Table S2. Non Missense IQSEC2 Mutations

No.	Mutation	Ref
1	c.55_151delinsAT p.Ala19Ile fs*32	1
2	c.83_85del p.Asp28del	1
3	c.88_90delATC p.Ile30del	1
4	c.97C>T p.Gln33*	1
5	c.184C>T p.Arg62*	1
6	c.267C>Gp. Thr89*	2
7	c.273_282del p.Asn91Lys fs*112	1
8	c.316C > T p.Gln106*	3
9	c.325delC insGC p. Gln1084Ala fs*22	4
10	c.443_4 44dup p.Ala149Gln fs*58	3
11	c.556C > A p. S189*	5
12	c.588_610del p.Arg197Ala fs*34	6
13	c.675dup p.Ser226 fs	7
14	c.737+10385del p.?	6
15	c.738-1G>A Splicing	1
16	c.804delC p.Try269Thr fs*3	1
17	c.847_848del insT, p.Gly283Ser fs*23	4
18	c.854del p.Pro285Leu fs*21	1
19	c.895C>T p.Gln299*	1
20	c.928G>T p.Glu310*	1
21	c.999+8A>G	2
22	c.1000_1034del splicing	1
23	c.1170dupG p.Gln391Ala fs*5	8
24	c.1405_1406 del p.Lys469Val fs*4	1
25	c.1417G> T p.Glu473*	3
26	chrX: 53251127_53251128 dup p.Ser484*	9
27	c.1459_1460delAT p.Met487Val fs*2	10
28	c.1510C>T p.Gln504*	1
29	c.1556_1599delACCT, p.Tyr519Trp fs*87	4
30	c.1567_2199 del ins GGC p.Thr523_Thr733 del ins Gly	1
31	c.1591C> T, p. Arg531*	4
32	c.1618C>T p.Gln540*	1
33	c.1744_1763 del p.Arg582Cys fs*9	1
34	c.1813_1814del, p.Asp605Pro fs*3	11
35	c.1861dup	12
36	c.1881delC p.His629Met fs*4	2
37	c.1983_1999 del p.Leu662Gln fs*25	1
38	c.2026del p.Ala676Leu fs*46	13
39	c.2052_2053 delCG p.Cys684*	1
40	c.2078delG p.Gly693Val*29	1
41	c.2184C > G p.Tyr728*	14
42	c.2203C>T p.Gln735*	1
43	c.2272C>T p.Arg758*	1
44	c.2295_2297del p.Asn765del	6

45	c.2317C>T p.Gln773*	1
46	c.2317_2332del p.Gln773Glyfs*25	1
47	c.2329G>T p.Glu777*	15
48	c.2459+21C>T splicing	16
49	c.2521C > T p.Glu841*	7
50	c.2563C>T p.Arg855*	1
51	c.2662dup p.Ile888Asn fs*16	1
52	c.2679_2680 ins A p.Asp894 fs*10	1
53	c.2776C>T p.Arg926*	1
54	c.2799C>G p.Try933*	1
55	c.2846_2852 del CCCAGGT p.Ser949Cys fs*7	1
56	c.2854C>T p.Gln952*	1
57	c.2911C > T, p. Arg971*	4
58	c.2962C>T p.Gln988*	1
59	c.3079delC p.Leu1027Ser fs*75	1
60	c.3097C>T p.Gln1033*	1
61	g.88032_88033del splicing	1
62	c.3163C>T p.Arg1055*	1
63	c.3277+2T>G Splicing	1
64	c.3277+5G>A Splicing	1
65	c.3278C>A p.Ser1093*	1
66	c.3300dup p.Met1101Tyr fs*5	16
67	c.3322C>T p.Gln1108*	1
68	c.3387C>A p.Tyr1129*	1
69	c.3433C>T p.Arg1145*	1
70	c.3457del p.Arg1153Gly fs*244	1
71	c.3613_3613delC p. Leu1205Trp fs*192	2
72	c.3669_3733del p.Thr1225Ser fs*4	1
73	c.3780delG p.Gln1261Ser fs*136	2
74	c.3801_3808dup p.Gln1270Arg fs*130 ,	10
75	c.3859C>T p.Gln1287*	2
76	c.4039dupG p.Ala1347Gly fs*40	1
77	c.4110-4111 del p. Tyr1371Gln fs*15	2
78	c.4164dupC p.Ile1389 His fs*218	17
79	c.4246_4247insG p.Ser1416 fs	18
80	c.4401del p.Gly1468 Ala fs*27	1
81	c.4419 Ser1474Val fs*21	19
82	c.4419_4420 insC Ser1474Gln fs*133	2
83	c.4419_4431del p.Ser1474Arg fs*17	2
84	chrX:52789239_53368927dup (579 kb)	1
85	chrX:52911287_53315010dup (403 kb)	1
86	chrX:52920728_53321125del (0.4 Mb)	1
87	chrX:52954520_53315542dup (361 kb)	1
88	chrX:53276030-53298472 dup/truncation	1
89	chrX:53283513- 53325282 dup/truncation	1

90	46 X,t(X;20)(p11.2;q11.2) translocation	1
91	Dup on chr4: g.183693432_18375617dup gain 62 kb Insertion point on chrX g. 53318362_53318363	1

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. * indicates introduction of a termination codon after the indicated number of amino acids where relevant. dup = duplication; ins=insertion; del=deletion; fs=frameshift.

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