

Location	cDNA	Protein	Mutation status	Reference	Patient	Number of patients	Sex
Exon 1	c.34_35delCT	p.Leu12Alafs*67	Hom	Cho et al., (2012)	Family 1	1	1 Female
Exon 1	c.34_35delCT	p.Leu12Alafs*67	Hom	Jaureguiberry et al., (2013)	Family 6	1	1 Male
Exon 1	c.34_35delCT	p.Leu12Alafs*67	Het	Jaureguiberry et al., (2013)	Family 16	2	2 Females
Exon 1	c.174-175ins29	p.Arg59Argfs*85	Hom	Cabral et al., (2013)	Family 1	12	10 Males; 2 Females
Exon 1	c.217C>T	p.Arg73*	Het	Jaureguiberry et al., (2013)	Family 11	2	1 Male; 1 Female
Exon1	c.349_367del19	p.Leu117Cysfs*22	Het	Kantaputra et al., (2013)	Family 1	1	1 Male
Intron 1	c.405-1G>C		Het	Wang et al. (2014)	Family 1	1	1 Female
Exon 2	c.406C>T	p.Arg136*	Hom	O'Sullivan et al., (2011)	Family 1	4	1 Male; 3 Females
Exon 2	c.406C>T	p.Arg136*	Het	Wang et al., (2013)	Family 3	2	2 Males
Exon 2	c.406C>T	p.Arg136*	Hom	Jaureguiberry et al., (2013)	Family 5	1	1 Female
Exon 2	c.518T>G	p.Leu173Arg	Hom	Jaureguiberry et al., (2013)	Family 9	1	1 Male
Intron 2	c.589+1G>A		Het	Jaureguiberry et al., (2013)	Family 2	2	1 Male; 1 Female
Intron 2	c.590-2A>G	p.Asp197_Ile214delinsVal	Het	Cho et al., (2012)	Family 4	1	1 Female
Exon 3	c.612delC	p.Leu205Cysfs*11	Het	Jaureguiberry et al., (2013)	Family 16	2	1 Female
Exon 4	c.641_719del	p.Ile214Asnfs*46	Het	Jaureguiberry et al., (2013)	Family 13	2	1 Male; 1 Female
Intron 4	c.719+1G>C		Het	Jaureguiberry et al., (2013)	Family 3	2	1Male; 1 Female
Intron 4	c.720-2A>G	p.Gln241_Arg271del	Hom	Wang et al., (2013)	Family 2	1	1 Male
Exon 5	c.727C>T	p.Arg243*	Het	Jaureguiberry et al., (2013)	Family 10	1	2 Female
Exon 5	c.727C>T	p.Arg243*	Het	Jaureguiberry et al., (2013)	Family 11	2	1 Male; 1 Female
Exon 5	c.755_757delTCT	p.Phe252del	Het	Jaureguiberry et al., (2013)	Family 13	2	1 Male; 1 Female
Intron 5	c.812+2T>G		Hom	Jaureguiberry et al., (2013)	Family 14	1	1 Female
Intron 5	c.813-2A>G	p.Arg271Serfx*70	Hom	Cho et al., (2012)	Family 2	4	3 Males; 1 Female
Exon 6	c.826C>T	p.Arg276*	Het	Cho et al., (2012)	Family 4	1	1 Female
Exon 6	c.907_908delAG	p.Ser303Cysfs*76	Hom	Jaureguiberry et al., (2013)	Family 15	3	2 Males; 1 Female
Exon 6	c.913_914delTT	p.Phe305Leufs*74	Het	Jaureguiberry et al., (2013)	Family 2	2	1 Male; 1 Female
Exon6	c.915_918delCTTT	p.Phe305Leufs*76	Hom	Jaureguiberry et al., (2013)	Family 1	1	1 Male
Exon 6	c.915_918delCTTT	p.Phe305Leufs*76	Het	Kantaputra et al., (2013)	Family 1	1	1 Male
Exon 7	c.992G>A	p.Gly331Asp	Hom	Wang et al., (2013)	Family 1	3	1 Male 2 Females
Exon 8	c.1175_1179delGGCTC	p.Arg392Profs*22	Hom	Cho et al., (2012)	Family 3	2	2 Males
Exon 8	c.1207G>A	p.Asp403Asn	Het	Wang et al. (2014)	Family 1	1	1 Female
Exon 9	c.1228_1229delGA	p.Asp410Profs*5	Het	Jaureguiberry et al., (2013)	Family 10	1	1 Female
Intron 9	c.1302-2A>G		Het	Kantaputra et al., (2013)	Family 2	1	1 Female
Exon10	c.1348_1349delTC	p.Ser450Profs*20	Het	Jaureguiberry et al., (2013)	Family 3	2	1 Male; 1 Female
Intron 10	c.1361+4del		Hom	Wang et al. (2014)	Family 2	2	1Male; 1 Female
Exon 11	c.1369A>T	p.Lys457*	Hom	Jaureguiberry et al., (2013)	Family 12	1	1 Female
Exon 11	c.1432C>T	p.Arg478*	Hom	Jaureguiberry et al., (2013)	Family 8	1	1 Male
Exon 11	c.1432C>T	p.Arg478*	Het	Wang et al., (2013)	Family 3	2	1 Male; 1 Female
Exon 11	c.1480delC	p.His494fs*13	Het	Kantaputra et al., (2013)	Family 2	1	1 Female
Exon 11	c.1475_1482dupAACCCAC	p.Leu495Asnfs*15	Hom	Jaureguiberry et al., (2013)	Family 4	2	1 Male; 1 Female
Exon 11	c.1513delA	p.Ile505Serfs*2	Hom	Jaureguiberry et al., (2013)	Family 7	2	1 Male; 1 Female
Total reported	40 (33 unique mutations)		18 Hom; 22 Het		29 Families	60 index cases	32 Males; 28 Females