Supp. Table S1. Previously reported pathogenic *BMPR2* mutations identified in pulmonary arterial hypertension

Location	Mutation category	Nucleotide change[†]	Amino acid change	Frequency in this study[‡]	Clinical classification	Reference(s)
5'UTR to intron 3	Deletion	c.-127936_418+7067del	p.?	1	H	Kataoka et al., 2013
5'UTR	Transition	c.-669G>A	p.?	1	H	Wang et al., 2009
5'UTR to exon 1	Deletion	c.?_-540_76+?del	p.?	[2]	H (n=2)	Pfarr et al., 2011
Exon 1	Deletion	c.1-?_76+?del	p.?	1	CHD-APAH (hereditary)	Pfarr et al., 2013
Exons 1-3	Deletion	c.1-?_418+?del	p.?	[2]	H, NK	Girerd et al., 2010; Kabata et al., 2013
Exons 1-13	Deletion	c.1-?_3117+?del	p.?	1	NK	Sztrymf et al., 2008
Exon 1	Nonsense	c.16C>T	p.Q6*	1	I	Kataoka et al., 2013
Exon 1	Frameshift	c.21delG	p.W9Gfs*38	1	I	Liu et al., 2012
Exon 1	Nonsense	c.27G>A	p.W9*	1	I	Liu et al., 2012
Exon 1	Nonsense	c.38G>A	p.W13*	1	P	Chida et al., 2012
Exon 1	Nonsense	c.39G>A	p.W13*	1	H	Hamid et al., 2010
Exon 1	Nonsense	c.47G>A	p.W16*	1	P	Kerstjens-Frederikse et al., 2013
Exon 1	Nonsense	c.48G>A	p.W16*	4	H, I, NK (n=2)	Sztrymf et al., 2008; Pfarr et al., 2011; Liu et al., 2012
Exons 1-6	Deletion	c.51_814del	p.I18Hfs*25	1	I	Machado et al., 2001
Intron 1	Splice-site	c.77-35_86del	p.?	1	I	Liu et al., 2012
Exon 2	Deletion	c.77-?_247+?del	p.?	1	NK	Girerd et al., 2010
Exon 2	Duplication	c.77-?_247+?dup	p.?	1	H	Cogan et al., 2006
Exons 2-3	Deletion	c.77-?_418+?del	p.?	1	H	Pfarr et al., 2011
Exons 2-3	Duplication	c.77-?_418+?dup	p.?	1	I	Liu et al., 2012
Exons 2-7	Duplication	c.77-?_967+?dup	p.?	1	I	Liu et al., 2012
Exons 2-13	Deletion	c.77-?_3117+?del	p.?	1	H	Cogan et al., 2006
Exon 2	Nonsense	c.88C>T	p.Q30*	1	I	Liu et al., 2012
Exon 2	Nonsense	c.91G>T	p.E31*	1	H	Pfarr et al., 2011
Exon 2	Frameshift	c.103delG	p.A35Rfs*12	1	I	Liu et al., 2012
Exon 2	Frameshift	c.116delC	p.P39Rfs*8	1	I	Rosenzweig et al., 2008
Exon 2	Missense	c.178T>G	p.C60G	1	I	Liu et al., 2012
Exon 2	Missense	c.196T>C	p.C66R	1	I	Wang et al., 2009
Exon 2	Missense	c.197G>A	p.C66Y	1	NK	Girerd et al., 2010
Exon 2	Missense	c.200A>G	p.Y67C	2	I, NK	Girerd et al., 2010; Wang et al., 2010
Exon 2	Frameshift	c.200dupA	p.Y67*	1	I	Kataoka et al., 2013
Exon 2	Frameshift	c.237delT	p.V80*	1	H	Kataoka et al., 2013
Exon 2	Frameshift	c.240_241insT	p.K81*	1	H	Johri et al., 2010
Exon 2	Nonsense	c.244C>T	p.Q82*	1	H	Pfarr et al., 2011

Location	Mutation category	Nucleotide change[†]	Amino acid change	Frequency in this study[‡]	Clinical classification	Reference(s)
Intron 2	Splice-site	c.247+2delC	p.G47_Q82del, G63_Q82del	1	H	Cogan et al., 2006
Intron 2	Splice-site	c.247+6T>G	p.G47_Q82del, G63_Q82del	1	H	Cogan et al., 2006
Intron 2 to exon 3	Deletion	c.248-592_413delinsGTAAAGTA	p.?	1	I	Aimi et al., 2013
Intron 2	Splice-site	c.248-3T>G	p.?	1	P	Chida et al., 2012
Exon 3	Deletion	c.248-?_418+?del	p.?	[2]	I (n=2)	Pfarr et al., 2011; Kabata et al., 2013
Exon 3	Missense	c.251G>T	p.C84F	1	I	Liu et al., 2012
Exon 3	Nonsense	c.255G>A	p.W85*	1	H	Kataoka et al., 2013
Exon 3	Nonsense	c.274C>T	p.Q92*	1	I	Kabata et al., 2013
Exon 3	Frameshift	c.277dupG	p.E93Gfs*5	1	I	Cogan et al., 2006
Exon 3	Nonsense	c.292G>T	p.E98*	1	I	Wang et al., 2009
Exon 3	Nonsense	c.320C>G	p.S107*	1	NK	Sztrymf et al., 2008
Exon 3	Missense	c.338A>G	p.Y113C	1	I	Liu et al., 2012
Exon 3	Frameshift	c.338dupA	p.Y113*	2	H, I	Kabata et al., 2013; Kataoka et al., 2013
Exon 3	Nonsense	c.339C>A	p.Y113*	1	I	Liu et al., 2012
Exon 3	Missense	c.349T>C	p.C117R	1	I	Liu et al., 2012
Exon 3	Missense	c.353G>A	p.C118Y	1	I	Pfarr et al., 2011
Exon 3	Missense	c.367T>C	p.C123R	1	NK	Girerd et al., 2010
Exon 3	Frameshift	c.371dupA	p.N124Kfs*6	1	NK	Girerd et al., 2010
Exon 3	Missense	c.377A>G	p.N126S	5	H, I (n=2), NK (n=2)	Girerd et al., 2010; Pfarr et al., 2011; Liu et al., 2012
Exon 3	Frameshift	c.399delT	p.P134Lfs*18	1	P	Kerstjens-Frederikse et al., 2013
Exon 3	Frameshift	c.407_408delCA	p.T136Nfs*10	1	NK	Girerd et al., 2010
Exon 3	Frameshift	c.408_412delAACAC	p.P138Qfs*7	1	NK	Girerd et al., 2010
Intron 3	Splice-site	c.418+3A>T	p.?	2	NK (n=2)	Sztrymf et al., 2008
Intron 3	Splice-site	c.418+5G>A	p.?	1	H	Pfarr et al., 2011
Intron 3	Splice-site	c.419-10T>C	p.?	1	P	Pfarr et al., 2013
Exons 4-5	Deletion	c.419-?_621+?del	p.?	1	P	Pfarr et al., 2013
Exons 4-8	Duplication	c.419-?_1128+?dup	p.?	1	NK	Girerd et al., 2010
Exons 4-13	Deletion	c.419-?_3117+?del	p.?	1	H	Pfarr et al., 2011
Exon 4	Frameshift	c.420_421insG	p.P141Afs*6	1	I	Arbustini et al., 2008
Exon 4	Nonsense	c.439C>T	p.R147*	7	H (n=2), I (n=2), NK (n=3)	Girerd et al., 2010; Wang et al., 2010; Pfarr et al., 2011; Liu et al., 2012; Momose et al., 2015
Exon 4	Frameshift	c.449dupC	p.I151Nfs*30	1	NK	Girerd et al., 2010
Exon 4	Frameshift	c.498delT	p.A167Pfs*9	2	I (n=2)	Kabata et al., 2013; Kataoka et al., 2013
Exon 4	Nonsense	c.507C>A	p.C169*	1	I	Pfarr et al., 2011
Exon 4	Nonsense	c.516C>G	p.Y172*	1	H	Cogan et al., 2006

Location	Mutation category	Nucleotide change[†]	Amino acid change	Frequency in this study[‡]	Clinical classification	Reference(s)
Exon 4	Frameshift	c.528delA	p.G177Efs*10	2	NK (n=2)	Girerd et al., 2010
Intron 4	Splice-site	c.529+2T>C	p.?	1	P	Chida et al., 2012
Exon 5	Frameshift	c.551_573del	p.H184Rfs*8	1	NK	Sztrymf et al., 2008
Exon 5	Frameshift	c.608_609delTG	p.L203Qfs*16	1	I	Wang et al., 2009
Exon 6	Deletion	c.622-?_852+?del	p.?	1	NK	Girerd et al., 2010
Exons 6-7	Deletion	c.622-?_967+?del	p.?	1	I-PAVM	Handa et al., 2014
Exon 6	Nonsense	c.631C>T	p.R211*	4	H (n=2), NK (n=2)	Girerd et al., 2010; Portillo et al., 2010; Pfarr et al., 2011
Exon 6	Nonsense	c.637C>T	p.R213*	1	H-IPF	Raamsteeboers et al., 2014
Exon 6	Frameshift	c.659dupG	p.S221Lfs*4	1	H	Pfarr et al., 2011
Exon 6	Missense	c.690A>T	p.K230N	1	P	Hayes et al., 2014
Exon 6	Missense	c.727G>A	p.E243K	1	I	Wang et al., 2010
Exon 6	Missense	c.727G>C	p.E243Q	1	P	Chida et al., 2012
Exon 6	Frameshift	c.782_783delTA	p.I261Sfs*4	1	NK	Girerd et al., 2010
Exon 6	Frameshift	c.796_799delAGAG	p.R266Sfs*12	1	H	Cogan et al., 2006
Exon 6	Frameshift	c.802dupA	p.T268Nfs*30	1	H	Wang et al., 2010
Exon 6	Frameshift	c.804delT	p.A269Qfs*10	1	H	Cogan et al., 2006
Exon 6	Missense	c.830T>C	p.L277P	2	I, NK	Sztrymf et al., 2008; Liu et al., 2012
Intron 6	Splice-site	c.852+1G>A	p.?	1	NK	Girerd et al., 2010
Intron 6	Splice-site	c.853-2A>G	p.?	1	I	Kataoka et al., 2013
Exon 7	Duplication	c.853-?_967+?dup	p.?	1	H	Aldred et al., 2006
Exon 7	Frameshift	c.872delT	p.L291*	1	H	Cogan et al., 2006
Exon 7	Nonsense	c.961C>T	p.R321*	6	I (n=5), NK	Girerd et al., 2010; Wang et al., 2010; Pfarr et al., 2011
Intron 7	Splice-site	c.967+5G>C	p.?	1	I	Liu et al., 2012
Intron 7	Splice-site	c.967+5G>T	p.?	1	NK	Girerd et al., 2010
Intron 7	Splice-site	c.968-2A>C	p.?	1	I	Wang et al., 2010
Exon 8	Deletion	c.968-?_1128+?del	p.?	[2]	NK (n=2)	Girerd et al., 2010
Exons 8-9	Deletion	c.968-?_1276+?del	p.?	1	I	Aldred et al., 2006
Exons 8-10	Duplication	c.968-?_1413+?dup	p.?	1	I	Liu et al., 2012
Exon 8	Missense	c.992A>G	p.H331R	1	I	Kabata et al., 2013
Exon 8	Nonsense	c.994C>T	p.R332*	3	I (n=2), NK	Girerd et al., 2010; Portillo et al., 2010; Liu et al., 2012
Exon 8	Missense	c.1016T>A	p.V339D	2	H, I	Kabata et al., 2013; Kataoka et al., 2013
Exon 8	Frameshift	c.1044delT	p.I349Lfs*8	1	H	Momose et al., 2015
Exon 8	Frameshift	c.1093_1098delinsG	p.R365Gfs*5	1	I	Liu et al., 2012
Intron 8	Splice-site	c.1128+1G>T	p.?	1	H	Pfarr et al., 2011

Location	Mutation category	Nucleotide change[†]	Amino acid change	Frequency in this study[‡]	Clinical classification	Reference(s)
Intron 8	Splice-site	c.1129-3C>G	p.?	1	I	Liu et al., 2012
Exon 9	Frameshift	c.1141dupA	p.R381Kfs*18	1	H	Hamid et al., 2009
Exon 9	Nonsense	c.1146T>G	p.Y382*	1	I	Rosenzweig et al., 2008
Exon 9	Missense	c.1151C>T	p.A384V	1	I	Kabata et al., 2013
Exon 9	Missense	c.1156G>A	p.E386K	1	I	Rosenzweig et al., 2008
Exon 9	Missense	c.1157A>C	p.E386A	1	I	Kabata et al., 2013
Exon 9	Missense	c.1157A>G	p.E386G	1	I	Pfarr et al., 2011
Exon 9	Missense	c.1175T>C	p.V392A	2	I (n=2)	Wang et al., 2010; Liu et al., 2012
Exon 9	Nonsense	c.1207C>T	p.Q403*	1	H	Kabata et al., 2013
Exon 9	Missense	c.1228G>A	p.G410R	1	H	Liu et al., 2012
Exon 9	Nonsense	c.1243G>T	p.E415*	1	I	Wang et al., 2010
Exon 9	Missense	c.1258T>C	p.C420R	1	H	Pfarr et al., 2011
Exon 9	Missense	c.1259G>A	p.C420Y	2	H, I	Pfarr et al., 2011; Liu et al., 2012
Intron 9	Splice-site	c.1276+3A>T	p.?	1	NK	Girerd et al., 2010
Introns 9-10	Deletion	c.1277-289_1413+4737del	p.?	1	I	Kataoka et al., 2013
Intron 9	Splice-site	c.1277-9A>G	p.?	1	NK	Girerd et al., 2010
Exon 10	Deletion	c.1277-?_1413+?del	p.?	[10]	I (n=3), NK (n=7)	Sztrymf et al., 2008; Girerd et al., 2010; Liu et al., 2012; Kabata et al., 2013
Exon 10	Duplication	c.1277-?_1413+?dup	p.?	1	I	Liu et al., 2012
Exon 10	Nonsense	c.1296C>G	p.Y432*	1	H	Pfarr et al., 2011
Exon 10	Nonsense	c.1297C>T	p.Q433*	2	I, P	Pfarr et al., 2011; Pfarr et al., 2013
Exon 10	Frameshift	c.1313_1316delCAGA	p.T438Rfs*35	1	I	Pfarr et al., 2011
Exon 10	Missense	c.1346T>G	p.M449R	1	H	Cogan et al., 2006
Exon 10	Nonsense	c.1348C>T	p.Q450*	2	I, NK	Girerd et al., 2010; Pfarr et al., 2011
Exon 10	Frameshift	c.1366delinsCA	p.E456Qfs*15	1	NK	Girerd et al., 2010
Exon 10	Frameshift	c.1371dupA	p.Q458Tfs*13	1	I	Liu et al., 2012
Exon 10	Frameshift	c.1387_1388insA	p.P463Hfs*8	1	I	Pfarr et al., 2011
Exon 10	Frameshift	c.1392delA	p.A465Pfs*9	1	NK	Girerd et al., 2010
Exon 10	Nonsense	c.1397G>A	p.W466*	1	I	Pfarr et al., 2011
Exon 10	Frameshift	c.1401delA	p.E468Kfs*6	1	NK	Girerd et al., 2010
Intron 10	Splice-site	c.1413+1G>A	p.?	2	I, NK	Girerd et al., 2010; Pfarr et al., 2011
Intron 10	Splice-site	c.1413+3A>T	p.?	1	NK	Pfarr et al., 2011
Exons 11-12	Deletion	c.1414-?_2866+?del	p.?	[2]	I, NK	Girerd et al., 2010; Pfarr et al., 2011
Exons 11-13	Deletion	c.1414-?_3117+?del	p.?	[3]	I, NK (n=2)	Girerd et al., 2010; Liu et al., 2012
Exon 11	Nonsense	c.1424C>A	p.S475*	1	NK	Girerd et al., 2010
Exon 11	Missense	c.1460A>T	p.D487V	1	H	Pfarr et al., 2011
Exon 11	Missense	c.1471C>T	p.R491W	13	H (n=5), I (n=3),	Girerd et al., 2010; Wang et al., 2010;

Location	Mutation category	Nucleotide change[†]	Amino acid change	Frequency in this study[‡]	Clinical classification	Reference(s)
					NK (n=5)	Pfarr et al., 2011; Liu et al., 2012
Exon 11	Missense	c.1472G>A	p.R491Q	5	H (n=2), I (n=2), P	Portillo et al., 2010; Pfarr et al., 2011; Liu et al., 2012; Pfarr et al., 2013
Exon 11	Nonsense	c.1483C>T	p.Q495*	1	NK	Pfarr et al., 2011
Exon 11	Nonsense	c.1523G>A	p.W508*	1	H	Pfarr et al., 2011
Exon 11	Nonsense	c.1525G>T	p.E509*	1	NK	Girerd et al., 2015
Exon 12	Nonsense	c.1750C>T	p.R584*	1	I	Pfarr et al., 2011
Exon 12	Nonsense	c.1771C>T	p.R591*	1	NK	Sztrymf et al., 2008
Exon 12	Frameshift	c.1968dupA	p.Q657Tfs*18	1	H	Kataoka et al., 2013
Exon 12	Nonsense	c.1978G>T	p.E660*	1	NK	Girerd et al., 2015
Exon 12	Frameshift	c.2009delC	p.P670Qfs*30	1	H	Kabata et al., 2013
Exon 12	Frameshift	c.2128delC	p.L710Sfs*2	2	I (n=2)	Kabata et al., 2013; Kataoka et al., 2013
Exon 12	Frameshift	c.2286delC	p.N764Ifs*8	1	P	Chida et al., 2012
Exon 12	Frameshift	c.2308delC	p.R770Gfs*2	2	H, I	Rosenzweig et al., 2008; Pfarr et al., 2011
Exon 12	Frameshift	c.2413dupA	p.T805Nfs*8	1	I	Girerd et al., 2015
Exon 12	Frameshift	c.2446_2447dupGT	p.N817Lfs*23	1	I	Liu et al., 2012
Exon 12	Frameshift	c.2503_2506delACAA	p.T835Pfs*3	2	I (n=2)	Liu et al., 2012; Kabata et al., 2013
Exon 12	Frameshift	c.2503dupA	p.T835Nfs*8	1	I	Kabata et al., 2013
Exon 12	Frameshift	c.2504delC	p.T835Kfs*4	1	I	Cogan et al., 2006
Exon 12	Frameshift	c.2521_2522dupCA	p.R842Ifs*18	1	NK	Sztrymf et al., 2008
Exon 12	Missense	c.2588G>A	p.S863N	1	H	Wang et al., 2009
Exon 12	Nonsense	c.2617C>T	p.R873*	7	I (n=5), NK (n=2)	Sztrymf et al., 2008; Pfarr et al., 2011; Liu et al., 2012; Kabata et al., 2013
Exon 12	Nonsense	c.2626C>T	p.Q876*	1	H	Pfarr et al., 2011
Exon 12	Frameshift	c.2668delA	p.R890Gfs*6	1	P	Pfarr et al., 2013
Exon 12	Nonsense	c.2695C>T	p.R899*	6	H (n=2), I (n=2), NK, P	Girerd et al., 2010; Wang et al., 2010; Pfarr et al., 2011; Liu et al., 2012; Kerstjens-Frederikse et al., 2013
Exon 12	Nonsense	c.2752C>T	p.Q918*	2	NK (n=2)	Girerd et al., 2015

[†]GenBank reference sequence and version number for *BMPR2*: NM_001204.6; numbering is from +1 as A of the ATG initiation codon

[‡]Total number of independent cases. Frequencies in square brackets denote chromosomal rearrangements for which the breakpoints are unknown and may therefore represent distinct mutations

Key to abbreviations: CHD-APAH: congenital heart disease-associated pulmonary arterial hypertension; H: heritable pulmonary arterial hypertension; I: idiopathic pulmonary arterial hypertension; IPF: idiopathic pulmonary fibrosis; NK: not known; P: pediatric pulmonary arterial hypertension; PAVM: pulmonary arteriovenous malformation

Supp. Table S2. Allelic series of 384 mutations identified across the *BMPR2* locus in pulmonary arterial hypertension

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
5'UTR to intron 3	Deletion	c.-127936_418+7067del	p.?	c.1-128k_418+7kdel
5'UTR	Indel	c.-947_-946delinsAT	p.?	c.*-944/5GC>AT
5'UTR	Transition	c.-669G>A	p.?	G-669A
5'UTR to exon 1	Deletion	c.?_-540_76+?del	p.?	c.?_-540_76_?del
5'UTR to exon 13	Deletion	c.?_-540_3117+?del	p.?	
Exon 1	Deletion	c.1-?_76+?del	p.?	?_IVS1 del
Exons 1-3	Deletion	c.1-?_418+?del	p.?	
Exons 1-4	Deletion	c.1-?_529+?del	p.?	c.1-?_419+?del
Exons 1-8	Deletion	c.1-?_1128+?del	p.?	del exon 1-8
Exons 1-13	Deletion	c.1-?_3117+?del	p.?	
Exon 1	Frameshift	c.9dupC	p.S4Lfs*34	
Exon 1	Nonsense	c.16C>T	p.Q6*	
Exon 1	Frameshift	c.16_20delCAGCG	p.Q6Afs*30	c.15_19delGCAGC
Exon 1	Frameshift	c.21delG	p.W9Gfs*38	
Exon 1	Frameshift	c.21_29delinsA	p.P8Gfs*27	c.21_29delGCCCTGGCGinsA
Exon 1	Nonsense	c.27G>A	p.W9*	
Exon 1	Missense	c.28C>T	p.R10W	p.T10W
Exon 1	Nonsense	c.38G>A	p.W13*	
Exon 1	Nonsense	c.39G>A	p.W13*	
Exon 1	Frameshift	c.44delC	p.P15Hfs*32	del44C
Exon 1	Nonsense	c.47G>A	p.W16*	
Exon 1	Nonsense	c.48G>A	p.W16*	
Exons 1-6	Deletion	c.51_814del	p.I18Hfs*25	51-814 del
Exon 1	Missense	c.71C>A	p.A24E	
Intron 1	Splice-site	c.76+1G>T	p.?	
Intron 1	Splice-site	c.76+2T>C	p.?	
Intron 1	Splice-site	c.76+5G>A	p.?	
Intron 1	Splice-site	c.77-35_86del	p.?	c.77-36_85del
Intron 1	Splice-site	c.77-1G>A	p.A26Efs*9	
Exon 2	Deletion	c.77-?_247+?del	p.A26_Q82del	IVS1_IVS2 del
Exon 2	Duplication	c.77-?_247+?dup	p.?	IVS1_IVS2 dup
Exons 2-3	Deletion	c.77-?_418+?del	p.?	c.76-?_420+?del Del c.77?-c.418?
Exons 2-3	Duplication	c.77-?_418+?dup	p.?	c.77-?_c.421+?dup
Exons 2-5	Deletion	c.77-?_621+?del	p.?	
Exons 2-7	Duplication	c.77-?_967+?dup	p.?	c.77-?_c.967+?dup

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exons 2-9	Deletion	c.77-?_1276+?del	p.?	
Exons 2-13	Deletion	c.77-?_3117+?del	p.?	IVS1_? Del
Exon 2	Nonsense	c.82C>T	p.Q28*	
Exon 2	Nonsense	c.88C>T	p.Q30*	
Exon 2	Nonsense	c.91G>T	p.E31*	
Exon 2	Missense	c.100T>C	p.C34R	
Exon 2	Frameshift	c.103delG	p.A35Rfs*12	
Exon 2	Frameshift	c.116delC	p.P39Rfs*8	
Exon 2	Nonsense	c.120T>G	p.Y40*	
Exon 2	Nonsense	c.124C>T	p.Q42*	
Exon 2	Missense	c.125A>G	p.Q42R	
Exon 2	Missense	c.140G>A	p.G47D	
Exon 2	Frameshift	c.156_157delTC	p.H53*	156-157delTC
Exon 2	Frameshift	c.168delG	p.T57Qfs*21	c.166delG
Exon 2	Missense	c.178T>C	p.C60R	
Exon 2	Missense	c.178T>G	p.C60G	
Exon 2	Missense	c.179G>A	p.C60Y	
Exon 2	Frameshift	c.186_187insTACC	p.G63Yfs*3	c.186insTACC
Exon 2	Frameshift	c.189_207delinsGGAGCATAATCAAA	p.S64Efs*32	c.189_207delins14
Exon 2	Deletion	c.189_209delTAGCACCTGCTATGGCCTTTG	p.S64_W70del	c.188-208delGTAGCACCTGCTATGGCCTTT c.188-208del21 c.189-209del21
Exon 2	Missense	c.196T>C	p.C66R	
Exon 2	Missense	c.196T>G	p.C66G	
Exon 2	Missense	c.197G>A	p.C66Y	
Exon 2	Missense	c.200A>G	p.Y67C	
Exon 2	Frameshift	c.200dupA	p.Y67*	c.201insA
Exon 2	Nonsense	c.201T>G	p.Y67*	
Exon 2	Missense	c.203G>A	p.G68D	
Exon 2	Nonsense	c.218C>G	p.S73*	
Exon 2	Frameshift	c.236_238delinsAAAAGGGGACA	p.L79Qfs*5	
Exon 2	Frameshift	c.237delT	p.V80*	
Exon 2	Frameshift	c.240_241insT	p.K81*	c.241insT
Exon 2	Nonsense	c.244C>T	p.Q82*	
Exon 2	Missense	c.246A>C	p.Q82H	
Exon 2	Frameshift	c.246dupA	p.G83Rfs*15	
Exon 2	Missense	c.247G>A	p.G83R	
Intron 2	Splice-site	c.247+1G>A	p.?	

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Intron 2	Splice-site	c.247+1_247+7delGCAAGTG	p.?	c.247+1delCAAGTG
Intron 2	Splice-site	c.247+2delC	p.G47_Q82del, G63_Q82del	IVS2 247+2delC
Intron 2	Splice-site	c.247+6T>G	p.G47_Q82del, G63_Q82del	IVS2 247+6T>G
Intron 2 to exon 3	Deletion	c.248-592_413delinsGTAAAGTA	p.?	756-bp deletion
Intron 2	Splice-site	c.248-5_248delinsAC	p.?	c.248-5delTATAGGinsAC c.-5-248delTATAGGinsAC
Intron 2	Splice-site	c.248-3T>G	p.?	
Intron 2	Splice-site	c.248-2A>G	p.?	
Intron 2	Splice-site	c.248-1G>A	p.?	c.2481-1G>A
Exon 3	Deletion	c.248-?_418+?del	p.?	IVS2_IVS3 del c.?_248-c.418_?del c.247-?_420+?del
Exon 3	Duplication	c.248-?_418+?dup	p.?	
Exon 3	Missense	c.248G>A	p.G83E	
Exon 3	Missense	c.250T>C	p.C84R	
Exon 3	Missense	c.250T>G	p.C84G	
Exon 3	Missense	c.251G>T	p.C84F	
Exon 3	Nonsense	c.255G>A	p.W85*	
Exon 3	Frameshift	c.260dupA	p.H87Qfs*11	c.261insA
Exon 3	Missense	c.266G>C	p.G89A	
Exon 3	Nonsense	c.274C>T	p.Q92*	
Exon 3	Missense	c.276A>C	p.Q92H	
Exon 3	Frameshift	c.277dupG	p.E93Gfs*5	277insG
Exon 3	Missense	c.280T>C	p.C94R	
Exon 3	Missense	c.280T>G	p.C94G	
Exon 3	Missense	c.292G>A	p.E98K	
Exon 3	Nonsense	c.292G>T	p.E98*	
Exon 3	Missense	c.295T>C	p.C99R	
Exon 3	Missense	c.296G>A	p.C99Y	
Exon 3	Missense	c.296G>T	p.C99F	
Exon 3	Missense	c.304A>G	p.T102A	
Exon 3	Missense	c.319T>C	p.S107P	
Exon 3	Nonsense	c.320C>G	p.S107*	
Exon 3	Missense	c.338A>G	p.Y113C	
Exon 3	Frameshift	c.338dupA	p.Y113*	c.399insA
Exon 3	Nonsense	c.339C>A	p.Y113*	
Exon 3	Nonsense	c.339C>G	p.Y113*	
Exon 3	Frameshift	c.339_340insAA	p.R114Nfs*39	

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exon 3	Frameshift	c.345_346delCT	p.F115Lfs*4	
Exon 3	Missense	c.349T>C	p.C117R	
Exon 3	Missense	c.350G>A	p.C117Y	
Exon 3	Missense	c.350G>C	p.C117S	
Exon 3	Missense	c.353G>A	p.C118Y	c.353C>T
Exon 3	Frameshift	c.353delG	p.C118Lfs*34	
Exon 3	Missense	c.354T>G	p.C118W	
Exon 3	Frameshift	c.354_355delinsAG	p.C118*	c.354-355TA>AG
Exon 3	Frameshift	c.355delA	p.S119Afs*33	
Exon 3	Frameshift	c.359_360delCA	p.T120Rfs*4	
Exon 3	Missense	c.367T>A	p.C123S	
Exon 3	Missense	c.367T>C	p.C123R	
Exon 3	Missense	c.370A>G	p.N124D	
Exon 3	Frameshift	c.371dupA	p.N124Kfs*6	c.371dup
Exon 3	Missense	c.377A>G	p.N126S	
Exon 3	Frameshift	c.399delT	p.P134Lfs*18	
Exon 3	Frameshift	c.407_408delCA	p.T136Nfs*10	c.407_408del
Exon 3	Frameshift	c.408_412delAACAC	p.P138Qfs*7	
Intron 3	Splice-site	c.418+1G>C	p.?	
Intron 3	Splice-site	c.418+2_418+4delinsGAG	p.?	c.418+2_418+4TAA>GAG
Intron 3	Splice-site	c.418+3A>T	p.?	
Intron 3	Splice-site	c.418+5G>A	p.?	
Intron 3	Splice-site	c.418+5_418+8delGTAA	p.C84_S140del	c.247+1_+4delGTAA
Intron 3	Splice-site	c.419-10T>C	p.?	
Exon 4	Deletion	c.419-?_529+?del	p.?	
Exons 4-5	Deletion	c.419-?_621+?del	p.?	IVS3_IVS5 del del exon 4-5
Exons 4-7	Deletion	c.419-?_967+?del	p.?	
Exons 4-8	Duplication	c.419-?_1128+?dup	p.?	
Exons 4-10	Deletion	c.419-?_1413+?del	p.?	
Exons 4-13	Deletion	c.419-?_3117+?del	p.?	Del c.419?-c.3017?
Exon 4	Frameshift	c.420_421insG	p.P141Afs*6	
Exon 4	Frameshift	c.435delT	p.F145Lfs*7	
Exon 4	Nonsense	c.439C>T	p.R147*	
Exon 4	Frameshift	c.449dupC	p.I151Nfs*30	c.449dup
Exon 4	Missense	c.461C>G	p.A154G	
Exon 4	Nonsense	c.482T>A	p.L161*	
Exon 4	Frameshift	c.498delT	p.A167Pfs*9	c.497delT

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exon 4	Frameshift	c.503dupT	p.L168Ffs*13	504insT c.504_505insT
Exon 4	Nonsense	c.507C>A	p.C169*	
Exon 4	Frameshift	c.507_510delinsAAA	p.C169*	c.507-510delCTTTinsAAA c.507_510delCTTTinsAAA
Exon 4	Nonsense	c.516C>G	p.Y172*	
Exon 4	Frameshift	c.528delA	p.G177Efs*10	
Intron 4	Splice-site	c.529+2T>C	p.?	
Exon 5	Deletion	c.530-?_621+?del	p.?	
Exons 5-7	Deletion	c.530-?_967+?del	p.?	del exon 5/6/7
Exon 5	Nonsense	c.541C>T	p.Q181*	
Exon 5	Missense	c.545G>A	p.G182D	
Exon 5	Frameshift	c.551_573delACAGTATGAACATGATGGAGGCA	p.H184Rfs*8	c.551_573del
Exon 5	Missense	c.556A>G	p.M186V	
Exon 5	Nonsense	c.583G>T	p.E195*	
Exon 5	Missense	c.604A>T	p.N202Y	
Exon 5	Frameshift	c.608_609delTG	p.L203Qfs*16	c.608-609delTG
Exon 5	Frameshift	c.612delA	p.K204Nfs*5	
Exon 6	Deletion	c.622-?_852+?del	p.?	
Exons 6-7	Deletion	c.622-?_967+?del	p.?	
Exon 6	Nonsense	c.631C>T	p.R211*	c.631G>A c.633C>T
Exon 6	Nonsense	c.637C>T	p.R213*	
Exon 6	Nonsense	c.642T>G	p.Y214*	
Exon 6	Frameshift	c.659dupG	p.S221Lfs*4	c.660insG
Exon 6	Frameshift	c.664_665delinsAAGG	p.L222Kfs*9	c.664_665delTTinsAAGG
Exon 6	Frameshift	c.673_679delCGTCCAG	p.R225Lfs*3	
Exon 6	Frameshift	c.689_690delAA	p.K230Sfs*25	c.689-690del c.689_690del c.689-690delAA
Exon 6	Missense	c.690A>T	p.K230N	
Exon 6	Frameshift	c.690_691delinsT	p.K230Nfs*22	c.690_691delAGinsT
Exon 6	Missense	c.727G>A	p.E243K	
Exon 6	Missense	c.727G>C	p.E243Q	
Exon 6	Nonsense	c.727G>T	p.E243*	
Exon 6	Frameshift	c.775delC	p.R259Afs*3	
Exon 6	Frameshift	c.782_783delTA	p.I261Sfs*4	c.782_783del
Exon 6	Frameshift	c.786dupT	p.G263Wfs*3	787insT
Exon 6	Frameshift	c.790delG	p.D264Mfs*15	

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exon 6	Missense	c.794A>G	p.E265G	
Exon 6	Frameshift	c.795_796delinsTT	p.E265_L1038delinsD	
Exon 6	Frameshift	c.796_799delAGAG	p.R266Sfs*12	796-799delAGAG
Exon 6	Missense	c.797G>C	p.R266T	
Exon 6	Frameshift	c.802dupA	p.T268Nfs*30	c.802-803insA
Exon 6	Frameshift	c.804delT	p.A269Qfs*10	
Exon 6	Missense	c.818T>G	p.M273R	
Exon 6	Missense	c.830T>C	p.L277P	
Intron 6	Splice-site	c.852+1G>A	p.?	
Intron 6	Splice-site	c.852+1G>C	p.?	
Intron 6	Splice-site	c.853-2A>G	p.?	
Intron 6	Splice-site	c.853-1G>A	p.?	
Intron 6	Splice-site	c.853-1G>C	p.?	
Exon 7	Duplication	c.853-?_967+?dup	p.?	
Exon 7	Frameshift	c.855delA	p.S286Lfs*6	
Exon 7	Nonsense	c.860T>A	p.L287*	
Exon 7	Nonsense	c.872T>G	p.L291*	
Exon 7	Frameshift	c.872delT	p.L291*	
Exon 7	Nonsense	c.893G>A	p.W298*	
Exon 7	Frameshift	c.894_895dupGG	p.V299Gfs*2	
Exon 7	Missense	c.901T>C	p.S301P	
Exon 7	Missense	c.908G>A	p.R303H	
Exon 7	Nonsense	c.928A>T	p.R310*	
Exon 7	Missense	c.932G>A	p.G311E	
Exon 7	Missense	c.937G>C	p.A313P	
Exon 7	Missense	c.954A>C	p.E318D	
Exon 7	Nonsense	c.961C>T	p.R321*	
Exon 7	Frameshift	c.961delC	p.R321Efs*14	
Intron 7	Splice-site	c.967+2T>C	p.?	
Intron 7	Splice-site	c.967+4delA	p.G285Ifs*12	IVS7 958+3delT c.968+3delA
Intron 7	Splice-site	c.967+5G>C	p.?	
Intron 7	Splice-site	c.967+5G>T	p.?	
Intron 7	Splice-site	c.968-5A>G	p.?	
Intron 7	Splice-site	c.968-3C>G	p.?	
Intron 7	Splice-site	c.968-2A>C	p.?	c.965-2A>C
Intron 7	Splice-site	c.968-1G>T	p.?	
Exon 8	Deletion	c.968-?_1128+?del	p.?	c.968-?_1129+?del

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exons 8-9	Deletion	c.968-?_1276+?del	p.?	c.967-?_1275+?del
Exons 8-10	Duplication	c.968-?_1413+?dup	p.?	c.968-?_c.1413+?dup
Exon 8	Frameshift	c.969dupT	p.H324Sfs*3	c.967_968insA c.968_969insT
Exon 8	Frameshift	c.980delC	p.P327Lfs*8	
Exon 8	Missense	c.992A>G	p.H331R	
Exon 8	Nonsense	c.994C>T	p.R332*	
Exon 8	Nonsense	c.1001T>G	p.L334*	
Exon 8	Frameshift	c.1011_1015delAAATG	p.R337Sfs*6	
Exon 8	Missense	c.1016T>A	p.V339D	
Exon 8	Missense	c.1019T>C	p.L340P	
Exon 8	Missense	c.1039T>C	p.C347R	
Exon 8	Missense	c.1040G>A	p.C347Y	1042G>A
Exon 8	Missense	c.1042G>A	p.V348I	
Exon 8	Frameshift	c.1044delT	p.I349Lfs*8	
Exon 8	Frameshift	c.1060delC	p.L354Cfs*3	
Exon 8	Missense	c.1066A>T	p.M356L	
Exon 8	Frameshift	c.1076delC	p.T359Mfs*16	
Exon 8	Frameshift	c.1093_1098delinsG	p.R365Gfs*5	
Exon 8	Frameshift	c.1097delC	p.P366Qfs*9	c.1095delC
Exon 8	Frameshift	c.1101_1105delGGAGG	p.E368Rfs*2	c.1099_1103del c.1099-1103delGGGGA c.1099_1103delGGGGA
Exon 8	Frameshift	c.1113dupT	p.A372Cfs*27	c.1113delT
Exon 8	Missense	c.1117G>C	p.A373P	
Exon 8	Frameshift	c.1120delA	p.I374*	
Exon 8	Nonsense	c.1126G>T	p.E376*	
Intron 8	Splice-site	c.1128+1G>A	p.?	
Intron 8	Splice-site	c.1128+1G>T	p.?	IVS8 +1G>T IVS8 1128+1G>T
Intron 8	Splice-site	c.1129-3C>G	p.?	IVS8 1129-3C>G
Exon 9	Frameshift	c.1129-1_1129dupGG	p.V377Gfs*13	
Exon 9	Frameshift	c.1141dupA	p.R381Kfs*18	c.1141_1142insA
Exon 9	Nonsense	c.1146T>G	p.Y382*	
Exon 9	Missense	c.1151C>T	p.A384V	
Exon 9	Missense	c.1156G>A	p.E386K	
Exon 9	Missense	c.1156G>C	p.E386Q	
Exon 9	Missense	c.1157A>C	p.E386A	

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exon 9	Missense	c.1157A>G	p.E386G	
Exon 9	Missense	c.1157A>T	p.E386V	
Exon 9	Missense	c.1171G>A	p.A391T	
Exon 9	Missense	c.1175T>C	p.V392A	
Exon 9	Frameshift	c.1191_1192delTG	p.C397*	1191/1192delTG c.1189-1190delTG
Exon 9	Nonsense	c.1196C>G	p.S399*	
Exon 9	Missense	c.1202T>C	p.L401S	
Exon 9	Nonsense	c.1207C>T	p.Q403*	C1207T
Exon 9	Frameshift	c.1214delA	p.D405Afs*7	
Exon 9	Missense	c.1220A>C	p.Y407S	
Exon 9	Nonsense	c.1221T>G	p.Y407*	
Exon 9	Missense	c.1228G>A	p.G410R	
Exon 9	Nonsense	c.1241G>A	p.W414*	
Exon 9	Nonsense	c.1243G>T	p.E415*	
Exon 9	Frameshift	c.1245_1246dupGA	p.I416Rfs*4	c.1246dupGA 1247/8insGA
Exon 9	Frameshift	c.1246_1247insG	p.I416Sfs*32	c.1246dupG
Exon 9	Frameshift	c.1247_1250delinsGA	p.I416Rfs*31	c.1246_1247dupGA; 1250_1253delTTAT
Exon 9	Frameshift	c.1248delA	p.F417Lfs*2	
Exon 9	Frameshift	c.1250_1253delTTAT	p.F417*	c.1248-1251delATTT
Exon 9	Missense	c.1257A>T	p.R419S	
Exon 9	Missense	c.1258T>C	p.C420R	
Exon 9	Missense	c.1259G>A	p.C420Y	
Exon 9	Frameshift	c.1268dupT	p.F424Lfs*24	
Exon 9	Frameshift	c.1271_1276delinsCGGAGA	p.F424_G426delinsSER	c.1271delTCCCAGinsCGGAGA
Exon 9	Frameshift	c.1274dupC	p.G426Rfs*22	c.1272insC c.1274dup
Exon 9	Missense	c.1276G>C	p.G426R	
Intron 9	Splice-site	c.1276+1G>A	p.?	
Intron 9	Splice-site	c.1276+3A>G	p.?	
Intron 9	Splice-site	c.1276+3A>T	p.?	
Intron 9	Splice-site	c.1276+4A>G	p.?	
Introns 9-10	Deletion	c.1277-289_1413+4737del	p.?	c.1277-291_1413+4735del
Intron 9	Splice-site	c.1277-9A>C	p.?	
Intron 9	Splice-site	c.1277-9A>G	p.?	
Intron 9	Splice-site	c.1277-8A>G	p.?	
Exon 10	Deletion	c.1277-?_1413+?del	p.?	c.1277-?_c.1413+?del

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exon 10	Duplication	c.1277-?_1413+?dup	p.?	IVS9_IVS10 dup c.1277-?.1413+?dup c.1277-?_c.1413+?dup
Exon 10	Frameshift	c.1279delG	p.E427Nfs*47	
Exon 10	Frameshift	c.1285_1286insGGATT	p.V429Gfs*47	
Exon 10	Frameshift	c.1293_1300delGTACCAGA	p.E431Dfs*14	
Exon 10	Nonsense	c.1296C>G	p.Y432*	
Exon 10	Nonsense	c.1297C>T	p.Q433*	
Exon 10	Frameshift	c.1313_1316delCAGA	p.T438Rfs*35	c.1313-1316delCAGA
Exon 10	Missense	c.1346T>G	p.M449R	
Exon 10	Nonsense	c.1348C>T	p.Q450*	
Exon 10	Frameshift	c.1366delinsCA	p.E456Qfs*15	
Exon 10	Frameshift	c.1371delA	p.K457Nfs*17	
Exon 10	Frameshift	c.1371dupA	p.Q458Tfs*13	c.1371dup
Exon 10	Frameshift	c.1376_1377delGA	p.R459Tfs*11	c.1375_1376delAG
Exon 10	Frameshift	c.1387_1388insA	p.P463Hfs*8	c.1388insA
Exon 10	Frameshift	c.1389dupA	p.E464Rfs*7	c.1388-1389insA c.1388_1389insA
Exon 10	Frameshift	c.1392delA	p.A465Pfs*9	
Exon 10	Nonsense	c.1397G>A	p.W466*	
Exon 10	Nonsense	c.1398G>A	p.W466*	
Exon 10	Frameshift	c.1401delA	p.E468Kfs*6	c.1399delA c.1401del
Exon 10	Nonsense	c.1402G>T	p.E468*	
Intron 10	Splice-site	c.1413+1G>A	p.?	
Intron 10	Splice-site	c.1413+3A>T	p.?	IVS10 +3A>T
Intron 10	Splice-site	c.1414-2A>T	p.?	
Exons 11-12	Deletion	c.1414-?_2866+?del	p.?	Del c.1414-?_2866+?
Exons 11-13	Deletion	c.1414-?_3117+?del	p.?	c.1414-?_3114+?del c.1414-?_c.3117+?del
Exon 11	Nonsense	c.1424C>A	p.S475*	
Exon 11	Frameshift	c.1426_1450delCTCAAGGAGACAATCGAAGACTGTT	p.L476Gfs*22	c.1426_1450del
Exon 11	Frameshift	c.1427delT	p.L476Pfs*30	c.1426delT
Exon 11	Nonsense	c.1441G>T	p.E481*	
Exon 11	Missense	c.1447T>C	p.C483R	
Exon 11	Nonsense	c.1451G>A	p.W484*	
Exon 11	Missense	c.1453G>A	p.D485N	
Exon 11	Missense	c.1454A>G	p.D485G	
Exon 11	Nonsense	c.1456C>T	p.Q486*	

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exon 11	Missense	c.1460A>T	p.D487V	
Exon 11	Missense	c.1469C>T	p.A490V	
Exon 11	Missense	c.1471C>T	p.R491W	
Exon 11	Missense	c.1472G>A	p.R491Q	
Exon 11	Frameshift	c.1477dupA	p.T493Nfs*6	
Exon 11	Nonsense	c.1483C>T	p.Q495*	
Exon 11	Missense	c.1486T>C	p.C496R	
Exon 11	Missense	c.1487G>A	p.C496Y	
Exon 11	Missense	c.1509A>C	p.E503D	
Exon 11	Missense	c.1516A>G	p.M506V	
Exon 11	Nonsense	c.1523G>A	p.W508*	
Exon 11	Nonsense	c.1525G>T	p.E509*	
Exon 11	Missense	c.1535A>C	p.K512T	
Exon 11	Missense	c.1557T>A	p.N519K	
Exon 11	Frameshift	c.1585delC	p.R529Afs*35	
Intron 11	Splice-site	c.1587-7_1587-4delCTTT	p.?	
Exon 12	Missense	c.1598A>G	p.H533R	
Exon 12	Nonsense	c.1629T>G	p.Y543*	
Exon 12	Missense	c.1687G>A	p.V563M	
Exon 12	Nonsense	c.1750C>T	p.R584*	
Exon 12	Missense	c.1766A>G	p.Y589C	
Exon 12	Nonsense	c.1771C>T	p.R591*	
Exon 12	Nonsense	c.1789C>T	p.R597*	
Exon 12	Frameshift	c.1954_1955dupAC	p.V654Lfs*6	c.1954_1955dup c.1954dupA c.1956insAC c.1956_1957insAC
Exon 12	Frameshift	c.1968dupA	p.Q657Tfs*18	c.1969insA c.1969_1970insA
Exon 12	Nonsense	c.1969C>T	p.Q657*	
Exon 12	Nonsense	c.1978G>T	p.E660*	
Exon 12	Nonsense	c.1981G>T	p.E661*	
Exon 12	Frameshift	c.2009delC	p.P670Qfs*30	
Exon 12	Nonsense	c.2124C>G	p.Y708*	
Exon 12	Frameshift	c.2128delC	p.L710Sfs*2	
Exon 12	Frameshift	c.2286delC	p.N764Ifs*8	c.2289delC
Exon 12	Frameshift	c.2291dupA	p.N764Kfs*49	2292insA
Exon 12	Missense	c.2296A>G	p.T766A	

Location	Mutation category	Nucleotide change[†]	Amino acid change	Alternative published nomenclature
Exon 12	Frameshift	c.2297delC	p.T766Kfs*6	delC2705
Exon 12	Frameshift	c.2303_2309delAGCCCCG	p.E768Gfs*2	
Exon 12	Frameshift	c.2308delC	p.R770Gfs*2	c.2305delC
Exon 12	Frameshift	c.2386delG	p.A796Qfs*7	
Exon 12	Frameshift	c.2407_2408insTG	p.T803Mfs*5	2408insTG c.2408_2409insTG
Exon 12	Frameshift	c.2410_2413delGTCA	p.V804Pfs*2	c.2410-2413delGTCA
Exon 12	Frameshift	c.2413dupA	p.T805Nfs*8	
Exon 12	Frameshift	c.2442_2443delCA	p.H814Qfs*3	c.2441-2442delAC
Exon 12	Frameshift	c.2446_2447dupGT	p.N817Lfs*23	c.2446_2447dup
Exon 12	Frameshift	c.2484delG	p.T829Qfs*10	
Exon 12	Frameshift	c.2503dupA	p.T835Nfs*8	c.2504insA
Exon 12	Frameshift	c.2503_2506delACAA	p.T835Pfs*3	c.2500delCAAA c.2503_2506del
Exon 12	Frameshift	c.2504delC	p.T835Kfs*4	
Exon 12	Frameshift	c.2506_2522delACCAACATAGTGACACA	p.T836*	c.2506_2522del17
Exon 12	Frameshift	c.2521_2522dupCA	p.R842Ifs*18	
Exon 12	Frameshift	c.2527delG	p.A843Pfs*16	
Exon 12	Nonsense	c.2533G>T	p.E845*	
Exon 12	Frameshift	c.2580delT	p.N861Ifs*11	c.2579delT 2579-2580 delT
Exon 12	Missense	c.2588G>A	p.S863N	
Exon 12	Frameshift	c.2609delT	p.L870Yfs*2	
Exon 12	Nonsense	c.2617C>T	p.R873*	
Exon 12	Missense	c.2618G>A	p.R873Q	
Exon 12	Nonsense	c.2620G>T	p.E874*	
Exon 12	Nonsense	c.2626C>T	p.Q876*	
Exon 12	Frameshift	c.2668delA	p.R890Gfs*6	
Exon 12	Nonsense	c.2695C>T	p.R899*	
Exon 12	Missense	c.2696G>C	p.R899P	
Exon 12	Missense	c.2708A>G	p.N903S	
Exon 12	Nonsense	c.2730T>A	p.C910*	
Exon 12	Nonsense	c.2737C>T	p.Q913*	
Exon 12	Nonsense	c.2752C>T	p.Q918*	
Exon 12	Nonsense	c.2789C>G	p.S930*	
Exon 13	Missense	c.2945A>G	p.K982R	

[†]GenBank reference sequence and version number for *BMPR2*: NM_001204.6; numbering is calculated from +1 as A of the ATG initiation codon. All nomenclature has been updated according to the most recent HGVS guidelines: http://www.hgvs.org/mutnomen.

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