

Table S1. Missense IQSEC2 Mutations

No.	Mutation	Ref
1	c.128G>C p.Arg43Pro	1
2	c.770G>A p.Ser257Asn	2
3	c.1048G>A p.Ala350Thr	3
4	c.1049C>T p.Ala350Val	4
5	c.1049C>A p.Ala350Asp	4
6	c.1075C>T p.Arg359Cys	4
7	c.1076 G>A p. Arg359His	5
8	c.1688G>A p.Arg563Gln	4
9	c.2117A > G p. Asn706Ser	6
10	c.2273G>A p.Arg758Gln	4
11	c.2278G>A p.Gly760Ser	4
12	c.2293A>G p.Asn765Asp	7
13	c.2312G>A p.Gly771Asp	4
14	c.2354C>T p.Pro785Leu	4
15	c.2366C>T p.Ala789Val	4
16	c.2402A>C p.Gln801Pro	4
17	c.2507C>T p.Ala836Val	4
18	c.2582G>C p.Ser861Thr	4
19	c.2587C>T p. Arg863Trp	4
20	c.2752G>T p.Val918Phe	8
21	c.2857G > A p.Ala953Thr	9
22	c.2909G > A p.Arg970His	9
23	c.2983C>T p.Arg995Trp	10
24	c.2995C>T p.Leu999Phe	4
25	c.3005A > G p.Asp1002Gly	9
26	c. 3011 T>C p. Leu1004Prp	5
27	c.3030C > G p.Phe1010Leu	9
28	c.3034A>G p. Lys1012Glu	8
29	c. 3065G>A p.Arg1022His	11
30	c.3206G>C p. Arg1069Pro	4
31	c.3206G > A p.Arg1069Gln	9
32	c.3235T > C p.Ser1079Pro	12
33	c.3364C>T p.Arg1122Cys	13
34	c.3412G > C p. Gly1138Arg	6
35	c.3415G>A p.Alal139Thr	8
36	c.3463C>T p.Arg1155Trp	8
37	c.4204 G>A p. Arg1402Thr	5

Numbers preceeded by c refer to nucleotide numbering of the IQSEC2 cDNA with +1 corresponding to the A of the ATG translation initiation codon in the reference sequence for the IQSEC2 gene [GenBank: NM_001111125.2]. Numbers proceeded by p refer to the amino acid number of the 1488 amino acid isoform of the IQSEC2 protein.

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