052C>T	p. Arg1018*	
574	Japan	c. 1456G>A	p. Asp486Asn	[2]
		c. 1868T>C	p. Leu623Pro	

575	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2686C>T	p. Arg896*	
576	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
577	Japan	c. 1A>T	p. Met1Leu	[2]
		c. 179C>T	p. Thr60Met	
578	Japan	c. 2537_2538delTT	p. Phe846*	[2]
		c. 1923C>G	p. Tyr641*	
579	Japan	c. 1077C>G	p. Asn359Lys	[2]
		c. 1709C>T	p. Ala570Val	
580	Japan	c. 1868T>C	p. Leu623Pro	[2]
		c. 2927C>T	p. Ser976Phe	
581	Japan	c. 2891G>A	p. Arg964Gln	[2]
		Exon 9-18 Duplication	NA	
582	Japan	c. 2573T>A	p. Leu858His	[2]
		Exon 7-8 Deletion	NA	
583	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 3053G>A	p. Arg1018Gln	
584	Japan	c. 1930delC	p. Gln644Serfs*28	[2]
		c. 2573T>A	p. Leu858His	
585	Japan	c. 1963C>T	p. Arg655Cys	[2]
		c. 2573T>A	p. Leu858His	
586	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
587	Japan	c. 539C>T	p. Thr180Lys	[2]
		c. 1698C>A	p. Asn566Lys	
588	Japan	c. 179C>T	p. Thr60Met	[2]
		c. 2099T>C	p. Leu700pro	
589	Japan	c. 1924C>T	p. Arg642Cys	[2]
		c. 2573T>A	p. Leu858His	
590	Japan	c. 238delC	p. Arg80Glyfs*34	[2]
		c. 2573T>A	p. Leu858His	
591	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 2891G>A	p. Arg964Gln	
592	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 1868T>C	p. Leu623Pro	
593	Japan	c. 1669+297T>G	Cryptic exon insertion	[2]
		c. 2927C>T	p. Ser976Phe	
594	Japan	c. 1897dupG	p. E633Gfs56	[2]
		c. 1924C>T	p. Arg642Cys	
595	Japan	c. 1456G>A	p. Asp486Asn	[2]
		c. 2927C>T	p. Ser976Phe	
596	Japan	c. 179C>T	p. Thr60met	[2]

		c. 2573T>A	p. Leu858His	
597	Japan	c. 1100C>T	p. Pro367Leu	[2]
		c. 1924C>T	p. Arg642Cys	
598	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 2573T>A	p. Leu858His	
599	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 1868T>C	p. Leu623Pro	
600	Japan	c. 539C>A	p. Thr180Lys	[2]
		c. 2891G>A	p. Arg964Gln	
601	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 3052C>T	p. Arg1018*	
602	Japan	c. 1924C>T	p. Leu642Pro	[2]
		c. 2573T>A	p. Leu858His	
603	Japan	c. 1924C>T	p. Leu642Pro	[2]
		c. 2573T>A	p. Leu858His	
604	Japan	c. 1868T>C	p. Leu623Pro	[2]
		c. 2927C>T	p. Ser976Phe	
605	Japan	c. 1195C>T	p. Arg399Cys	[2]
		c. 2573A>C	p. Leu858His	
606	Japan	c. 2573T>A	p. Leu858His	[2]
		c. 3053G>A	p. Arg1018Gln	
607	Japan	c. 788_805dup	p. Ile263_Val 268dup	[2]
		c. 1132G>A	p. Ala378Thr	
608	Japan	c. 1163C>A	p. Ala388Asp	[2]
		c. 2573A>C	p. Leu858His	
609	Japan	c. 139delc	p. His47Thrfs*67	[2]
		c. 2660+1G>A	NA	
610	Japan	c.1163C>A	p. Ala388Asp	[2]
		c.2573A>C	p. Leu858His	
611	Japan	c.539C>A	p. Thr180Lys	[2]
		c. 805_806ins18	p. Thr269delinsAsn TrpArgGlyLeuGlyPro	
612	Japan	c.248G>A	p. Arg83Gln	[2]
		c. 2537_2538delTT	p. Phe846*	
613	Japan	c.2573T>A	p. Leu858His	[2]
		c.2891G>A	p. Arg964Gln	
614	Japan	c.2573T>A	p. Leu858His	[2]
		c.2891G>A	p. Arg964Gln	
615	Japan	c.1930delC	p. Gln644Serfs*28	[2]
		c.1948G>C	p. Gly650Arg	
616	Japan	c.539C>A	p. Thr180Lys	[2]
		c.1923C>G	p. Tyr641	

617	Japan	c. 1925G>A	p. Arg642His	[87, 88]
		c. 1924C>T	p. Arg642Cys	
618	Japan	c. 1868T>C	p. Leu623Pro	[89]
		c. 2543-2544del TT	NA	
619	Japan	c. 1315G>A	p. Gly439Ser	[89]
		c. 1664C>T	p. Ser555Leu	
620	Japan	NA	p. Thr180Lys	[90]
		NA	p. Leu858His	
621	Japan	c. 2927C>T	p. Ser976Phe	[91]
		c. 1567+297T>G	NA	
622	Japan	c. 539C>A	p. Thr180Lys	[92]
		c. 1844C>T	p. Ser615Leu	
623	Japan	NA	p. Arg655Cys	[93]
		NA	p. Arg955Gln	
624	Japan	NA	p. Thr180Lys	[94]
		c. 2552T>A	p. Leu849His	
625	Japan	NA	p. Thr180Lys	[95]
		NA	p. Val677Met	
626	Japan	NA	p. Thr180Lys	[95]
		NA	p. Leu849Met	
627	Japan	c. 539C>A	p. Thr180Lys	[6]
		c. 1732G>A	p. Val578Met	
628	Japan	c. 1045C>T	p. Pro349Ser	[6]
		c. 1706C>T	p. Ala569Val	
629	Japan	c. 2552T>A	p. Leu849His	[96]
		c. 2561G>A	p. Arg852His	
630	Japan	c. 1732G>A	p. Val578Met	[97]
		c. 2573T>A	p. Leu858His	
631	Japan	c. 2552T>A	p. Leu849His	[7]
		NA	p. Arg852His	
632	Japan	NA	p. Arg261Cys	[98]
		NA	p. Leu623Pro	
633	Japan	c. 185C>T	p. Thr60Met	[70]
		c. 1181_1186+20del26	Deletion of exon 9 and frameshift at exon 10	
634	Japan	c. 1811_1812delAT	Frameshift	[70]
		c. 2552T>A	p. Leu849His	
635	Japan	c. 1811_1812delAT	Frameshift	[70]
		c. 1930C>T	p. Arg642Cys	
636	Japan	c. 1930C>T	p. Arg642Cys	[70]
		c. 2552T>A	p. Leu849His	
637	Japan	c. 7A>T	p. Met1Leu	[70]
		c. IVS16+1G>A	Additional 32 amino acids	

638	Japan	c. 7A>T	p. Met1Leu	[70]
		c. IVS16+1G>A	Additional 32 amino acids	
639	Japan	c. 1932delC	Frameshift	[70]
		c. 2552T>A	p. Leu849His	
640	Japan	c. 731_732 delTG	NA	[99]
		c. 2543_2544delTT	NA	
641	Japan	NA	p. Leu623Pro	[99]
		In-frame 18 base insertion	NA	
642	UK	c. 2221G>A	p. Gly741Arg	[29]
		c. 3002C>A	p. Ala1001Asp	
643	UK	c. 1202C>A	p. Ala401Asp	[29]
		c. 2965	p. Gly989Arg	
644	UK	c. 2221G>A	p. Gly741Arg	[29]
		c. 3052C>T	p. Arg1018*	
645	UK	c. 626G>A	p. Arg209Gln	[29]
		c. 1577A>G	p. Asn526Ser	
646	UK	c. 626G>A	p. Arg209Gln	[29]
		c. 1577A>G	p. Asn526Ser	
647	UK	c. 647G>A	p. Gly216Glu	[29]
		c. 2221G>A	p. Gly741Arg	
648	UK	c. 424G>T	p. Val142Leu	[29]
		c. 2952-?_*1+?del	NA	
649	UK	c. 506-1G>A	SP	[29]
		c. 1180 + 1G>T	SP	
650	UK	c. 1664C>T	p. Ser555Leu	[30]
		Exon deleted	p. Thr985X	
651	UK	c. 1196_1202dup GTGATGC	p. Ser402X	[30]
		c. 2576T>C	p. Leu859Pro	
652	UK	c. 363G>C	p. Glu121Asp	[30]
		c. 626G>A	p. Arg209Gln	
653	UK	c. 1763C>T	p. Ala588Val	[30]
		c. 2893C>T	p. Gln965X	
654	UK	c. 497C>T	p. Ala166Val	[30]
		c. 2576T>C	p. Leu859Pro	
655	UK	c. 2221G>A	p. Gly741Arg	[30]
		c. 2981G>A	p. Cys994Tyr	
656	UK	c. 1825+1delG	Intron 14 5′ SP	[30]
		c. 2981G>A	p. Cys994Tyr	
657	UK	c. 961C>T	p. Arg321Trp	[30]
		c. 2221G>A	p. Gly741Arg	
658	UK	c. 911C>T	p. Thr304Met	[30]
		c. 2883+1G>T	Intron 24 5′ SP	

659	UK	c. 1963C>T	p. Arg655Cys	[30]
		c. 2990_2993 dupCGCT	p. Leu998fsc	
660	UK	c. 1664C>T	p. Ser555Leu	[30]
		c. 2221G>A	p. Gly741Arg	
661	UK	c. 2221G>A	p. Gly741Arg	[30]
		c. 2883+1G>T	Intron 24 5′ SP	
662	UK	c. 2996A>G	p. Tyr999Cys	[30]
		c. 3089A>G	p. Gln1030Arg	
663	UK	c. 1145C>T	p. Thr382Met	[30]
		c. 2221G>A	p. Gly741Arg	
664	UK	c. 710G>A	p. Gly237Asp	[30]
		c. 2576T>C	p. Leu859Pro	
665	UK	c. 1196G>T	p. Arg399Leu	[30]
		c. 1825+1G>T	Intron 14 5′ SP	
666	UK	c. 1196G>T	p. Arg399Leu	[30]
		c. 1825+1G>T	Intron 14 5′ SP	
667	UK	c. 539-543delCGGTG	p. Thr180fs	[30]
		c. 2883+1G>T	Intron 24 5′ SP	
668	UK	c. 1390G>A	p. Ala464Thr	[30]
		c. 2576T>C	p. Leu859Pro	
669	UK	c. 2221G>A	p. Gly741Arg	[30]
		c. 3053G>C	p. Arg1018Pro	
670	UK	c. 506-1G>A	SP	[30]
		c. 2089_2095del ACCAAGT	p. Thr697fs	
671	UK	c. 1763C>T	p. Ala588Val	[30]
		c. 2576T>C	p. Leu859Pro	
672	UK	c. 1258G>A	p. Ala420Thr	[30]
		c. 2947G>A	p. Val983Ile	
673	UK	c.1061_1062dupTC	p. Leu355fs	[30]
		c. 2965G>A	p. Gly989Arg	
674	UK	c. 1315G>A	p. Gly439Ser	[30]
		c. 2221G>A	p. Gly741Arg	
675	UK	c. 2221G>A	p. Gly741Arg	[30]
		c. 2981G>A	p. Cys994Tyr	
676	UK	c. 2037+1G>A	Intron 16 5′ SP	[30]
		c. 3053G>A	p. Arg1018Gln	
677	UK	Exons deleted	Probable loss	[30]
		c. 2221G>A	p. Gly741Arg	
678	UK	c. 626G>A	p. Arg209Gln	[30]
		c. 1763C>A	p. Ala588Glu	
679	UK	c. 2581C>T	p. Arg861Cys	[30]

		c. 2723T>C	p. Ile908Thr	
680	UK	c. 1351T>A	p. Ser451Thr	[30]
		c. 2221G>A	p. Gly741Arg	
681	UK	c. 1315G>A	p. Gly439Ser	[30]
		c. 2883+1G>T	Intron 24 5′ SP	
682	UK	c. 626G>A	p. Arg209Gln	[30]
		c. 2981G>A	p. Cys994Tyr	
683	UK	NA	p. Arg209Gln	[100]
		NA	p. Ser615Leu	
684	UK	c. IVS24+1: G>T	NA	[101]
		NA	p. Gly439Ser	
685	France	c. 2581C>T	p. Arg861Cys	[38]
		c. 3077C>T	p. Thr1026Ile	
686	France	c. 644T>C	p. Leu215Pro	[38]
		c. 1315G>A	p. Gly439Ser	
687	France	c. 1262G>A	p. Cys421Tyr	[38]
		c. 2221G>A	p. Gly741Arg	
688	France	c. 1262G>A	p. Cys421Tyr	[38]
		c. 2221G>A	p. Gly741Arg	
689	France	c. 1196_1202dup	p. Ser402X	[102]
		c. 2747 + 1G > A	SP	
690	France	c. 1805_1806del	p. Tyr602CysfsX31	[102]
		c. 2660 + 1G > A	SP	
		c. 2747 + 1G > A	SP	
691	France	c. 1805_1806del	p. Tyr602CysfsX31	[102]
		c. 2660 + 1G > A	SP	
		c. 2747 + 1G > A	SP	
692	France	c. 1805_1806del	p. Tyr602CysfsX31	[102]
		c. 2660 + 1G > A	SP	
		c. 2747 + 1G > A	SP	
693	France	c. 1196_1202dup	p. Ser402X	[102]
		c. 1805_1806del	p. Tyr602CysfsX31	
		c. 2660 + 1G > A	SP	
694	France	c. 911C>T	p. Thr304Met	[103]
		c. 1315G>A	p. Gly439Ser	
695	France	c. 2221G>A	p. Gly741Arg	[104]
		c. 247C>T	p. Arg83Trp	
696	France	c. 2221G>A	p. Gly741Arg	[104]
		c. 247C>T	p. Arg83Trp	
697	France	c. 1484T>	p. Phe495Ser	[39]
		c. 2221G>A	p. Gly741Arg	
698	France	c. 1840T>C	p. Ser614Pro	[39]
		c. 1390G>A	p. Ala464Thr	

699	France	c. 679A>G	p. Asn227Asp	[39]
		c. 1664C>T	p. Ser555Leu	
700	France	c. 1390G>A	p. Ala464Thr	[39]
		c. 2221G>A	p. Gly741Arg	
701	France	c. 160C>T	p. Arg54Cys	[39]
		c. 2782C>T	p. Arg928Cys	
702	France	c. 2747+1G>C	NA	[39]
		c. 2883+1G>T	SP	
703	France	c. 2576T>C	p. Leu859Pro	[39]
		c. 2581C>T	p. Arg861Cys	
704	France	c. 1928C>T	p. Pro643Leu	[39]
		c. 964+1G>T	NA	
705	France	c. 1145C>T	p. Thr382Met	[39]
		c. 2807_2810dup	p. Thr938GlyfsX17	
706	France	c. 2883+1G>T	SP	[39]
		c. 2981G>A	p. Cys994Tyr	
707	France	c. 2120C>T	p. Ala707Val	[39]
		c. 1145C>T	p. Thr382Met	
708	France	c. 910A>C	p. Thr304Pro	[39]
		c. 2120C>T	p. Ala707Val	
709	France	c. 2310+2T>G	SP	[34]
		c. 2167C>T	p. Ala714Ala	
710	France	c. 2579C>T	p. Arg852Cys	[34]
		c. 2736G>A	p. Arg904Gln	
		c. 1909G>A	p. Ser628Ser	
711	Korea	c. 179C>T	p. Thr60Met	[25]
		c. 1216A>C	p. Asn406His	
712	Korea	c. 268C>T	p. His90Tyr	[25]
		c. 1216A>C	p. Asn406His	
713	Korea	c. 433C>T	p. Arg145Cys	[25, 105]
		c. 1174A>C	p. Thr392Pro	
714	Korea	c. 433C>T	p. Arg145Cys	[25, 105]
		c. 1174A>C	p. Thr392Pro	
715	Korea	c. 506-1G>A	SP	[25]
		c. 1456G>A	p. Asp486Asn	
716	Korea	c. 536T>A	p. Val179Asp	[25]
		c. 1762delG	p. Ala588fs*23	
717	Korea	c. 784_785ins13	p. Ile262Rfs	[25]
		c. 1456G>A	p. Asp486Asn	
718	Korea	c. 964+1G>A	NA	[25]
		c. 1216A>C	p. Asn406His	
719	Korea	c. 964+1G>T	NA	[25]
		c. 1844C>T	p. Ser615Leu	

720	Korea	c. 964+1G>A	NA	[25]
		c. 2927C>T	p. Ser976Phe	
721	Korea	c. 1897_1898insG	p. Glu633Gfs*56	[25]
		c. 3052C>T	p. Arg1018*	
722	Korea	c. 1924C>T	p. Arg642Cys	[25]
		c. 2243C>T	p. Ser748Leu	
723	Korea	c. 1924C>T	p. Arg642Cys	[25]
		c. 2243C>T	p. Ser748Leu	
724	Korea	c. 1924C>T	p. Arg642Cys	[25]
		c. 2573T>A	p. Leu858His	
725	Korea	c. 2542G>A	p. Asp848Asn	[25]
		c. 2963T>C	p. Ile988Thr	
726	Korea	c. 2573T>A	p. Leu858His	[25]
		c. 2927C>T	p. Ser976Phe	
727	Korea	c. 494A>T	p. Gln165Leu	[106]
		c. 1868T>C	p. Leu623Pro	
728	Korea	NA	p. Asp848Asn	[26]
		NA	p. Ile988Thr	
729	Korea	NA	p. Asp839Asn	[27]
		NA	p. Ile979Thr	
730	Korea	c. 372A>G	p. Ala122Ala	[107]
		c. 545C>A	p. Thr180lys	
		c. 1092T>G	p. Gly362Gly	
		c. 1222A>C	p. Asn406His	
		c. 1715C>T	p. Ala570Val	
731	Korea	c. 372A>G	p. Ala122Ala	[107]
		c. 545C>A	p. Thr180Lys	
		c. +5th, ins A	5’ SP	
		c. 1715C>T	p. Ala570Val	
		c. 2631C>T	p. Gly866Gly	
732	Philippines	c. GCA>ACA	p. Ala728Thr	[8]
		c. GGC>TGC	p. Gly496Cys	
733	CA	NA	p. Pro643Leu	[33]
		NA	p. Gly741Arg	
734	CA	c. 473G>A	p. Arg158Gln	[108]
		c. 631_642del	p. Arg211_ Glu214del	
735	United States	c. 2246G>A	p. Gly741Arg	[34]
		c. 2881+1G>T	SP	
736	United States	c. TGC>CGC	p. Cys421Arg	[8]
		c. CGG>TGG	p. Arg209Trp	
737	United States	c. TGC>CGC	p. Cys421Arg	[8]
		c. CGG>TGG	p. Arg209Trp	

738	United States	c. TGC>CGC	p. Cys421Arg	[8]
		c. CGG>TGG	p. Arg209Trp	
739	United States	c. TGC>CGC	p. Cys421Arg	[8]
		c. CAG>CAT	3′ SP	
740	Portugal	c. 602–16G>A	NA	[109]
		c. 2221G>A	p. Gly741Arg	
741	South Africa	exon 13+69A>G	p. Ser546Gly	[110]
		c. 1930insAGCCCC	NA	
742	South Africa	exon 13+69A>G	p. Ser546Gly	[110]
		c. 1930insAGCCCC	NA	
743	South Africa	exon 13+69A>G	p. Ser546Gly	[110]
		c. 1930insAGCCCC	NA	
744	South Africa	exon 13+69A>G	p. Ser546Gly	[110]
		c. 1930insAGCCCC	NA	
745	South Africa	exon 13+69A>G	p. Ser546Gly	[110]
		c. 1930insAGCCCC	NA	
746	Italy	c. 2581C>T	p. Arg861Cys	[111]
		c. 283delC	p. Gln95ArgfsX19	
747	Italy	c. 2581C>T	p. Arg861Cys	[111]
		c. 283delC	p. Gln95ArgfsX19	
		c. 2738 G>A	p. Arg913Gln	
748	Italy	c. 1925G>A	p. Arg642His	[112]
		c.1181G>A	p. Gly394Asp	
749	Italy	c. 1925G>A	p. Arg642His	[112]
		c.1181G>A	p. Gly394Asp	
750	Italy	c. 269A > C	p. His90Pro	[113]
		c. 1205C > A	p. Ser402Tyr	
751	Italy	NA	p. Gly439Ser	[114]
		NA	p. Arg1018Term	
752	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 1924C>G	p. Arg642Gly	
753	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 20_21delCA	p. Thr7fs	
754	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 1175C>T	p. Thr392Ile	
755	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 2981G>A	p. Cys994Tyr	
756	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 2542G>T	p. Asp848Tyr	
757	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 433C>A	p. Arg145Ser	
758	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 1424C>G	p. Ser475Cys	

759	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		?	?	
760	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 248G>A	p. Arg83Gln	
761	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 1881C>G	p. Tyr627X	
762	Italy	c. 1196_1202dup7bp	p. Ser402X	[41]
		c. 2981G>A	p. Cys994Tyr	
763	Italy	c. 1196_1202dup7bp	p. Ser402X	[115]
		c. 1424C>G	p. Ser475Cys	
764	Italy	c. 1196_1202dup7bp	p. Ser402X	[115]
		c. 1424C>G	p. Ser475Cys	
765	Italy	c. 1175C>T	p. Thr392Ile	[115]
		c. 1844C>T	p. Ser615Leu	
766	Italy	c. 1175C>T	p. Thr392Ile	[115]
		c. 1844C>T	p. Ser615Leu	
767	Italy	c. 1413G>A	p. Gly463Glu	[43]
		c. 1869C>T	p. Ser615Leu	
768	Italy	c. 2780C>T	p. Arg919Cys	[43]
		c. 2979C>T	p. Cys985Tyr	
769	Italy	c. 650C>T	p. Arg209Trp	[43]
		c. 2054G>A	p. Val677Met	
770	Italy	c. 539T>C	p. Trp172Arg	[43]
		c. 650C>T	p. Arg209Trp	
771	Italy	c. 498G>A	p. Arg158Gln	[43]
		2114_2120delACCAAGT	p. Thr697fsX698	
772	Italy	c. 539T>C	p. Trp172Arg	[43]
		c. 1146G>T	p. Gly374Val	
773	Italy	c. 1950G>A	p. Arg642His	[43]
		c. 2579C>A	p. Arg852Ser	
774	Italy	c. 972G>T	p. Gly316Val	[43]
		c. 1949C>G	p. Arg642Gly	
775	Italy	NA	p. Arg209Trp	[45]
		NA	p. Gly186Asp	
776	Italy	NA	p. Pro349Leu	[45]
		NA	p. Lys478Glu	
777	Italy	c. 2216_2217del2-bp	NA	[45]
		NA	p. Gly439Ser	
778	Italy	c. 1228insGTGATGC	NA	[45]
		NA	p. Gly731Arg	
779	Cyprus	NA	p. Met581Lys	[116]
		NA	E9_E14dup	
780	Cyprus	NA	p. Met581Lys	[116]

		NA	E9_E14dup	
781	Spain	c. 1939G>A	p. Val647Met	[48]
		Intron 9 +1G>T	NA	
782	Netherlands	c. 815T>C	p. Leu272Pro	[52]
		c. 1670C>T	NA	
783	Netherlands	c. 815T>C	p. Leu272Pro	[52]
		c. 1315G>A	p. Gly439Ser	
784	Netherlands	c. 2246G>A	p. Gly741Arg	[34]
		c. 2780C>T	p. Arg919Cys	
		c. 2927C>T	p. Arg968Stop	
785	Netherlands	c. 2246G>A	p. Gly741Arg	[34]
		c. 2927C>T	p. Arg968Stop	
786	Netherlands	c. CGA>TGA	p. Arg968stop	[8]
		c. GGG>AGG	p. Gly741Arg	
787	Netherlands	c. CGA>TGA	p. Arg968stop	[8]
		c. GGG>AGG	p. Gly741Arg	
788	Netherlands	c. CGA>TGA	p. Arg968stop	[8]
		c. GGG>AGG	p. Gly741Arg	
789	Poland	c. 35_36insA	p. D12EfsX17	[117]
		c. 1095+5G>A	p. IVS8+5G>A	
790	Czech Republic	c. 2576T>C	p. Leu859Pro	[118]
		c. 2929C>T	p. Arg977X	
791	Czech Republic	c. 1315G>A	p. Gly439Ser	[57]
		c. 2213T>G	p. Leu738Arg	
792	Czech Republic	c. 1315G>A	p. Gly439Ser	[57]
		c. 2213T>G	p. Leu738Arg	
793	Czech Republic	c. 480ins.C	Pro160fsX97	[57]
		c. 741+1G>A	Exon/intron boundary change	
794	Czech Republic	c. 1315G>A	p. Gly439Ser	[57]
		c. 1202insGTGATGC	p. Ala401fs2X	
795	Czech Republic	c. 1261T>C	p. Cys421Arg	[57]
		c. 2221G>A	p. Gly741Arg	
796	Czech Republic	c. 2221G>A	p. Gly741Arg	[57]
		c. 2549T>C	p. Leu850Pro	
797	Czech Republic	c. 2221G>A	p. Gly741Arg	[57]
		c. 2549T>C	p. Leu850Pro	
798	Czech Republic	c. 1315G>A	p. Gly439Ser	[57]
		c. 1664C>T	p. Ser555Leu	
		c. 2711G>A	p. Arg904Gln	
799	Australia	c. 2186G>T	p. Gly729Val	[119]
		c. 2872A>T	NA	
800	Germany	c. 738T>G	NA	[120]

		c. 741G>A	NA	
801	Germany	NA	p. Ser555Leu	[55]
		NA	p. Leu859Pro	
802	Germany	NA	p. Ala356Val	[55]
		NA	p. Gly439Ser	
803	Germany	c. 966C>T	p. Ser314Phe	[121]
		c. 1635_46 del TCTGCTCCTATG ins AACAACTTCCT	NA	
804	Germany	c. 1230C>T	p. Ser402Phe	[121]
		c. 650C>T	p. Arg209Trp	
805	Germany	c. 2238T>G	p. Leu738Arg	[34]
		c. 2246G>A	p. Gly741Arg	
806	Belgium	c. 1664C>T	p. Ser555Leu	[122]
		c. 2633+1G>C	r. 2521_2634del	
807	Denmark	c. 1606 T>C	p. Phe536Leu	[123]
		c. 2221 G>A	p. Gly741Arg	
808	Sweden	c. 935A>C	p. Thr304Pro	[124]
		c. 2207G>A	p. Ala728Thr	
		c. 2216G>A	p. Gly731Arg	
809	Sweden	c. 935A>C	p. Thr304Pro	[124]
		c. 2207G>A	p. Ala728Thr	
		c. 2216G>A	p. Gly731Arg	
810	Sweden	c. 1340G>A	p. Gly439Ser	[124]
		c. 2246G>A	p. Gly741Arg	
811	Sweden	c. 2246G>A	p. Gly741Arg	[124]
		c. 2745insAGCA	NA	
812	Sweden	c. 1934C>T	p. Pro643Leu	[56]
		c. 834_835insG	Sequence shift	
813	Sweden	c. 1931G>A	p. Arg642Gln	[56]
		c. 463G>A	p. Vall53Met	
814	Sweden	c. 520T>C	p. Trp172Arg	[56]
		c. 2859+1G>T	SP	
815	Sweden	c. 1321G>C	p. Gly439Ser	[56]
		c. 2227G>A	p. Gly741Arg	
816	Sweden	c. 1321G>C	p. Gly439Ser	[56]
		c. 2227G>A	p. Gly741Arg	
817	Sweden	c. 1321G>C	p. Gly439Ser	[56]
		c. 2227G>A	p. Gly741Arg	
818	Sweden	c. 2192G>T	p. Gly729Val	[56]
		c. 2859+1G>T	SP	
819	Sweden	c. 2897G>A	p. Arg964Gln	[56]
		c. 1321G>C	p. Gly439Ser	

820	Sweden	c. 2035G>C	p. Val677Leu	[56]
		c. 211OA	p. His69Asn	
821	Sweden	c. 2035G>C	p. Val677Leu	[56]
		c. 211OA	p. His69Asn	
822	Sweden	c. 2035G>A	p. Val677Met	[56]
		c. 2192G>T	p. Gly729Val	
823	Sweden	c. 2192G>T	p. Gly729Val	[56]
		c. 2897G>A	p. Arg964Gln	
824	Sweden	c.2114_2120del TGAAACCA	Sequence shift	[56]
		c. 2192G>T	p. Gly729Val	
825	Sweden	c. 2897G>A	p. Arg964Gln	[56]
		c. 2859+1G>T	SP	
826	Sweden	c. 1321G>C	p. Gly439Ser	[56]
		c. 2227G>A	p. Gly741Arg	
827	Sweden	c. 1321G>C	p. Gly439Ser	[56]
		c. 2227G>A	p. Gly741Arg	
828	Sweden	c. 208G>A	p. Glu68Lys	[56]
		c. 440G>A	p. Arg145His	
829	Sweden	c. 2035G>A	p. Val677Met	[56]
		c. 1031G>C	p. Gly342Ala	
830	Sweden	c. 1321G>C	p. Gly439Ser	[56]
		c. 2227G>A	p. Gly741Arg	
831	Sweden	c. 2632G>A	p. Glv876Ser	[56]
		C. 1569-1G>A	SP	
832	Sweden	c. 2192G>T	p. Gly729Aal	[56]
		c. 2227G>A	p. Gly741Arg	
833	Sweden	c. 2192G>T	p. Gly729Val	[56]
		c.2859+1G>T	SP	
834	Sweden	c. CCTTCA>CCA	p. ProSer561Pro	[8]
		c. GGC>GTC	p. Gly630Val	
835	Sweden	c. CCTTCA>CCA	p. ProSer561Pro	[8]
		c. GGC>GTC	p. Gly630Val	
836	Sweden	c. CCTTCA>CCA	p. ProSer561Pro	[8]
		c. GGC>GTC	p. Gly630Val	
837	Sweden	c. CCTTCA>CCA	p. ProSer561Pro	[8]
		c. GGC>GTC	p. Gly630Val	
838	Israel	NA	p. Pro643Leu	[125]
		NA	p. Trp1002Cys	
839	Israel	NA	p. Pro643Leu	[125]
		NA	p. Trp1002Cys	
840	Europe[1]	c. 1175C>T	p. Thr392Ile	[58]
		c. 2965G>A	p. Gly989Arg	

841	Europe[1]	c. 2576T>C	p. Leu859Pro	[58]
		c. 2883+1G>T	SP	
842	Europe[1]	c. 581C>T	p. Thr194Ile	[58]
		c. 2899A>G	p. Arg967Gly	
843	Europe[1]	c. 1465T>C	p. Tyr489His	[58]
		c. 1567G>A	p. Ala523Thr	
844	Europe[1]	c. 1121G>A	p. Gly374Glu	[58]
		c. 1335+1G>A	SP	
845	Europe[1]	c. 248G>A	p. Arg83Gln	[58]
		c. 433C>T	p. Arg145Cys	
846	Europe[1]	c. 2883+1G>T	SP	[58]
		c. 2179-?_2285+?del	p. Ala727X	
847	Europe[1]	c. 704C>G	p. Thr235Arg	[58]
		c. 1145C>T	p. Thr382Met	
848	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 1145C>T	p. Thr382Met	
849	Europe[1]	c. 1924C>T	p. Arg642Cys	[58]
		c. 2368+5G>A	SP	
850	Europe[1]	c. 1315G>A	p. Gly439Ser	[58]
		c. 2965G>A	p. Gly989Arg	
851	Europe[1]	c. 741+1G>A	SP	[58]
		c. 2687G>A	p. Arg896Gln	
852	Europe[1]	c. 2581C>T	p. Arg861Cys	[58]
		c. 2981G>A	p. Cys994Tyr	
853	Europe[1]	c. 1925G>A	p. Arg642His	[58]
		c. 2221G>A	p. Gly741Arg	
854	Europe[1]	c. 1925G>A	p. Arg642His	[58]
		c. 2221G>A	p. Gly741Arg	
855	Europe[1]	c. 1606T>C	p. Phe536Leu	[58]
		c. 2221G>A	p. Gly741Arg	
856	Europe[1]	c. -66_23del	Probably no protein is produced, because the start codon is deleted.	[58]
		c. 1180+1G>T	SP	
857	Europe[1]	c. 1196_1202dup	p. Ser402X	[58]
		c. 2965G>A	p. Gly989Arg	
858	Europe[1]	c. 938C>T	p. Ala313Val	[58]
		c. 2221G>A	p. Gly741Arg	
859	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		Deletion of exons 1 to 6	Probably no protein is produced, because the start codon is deleted.	
860	Europe[1]	c. 363G>C	p. Glu121Asp	[58]

		c. 602-16G>A	SP	
861	Europe[1]	c. 1636A>G	p. Ser546Gly	[58]
		c. 1925G>A	p. Arg642His	
862	Europe[1]	c. 1664C>T	p. Ser555Leu	[58]
		c. 2221G>A	p. Gly741Arg	
863	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2965G>A	p. Gly989Arg	
864	Europe[1]	c. 457G>A	p. Val153Met	[58]
		c. 1925G>A	p. Arg642His	
865	Europe[1]	c. 2929C>T	p. Arg977X	[58]
		c. 2748-?_2951+?deli	p. Lys918_Thr 985del	
866	Europe[1]	c. 514T>C	p. Trp172Arg	[58]
		c. 1664C>T	p. Ser555Leu	
867	Europe[1]	c. 2883+1G>T	SP	[58]
		c. 1196_1202dup	p. Ser402X	
868	Europe[1]	c. 2687G>A	p. Arg896Gln	[58]
		Deletion of exons 1 to 6	Probably no protein is produced, because the start codon is deleted.	
869	Europe[1]	c. 2687G>A	p. Arg896Gln	[58]
		c. 2221G>A	p. Gly741Arg	
870	Europe[1]	c. 2660+1G>C	SP	[58]
		c. 2891G>A	p. Arg964Gln	
871	Europe[1]	c. 1924C>G	p. Arg642Gly	[58]
		c. 2642T>C	p. Met881Thr	
872	Europe[1]	c. 1325A>G	p. Asn442Ser	[58]
		c. 2869A>T	p. Lys957X	
873	Europe[1]	c. 247C>T	p. Arg83Trp	[58]
		c. 2204C>G	p. Pro735Arg	
874	Europe[1]	c. 626G>A	p. Arg209Gln	[58]
		c. 2186G>T	p. Gly729Val	
875	Europe[1]	c. 1928C>T	p. Pro643Leu	[58]
		c. 2186G>T	p. Gly729Val	
876	Europe[1]	c. 2037+4A>G	SP	[58]
		c. 2883+1G>T	SP	
877	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2891G>A	p. Arg964Gln	
878	Europe[1]	c. 2965G>A	p. Gly989Arg	[58]
		c. 3089A>G	p. Gln1030Arg	
879	Europe[1]	c. 1387G>A	p. Gly463Arg	[58]
		c. 2581C>T	p. Arg861Cys	
880	Europe[1]	c. 1315G>A	p. Gly439Ser	[58]

		c. 2883+1G>T	SP	
881	Europe[1]	c. 1424C>G	p. Ser475Cys	[58]
		c. 1925G>A	p. Arg642His	
882	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2581C>T	p. Arg861Cys	
883	Europe[1]	c. 179C>T	p. Thr60Met	[58]
		c. 2221G>A	p. Gly741Arg	
884	Europe[1]	c. 1669+5G>T	SP	[58]
		c.2952-?_3077+?del Deletion of exon 26	p. Ile984fs	
885	Europe[1]	c. 473G>T	p. Arg158Leu	[58]
		c. 1180+1G>T	SP	
886	Europe[1]	c. 247C>T	p. Arg83Trp	[58]
		c. 514T>C	p. Trp172Arg	
887	Europe[1]	c. 2186G>T	p. Gly729Val	[58]
		c. 3052C>T	p. Arg1018X	
888	Europe[1]	c. 2883+1G>T	SP	[58]
		c. 2965G>A	p. Gly989Arg	
889	Europe[1]	c. 470T>C	p. Leu157Pro	[58]
		c. 2221G>A	p. Gly741Arg	
890	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2981G>A	p. Cys994Tyr	
891	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 2883+1G>T	SP	
892	Europe[1]	c. 450T>G	p. Ile150Met	[58]
		c. 2883+1G>T	SP	
893	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2883+1G>T	SP	
894	Europe[1]	c. 1519C>T	p. Arg507Cys	[58]
		c. 506-?_852+? Deletion of exons 4 to 6	p. Val169fs	
895	Europe[1]	c. 2542G>A	p. Asp848Asn	[58]
		c. 2965G>A	p. Gly989Arg	
896	Europe[1]	c. 910A>C	p. Thr304Pro	[58]
		c. 2883+1G>T	SP	
897	Europe[1]	c. 961C>T	p. Arg321Trp	[58]
		c. 1844C>T	p. Ser615Leu	
898	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 248G>A	p. Arg83Gln	
899	Europe[1]	c. 1964G>A	p. Arg655His	[58]
		c. 3053G>A	p. Arg1018Gln	
900	Europe[1]	c. 1175C>T	p. Thr392Ile	[58]
		c. 1325A>G	p. Asn442Ser	

901	Europe[1]	c. 2576T>C	p. Leu859Pro	[58]
		c. 3053G>A	p. Arg1018Gln	
902	Europe[1]	c. 1000C>T	p. Arg334Trp	[58]
		c. 2221G>A	p. Gly741Arg	
903	Europe[1]	c. 184G>C	p. Asp62His	[58]
		c. 237_238dup	p. Arg80fs	
904	Europe[1]	c. 533C>T	p. Ser178Leu	[58]
		c. 2827C>T	p. Arg943Trp	
905	Europe[1]	c. 505+5G>A	SP	[58]
		c. 2221G>A	p. Gly741Arg	
906	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 3052C>T	p. Arg1018X	
907	Europe[1]	c. 965-1_976delins12	p. Asp323fs	[58]
		c. 2532G>A	p. Trp844X	
908	Europe[1]	c. 1664C>T	p. Ser555Leu	[58]
		c. 2883+1G>T	SP	
909	Europe[1]	c. 2883+1G>T	SP	[58]
		c. 2221G>A	p. Gly741Arg	
910	Europe[1]	c. 2883+1G>T	SP	[58]
		c. 2221G>A	p. Gly741Arg	
911	Europe[1]	c. 2877_2878del	p. Arg959fs	[58]
		c. 2929C>T	p. Arg977X	
912	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2981G>A	p. Cys994Tyr	
913	Europe[1]	c. 2965G>A	p. Gly989Arg	[58]
		c. 1195C>T	p. Arg399Cys	
914	Europe[1]	c. 1956del	p. Asn653fs	[58]
		c. 626G>A	p. Arg209Gln	
915	Europe[1]	c. 2883+1G>T	SP	[58]
		c. 1601A>G	p. Asn534Ser	
916	Europe[1]	c. 247C>T	p. Arg83Trp	[58]
		c. 2221G>A	p. Gly741Arg	
917	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 1844C>T	p. Ser615Leu	
918	Europe[1]	c. 497C>T	p. Ala166Val	[58]
		c. 2981G>A	p. Cys994Tyr	
919	Europe[1]	c. 1924C>T	p. Arg642Cys	[58]
		c. 2965G>A	p. Gly989Arg	
920	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 363G>C	p. Glu121Asp	
921	Europe[1]	c. 2576T>C	p. Leu859Pro	[58]
		c. 2221G>A	p. Gly741Arg	
922	Europe[1]	c. 506-1G>A	SP	[58]

		c. 505+5G>A	SP	
923	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 2883+1G>T	SP	
924	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 1664C>T	p. Ser555Leu	
925	Europe[1]	c. 2089_2095del	p. Thr697fs	[58]
		c. 947G>T	p. Gly316Val	
926	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2821G>T	p. Glu941X	
927	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 2929C>T	p. Arg977X	
928	Europe[1]	c. 2204C>G	p. Pro735Arg	[58]
		c. 602-16G>A	SP	
929	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 817dup	p. Ala273fs	
930	Europe[1]	c. 1390G>A	p. Ala464Thr	[58]
		c. 2548+1G>T	SP	
931	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 1335+1G>T	SP	
932	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2981G>A	p. Cys994Tyr	
933	Europe[1]	c. 1298_1308delinsTG	p. Gln433_Cys436 delinsLeu	[58]
		c. 815T>C	p. Leu272Pro	
934	Europe[1]	c. 2611C>T	p. Arg871Cys	[58]
		c. 247C>T	p. Arg83Trp	
935	Europe[1]	c. 2115_2139dup	p. Lys706_Ile 713dup	[58]
		c. 2883+1G>T	SP	
936	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 2883+1G>T	SP	
937	Europe[1]	c. 2221G>A	p. Gly741Arg	[58]
		c. 2883+1G>A	SP	
938	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 1274G>A	p. Trp425X	
939	Europe[1]	c. 815T>C	p. Leu272Pro	[58]
		c. 1664C>T	p. Ser555Leu	

Abbreviations and note: Homo = Homozygosity; Hetero = Heterozygosity; Co-hetero = Compound heterozygosity; Co-homo = Compound homozygosity; SP = splicing mutation; del = deletion; ins = insertion; NA: Not available; Europe[1] = Most were from Netherlands, others from Belgium, Sweden and Italy. GenBank accession number NM_000339.3 is used as a reference sequence. Intron sequences can be found in NT_010498.15.

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