Reported point mutations in the medical literature and public databases (ClinVar and LOVD), updated 2/2/2017. Builds NM_015265.1, NM_001172517.1, and NM_001172509.1 correspond to the same locations. Exon locations according to NM_015265 are provided

Exon	Nucleotide	Protein	Inheritance	Reference
3	c.83delC	p.P28Qfs*2	de novo	9
3	c.124G>T	p.G42*	de novo	7
4-5	c.346+2T>G	p.G115fs*15	de novo	2
4-5	c.346G>C	p.G116R	de novo	7
5	c.400delG	p.A134Hfs*17	de novo	7,10
6	c.482delA	p.K161SfsX19	de novo	7,10
6	c.583dupT	p.C195Lfs*14	de novo	7,10
6	c.593_596delInsA	p.S198*	de novo	10,11
8	c.715C>T	p.R239*	de novo	1,9
8	c.715C>T	p.R239*	de novo	3,9
8	c.748C>T	p.Q250*	de novo	2
8	c.816delT	p.H273Tfs*21	de novo	7,10
8	c.847C>T	p.R283*	de novo	2,9
8	c.847C>T	p.R283*	de novo	7,10
8	c.868C>T	p.Q290*	de novo	9
8	c.1131_1132delGT	p.S378Pfs*18	?	9
8	c.1142T>G	p.V381G	de novo	4
8	c.1165C>T	p.R389C	de novo	5
8	c.1166G>T	p.R389L	de novo	10,11
8	c.1169C>T	p.T390I	de novo	9
8	c.1171C>T	p.Q391*	de novo	2
8-9	c.1173+2T>C	p.?	de novo	10,11
9	c.1186G>C	p.E396Q	de novo	6
9	c.1196G>C	p.R399H	de novo	9
9	c.1196G>C	p.R399H	de novo	10,11
9	c.1198A>G	p.K400E	de novo	9
9	c.1204G>A	p.E402K	de novo	10,11
9	c.1255C>T	p.Q419*	de novo	7,10
9	c.1285C>T	p.R429*	de novo	9
9	c.1286G>A	p.R429Q	de novo	7,9
9	c.1286G>A	p.R429Q	de novo	7
9	c.1375C>T	p.R459*	de novo	6
9	c.1375C>T	p.R459*	de novo	10,11
10	c.1495A>T	p.K499*	de novo	9
10	c.1515delT	p.505Lfs*41	de novo	9
10-11	c.1542+1G>A	p.?	de novo	10

11	c.1543G>A	p.G515S	de novo	10,11
11	c.1728delT	p.E577Sfs*47	?	7,10
12	c.1942_1943delCT	p.L648Ffs*40	de novo	9
12	c.1945dupT	p.S649Ffs*40	de novo	2
12	c.1964C>T	p.P655L	de novo	7
12	c.2018dupA	p.H673Qfs*16	de novo	8

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