

GENE	HGMD ACCESSION	HGMD codon CHANGE	HGMD amino CHANGE	HGVS (nucleotide)	HGVS (protein)	Phenotype	Reference	Source
ASPA	CM100918	ATG-ACG	Met1Thr	2T>C	M1T	Canavan disease	Zhang (2010) <i>Brain Dev</i> 32:879	PubMed 20129749
ASPA	CM063852	GTT-GGT	Val14Gly	41T>G	V14G	Canavan disease	Zeng (2006) <i>Mol Genet Metab</i> 89:156	PubMed 16854607
ASPA	CM960084	ATC-ACC	Ile16Thr	47T>C	I16T	Canavan disease	Kaul (1996) <i>Am J Hum Genet</i> 59:95	PubMed 8659549
ASPA	CM067343	GGA-AGA	Gly18Arg	52G>A	G18R	Canavan disease	Zeng (2006) <i>Adv Exp Med Biol</i> 576:165	PubMed 16802711
ASPA	CM001608	CAT-CCT	His21Pro	62A>C	H21P	Canavan disease	Sistermans (2000) <i>Eur J Hum Genet</i> 8:557	PubMed 10909858
ASPA	CM023602	GAG-GGG	Glu24Gly	71A>G	E24G	Canavan disease	Zeng (2002) <i>J Inherit Metab Dis</i> 25:557	PubMed 12638939
ASPA	CM960085	GGA-AGA	Gly27Arg	79G>A	G27R	Canavan disease	Kaul (1996) <i>Am J Hum Genet</i> 59:95	PubMed 8659549
ASPA	CM127193	AAC-AAA	Asn54Lys	162C>A	N54K	Canavan disease	Howell (2004) <i>J Inherit Metab Dis</i> 27:289	PubMed 15243987
ASPA	CM001609	GCA-ACA	Ala57Thr	169G>A	A57T	Canavan disease	Sistermans (2000) <i>Eur J Hum Genet</i> 8:557	PubMed 10909858
ASPA	CM023603	GAC-GCC	Asp68Ala	203A>C	D68A	Canavan disease	Zeng (2002) <i>J Inherit Metab Dis</i> 25:557	PubMed 12638939
ASPA	CM100919	AAT-AGT	Asn70Ser	209A>G	N70S	Canavan disease	Zhang (2010) <i>Brain Dev</i> 32:879	PubMed 20129749
ASPA	CM060201	CGC-CAC	Arg71His	212G>A	R71H	Canavan disease	Janson (2006) <i>Ann Neurol</i> 59:428	PubMed 16437572
ASPA	CM126484	TAT-TAA	Tyr88Term	264T>A	Y88*	Canavan disease	Durmaz (2012) <i>Fetal Pediatr Pathol</i> 31:236	PubMed 22468686
ASPA	CM990192	TAT-TAG	Tyr109Term	327T>G	Y109*	Canavan disease	Elpeleg (1999) <i>J Inherit Metab Dis</i> 22:531	PubMed 10407784
ASPA	CM960086	GAC-GAA	Asp114Glu	342C>A	D114E	Canavan disease	Kaul (1996) <i>Am J Hum Genet</i> 59:95	PubMed 8659549
ASPA	CM023014	GAC-TAC	Asp114Tyr	340G>T	D114Y	Canavan disease	Olsen (2002) <i>J Med Genet</i> 39:e55	PubMed 12205125
ASPA	CM063846	AAC-ATC	Asn121Ile	362A>T	N121I	Canavan disease	Zeng (2006) <i>Mol Genet Metab</i> 89:156	PubMed 16854607
ASPA	CM960087	GGG-GAG	Gly123Glu	368G>A	G123E	Canavan disease	Kaul (1996) <i>Am J Hum Genet</i> 59:95	PubMed 8659549
ASPA	CM063849	ATT-TTT	Ile143Phe	427A>T	I143F	Canavan disease	Zeng (2006) <i>Mol Genet Metab</i> 89:156	PubMed 16854607
ASPA	CM980125	ATT-ACT	Ile143Thr	428T>C	I143T	Canavan disease	Kobayashi (1998) <i>Hum Mutat</i> S1:S308	PubMed 9452117
ASPA	CM950102	TGC-CGC	Cys152Arg	454T>C	C152R	Canavan disease	Kaul (1995) <i>Hum Mutat</i> 5:269	PubMed 7599639
ASPA	CM023604	TGC-TGG	Cys152Trp	456C>G	C152W	Canavan disease	Zeng (2002) <i>J Inherit Metab Dis</i> 25:557	PubMed 12638939
ASPA	CM960088	TGC-TAC	Cys152Tyr	455G>A	C152Y	Canavan disease	Kaul (1996) <i>Am J Hum Genet</i> 59:95	PubMed 8659549
ASPA	CM063847	ACC-ATC	Thr166Ile	497C>T	T166I	Canavan disease	Zeng (2006) <i>Mol Genet Metab</i> 89:156	PubMed 16854607
ASPA	CM960089	CGT-TGT	Arg168Cys	502C>T	R168C	Canavan disease	Kaul (1996) <i>Am J Hum Genet</i> 59:95	PubMed 8659549
ASPA	CM001610	CGT-CAT	Arg168His	503G>A	R168H	Canavan disease	Sistermans (2000) <i>Eur J Hum Genet</i> 8:557	PubMed 10909858
ASPA	CM138792	ATA-ACA	Ile177Thr	530T>C	I177T	Canavan disease	Di Pietro (2013) <i>Clin Biochem</i> 46:1902	PubMed 24036223
ASPA	CM063850	CCT-CTT	Pro181Leu	542C>T	P181L	Canavan disease	Zeng (2006) <i>Mol Genet Metab</i> 89:156	PubMed 16854607
ASPA	CM001611	CCT-ACT	Pro181Thr	541C>A	P181T	Canavan disease	Sistermans (2000) <i>Eur J Hum Genet</i> 8:557	PubMed 10909858
ASPA	CM990193	CCT-CAT	Pro183His	548C>A	P183H	Canavan disease	Elpeleg (1999) <i>J Inherit Metab Dis</i> 22:531	PubMed 10407784
ASPA	CM023605	CAA-TAA	Gln184Term	550C>T	Q184*	Canavan disease	Zeng (2002) <i>J Inherit Metab Dis</i> 25:557	PubMed 12638939
ASPA	CM990194	GTT-TTT	Val186Phe	556G>T	V186F	Canavan disease	Elpeleg (1999) <i>J Inherit Metab Dis</i> 22:531	PubMed 10407784
ASPA	CM990195	ATG-AGG	Met195Arg	584T>G	M195R	Canavan disease	Elpeleg (1999) <i>J Inherit Metab Dis</i> 22:531	PubMed 10407784
ASPA	CM055097	AAA-GAA	Lys213Glu	637A>G	K213E	Canavan disease	Tacke (2005) <i>Neuropediatrics</i> 36:252	PubMed 16138249
ASPA	CM023606	GAA-TAA	Glu214Term	640G>T	E214*	Canavan disease	Zeng (2002) <i>J Inherit Metab Dis</i> 25:557	PubMed 12638939
ASPA	CM950103	TGC-TGA	Cys218Term	654C>A	C218*	Canavan disease	Shaag (1995) <i>Am J Hum Genet</i> 57:572	PubMed 7668285
ASPA	CM086530	ATA-ACA	Ile226Thr	677T>C	I226T	Canavan disease	Di Pietro (2008) <i>Clin Biochem</i> 41:611	PubMed 18280251
ASPA	CM994594	TAC-TGC	Tyr231Cys	692A>G	Y231C	Canavan disease	Rady (1999) <i>Am J Med Genet</i> 87:273	PubMed 10564886
ASPA	CM940123	TAC-TAA	Tyr231Term	693C>A	Y231*	Canavan disease	Kaul (1994) <i>Am J Hum Genet</i> 55:34	PubMed 8023850
ASPA	CM127191	ATC-AGC	Ile243Ser	728T>G	I243S	Canavan disease	Bijarnia (2013) <i>Indian J Pediatr</i> 80:26	PubMed 22878930
ASPA	CM023607	CAT-CGT	His244Arg	731A>G	H244R	Canavan disease	Zeng (2002) <i>J Inherit Metab Dis</i> 25:557	PubMed 12638939

ASPA	CM063848	CAT-CTT	His244Leu	731A>T	H244L	Canavan disease	Zeng (2006) <i>Mol Genet Metab</i> 89:156	PubMed 16854607
ASPA	CM023015	GAT-GTT	Asp249Val	746A>T	D249V	Canavan disease	Olsen (2002) <i>J Med Genet</i> 39:e55	PubMed 12205125
ASPA	CM127076	CCT-CGT	Pro257Arg	770C>G	P257R	Canavan disease	Zano (2013) <i>J Inherit Metab Dis</i> 36:1	PubMed 22850825
ASPA	CM063851	CTG-CCG	Leu272Pro	815T>C	L272P	Canavan disease	Zeng (2006) <i>Mol Genet Metab</i> 89:156	PubMed 16854607
ASPA	CM950104	GGA-AGA	Gly274Arg	820G>A	G274R	Canavan disease	Shaag (1995) <i>Am J Hum Genet</i> 57:572	PubMed 7668285
ASPA	CM990196	CCC-CTC	Pro280Leu	839C>T	P280L	Canavan disease	Elpeleg (1999) <i>J Inherit Metab Dis</i> 22:531	PubMed 10407784
ASPA	CM990197	CCC-TCC	Pro280Ser	838C>T	P280S	Canavan disease	Elpeleg (1999) <i>J Inherit Metab Dis</i> 22:531	PubMed 10407784
ASPA	CM930046	GAG-GCG	Glu285Ala	854A>C	E285A	Canavan disease	Kaul (1993) <i>Nat Genet</i> 5:118	PubMed 8252036
ASPA	CM990198	GCA-ACA	Ala287Thr	859G>A	A287T	Canavan disease	Elpeleg (1999) <i>J Inherit Metab Dis</i> 22:531	PubMed 10407784
ASPA	CM034717	TAT-TGT	Tyr288Cys	863A>G	Y288C	Canavan disease	Surendran (2003) <i>Mol Genet Metab</i> 80:74	PubMed 14567959
ASPA	CM950105	TTT-TCT	Phe295Ser	884T>C	F295S	Canavan disease	Shaag (1995) <i>Am J Hum Genet</i> 57:572	PubMed 7668285
ASPA	CM127192	CTA-CCA	Leu301Pro	902T>C	L301P	Canavan disease	Bijarnia (2013) <i>Indian J Pediatr</i> 80:26	PubMed 22878930
ASPA	CM940124	GCA-GAA	Ala305Glu	914C>A	A305E	Canavan disease	Kaul (1994) <i>Am J Hum Genet</i> 55:34	PubMed 8023850
ASPA	CM023608	TAG-TGG	Term314Trp	941A>G	*314W	Canavan disease	Zeng (2002) <i>J Inherit Metab Dis</i> 25:557	PubMed 12638939
BRAF	CM072907	ACC-CCC	Thr244Pro	730A>C	T244P	syndrome	Gripp (2007) <i>Am J Med Genet A</i> 143A:1472	PubMed 17551924
BRAF	CM092081	TTA-TTT	Leu245Phe	735A>T	L245F	syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	CM060871	GCA-CCA	Ala246Pro	736G>C	A246P	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404
BRAF	CM060874	CAG-CGG	Gln257Arg	770A>G	Q257R	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404
BRAF	CM071572	CAG-AAG	Gln257Lys	769C>A	Q257K	syndrome	Narumi (2007) <i>Am J Med Genet A</i> 143A:799	PubMed 17366577
BRAF	CM076029	CAA-CGA	Gln262Arg	785A>G	Q262R	syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
BRAF	CM080111	CAA-AAA	Gln262Lys	784C>A	Q262K	syndrome	Schulz (2008) <i>Clin Genet</i> 73:62	PubMed 18042262
BRAF	CM092082	GAA-AAA	Glu275Lys	823G>A	E275K	syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	HM070099	GGA-CGA	Gly464Arg	1390G>C	G464R	Cardio-facio-cutaneous syndrome	Cave (2008) <i>Hum Genet</i> 123:108	Source: Online submission
BRAF	CM086781	GGA-GTA	Gly464Val	1391G>T	G464V	Cardio-facio-cutaneous syndrome	Rodriguez-Viciana (2008) <i>Methods Enzymol</i> 438:277	PubMed 18413255
BRAF	CM115083	GGA-GAA	Gly466Glu	1397G>A	G466E	syndrome	Siegel (2011) <i>Br J Dermatol</i> 164:521	PubMed 21062266
BRAF	CM060872	TCA-GCA	Ser467Ala	1399T>G	S467A	syndrome	Rodriguez-Viciana (2006) <i>Science</i> 311:1287	PubMed 16439621
BRAF	CM060881	TTT-TCT	Phe468Ser	1403T>C	F468S	syndrome	Rodriguez-Viciana (2006) <i>Science</i> 311:1287	PubMed 16439621
BRAF	BM1457681	GGA-CGA	Gly469Arg	1405G>C	G469R	syndrome	Terry (2014) <i>Pediatr Dev Pathol</i> 17:59	PubMed 24303953
BRAF	CM060876	GGA-GAA	Gly469Glu	1406G>A	G469E	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404
BRAF	CM123144	ACA-CCA	Thr470Pro	1408A>C	T470P	syndrome	Quaio (2012) <i>Am J Med Genet A</i> 158A:1077	PubMed 22488759
BRAF	CM123146	GTC-TTC	Val471Phe	1411G>T	V471F	syndrome	Abe (2012) <i>Am J Med Genet A</i> 158A:1083	PubMed 22495831
BRAF	CM1314446	GCA-GAA	Ala481Glu	1442C>A	A481E	syndrome	Hazan (2013) <i>Iran J Pediatr</i> 23:608	PubMed 24800029
BRAF	CM060880	TTG-TTC	Leu485Phe	1455G>C	L485F	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404
BRAF	CM086782	TTG-TCG	Leu485Ser	1454T>C	L485S	Cardio-facio-cutaneous syndrome	Rodriguez-Viciana (2008) <i>Methods Enzymol</i> 438:277	PubMed 18413255
BRAF	CM071573	GTG-GGG	Val487Gly	1460T>G	V487G	syndrome	Narumi (2007) <i>Am J Med Genet A</i> 143A:799	PubMed 17366577
BRAF	CM080110	AAA-AAC	Lys499Asn	1497A>C	K499N	syndrome	Schulz (2008) <i>Clin Genet</i> 73:62	PubMed 18042262
BRAF	CM060873	AAA-GAA	Lys499Glu	1495A>G	K499E	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404
BRAF	CM060875	GAA-GGA	Glu501Gly	1502A>G	E501G	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404
BRAF	CM060879	GAA-AAA	Glu501Lys	1501G>A	E501K	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404
BRAF	CM076030	GAA-GTA	Glu501Val	1502A>T	E501V	syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
BRAF	CM072908	CTG-CCG	Leu525Pro	1574T>C	L525P	syndrome	Gripp (2007) <i>Am J Med Genet A</i> 143A:1472	PubMed 17551924

BRAF	CM062462	GGC-CGC	Gly534Arg	1600G>C	G534R	syndrome	Rauen (2006) <i>Am J Med Genet A</i> 140A:1681	PubMed 16804887
BRAF	CM072909	GAT-GAG	Asp565Glu	1695T>G	D565E	syndrome	Gripp (2007) <i>Am J Med Genet A</i> 143A:1472	PubMed 17551924
BRAF	CM071574	AAT-GAT	Asn580Asp	1738A>G	N580D	syndrome	Narumi (2007) <i>Am J Med Genet A</i> 143A:799	PubMed 17366577
BRAF	CM060878	AAT-GAT	Asn581Asp	1741A>G	N581D	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404
BRAF	CM080112	TTT-TTA	Phe595Leu	1785T>A	F595L	syndrome	Schulz (2008) <i>Clin Genet</i> 73:62	PubMed 18042262
BRAF	CM060882	TTT-TTG	Phe595Leu	1785T>G	F595L	syndrome	Rodriguez-Viciana (2006) <i>Science</i> 311:1287	PubMed 16439621
BRAF	CM060877	GGT-GTT	Gly596Val	1787G>T	G596V	syndrome Cardio-facio-cutaneous	Rodriguez-Viciana (2006) <i>Science</i> 311:1287	PubMed 16439621
BRAF	CM086780	ACA-AGA	Thr599Arg	1796C>G	T599R	syndrome	Rodriguez-Viciana (2008) <i>Methods Enzymol</i> 438:277	PubMed 18413255
BRAF	CM112509	GTG-GGG	Val600Gly	1799T>G	V600G	syndrome	Champion (2011) <i>Clin Genet</i> 79:468	PubMed 20735442
BRAF	CM092083	AAA-CAA	Lys601Gln	1801A>C	K601Q	syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	CM140993	AAA-ATA	Lys601Ile	1802A>T	K601I	syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
BRAF	CM123147	AAA-ACA	Lys601Thr	1802A>C	K601T	syndrome	Abe (2012) <i>Am J Med Genet A</i> 158A:1083	PubMed 22495831
BRAF	CM062461	GAT-GAA	Asp638Glu	1914T>A	D638E	syndrome	Rauen (2006) <i>Am J Med Genet A</i> 140A:1681	PubMed 16804887
BRAF	CM092084	GAT-GAG	Asp638Glu	1914T>G	D638E	syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	CM092085	CAA-CGA	Gln709Arg	2126A>G	Q709R	syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	CM125906	TCT-TGT	Ser36Cys	107C>G	S36C	hypertrophic	Kaski (2012) <i>Circ Cardiovasc Genet</i> 5:317	PubMed 22589294
BRAF	CM076028	ACG-CCG	Thr241Pro	721A>C	T241P	Costello syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
BRAF	CM122892	ACA-GCA	Thr599Ala	1795A>G	T599A	Langerhans cell histiocytosis	Satoh (2012) <i>PLoS One</i> 7:	PubMed 22506009
BRAF	CM092078	ACG-AGG	Thr241Arg	722C>G	T241R	Noonan syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	CM092077	ACG-ATG	Thr241Met	722C>T	T241M	Noonan syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	CM137740	AGA-GGA	Arg384Gly	1150A>G	R384G	Noonan syndrome	Croonen (2013) <i>Eur J Hum Genet</i> 21:936	PubMed 23321623
BRAF	CM1211842	TTC-TAC	Phe498Tyr	1493T>A	F498Y	Noonan syndrome	Cheon (2012) <i>Horm Res</i> 78S1:203	PubMed: No PubMed ID
BRAF	CM092079	TGG-TGC	Trp531Cys	1593G>C	W531C	Noonan syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	CM092080	CTA-GTA	Leu597Val	1789C>G	L597V	Noonan syndrome Noonan syndrome / cardio-facio-cutaneous syndrome	Sarkozy (2009) <i>Hum Mutat</i> 30:695	PubMed 19206169
BRAF	CM140992	GAT-GGT	Asp565Gly	1694A>G	D565G	facio-cutaneous syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
CBL	CM105116	CAA-CCA	Gln367Pro	1100A>C	Q367P	Noonan-like syndrome	Martinelli (2010) <i>Am J Hum Genet</i> 87:250	PubMed 20619386
CBL	CM105117	AAA-GAA	Lys382Glu	1144A>G	K382E	Noonan-like syndrome	Martinelli (2010) <i>Am J Hum Genet</i> 87:250	PubMed 20619386
CBL	CM105118	GAT-TAT	Asp390Tyr	1168G>T	D390Y	Noonan-like syndrome	Martinelli (2010) <i>Am J Hum Genet</i> 87:250	PubMed 20619386
CBL	CM142934	GAT-GTT	Asp390Val	1169A>T	D390V	Noonan-like syndrome	Becker (2014) <i>Blood</i> 123:1883	PubMed 24493670
CBL	CM105119	CGA-CAA	Arg420Gln	1259G>A	R420Q	Noonan-like syndrome	Martinelli (2010) <i>Am J Hum Genet</i> 87:250	PubMed 20619386
CBL	CM140995	GTG-ATG	Val784Met	2350G>A	V784M	Noonan-like syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
CFTR	CM074735	GCT-GGT	Ala107Gly	320C>G	A107G	Asthenospermia	Anguis (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM067757	ATG-TTG	Met152Leu	454A>T	M152L	Asthenospermia	Angius (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM010791	ATG-AGG	Met1Arg	2T>G	M1R	Asthma	Tzetzis (2001) <i>Hum Genet</i> 108:216	PubMed 11354633
CFTR	CM010792	GTT-ATT	Val11Ile	31G>A	V11I	Asthma	Tzetzis (2001) <i>Hum Genet</i> 108:216	PubMed 11354633
CFTR	CM010793	ACG-ATG	Thr388Met	1163C>T	T388M	Asthma	Tzetzis (2001) <i>Hum Genet</i> 108:216	PubMed 11354633
CFTR	CM015356	TTT-TTG	Phe693Leu	2079T>G	F693L	Asthma	Meyer (2001) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM004893	ACA-ATA	Thr1142Ile	3425C>T	T1142I	Asthma	Lazaro (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM043476	ACT-ATT	Thr1263Ile	3788C>T	T1263I	Asthma	Seia (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM0910748	TCC-TAC	Ser13Tyr	38C>A	S13Y	Azoospermia	Gallati (2009) <i>Reprod Biomed Online</i> 19:685	PubMed 20021716
CFTR	CM0910749	AGT-ATT	Ser158Ile	473G>T	S158I	Azoospermia	Gallati (2009) <i>Reprod Biomed Online</i> 19:685	PubMed 20021716

CFTR	CM067742	ACT-GCT	Thr1263Ala	3787A>G	T1263A	Azoospermia	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM129622	GTT-GGT	Val1318Gly	3953T>G	V1318G	Azoospermia	Poulou (2012) <i>J Cyst Fibros</i> 11:344	PubMed 22326559
CFTR	CM147382	AAG-AGG	Lys1351Arg	4052A>G	K1351R	Azoospermia	Sharma (2014) <i>Gene</i> 548:43	PubMed 25010724
CFTR	CM147393	AGA-ATA	Arg1358Ile	4073G>T	R1358I	Azoospermia	Sharma (2014) <i>Gene</i> 548:43	PubMed 25010724
CFTR	CM024685	GGC-GTC	Gly723Val	2168G>T	G723V	Bronchiectasis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM024689	AGC-AGG	Ser902Arg	2706C>G	S902R	Bronchiectasis	Claustres (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM003258	GAG-GGG	Glu1228Gly	3683A>G	E1228G	Bronchiectasis	Ninis (2003) <i>J Med Genet</i> 40:530	PubMed 12843327
CFTR	CM034795	CCC-ACC	Pro1372Thr	4114C>A	P1372T	Bronchiectasis	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM129623	AAG-ACG	Lys1165Thr	3494A>C	K1165T	Bronchitis	Poulou (2012) <i>J Cyst Fibros</i> 11:344	PubMed 22326559
CFTR	CM135530	CTT-CCT	Leu15Pro	44T>C	L15P	CFTR-related disorder	Trujillano (2013) <i>J Med Genet</i> 50:455	PubMed 23687349
CFTR	CM135529	ACT-AGT	Thr351Ser	1052C>G	T351S	CFTR-related disorder	Trujillano (2013) <i>J Med Genet</i> 50:455	PubMed 23687349
CFTR	CM135528	ATG-AAG	Met645Lys	1934T>A	M645K	CFTR-related disorder	Trujillano (2013) <i>J Med Genet</i> 50:455	PubMed 23687349
CFTR	CM920985	ATT-GTT	Ile556Val	1666A>G	I556V	Chronic pulmonary disease	Ngiam (2006) <i>J Cyst Fibros</i> 5:159	PubMed 16678503
CFTR	CM970260	GAA-AAA	Glu56Lys	166G>A	E56K	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM970261	GAT-AAT	Asp58Asn	172G>A	D58N	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM980331	CGA-CAA	Arg75Gln	224G>A	R75Q	deferens	Jarvi (1998) <i>Fertil Steril</i> 70:724	PubMed 9797105
CFTR	CM091992	TTT-ATT	Phe87Ile	259T>A	F87I	deferens	Sharma (2009) <i>Hum Reprod</i> 24:1229	PubMed 19181743
CFTR	CM015351	CAG-CCG	Gln98Pro	293A>C	Q98P	deferens	Schrijver (2005) <i>J Mol Diagn</i> 7:375	PubMed 16049310
CFTR	CM034779	TTA-TCA	Leu101Ser	302T>C	L101S	deferens	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM970263	CCG-CTG	Pro111Leu	332C>T	P111L	deferens	de Meeus (1997) <i>Hum Mutat</i> 11:480	PubMed 10200050
CFTR	CM074727	AAC-ATC	Asn113Ile	338A>T	N113I	deferens	Omar (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM005456	CGC-GGC	Arg117Gly	349C>G	R117G	deferens	Daudin (2000) <i>Fertil Steril</i> 74:1164	PubMed 11119745
CFTR	CM147305	TCT-CCT	Ser118Pro	352T>C	S118P	deferens	Sharma (2014) <i>Mol Hum Reprod</i> epub:epub	PubMed 24958810
CFTR	CM065998	TAT-CAT	Tyr122His	364T>C	Y122H	deferens	Radpour (2006) <i>Mol Hum Reprod</i> 12:717	PubMed 16973827
CFTR	CM147304	GGC-TGC	Gly126Cys	376G>T	G126C	deferens	Sharma (2014) <i>Mol Hum Reprod</i> epub:epub	PubMed 24958810
CFTR	CM091993	GGC-AGC	Gly126Ser	376G>A	G126S	deferens	Sharma (2009) <i>Hum Reprod</i> 24:1229	PubMed 19181743
CFTR	CM962455	CAT-CGT	His146Arg	437A>G	H146R	deferens	Bienvenu (1996) <i>CFTR LSDB entry</i> Newsletter:#68	Source: LSDB
CFTR	CM043460	CAG-AAG	Gln151Lys	451C>A	Q151K	deferens	Girodon (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM091994	TTT-TGT	Phe157Cys	470T>G	F157C	deferens	Sharma (2009) <i>Hum Reprod</i> 24:1229	PubMed 19181743
CFTR	CM994783	AGT-CGT	Ser158Arg	472A>C	S158R	deferens	Girodon (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM015353	CGT-CAT	Arg170His	509G>A	R170H	deferens	Brownsell (2001) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM024664	TTC-ATC	Phe200Ile	598T>A	F200I	deferens	Claustres (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM043462	ATC-ATG	Ile203Met	609C>G	I203M	deferens	Claustres (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067761	GCT-GTT	Ala238Val	713C>T	A238V	deferens	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM970267	ATG-AAG	Met244Lys	731T>A	M244K	deferens	de Meeus (1997) <i>Hum Mutat</i> 11:480	PubMed 10200050
CFTR	CM972940	AGA-ACA	Arg248Thr	743G>C	R248T	deferens	Scheffer (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM970269	ATG-AGG	Met265Arg	794T>G	M265R	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM147306	ATG-AGG	Met281Arg	842T>G	M281R	deferens	Sharma (2014) <i>Mol Hum Reprod</i> epub:epub	PubMed 24958810
CFTR	CM058372	AAC-AAG	Asn287Lys	861C>G	N287K	deferens	Wu (2005) <i>Hum Reprod</i> 20:2470	PubMed 15905293
CFTR	CM970271	CGG-TGG	Arg297Trp	889C>T	R297W	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM962461	TTA-GTA	Leu320Val	958T>G	L320V	deferens	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM970273	CGG-CTG	Arg334Leu	1001G>T	R334L	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM065996	ACC-GCC	Thr338Ala	1012A>G	T338A	deferens	Radpour (2006) <i>Mol Hum Reprod</i> 12:717	PubMed 16973827

CFTR	CM123027	GCT-ACT	Ala357Thr	1069G>A	A357T	deferens	Li (2012) <i>J Cyst Fibros</i> epub:epub	PubMed 22483971
CFTR	CM024673	GAA-AAA	Glu379Lys	1135G>A	E379K	deferens	Claustres (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM962463	GTA-GCA	Val392Ala	1175T>C	V392A	deferens Congenital absence of vas	Bienvenu (1996) <i>CFTR LSDB entry</i> Newsletter:#68	Source: LSDB
CFTR	CM024674	GCC-GAC	Ala399Asp	1196C>A	A399D	deferens	Bernardino (2003) <i>Genet Mol Biol</i> 26:1	PubMed: No PubMed ID
CFTR	CM123029	AGA-ATA	Arg419Ile	1256G>T	R419I	deferens	Li (2012) <i>J Cyst Fibros</i> epub:epub	PubMed 22483971
CFTR	CM970275	GAT-TAT	Asp443Tyr	1327G>T	D443Y	deferens	de Meeus (1997) <i>Hum Mutat</i> 11:480	PubMed 10200050
CFTR	CM952049	TCA-TTA	Ser466Leu	1397C>T	S466L	deferens	Costes (1995) <i>CFTR LSDB entry</i> Newsletter:#66	Source: LSDB
CFTR	CM056306	ATG-ATT	Met469Ile	1407G>T	M469I	deferens	Wu (2005) <i>Hum Reprod</i> 20:2470	PubMed 15905293
CFTR	CM034785	ATG-GTG	Met469Val	1405A>G	M469V	deferens	Lu (2013) <i>Urology</i> 82:824	PubMed 23953609
CFTR	CM972946	CAC-TAC	His484Tyr	1450C>T	H484Y	deferens	Casals (2000) <i>Hum Reprod</i> 15:1476	PubMed 10875853
CFTR	CM123030	TGT-TTT	Cys491Phe	1472G>T	C491F	deferens	Li (2012) <i>J Cyst Fibros</i> 11:316	PubMed 22483971
CFTR	CM129626	TGT-TCT	Cys491Ser	1472G>C	C491S	deferens	Poulou (2012) <i>J Cyst Fibros</i> 11:344	PubMed 22326559
CFTR	CM980335	CCT-GCT	Pro499Ala	1495C>G	P499A	deferens	Arduino (1998) <i>Clin Genet</i> 53:202	PubMed 9630075
CFTR	CM147278	ACC-AAC	Thr501Asn	1502C>A	T501N	deferens	Lu (2014) <i>Fertil Steril</i> 101:1255	PubMed 24559724
CFTR	CM147279	ATC-AAC	Ile507Asn	1520T>A	I507N	deferens	Lu (2014) <i>Fertil Steril</i> 101:1255	PubMed 24559724
CFTR	CM068683	ATC-GTC	Ile507Val	1519A>G	I507V	deferens	Radpour (2006) <i>Mol Hum Reprod</i> 12:469	PubMed 16714368
CFTR	CM980336	GAT-GGT	Asp513Gly	1538A>G	D513G	deferens	Bienvenu (1998) <i>Hum Mutat</i> 12:213	PubMed 10651488
CFTR	CM137997	GAA-AAA	Glu527Lys	1579G>A	E527K	deferens	Lu (2013) <i>Urology</i> epub:epub	PubMed 23953609
CFTR	CM065997	AAA-TAA	Lys536Term	1606A>T	K536*	deferens	Radpour (2006) <i>Mol Hum Reprod</i> 12:717	PubMed 16973827
CFTR	CM995324	CTT-CCT	Leu541Pro	1622T>C	L541P	deferens	Lissens (1999) <i>Mol Hum Reprod</i> 5:10	PubMed 10050655
CFTR	CM091995	GAA-GCA	Glu543Ala	1628A>C	E543A	deferens	Sharma (2009) <i>Hum Reprod</i> 24:1229	PubMed 19181743
CFTR	CM970277	GGT-GTT	Gly544Val	1631G>T	G544V	deferens	de Meeus (1997) <i>Hum Mutat</i> 11:480	PubMed 10200050
CFTR	CM980337	GGT-GCT	Gly551Ala	1652G>C	G551A	deferens	Jarvi (1998) <i>Fertil Steril</i> 70:724	PubMed 9797105
CFTR	CM983666	GAT-GGT	Asp565Gly	1694A>G	D565G	deferens	Kanavakis (1998) <i>Mol Hum Reprod</i> 4:333	PubMed 9620832
CFTR	CM970281	TTG-TTT	Leu568Phe	1704G>T	L568F	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM067770	CCT-TCT	Pro574Ser	1720C>T	P574S	deferens	Fresquet (2011) <i>J Mol Diagn</i> 13:520	PubMed 21708286
CFTR	CM920164	GGA-GCA	Gly576Ala	1727G>C	G576A	deferens	Anguiano (1992) <i>JAMA</i> 267:1794	PubMed 1545465
CFTR	CM024680	ACA-TCA	Thr582Ser	1744A>T	T582S	deferens	Seia (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM043467	TTT-ATT	Phe587Ile	1759T>A	F587I	deferens	Polizzi (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM123026	TGT-TTT	Cys592Phe	1775G>T	C592F	deferens	Li (2012) <i>J Cyst Fibros</i> epub:epub	PubMed 22483971
CFTR	CM994786	ACT-ATT	Thr604Ile	1811C>T	T604I	deferens	Girodon (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM077237	AGC-AGG	Ser753Arg	2259C>G	S753R	deferens	Pieri (2007) <i>Clinics (Sao Paulo)</i> 62:385	PubMed 17823699
CFTR	CM980348	AGG-ATG	Arg766Met	2297G>T	R766M	deferens	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM980347	CGA-GGA	Arg792Gly	2374C>G	R792G	deferens	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM004239	GAA-GTA	Glu804Val	2411A>T	E804V	deferens	Ravnik-Glavac (2000) <i>Pflugers Arch</i> 439:R53	PubMed 10653141
CFTR	CM980349	ATA-ATG	Ile807Met	2421A>G	I807M	deferens Congenital absence of vas	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM015358	AGA-GGA	Arg810Gly	2428A>G	R810G	deferens	Sobczynska-Tomaczewsk (2001) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM129625	GGC-GTC	Gly817Val	2450G>T	G817V	deferens	Poulou (2012) <i>J Cyst Fibros</i> 11:344	PubMed 22326559
CFTR	CM004892	CCA-CGA	Pro841Arg	2522C>G	P841R	deferens	Fresquet (2011) <i>J Mol Diagn</i> 13:520	PubMed 21708286
CFTR	CM091996	TAT-TTT	Tyr852Phe	2555A>T	Y852F	deferens	Sharma (2009) <i>Hum Reprod</i> 24:1229	PubMed 19181743
CFTR	CM044595	ATT-TTT	Ile853Phe	2557A>T	I853F	deferens	Dayangac (2004) <i>Hum Reprod</i> 19:1094	PubMed 15070876
CFTR	CM043469	GTG-TTG	Val920Leu	2758G>T	V920L	deferens	Girodon (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB

CFTR	CM034788	AGA-GGA	Arg933Gly	2797A>G	R933G	deferens	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM970289	AGA-AGT	Arg933Ser	2799A>T	R933S	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM147307	AGA-ACA	Arg933Thr	2798G>C	R933T	deferens	Sharma (2014) <i>Mol Hum Reprod</i> epub:epub	PubMed 24958810
CFTR	CM970290	GTG-GGG	Val938Gly	2813T>G	V938G	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM034789	GCA-GTA	Ala959Val	2876C>T	A959V	deferens	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM970291	GAT-GCT	Asp979Ala	2936A>C	D979A	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM920171	TTG-TTC	Leu997Phe	2991G>C	L997F	deferens	Fanen (1992) <i>Genomics</i> 13:770	PubMed 1379210
CFTR	CM972957	CCA-TCA	Pro1021Ser	3061C>T	P1021S	deferens	Casals (2000) <i>Hum Reprod</i> 15:1476	PubMed 10875853
CFTR	CM970293	TAT-TGT	Tyr1032Cys	3095A>G	Y1032C	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
						Congenital absence of vas	Zeng (2000) <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i>	
CFTR	CM003805	CTT-ATT	Leu1055Ile	3163C>A	L1055I	deferens	17:241	PubMed 10932005
CFTR	CM074736	GCC-CCC	Ala1067Pro	3199G>C	A1067P	deferens	Omar (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM034791	CAG-TAG	Gln1071Term	3211C>T	Q1071*	deferens	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM015359	GCT-CCT	Ala1081Pro	3241G>C	A1081P	deferens	Ngukam (2004) <i>J Trop Pediatr</i> 50:239	PubMed 15357566
CFTR	BM1272737	CAA-AAA	Gln1100Lys	3298C>A	Q1100K	deferens	Akin (2012) <i>Andrologia</i> epub:epub	PubMed 23240968
CFTR	CM071597	GTC-CTC	Val1108Leu	3322G>C	V1108L	deferens	Grangeia (2007) <i>Genet Med</i> 9:163	PubMed 17413420
CFTR	CM015360	GGT-GCT	Gly1130Ala	3389G>C	G1130A	deferens	Dayangac (2004) <i>Hum Reprod</i> 19:1094	PubMed 15070876
CFTR	CM086582	TGG-CGG	Trp1145Arg	3433T>C	W1145R	deferens	Alibakhshi (2008) <i>J Cyst Fibros</i> 7:102	PubMed 17662673
CFTR	CM970296	GTG-GAG	Val1153Glu	3458T>A	V1153E	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM941983	GAT-GGT	Asp1154Gly	3461A>G	D1154G	deferens	Costes (1994) <i>CFTR LSDB entry</i> Newsletter:#64	Source: LSDB
CFTR	CM091997	GAT-GAA	Asp1270Glu	3810T>A	D1270E	deferens	Sharma (2009) <i>Hum Reprod</i> 24:1229	PubMed 19181743
						Congenital absence of vas		
CFTR	CM113212	GCC-GTC	Ala1285Val	3854C>T	A1285V	deferens	Sachdeva (2011) <i>Genet Test Mol Biomarkers</i> 15:307	PubMed 21254931
CFTR	CM055122	CCA-TCA	Pro1290Ser	3868C>T	P1290S	deferens	Grangeia (2005) <i>Fertil Steril</i> 83:448	PubMed 15705389
CFTR	CM034794	TAT-TGT	Tyr1307Cys	3920A>G	Y1307C	deferens	Casals (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067747	CAG-CAT	Gln1309His	3927G>T	Q1309H	deferens	Girodon (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM973398	TTT-GTT	Phe1337Val	4009T>G	F1337V	deferens	Scheffer (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM994789	CTT-TTT	Leu1339Phe	4015C>T	L1339F	deferens	Girodon (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM970298	AAG-GAG	Lys1351Glu	4051A>G	K1351E	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM970299	GCG-GTG	Ala1364Val	4091C>T	A1364V	deferens	de Meeus (1997) <i>Hum Mutat</i> 11:480	PubMed 10200050
CFTR	CM950257	GAT-CAT	Asp1377His	4129G>C	D1377H	deferens	Costes (1995) <i>Eur J Hum Genet</i> 3:285	PubMed 8556303
CFTR	CM970300	CTA-CAA	Leu1388Gln	4163T>A	L1388Q	deferens	Doerk (1997) <i>Hum Genet</i> 100:365	PubMed 9272157
CFTR	CM067755	GAA-GGA	Glu1401Gly	4202A>G	E1401G	deferens	Girodon (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM071596	GAA-AAA	Glu1401Lys	4201G>A	E1401K	deferens	Grangeia (2007) <i>Genet Med</i> 9:163	PubMed 17413420
CFTR	CM034796	GAA-AAA	Glu1409Lys	4225G>A	E1409K	deferens	Alonso (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM931253	CAA-TAA	Gln1411Term	4231C>T	Q1411*	deferens	Costes (1993) <i>CFTR LSDB entry</i> Newsletter:#58	Source: LSDB
CFTR	CM067773	TTG-TCG	Leu1414Ser	4241T>C	L1414S	deferens	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM034797	GAA-TAA	Glu1473Term	4417G>T	E1473*	deferens	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM941965	ATG-ATA	Met1Ile	3G>A	M1I	Cystic fibrosis	Axton (1994) <i>CFTR LSDB entry</i> Newsletter:#61	Source: LSDB
CFTR	CM960274	ATG-ATT	Met1Ile	3G>T	M1I	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM930093	ATG-AAG	Met1Lys	2T>A	M1K	Cystic fibrosis	Claustres (1993) <i>Hum Mol Genet</i> 2:1209	PubMed 7691344
CFTR	CM994781	ATG-ACG	Met1Thr	2T>C	M1T	Cystic fibrosis	Bienvenu (2005) <i>Hum Biol</i> 77:705	PubMed 16596947
CFTR	CM930094	ATG-GTG	Met1Val	1A>G	M1V	Cystic fibrosis	Cheadle (1993) <i>Hum Mol Genet</i> 2:1551	PubMed 7505689
CFTR	CM940232	CAG-TAG	Gln2Term	4C>T	Q2*	Cystic fibrosis	Savov (1994) <i>Hum Mol Genet</i> 3:57	PubMed 7512860

CFTR	CM940233	AGG-TGG	Arg3Trp	7A>T	R3W	Cystic fibrosis	Savov (1994) <i>Hum Mol Genet</i> 3:57	PubMed 7512860
CFTR	CM930095	TCG-TAG	Ser4Term	11C>A	S4*	Cystic fibrosis	Glavac (1993) <i>Hum Mol Genet</i> 2:315	PubMed 7684643
CFTR	CM970256	CCT-CTT	Pro5Leu	14C>T	P5L	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM021068	CTG-GTG	Leu6Val	16C>G	L6V	Cystic fibrosis	Visich (2002) <i>Clin Genet</i> 61:207	PubMed 12000363
CFTR	CM031655	GAA-TAA	Glu7Term	19G>T	E7*	Cystic fibrosis	Wong (2003) <i>Am J Med Genet</i> 120A:296	PubMed 12833420
CFTR	CM952250	AGC-CGC	Ser10Arg	28A>C	S10R	Cystic fibrosis	Hughes (1995) <i>CFTR LSDB entry Newsletter</i> :#65	Source: LSDB
CFTR	CM962449	TCC-TTC	Ser13Phe	38C>T	S13F	Cystic fibrosis	Cao (1996) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM931138	AAA-TAA	Lys14Term	40A>T	K14*	Cystic fibrosis	Ferec (1993) <i>CFTR LSDB entry Newsletter</i> :#56	Source: LSDB
CFTR	CM067745	AGC-GGC	Ser18Gly	52A>G	S18G	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM970257	TGG-TGT	Trp19Cys	57G>T	W19C	Cystic fibrosis	Macek (1997) <i>Am J Hum Genet</i> 60:1122 Chevalier-Porst (1998) <i>CFTR LSDB entry Newsletter</i> :#70	PubMed 9150159 Source: LSDB
CFTR	CM983534	TGG-TGA	Trp19Term	57G>A	W19*	Cystic fibrosis	Newsletters:#70	Source: LSDB
CFTR	CM043456	TTG-TTC	Leu24Phe	72G>C	L24F	Cystic fibrosis	Seia (2004) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM043457	GGA-AGA	Gly27Arg	79G>A	G27R	Cystic fibrosis	Oller Ramirez (2006) <i>Mol Genet Metab</i> 87:370	PubMed 16423550
CFTR	CM057570	GGA-CGA	Gly27Arg	79G>C	G27R	Cystic fibrosis	Herova (2005) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM940234	GGA-GAA	Gly27Glu	80G>A	G27E	Cystic fibrosis	Bienvenu (1994) <i>Hum Mol Genet</i> 3:365	PubMed 7516232
CFTR	CM920138	GGA-TGA	Gly27Term	79G>T	G27*	Cystic fibrosis	Shackleton (1992) <i>Hum Mol Genet</i> 1:445	PubMed 1284531
CFTR	CM141622	GGA-GTA	Gly27Val	80G>T	G27V	Cystic fibrosis	Ziętkiewicz (2014) <i>PLoS One</i> 9:e89094	PubMed 24586523
CFTR	CM940235	CAG-TAG	Gln30Term	88C>T	Q30*	Cystic fibrosis	Chillon (1994) <i>Hum Genet</i> 93:447	PubMed 7513293
CFTR	CM950231	CGC-CTC	Arg31Leu	92G>T	R31L	Cystic fibrosis	Zielenski (1995) <i>Hum Mutat</i> 5:43	PubMed 7537150
CFTR	CM136671	CTG-ATG	Leu32Met	94C>A	L32M	Cystic fibrosis	Prach (2013) <i>J Mol Diagn</i> epub:epub Sobczynska-Tomaszewska (2013) <i>Eur J Hum Genet</i> 21:391	PubMed 23810505 PubMed 22892530
CFTR	CM132377	GAA-TAA	Glu33Term	97G>T	E33*	Cystic fibrosis	21:391	PubMed 22892530
CFTR	CM034776	GAC-AAC	Asp36Asn	106G>A	D36N	Cystic fibrosis	Girodon (2003) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM920139	CAA-TAA	Gln39Term	115C>T	Q39*	Cystic fibrosis	Cutting (1992) <i>Am J Hum Genet</i> 50:1185	PubMed 1376017
CFTR	CM950232	TCT-TTT	Ser42Phe	125C>T	S42F	Cystic fibrosis	Ferec (1995) <i>Mol Cell Probes</i> 9:135	PubMed 7541510
CFTR	CM920140	GAT-GGT	Asp44Gly	131A>G	D44G	Cystic fibrosis	Fanen (1992) <i>Genomics</i> 13:770	PubMed 1379210
CFTR	CM970258	GCT-GAT	Ala46Asp	137C>A	A46D	Cystic fibrosis	Tzetic (1997) <i>Hum Genet</i> 99:121	PubMed 9003508
CFTR	CM952045	TCT-CCT	Ser50Pro	148T>C	S50P	Cystic fibrosis	Casals (2000) <i>Hum Reprod</i> 15:1476	PubMed 10875853
CFTR	CM970259	TCT-TAT	Ser50Tyr	149C>A	S50Y	Cystic fibrosis	Zielenski (1997) <i>Hum Mutat</i> 9:183	PubMed 9067761
CFTR	CM067734	AGA-AAA	Arg55Lys	164G>A	R55K	Cystic fibrosis	Pasquet (2006) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM972934	TGG-CGG	Trp57Arg	169T>C	W57R	Cystic fibrosis	Malone (1997) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM950233	TGG-GGG	Trp57Gly	169T>G	W57G	Cystic fibrosis	Brancolini (1995) <i>Hum Genet</i> 96:312	PubMed 7544319
CFTR	CM930096	TGG-TAG	Trp57Term	170G>A	W57*	Cystic fibrosis	Audrezet (1993) <i>Hum Mol Genet</i> 2:51	PubMed 7683952
CFTR	CM994782	TGG-TGA	Trp57Term	171G>A	W57*	Cystic fibrosis	Le Maréchal (2001) <i>Hum Genet</i> 108:290	PubMed 11379874
CFTR	CM003234	GAT-GGT	Asp58Gly	173A>G	D58G	Cystic fibrosis	Claustres (2000) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM1213091	AGA-TGA	Arg59Term	175A>T	R59*	Cystic fibrosis	Deepak (2012) <i>J Pediatr Genet</i> 1:221	PubMed: No PubMed ID
CFTR	CM003235	GAG-AAG	Glu60Lys	178G>A	E60K	Cystic fibrosis	Claustres (2000) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM920141	GAG-TAG	Glu60Term	178G>T	E60*	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM057577	CTG-CCG	Leu61Pro	182T>C	L61P	Cystic fibrosis	Stark (2005) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM034777	AAG-GAG	Lys64Glu	190A>G	K64E	Cystic fibrosis	Girodon (2003) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM034778	AAT-AGT	Asn66Ser	197A>G	N66S	Cystic fibrosis	Cashman (2003) <i>CFTR LSDB entry Newsletter</i> :#55	Source: LSDB
CFTR	CM930097	CCT-CTT	Pro67Leu	200C>T	P67L	Cystic fibrosis	Claustres (1993) <i>Hum Mol Genet</i> 2:1209	PubMed 7691344

CFTR	CM076095	CCT-TCT	Pro67Ser	199C>T	P67S	Cystic fibrosis	Kraus (2007) <i>Cell Physiol Biochem</i> 19:239	PubMed 17495464
CFTR	CM920979	AAA-AAT	Lys68Asn	204A>T	K68N	Cystic fibrosis	Doerk (1992) <i>CFTR LSDB entry Newsletter</i> :#48	Source: LSDB
CFTR	CM972935	AAA-GAA	Lys68Glu	202A>G	K68E	Cystic fibrosis	Kilinc (2002) <i>Am J Med Genet</i> 113:250	PubMed 12439892
CFTR	CM090068	CTC-CAC	Leu69His	206T>A	L69H	Cystic fibrosis	Sharma (2009) <i>Ann Hum Genet</i> 73:26	PubMed 18782298
CFTR	CM962450	GCC-GAC	Ala72Asp	215C>A	A72D	Cystic fibrosis	Le Gall (1996) <i>CFTR LSDB entry Newsletter</i> :#68	Source: LSDB
CFTR	CM993844	GCC-ACC	Ala72Thr	214G>A	A72T	Cystic fibrosis	Pacheco (1999) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM003236	CGG-CAG	Arg74Gln	221G>A	R74Q	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:375	PubMed 16049310
CFTR	CM930098	CGG-TGG	Arg74Trp	220C>T	R74W	Cystic fibrosis	Claustres (1993) <i>Hum Mol Genet</i> 2:1209	PubMed 7691344
CFTR	CM126469	CGA-GGA	Arg75Gly	223C>G	R75G	Cystic fibrosis	Sachdeva (2012) <i>Genet Test Mol Biomarkers</i> 16:798	PubMed 22299590
CFTR	CM950234	CGA-CTA	Arg75Leu	224G>T	R75L	Cystic fibrosis	Chillon (1995) <i>N Engl J Med</i> 332:1475	PubMed 7739684
CFTR	CM940236	CGA-TGA	Arg75Term	223C>T	R75*	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM931140	TGG-CGG	Trp79Arg	235T>C	W79R	Cystic fibrosis	Macek (1993) <i>CFTR LSDB entry Newsletter</i> :#56	Source: LSDB
CFTR	CM931141	TGG-TAG	Trp79Term	236G>A	W79*	Cystic fibrosis	Ferec (1993) <i>CFTR LSDB entry Newsletter</i> :#58	Source: LSDB
CFTR	CM067768	ATG-GTG	Met82Val	244A>G	M82V	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM067752	TAT-CAT	Tyr84His	250T>C	Y84H	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM910068	GGA-GAA	Gly85Glu	254G>A	G85E	Cystic fibrosis	Zielenski (1991) <i>Genomics</i> 10:229	PubMed 1710599
CFTR	CM970262	GGA-GTA	Gly85Val	254G>T	G85V	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM940237	TTT-CTT	Phe87Leu	259T>C	F87L	Cystic fibrosis	Bienvenu (1994) <i>Hum Mutat</i> 3:395	PubMed 8081395
CFTR	CM960275	TTA-TCA	Leu88Ser	263T>C	L88S	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM940238	TTA-TAA	Leu88Term	263T>A	L88*	Cystic fibrosis	Savov (1994) <i>Hum Mol Genet</i> 3:57	PubMed 7512860
CFTR	CM920142	TTA-TGA	Leu88Term	263T>G	L88*	Cystic fibrosis	Macek (1992) <i>Hum Mutat</i> 1:501	PubMed 1284542
CFTR	CM993845	TAT-TGT	Tyr89Cys	266A>G	Y89C	Cystic fibrosis	Giusti (2007) <i>Pediatrics</i> 119:e460	PubMed 17272608
CFTR	CM983535	TTA-TCA	Leu90Ser	269T>C	L90S	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM930099	GGG-AGG	Gly91Arg	271G>A	G91R	Cystic fibrosis	Guillermi (1993) <i>Hum Genet</i> 91:233	PubMed 7682984
CFTR	CM043458	GAA-GAT	Glu92Asp	276A>T	E92D	Cystic fibrosis	Girodon (2004) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM920143	GAA-AAA	Glu92Lys	274G>A	E92K	Cystic fibrosis	Shackleton (1992) <i>Hum Mol Genet</i> 1:439	PubMed 1284529
CFTR	CM940239	GAA-TAA	Glu92Term	274G>T	E92*	Cystic fibrosis	Will (1994) <i>J Clin Invest</i> 93:1852	PubMed 7512993
CFTR	CM983536	GCA-GAA	Ala96Glu	287C>A	A96E	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM950235	CAG-CGG	Gln98Arg	293A>G	Q98R	Cystic fibrosis	Romey (1995) <i>Hum Mutat</i> 6:190	PubMed 7581407
CFTR	CM980332	CAG-TAG	Gln98Term	292C>T	Q98*	Cystic fibrosis	Malone (1998) <i>Hum Mutat</i> 11:152	PubMed 9482579
CFTR	CM920980	CCT-CTT	Pro99Leu	296C>T	P99L	Cystic fibrosis	Schwartz (1992) <i>CFTR LSDB entry Newsletter</i> :#50	Source: LSDB
CFTR	CM962451	TTA-TGA	Leu101Term	302T>G	L101*	Cystic fibrosis	Casals (1996) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM057578	CTG-CGG	Leu102Arg	305T>G	L102R	Cystic fibrosis	Torricelli (2005) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM015352	CTG-CCG	Leu102Pro	305T>C	L102P	Cystic fibrosis	Fresquet (2011) <i>J Mol Diagn</i> 13:520	PubMed 21708286
CFTR	CM920981	GGA-TGA	Gly103Term	307G>T	G103*	Cystic fibrosis	Edkins (1992) <i>CFTR LSDB entry Newsletter</i> :#47	Source: LSDB
CFTR	CM003237	ATC-AAC	Ile105Asn	314T>A	I105N	Cystic fibrosis	Claustres (2000) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM087273	GCT-GTT	Ala107Val	320C>T	A107V	Cystic fibrosis	Rana-Díez (2008) <i>J Cyst Fibros</i> 7:520	PubMed 18676185
CFTR	CM950236	TCC-TTC	Ser108Phe	323C>T	S108F	Cystic fibrosis	Seydewitz (1995) <i>Hum Mutat</i> 6:278	PubMed: No PubMed ID
CFTR	CM983537	TAT-AAT	Tyr109Asn	325T>A	Y109N	Cystic fibrosis	Schaedel (1998) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM940240	TAT-TGT	Tyr109Cys	326A>G	Y109C	Cystic fibrosis	Schaedel (1994) <i>Hum Mol Genet</i> 3:1001	PubMed 7524909
CFTR	CM040400	TAT-TAA	Tyr109Term	327T>A	Y109*	Cystic fibrosis	Feuillet-Fieux (2004) <i>Clin Genet</i> 65:284	PubMed 15025720
CFTR	CM993846	GAC-GAA	Asp110Glu	330C>A	D110E	Cystic fibrosis	Padoan (2002) <i>Acta Paediatr</i> 91:82	PubMed 11883825

CFTR	CM900042	GAC-CAC	Asp110His	328G>C	D110H	Cystic fibrosis	Dean (1990) <i>Cell</i> 61:863	PubMed 2344617
CFTR	CM003238	GAC-TAC	Asp110Tyr	328G>T	D110Y	Cystic fibrosis	Casals (2000) <i>Hum Reprod</i> 15:1476	PubMed 10875853
CFTR	CM962452	CCG-GCG	Pro111Ala	331C>G	P111A	Cystic fibrosis	Ferec (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM043459	AAG-TAG	Lys114Term	340A>T	K114*	Cystic fibrosis	Bonizzato (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM001648	GAG-TAG	Glu115Term	343G>T	E115*	Cystic fibrosis	Banjar (1999) <i>Ann Trop Paediatr</i> 19:69	PubMed 10605524
CFTR	CM003239	GAA-CAA	Glu116Gln	346G>C	E116Q	Cystic fibrosis	Walker (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM941966	GAA-AAA	Glu116Lys	346G>A	E116K	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM066753	GAA-TAA	Glu116Term	346G>T	E116*	Cystic fibrosis	Munthe-Kaas (2006) <i>Respir Med</i> 100:2121	PubMed 16678395
CFTR	CM940241	CGC-TGC	Arg117Cys	349C>T	R117C	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM900043	CGC-CAC	Arg117His	350G>A	R117H	Cystic fibrosis	Dean (1990) <i>Cell</i> 61:863	PubMed 2344617
CFTR	CM950237	CGC-CTC	Arg117Leu	350G>T	R117L	Cystic fibrosis	Ferec (1995) <i>Mol Cell Probes</i> 9:135	PubMed 7541510
CFTR	CM930100	CGC-CCC	Arg117Pro	350G>C	R117P	Cystic fibrosis	Reiss (1993) <i>Hum Mol Genet</i> 2:809	PubMed 7689013
CFTR	CM024661	ATC-GTC	Ile119Val	355A>G	I119V	Cystic fibrosis	Schwarz (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM940242	GCG-ACG	Ala120Thr	358G>A	A120T	Cystic fibrosis	Chillon (1994) <i>Hum Genet</i> 93:447	PubMed 7513293
CFTR	CM034780	GCG-GTG	Ala120Val	359C>T	A120V	Cystic fibrosis	Casals (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067766	TAT-TGT	Tyr122Cys	365A>G	Y122C	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM920144	TAT-TAA	Tyr122Term	366T>A	Y122*	Cystic fibrosis	Chevalier (1992) <i>Hum Mol Genet</i> 1:647	PubMed 1284471
CFTR	CM983538	ATA-ACA	Ile125Thr	374T>C	I125T	Cystic fibrosis	Kilinc (2002) <i>Am J Med Genet</i> 113:250	PubMed 12439892
CFTR	CM940243	GGC-GAC	Gly126Asp	377G>A	G126D	Cystic fibrosis	Wagner (1994) <i>Hum Hered</i> 44:56	PubMed 8163293
CFTR	CM962453	TTA-TGA	Leu127Term	380T>G	L127*	Cystic fibrosis	Ferec (1996) <i>CFTR LSDB entry</i> Newsletter:#67	Source: LSDB
CFTR	CM972936	CTC-CGC	Leu137Arg	410T>G	L137R	Cystic fibrosis	Chevalier-Porst (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM962454	CTC-CAC	Leu137His	410T>A	L137H	Cystic fibrosis	Wallace (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM051410	CTC-CCC	Leu137Pro	410T>C	L137P	Cystic fibrosis	Patrinis (2005) <i>CFTR LSDB entry</i> :	Source: LSDB
CFTR	CM950238	CAC-CGC	His139Arg	416A>G	H139R	Cystic fibrosis	Ferec (1995) <i>Mol Cell Probes</i> 9:135	PubMed 7541510
CFTR	CM001649	CAC-CTC	His139Leu	416A>T	H139L	Cystic fibrosis	Banjar (1999) <i>Ann Trop Paediatr</i> 19:69	PubMed 10605524
CFTR	CM983539	CCA-CTA	Pro140Leu	419C>T	P140L	Cystic fibrosis	Tzetzis (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM941967	CCA-TCA	Pro140Ser	418C>T	P140S	Cystic fibrosis	Ferec (1994) <i>CFTR LSDB entry</i> Newsletter:#61	Source: LSDB
CFTR	CM970264	GCC-GAC	Ala141Asp	422C>A	A141D	Cystic fibrosis	Gouya (1997) <i>Hum Mutat</i> 10:86	PubMed 9222768
CFTR	CM057579	CTT-CAT	Leu145His	434T>A	L145H	Cystic fibrosis	Wang (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM962456	ATT-AAT	Ile148Asn	443T>A	I148N	Cystic fibrosis	Hirtz (2004) <i>Gastroenterology</i> 127:1085	PubMed 15480987
CFTR	CM950239	GGA-AGA	Gly149Arg	445G>A	G149R	Cystic fibrosis	Mercier (1995) <i>Am J Hum Genet</i> 56:272	PubMed 7529962
CFTR	CM086804	GGA-TGA	Gly149Term	445G>T	G149*	Cystic fibrosis	Shastri (2008) <i>J Cyst Fibros</i> 7:110	PubMed 17716958
CFTR	CM067763	GGA-GTA	Gly149Val	446G>T	G149V	Cystic fibrosis	Pacheco (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM063899	ATG-AAG	Met150Lys	449T>A	M150K	Cystic fibrosis	Kammesheidt (2006) <i>Genet Med</i> 8:557	PubMed 16980811
CFTR	CM920146	CAG-TAG	Gln151Term	451C>T	Q151*	Cystic fibrosis	Shackleton (1992) <i>Hum Mol Genet</i> 1:439	PubMed 1284529
CFTR	CM983540	ATG-AGG	Met152Arg	455T>G	M152R	Cystic fibrosis	Morokawa (2000) <i>Hum Mutat</i> 15:485	PubMed 10790220
CFTR	CM931142	ATG-GTG	Met152Val	454A>G	M152V	Cystic fibrosis	Wong (2004) <i>Electrophoresis</i> 25:2593	PubMed 15300780
CFTR	CM141625	AGA-ATA	Arg153Ile	458G>T	R153I	Cystic fibrosis	Ziętkiewicz (2014) <i>PLoS One</i> 9:e89094	PubMed 24586523
CFTR	CM972937	GCT-CCT	Ala155Pro	463G>C	A155P	Cystic fibrosis	Zielenski (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM034009	AGT-AAT	Ser158Asn	473G>A	S158N	Cystic fibrosis	Hicks (2003) <i>Genet Test</i> 7:73	PubMed 12820707
CFTR	CM055123	AGT-ACT	Ser158Thr	473G>C	S158T	Cystic fibrosis	McGinniss (2005) <i>Hum Genet</i> 118:331	PubMed 16189704
CFTR	CM024662	TTG-TCG	Leu159Ser	476T>C	L159S	Cystic fibrosis	Alonso (2006) <i>Ann Hum Genet</i> 71:194	PubMed 17331079
CFTR	CM003240	TTG-TAG	Leu159Term	476T>A	L159*	Cystic fibrosis	Andrew (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB

CFTR	CM003241	TAT-AAT	Tyr161Asn	481T>A	Y161N	Cystic fibrosis	des Georges (2004) <i>J Cyst Fibros</i> 3:265	PubMed 15698946
CFTR	CM993847	TAT-GAT	Tyr161Asp	481T>G	Y161D	Cystic fibrosis	Strandvik (2001) <i>Genet Test</i> 5:235	PubMed 11788090
CFTR	CM044864	TAT-TGT	Tyr161Cys	482A>G	Y161C	Cystic fibrosis	Hirtz (2004) <i>Gastroenterology</i> 127:1085	PubMed 15480987
CFTR	CM993848	TAT-TCT	Tyr161Ser	482A>C	Y161S	Cystic fibrosis	Andrew (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983541	AAG-GAG	Lys162Glu	484A>G	K162E	Cystic fibrosis	Tzetis (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM920982	TTA-TCA	Leu165Ser	494T>C	L165S	Cystic fibrosis	Quint (2005) <i>Am J Med Genet</i> 136A:246	PubMed 15948195
CFTR	CM141626	TTA-TAA	Leu165Term	494T>A	L165*	Cystic fibrosis	Ziętkiewicz (2014) <i>PLoS One</i> 9:e89094	PubMed 24586523
CFTR	CM970265	AAG-GAG	Lys166Glu	496A>G	K166E	Cystic fibrosis	Macek (1997) <i>Hum Mutat</i> 9:136	PubMed 9067754
CFTR	CM116798	CTG-CGG	Leu167Arg	500T>G	L167R	Cystic fibrosis	Fresquet (2011) <i>J Mol Diagn</i> 13:520	PubMed 21708286
CFTR	CM1213092	AGC-GGC	Ser169Gly	505A>G	S169G	Cystic fibrosis	Deepak (2012) <i>J Pediatr Genet</i> 1:221	PubMed: No PubMed ID
CFTR	CM941968	CGT-TGT	Arg170Cys	508C>T	R170C	Cystic fibrosis	Ferec (1994) <i>CFTR LSDB entry</i> Newsletter:#62	Source: LSDB
CFTR	CM920983	CGT-GGT	Arg170Gly	508C>G	R170G	Cystic fibrosis	Claustres (1992) <i>CFTR LSDB entry</i> Newsletter:#49	Source: LSDB
CFTR	CM940244	ATA-GTA	Ile175Val	523A>G	I175V	Cystic fibrosis	Romey (1994) <i>Hum Mol Genet</i> 3:661	PubMed 7520799
CFTR	CM962457	ATT-ACT	Ile177Thr	530T>C	I177T	Cystic fibrosis	Bienvenu (1996) <i>CFTR LSDB entry</i> Newsletter:#68	Source: LSDB
CFTR	CM910069	GGA-AGA	Gly178Arg	532G>A	G178R	Cystic fibrosis	Zielenski (1991) <i>Genomics</i> 10:229	PubMed 1710599
CFTR	CM043461	GGA-GAA	Gly178Glu	533G>A	G178E	Cystic fibrosis	Soldatova (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM003242	CAA-AAA	Gln179Lys	535C>A	Q179K	Cystic fibrosis	Wong (2004) <i>Electrophoresis</i> 25:2593	PubMed 15300780
CFTR	CM057583	CTC-ATC	Leu183Ile	547C>A	L183I	Cystic fibrosis	Shastri (2008) <i>J Cyst Fibros</i> 7:110	PubMed 17716958
CFTR	CM983542	AAC-AAA	Asn186Lys	558C>A	N186K	Cystic fibrosis	Claustres (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM1213093	AAC-GAC	Asn187Asp	559A>G	N187D	Cystic fibrosis	Deepak (2012) <i>J Pediatr Genet</i> 1:221	PubMed: No PubMed ID
CFTR	CM983543	AAC-AAA	Asn187Lys	561C>A	N187K	Cystic fibrosis	Gaia (2002) <i>Dig Dis Sci</i> 47:2416	PubMed 12452372
CFTR	CM060896	AAC-AAA	Asn189Lys	567C>A	N189K	Cystic fibrosis	Li (2006) <i>Chin Med J (Engl)</i> 119:103	PubMed 16454991
CFTR	CM021553	TTT-GTT	Phe191Val	571T>G	F191V	Cystic fibrosis	Groman (2002) <i>N Engl J Med</i> 347:401	PubMed 12167682
CFTR	CM941969	GAT-AAT	Asp192Asn	574G>A	D192N	Cystic fibrosis	Hubert (2004) <i>J Cyst Fibros</i> 3:15	PubMed 15463882
CFTR	CM940245	GAT-GGT	Asp192Gly	575A>G	D192G	Cystic fibrosis	Audrezet (1994) <i>Hum Genet</i> 93:659	PubMed 7516305
CFTR	CM950240	GAA-AAA	Glu193Lys	577G>A	E193K	Cystic fibrosis	Mercier (1995) <i>Am J Hum Genet</i> 56:272	PubMed 7529962
CFTR	CM941970	GAA-TAA	Glu193Term	577G>T	E193*	Cystic fibrosis	Ferec (1994) <i>CFTR LSDB entry</i> Newsletter:#62	Source: LSDB
CFTR	CM983544	GGA-GTA	Gly194Val	581G>T	G194V	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993849	GCA-CCA	Ala198Pro	592G>C	A198P	Cystic fibrosis	Walker (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM024663	CAT-CGT	His199Arg	596A>G	H199R	Cystic fibrosis	D'Apice (2004) <i>BMC Med Genet</i> 5:8	PubMed 15084222
CFTR	CM920147	CAT-CAG	His199Gln	597T>G	H199Q	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM940246	CAT-TAT	His199Tyr	595C>T	H199Y	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM001961	GTG-ATG	Val201Met	601G>A	V201M	Cystic fibrosis	Bernardino (2000) <i>Genet Test</i> 4:69	PubMed 10794365
CFTR	CM034781	TGG-TGA	Trp202Term	606G>A	W202*	Cystic fibrosis	Alonso (2006) <i>Ann Hum Genet</i> 71:194	PubMed 17331079
CFTR	CM087336	GCT-ACT	Ala204Thr	610G>A	A204T	Cystic fibrosis	Mutesa (2008) <i>Chest</i> 135:1233	PubMed 19017867
CFTR	CM024665	CCT-CGT	Pro205Arg	614C>G	P205R	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM930101	CCT-TCT	Pro205Ser	613C>T	P205S	Cystic fibrosis	Chillon (1993) <i>Hum Mol Genet</i> 2:1741	PubMed 7505694
CFTR	CM972938	TTG-TTT	Leu206Phe	618G>T	L206F	Cystic fibrosis	Ferec (1997) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM930102	TTG-TGG	Leu206Trp	617T>G	L206W	Cystic fibrosis	Claustres (1993) <i>Hum Mol Genet</i> 2:1209	PubMed 7691344
CFTR	CM931143	CAA-TAA	Gln207Term	619C>T	Q207*	Cystic fibrosis	Zielenski (1993) <i>CFTR LSDB entry</i> Newsletter:#58	Source: LSDB
CFTR	CM983545	GCA-TCA	Ala209Ser	625G>T	A209S	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM034782	CTC-CCC	Leu210Pro	629T>C	L210P	Cystic fibrosis	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB

CFTR	CM094116	GGG-GAG	Gly213Glu	638G>A	G213E	Cystic fibrosis	Seia (2009) <i>Clin Biochem</i> 42:611	PubMed 19318035
CFTR	CM024666	GGG-GTG	Gly213Val	638G>T	G213V	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993850	TGG-TAG	Trp216Term	647G>A	W216*	Cystic fibrosis	Yoshimura (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM972939	GAG-GGG	Glu217Gly	650A>G	E217G	Cystic fibrosis	Zielenski (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM962458	TTG-TAG	Leu218Term	653T>A	L218*	Cystic fibrosis	Zielenski (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM983546	CAG-CGG	Gln220Arg	659A>G	Q220R	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM940247	CAG-TAG	Gln220Term	658C>T	Q220*	Cystic fibrosis	Shackleton (1994) <i>Hum Mutat</i> 3:141	PubMed 7515303
CFTR	CM121017	GCC-GTC	Ala223Val	668C>T	A223V	Cystic fibrosis	Carr (2012) <i>Hum Mutat</i> 33:338	PubMed 22052625
CFTR	CM920148	TGT-CGT	Cys225Arg	673T>C	C225R	Cystic fibrosis	Fanen (1992) <i>Genomics</i> 13:770	PubMed 1379210
CFTR	CM962459	TGT-TGA	Cys225Term	675T>A	C225*	Cystic fibrosis	Mercier (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM002751	CTT-CGT	Leu227Arg	680T>G	L227R	Cystic fibrosis	Dequeker (2000) <i>Eur J Hum Genet</i> 8:S2	PubMed 11108532
CFTR	CM970266	GTC-GAC	Val232Asp	695T>A	V232D	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM941971	CAG-GAG	Gln237Glu	709C>G	Q237E	Cystic fibrosis	Hubert (2004) <i>J Cyst Fibros</i> 3:15	PubMed 15463882
CFTR	CM950241	GGG-AGG	Gly239Arg	715G>A	G239R	Cystic fibrosis	Bienvenu (1995) <i>Hum Hered</i> 45:53	PubMed 7534748
CFTR	CM011283	GGG-AGG	Gly241Arg	721G>A	G241R	Cystic fibrosis	Le Maréchal (2001) <i>Hum Genet</i> 108:290	PubMed 11379874
CFTR	CM993851	ATG-CTG	Met243Leu	727A>C	M243L	Cystic fibrosis	Yoshimura (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM970268	TAC-TAG	Tyr247Term	741C>G	Y247*	Cystic fibrosis	Tzetic (1997) <i>Hum Genet</i> 99:121	PubMed 9003508
CFTR	CM063902	GAA-TAA	Glu257Term	769G>T	E257*	Cystic fibrosis	Kammesheidt (2006) <i>Genet Med</i> 8:557	PubMed 16980811
CFTR	CM950242	AGA-GGA	Arg258Gly	772A>G	R258G	Cystic fibrosis	Mercier (1995) <i>Am J Hum Genet</i> 56:272	PubMed 7529962
CFTR	CM001962	TAC-TAG	Tyr275Term	825C>G	Y275*	Cystic fibrosis	Bernardino (2000) <i>Genet Test</i> 4:69	PubMed 10794365
CFTR	CM950243	TGC-TGA	Cys276Term	828C>A	C276*	Cystic fibrosis	Ferec (1995) <i>Mol Cell Probes</i> 9:135	PubMed 7541510
CFTR	CM043463	TGG-CGG	Trp277Arg	829T>C	W277R	Cystic fibrosis	Oller Ramirez (2006) <i>Mol Genet Metab</i> 87:370	PubMed 16423550
CFTR	CM067756	GAA-GAT	Glu279Asp	837A>T	E279D	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM066751	GAA-TAA	Glu279Term	835G>T	E279*	Cystic fibrosis	Munthe-Kaas (2006) <i>Respir Med</i> 100:2121	PubMed 16678395
CFTR	CM024667	ATG-ACG	Met281Thr	842T>C	M281T	Cystic fibrosis	Casals (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM055925	ATT-TTT	Ile285Phe	853A>T	I285F	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM970270	AAC-TAC	Asn287Tyr	859A>T	N287Y	Cystic fibrosis	Shrimpton (1997) <i>Hum Mutat</i> 10:436	PubMed 9401006
CFTR	CM952046	CAA-TAA	Gln290Term	868C>T	Q290*	Cystic fibrosis	Ferec (1995) <i>CFTR LSDB entry</i> Newsletter:#65	Source: LSDB
CFTR	CM962763	GAA-AAA	Glu292Lys	874G>A	E292K	Cystic fibrosis	Bienvenu (1996) <i>CFTR LSDB entry</i> Newsletter:#65	Source: LSDB
CFTR	CM024668	CTG-ATG	Leu293Met	877C>A	L293M	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM066752	CTG-CAG	Leu295Gln	884T>A	L295Q	Cystic fibrosis	Munthe-Kaas (2006) <i>Respir Med</i> 100:2121	PubMed 16678395
CFTR	CM920149	CGG-CAG	Arg297Gln	890G>A	R297Q	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM993852	GCA-ACA	Ala299Thr	895G>A	A299T	Cystic fibrosis	Ferec (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM962460	TAC-TAG	Tyr304Term	912C>G	Y304*	Cystic fibrosis	Ferec (1996) <i>CFTR LSDB entry</i> Newsletter:#67	Source: LSDB
CFTR	CM067754	TTC-GTC	Phe305Val	913T>G	F305V	Cystic fibrosis	Poulou (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB Source: Online submission
CFTR	HM971268	AGC-AAC	Ser307Asn	920G>A	S307N	Cystic fibrosis	Onay (1998) <i>Hum Genet</i> 102:710	PubMed 9401006
CFTR	CM941972	GCC-GAC	Ala309Asp	926C>A	A309D	Cystic fibrosis	Ferrari (1994) <i>CFTR LSDB entry</i> Newsletter:#64	Source: LSDB
CFTR	CM980334	GCC-GGC	Ala309Gly	926C>G	A309G	Cystic fibrosis	Cartault (1998) <i>Clin Genet</i> 54:437	PubMed 9842999
CFTR	CM024669	GCC-ACC	Ala309Thr	925G>A	A309T	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM920150	TTC-CTC	Phe311Leu	931T>C	F311L	Cystic fibrosis	Ferec (1992) <i>Nat Genet</i> 1:188	PubMed 1284639
CFTR	CM057582	TCA-TAA	Ser313Term	938C>A	S313*	Cystic fibrosis	Torricelli (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM141627	GGG-AGG	Gly314Arg	940G>A	G314R	Cystic fibrosis	Ziętkiewicz (2014) <i>PLoS One</i> 9:e89094	PubMed 24586523
CFTR	CM960276	GGG-CGG	Gly314Arg	940G>C	G314R	Cystic fibrosis	Nasr (1996) <i>Hum Mutat</i> 7:151	PubMed 8829633

CFTR	CM920151	GGG-GAG	Gly314Glu	941G>A	G314E	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197 Chevalier-Porst (1997) <i>CFTR LSDB entry</i> Newsletter:#70	PubMed 1284534
CFTR	CM972941	GGG-GTG	Gly314Val	941G>T	G314V	Cystic fibrosis		Source: LSDB
CFTR	CM931144	GTG-GCG	Val317Ala	950T>C	V317A	Cystic fibrosis	Ferec (1993) <i>CFTR LSDB entry</i> Newsletter:#55	Source: LSDB
CFTR	CM941973	TTA-TTT	Leu320Phe	960A>T	L320F	Cystic fibrosis	Macek (1994) <i>CFTR LSDB entry</i> Newsletter:#64	Source: LSDB
CFTR	CM015354	TTA-TAA	Leu320Term	959T>A	L320*	Cystic fibrosis	Iron (2001) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM043464	TCT-CCT	Ser321Pro	961T>C	S321P	Cystic fibrosis	Seia (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM941974	GTG-GCG	Val322Ala	965T>C	V322A	Cystic fibrosis	Ferec (1994) <i>CFTR LSDB entry</i> Newsletter:#63	Source: LSDB
CFTR	CM011284	GTG-ATG	Val322Met	964G>A	V322M	Cystic fibrosis	Le Maréchal (2001) <i>Hum Genet</i> 108:290	PubMed 11379874
CFTR	CM136672	CTT-CCT	Leu323Pro	968T>C	L323P	Cystic fibrosis	Prach (2013) <i>J Mol Diagn</i> epub:epub	PubMed 23810505
CFTR	CM970272	GGA-TGA	Gly330Term	988G>T	G330*	Cystic fibrosis	Macek (1997) <i>Am J Hum Genet</i> 60:1122	PubMed 9150159
CFTR	CM952047	CGG-CAG	Arg334Gln	1001G>A	R334Q	Cystic fibrosis	Ferec (1995) <i>CFTR LSDB entry</i> Newsletter:#65	Source: LSDB
CFTR	CM910070	CGG-TGG	Arg334Trp	1000C>T	R334W	Cystic fibrosis	Gasparini (1991) <i>Genomics</i> 10:193	PubMed 2045102
CFTR	CM930103	ATA-AAA	Ile336Lys	1007T>A	I336K	Cystic fibrosis	Cuppens (1993) <i>Genomics</i> 18:693	PubMed 7508414
CFTR	CM930104	ACC-ATC	Thr338Ile	1013C>T	T338I	Cystic fibrosis	Saba (1993) <i>Hum Mol Genet</i> 2:1739	PubMed 7505693
CFTR	CM024671	ATC-AAC	Ile340Asn	1019T>A	I340N	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM057576	TCA-CCA	Ser341Pro	1021T>C	S341P	Cystic fibrosis	Wang (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM940248	CTG-CCG	Leu346Pro	1037T>C	L346P	Cystic fibrosis	Boteva (1994) <i>Hum Genet</i> 93:529	PubMed 7513296
CFTR	CM931146	CGC-TGC	Arg347Cys	1039C>T	R347C	Cystic fibrosis	Ferec (1993) <i>CFTR LSDB entry</i> Newsletter:#56	Source: LSDB
CFTR	CM920152	CGC-CAC	Arg347His	1040G>A	R347H	Cystic fibrosis	Cremonesi (1992) <i>Hum Mutat</i> 1:314	PubMed 1284538
CFTR	CM930105	CGC-CTC	Arg347Leu	1040G>T	R347L	Cystic fibrosis	Audrezet (1993) <i>Hum Mol Genet</i> 2:51	PubMed 7683952
CFTR	CM900044	CGC-CCC	Arg347Pro	1040G>C	R347P	Cystic fibrosis	Dean (1990) <i>Cell</i> 61:863	PubMed 2344617
CFTR	CM930106	ATG-AAG	Met348Lys	1043T>A	M348K	Cystic fibrosis	Audrezet (1993) <i>Hum Hered</i> 43:295	PubMed 8406518
CFTR	CM067738	ATG-ACG	Met348Thr	1043T>C	M348T	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM930107	GCG-GTG	Ala349Val	1046C>T	A349V	Cystic fibrosis	Audrezet (1993) <i>Hum Mol Genet</i> 2:51	PubMed 7683952
CFTR	CM920153	CGG-CAG	Arg352Gln	1055G>A	R352Q	Cystic fibrosis	Cremonesi (1992) <i>Hum Mutat</i> 1:314	PubMed 1284538
CFTR	CM034783	CGG-GGG	Arg352Gly	1054C>G	R352G	Cystic fibrosis	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM962462	CGG-TGG	Arg352Trp	1054C>T	R352W	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM952048	CAA-CAC	Gln353His	1059A>C	Q353H	Cystic fibrosis	Ferec (1995) <i>CFTR LSDB entry</i> Newsletter:#65	Source: LSDB
CFTR	CM003243	CAA-TAA	Gln353Term	1057C>T	Q353*	Cystic fibrosis	Seia (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067769	CCC-TCC	Pro355Ser	1063C>T	P355S	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM043465	TGG-TCG	Trp356Ser	1067G>C	W356S	Cystic fibrosis	Barbolina (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983547	TGG-TGA	Trp356Term	1068G>A	W356*	Cystic fibrosis	Ellis (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM993853	CAA-CGA	Gln359Arg	1076A>G	Q359R	Cystic fibrosis	Ferec (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983548	ACA-AGA	Thr360Arg	1079C>G	T360R	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983549	TGG-AGG	Trp361Arg	1081T>A	W361R	Cystic fibrosis	Alonso (2006) <i>Ann Hum Genet</i> 71:194	PubMed 17331079
CFTR	CM931147	TGG-CGG	Trp361Arg	1081T>C	W361R	Cystic fibrosis	Bienvenu (1993) <i>CFTR LSDB entry</i> Newsletter:#56	Source: LSDB
CFTR	CM104904	TAT-TAA	Tyr362Term	1086T>A	Y362*	Cystic fibrosis	Makukh (2010) <i>J Cyst Fibros</i> 9:371	PubMed 20659818
CFTR	CM021069	TAT-TAG	Tyr362Term	1086T>G	Y362*	Cystic fibrosis	Visich (2002) <i>Clin Genet</i> 61:207	PubMed 12000363
CFTR	CM970274	TCT-CCT	Ser364Pro	1090T>C	S364P	Cystic fibrosis	Macek (1997) <i>Am J Hum Genet</i> 60:1122	PubMed 9150159
CFTR	CM003244	CTT-CCT	Leu365Pro	1094T>C	L365P	Cystic fibrosis	Alonso (2006) <i>Ann Hum Genet</i> 71:194	PubMed 17331079
CFTR	CM1212240	CAG-TAG	Gln372Term	1114C>T	Q372*	Cystic fibrosis	Křenková (2013) <i>J Cyst Fibros</i> 12:532	PubMed 23276700
CFTR	CM067758	GAT-GAG	Asp373Glu	1119T>G	D373E	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM960277	TTA-TTC	Leu375Phe	1125A>C	L375F	Cystic fibrosis	Jezequel (1996) <i>Hum Genet</i> 97:548	PubMed 8834261

CFTR	CM024672	CAA-CGA	Gln378Arg	1133A>G	Q378R	Cystic fibrosis	Tzetzis (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM003245	GAA-TAA	Glu379Term	1135G>T	E379*	Cystic fibrosis	Glaeser (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM972942	TTG-TCG	Leu383Ser	1148T>C	L383S	Cystic fibrosis	Casals (2000) <i>Hum Reprod</i> 15:1476	PubMed 10875853
CFTR	CM972943	GTA-GGA	Val392Gly	1175T>G	V392G	Cystic fibrosis	Zielenski (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM983550	ATG-AGG	Met394Arg	1181T>G	M394R	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM003246	GCC-GTC	Ala399Val	1196C>T	A399V	Cystic fibrosis	Yoshimura (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM972944	TGG-TAG	Trp401Term	1202G>A	W401*	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:375	PubMed 16049310
CFTR	CM930111	TGG-TGA	Trp401Term	1203G>A	W401*	Cystic fibrosis	Cuppens (1993) <i>Genomics</i> 18:693	PubMed 7508414
CFTR	CM993854	GAG-GAC	Glu403Asp	1209G>C	E403D	Cystic fibrosis	Ferec (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993855	GAA-GTA	Glu407Val	1220A>T	E407V	Cystic fibrosis	Zielenski (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM940249	CAA-TAA	Gln414Term	1240C>T	Q414*	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 93:67	PubMed 7505767
CFTR	CM067729	AAT-AGT	Asn416Ser	1247A>G	N416S	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM941975	AAT-AGT	Asn418Ser	1253A>G	N418S	Cystic fibrosis	Sava (1994) <i>CFTR LSDB entry</i> Newsletter:#64	Source: LSDB
CFTR	CM003247	GGT-AGT	Gly424Ser	1270G>A	G424S	Cystic fibrosis	Bienvenu (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067748	AGT-GGT	Ser431Gly	1291A>G	S431G	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM983551	TCA-TGA	Ser434Term	1301C>G	S434*	Cystic fibrosis	Leymarie (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM087536	GGT-GAT	Gly437Asp	1310G>A	G437D	Cystic fibrosis	Kolesár (2008) <i>Gen Physiol Biophys</i> 27:299	PubMed 19202204
CFTR	CM055927	CCT-TCT	Pro439Ser	1315C>T	P439S	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM102814	CTG-CCG	Leu441Pro	1322T>C	L441P	Cystic fibrosis	Gee (2010) <i>J Korean Med Sci</i> 25:166	PubMed 20052366
CFTR	CM993856	ATT-AGT	Ile444Ser	1331T>G	I444S	Cystic fibrosis	Zielenski (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM074731	ATT-ACT	Ile444Thr	1331T>C	I444T	Cystic fibrosis	Pasquet (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM055125	GGA-GTA	Gly451Val	1352G>T	G451V	Cystic fibrosis	McGinniss (2005) <i>Hum Genet</i> 118:331	PubMed 16189704
CFTR	CM972945	CAG-CCG	Gln452Pro	1355A>C	Q452P	Cystic fibrosis	Claustres (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM900045	GCG-GAG	Ala455Glu	1364C>A	A455E	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM022334	GTT-GCT	Val456Ala	1367T>C	V456A	Cystic fibrosis	McCormick (2002) <i>Eur J Hum Genet</i> 10:583	PubMed 12357328
CFTR	CM940250	GTT-TTT	Val456Phe	1366G>T	V456F	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 93:67	PubMed 7505767
CFTR	CM1110465	GCT-CCT	Ala457Pro	1369G>C	A457P	Cystic fibrosis	Cole (2011) <i>Case Report Med</i> 2011:	PubMed 22194755
CFTR	CM920154	GGA-GTA	Gly458Val	1373G>T	G458V	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM021554	GGC-GAC	Gly463Asp	1388G>A	G463D	Cystic fibrosis	Groman (2002) <i>N Engl J Med</i> 347:401	PubMed 12167682
CFTR	CM034784	AAG-AAT	Lys464Asn	1392G>T	K464N	Cystic fibrosis	Bieth (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM063897	ACT-AAT	Thr465Asn	1394C>A	T465N	Cystic fibrosis	Kammesheidt (2006) <i>Genet Med</i> 8:557	PubMed 16980811
CFTR	CM960278	TCA-TAA	Ser466Term	1397C>A	S466*	Cystic fibrosis	Mittre (1996) <i>Hum Mutat</i> 8:392	PubMed 8956053
CFTR	CM940251	TCA-TGA	Ser466Term	1397C>G	S466*	Cystic fibrosis	Deufel (1994) <i>Hum Mutat</i> 3:64	PubMed 7509683
CFTR	CM063898	CTT-TTT	Leu467Phe	1399C>T	L467F	Cystic fibrosis	Elahi (2006) <i>J Mol Diagn</i> 8:119	PubMed 16436643
CFTR	CM107799	CTT-CCT	Leu467Pro	1400T>C	L467P	Cystic fibrosis	Jambhekar (2010) <i>J Cyst Fibros</i> 9:269	PubMed 20510657
CFTR	CM994784	GAA-AAA	Glu474Lys	1420G>A	E474K	Cystic fibrosis	Gironon (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM074728	GAG-TAG	Glu479Term	1435G>T	E479*	Cystic fibrosis	Narzi (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM952251	GGT-GAT	Gly480Asp	1439G>A	G480D	Cystic fibrosis	Hawthorn (1995) <i>CFTR LSDB entry</i> Newsletter:#66	Source: LSDB
CFTR	CM920155	GGT-TGT	Gly480Cys	1438G>T	G480C	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM015355	GGT-AGT	Gly480Ser	1438G>A	G480S	Cystic fibrosis	Kawasoe (2001) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM055126	AAA-GAA	Lys481Glu	1441A>G	K481E	Cystic fibrosis	McGinniss (2005) <i>Hum Genet</i> 118:331	PubMed 16189704
CFTR	CM983552	CAC-CGC	His484Arg	1451A>G	H484R	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993857	AGT-TGT	Ser485Cys	1453A>T	S485C	Cystic fibrosis	Andrew (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB

CFTR	CM952050	GGA-TGA	Gly486Term	1456G>T	G486*	Cystic fibrosis	Hughes (1995) <i>CFTR LSDB entry Newsletter</i> :#65	Source: LSDB
CFTR	CM032555	ATT-ATG	Ile488Met	1464T>G	I488M	Cystic fibrosis	Korytina (2003) <i>Mol Biol (Mosk)</i> 37:61	PubMed 12624947
CFTR	CM970276	TCA-TAA	Ser489Term	1466C>A	S489*	Cystic fibrosis	Plouvier (1997) <i>Ann Genet</i> 40:185 Chevalier-Porst (1998) <i>CFTR LSDB entry Newsletter</i> :#70	PubMed 9401110 Source: LSDB
CFTR	CM983553	TGT-CGT	Cys491Arg	1471T>C	C491R	Cystic fibrosis	Ferec (1992) <i>Nat Genet</i> 1:188	PubMed 1284639
CFTR	CM920156	TCT-TTT	Ser492Phe	1475C>T	S492F	Cystic fibrosis	Savov (1994) <i>Hum Mol Genet</i> 3:57	PubMed 7512860
CFTR	CM940252	CAG-CGG	Gln493Arg	1478A>G	Q493R	Cystic fibrosis	Sharma (2009) <i>Ann Hum Genet</i> 73:26	PubMed 18782298
CFTR	CM090069	CAG-CTG	Gln493Leu	1478A>T	Q493L	Cystic fibrosis	Kilinc (2002) <i>Am J Med Genet</i> 113:250	PubMed 12439892
CFTR	CM024232	CAG-CCG	Gln493Pro	1478A>C	Q493P	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM900046	CAG-TAG	Gln493Term	1477C>T	Q493*	Cystic fibrosis	Balassopoulou (1994) <i>Hum Mol Genet</i> 3:1887	PubMed 7531541
CFTR	CM940253	TGG-TAG	Trp496Term	1487G>A	W496*	Cystic fibrosis	Bienvenu (2005) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM057571	ATG-ATC	Met498Ile	1494G>C	M498I	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM993858	ACC-GCC	Thr501Ala	1501A>G	T501A	Cystic fibrosis	Hinks (2002) <i>CFTR LSDB entry Pers comm</i> : Chevalier-Porst (1998) <i>CFTR LSDB entry Newsletter</i> :#70	Source: LSDB PubMed 1284534
CFTR	CM024676	ATT-AAT	Ile502Asn	1505T>A	I502N	Cystic fibrosis	Casals (1997) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM983554	ATT-ACT	Ile502Thr	1505T>C	I502T	Cystic fibrosis	Strandvik (2001) <i>Genet Test</i> 5:235	PubMed 11788090
CFTR	CM920157	GAA-CAA	Glu504Gln	1510G>C	E504Q	Cystic fibrosis	Chevalier-Porst (1994) <i>J Med Genet</i> 31:541	PubMed 7525963
CFTR	CM972947	GAA-TAA	Glu504Term	1510G>T	E504*	Cystic fibrosis	Deufel (1994) <i>Hum Mutat</i> 3:64	PubMed 7509683
CFTR	CM983555	ATC-CTC	Ile506Leu	1516A>C	I506L	Cystic fibrosis	Desgeorges (1995) <i>Hum Mutat</i> 6:279	PubMed: No PubMed ID
CFTR	CM940255	ATC-ATG	Ile506Met	1518C>G	I506M	Cystic fibrosis	Kobayashi (1990) <i>Am J Hum Genet</i> 47:611	PubMed 1977306
CFTR	CM940254	ATC-AGC	Ile506Ser	1517T>G	I506S	Cystic fibrosis	Kobayashi (1990) <i>Am J Hum Genet</i> 47:611	PubMed 1977306
CFTR	CM950244	ATC-ACC	Ile506Thr	1517T>C	I506T	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM900047	ATC-GTC	Ile506Val	1516A>G	I506V	Cystic fibrosis	Mittre (2002) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM900048	TTT-TGT	Phe508Cys	1523T>G	F508C	Cystic fibrosis	Arduino (2002) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM983556	TTT-TCT	Phe508Ser	1523T>C	F508S	Cystic fibrosis	Arduino (1997) <i>CFTR LSDB entry Newsletter</i> :#70	Source: LSDB
CFTR	CM024677	TAT-CAT	Tyr515His	1543T>C	Y515H	Cystic fibrosis	Bienvenu (2005) <i>Hum Biol</i> 77:705	PubMed 16596947
CFTR	CM024678	AGA-GGA	Arg516Gly	1546A>G	R516G	Cystic fibrosis	Jones (1992) <i>Hum Mol Genet</i> 1:11	PubMed 1284466
CFTR	CM972948	TAC-TGC	Tyr517Cys	1550A>G	Y517C	Cystic fibrosis	Patrinos (2005) <i>CFTR LSDB entry</i> :	Source: LSDB
CFTR	CM941976	GTC-ATC	Val520Ile	1558G>A	V520I	Cystic fibrosis	Jones (1992) <i>Hum Mol Genet</i> 1:11	PubMed 1284466
CFTR	CM920158	GTC-TTC	Val520Phe	1558G>T	V520F	Cystic fibrosis	Shackleton (1994) <i>Hum Mutat</i> 3:141	PubMed 7515303
CFTR	CM051411	ATC-TTC	Ile521Phe	1561A>T	I521F	Cystic fibrosis	Byrne (1997) <i>CFTR LSDB entry Newsletter</i> :#70	Source: LSDB
CFTR	CM920159	TGC-TGA	Cys524Term	1572C>A	C524*	Cystic fibrosis	Brakensiek (2007) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM940256	CAA-TAA	Gln525Term	1573C>T	Q525*	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM972949	GAA-CAA	Glu527Gln	1579G>C	E527Q	Cystic fibrosis	Sharma (2009) <i>Ann Hum Genet</i> 73:26	PubMed 18782298
CFTR	CM074733	GAC-GGC	Asp529Gly	1586A>G	D529G	Cystic fibrosis	Laguna (2008) <i>J Cyst Fibros</i> 7:454	PubMed 18463004
CFTR	CM983557	GAC-CAC	Asp529His	1585G>C	D529H	Cystic fibrosis	Kambouris (2000) <i>Eur J Pediatr</i> 159:303	PubMed 10834512
CFTR	CM090070	ATC-CTC	Ile530Leu	1588A>C	I530L	Cystic fibrosis	Audrezet (1993) <i>Hum Mol Genet</i> 2:51	PubMed 7683952
CFTR	CM086646	TCC-CCC	Ser531Pro	1591T>C	S531P	Cystic fibrosis	Chomel (1995) <i>CFTR LSDB entry Newsletter</i> :#66	Source: LSDB
CFTR	CM001650	TTT-CTT	Phe533Leu	1597T>C	F533L	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM930112	GCA-GAA	Ala534Glu	1601C>A	A534E	Cystic fibrosis	Ferec (1994) <i>CFTR LSDB entry Newsletter</i> :#61	Source: LSDB
CFTR	CM952051	ATA-ACA	Ile539Thr	1616T>C	I539T	Cystic fibrosis		
CFTR	CM900049	GGA-TGA	Gly542Term	1624G>T	G542*	Cystic fibrosis		
CFTR	CM941977	GGT-AGT	Gly544Ser	1630G>A	G544S	Cystic fibrosis		

CFTR	CM900052	AGT-AGG	Ser549Arg	1647T>G	S549R	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM910071	AGT-CGT	Ser549Arg	1645A>C	S549R	Cystic fibrosis	Sanguuolo (1991) <i>Genomics</i> 9:788	PubMed 1903761
CFTR	CM900050	AGT-AAT	Ser549Asn	1646G>A	S549N	Cystic fibrosis	Cutting (1990) <i>Nature</i> 346:366	PubMed 1695717
CFTR	CM900051	AGT-ATT	Ser549Ile	1646G>T	S549I	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM952052	GGA-AGA	Gly550Arg	1648G>A	G550R	Cystic fibrosis	Ferec (1995) <i>CFTR LSDB entry Newsletter</i> :#66	Source: LSDB
CFTR	CM920984	GGA-TGA	Gly550Term	1648G>T	G550*	Cystic fibrosis	Deiman (1992) <i>CFTR LSDB entry Newsletter</i> :#43	Source: LSDB
CFTR	CM900053	GGT-GAT	Gly551Asp	1652G>A	G551D	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM920160	GGT-AGT	Gly551Ser	1651G>A	G551S	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM962464	CAA-AAA	Gln552Lys	1654C>A	Q552K	Cystic fibrosis	Faucz (1996) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM910072	CAA-TAA	Gln552Term	1654C>T	Q552*	Cystic fibrosis	Devoto (1991) <i>Am J Hum Genet</i> 48:1127	PubMed 1709778
CFTR	CM920161	CGA-CAA	Arg553Gln	1658G>A	R553Q	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM931148	CGA-GGA	Arg553Gly	1657C>G	R553G	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:375	PubMed 16049310
CFTR	CM900054	CGA-TGA	Arg553Term	1657C>T	R553*	Cystic fibrosis	Cutting (1990) <i>Nature</i> 346:366	PubMed 1695717
CFTR	CM930113	GCA-GAA	Ala554Glu	1661C>A	A554E	Cystic fibrosis	Audrezet (1993) <i>Hum Mol Genet</i> 2:51	PubMed 7683952
CFTR	CM993859	AGA-GGA	Arg555Gly	1663A>G	R555G	Cystic fibrosis	Zielenski (1999) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM920162	TTA-TCA	Leu558Ser	1673T>C	L558S	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM994785	GCA-GAA	Ala559Glu	1676C>A	A559E	Cystic fibrosis	Girodon (1999) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM900055	GCA-ACA	Ala559Thr	1675G>A	A559T	Cystic fibrosis	Cutting (1990) <i>Nature</i> 346:366	PubMed 1695717
CFTR	CM067759	GCA-GTA	Ala559Val	1676C>T	A559V	Cystic fibrosis	Girodon (2006) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM024679	AGA-GGA	Arg560Gly	1678A>G	R560G	Cystic fibrosis	Alonso (2006) <i>Ann Hum Genet</i> 71:194	PubMed 17331079
CFTR	CM920163	AGA-AAA	Arg560Lys	1679G>A	R560K	Cystic fibrosis	Ferec (1992) <i>Nat Genet</i> 1:188	PubMed 1284639
CFTR	CM980338	AGA-AGC	Arg560Ser	1680A>C	R560S	Cystic fibrosis	Malone (1998) <i>Hum Mutat</i> 11:152	PubMed 9482579
CFTR	CM992306	AGA-AGT	Arg560Ser	1680A>T	R560S	Cystic fibrosis	Liechti (1999) <i>Eur J Hum Genet</i> 7:590	PubMed 10439967
CFTR	CM900056	AGA-ACA	Arg560Thr	1679G>C	R560T	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM970278	GCA-GAA	Ala561Glu	1682C>A	A561E	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM990362	GTA-ATA	Val562Ile	1684G>A	V562I	Cystic fibrosis	Telleria (1999) <i>Hum Mutat</i> 14:89	PubMed 10447267
CFTR	CM960279	GTA-CTA	Val562Leu	1684G>C	V562L	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM900057	TAC-AAC	Tyr563Asn	1687T>A	Y563N	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM970279	TAC-GAC	Tyr563Asp	1687T>G	Y563D	Cystic fibrosis	Macek (1997) <i>Am J Hum Genet</i> 60:1122	PubMed 9150159
CFTR	CM962465	TAC-TGC	Tyr563Cys	1688A>G	Y563C	Cystic fibrosis	Delhaize (1996) <i>CFTR LSDB entry Newsletter</i> :#67	Source: LSDB
CFTR	CM1212241	TAC-CAC	Tyr563His	1687T>C	Y563H	Cystic fibrosis	Křenková (2013) <i>J Cyst Fibros</i> 12:532	PubMed 23276700
CFTR	CM063900	TAC-TAA	Tyr563Term	1689C>A	Y563*	Cystic fibrosis	Elahi (2006) <i>J Mol Diagn</i> 8:119	PubMed 16436643
CFTR	CM078046	GCT-GAT	Ala566Asp	1697C>A	A566D	Cystic fibrosis	Alibakhshi (2008) <i>J Cyst Fibros</i> 7:102	PubMed 17662673
CFTR	CM034786	GCT-ACT	Ala566Thr	1696G>A	A566T	Cystic fibrosis	Wallace (2003) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM970280	TTG-TAG	Leu568Term	1703T>A	L568*	Cystic fibrosis	Macek (1997) <i>Hum Mutat</i> 9:136	PubMed 9067754
CFTR	CM980339	TAT-GAT	Tyr569Asp	1705T>G	Y569D	Cystic fibrosis	Malone (1998) <i>Hum Mutat</i> 11:152	PubMed 9482579
CFTR	CM960280	TAT-TGT	Tyr569Cys	1706A>G	Y569C	Cystic fibrosis	Petreska (1996) <i>Hum Mutat</i> 7:374	PubMed 8723693
CFTR	CM001651	TAT-CAT	Tyr569His	1705T>C	Y569H	Cystic fibrosis	Kabra (2000) <i>Am J Med Genet</i> 93:161	PubMed 10869121
CFTR	CM931149	TAT-TAA	Tyr569Term	1707T>A	Y569*	Cystic fibrosis	Ferec (1993) <i>CFTR LSDB entry Newsletter</i> :#54	Source: LSDB
CFTR	CM970282	TTA-TCA	Leu571Ser	1712T>C	L571S	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM950245	GAC-AAC	Asp572Asn	1714G>A	D572N	Cystic fibrosis	Verlingue (1995) <i>Hum Mutat</i> 5:205	PubMed 7541273
CFTR	CM900058	CCT-CAT	Pro574His	1721C>A	P574H	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM962466	GGA-TGA	Gly576Term	1726G>T	G576*	Cystic fibrosis	Girodon (1996) <i>CFTR LSDB entry Newsletter</i> :#67	Source: LSDB

CFTR	CM962467	TAC-TTC	Tyr577Phe	1730A>T	Y577F	Cystic fibrosis	Pagani (2003) <i>Hum Mol Genet</i> 12:1111	PubMed 12719375
CFTR	CM983558	GAT-GCT	Asp579Ala	1736A>C	D579A	Cystic fibrosis	Pacheco (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM950246	GAT-GGT	Asp579Gly	1736A>G	D579G	Cystic fibrosis	Brancolini (1995) <i>Hum Genet</i> 96:312	PubMed 7544319
CFTR	CM941978	GAT-TAT	Asp579Tyr	1735G>T	D579Y	Cystic fibrosis	Harris (1994) <i>CFTR LSDB entry</i> Newsletter:#63	Source: LSDB
CFTR	CM940257	ACA-AGA	Thr582Arg	1745C>G	T582R	Cystic fibrosis	Chillon (1994) <i>Hum Genet</i> 93:447	PubMed 7513293
CFTR	CM962468	ACA-ATA	Thr582Ile	1745C>T	T582I	Cystic fibrosis	des Georges (2004) <i>J Cyst Fibros</i> 3:265	PubMed 15698946
CFTR	CM920165	GAA-TAA	Glu585Term	1753G>T	E585*	Cystic fibrosis	Cremonesi (1992) <i>Hum Mutat</i> 1:314	PubMed 1284538
CFTR	CM067730	ATA-GTA	Ile586Val	1756A>G	I586V	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM022335	GAA-TAA	Glu588Term	1762G>T	E588*	Cystic fibrosis	Spitzer (2002) <i>Eur Respir J</i> 19:374	PubMed 11866018
CFTR	CM055924	GAA-GTA	Glu588Val	1763A>T	E588V	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM056941	AGC-AAC	Ser589Asn	1766G>A	S589N	Cystic fibrosis	Stanziale (2005) <i>Genet Test</i> 9:285	PubMed 16379540
CFTR	CM056942	AGC-ACC	Ser589Thr	1766G>C	S589T	Cystic fibrosis	Kinnunen (2005) <i>J Cyst Fibros</i> 4:233	PubMed 16051530
CFTR	CM074732	CTG-CCG	Leu594Pro	1781T>C	L594P	Cystic fibrosis	Omar (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM067739	ATG-ACG	Met595Thr	1784T>C	M595T	Cystic fibrosis	Pasquet (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM067741	AAA-TAA	Lys598Term	1792A>T	K598*	Cystic fibrosis	Seia (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM972950	AGG-GGG	Arg600Gly	1798A>G	R600G	Cystic fibrosis	Hubert (2004) <i>J Cyst Fibros</i> 3:15	PubMed 15463882
CFTR	CM980340	ATT-TTT	Ile601Phe	1801A>T	I601F	Cystic fibrosis	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM057326	ATT-ACT	Ile601Thr	1802T>C	I601T	Cystic fibrosis	Bienvenu (2005) <i>Hum Biol</i> 77:705	PubMed 16596947
CFTR	CM983559	GTC-TTC	Val603Phe	1807G>T	V603F	Cystic fibrosis	Strandvik (2001) <i>Genet Test</i> 5:235	PubMed 11788090
CFTR	CM055922	ACT-AGT	Thr604Ser	1811C>G	T604S	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM024233	GAA-GGA	Glu608Gly	1823A>G	E608G	Cystic fibrosis	Kilinc (2002) <i>Am J Med Genet</i> 113:250	PubMed 12439892
CFTR	CM972951	CAT-CGT	His609Arg	1826A>G	H609R	Cystic fibrosis	Padoan (2002) <i>Acta Paediatr</i> 91:82	PubMed 11883825
CFTR	CM067750	CAT-CTT	His609Leu	1826A>T	H609L	Cystic fibrosis	Petrocchi (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM980341	TTA-TCA	Leu610Ser	1829T>C	L610S	Cystic fibrosis	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM980342	GCT-ACT	Ala613Thr	1837G>A	A613T	Cystic fibrosis	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM930114	GAC-GGC	Asp614Gly	1841A>G	D614G	Cystic fibrosis	Audrezet (1993) <i>Hum Hered</i> 43:295	PubMed 8406518
CFTR	CM994787	GAC-TAC	Asp614Tyr	1840G>T	D614Y	Cystic fibrosis	Girodon (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM970283	ATT-ACT	Ile618Thr	1853T>C	I618T	Cystic fibrosis	Macek (1997) <i>Am J Hum Genet</i> 60:1122	PubMed 9150159
CFTR	CM940258	TTG-TCG	Leu619Ser	1856T>C	L619S	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM980344	CAT-CAG	His620Gln	1860T>G	H620Q	Cystic fibrosis	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM980343	CAT-CCT	His620Pro	1859A>C	H620P	Cystic fibrosis	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM980345	GGT-GAT	Gly622Asp	1865G>A	G622D	Cystic fibrosis	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM920166	GGG-AGG	Gly628Arg	1882G>A	G628R	Cystic fibrosis	Fanen (1992) <i>Genomics</i> 13:770	PubMed 1379210
CFTR	CM931150	GGG-CGG	Gly628Arg	1882G>C	G628R	Cystic fibrosis	Cuppens (1993) <i>Genomics</i> 18:693	PubMed 7508414
CFTR	CM067733	CTC-ATC	Leu633Ile	1897C>A	L633I	Cystic fibrosis	Pacheco (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM980346	CTC-CCC	Leu633Pro	1898T>C	L633P	Cystic fibrosis	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
							Michel-Calemard (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM962764	CAA-TAA	Gln634Term	1900C>T	Q634*	Cystic fibrosis	Bombieri (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM983560	CTA-CCA	Leu636Pro	1907T>C	L636P	Cystic fibrosis	Hirtz (2004) <i>Gastroenterology</i> 127:1085	PubMed 15480987
CFTR	CM993860	CAG-TAG	Gln637Term	1909C>T	Q637*	Cystic fibrosis		
CFTR	CM024681	GAC-TAC	Asp639Tyr	1915G>T	D639Y	Cystic fibrosis	Aulehla-Scholz (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM940259	GAT-GTT	Asp648Val	1943A>T	D648V	Cystic fibrosis	Mercier (1994) <i>Hum Genet</i> 94:629	PubMed 7527370
CFTR	CM983561	GAC-AAC	Asp651Asn	1951G>A	D651N	Cystic fibrosis	Bombieri (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB

CFTR	CM972952	GAA-TAA	Glu656Term	1966G>T	E656*	Cystic fibrosis	Aznarez (2003) <i>Hum Mol Genet</i> 12:2031	PubMed 12913074
CFTR	CM067772	TCA-ACA	Ser660Thr	1978T>A	S660T	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM970284	GAG-TAG	Glu664Term	1990G>T	E664*	Cystic fibrosis	Clavel (1997) <i>Hum Mutat</i> 9:368	PubMed 9101301
CFTR	CM960281	ACC-TCC	Thr665Ser	1993A>T	T665S	Cystic fibrosis	Messaoud (1996) <i>Eur J Hum Genet</i> 4:20	PubMed 8800923
CFTR	CM950247	CGT-TGT	Arg668Cys	2002C>T	R668C	Cystic fibrosis	Chillon (1995) <i>N Engl J Med</i> 332:1475	PubMed 7739684
CFTR	CM940260	GGA-TGA	Gly673Term	2017G>T	G673*	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM993861	TGG-TAG	Trp679Term	2036G>A	W679*	Cystic fibrosis	Walker (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM003248	AAA-AGA	Lys683Arg	2048A>G	K683R	Cystic fibrosis	Chevalier-Porst (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM940261	CAA-TAA	Gln685Term	2053C>T	Q685*	Cystic fibrosis	Audrezet (1994) <i>Hum Genet</i> 93:659	PubMed 7516305
CFTR	CM067765	AAA-TAA	Lys688Term	2062A>T	K688*	Cystic fibrosis	Gimenez (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM034787	CAG-TAG	Gln689Term	2065C>T	Q689*	Cystic fibrosis	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM970285	GAG-TAG	Glu692Term	2074G>T	E692*	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM930115	TTT-CTT	Phe693Leu	2077T>C	F693L	Cystic fibrosis	Audrezet (1993) <i>Hum Hered</i> 43:295	PubMed 8406518
CFTR	CM116799	AAA-AGA	Lys696Arg	2087A>G	K696R	Cystic fibrosis	Fresquet (2011) <i>J Mol Diagn</i> 13:520	PubMed 21708286
CFTR	CM972953	AAG-AGG	Lys698Arg	2093A>G	K698R	Cystic fibrosis	Ferec (1997) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM024683	CGA-CAA	Arg709Gln	2126G>A	R709Q	Cystic fibrosis	Wagner (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM950248	CGA-TGA	Arg709Term	2125C>T	R709*	Cystic fibrosis	Bonizzato (1995) <i>Hum Genet</i> 95:397	PubMed 7535742
CFTR	CM920167	AAA-TAA	Lys710Term	2128A>T	K710*	Cystic fibrosis	Fanen (1992) <i>Genomics</i> 13:770	PubMed 1379210
CFTR	CM024684	TCC-TGC	Ser712Cys	2135C>G	S712C	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM930116	AAG-TAG	Lys716Term	2146A>T	K716*	Cystic fibrosis	Audrezet (1993) <i>Hum Mol Genet</i> 2:51	PubMed 7683952
CFTR	CM940262	TTA-TAA	Leu719Term	2156T>A	L719*	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM993862	CAA-TAA	Gln720Term	2158C>T	Q720*	Cystic fibrosis	Malone (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983562	GAA-AAA	Glu725Lys	2173G>A	E725K	Cystic fibrosis	des Georges (2004) <i>J Cyst Fibros</i> 3:265	PubMed 15698946
CFTR	CM930117	GAG-TAG	Glu730Term	2188G>T	E730*	Cystic fibrosis	Cuppens (1993) <i>Genomics</i> 18:693	PubMed 7508414
CFTR	CM970286	TTA-TGA	Leu732Term	2195T>G	L732*	Cystic fibrosis	Tzetis (1997) <i>Hum Genet</i> 99:121	PubMed 9003508
CFTR	CM015357	AGG-AAG	Arg735Lys	2204G>A	R735K	Cystic fibrosis	Frentescu (2008) <i>J Cyst Fibros</i> 7:423	PubMed 18467194
CFTR	CM057581	GGA-TGA	Gly745Term	2233G>T	G745*	Cystic fibrosis	Bienvenu (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM000667	CCT-CTT	Pro750Leu	2249C>T	P750L	Cystic fibrosis	Orozco (2000) <i>Hum Genet</i> 106:360	PubMed 10798368
CFTR	CM024686	CGC-CCC	Arg751Pro	2252G>C	R751P	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM000668	GTG-ATG	Val754Met	2260G>A	V754M	Cystic fibrosis	Orozco (2000) <i>Hum Genet</i> 106:360	PubMed 10798368
CFTR	CM993863	ACG-ATG	Thr760Met	2279C>T	T760M	Cystic fibrosis	Zielenski (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM960282	CGA-TGA	Arg764Term	2290C>T	R764*	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM972954	TCA-TGA	Ser776Term	2327C>G	S776*	Cystic fibrosis	des Georges (2004) <i>J Cyst Fibros</i> 3:265	PubMed 15698946
CFTR	CM972955	CAG-TAG	Gln781Term	2341C>T	Q781*	Cystic fibrosis	Zhou (1997) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM023615	AAC-AAA	Asn782Lys	2346C>A	N782K	Cystic fibrosis	Girodon (2002) <i>J Hepatol</i> 37:192	PubMed 12127423
CFTR	CM941979	CGA-TGA	Arg785Term	2353C>T	R785*	Cystic fibrosis	Frentescu (2008) <i>J Cyst Fibros</i> 7:423	PubMed 18467194
CFTR	CM067731	ACA-ATA	Thr788Ile	2363C>T	T788I	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM930118	CGA-TGA	Arg792Term	2374C>T	R792*	Cystic fibrosis	Claustres (1993) <i>Hum Mol Genet</i> 2:1209	PubMed 7691344
CFTR	CM950249	GCA-GGA	Ala800Gly	2399C>G	A800G	Cystic fibrosis	Mercier (1995) <i>Am J Hum Genet</i> 56:272	PubMed 7529962
CFTR	CM072949	GAT-GGT	Asp806Gly	2417A>G	D806G	Cystic fibrosis	Narzi (2007) <i>Clin Genet</i> 72:39	PubMed 17594398
CFTR	CM067767	ATA-GTA	Ile807Val	2419A>G	I807V	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM024687	TCT-CCT	Ser813Pro	2437T>C	S813P	Cystic fibrosis	Tzetis (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM035676	CAA-TAA	Gln814Term	2440C>T	Q814*	Cystic fibrosis	Ahmed (2003) <i>Gut</i> 52:1159	PubMed 12865275

CFTR	CM930119	GAA-AAA	Glu822Lys	2464G>A	E822K	Cystic fibrosis	Mercier (1993) <i>Hum Mutat</i> 2:16	PubMed 8477260
CFTR	CM970287	GAA-TAA	Glu822Term	2464G>T	E822*	Cystic fibrosis	Tzetis (1997) <i>Hum Genet</i> 99:121	PubMed 9003508
CFTR	CM983563	GAA-TAA	Glu823Term	2467G>T	E823*	Cystic fibrosis	Tzetis (1998) <i>CFTR LSDB entry Newsletter</i> :#70	Source: LSDB
CFTR	CM980350	GAA-AAA	Glu826Lys	2476G>A	E826K	Cystic fibrosis	Vankeerberghen (1998) <i>Hum Mol Genet</i> 7:1761	PubMed 9736778
CFTR	CM920168	GAA-TAA	Glu827Term	2479G>T	E827*	Cystic fibrosis	Ferec (1992) <i>Nat Genet</i> 1:188	PubMed 1284639
CFTR	CM024688	GAC-GGC	Asp828Gly	2483A>G	D828G	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM930120	AAG-TAG	Lys830Term	2488A>T	K830*	Cystic fibrosis	Mercier (1993) <i>Hum Mutat</i> 2:16	PubMed 8477260
CFTR	CM960283	GAG-TAG	Glu831Term	2491G>T	E831*	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM920986	GAT-TAT	Asp836Tyr	2506G>T	D836Y	Cystic fibrosis	des Georges (2004) <i>J Cyst Fibros</i> 3:265	PubMed 15698946
CFTR	CM067736	ATA-ACA	Ile840Thr	2519T>C	I840T	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM930121	TGG-TAG	Trp846Term	2537G>A	W846*	Cystic fibrosis	Cheadle (1993) <i>Hum Mol Genet</i> 2:1067	PubMed 7689897
CFTR	CM900059	TGG-TGA	Trp846Term	2538G>A	W846*	Cystic fibrosis	Vidaud (1990) <i>Hum Genet</i> 85:446	PubMed 2210768
CFTR	CM994348	TAC-TAA	Tyr849Term	2547C>A	Y849*	Cystic fibrosis	Castaldo (1999) <i>Hum Mutat</i> 14:272	PubMed 10477439
CFTR	CM970288	CGA-CTA	Arg851Leu	2552G>T	R851L	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM910073	CGA-TGA	Arg851Term	2551C>T	R851*	Cystic fibrosis	White (1991) <i>Genomics</i> 11:778	PubMed 1723056
CFTR	CM088455	TAT-TAG	Tyr852Term	2556T>G	Y852*	Cystic fibrosis	Bulman (2008) <i>CFTR LSDB entry Unpublished</i> ..	Source: LSDB
CFTR	CM094117	GTC-ATC	Val855Ile	2563G>A	V855I	Cystic fibrosis	Seia (2009) <i>Clin Biochem</i> 42:611	PubMed 19318035
CFTR	CM920169	TGC-TAC	Cys866Tyr	2597G>A	C866Y	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM962469	TTA-TAA	Leu867Term	2600T>A	L867*	Cystic fibrosis	Haworth (1996) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM962470	TGG-TAG	Trp882Term	2645G>A	W882*	Cystic fibrosis	Ferec (1996) <i>CFTR LSDB entry Newsletter</i> :#67	Source: LSDB
CFTR	CM983564	CAA-CGA	Gln890Arg	2669A>G	Q890R	Cystic fibrosis	Casals (2000) <i>Hum Reprod</i> 15:1476	PubMed 10875853
CFTR	CM920170	CAA-TAA	Gln890Term	2668C>T	Q890*	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM983565	GAC-GGC	Asp891Gly	2672A>G	D891G	Cystic fibrosis	Kilinc (1998) <i>CFTR LSDB entry Newsletter</i> :#70	Source: LSDB
CFTR	CM004880	AGT-AAT	Ser895Asn	2684G>A	S895N	Cystic fibrosis	Wu (2000) <i>J Formos Med Assoc</i> 99:564	PubMed 10925568
CFTR	CM003249	ACT-ATT	Thr896Ile	2687C>T	T896I	Cystic fibrosis	Lazaro (2000) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM004462	ACC-AAC	Thr908Asn	2723C>A	T908N	Cystic fibrosis	Hammerle (2000) <i>Glycoconj J</i> 17:807	PubMed 11443282
CFTR	CM940263	TCG-TTG	Ser912Leu	2735C>T	S912L	Cystic fibrosis	Ghanem (1994) <i>Genomics</i> 21:434	PubMed 7522211
CFTR	CM930122	TCG-TAG	Ser912Term	2735C>A	S912*	Cystic fibrosis	Saba (1993) <i>Hum Mol Genet</i> 2:1739	PubMed 7505693
CFTR	CM900060	TAT-TGT	Tyr913Cys	2738A>G	Y913C	Cystic fibrosis	Vidaud (1990) <i>Hum Genet</i> 85:446	PubMed 2210768
CFTR	CM055926	TAT-TAA	Tyr913Term	2739T>A	Y913*	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM063901	TAT-TAA	Tyr914Term	2742T>A	Y914*	Cystic fibrosis	Kammesheidt (2006) <i>Genet Med</i> 8:557	PubMed 16980811
CFTR	CM962471	TAC-GAC	Tyr917Asp	2749T>G	Y917D	Cystic fibrosis	Schwarz (1996) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM960284	TAC-TGC	Tyr917Cys	2750A>G	Y917C	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM940264	TAC-TGC	Tyr919Cys	2756A>G	Y919C	Cystic fibrosis	Savov (1994) <i>Hum Mol Genet</i> 3:57	PubMed 7512860
CFTR	CM980351	GTG-ATG	Val920Met	2758G>A	V920M	Cystic fibrosis	Steffann (1998) <i>Ann Genet</i> 41:213	PubMed 9881185
CFTR	CM074093	GAC-AAC	Asp924Asn	2770G>A	D924N	Cystic fibrosis	Alonso (2006) <i>Ann Hum Genet</i> 71:194	PubMed 17331079
CFTR	CM940265	CTT-CCT	Leu927Pro	2780T>C	L927P	Cystic fibrosis	Hermans (1994) <i>Hum Mol Genet</i> 3:1199	PubMed 7526927
CFTR	CM993864	TTC-TCC	Phe932Ser	2795T>C	F932S	Cystic fibrosis	Ferec (1999) <i>CFTR LSDB entry Pers comm</i> :	Source: LSDB
CFTR	CM962472	CAT-CGT	His939Arg	2816A>G	H939R	Cystic fibrosis	Ferec (1996) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM931151	CAT-GAT	His939Asp	2815C>G	H939D	Cystic fibrosis	Ferec (1993) <i>CFTR LSDB entry Newsletter</i> :#54	Source: LSDB
CFTR	CM930123	TCG-TTG	Ser945Leu	2834C>T	S945L	Cystic fibrosis	Claustres (1993) <i>Hum Mol Genet</i> 2:1209	PubMed 7691344
CFTR	CM962473	AAA-TAA	Lys946Term	2836A>T	K946*	Cystic fibrosis	Haworth (1996) <i>CFTR LSDB entry Newsletter</i> :#69	Source: LSDB
CFTR	CM952053	CAC-CGC	His949Arg	2846A>G	H949R	Cystic fibrosis	Ferec (1995) <i>CFTR LSDB entry Newsletter</i> :#65	Source: LSDB

CFTR	CM055124	CAC-CTC	His949Leu	2846A>T	H949L	Cystic fibrosis	McGinniss (2005) <i>Hum Genet</i> 118:331	PubMed 16189704
CFTR	CM940266	CAC-TAC	His949Tyr	2845C>T	H949Y	Cystic fibrosis	Ghanem (1994) <i>Genomics</i> 21:434	PubMed 7522211
CFTR	CM980352	ATG-ATC	Met952Ile	2856G>C	M952I	Cystic fibrosis	Desgeorges (1997) <i>Hum Genet</i> 100:279	PubMed 9254864
CFTR	CM993865	ATG-ACG	Met952Thr	2855T>C	M952T	Cystic fibrosis	Casals (2000) <i>Hum Reprod</i> 15:1476	PubMed 10875853
CFTR	CM087537	CAT-CCT	His954Pro	2861A>C	H954P	Cystic fibrosis	Kolesár (2008) <i>Gen Physiol Biophys</i> 27:299	PubMed 19202204
CFTR	CM003250	ATG-ATT	Met961Ile	2883G>T	M961I	Cystic fibrosis	Malone (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM930124	GGT-CGT	Gly970Arg	2908G>C	G970R	Cystic fibrosis	Cuppens (1993) <i>Genomics</i> 18:693	PubMed 7508414
CFTR	CM990363	GGT-GAT	Gly970Asp	2909G>A	G970D	Cystic fibrosis	Wagner (1999) <i>Hum Genet</i> 104:511	PubMed 10453741
CFTR	CM067735	GGT-AGT	Gly970Ser	2908G>A	G970S	Cystic fibrosis	Seia (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM983566	CTT-CCT	Leu973Pro	2918T>C	L973P	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM972956	TCC-TTC	Ser977Phe	2930C>T	S977F	Cystic fibrosis	Ferec (1997) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM960285	TCC-CCC	Ser977Pro	2929T>C	S977P	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM970292	GAT-GTT	Asp979Val	2936A>T	D979V	Cystic fibrosis	Plouvier (1997) <i>Ann Genet</i> 40:185	PubMed 9401110
CFTR	CM960286	ATA-AAA	Ile980Lys	2939T>A	I980K	Cystic fibrosis	Bienvenu (1996) <i>Hum Mutat</i> 7:182	PubMed 8829643
CFTR	CM983567	GAC-CAC	Asp985His	2953G>C	D985H	Cystic fibrosis	Claustres (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM941980	GAC-TAC	Asp985Tyr	2953G>T	D985Y	Cystic fibrosis	Bienvenu (1994) <i>CFTR LSDB entry</i> Newsletter:#63	Source: LSDB
CFTR	CM962474	GAC-TAC	Asp993Tyr	2977G>T	D993Y	Cystic fibrosis	des Georges (2004) <i>J Cyst Fibros</i> 3:265	PubMed 15698946
CFTR	CM983568	TTC-TGC	Phe994Cys	2981T>G	F994C	Cystic fibrosis	Claustres (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM003251	GGA-TGA	Gly1003Term	3007G>T	G1003*	Cystic fibrosis	Ferec (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM940267	ATA-AGA	Ile1005Arg	3014T>G	I1005R	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM950250	GCA-GAA	Ala1006Glu	3017C>A	A1006E	Cystic fibrosis	Ferec (1995) <i>Mol Cell Probes</i> 9:135	PubMed 7541510
CFTR	CM983569	GTC-GAC	Val1008Asp	3023T>A	V1008D	Cystic fibrosis	Alonso (2006) <i>Ann Hum Genet</i> 71:194	PubMed 17331079
CFTR	CM003252	GCA-ACA	Ala1009Thr	3025G>A	A1009T	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM980353	CCC-CTC	Pro1013Leu	3038C>T	P1013L	Cystic fibrosis	Onay (1998) <i>Hum Genet</i> 102:224	PubMed 9521595
CFTR	CM983570	TAC-TGC	Tyr1014Cys	3041A>G	Y1014C	Cystic fibrosis	Casals (2000) <i>Hum Reprod</i> 15:1476	PubMed 10875853
CFTR	CM042689	TTT-TCT	Phe1016Ser	3047T>C	F1016S	Cystic fibrosis	Alper (2004) <i>Hum Mutat</i> 24:353	PubMed 15365999
CFTR	CM067764	GTG-GAG	Val1020Glu	3059T>A	V1020E	Cystic fibrosis	Girodon (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM024690	CCA-GCA	Pro1021Ala	3061C>G	P1021A	Cystic fibrosis	Tzetic (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM960287	ATT-ACT	Ile1027Thr	3080T>C	I1027T	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM003253	ATG-AGG	Met1028Arg	3083T>G	M1028R	Cystic fibrosis	Lazaro (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM980354	ATG-ATT	Met1028Ile	3084G>T	M1028I	Cystic fibrosis	Onay (1998) <i>Hum Genet</i> 102:224	PubMed 9521595
CFTR	CM043471	TAT-AAT	Tyr1032Asn	3094T>A	Y1032N	Cystic fibrosis	Seia (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM043472	CAA-TAA	Gln1035Term	3103C>T	Q1035*	Cystic fibrosis	Bogdanova (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM055121	ACC-AAC	Thr1036Asn	3107C>A	T1036N	Cystic fibrosis	McGinniss (2005) <i>Hum Genet</i> 118:331	PubMed 16189704
CFTR	CM078047	ACC-ATC	Thr1036Ile	3107C>T	T1036I	Cystic fibrosis	Alibakhshi (2008) <i>J Cyst Fibros</i> 7:102	PubMed 17662673
CFTR	CM983571	CAA-TAA	Gln1042Term	3124C>T	Q1042*	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM056563	CTG-CGG	Leu1043Arg	3128T>G	L1043R	Cystic fibrosis	Messaoud (2005) <i>Ann Biol Clin (Paris)</i> 63:627	PubMed 16330381
CFTR	CM087272	TCT-TAT	Ser1045Tyr	3134C>A	S1045Y	Cystic fibrosis	Rana-Díez (2008) <i>J Cyst Fibros</i> 7:520	PubMed 18676185
CFTR	CM1212242	GAA-TAA	Glu1046Term	3136G>T	E1046*	Cystic fibrosis	Křenková (2013) <i>J Cyst Fibros</i> 12:532	PubMed 23276700
CFTR	CM024691	GGC-CGC	Gly1047Arg	3139G>C	G1047R	Cystic fibrosis	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067746	AGG-GGG	Arg1048Gly	3142A>G	R1048G	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM043473	ATT-GTT	Ile1051Val	3151A>G	I1051V	Cystic fibrosis	Pasquet (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM930125	TTC-GTC	Phe1052Val	3154T>G	F1052V	Cystic fibrosis	Mercier (1993) <i>Genomics</i> 16:296	PubMed 7683628

CFTR	CM983572	ACT-ATT	Thr1053Ile	3158C>T	T1053I	Cystic fibrosis	Bienvenu (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067749	CAT-CGT	His1054Arg	3161A>G	H1054R	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM940268	CAT-GAT	His1054Asp	3160C>G	H1054D	Cystic fibrosis	Mercier (1994) <i>J Med Genet</i> 31:731	PubMed 7529319
CFTR	CM067751	CAT-CTT	His1054Leu	3161A>T	H1054L	Cystic fibrosis	Pacheco (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM962475	ACA-GCA	Thr1057Ala	3169A>G	T1057A	Cystic fibrosis	Dal'Maso (2013) <i>J Bras Pneumol</i> 39:181	PubMed 23670503
CFTR	CM940269	TTA-TGA	Leu1059Term	3176T>G	L1059*	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM950251	AAA-ACA	Lys1060Thr	3179A>C	K1060T	Cystic fibrosis	Casals (1995) <i>Hum Genet</i> 95:205	PubMed 7532150
CFTR	CM960288	GGA-AGA	Gly1061Arg	3181G>A	G1061R	Cystic fibrosis	Bienvenu (1996) <i>Hum Mutat</i> 7:376	PubMed 8723695
CFTR	CM930126	GGA-CGA	Gly1061Arg	3181G>C	G1061R	Cystic fibrosis	Mercier (1993) <i>Genomics</i> 16:296	PubMed 7683628
CFTR	CM920172	TGG-TGA	Trp1063Term	3189G>A	W1063*	Cystic fibrosis	Fanen (1992) <i>Genomics</i> 13:770	PubMed 1379210
CFTR	CM980355	CTT-CGT	Leu1065Arg	3194T>G	L1065R	Cystic fibrosis	Casals (1998) <i>Hum Mutat</i> S1:S99	PubMed 9452054
CFTR	CM984227	CTT-TTT	Leu1065Phe	3193C>T	L1065F	Cystic fibrosis	Tzetzis (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM920173	CTT-CCT	Leu1065Pro	3194T>C	L1065P	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM920175	CGT-TGT	Arg1066Cys	3196C>T	R1066C	Cystic fibrosis	Fanen (1992) <i>Genomics</i> 13:770	PubMed 1379210
CFTR	CM1212243	CGT-GGT	Arg1066Gly	3196C>G	R1066G	Cystic fibrosis	Křenková (2013) <i>J Cyst Fibros</i> 12:532	PubMed 23276700
CFTR	CM920174	CGT-CAT	Arg1066His	3197G>A	R1066H	Cystic fibrosis	Ferec (1992) <i>Nat Genet</i> 1:188	PubMed 1284639
CFTR	CM930127	CGT-CTT	Arg1066Leu	3197G>T	R1066L	Cystic fibrosis	Mercier (1993) <i>Genomics</i> 16:296	PubMed 7683628
CFTR	CM952054	CGT-AGT	Arg1066Ser	3196C>A	R1066S	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:375	PubMed 16049310
CFTR	CM994788	GCC-GAC	Ala1067Asp	3200C>A	A1067D	Cystic fibrosis	Girodon (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM920176	GCC-ACC	Ala1067Thr	3199G>A	A1067T	Cystic fibrosis	Ferec (1992) <i>Nat Genet</i> 1:188	PubMed 1284639
CFTR	CM950252	GCC-GTC	Ala1067Val	3200C>T	A1067V	Cystic fibrosis	Jezequel (1995) <i>Clin Chem</i> 41:833	PubMed 7539342
CFTR	CM940270	GGA-AGA	Gly1069Arg	3205G>A	G1069R	Cystic fibrosis	Savov (1994) <i>Hum Mol Genet</i> 3:57	PubMed 7512860
CFTR	CM930128	CGG-CAG	Arg1070Gln	3209G>A	R1070Q	Cystic fibrosis	Mercier (1993) <i>Genomics</i> 16:296	PubMed 7683628
CFTR	CM970294	CGG-CCG	Arg1070Pro	3209G>C	R1070P	Cystic fibrosis	Shrimpton (1997) <i>Hum Mutat</i> 10:436	PubMed 9401006
CFTR	CM950253	CGG-TGG	Arg1070Trp	3208C>T	R1070W	Cystic fibrosis	Jezequel (1995) <i>Clin Chem</i> 41:833	PubMed 7539342
CFTR	CM003254	CAG-CAT	Gln1071His	3213G>T	Q1071H	Cystic fibrosis	Claustres (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM940271	CAG-CCG	Gln1071Pro	3212A>C	Q1071P	Cystic fibrosis	Ghanem (1994) <i>Genomics</i> 21:434	PubMed 7522211
CFTR	CM983573	CCT-CTT	Pro1072Leu	3215C>T	P1072L	Cystic fibrosis	Bombieri (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM970295	TTT-TTA	Phe1074Leu	3222T>A	F1074L	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM920177	CTG-CCG	Leu1077Pro	3230T>C	L1077P	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM057573	CAC-CCC	His1079Pro	3236A>C	H1079P	Cystic fibrosis	Wang (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM930129	CAT-CGT	His1085Arg	3254A>G	H1085R	Cystic fibrosis	Mercier (1993) <i>Genomics</i> 16:296	PubMed 7683628
CFTR	CM034792	ACT-GCT	Thr1086Ala	3256A>G	T1086A	Cystic fibrosis	Girodon (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM962476	ACT-ATT	Thr1086Ile	3257C>T	T1086I	Cystic fibrosis	Bienvenu (1996) <i>CFTR LSDB entry</i> Newsletter:#67	Source: LSDB
CFTR	CM983574	AAC-GAC	Asn1088Asp	3262A>G	N1088D	Cystic fibrosis	Strandvik (2001) <i>Genet Test</i> 5:235	PubMed 11788090
CFTR	CM950254	TGG-CGG	Trp1089Arg	3265T>C	W1089R	Cystic fibrosis	Zielenski (1995) <i>Hum Mutat</i> 5:43	PubMed 7537150
CFTR	CM920178	TGG-TAG	Trp1089Term	3266G>A	W1089*	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	HM030019	TAC-TGC	Tyr1092Cys	3275A>G	Y1092C	Cystic fibrosis	Trujillo-Tiebas (2004) <i>Hum Genet</i> 114:403	Source: Online submission
CFTR	CM972958	TAC-CAC	Tyr1092His	3274T>C	Y1092H	Cystic fibrosis	Trujillano (2013) <i>J Med Genet</i> 50:455	PubMed 23687349
CFTR	CM920179	TAC-TAA	Tyr1092Term	3276C>A	Y1092*	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM941981	TAC-TAG	Tyr1092Term	3276C>G	Y1092*	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:375	PubMed 16049310
CFTR	CM993438	CTG-CCG	Leu1093Pro	3278T>C	L1093P	Cystic fibrosis	Yee (1999) <i>Hum Mutat</i> 15:208	PubMed 10649505
CFTR	CM983575	CTG-CGG	Leu1096Arg	3287T>G	L1096R	Cystic fibrosis	Claustres (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB

CFTR	CM056307	TGG-CGG	Trp1098Arg	3292T>C	W1098R	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:375	PubMed 16049310
CFTR	CM000669	TGG-TGT	Trp1098Cys	3294G>T	W1098C	Cystic fibrosis	Orozco (2000) <i>Hum Genet</i> 106:360	PubMed 10798368
CFTR	CM074737	TGG-TTG	Trp1098Leu	3293G>T	W1098L	Cystic fibrosis	Brakensiek (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM940272	TGG-TAG	Trp1098Term	3293G>A	W1098*	Cystic fibrosis	Chillon (1994) <i>Hum Genet</i> 93:447 Michel-Calemard (1996) <i>CFTR LSDB entry</i>	PubMed 7513293
CFTR	CM962477	TGG-TGA	Trp1098Term	3294G>A	W1098*	Cystic fibrosis	Newsletter:#69	Source: LSDB
CFTR	CM055127	TTC-TTA	Phe1099Leu	3297C>A	F1099L	Cystic fibrosis	McGinniss (2005) <i>Hum Genet</i> 118:331	PubMed 16189704
CFTR	CM940273	CAA-CCA	Gln1100Pro	3299A>C	Q1100P	Cystic fibrosis	Chillon (1994) <i>Hum Genet</i> 93:447	PubMed 7513293
CFTR	CM930131	ATG-AGG	Met1101Arg	3302T>G	M1101R	Cystic fibrosis	Mercier (1993) <i>Genomics</i> 16:296	PubMed 7683628
CFTR	CM930130	ATG-AAG	Met1101Lys	3302T>A	M1101K	Cystic fibrosis	Zielenski (1993) <i>Am J Hum Genet</i> 52:609 Sobczynska-Tomaszewska (2013) <i>Eur J Hum Genet</i>	PubMed 7680525
CFTR	CM043474	AGA-TGA	Arg1102Term	3304A>T	R1102*	Cystic fibrosis	21:391	PubMed 22892530
CFTR	CM1212244	GAA-AAA	Glu1104Lys	3310G>A	E1104K	Cystic fibrosis	Křenková (2013) <i>J Cyst Fibros</i> 12:532	PubMed 23276700
CFTR	CM950255	GAA-TAA	Glu1104Term	3310G>T	E1104*	Cystic fibrosis	Zielenski (1995) <i>Hum Mutat</i> 5:43	PubMed 7537150
CFTR	CM031656	ATG-AGG	Met1105Arg	3314T>G	M1105R	Cystic fibrosis	Scotet (2003) <i>Hum Mutat</i> 22:105	PubMed 12815607
CFTR	CM136673	TTT-CTT	Phe1107Leu	3319T>C	F1107L	Cystic fibrosis	Prach (2013) <i>J Mol Diagn</i> epub:epub	PubMed 23810505
CFTR	CM993866	TCC-TGC	Ser1118Cys	3353C>G	S1118C	Cystic fibrosis	Zielenski (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983576	TCC-TTC	Ser1118Phe	3353C>T	S1118F	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM960289	GGA-CGA	Gly1123Arg	3367G>C	G1123R	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM941982	GGA-GAA	Gly1127Glu	3380G>A	G1127E	Cystic fibrosis	Bienvenu (1994) <i>CFTR LSDB entry</i> Newsletter:#63	Source: LSDB
CFTR	CM962478	AGA-TGA	Arg1128Term	3382A>T	R1128*	Cystic fibrosis	Ghanem (1996) <i>CFTR LSDB entry</i> Newsletter:#68	Source: LSDB
CFTR	CM024693	GTT-GGT	Val1129Gly	3386T>G	V1129G	Cystic fibrosis	Claustres (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993867	ATT-GTT	Ile1131Val	3391A>G	I1131V	Cystic fibrosis	Giorgi (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM003255	GCC-ACC	Ala1136Thr	3406G>A	A1136T	Cystic fibrosis	Ferec (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM952055	ATG-AGG	Met1137Arg	3410T>G	M1137R	Cystic fibrosis	Hirtz (2004) <i>Gastroenterology</i> 127:1085	PubMed 15480987
CFTR	CM067740	ATG-ACG	Met1137Thr	3410T>C	M1137T	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM931152	ATG-GTG	Met1137Val	3409A>G	M1137V	Cystic fibrosis	Zielenski (1993) <i>CFTR LSDB entry</i> Newsletter:#59	Source: LSDB
CFTR	CM940274	ATC-GTC	Ile1139Val	3415A>G	I1139V	Cystic fibrosis	Teng (1994) <i>Hum Mol Genet</i> 3:2249	PubMed 7881429
CFTR	CM983577	ATG-AAG	Met1140Lys	3419T>A	M1140K	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM962479	CAG-TAG	Gln1144Term	3430C>T	Q1144*	Cystic fibrosis	Schwarz (1996) <i>CFTR LSDB entry</i> Newsletter:#67	Source: LSDB
CFTR	CM003256	TGG-TGA	Trp1145Term	3435G>A	W1145*	Cystic fibrosis	Seia (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983578	GTA-ATA	Val1147Ile	3439G>A	V1147I	Cystic fibrosis	Kilinc (2002) <i>Am J Med Genet</i> 113:250	PubMed 12439892
CFTR	CM003257	AAC-AAA	Asn1148Lys	3444C>A	N1148K	Cystic fibrosis	Casals (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM950256	GAT-CAT	Asp1152His	3454G>C	D1152H	Cystic fibrosis	Chillon (1995) <i>N Engl J Med</i> 332:1475	PubMed 7739684
CFTR	CM920180	CGA-TGA	Arg1158Term	3472C>T	R1158*	Cystic fibrosis	Ronchetto (1992) <i>Genomics</i> 12:417	PubMed 1371265
CFTR	CM993868	TCT-TTT	Ser1159Phe	3476C>T	S1159F	Cystic fibrosis	Hirtz (2004) <i>Gastroenterology</i> 127:1085	PubMed 15480987
CFTR	CM931153	TCT-CCT	Ser1159Pro	3475T>C	S1159P	Cystic fibrosis	Macek (1993) <i>CFTR LSDB entry</i> Newsletter:#55	Source: LSDB
CFTR	CM011285	AGC-AGG	Ser1161Arg	3483C>G	S1161R	Cystic fibrosis	Le Maréchal (2001) <i>Hum Genet</i> 108:290	PubMed 11379874
CFTR	CM014892	CGA-CAA	Arg1162Gln	3485G>A	R1162Q	Cystic fibrosis	Strandvik (2001) <i>Genet Test</i> 5:235	PubMed 11788090
CFTR	CM021555	CGA-CTA	Arg1162Leu	3485G>T	R1162L	Cystic fibrosis	Groman (2002) <i>N Engl J Med</i> 347:401	PubMed 12167682
CFTR	CM910074	CGA-TGA	Arg1162Term	3484C>T	R1162*	Cystic fibrosis	Gasparini (1991) <i>Genomics</i> 10:193	PubMed 2045102
CFTR	CM034793	TTC-TGC	Phe1166Cys	3497T>G	F1166C	Cystic fibrosis	Messaoud (2005) <i>Ann Biol Clin (Paris)</i> 63:627	PubMed 16330381
CFTR	CM931154	GAC-GGC	Asp1168Gly	3503A>G	D1168G	Cystic fibrosis	Macek (1993) <i>CFTR LSDB entry</i> Newsletter:#58	Source: LSDB
CFTR	CM941984	AAG-AGG	Lys1177Arg	3530A>G	K1177R	Cystic fibrosis	Baralle (1994) <i>CFTR LSDB entry</i> Newsletter:#61	Source: LSDB

CFTR	CM001653	AAG-TAG	Lys1177Term	3529A>T	K1177*	Cystic fibrosis	Banjar (1999) <i>Ann Trop Paediatr</i> 19:69	PubMed 10605524
CFTR	CM941985	TAC-TAG	Tyr1182Term	3546C>G	Y1182*	Cystic fibrosis	Wallace (1994) <i>CFTR LSDB entry</i> Newsletter:#64	Source: LSDB
CFTR	CM962480	CAA-TAA	Gln1186Term	3556C>T	Q1186*	Cystic fibrosis	Haworth (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM941986	GTT-GAT	Val1190Asp	3569T>A	V1190D	Cystic fibrosis	Glavac (1994) <i>CFTR LSDB entry</i> Newsletter:#64	Source: LSDB
CFTR	CM024694	AAT-ACT	Asn1195Thr	3584A>C	N1195T	Cystic fibrosis	Savov (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM930132	TCA-TGA	Ser1196Term	3587C>G	S1196*	Cystic fibrosis	Ivaschenko (1993) <i>Hum Genet</i> 91:63	PubMed 7681034
CFTR	CM920181	AAA-GAA	Lys1200Glu	3598A>G	K1200E	Cystic fibrosis	Fanen (1992) <i>Genomics</i> 13:770	PubMed 1379210
CFTR	CM920182	ATC-GTC	Ile1203Val	3607A>G	I1203V	Cystic fibrosis	Cutting (1992) <i>Am J Hum Genet</i> 50:1185	PubMed 1376017
CFTR	CM920183	TGG-TAG	Trp1204Term	3611G>A	W1204*	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM067771	TGG-TGA	Trp1204Term	3612G>A	W1204*	Cystic fibrosis	Schrijver (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM141631	TCA-TAA	Ser1206Term	3617C>A	S1206*	Cystic fibrosis	Ziętkiewicz (2014) <i>PLoS One</i> 9:e89094	PubMed 24586523
CFTR	CM962481	TCA-TGA	Ser1206Term	3617C>G	S1206*	Cystic fibrosis	Ferec (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM931155	ATG-ATA	Met1210Ile	3630G>A	M1210I	Cystic fibrosis	Nukiwa (1993) <i>CFTR LSDB entry</i> Newsletter:#55	Source: LSDB
CFTR	CM043475	ATG-AAG	Met1210Lys	3629T>A	M1210K	Cystic fibrosis	Seia (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM931156	GTC-ATC	Val1212Ile	3634G>A	V1212I	Cystic fibrosis	Macek (1993) <i>CFTR LSDB entry</i> Newsletter:#55	Source: LSDB
CFTR	CM032556	ATA-AGA	Ile1226Arg	3677T>G	I1226R	Cystic fibrosis	Korytina (2003) <i>Mol Biol (Mosk)</i> 37:61	PubMed 12624947
CFTR	CM972959	TTA-TCA	Leu1227Ser	3680T>C	L1227S	Cystic fibrosis	Jezequel (2000) <i>Mol Hum Reprod</i> 6:1063	PubMed 11101688
CFTR	CM972960	ATT-ACT	Ile1230Thr	3689T>C	I1230T	Cystic fibrosis	Claustres (1997) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM920184	ATA-GTA	Ile1234Val	3700A>G	I1234V	Cystic fibrosis	Claustres (1992) <i>Hum Mutat</i> 1:310	PubMed 1284537
CFTR	CM930133	AGT-AGG	Ser1235Arg	3705T>G	S1235R	Cystic fibrosis	Cuppens (1993) <i>Genomics</i> 18:693	PubMed 7508414
CFTR	CM003259	GGC-AGC	Gly1237Ser	3709G>A	G1237S	Cystic fibrosis	Casals (2000) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM931157	CAG-CGG	Gln1238Arg	3713A>G	Q1238R	Cystic fibrosis	Ferec (1993) <i>CFTR LSDB entry</i> Newsletter:#58	Source: LSDB
CFTR	CM930134	CAG-TAG	Gln1238Term	3712C>T	Q1238*	Cystic fibrosis	Audrezet (1993) <i>Hum Mol Genet</i> 2:51	PubMed 7683952
CFTR	CM993869	GTG-GGG	Val1240Gly	3719T>G	V1240G	Cystic fibrosis	Sobczynska-Tomaszewska (2013) <i>Eur J Hum Genet</i> 21:391	PubMed 22892530
CFTR	CM057580	GGA-AGA	Gly1244Arg	3730G>A	G1244R	Cystic fibrosis	Lucarelli (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM141632	GGA-CGA	Gly1244Arg	3730G>C	G1244R	Cystic fibrosis	Ziętkiewicz (2014) <i>PLoS One</i> 9:e89094	PubMed 24586523
CFTR	CM910075	GGA-GAA	Gly1244Glu	3731G>A	G1244E	Cystic fibrosis	Devoto (1991) <i>Am J Hum Genet</i> 48:1127	PubMed 1709778
CFTR	CM940275	GGA-GTA	Gly1244Val	3731G>T	G1244V	Cystic fibrosis	Savov (1994) <i>Hum Mol Genet</i> 3:513	PubMed 7516777
CFTR	CM941987	ACT-ATT	Thr1246Ile	3737C>T	T1246I	Cystic fibrosis	Ferec (1994) <i>CFTR LSDB entry</i> Newsletter:#64	Source: LSDB
CFTR	CM962482	GGA-AGA	Gly1247Arg	3739G>A	G1247R	Cystic fibrosis	Casals (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM057584	GGA-CGA	Gly1247Arg	3739G>C	G1247R	Cystic fibrosis	Lucarelli (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM1210538	GGA-TGA	Gly1247Term	3739G>T	G1247*	Cystic fibrosis	Eminoglu (2012) <i>Indian J Pediatr</i> epub:epub	PubMed 22798282
CFTR	CM940277	GGG-AGG	Gly1249Arg	3745G>A	G1249R	Cystic fibrosis	Dijkstra (1994) <i>Hum Mutat</i> 4:161	PubMed 7526929
CFTR	CM940276	GGG-GAG	Gly1249Glu	3746G>A	G1249E	Cystic fibrosis	Greil (1994) <i>Hum Hered</i> 44:238	PubMed 7520022
CFTR	CM920185	AGT-AAT	Ser1251Asn	3752G>A	S1251N	Cystic fibrosis	Kaelin (1992) <i>Hum Mutat</i> 1:204	PubMed 1284535
CFTR	CM962483	ACT-CCT	Thr1252Pro	3754A>C	T1252P	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:375	PubMed 16049310
CFTR	CM960290	TTA-TAA	Leu1254Term	3761T>A	L1254*	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM980356	TCA-TTA	Ser1255Leu	3764C>T	S1255L	Cystic fibrosis	Cartault (1998) <i>Clin Genet</i> 54:437	PubMed 9842999
CFTR	CM920187	TCA-CCA	Ser1255Pro	3763T>C	S1255P	Cystic fibrosis	Lissens (1992) <i>Hum Mol Genet</i> 1:441	PubMed 1284530
CFTR	CM920186	TCA-TAA	Ser1255Term	3764C>A	S1255*	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM983579	TTT-TTG	Phe1257Leu	3771T>G	F1257L	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM024695	CTA-CGA	Leu1260Arg	3779T>G	L1260R	Cystic fibrosis	Seia (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067743	CAG-CGG	Gln1268Arg	3803A>G	Q1268R	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB

CFTR	CM952056	ATC-AAC	Ile1269Asn	3806T>A	I1269N	Cystic fibrosis	McDowell (1995) <i>CFTR LSDB entry</i> Newsletter:#66	Source: LSDB
CFTR	CM920188	GAT-AAT	Asp1270Asn	3808G>A	D1270N	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM057575	GAT-TAT	Asp1270Tyr	3808G>T	D1270Y	Cystic fibrosis	Wang (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM993870	TGG-TGA	Trp1274Term	3822G>A	W1274*	Cystic fibrosis	Zielenski (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM970297	CAG-TAG	Gln1281Term	3841C>T	Q1281*	Cystic fibrosis	Casals (1997) <i>Hum Genet</i> 101:365	PubMed 9439669
CFTR	CM930135	TGG-CGG	Trp1282Arg	3844T>C	W1282R	Cystic fibrosis	Ivaschenko (1993) <i>Hum Genet</i> 91:63	PubMed 7681034
CFTR	CM962484	TGG-TGT	Trp1282Cys	3846G>T	W1282C	Cystic fibrosis	Ferec (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM962485	TGG-GGG	Trp1282Gly	3844T>G	W1282G	Cystic fibrosis	Faucz (2007) <i>Clin Genet</i> 72:218	PubMed 17718859
CFTR	CM900061	TGG-TGA	Trp1282Term	3846G>A	W1282*	Cystic fibrosis	Kerem (1990) <i>Proc Natl Acad Sci U S A</i> 87:8447	PubMed 2236053
CFTR	CM940278	AGG-AAG	Arg1283Lys	3848G>A	R1283K	Cystic fibrosis	Chevalier-Porst (1994) <i>J Med Genet</i> 31:541	PubMed 7525963
CFTR	CM920189	AGG-ATG	Arg1283Met	3848G>T	R1283M	Cystic fibrosis	Cheadle (1992) <i>Hum Mol Genet</i> 1:123	PubMed 1284468
							Sobczynska-Tomaszewska (2013) <i>Eur J Hum Genet</i> 21:391	PubMed 22892530
CFTR	CM132378	TTT-TGT	Phe1286Cys	3857T>G	F1286C	Cystic fibrosis	Dorval (1993) <i>Hum Genet</i> 91:254	PubMed 8097485
CFTR	CM930136	TTT-TCT	Phe1286Ser	3857T>C	F1286S	Cystic fibrosis	Chou (2005) <i>J Mol Diagn</i> 7:111	PubMed 15681482
CFTR	CM055928	GGA-AGA	Gly1287Arg	3859G>A	G1287R	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM067732	CCA-ACA	Pro1290Thr	3868C>A	P1290T	Cystic fibrosis	Doerk (1994) <i>Hum Genet</i> 94:533	PubMed 7525450
CFTR	CM940279	CAG-CGG	Gln1291Arg	3872A>G	Q1291R	Cystic fibrosis	Jones (1992) <i>Hum Mol Genet</i> 1:11	PubMed 1284466
CFTR	CM920190	CAG-CAC	Gln1291His	3873G>C	Q1291H	Cystic fibrosis	Feldmann (2001) <i>Hum Mutat</i> 17:356	PubMed 11295849
CFTR	CM003260	CAG-TAG	Gln1291Term	3871C>T	Q1291*	Cystic fibrosis	Ferec (1997) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM972961	GTA-ATA	Val1293Ile	3877G>A	V1293I	Cystic fibrosis	Liechti (1999) <i>Eur J Hum Genet</i> 7:590	PubMed 10439967
CFTR	CM992307	ACA-ATA	Thr1299Ile	3896C>T	T1299I	Cystic fibrosis	Poncin (1996) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM962486	TTT-CTT	Phe1300Leu	3898T>C	F1300L	Cystic fibrosis	Bonyadi (2011) <i>Genet Test Mol Biomarkers</i> 15:89	PubMed 21198395
CFTR	CM111172	AAA-TAA	Lys1302Term	3904A>T	K1302*	Cystic fibrosis	Claustres (1992) <i>Hum Mutat</i> 1:310	PubMed 1284537
CFTR	CM920191	AAC-CAC	Asn1303His	3907A>C	N1303H	Cystic fibrosis	Lissens (1995) <i>CFTR LSDB entry</i> Newsletter:#66	Source: LSDB
CFTR	CM952057	AAC-ATC	Asn1303Ile	3908A>T	N1303I	Cystic fibrosis	Osborne (1991) <i>Am J Hum Genet</i> 48:608	PubMed 1998343
CFTR	CM910076	AAC-AAG	Asn1303Lys	3909C>G	N1303K	Cystic fibrosis	Claustres (1997) <i>CFTR LSDB entry</i> Newsletter:#69	Source: LSDB
CFTR	CM972962	GAT-GAA	Asp1305Glu	3915T>A	D1305E	Cystic fibrosis	Zielenski (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993871	TAT-TAA	Tyr1307Term	3921T>A	Y1307*	Cystic fibrosis	Alonso (2006) <i>Ann Hum Genet</i> 71:194	PubMed 17331079
CFTR	CM972963	GAA-TAA	Glu1308Term	3922G>T	E1308*	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM920192	TGG-TAG	Trp1310Term	3929G>A	W1310*	Cystic fibrosis	Malone (1996) <i>CFTR LSDB entry</i> Newsletter:#68	Source: LSDB
CFTR	CM962487	CAA-AAA	Gln1313Lys	3937C>A	Q1313K	Cystic fibrosis	Audrezet (1993) <i>Hum Mol Genet</i> 2:51	PubMed 7683952
CFTR	CM930137	CAA-TAA	Gln1313Term	3937C>T	Q1313*	Cystic fibrosis	Cutting (1990) <i>N Engl J Med</i> 323:1685	PubMed 2233965
CFTR	CM900062	TGG-TAG	Trp1316Term	3947G>A	W1316*	Cystic fibrosis	Ferec (1998) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983580	GTT-GCT	Val1318Ala	3953T>C	V1318A	Cystic fibrosis	Ferec (1994) <i>CFTR LSDB entry</i> Newsletter:#64	Source: LSDB
CFTR	CM941988	GAG-CAG	Glu1321Gln	3961G>C	E1321Q	Cystic fibrosis	Bienvenu (2005) <i>Hum Biol</i> 77:705	PubMed 16596947
CFTR	CM043477	CTC-CCC	Leu1324Pro	3971T>C	L1324P	Cystic fibrosis	Sharma (2009) <i>Ann Hum Genet</i> 73:26	PubMed 18782298
CFTR	CM090071	GAA-CAA	Glu1329Gln	3985G>C	E1329Q	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM067762	CTT-TTT	Leu1335Phe	4003C>T	L1335F	Cystic fibrosis	Zielenski (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM972964	CTT-CCT	Leu1335Pro	4004T>C	L1335P	Cystic fibrosis	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM920193	GGC-GAC	Gly1349Asp	4046G>A	G1349D	Cystic fibrosis	Yoshimura (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993872	GGC-AGC	Gly1349Ser	4045G>A	G1349S	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM067760	CAG-GAG	Gln1352Glu	4054C>G	Q1352E	Cystic fibrosis	Lee (2005) <i>Dig Dis Sci</i> 50:1852	PubMed 16187186
CFTR	CM931158	CAG-CAC	Gln1352His	4056G>C	Q1352H	Cystic fibrosis	Ferec (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993873	AGA-AGT	Arg1358Ser	4074A>T	R1358S	Cystic fibrosis		

CFTR	CM1212245	ATC-AAC	Ile1366Asn	4097T>A	I1366N	Cystic fibrosis	Křenková (2013) <i>J Cyst Fibros</i> 12:532	PubMed 23276700
CFTR	CM993874	ATC-ACC	Ile1366Thr	4097T>C	I1366T	Cystic fibrosis	Ferec (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM920194	GAA-TAA	Glu1371Term	4111G>T	E1371*	Cystic fibrosis	Cutting (1992) <i>Am J Hum Genet</i> 50:1185	PubMed 1376017
CFTR	CM055923	CCC-CTC	Pro1372Leu	4115C>T	P1372L	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM087538	CAT-AAT	His1375Asn	4123C>A	H1375N	Cystic fibrosis	Kolesár (2008) <i>Gen Physiol Biophys</i> 27:299	PubMed 19202204
CFTR	CM057574	CAT-CCT	His1375Pro	4124A>C	H1375P	Cystic fibrosis	Narzi (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM067737	TAC-CAC	Tyr1381His	4141T>C	Y1381H	Cystic fibrosis	Sharma (2009) <i>Ann Hum Genet</i> 73:26	PubMed 18782298
CFTR	CM024696	TAC-TAA	Tyr1381Term	4143C>A	Y1381*	Cystic fibrosis	Casals (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983581	CAA-TAA	Gln1382Term	4144C>T	Q1382*	Cystic fibrosis	Claustres (1998) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM940280	GTA-GAA	Val1397Glu	4190T>A	V1397E	Cystic fibrosis	Petreska (1994) <i>Hum Mol Genet</i> 3:999	PubMed 7524913
CFTR	CM057572	ATT-AGT	Ile1398Ser	4193T>G	I1398S	Cystic fibrosis	Wang (2005) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM024697	GAA-GCA	Glu1401Ala	4202A>C	E1401A	Cystic fibrosis	Valaskova (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM983582	GAA-TAA	Glu1401Term	4201G>T	E1401*	Cystic fibrosis	Strandvik (2001) <i>Genet Test</i> 5:235	PubMed 11788090
CFTR	CM043478	ATG-ACG	Met1407Thr	4220T>C	M1407T	Cystic fibrosis	Girodon (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM931159	GAA-GTA	Glu1409Val	4226A>T	E1409V	Cystic fibrosis	Claustres (1993) <i>CFTR LSDB entry</i> Newsletter:#55	Source: LSDB
CFTR	CM960291	CAA-TAA	Gln1412Term	4234C>T	Q1412*	Cystic fibrosis	Hughes (1996) <i>Hum Mutat</i> 8:340	PubMed 8956039
CFTR	CM043479	GAG-TAG	Glu1418Term	4252G>T	E1418*	Cystic fibrosis	Girodon (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM972965	CGG-TGG	Arg1422Trp	4264C>T	R1422W	Cystic fibrosis	Claustres (1997) <i>CFTR LSDB entry</i> Newsletter:#70	Source: LSDB
CFTR	CM993875	TCC-CCC	Ser1426Pro	4276T>C	S1426P	Cystic fibrosis	Ferec (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM043480	CGG-TGG	Arg1438Trp	4312C>T	R1438W	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM962488	GAC-AAC	Asp1445Asn	4333G>A	D1445N	Cystic fibrosis	Schrijver (2005) <i>J Mol Diagn</i> 7:289	PubMed 15858154
CFTR	CM993876	CGG-TGG	Arg1453Trp	4357C>T	R1453W	Cystic fibrosis	Yoshimura (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM993877	CAA-TAA	Gln1476Term	4426C>T	Q1476*	Cystic fibrosis	Claustres (1999) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM067753	CTT-CCT	Leu1480Pro	4439T>C	L1480P	Cystic fibrosis	Picci (2006) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM031947	TAG-TGG	Term1481Trp	4442A>G	*1481W	Cystic fibrosis	Bienvenu (2003) <i>Clin Genet</i> 64:266	PubMed 12919146
CFTR	CM146100	TGT-TAT	Cys592Tyr	1775G>A	C592Y	disorder	Ramos (2014) <i>Clin Genet</i> 86:91	PubMed 23837941
CFTR	CM1314598	CAG-CAC	Gln237His	711G>C	Q237H	Cystic fibrosis, atypical	Michl (2013) <i>Klin Padiatr</i> 225:288	PubMed 24022636
CFTR	CM129624	TTT-GTT	Phe319Val	955T>G	F319V	Cystic fibrosis, atypical	Poulou (2012) <i>J Cyst Fibros</i> 11:344	PubMed 22326559
CFTR	CM088478	ATC-AGC	Ile752Ser	2255T>G	I752S	Cystic fibrosis, atypical	Paranjape (2008) <i>Clin Rev Allergy Immunol</i> 35:116	PubMed 18493878
CFTR	CM980357	TCA-TGA	Ser1455Term	4364C>G	S1455*	concentration	Mickle (1998) <i>Hum Mol Genet</i> 7:729	PubMed 9499426
CFTR	CM043466	CTG-CAG	Leu548Gln	1643T>A	L548Q	bowel	Yamamoto (2006) <i>Prenat Diagn</i> 26:6	PubMed 16378323
CFTR	CM990360	TAT-TGT	Tyr301Cys	902A>G	Y301C	sweat chloride	Castellani (1999) <i>Am J Hum Genet</i> 64:303	PubMed 9915972
CFTR	CM990361	GAA-GGA	Glu527Gly	1580A>G	E527G	sweat chloride	Puget (1999) <i>Am J Hum Genet</i> 64:303	PubMed 9915972
CFTR	CM001652	TTG-TCG	Leu967Ser	2900T>C	L967S	sweat chloride	Boyne (2000) <i>J Med Genet</i> 37:543	PubMed 10970190
CFTR	CM024660	GAC-AAC	Asp110Asn	328G>A	D110N	Hypertrypsinaemia, neonatal	Seia (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM024670	GCC-GTC	Ala309Val	926C>T	A309V	Hypertrypsinaemia, neonatal	Ferec (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM024692	TAC-TGC	Tyr1073Cys	3218A>G	Y1073C	Hypertrypsinaemia, neonatal	Seia (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM043470	GCT-GAT	Ala1025Asp	3074C>A	A1025D	Hypochloraemic alkalosis	Schrijver (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM129621	CCA-ACA	Pro936Thr	2806C>A	P936T	Inadequate weight gain	Poulou (2012) <i>J Cyst Fibros</i> 11:344	PubMed 22326559
CFTR	CM140666	TGC-TAC	Cys524Tyr	1571G>A	C524Y	Meconium ileus, lethal	Puzik (2014) <i>BMC Pediatr</i> 14:13	PubMed 24433235
CFTR	CM129620	CAA-CAC	Gln1209His	3627A>C	Q1209H	Metabolic alkalosis	Poulou (2012) <i>J Cyst Fibros</i> 11:344	PubMed 22326559
CFTR	CM024682	GAC-CAC	Asp651His	1951G>C	D651H	Obstructive azoospermia	Mittre (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM024698	TCC-TTC	Ser1426Phe	4277C>T	S1426F	Obstructive azoospermia	Casals (2002) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB

CFTR	CM074729	ATA-ATG	Ile980Met	2940A>G	I980M	Oligospermia	Pasquet (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM931139	CGC-TGC	Arg31Cys	91C>T	R31C	Pancreatitis	Gomez Lira (2001) <i>Pancreatology</i> 1:538	PubMed 12120234
CFTR	CM931145	CTA-CGA	Leu327Arg	980T>G	L327R	Pancreatitis	Ravnik-Glavak (1996) <i>Pflugers Arch</i> 431:R191	PubMed 8992448
CFTR	CM053173	ATG-GTG	Met348Val	1042A>G	M348V	Pancreatitis	Weiss (2005) <i>Gut</i> 54:1456	PubMed 15987793
CFTR	CM108945	CCT-CGT	Pro439Arg	1316C>G	P439R	Pancreatitis	Pelletier (2010) <i>Pancreatology</i> 10:158	PubMed 20460946
CFTR	CM024675	ATT-GTT	Ile497Val	1489A>G	I497V	Pancreatitis	Gaia (2002) <i>Dig Dis Sci</i> 47:2416	PubMed 12452372
CFTR	CM043468	TCC-TTC	Ser737Phe	2210C>T	S737F	Pancreatitis	Centrone (2004) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM034790	CCC-CAC	Pro1013His	3038C>A	P1013H	Pancreatitis	Bieth (2003) <i>CFTR LSDB entry</i> Pers comm:	Source: LSDB
CFTR	CM053174	GCC-CCC	Ala1087Pro	3259G>C	A1087P	Pancreatitis	Weiss (2005) <i>Gut</i> 54:1456	PubMed 15987793
CFTR	CM136421	GAT-GGT	Asp1312Gly	3935A>G	D1312G	Pancreatitis	Hamoir (2013) <i>Digestion</i> 87:229	PubMed 23751316
CFTR	CM087308	CTG-CCG	Leu188Pro	563T>C	L188P	Pancreatitis, chronic	Audrezet (2008) <i>J Mol Diagn</i> 10:424	PubMed 18687795
CFTR	CM066544	GGA-AGA	Gly194Arg	580G>A	G194R	Pancreatitis, chronic	Keiles (2006) <i>Pancreas</i> 33:221	PubMed 17003641
CFTR	CM087309	ATG-ACG	Met284Thr	851T>C	M284T	Pancreatitis, chronic	Audrezet (2008) <i>J Mol Diagn</i> 10:424	PubMed 18687795
CFTR	CM066547	TCT-TGT	Ser573Cys	1718C>G	S573C	Pancreatitis, chronic	Keiles (2006) <i>Pancreas</i> 33:221	PubMed 17003641
CFTR	CM066548	TTT-TAT	Phe575Tyr	1724T>A	F575Y	Pancreatitis, chronic	Keiles (2006) <i>Pancreas</i> 33:221	PubMed 17003641
CFTR	CM066545	AAA-GAA	Lys598Glu	1792A>G	K598E	Pancreatitis, chronic	Keiles (2006) <i>Pancreas</i> 33:221	PubMed 17003641
CFTR	CM133354	CAC-TAC	His667Tyr	1999C>T	H667Y	Pancreatitis, chronic	Rosendahl (2013) <i>Gut</i> 62:582	PubMed 22427236
CFTR	CM066549	TTT-TTG	Phe834Leu	2502T>G	F834L	Pancreatitis, chronic	Keiles (2006) <i>Pancreas</i> 33:221	PubMed 17003641
CFTR	CM140645	CAA-CGA	Gln1042Arg	3125A>G	Q1042R	Pancreatitis, chronic	Sinha (2014) <i>Pancreas</i> 43:146	PubMed 24326373
CFTR	CM066546	CTA-CCA	Leu1260Pro	3779T>C	L1260P	Pancreatitis, chronic	Keiles (2006) <i>Pancreas</i> 33:221	PubMed 17003641
CFTR	CM920145	ATT-ACT	Ile148Thr	443T>C	I148T	Phenotype modifier	Tsui (1992) <i>Hum Mutat</i> 1:197	PubMed 1284534
CFTR	CM031945	TCT-TAT	Ser686Tyr	2057C>A	S686Y	cholangitis	Sheth (2003) <i>Hum Genet</i> 113:286	PubMed 12783301
CFTR	CM031946	ATC-TTC	Ile1366Phe	4096A>T	I1366F	cholangitis	Sheth (2003) <i>Hum Genet</i> 113:286	PubMed 12783301
CFTR	CM074730	ATG-ATA	Met595Ile	1785G>A	M595I	cystic fibrosis	Rana-Díez (2008) <i>J Cyst Fibros</i> 7:520	PubMed 18676185
CFTR	CM074734	GAC-GGC	Asp993Gly	2978A>G	D993G	cystic fibrosis Respiratory/pancreatic	Omar (2007) <i>CFTR LSDB entry</i> Unpublished:.	Source: LSDB
CFTR	CM034388	ATG-GTG	Met470Val	1408A>G	M470V	disease, association with	Lee (2003) <i>Hum Mol Genet</i> 12:2321	PubMed 12952861
CFTR	CM001963	ATA-GTA	Ile991Val	2971A>G	I991V	Sarcoidosis, susceptibility to	Bombieri (2000) <i>Eur J Hum Genet</i> 8:717	PubMed 10980579
COL1A1	CM137809	ATG-ACG	Met1Thr	2T>C	M1T	Osteogenesis imperfecta	Ben Amor (2013) <i>J Bone Miner Res</i> 28:2001	PubMed 23529829
COL1A1	CM117851	GGC-GAC	Gly26Asp	77G>A	G26D	Osteogenesis imperfecta	Fuccio (2011) <i>J Mol Diagn</i> 13:648	PubMed 21884818
COL1A1	CM137813	GGA-TGA	Gly148Term	442G>T	G148*	Osteogenesis imperfecta	Ben Amor (2013) <i>J Bone Miner Res</i> 28:2001	PubMed 23529829
COL1A1	CM086454	GGC-GTC	Gly200Val	599G>T	G200V	Osteogenesis imperfecta	Witecka (2008) <i>J Appl Genet</i> 49:283	PubMed 18670065
COL1A1	CM062530	GGT-GAT	Gly242Asp	725G>A	G242D	Osteogenesis imperfecta	Lee (2006) <i>Hum Mutat</i> 27:599	PubMed 16705691
COL1A1	CM040707	CGA-TGA	Arg253Term	757C>T	R253*	Osteogenesis imperfecta	Ries-Levavi (2004) <i>Hum Mutat</i> 23:399	PubMed 15024745
COL1A1	CM045285	GGT-AGT	Gly299Ser	895G>A	G299S	Osteogenesis imperfecta	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A1	CM124995	GGA-TGA	Gly323Term	967G>T	G323*	Osteogenesis imperfecta	Nadyrshina (2012) <i>Genetika</i> 48:372	PubMed 22679784
COL1A1	CM086455	GTT-TTT	Val349Phe	1045G>T	V349F	Osteogenesis imperfecta	Witecka (2008) <i>J Appl Genet</i> 49:283	PubMed 18670065
COL1A1	CM137814	CAG-TAG	Gln367Term	1099C>T	Q367*	Osteogenesis imperfecta	Ben Amor (2013) <i>J Bone Miner Res</i> 28:2001	PubMed 23529829
COL1A1	CM110790	GGT-GAT	Gly437Asp	1310G>A	G437D	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM094109	CGA-TGA	Arg469Term	1405C>T	R469*	Osteogenesis imperfecta	Swinnen (2009) <i>Laryngoscope</i> 119:1171	PubMed 19358256
COL1A1	CM033357	GGT-GCT	Gly659Ala	1976G>C	G659A	Osteogenesis imperfecta	Benusiene (2003) <i>J Appl Genet</i> 44:95	PubMed 12590186
COL1A1	CM137808	CAG-TAG	Gln661Term	1981C>T	Q661*	Osteogenesis imperfecta	Ben Amor (2013) <i>J Bone Miner Res</i> 28:2001	PubMed 23529829
COL1A1	CM110794	GGC-AGC	Gly668Ser	2002G>A	G668S	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM033358	GAG-TAG	Glu678Term	2032G>T	E678*	Osteogenesis imperfecta	Benusiene (2003) <i>J Appl Genet</i> 44:95	PubMed 12590186

COL1A1	CM062536	GGC-AGC	Gly683Ser	2047G>A	G683S	Osteogenesis imperfecta	Venturi (2006) <i>Clin Genet</i> 70:131	PubMed 16879195
COL1A1	CM094476	GGT-GCT	Gly686Ala	2057G>C	G686A	Osteogenesis imperfecta	Wang (2009) <i>J Investig Med</i> 57:662	PubMed 19491628
COL1A1	CM110795	GGT-GTT	Gly731Val	2192G>T	G731V	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM110796	GGT-GAT	Gly743Asp	2228G>A	G743D	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM110797	GGT-AGT	Gly764Ser	2290G>A	G764S	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM110792	GGT-GCT	Gly824Ala	2471G>C	G824A	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM110793	GGT-GTT	Gly863Val	2588G>T	G863V	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM040708	CAG-TAG	Gln957Term	2869C>T	Q957*	Osteogenesis imperfecta	Ries-Levavi (2004) <i>Hum Mutat</i> 23:399	PubMed 15024745
COL1A1	CM094107	GGT-GCT	Gly974Ala	2921G>C	G974A	Osteogenesis imperfecta	Swinnen (2009) <i>Laryngoscope</i> 119:1171	PubMed 19358256
COL1A1	CM110798	GGT-AGT	Gly1109Ser	3325G>A	G1109S	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM137810	CAA-TAA	Gln1129Term	3385C>T	Q1129*	Osteogenesis imperfecta	Ben Amor (2013) <i>J Bone Miner Res</i> 28:2001	PubMed 23529829
COL1A1	CM068181	GGT-GAT	Gly1157Asp	3470G>A	G1157D	Osteogenesis imperfecta	Wang (2006) <i>Zhonghua Yi Xue Za Zhi</i> 86:170	PubMed 16638323
COL1A1	CM137811	CAG-TAG	Gln1203Term	3607C>T	Q1203*	Osteogenesis imperfecta	Ben Amor (2013) <i>J Bone Miner Res</i> 28:2001	PubMed 23529829
COL1A1	CM114195	GCT-ACT	Ala1218Thr	3652G>A	A1218T	Osteogenesis imperfecta	Lindahl (2011) <i>Hum Mutat</i> 32:598	PubMed 21344539
COL1A1	CM114190	GAT-AAT	Asp1219Asn	3655G>A	D1219N	Osteogenesis imperfecta	Lindahl (2011) <i>Hum Mutat</i> 32:598	PubMed 21344539
COL1A1	CM110799	GAC-GAG	Asp1277Glu	3831C>G	D1277E	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A1	CM142714	AAC-CAC	Asn1279His	3835A>C	N1279H	Osteogenesis imperfecta	Lu (2014) <i>Clin Endocrinol (Oxf)</i> 80:524	PubMed 24147872
COL1A1	CM142715	ACC-AAC	Thr1298Asn	3893C>A	T1298N	Osteogenesis imperfecta	Lu (2014) <i>Clin Endocrinol (Oxf)</i> 80:524	PubMed 24147872
COL1A1	CM142716	TGC-TGA	Cys1299Term	3897C>A	C1299*	Osteogenesis imperfecta	Lu (2014) <i>Clin Endocrinol (Oxf)</i> 80:524	PubMed 24147872
COL1A1	CM012085	TGC-TGG	Cys1299Trp	3897C>G	C1299W	Osteogenesis imperfecta	Pace (2001) <i>J Med Genet</i> 38:443	PubMed 11432962
COL1A1	CM094108	CAG-TAG	Gln1304Term	3910C>T	Q1304*	Osteogenesis imperfecta	Swinnen (2009) <i>Laryngoscope</i> 119:1171	PubMed 19358256
COL1A1	CM040710	CAT-TAT	His1323Tyr	3967C>T	H1323Y	Osteogenesis imperfecta	Ries-Levavi (2004) <i>Hum Mutat</i> 23:399	PubMed 15024745
COL1A1	CM137812	TGG-TGA	Trp1325Term	3975G>A	W1325*	Osteogenesis imperfecta	Ben Amor (2013) <i>J Bone Miner Res</i> 28:2001	PubMed 23529829
COL1A1	CM142717	CAG-TAG	Gln1341Term	4021C>T	Q1341*	Osteogenesis imperfecta	Lu (2014) <i>Clin Endocrinol (Oxf)</i> 80:524	PubMed 24147872
COL1A1	CM117852	ACC-ATC	Thr1432Ile	4295C>T	T1432I	Osteogenesis imperfecta	Fuccio (2011) <i>J Mol Diagn</i> epub:epub	PubMed 21884818
COL1A1	CM123296	TGC-TTC	Cys61Phe	182G>T	C61F	Osteogenesis imperfecta I	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A1	CM1211671	GGT-GCT	Gly194Ala	581G>C	G194A	Osteogenesis imperfecta I	Valentina Gentile (2012) <i>Hum Mutat</i> 33:1697	PubMed 22753364
COL1A1	CM062538	GGT-CGT	Gly194Arg	580G>C	G194R	Osteogenesis imperfecta I	Lee (2006) <i>Hum Mutat</i> 27:599	PubMed 16705691
COL1A1	CM983820	GGT-TGT	Gly197Cys	589G>T	G197C	Osteogenesis imperfecta I	Lund (1998) <i>J Craniofac Genet Dev Biol</i> 18:30	PubMed 9594376
COL1A1	CM070748	GGT-AGT	Gly197Ser	589G>A	G197S	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM123297	GGT-AGT	Gly203Ser	607G>A	G203S	Osteogenesis imperfecta I	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A1	CM145060	GGC-GAC	Gly209Asp	626G>A	G209D	Osteogenesis imperfecta I	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A1	CM980389	CGA-TGA	Arg220Term	658C>T	R220*	Osteogenesis imperfecta I	Korkko (1998) <i>Am J Hum Genet</i> 62:98	PubMed 9443882
COL1A1	CM920198	GGT-TGT	Gly221Cys	661G>T	G221C	Osteogenesis imperfecta I	Shapiro (1992) <i>J Clin Invest</i> 89:567	PubMed 1737847
COL1A1	CM910077	GGT-TGT	Gly224Cys	670G>T	G224C	Osteogenesis imperfecta I	Byers (1991) <i>J Med Genet</i> 28:433	PubMed 1895312
COL1A1	CM1110798	GGT-AGT	Gly224Ser	670G>A	G224S	Osteogenesis imperfecta I	Swinnen (2011) <i>Orphanet J Rare Dis</i> 6:88	PubMed 22206639
COL1A1	CM070715	GGT-TGT	Gly251Cys	751G>T	G251C	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM960320	GGA-AGA	Gly257Arg	769G>A	G257R	Osteogenesis imperfecta I	Redford-Badwal (1996) <i>J Clin Invest</i> 97:1035	PubMed 8613526
COL1A1	CM910078	GGA-AGA	Gly263Arg	787G>A	G263R	Osteogenesis imperfecta I	Deak (1991) <i>J Biol Chem</i> 266:21827	PubMed 1718984
COL1A1	CM930140	GGA-GTA	Gly263Val	788G>T	G263V	Osteogenesis imperfecta I	Valli (1993) <i>Eur J Biochem</i> 217:77	PubMed 8223589
COL1A1	CM890180	GGT-TGT	Gly272Cys	814G>T	G272C	Osteogenesis imperfecta I	Starman (1989) <i>J Clin Invest</i> 84:1206	PubMed 2794057
COL1A1	CM077240	GGT-AGT	Gly287Ser	859G>A	G287S	Osteogenesis imperfecta I	Kataoka (2007) <i>Pediatr Int</i> 49:564	PubMed 17875077
COL1A1	CM062534	GAG-AAG	Glu288Lys	862G>A	E288K	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509

COL1A1	CM043267	GGC-CGC	Gly305Arg	913G>C	G305R	Osteogenesis imperfecta I	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A1	CM062531	GGT-GTT	Gly320Val	959G>T	G320V	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM070686	GGC-AGC	Gly338Ser	1012G>A	G338S	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM980390	CGA-TGA	Arg361Term	1081C>T	R361*	Osteogenesis imperfecta I	Korkko (1998) <i>Am J Hum Genet</i> 62:98	PubMed 9443882
COL1A1	CM900063	GGC-TGC	Gly383Cys	1147G>T	G383C	Osteogenesis imperfecta I	Byers (1990) <i>Trends Genet</i> 6:293	PubMed 2238087
COL1A1	CM900064	GGT-TGT	Gly401Cys	1201G>T	G401C	Osteogenesis imperfecta I	Byers (1990) <i>Trends Genet</i> 6:293	PubMed 2238087
COL1A1	CM960321	CGA-TGA	Arg415Term	1243C>T	R415*	Osteogenesis imperfecta I	Willing (1996) <i>Am J Hum Genet</i> 59:799	PubMed 8808594
COL1A1	CM062533	CGA-TGA	Arg472Term	1414C>T	R472*	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM1211672	GGT-AGT	Gly491Ser	1471G>A	G491S	Osteogenesis imperfecta I	Valentina Gentile (2012) <i>Hum Mutat</i> 33:1697	PubMed 22753364
COL1A1	CM042008	GGT-GCT	Gly527Ala	1580G>C	G527A	Osteogenesis imperfecta I	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A1	CM062523	CCC-CGC	Pro555Arg	1664C>G	P555R	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM043754	CGA-TGA	Arg598Term	1792C>T	R598*	Osteogenesis imperfecta I	Siegert (2004) <i>Klin Padiatr</i> 216:91	PubMed 15106082
COL1A1	CM062537	GGT-AGT	Gly647Ser	1939G>A	G647S	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM960322	CGA-TGA	Arg697Term	2089C>T	R697*	Osteogenesis imperfecta I	Willing (1996) <i>Am J Hum Genet</i> 59:799	PubMed 8808594
COL1A1	CM062529	GGC-AGC	Gly722Ser	2164G>A	G722S	Osteogenesis imperfecta I	Lee (2006) <i>Hum Mutat</i> 27:599	PubMed 16705691
COL1A1	CM070709	GGC-GCC	Gly815Ala	2444G>C	G815A	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM071622	CAA-TAA	Gln822Term	2464C>T	Q822*	Osteogenesis imperfecta I	Liu (2007) <i>Mol Vis</i> 13:360	PubMed 17392686
COL1A1	CM070771	GGT-TGT	Gly824Cys	2470G>T	G824C	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM960323	GGT-AGT	Gly839Ser	2515G>A	G839S	Osteogenesis imperfecta I	Nuytinck (1996) <i>Hum Genet</i> 97:324	PubMed 8786074
COL1A1	CM139321	GGT-GAT	Gly878Asp	2633G>A	G878D	Osteogenesis imperfecta I	Cianci (2013) <i>Am J Med Genet A</i> 161:2681	PubMed 23918677
COL1A1	CM042010	CGA-TGA	Arg882Term	2644C>T	R882*	Osteogenesis imperfecta I	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A1	CM870006	GGT-TGT	Gly926Cys	2776G>T	G926C	Osteogenesis imperfecta I	Vogel (1987) <i>J Biol Chem</i> 262:14737	PubMed 3667599
COL1A1	CM040709	CGA-TGA	Arg1026Term	3076C>T	R1026*	Osteogenesis imperfecta I	Ries (2000) <i>Prenat Diagn</i> 20:876	PubMed 11113887
COL1A1	CM920204	GGC-AGC	Gly1079Ser	3235G>A	G1079S	Osteogenesis imperfecta I	Mottes (1992) <i>Hum Genet</i> 89:480	PubMed 1634225
COL1A1	CM144919	GGA-GAA	Gly1088Glu	3263G>A	G1088E	Osteogenesis imperfecta I	Xia (2014) <i>Mol Med Rep</i> 9:2187	PubMed 24682174
COL1A1	CM123298	CAG-TAG	Gln1117Term	3349C>T	Q1117*	Osteogenesis imperfecta I	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A1	CM940296	CGA-TGA	Arg1141Term	3421C>T	R1141*	Osteogenesis imperfecta I	Willing (1994) <i>Am J Hum Genet</i> 55:638	PubMed 7942841
COL1A1	CM970354	GGC-AGC	Gly1151Ser	3451G>A	G1151S	Osteogenesis imperfecta I	Korkko (1997) <i>Hum Mutat</i> 9:148	PubMed 9067755
COL1A1	CM880016	GGT-TGT	Gly1195Cys	3583G>T	G1195C	Osteogenesis imperfecta I	Labhard (1988) <i>Mol Biol Med</i> 5:197	PubMed 3244312
COL1A1	CM123299	AGC-GGC	Ser1199Gly	3595A>G	S1199G	Osteogenesis imperfecta I	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A1	CM062527	GAT-GAG	Asp1219Glu	3657T>G	D1219E	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM123300	GCC-ACC	Ala1256Thr	3766G>A	A1256T	Osteogenesis imperfecta I	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A1	CM1110821	CAG-TAG	Gln1309Term	3925C>T	Q1309*	Osteogenesis imperfecta I	Swinnen (2011) <i>Orphanet J Rare Dis</i> 6:88	PubMed 22206639
COL1A1	CM042011	TGG-TAG	Trp1325Term	3974G>A	W1325*	Osteogenesis imperfecta I	Hartikka (2004) <i>Hum Mutat</i> 24:147 Ke (2009) <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i>	PubMed 15241796
COL1A1	CM091682	GAT-CAT	Asp1441His	4321G>C	D1441H	Osteogenesis imperfecta I Osteogenesis imperfecta I &	26:50 Vandersteen (2013) <i>Am J Med Genet A</i> 164A:386	PubMed 19199251
COL1A1	CM1313371	GGT-CGT	Gly215Arg	643G>C	G215R	valvular heart disease	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 24311407
COL1A1	CM070679	GGT-AGT	Gly269Ser	805G>A	G269S	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070723	GGT-GCT	Gly284Ala	851G>C	G284A	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070736	GGA-AGA	Gly314Arg	940G>A	G314R	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070755	GGT-AGT	Gly854Ser	2560G>A	G854S	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070727	GGT-GCT	Gly1022Ala	3065G>C	G1022A	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM062525	GGC-CGC	Gly22Arg	64G>C	G22R	Osteogenesis imperfecta II	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509

COL1A1	CM090266	CCC-ACC	Pro146Thr	436C>A	P146T	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM920199	GGT-GAT	Gly275Asp	824G>A	G275D	Osteogenesis imperfecta II	Lightfoot (1992) <i>J Biol Chem</i> 267:25521	PubMed 1460046
COL1A1	CM070718	GGT-GAT	Gly281Asp	842G>A	G281D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090696	GAG-GCG	Glu288Ala	863A>C	E288A	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM940290	GGT-CGT	Gly350Arg	1048G>C	G350R	Osteogenesis imperfecta II	MacKay (1994) <i>Hum Mutat</i> 3:324	PubMed 8019571
COL1A1	CM090246	GGT-GAT	Gly353Asp	1058G>A	G353D	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM070730	GGT-GTT	Gly365Val	1094G>T	G365V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090247	GGT-GTT	Gly368Val	1103G>T	G368V	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM942084	GGT-CGT	Gly389Arg	1165G>C	G389R	Osteogenesis imperfecta II	Sztrolowics (1994) <i>Bone</i> 15:321	PubMed 7520724
COL1A1	CM090256	GCT-ACT	Ala390Thr	1168G>A	A390T	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM070696	GGA-GAA	Gly392Glu	1175G>A	G392E	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM952153	GGT-GAT	Gly398Asp	1193G>A	G398D	Osteogenesis imperfecta II	Culbert (1995) <i>Biochem J</i> 311:815	PubMed 7487936
COL1A1	CM070722	GGT-GAT	Gly401Asp	1202G>A	G401D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM055136	CCC-GCC	Pro417Ala	1249C>G	P417A	Osteogenesis imperfecta II	Reis (2005) <i>J Appl Genet</i> 46:105	PubMed 15741671
COL1A1	CM930142	GGC-TGC	Gly422Cys	1264G>T	G422C	Osteogenesis imperfecta II	Fertala (1993) <i>Biochem J</i> 289:195	PubMed 8424758
COL1A1	CM930143	GGC-AGC	Gly425Ser	1273G>A	G425S	Osteogenesis imperfecta II	Mackay (1993) <i>Hum Mol Genet</i> 2:1155	PubMed 7691343
COL1A1	CM070765	GGT-TGT	Gly428Cys	1282G>T	G428C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070732	GGT-GTT	Gly431Val	1292G>T	G431V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM890027	GGT-GTT	Gly434Val	1301G>T	G434V	Osteogenesis imperfecta II	Patterson (1989) <i>J Biol Chem</i> 264:10083	PubMed 2470760
COL1A1	CM090248	GGT-GAT	Gly455Asp	1364G>A	G455D	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM070766	GGT-TGT	Gly455Cys	1363G>T	G455C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070741	GGC-AGC	Gly461Ser	1381G>A	G461S	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070697	GGA-GAA	Gly464Glu	1391G>A	G464E	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090249	GGA-GTA	Gly470Val	1409G>T	G470V	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM070683	GGA-AGA	Gly476Arg	1426G>A	G476R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM910081	GGA-CGA	Gly476Arg	1426G>C	G476R	Osteogenesis imperfecta II	Byers (1991) <i>J Med Genet</i> 28:433	PubMed 1895312
COL1A1	CM070684	GGA-AGA	Gly482Arg	1444G>A	G482R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070768	GGT-TGT	Gly503Cys	1507G>T	G503C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090250	GGT-GTT	Gly509Val	1526G>T	G509V	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM941957	GGT-GAT	Gly533Asp	1598G>A	G533D	Osteogenesis imperfecta II	Raghunath (1994) <i>Pediatr Res</i> 36:441	PubMed 7816518
COL1A1	CM070769	GGT-TGT	Gly533Cys	1597G>T	G533C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090251	GGT-GCT	Gly548Ala	1643G>C	G548A	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM070708	GGC-GCC	Gly554Ala	1661G>C	G554A	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM962422	GGT-CGT	Gly560Arg	1678G>C	G560R	Osteogenesis imperfecta II	Cohen-Solal (1996) <i>Mol Cell Probes</i> 10:219	PubMed 8799376
COL1A1	CM024728	GGA-AGA	Gly566Arg	1696G>A	G566R	Osteogenesis imperfecta II	Galicka (2002) <i>Acta Biochim Pol</i> 49:443	PubMed 12362986
COL1A1	CM070685	GGA-CGA	Gly566Arg	1696G>C	G566R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM870040	GGC-CGC	Gly569Arg	1705G>C	G569R	Osteogenesis imperfecta II	Bateman (1987) <i>J Biol Chem</i> 262:7021	PubMed 3108247
COL1A1	CM062528	GGA-AGA	Gly581Arg	1741G>A	G581R	Osteogenesis imperfecta II	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM070680	GGT-AGT	Gly587Ser	1759G>A	G587S	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070701	GGC-GAC	Gly593Asp	1778G>A	G593D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090252	GGA-AGA	Gly602Arg	1804G>A	G602R	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM090253	GGC-GAC	Gly605Asp	1814G>A	G605D	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM090254	GGA-CGA	Gly614Arg	1840G>C	G614R	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919

COL1A1	CM070738	GGA-AGA	Gly617Arg	1849G>A	G617R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM973108	GGT-TGT	Gly626Cys	1876G>T	G626C	Osteogenesis imperfecta II	Pepin (1997) <i>Prenat Diagn</i> 17:559	PubMed 9203215
COL1A1	CM973110	GGC-AGC	Gly629Ser	1885G>A	G629S	Osteogenesis imperfecta II	Pepin (1997) <i>Prenat Diagn</i> 17:559	PubMed 9203215
COL1A1	CM070678	GGC-AGC	Gly635Ser	1903G>A	G635S	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070711	GGC-GTC	Gly638Val	1913G>T	G638V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070762	GGT-CGT	Gly650Arg	1948G>C	G650R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM970347	GGC-AGC	Gly656Ser	1966G>A	G656S	Osteogenesis imperfecta II	Mottes (1997) <i>Hum Mutat</i> 12:71	PubMed 10627137
COL1A1	CM070733	GGT-GTT	Gly686Val	2057G>T	G686V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM910083	GGT-GAT	Gly719Asp	2156G>A	G719D	Osteogenesis imperfecta II	Zhuang (1991) <i>Am J Hum Genet</i> 48:1186	PubMed 2035536
COL1A1	CM900066	GGA-AGA	Gly728Arg	2182G>A	G728R	Osteogenesis imperfecta II	Wallis (1990) <i>Am J Hum Genet</i> 46:1034	PubMed 2339700
COL1A1	CM062532	GGT-GTT	Gly734Val	2201G>T	G734V	Osteogenesis imperfecta II	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM900325	GGT-GAT	Gly737Asp	2210G>A	G737D	Osteogenesis imperfecta II	Cohn (1990) <i>COL1A1 LSDB entry</i> Unpublished:	Source: LSDB
COL1A1	CM090255	GGG-CGG	Gly740Arg	2218G>C	G740R	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM920202	GGT-AGT	Gly743Ser	2227G>A	G743S	Osteogenesis imperfecta II	Bateman (1992) <i>Biochem J</i> 288:131	PubMed 1445258
COL1A1	CM930146	GGT-GTT	Gly743Val	2228G>T	G743V	Osteogenesis imperfecta II	Mackay (1993) <i>Hum Genet</i> 91:439	PubMed 8100209
COL1A1	CM970348	GGT-GTT	Gly764Val	2291G>T	G764V	Osteogenesis imperfecta II	Lund (1997) <i>Hum Mutat</i> 9:431	PubMed 9143923
COL1A1	CM070770	GGT-TGT	Gly770Cys	2308G>T	G770C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM900067	GGT-AGT	Gly776Ser	2326G>A	G776S	Osteogenesis imperfecta II	Westerhausen (1990) <i>J Biol Chem</i> 265:13995	PubMed 2116413
COL1A1	CM900068	GGC-AGC	Gly809Ser	2425G>A	G809S	Osteogenesis imperfecta II	Westerhausen (1990) <i>J Biol Chem</i> 265:13995	PubMed 2116413
COL1A1	CM910084	GGC-GTC	Gly815Val	2444G>T	G815V	Osteogenesis imperfecta II	Tsuneyoshi (1991) <i>J Biol Chem</i> 266:15608	PubMed 1874719
COL1A1	CM070688	GGC-CGC	Gly821Arg	2461G>C	G821R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090257	GGT-CGT	Gly824Arg	2470G>C	G824R	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM060927	GGT-GAT	Gly833Asp	2498G>A	G833D	Osteogenesis imperfecta II	Aerts (2006) <i>Prenat Diagn</i> 26:394	PubMed 16566045
COL1A1	CM070734	GGT-GTT	Gly833Val	2498G>T	G833V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM952154	GGC-CGC	Gly842Arg	2524G>C	G842R	Osteogenesis imperfecta II	Culbert (1995) <i>Biochem J</i> 311:815	PubMed 7487936
COL1A1	CM070702	GGC-GAC	Gly842Asp	2525G>A	G842D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM880015	GGA-AGA	Gly845Arg	2533G>A	G845R	Osteogenesis imperfecta II	Bateman (1988) <i>J Biol Chem</i> 263:11627	PubMed 3403550
COL1A1	CM090258	GGA-CGA	Gly845Arg	2533G>C	G845R	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM090259	GGA-CGA	Gly848Arg	2542G>C	G848R	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM900326	GGC-GAC	Gly851Asp	2552G>A	G851D	Osteogenesis imperfecta II	Cohn (1990) <i>COL1A1 LSDB entry</i> Unpublished:	Source: LSDB
COL1A1	CM090262	AAT-CAT	Asn855His	2563A>C	N855H	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM910085	GGT-TGT	Gly869Cys	2605G>T	G869C	Osteogenesis imperfecta II	Steinmann (1991) <i>Biochem J</i> 279:747	PubMed 1953667
COL1A1	CM070756	GGT-AGT	Gly875Ser	2623G>A	G875S	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070757	GGT-AGT	Gly878Ser	2632G>A	G878S	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090260	GGT-AGT	Gly884Ser	2650G>A	G884S	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM070743	GGC-AGC	Gly887Ser	2659G>A	G887S	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090261	GGC-GAC	Gly896Asp	2687G>A	G896D	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM890182	GGC-TGC	Gly896Cys	2686G>T	G896C	Osteogenesis imperfecta II	Starman (1989) <i>J Clin Invest</i> 84:1206	PubMed 2794057
COL1A1	CM070698	GGA-GAA	Gly917Glu	2750G>A	G917E	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070773	GGT-TGT	Gly944Cys	2830G>T	G944C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070774	GGT-TGT	Gly947Cys	2839G>T	G947C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070689	GGG-AGG	Gly950Arg	2848G>A	G950R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM090263	GGC-GAC	Gly977Asp	2930G>A	G977D	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919

COL1A1	CM920203	GGT-GTT	Gly980Val	2939G>T	G980V	Osteogenesis imperfecta II	Bonaventure (1992) <i>Hum Genet</i> 89:640	PubMed 1511982
COL1A1	CM070712	GGC-TGC	Gly1001Cys	3001G>T	G1001C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM970350	GGT-GTT	Gly1022Val	3065G>T	G1022V	Osteogenesis imperfecta II	Korkko (1997) <i>Hum Mutat</i> 9:148	PubMed 9067755
COL1A1	CM900069	GGA-AGA	Gly1025Arg	3073G>A	G1025R	Osteogenesis imperfecta II	Wallis (1990) <i>J Biol Chem</i> 265:18628	PubMed 2211725
COL1A1	CM940293	GGC-AGC	Gly1040Ser	3118G>A	G1040S	Osteogenesis imperfecta II	Virdi (1994) <i>Hum Genet</i> 93:287	PubMed 8125479
COL1A1	CM090264	GGC-GAC	Gly1055Asp	3164G>A	G1055D	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM900070	GGC-GAC	Gly1061Asp	3182G>A	G1061D	Osteogenesis imperfecta II	Cohn (1990) <i>Am J Hum Genet</i> 46:591	PubMed 2309707
COL1A1	CM940294	GGC-AGC	Gly1061Ser	3181G>A	G1061S	Osteogenesis imperfecta II	Lightfoot (1994) <i>J Biol Chem</i> 269:30352	PubMed 7982948
COL1A1	CM070693	GGT-TGT	Gly1076Cys	3226G>T	G1076C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM890030	GGC-TGC	Gly1082Cys	3244G>T	G1082C	Osteogenesis imperfecta II	Constantinou (1989) <i>J Clin Invest</i> 83:574	PubMed 2913053
COL1A1	CM930147	GGA-GCA	Gly1088Ala	3263G>C	G1088A	Osteogenesis imperfecta II	Valli (1993) <i>Eur J Biochem</i> 211:415	PubMed 7679635
COL1A1	CM900071	GGC-AGC	Gly1091Ser	3271G>A	G1091S	Osteogenesis imperfecta II	Cohn (1990) <i>Matrix</i> 10:236A	Source: Meeting abstract
COL1A1	CM090265	GGT-AGT	Gly1094Ser	3280G>A	G1094S	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A1	CM070704	GGC-GAC	Gly1100Asp	3299G>A	G1100D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM890031	GGC-GCC	Gly1106Ala	3317G>C	G1106A	Osteogenesis imperfecta II	Lamande (1989) <i>J Biol Chem</i> 264:15809	PubMed 2777764
COL1A1	CM940295	GGC-TGC	Gly1124Cys	3370G>T	G1124C	Osteogenesis imperfecta II	Kwasaka (1994) <i>J Biochem Tokyo</i> 115:853	PubMed 7961597
COL1A1	CM890032	GGT-AGT	Gly1142Ser	3424G>A	G1142S	Osteogenesis imperfecta II	Wallis (1989) <i>Am J Hum Genet</i> 45:A228	Source: Meeting abstract
COL1A1	CM070705	GGC-GAC	Gly1145Asp	3434G>A	G1145D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM890033	GGC-GTC	Gly1151Val	3452G>T	G1151V	Osteogenesis imperfecta II	Lamande (1989) <i>J Biol Chem</i> 264:15809	PubMed 2777764
COL1A1	CM890034	GGA-AGA	Gly1154Arg	3460G>A	G1154R	Osteogenesis imperfecta II	Lamande (1989) <i>J Biol Chem</i> 264:15809	PubMed 2777764
COL1A1	CM860006	GGT-TGT	Gly1166Cys	3496G>T	G1166C	Osteogenesis imperfecta II	Cohn (1986) <i>Proc Natl Acad Sci U S A</i> 83:6045	PubMed 3016737
COL1A1	CM070694	GGT-TGT	Gly1169Cys	3505G>T	G1169C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070735	GGT-GTT	Gly1178Val	3533G>T	G1178V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM890035	GGA-GTA	Gly1184Val	3551G>T	G1184V	Osteogenesis imperfecta II	Lamande (1989) <i>J Biol Chem</i> 264:15809	PubMed 2777764
COL1A1	CM900072	GGT-GTT	Gly1187Val	3560G>T	G1187V	Osteogenesis imperfecta II	Cohn (1990) <i>Matrix</i> 10:236A	Source: Meeting abstract
COL1A1	CM045284	ATG-GTG	Met1264Val	3790A>G	M1264V	Osteogenesis imperfecta II	Symoens (2004) <i>J Med Genet</i> 41:e96	PubMed 15235039
COL1A1	CM930148	GAC-CAC	Asp1277His	3829G>C	D1277H	Osteogenesis imperfecta II	Chessler (1993) <i>J Biol Chem</i> 268:18218	PubMed 8349697
COL1A1	CM930149	TGG-TGT	Trp1312Cys	3936G>T	W1312C	Osteogenesis imperfecta II	Bateman (1993) <i>Am J Med Genet</i> 45:233	PubMed 8456808
COL1A1	CM930150	CTG-CGG	Leu1388Arg	4163T>G	L1388R	Osteogenesis imperfecta II	Chessler (1993) <i>J Biol Chem</i> 268:18218	PubMed 8349697
COL1A1	CM062524	GAT-AAT	Asp1413Asn	4237G>A	D1413N	Osteogenesis imperfecta II	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM020370	GAT-TAT	Asp1441Tyr	4321G>T	D1441Y	Osteogenesis imperfecta II	Pace (2002) <i>J Med Genet</i> 39:23	PubMed 11826020
COL1A1	CM070677	GGA-AGA	Gly254Arg	760G>A	G254R	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070714	GGT-AGT	Gly368Ser	1102G>A	G368S	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070731	GGT-GTT	Gly380Val	1139G>T	G380V	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070742	GGC-AGC	Gly755Ser	2263G>A	G755S	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070759	GGT-AGT	Gly1052Ser	3154G>A	G1052S	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM055137	GGC-GAC	Gly827Asp	2480G>A	G827D	Osteogenesis imperfecta IIA	Reis (2005) <i>J Appl Genet</i> 46:105	PubMed 15741671
COL1A1	CM970355	GGT-GAT	Gly1172Asp	3515G>A	G1172D	Osteogenesis imperfecta IIC	Mottes (1997) <i>Hum Mutat</i> 12:71	PubMed 10627137
COL1A1	CM116611	GCC-GTC	Ala1387Val	4160C>T	A1387V	Osteogenesis imperfecta IIC	Takagi (2011) <i>Am J Med Genet A</i> 155:2269	PubMed 21834035
COL1A1	CM070747	GGG-AGG	Gly248Arg	742G>A	G248R	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM145061	GGT-CGT	Gly251Arg	751G>C	G251R	Osteogenesis imperfecta III	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929

COL1A1	CM012742	GGA-GAA	Gly254Glu	761G>A	G254E	Osteogenesis imperfecta III	Cabral (2001) <i>Mol Genet Metab</i> 72:326	PubMed 11286507
COL1A1	CM070750	GGT-AGT	Gly284Ser	850G>A	G284S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070719	GGT-GAT	Gly299Asp	896G>A	G299D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070720	GGT-GAT	Gly311Asp	932G>A	G311D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070695	GGA-GAA	Gly323Glu	968G>A	G323E	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070721	GGT-GAT	Gly326Asp	977G>A	G326D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209 Krakow (1996) <i>COL1A1 LSDB entry</i> Meeting abstract:	PubMed 17078022 Source: LSDB
COL1A1	CM962767	GGG-GCG	Gly332Ala	995G>C	G332A	Osteogenesis imperfecta III		
COL1A1	CM910079	GGG-AGG	Gly332Arg	994G>A	G332R	Osteogenesis imperfecta III	Pruchno (1991) <i>Hum Genet</i> 87:33	PubMed 2037280
COL1A1	CM070713	GGT-AGT	Gly353Ser	1057G>A	G353S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070751	GGT-AGT	Gly371Ser	1111G>A	G371S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM145062	GGT-AGT	Gly380Ser	1138G>A	G380S	Osteogenesis imperfecta III	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A1	CM070752	GGT-AGT	Gly389Ser	1165G>A	G389S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070753	GGT-AGT	Gly395Ser	1183G>A	G395S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070754	GGT-AGT	Gly404Ser	1210G>A	G404S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM011291	GGT-AGT	Gly407Ser	1219G>A	G407S	Osteogenesis imperfecta III	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A1	CM070740	GGC-AGC	Gly410Ser	1228G>A	G410S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070725	GGT-GCT	Gly413Ala	1238G>C	G413A	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070737	GGA-AGA	Gly464Arg	1390G>A	G464R	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070699	GGA-GCA	Gly482Ala	1445G>C	G482A	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM034878	GGT-AGT	Gly689Ser	2065G>A	G689S	Osteogenesis imperfecta III	Galicka (2003) <i>Mol Cell Biochem</i> 248:49	PubMed 12870654
COL1A1	CM070690	GGT-AGT	Gly701Ser	2101G>A	G701S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM890181	GGC-TGC	Gly704Cys	2110G>T	G704C	Osteogenesis imperfecta III	Starman (1989) <i>J Clin Invest</i> 84:1206	PubMed 2794057
COL1A1	CM011292	GGC-AGC	Gly704Ser	2110G>A	G704S	Osteogenesis imperfecta III	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A1	CM930145	GGT-AGT	Gly719Ser	2155G>A	G719S	Osteogenesis imperfecta III	Mackay (1993) <i>Hum Mol Genet</i> 2:1155	PubMed 7691343
COL1A1	CM940291	GGC-AGC	Gly767Ser	2299G>A	G767S	Osteogenesis imperfecta III	Forlino (1994) <i>Hum Mol Genet</i> 3:2201	PubMed 7881420
COL1A1	CM055138	GGT-AGT	Gly770Ser	2308G>A	G770S	Osteogenesis imperfecta III	Reis (2005) <i>J Appl Genet</i> 46:105	PubMed 15741671
COL1A1	CM043755	GGC-AGC	Gly773Ser	2317G>A	G773S	Osteogenesis imperfecta III	Chamberlain (2004) <i>Science</i> 303:1198	PubMed 14976317
COL1A1	CM970349	GGC-AGC	Gly821Ser	2461G>A	G821S	Osteogenesis imperfecta III	Lund (1997) <i>Hum Mutat</i> 9:378	PubMed 9101304
COL1A1	CM070710	GGC-GCC	Gly836Ala	2507G>C	G836A	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM011293	GGC-TGC	Gly866Cys	2596G>T	G866C	Osteogenesis imperfecta III	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A1	CM990379	GGC-AGC	Gly866Ser	2596G>A	G866S	Osteogenesis imperfecta III	Lund (1999) <i>Hum Mutat</i> 13:503	PubMed 10408781
COL1A1	CM145064	GGT-AGT	Gly911Ser	2731G>A	G911S	Osteogenesis imperfecta III	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A1	CM070703	GGC-GAC	Gly962Asp	2885G>A	G962D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM890029	GGT-AGT	Gly1022Ser	3064G>A	G1022S	Osteogenesis imperfecta III	Pack (1989) <i>J Biol Chem</i> 264:19694	PubMed 2511192
COL1A1	CM973112	GGT-TGT	Gly1046Cys	3136G>T	G1046C	Osteogenesis imperfecta III	Pepin (1997) <i>Prenat Diagn</i> 17:559	PubMed 9203215
COL1A1	CM070758	GGT-AGT	Gly1046Ser	3136G>A	G1046S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM970351	GGT-AGT	Gly1049Ser	3145G>A	G1049S	Osteogenesis imperfecta III	Lund (1997) <i>Hum Mutat</i> 9:378	PubMed 9101304
COL1A1	CM970353	GGT-AGT	Gly1076Ser	3226G>A	G1076S	Osteogenesis imperfecta III	Lund (1997) <i>Hum Mutat</i> 9:378	PubMed 9101304
COL1A1	CM070746	GGC-TGC	Gly1145Cys	3433G>T	G1145C	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM123295	GCT-GTT	Ala1174Val	3521C>T	A1174V	Osteogenesis imperfecta III	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A1	CM070775	GGT-TGT	Gly1175Cys	3523G>T	G1175C	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070760	GGT-AGT	Gly1175Ser	3523G>A	G1175S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM910086	GGC-AGC	Gly1181Ser	3541G>A	G1181S	Osteogenesis imperfecta III	Pruchno (1991) <i>Hum Genet</i> 87:33	PubMed 2037280

COL1A1	CM900073	GGT-AGT	Gly1187Ser	3559G>A	G1187S	Osteogenesis imperfecta III	Cohn (1990) <i>Matrix</i> 10:236A	Source: Meeting abstract
COL1A1	CM088316	ACC-ATC	Thr1298Ile	3893C>T	T1298I	Osteogenesis imperfecta III	Obafemi (2008) <i>Am J Med Genet A</i> 146A:2725	PubMed 18798308
COL1A1	CM145065	GTC-GAC	Val1410Asp	4229T>A	V1410D	Osteogenesis imperfecta III	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A1	CM960324	CTG-CCG	Leu1464Pro	4391T>C	L1464P	Osteogenesis imperfecta III	Oliver (1996) <i>Hum Mutat</i> 7:318	PubMed 8723681
COL1A1	CM070681	GGT-CGT	Gly203Arg	607G>C	G203R	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070729	GGT-GTT	Gly269Val	806G>T	G269V	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM930141	GGT-TGT	Gly353Cys	1057G>T	G353C	III/IV	Wirtz (1993) <i>Connect Tissue Res</i> 29:1	PubMed 8339541
COL1A1	CM070724	GGT-GCT	Gly365Ala	1094G>C	G365A	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070706	GGC-GCC	Gly377Ala	1130G>C	G377A	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070739	GGC-AGC	Gly383Ser	1147G>A	G383S	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
						Osteogenesis imperfecta		
COL1A1	CM942085	GGT-TGT	Gly389Cys	1165G>T	G389C	III/IV	Wilcox (1994) <i>COL1A1 LSDB entry</i> Meeting abstract: Source: LSDB	
COL1A1	CM070682	GGT-TGT	Gly398Cys	1192G>T	G398C	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
						Osteogenesis imperfecta		
COL1A1	CM942086	GGT-TGT	Gly404Cys	1210G>T	G404C	III/IV	Wilcox (1994) <i>COL1A1 LSDB entry</i> Meeting abstract: Source: LSDB	
COL1A1	CM070707	GGC-GCC	Gly422Ala	1265G>C	G422A	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM910082	GGC-TGC	Gly593Cys	1777G>T	G593C	III/IV	Nicholls (1991) <i>J Med Genet</i> 28:757	PubMed 1770532
COL1A1	CM920201	GGC-AGC	Gly593Ser	1777G>A	G593S	III/IV	Bateman (1992) <i>Biochem J</i> 288:131	PubMed 1445258
COL1A1	CM973111	GGT-AGT	Gly779Ser	2335G>A	G779S	III/IV	Lund (1997) <i>Acta Paediatr</i> 86:711	PubMed 9240878
COL1A1	CM070687	GGC-AGC	Gly788Ser	2362G>A	G788S	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070772	GGT-TGT	Gly857Cys	2569G>T	G857C	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070700	GGA-GCA	Gly860Ala	2579G>C	G860A	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM062539	GGT-CGT	Gly197Arg	589G>C	G197R	Osteogenesis imperfecta IV	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM123293	GGA-GAA	Gly212Glu	635G>A	G212E	Osteogenesis imperfecta IV	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A1	CM070717	GGT-GAT	Gly239Asp	716G>A	G239D	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070761	GGT-CGT	Gly275Arg	823G>C	G275R	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070749	GGT-AGT	Gly281Ser	841G>A	G281S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM011289	GGC-AGC	Gly305Ser	913G>A	G305S	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A1	CM011290	GGT-CGT	Gly311Arg	931G>C	G311R	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A1	CM062526	GGC-TGC	Gly338Cys	1012G>T	G338C	Osteogenesis imperfecta IV	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A1	CM910080	GGT-TGT	Gly356Cys	1066G>T	G356C	Osteogenesis imperfecta IV	Valli (1991) <i>J Biol Chem</i> 266:1872	PubMed 1988452
COL1A1	CM070763	GGT-TGT	Gly380Cys	1138G>T	G380C	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM980391	GGT-GCT	Gly398Ala	1193G>C	G398A	Osteogenesis imperfecta IV	Sarafova (1998) <i>Hum Mutat</i> 11:395	PubMed 9600458
COL1A1	CM070764	GGT-TGT	Gly407Cys	1219G>T	G407C	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM123294	GGA-GCA	Gly470Ala	1409G>C	G470A	Osteogenesis imperfecta IV	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A1	CM070767	GGT-TGT	Gly491Cys	1471G>T	G491C	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM980392	GGT-TGT	Gly527Cys	1579G>T	G527C	Osteogenesis imperfecta IV	Sarafova (1998) <i>Hum Mutat</i> 11:395	PubMed 9600458
COL1A1	CM962604	GGT-TGT	Gly530Cys	1588G>T	G530C	Osteogenesis imperfecta IV	Cohen (1996) <i>COL1A1 LSDB entry</i> Meeting abstract: Source: LSDB	
COL1A1	CM920200	GGT-AGT	Gly530Ser	1588G>A	G530S	Osteogenesis imperfecta IV	Bateman (1992) <i>Biochem J</i> 288:131	PubMed 1445258
COL1A1	CM900065	GGT-TGT	Gly560Cys	1678G>T	G560C	Osteogenesis imperfecta IV	Byers (1990) <i>Trends Genet</i> 6:293	PubMed 2238087
COL1A1	CM930144	GGT-AGT	Gly560Ser	1678G>A	G560S	Osteogenesis imperfecta IV	Mackay (1993) <i>Hum Mol Genet</i> 2:1155	PubMed 7691343
COL1A1	CM070726	GGT-GCT	Gly587Ala	1760G>C	G587A	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022

COL1A1	CM973109	GGT-AGT	Gly626Ser	1876G>A	G626S	Osteogenesis imperfecta IV	Pepin (1997) <i>Prenat Diagn</i> 17:559	PubMed 9203215
COL1A1	CM980393	GGT-TGT	Gly701Cys	2101G>T	G701C	Osteogenesis imperfecta IV	Sarafova (1998) <i>Hum Mutat</i> 11:395	PubMed 9600458
COL1A1	CM145063	GCC-GTC	Ala723Val	2168C>T	A723V	Osteogenesis imperfecta IV	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A1	CM070745	GGC-TGC	Gly758Cys	2272G>T	G758C	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM042009	GGC-TGC	Gly788Cys	2362G>T	G788C	Osteogenesis imperfecta IV	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A1	CM890028	GGT-AGT	Gly1010Ser	3028G>A	G1010S	Osteogenesis imperfecta IV	Marini (1989) <i>J Biol Chem</i> 264:11893	PubMed 2745420
COL1A1	CM070691	GGT-AGT	Gly1028Ser	3082G>A	G1028S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM970352	GGC-AGC	Gly1058Ser	3172G>A	G1058S	Osteogenesis imperfecta IV	Lund (1997) <i>Hum Mutat</i> 9:378	PubMed 9101304
COL1A1	CM070728	GGT-GCT	Gly1064Ala	3191G>C	G1064A	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070744	GGC-AGC	Gly1085Ser	3253G>A	G1085S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM070692	GGT-AGT	Gly1169Ser	3505G>A	G1169S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A1	CM062535	TGG-TAG	Trp1269Term	3806G>A	W1269*	Osteogenesis imperfecta IV	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM062541	GGT-AGT	Gly193Ser	577G>A	G193S	Osteogenesis imperfecta	Venturi (2006) <i>Clin Genet</i> 70:131	PubMed 16879195
COL1A2	CM137632	GGT-AGT	Gly259Ser	775G>A	G259S	Osteogenesis imperfecta	Adur (2013) <i>PLoS One</i> 8:e69186	PubMed 23869235
COL1A2	CM011298	GGT-TGT	Gly328Cys	982G>T	G328C	Osteogenesis imperfecta	Trummer (2001) <i>Clin Genet</i> 59:338	PubMed 11359465
COL1A2	CM072955	GGT-GTT	Gly346Val	1037G>T	G346V	Osteogenesis imperfecta	Parmar (2007) <i>J Neurosurg</i> 106:57	PubMed 17233315
COL1A2	CM1311796	GGT-GAT	Gly439Asp	1316G>A	G439D	Osteogenesis imperfecta	Reuter (2013) <i>Eur J Med Genet</i> 56:669	PubMed 24140640
COL1A2	CM1313963	GGA-AGA	Gly487Arg	1459G>A	G487R	Osteogenesis imperfecta	McInerney-Leo (2013) <i>Bonekey Rep</i> 2:456	PubMed 24501682
COL1A2	CM980394	GGT-GAT	Gly511Asp	1532G>A	G511D	Osteogenesis imperfecta	Forlino (1998) <i>Matrix Biol</i> 17:575	PubMed 9923651
COL1A2	CM062548	GGT-GAT	Gly625Asp	1874G>A	G625D	Osteogenesis imperfecta	Venturi (2006) <i>Clin Genet</i> 70:131	PubMed 16879195
COL1A2	CM062558	GGT-TGT	Gly754Cys	2260G>T	G754C	Osteogenesis imperfecta	Venturi (2006) <i>Clin Genet</i> 70:131	PubMed 16879195
COL1A2	CM110801	GGT-GAT	Gly787Asp	2360G>A	G787D	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A2	CM117850	CCT-TCT	Pro810Ser	2428C>T	P810S	Osteogenesis imperfecta	Fuccio (2011) <i>J Mol Diagn</i> epub:epub	PubMed 21884818
COL1A2	CM970358	GGC-AGC	Gly841Ser	2521G>A	G841S	Osteogenesis imperfecta	De Paepe (1997) <i>Hum Genet</i> 99:478	PubMed 9099837
COL1A2	CM062559	GGT-TGT	Gly925Cys	2773G>T	G925C	Osteogenesis imperfecta	Venturi (2006) <i>Clin Genet</i> 70:131	PubMed 16879195
COL1A2	CM062551	GGT-GTT	Gly991Val	2972G>T	G991V	Osteogenesis imperfecta	Venturi (2006) <i>Clin Genet</i> 70:131	PubMed 16879195
COL1A2	CM110802	GGC-GAC	Gly994Asp	2981G>A	G994D	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A2	CM110803	GGA-CGA	Gly1024Arg	3070G>C	G1024R	Osteogenesis imperfecta	Pyott (2011) <i>Genet Med</i> 13:125	PubMed 21239989
COL1A2	CM112879	GGT-GCT	Gly1102Ala	3305G>C	G1102A	Osteogenesis imperfecta	Xu (2011) <i>J Genet Genomics</i> 38:149	PubMed 21530898
COL1A2	CM112880	TAC-TGC	Tyr1117Cys	3350A>G	Y1117C	Osteogenesis imperfecta	Xu (2011) <i>J Genet Genomics</i> 38:149	PubMed 21530898
COL1A2	CM114192	GCT-ACT	Ala1119Thr	3355G>A	A1119T	Osteogenesis imperfecta	Lindahl (2011) <i>Hum Mutat</i> 32:598	PubMed 21344539
COL1A2	CM114194	GAC-GCC	Asp1120Ala	3359A>C	D1120A	Osteogenesis imperfecta	Lindahl (2011) <i>Hum Mutat</i> 32:598	PubMed 21344539
COL1A2	CM137633	AGA-AAA	Arg1156Lys	3467G>A	R1156K	Osteogenesis imperfecta	Adur (2013) <i>PLoS One</i> 8:e69186	PubMed 23869235
COL1A2	CM1311797	CAG-TAG	Gln1354Term	4060C>T	Q1354*	Osteogenesis imperfecta	Reuter (2013) <i>Eur J Med Genet</i> 56:669	PubMed 24140640
COL1A2	CM062540	TAA-CAA	Term1367Gln	4099T>C	*1367Q	Osteogenesis imperfecta	Venturi (2006) <i>Clin Genet</i> 70:131	PubMed 16879195
COL1A2	CM070817	GGT-GAT	Gly118Asp	353G>A	G118D	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070834	GGT-GTT	Gly166Val	497G>T	G166V	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM960325	GGT-GAT	Gly211Asp	632G>A	G211D	Osteogenesis imperfecta I	Zhuang (1996) <i>Hum Mutat</i> 7:89	PubMed 8829649
COL1A2	CM062556	GGT-CGT	Gly247Arg	739G>C	G247R	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM141560	GGC-GAC	Gly292Asp	875G>A	G292D	Osteogenesis imperfecta I	Fratzl-Zelman (2014) <i>Bone</i> 60:122	PubMed 24296239
COL1A2	CM070815	GGT-CGT	Gly310Arg	928G>C	G310R	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM1211689	GGC-AGC	Gly316Ser	946G>A	G316S	Osteogenesis imperfecta I	Valentina Gentile (2012) <i>Hum Mutat</i> 33:1697	PubMed 22753364
COL1A2	CM062553	GGG-AGG	Gly319Arg	955G>A	G319R	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509

COL1A2	CM042012	GGC-AGC	Gly322Ser	964G>A	G322S	Osteogenesis imperfecta I	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A2	CM970356	GGT-TGT	Gly337Cys	1009G>T	G337C	Osteogenesis imperfecta I	Korkko (1997) <i>Hum Mutat</i> 9:148	PubMed 9067755
COL1A2	CM960326	GGT-AGT	Gly337Ser	1009G>A	G337S	Osteogenesis imperfecta I	Zhuang (1996) <i>Hum Mutat</i> 7:89	PubMed 8829649
COL1A2	CM070790	GGA-GAA	Gly484Glu	1451G>A	G484E	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070842	GGT-GTT	Gly661Val	1982G>T	G661V	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM011307	GGT-CGT	Gly676Arg	2026G>C	G676R	Osteogenesis imperfecta I	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM062557	GGT-TGT	Gly733Cys	2197G>T	G733C	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM910089	GGT-TGT	Gly736Cys	2206G>T	G736C	Osteogenesis imperfecta I	Wenstrup (1991) <i>J Biol Chem</i> 266:2590	PubMed 1990009
COL1A2	CM145071	GGT-AGT	Gly748Ser	2242G>A	G748S	Osteogenesis imperfecta I	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A2	CM070865	GGT-AGT	Gly766Ser	2296G>A	G766S	Osteogenesis imperfecta I	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM910091	GGT-CGT	Gly784Arg	2350G>C	G784R	Osteogenesis imperfecta I	Tsuneyoshi (1991) <i>J Biol Chem</i> 266:15608	PubMed 1874719
COL1A2	CM1110848	GGC-CGC	Gly805Arg	2413G>C	G805R	Osteogenesis imperfecta I	Swinnen (2011) <i>Orphanet J Rare Dis</i> 6:88	PubMed 22206639
COL1A2	CM1110831	GGT-GCT	Gly811Ala	2432G>C	G811A	Osteogenesis imperfecta I	Swinnen (2011) <i>Orphanet J Rare Dis</i> 6:88	PubMed 22206639
COL1A2	HM090040	GGT-TGT	Gly823Cys	2467G>T	G823C	Osteogenesis imperfecta I	Lin (2010) <i>Hum Genet</i> 127:467	PubMed 21488276
COL1A2	CM960328	GGT-AGT	Gly835Ser	2503G>A	G835S	Osteogenesis imperfecta I	Zhuang (1996) <i>Hum Mutat</i> 7:89	PubMed 8829649
COL1A2	CM042017	GGC-AGC	Gly859Ser	2575G>A	G859S	Osteogenesis imperfecta I	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A2	CM042018	GGG-CGG	Gly904Arg	2710G>C	G904R	Osteogenesis imperfecta I	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A2	CM1110799	GGA-AGA	Gly916Arg	2746G>A	G916R	Osteogenesis imperfecta I	Swinnen (2011) <i>Orphanet J Rare Dis</i> 6:88	PubMed 22206639
COL1A2	HM090041	GGT-GAT	Gly934Asp	2801G>A	G934D	Osteogenesis imperfecta I	Lin (2010) <i>Hum Genet</i> 127:468	PubMed 21488295
COL1A2	CM890036	GGT-GAT	Gly997Asp	2990G>A	G997D	Osteogenesis imperfecta I	Baldwin (1989) <i>J Biol Chem</i> 264:3002	PubMed 2914942
COL1A2	CM042019	GGT-AGT	Gly1102Ser	3304G>A	G1102S	Osteogenesis imperfecta I	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A2	CM123314	GGT-AGT	Gly1105Ser	3313G>A	G1105S	Osteogenesis imperfecta I	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A2	CM062560	TGT-TAT	Cys1195Tyr	3584G>A	C1195Y	Osteogenesis imperfecta I	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM123315	TTC-TTA	Phe1365Leu	4095C>A	F1365L	Osteogenesis imperfecta I	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A2	CM070794	GGA-GTA	Gly190Val	569G>T	G190V	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070864	GGT-AGT	Gly661Ser	1981G>A	G661S	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070843	GGT-GTT	Gly853Val	2558G>T	G853V	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070801	GGC-GAC	Gly973Asp	2918G>A	G973D	Osteogenesis imperfecta I/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM034879	CAC-GAC	His182Asp	544C>G	H182D	Osteogenesis imperfecta II	Galicka (2003) <i>Acta Biochim Pol</i> 50:481	PubMed 12833172
COL1A2	CM090272	CGT-TGT	Arg234Cys	700C>T	R234C	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A2	CM062544	GGC-GAC	Gly253Asp	758G>A	G253D	Osteogenesis imperfecta II	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM090273	GGT-CGT	Gly283Arg	847G>C	G283R	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A2	CM090274	GGG-GAG	Gly397Glu	1190G>A	G397E	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A2	CM983821	GGA-GTA	Gly409Val	1226G>T	G409V	Osteogenesis imperfecta II	Mottes (1997) <i>Hum Mutat</i> 12:71	PubMed 10627137
COL1A2	CM930151	GGA-GAA	Gly433Glu	1298G>A	G433E	Osteogenesis imperfecta II	Rose (1993) <i>Hum Mol Genet</i> 2:2175	PubMed 7906591
COL1A2	CM070869	GGT-CGT	Gly445Arg	1333G>C	G445R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070872	GGT-TGT	Gly454Cys	1360G>T	G454C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070806	GGC-GTC	Gly499Val	1496G>T	G499V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070838	GGT-GTT	Gly511Val	1532G>T	G511V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070781	GGT-TGT	Gly541Cys	1621G>T	G541C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070839	GGT-GTT	Gly544Val	1631G>T	G544V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM920205	GGT-CGT	Gly547Arg	1639G>C	G547R	Osteogenesis imperfecta II	Bateman (1992) <i>Hum Mutat</i> 1:55	PubMed 1284475
COL1A2	CM070800	GGC-GAC	Gly553Asp	1658G>A	G553D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022

COL1A2	CM920206	GGT-TGT	Gly562Cys	1684G>T	G562C	Osteogenesis imperfecta II	Edwards (1992) <i>Hum Mutat</i> 1:47	PubMed 1301191
COL1A2	CM090275	GGT-GTT	Gly562Val	1685G>T	G562V	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A2	CM900327	GGT-CGT	Gly586Arg	1756G>C	G586R	Osteogenesis imperfecta II	Bateman (1990) <i>COL1A2 LSDB entry</i> Unpublished:	Source: LSDB
COL1A2	CM940297	GGT-AGT	Gly592Ser	1774G>A	G592S	Osteogenesis imperfecta II	Rose (1994) <i>Hum Genet</i> 94:497	PubMed 7959683
COL1A2	CM070782	GGT-TGT	Gly595Cys	1783G>T	G595C	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM880017	GGT-GAT	Gly637Asp	1910G>A	G637D	Osteogenesis imperfecta II	Bonadio (1988) <i>Coll Relat Res</i> 8:506A	Source: Meeting abstract
COL1A2	CM070792	GGA-GAA	Gly640Glu	1919G>A	G640E	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM920207	GGC-GAC	Gly670Asp	2009G>A	G670D	Osteogenesis imperfecta II	Niyibizi (1992) <i>J Biol Chem</i> 267:23108	PubMed 1385413
COL1A2	CM070829	GGT-GAT	Gly712Asp	2135G>A	G712D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM931214	GGT-GAT	Gly715Asp	2144G>A	G715D	Osteogenesis imperfecta II	Byers (1993) <i>COL1A2 LSDB entry</i> Unpublished:	Source: LSDB
COL1A2	CM070787	GGT-AGT	Gly718Ser	2152G>A	G718S	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070853	GGC-AGC	Gly721Ser	2161G>A	G721S	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM127973	GGT-CGT	Gly730Arg	2188G>C	G730R	Osteogenesis imperfecta II	Hachiya (2012) <i>Neuropediatrics</i> 43:225	PubMed 22911485
COL1A2	CM090276	GGA-CGA	Gly739Arg	2215G>C	G739R	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A2	CM090277	GGT-GTT	Gly748Val	2243G>T	G748V	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A2	CM970357	GGT-TGT	Gly787Cys	2359G>T	G787C	Osteogenesis imperfecta II	Mottes (1997) <i>Hum Mutat</i> 12:71	PubMed 10627137
COL1A2	CM940298	GGT-GAT	Gly790Asp	2369G>A	G790D	Osteogenesis imperfecta II	Cohen-Solal (1994) <i>J Biol Chem</i> 269:14751	PubMed 8182080
COL1A2	CM077241	GGT-TGT	Gly796Cys	2386G>T	G796C	Osteogenesis imperfecta II	Kataoka (2007) <i>Pediatr Int</i> 49:564	PubMed 17875077
COL1A2	CM930154	GGT-AGT	Gly796Ser	2386G>A	G796S	Osteogenesis imperfecta II	Wang (1993) <i>J Biol Chem</i> 268:25162	PubMed 7693712
COL1A2	CM070816	GGT-CGT	Gly847Arg	2539G>C	G847R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM090278	GGA-GTA	Gly856Val	2567G>T	G856V	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A2	CM930155	GGC-TGC	Gly877Cys	2629G>T	G877C	Osteogenesis imperfecta II	Fertala (1993) <i>Biochem J</i> 289:195	PubMed 8424758
COL1A2	CM900075	GGT-GAT	Gly895Asp	2684G>A	G895D	Osteogenesis imperfecta II	Grange (1990) <i>Nucleic Acids Res</i> 18:4227	PubMed 1696002
COL1A2	CM070811	GGA-AGA	Gly946Arg	2836G>A	G946R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM090279	GGT-GAT	Gly955Asp	2864G>A	G955D	Osteogenesis imperfecta II	Bodian (2009) <i>Hum Mol Genet</i> 18:463	PubMed 18996919
COL1A2	CM930156	GGT-AGT	Gly955Ser	2863G>A	G955S	Osteogenesis imperfecta II	Lamande (1989) <i>J Biol Chem</i> 264:15809	PubMed 2777764
COL1A2	CM070830	GGT-GAT	Gly970Asp	2909G>A	G970D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM062550	GGT-GAT	Gly982Asp	2945G>A	G982D	Osteogenesis imperfecta II	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM070844	GGT-GTT	Gly982Val	2945G>T	G982V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM062545	GGC-GAC	Gly1003Asp	3008G>A	G1003D	Osteogenesis imperfecta II	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM070849	GGA-AGA	Gly1027Arg	3079G>A	G1027R	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070793	GGA-GAA	Gly1027Glu	3080G>A	G1027E	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070831	GGT-GAT	Gly1039Asp	3116G>A	G1039D	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM900076	GGT-GAT	Gly1066Asp	3197G>A	G1066D	Osteogenesis imperfecta II	Byers (1990) <i>Trends Genet</i> 6:293	PubMed 2238087
COL1A2	CM070846	GGT-GTT	Gly1084Val	3251G>T	G1084V	Osteogenesis imperfecta II	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	HM060012	GGT-GAT	Gly1111Asp	3332G>A	G1111D	Osteogenesis imperfecta II	Tinkle (2006) <i>Hum Genet</i> 119:677	Source: Online submission
COL1A2	CM070836	GGT-GTT	Gly286Val	857G>T	G286V	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070809	GGG-GAG	Gly385Glu	1154G>A	G385E	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070796	GGA-GTA	Gly388Val	1163G>T	G388V	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070798	GGC-GAC	Gly418Asp	1253G>A	G418D	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070812	GGC-AGC	Gly418Ser	1252G>A	G418S	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070789	GGA-GAA	Gly448Glu	1343G>A	G448E	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022

COL1A2	CM070799	GGC-GAC	Gly478Asp	1433G>A	G478D	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070805	GGC-GTC	Gly478Val	1433G>T	G478V	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070808	GGC-GTC	Gly1081Val	3242G>T	G1081V	II/III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070820	GGT-GAT	Gly193Asp	578G>A	G193D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070797	GGC-GAC	Gly256Asp	767G>A	G256D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM973113	GGT-GAT	Gly265Asp	794G>A	G265D	Osteogenesis imperfecta III	Lund (1997) <i>Acta Paediatr</i> 86:711	PubMed 9240878
COL1A2	CM070835	GGT-GTT	Gly274Val	821G>T	G274V	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM088317	GGT-GTT	Gly280Val	839G>T	G280V	Osteogenesis imperfecta III	Obafemi (2008) <i>Am J Med Genet A</i> 146A:2725	PubMed 18798308
COL1A2	CM070803	GGC-GTC	Gly289Val	866G>T	G289V	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070795	GGA-GTA	Gly295Val	884G>T	G295V	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070785	GGC-CGC	Gly316Arg	946G>C	G316R	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070824	GGT-GAT	Gly328Asp	983G>A	G328D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM950287	GGT-AGT	Gly328Ser	982G>A	G328S	Osteogenesis imperfecta III	Rose (1995) <i>Hum Genet</i> 95:215	PubMed 7860070
COL1A2	CM990380	GGC-GAC	Gly331Asp	992G>A	G331D	Osteogenesis imperfecta III	Lund (1999) <i>Hum Mutat</i> 13:503	PubMed 10408781
COL1A2	CM070804	GGC-GTC	Gly331Val	992G>T	G331V	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070860	GGT-AGT	Gly340Ser	1018G>A	G340S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070861	GGT-AGT	Gly346Ser	1036G>A	G346S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM973114	GGT-TGT	Gly358Cys	1072G>T	G358C	Osteogenesis imperfecta III	Lund (1997) <i>Acta Paediatr</i> 86:711	PubMed 9240878
COL1A2	CM062542	GGT-AGT	Gly358Ser	1072G>A	G358S	Osteogenesis imperfecta III	Lee (2006) <i>Hum Mutat</i> 27:599	PubMed 16705691
COL1A2	CM145066	GGG-GAG	Gly367Glu	1100G>A	G367E	Osteogenesis imperfecta III	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A2	CM001074	GGG-TGG	Gly367Trp	1099G>T	G367W	Osteogenesis imperfecta III	Nuytinck (2000) <i>J Med Genet</i> 37:371	PubMed 10807697
COL1A2	CM973115	GGT-AGT	Gly370Ser	1108G>A	G370S	Osteogenesis imperfecta III	Lund (1997) <i>Acta Paediatr</i> 86:711	PubMed 9240878
COL1A2	CM1211690	GGG-AGG	Gly385Arg	1153G>A	G385R	Osteogenesis imperfecta III	Valentina Gentile (2012) <i>Hum Mutat</i> 33:1697	PubMed 22753364
COL1A2	CM070784	GGC-AGC	Gly391Ser	1171G>A	G391S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM960327	GGC-AGC	Gly460Ser	1378G>A	G460S	Osteogenesis imperfecta III	Zhuang (1996) <i>Hum Mutat</i> 7:89	PubMed 8829649
COL1A2	CM070825	GGT-GAT	Gly508Asp	1523G>A	G508D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM942087	GGT-CGT	Gly517Arg	1549G>C	G517R	Osteogenesis imperfecta III	Sztrolovics (1994) <i>Bone</i> 15:321	PubMed 7520724
COL1A2	CM133814	GGT-GAT	Gly517Asp	1550G>A	G517D	Osteogenesis imperfecta III	Majka (2013) <i>Open J Genet</i> 3:49	PubMed: No PubMed ID
COL1A2	CM145068	GGT-AGT	Gly517Ser	1549G>A	G517S	Osteogenesis imperfecta III	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A2	CM011301	GGA-GAA	Gly526Glu	1577G>A	G526E	Osteogenesis imperfecta III	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM145069	GGC-GTC	Gly553Val	1658G>T	G553V	Osteogenesis imperfecta III	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A2	CM070841	GGT-GTT	Gly646Val	1937G>T	G646V	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM011306	GGT-GAT	Gly664Asp	1991G>A	G664D	Osteogenesis imperfecta III	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM062549	GGT-GAT	Gly676Asp	2027G>A	G676D	Osteogenesis imperfecta III	Lee (2006) <i>Hum Mutat</i> 27:599	PubMed 16705691
COL1A2	CM070827	GGT-GAT	Gly682Asp	2045G>A	G682D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070828	GGT-GAT	Gly700Asp	2099G>A	G700D	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM973118	GGT-AGT	Gly712Ser	2134G>A	G712S	Osteogenesis imperfecta III	Lund (1997) <i>Acta Paediatr</i> 86:711	PubMed 9240878
COL1A2	CM070776	GGA-AGA	Gly739Arg	2215G>A	G739R	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070810	GGG-GAG	Gly745Glu	2234G>A	G745E	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM145070	GGT-GTT	Gly751Val	2252G>T	G751V	Osteogenesis imperfecta III	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A2	CM070807	GGC-GTC	Gly757Val	2270G>T	G757V	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070866	GGT-AGT	Gly784Ser	2350G>A	G784S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070848	GGA-AGA	Gly793Arg	2377G>A	G793R	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022

COL1A2	CM062554	GGT-AGT	Gly820Ser	2458G>A	G820S	Osteogenesis imperfecta III	Lee (2006) <i>Hum Mutat</i> 27:599	PubMed 16705691
COL1A2	HM090039	GGT-GAT	Gly853Asp	2558G>A	G853D	Osteogenesis imperfecta III	Lin (2010) <i>Hum Genet</i> 127:467	PubMed 21488231
COL1A2	CM062552	GGA-AGA	Gly856Arg	2566G>A	G856R	Osteogenesis imperfecta III	Lee (2006) <i>Hum Mutat</i> 27:599	PubMed 16705691
COL1A2	CM940299	GGT-AGT	Gly949Ser	2845G>A	G949S	Osteogenesis imperfecta III	Rose (1994) <i>Hum Mutat</i> 3:391	PubMed 8081394
COL1A2	CM070873	GGT-TGT	Gly955Cys	2863G>T	G955C	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM990382	GGC-GTC	Gly973Val	2918G>T	G973V	Osteogenesis imperfecta III	Lund (1999) <i>Hum Mutat</i> 13:503	PubMed 10408781
COL1A2	CM070845	GGT-GTT	Gly988Val	2963G>T	G988V	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070813	GGG-CGG	Gly1015Arg	3043G>C	G1015R	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070870	GGT-CGT	Gly1036Arg	3106G>C	G1036R	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070780	GGT-AGT	Gly1042Ser	3124G>A	G1042S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070867	GGT-AGT	Gly1051Ser	3151G>A	G1051S	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070874	GGT-TGT	Gly1057Cys	3169G>T	G1057C	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM062546	GGC-GAC	Gly1087Asp	3260G>A	G1087D	Osteogenesis imperfecta III	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM091167	GGC-GAC	Gly1090Asp	3269G>A	G1090D	Osteogenesis imperfecta III	Faqeih (2009) <i>Am J Med Genet A</i> 149A:461	PubMed 19208385
COL1A2	CM950289	GGC-GCC	Gly1096Ala	3287G>C	G1096A	Osteogenesis imperfecta III	Lu (1995) <i>Hum Mutat</i> 5:175	PubMed 7749416
COL1A2	CM070850	GGA-AGA	Gly1099Arg	3295G>A	G1099R	Osteogenesis imperfecta III	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070822	GGT-GAT	Gly247Asp	740G>A	G247D	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070788	GGT-CGT	Gly337Arg	1009G>C	G337R	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070871	GGT-TGT	Gly346Cys	1036G>T	G346C	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM910087	GGT-TGT	Gly349Cys	1045G>T	G349C	III/IV	Wenstrup (1991) <i>J Biol Chem</i> 266:2590	PubMed 1990009
COL1A2	CM973116	GGT-GAT	Gly376Asp	1127G>A	G376D	III/IV	Lund (1997) <i>Acta Paediatr</i> 86:711	PubMed 9240878
COL1A2	CM070852	GGC-AGC	Gly427Ser	1279G>A	G427S	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM973117	GGA-GTA	Gly484Val	1451G>T	G484V	III/IV	Lund (1997) <i>Acta Paediatr</i> 86:711	PubMed 9240878
COL1A2	CM070840	GGT-GTT	Gly598Val	1793G>T	G598V	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM942088	GGT-TGT	Gly730Cys	2188G>T	G730C	III/IV	Gomez-Lira (1994) <i>J Med Genet</i> 31:965	PubMed 7891382
COL1A2	CM973119	GGC-GTC	Gly763Val	2288G>T	G763V	III/IV	Lund (1997) <i>Acta Paediatr</i> 86:711	PubMed 9240878
COL1A2	CM950288	GGT-AGT	Gly778Ser	2332G>A	G778S	III/IV	Raghunath (1995) <i>Eur J Pediatr</i> 154:123	PubMed 7720740
COL1A2	CM042016	GGA-CGA	Gly793Arg	2377G>C	G793R	III/IV	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A2	CM960329	GGT-GAT	Gly892Asp	2675G>A	G892D	III/IV	Lund (1996) <i>Eur J Hum Genet</i> 4:39	PubMed 8800927
COL1A2	CM070855	GGC-AGC	Gly1006Ser	3016G>A	G1006S	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070857	GGC-TGC	Gly1090Cys	3268G>T	G1090C	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070832	GGT-GAT	Gly1102Asp	3305G>A	G1102D	III/IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070818	GGT-GAT	Gly124Asp	371G>A	G124D	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070819	GGT-GAT	Gly130Asp	389G>A	G130D	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070858	GGT-AGT	Gly133Ser	397G>A	G133S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM1211686	GGT-GAT	Gly196Asp	587G>A	G196D	Osteogenesis imperfecta IV	Valentina Gentile (2012) <i>Hum Mutat</i> 33:1697	PubMed 22753364
COL1A2	CM123311	GGT-TGT	Gly196Cys	586G>T	G196C	Osteogenesis imperfecta IV	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A2	CM062555	GGT-CGT	Gly202Arg	604G>C	G202R	Osteogenesis imperfecta IV	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM070821	GGT-GAT	Gly202Asp	605G>A	G202D	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM011294	GGG-GTG	Gly217Val	650G>T	G217V	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM062547	GGC-GTC	Gly256Val	767G>T	G256V	Osteogenesis imperfecta IV	Pollitt (2006) <i>Hum Mutat</i> 27:716	PubMed 16786509
COL1A2	CM1211688	GGT-CGT	Gly271Arg	811G>C	G271R	Osteogenesis imperfecta IV	Valentina Gentile (2012) <i>Hum Mutat</i> 33:1697	PubMed 22753364
COL1A2	CM011295	GGT-GAT	Gly274Asp	821G>A	G274D	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364

COL1A2	CM070786	GGT-AGT	Gly280Ser	838G>A	G280S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM011296	GGT-GCT	Gly286Ala	857G>C	G286A	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434 Krakow (1996) <i>COL1A2 LSDB entry</i> Meeting abstract:	PubMed 11317364
COL1A2	CM962605	GGC-GCC	Gly289Ala	866G>C	G289A	Osteogenesis imperfecta IV		Source: LSDB
COL1A2	CM011297	GGT-CGT	Gly298Arg	892G>C	G298R	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM070823	GGT-GAT	Gly307Asp	920G>A	G307D	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM062543	GGA-GAA	Gly325Glu	974G>A	G325E	Osteogenesis imperfecta IV	Lee (2006) <i>Hum Mutat</i> 27:599	PubMed 16705691
COL1A2	CM070859	GGT-AGT	Gly334Ser	1000G>A	G334S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070868	GGT-CGT	Gly340Arg	1018G>C	G340R	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070862	GGT-AGT	Gly349Ser	1045G>A	G349S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM011299	GGC-AGC	Gly364Ser	1090G>A	G364S	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM042013	GGT-GCT	Gly370Ala	1109G>C	G370A	Osteogenesis imperfecta IV	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A2	CM070837	GGT-GTT	Gly376Val	1127G>T	G376V	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM011300	GGC-TGC	Gly391Cys	1171G>T	G391C	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM042014	GGT-AGT	Gly406Ser	1216G>A	G406S	Osteogenesis imperfecta IV	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A2	CM070863	GGT-AGT	Gly424Ser	1270G>A	G424S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM020371	GGC-GCC	Gly469Ala	1406G>C	G469A	Osteogenesis imperfecta IV	Johnson (2002) <i>J Med Genet</i> 39:128	PubMed 11836364
COL1A2	CM070791	GGA-GAA	Gly493Glu	1478G>A	G493E	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM145067	GGA-GTA	Gly493Val	1478G>T	G493V	Osteogenesis imperfecta IV	Stephen (2014) <i>Am J Med Genet A</i> 164:1482	PubMed 24668929
COL1A2	CM070777	GGT-AGT	Gly511Ser	1531G>A	G511S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070814	GGT-AGT	Gly520Ser	1558G>A	G520S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM962606	GGT-TGT	Gly529Cys	1585G>T	G529C	Osteogenesis imperfecta IV	Mottes (1996) <i>COL1A2 LSDB entry</i> Unpublished:	Source: LSDB
COL1A2	CM011302	GGA-GAA	Gly532Glu	1595G>A	G532E	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM070778	GGT-AGT	Gly544Ser	1630G>A	G544S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	HM090036	GGT-AGT	Gly550Ser	1648G>A	G550S	Osteogenesis imperfecta IV	Lin (2010) <i>Hum Genet</i> 127:466	PubMed 21488275
COL1A2	CM011303	GGA-AGA	Gly571Arg	1711G>A	G571R	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM042015	GGT-AGT	Gly595Ser	1783G>A	G595S	Osteogenesis imperfecta IV	Hartikka (2004) <i>Hum Mutat</i> 24:147	PubMed 15241796
COL1A2	CM070826	GGT-GAT	Gly601Asp	1802G>A	G601D	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM011304	GGT-AGT	Gly601Ser	1801G>A	G601S	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM011305	GGT-CGT	Gly628Arg	1882G>C	G628R	Osteogenesis imperfecta IV	Ward (2001) <i>Hum Mutat</i> 17:434	PubMed 11317364
COL1A2	CM930152	GGT-GTT	Gly634Val	1901G>T	G634V	Osteogenesis imperfecta IV	Sztrolovics (1993) <i>Hum Mol Genet</i> 2:1319	PubMed 8401517
COL1A2	HM090037	GGT-TGT	Gly646Cys	1936G>T	G646C	Osteogenesis imperfecta IV	Lin (2010) <i>Hum Genet</i> 127:466	PubMed 21488249
COL1A2	CM123312	GGT-TGT	Gly661Cys	1981G>T	G661C	Osteogenesis imperfecta IV	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A2	CM910088	GGT-GTT	Gly676Val	2027G>T	G676V	Osteogenesis imperfecta IV	Bateman (1991) <i>Biochem J</i> 276:765	PubMed 2064612
COL1A2	CM994119	GGT-TGT	Gly700Cys	2098G>T	G700C	Osteogenesis imperfecta IV	McBride (1999) <i>COL1A2 LSDB entry</i> Unpublished:	Source: LSDB
COL1A2	CM133325	GGA-GAA	Gly760Glu	2279G>A	G760E	Osteogenesis imperfecta IV	Chen (2013) <i>Taiwan J Obstet Gynecol</i> 52:152	PubMed 23548243
COL1A2	HM090038	GGC-GAC	Gly763Asp	2288G>A	G763D	Osteogenesis imperfecta IV	Lin (2010) <i>Hum Genet</i> 127:467	PubMed 21488294
COL1A2	CM930153	GGT-GTT	Gly766Val	2297G>T	G766V	Osteogenesis imperfecta IV	Wang (1993) <i>J Biol Chem</i> 268:25162	PubMed 7693712
COL1A2	CM973120	GGT-AGT	Gly772Ser	2314G>A	G772S	Osteogenesis imperfecta IV	Nuytinck (1997) <i>Eur J Hum Genet</i> 5:161	PubMed 9272740
COL1A2	CM070833	GGT-GCT	Gly796Ala	2387G>C	G796A	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070856	GGC-TGC	Gly805Cys	2413G>T	G805C	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM123313	GGG-GTG	Gly817Val	2450G>T	G817V	Osteogenesis imperfecta IV	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
COL1A2	CM070854	GGC-AGC	Gly829Ser	2485G>A	G829S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070779	GGT-AGT	Gly880Ser	2638G>A	G880S	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022

COL1A2	CM973121	GGC-AGC	Gly901Ser	2701G>A	G901S	Osteogenesis imperfecta IV	Nuytinck (1997) <i>Eur J Hum Genet</i> 5:161	PubMed 9272740
COL1A2	CM070851	GGA-CGA	Gly916Arg	2746G>C	G916R	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM070783	GGT-TGT	Gly937Cys	2809G>T	G937C	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM930157	GGT-AGT	Gly1012Ser	3034G>A	G1012S	Osteogenesis imperfecta IV	Marini (1993) <i>J Biol Chem</i> 268:2667	PubMed 8094076
COL1A2	CM1110830	GGG-AGG	Gly1015Arg	3043G>A	G1015R	Osteogenesis imperfecta IV	Swinnen (2011) <i>Orphanet J Rare Dis</i> 6:88	PubMed 22206639
COL1A2	CM070802	GGC-GAC	Gly1078Asp	3233G>A	G1078D	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM880018	GGT-CGT	Gly1102Arg	3304G>C	G1102R	Osteogenesis imperfecta IV	Wenstrup (1988) <i>J Biol Chem</i> 263:7734	PubMed 2897363
COL1A2	CM070847	GGT-GTT	Gly1102Val	3305G>T	G1102V	Osteogenesis imperfecta IV	Marini (2007) <i>Hum Mutat</i> 28:209	PubMed 17078022
COL1A2	CM136399	CTT-CCT	Leu1313Pro	3938T>C	L1313P	Osteogenesis imperfecta IV	Sule (2013) <i>Osteoporos Int</i> 24:2253	PubMed 23443412
COL2A1	CM001075	GGT-GAT	Gly453Asp	1358G>A	G453D	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM001076	GGT-GTT	Gly453Val	1358G>T	G453V	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM950291	GGT-GAT	Gly510Asp	1529G>A	G510D	Achondrogenesis 2	Bonaventure (1995) <i>Biochem J</i> 307:823	PubMed 7741714
COL2A1	CM041750	GGT-GAT	Gly516Asp	1547G>A	G516D	Achondrogenesis 2	Faivre (2004) <i>Am J Med Genet</i> 126A:308	PubMed 15054848
COL2A1	CM076122	GGT-GTT	Gly546Val	1637G>T	G546V	Achondrogenesis 2	Forzano (2007) <i>Am J Med Genet A</i> 143A:2815	PubMed 17994563
COL2A1	CM103749	GGT-GAT	Gly768Asp	2303G>A	G768D	Achondrogenesis 2	Comstock (2010) <i>Am J Med Genet A</i> 152A:1822	PubMed 20583175
COL2A1	CM001080	GGT-GAT	Gly771Asp	2312G>A	G771D	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM001081	GGA-AGA	Gly780Arg	2338G>A	G780R	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM001082	GGC-CGC	Gly795Arg	2383G>C	G795R	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM001083	GGA-GAA	Gly894Glu	2681G>A	G894E	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM001084	GGT-GAT	Gly948Asp	2843G>A	G948D	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM950294	GGT-AGT	Gly969Ser	2905G>A	G969S	Achondrogenesis 2	Chan (1995) <i>J Biol Chem</i> 270:1747	PubMed 7829510
COL2A1	CM001085	GGC-AGC	Gly981Ser	2941G>A	G981S	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM972870	GGT-GTT	Gly1017Val	3050G>T	G1017V	Achondrogenesis 2	Kuivaniemi (1997) <i>Hum Mutat</i> 9:300	PubMed 9101290
COL2A1	CM001086	GGA-GTA	Gly1065Val	3194G>T	G1065V	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM972871	GGT-TGT	Gly1110Cys	3328G>T	G1110C	Achondrogenesis 2	Kuivaniemi (1997) <i>Hum Mutat</i> 9:300	PubMed 9101290
COL2A1	CM001087	GGA-AGA	Gly1119Arg	3355G>A	G1119R	Achondrogenesis 2	Korkko (2000) <i>Am J Med Genet</i> 92:95	PubMed 10797431
COL2A1	CM972872	GGC-TGC	Gly1143Cys	3427G>T	G1143C	Achondrogenesis 2	Kuivaniemi (1997) <i>Hum Mutat</i> 9:300	PubMed 9101290
CRTAP	CM067661	ATG-ATA	Met1Ile	3G>A	M1I	recessive	Barnes (2006) <i>N Engl J Med</i> 355:2757	PubMed 17192541
CRTAP	CM098071	GCG-GAG	Ala13Glu	38C>A	A13E	recessive	Van Dijk (2009) <i>Eur J Hum Genet</i> 17:1560	PubMed 19550437
CRTAP	CM098070	TAC-TAA	Tyr66Term	198C>A	Y66*	recessive	Van Dijk (2009) <i>Eur J Hum Genet</i> 17:1560	PubMed 19550437
CRTAP	CM086807	CTG-CCG	Leu67Pro	200T>C	L67P	recessive	Baldrige (2008) <i>Hum Mutat</i> 29:1435	PubMed 18566967
CRTAP	CM098072	AAG-GAG	Lys157Glu	469A>G	K157E	recessive	Van Dijk (2009) <i>Eur J Hum Genet</i> 17:1560	PubMed 19550437
CRTAP	CM1210078	TAT-TAG	Tyr187Term	561T>G	Y187*	Osteogenesis imperfecta, autosomal recessive	Shaheen (2012) <i>J Med Genet</i> 49:630	PubMed 23054245
CRTAP	CM067660	CAG-TAG	Gln276Term	826C>T	Q276*	recessive	Barnes (2006) <i>N Engl J Med</i> 355:2757	PubMed 17192541
FBN1	CM091204	ATG-TTG	Met1Leu	1A>T	M1L	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM117701	CTG-CGG	Leu11Arg	32T>G	L11R	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM054704	TAC-TGC	Tyr20Cys	59A>G	Y20C	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM130604	GGA-GAA	Gly55Glu	164G>A	G55E	Marfan syndrome	Wang (2013) <i>J Mol Med (Berl)</i> 91:37	PubMed 22772377
FBN1	CM093608	AAT-GAT	Asn57Asp	169A>G	N57D	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM1110388	TGT-CGT	Cys59Arg	175T>C	C59R	Marfan syndrome	Wei (2011) <i>PLoS One</i> 6:e29500	PubMed 22216297
FBN1	CM021992	CGT-TGT	Arg62Cys	184C>T	R62C	Marfan syndrome	Korkko (2002) <i>J Med Genet</i> 39:34	PubMed 11826022
FBN1	CM145706	CGT-CAT	Arg62His	185G>A	R62H	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM096435	TGT-CGT	Cys67Arg	199T>C	C67R	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986

FBN1	CM098631	TGC-AGC	Cys68Ser	202T>A	C68S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM062701	TGG-CGG	Trp71Arg	211T>C	W71R	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM131911	AAA-TAA	Lys72Term	214A>T	K72*	Marfan syndrome	Kuchtey (2013) <i>Am J Med Genet A</i> 161:880	PubMed 23444230
FBN1	CM095634	GGA-TGA	Gly77Term	229G>T	G77*	Marfan syndrome	Magyar (2009) <i>Hum Mutat</i> 30:1355	PubMed 19618372
FBN1	CM098632	TGT-CGT	Cys80Arg	238T>C	C80R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM117702	TGT-GGT	Cys80Gly	238T>G	C80G	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM077273	TGT-TAT	Cys80Tyr	239G>A	C80Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM013916	TGT-TTT	Cys89Phe	266G>T	C89F	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM098633	TGT-TGG	Cys89Trp	267T>G	C89W	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM067403	TGT-TAT	Cys89Tyr	266G>A	C89Y	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM093609	TGC-TAC	Cys100Tyr	299G>A	C100Y	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM098635	ACT-GCT	Thr101Ala	301A>G	T101A	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM970504	TGT-CGT	Cys111Arg	331T>C	C111R	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM021993	TCC-TGC	Ser115Cys	344C>G	S115C	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM940756	CGC-TGC	Arg122Cys	364C>T	R122C	Marfan syndrome	Stahl-Hallengren (1994) <i>J Clin Invest</i> 94:709	PubMed 8040326
FBN1	CM054715	TGT-TAT	Cys123Tyr	368G>A	C123Y	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM118483	ATG-GTG	Met124Val	370A>G	M124V	Marfan syndrome	Zadeh (2011) <i>Am J Med Genet A</i> 155:2661	PubMed 21932315
FBN1	CM126925	TGC-GGC	Cys129Gly	385T>G	C129G	Marfan syndrome	Haji-Seyed-Javadi (2012) <i>Hum Mutat</i> 33:1182	PubMed 22539340
FBN1	CM950436	TGC-TAC	Cys129Tyr	386G>A	C129Y	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM138429	TGT-CGT	Cys134Arg	400T>C	C134R	Marfan syndrome	Attanasio (2013) <i>Eur J Med Genet</i> 56:356	PubMed 23684891
FBN1	CM062707	TGT-GGT	Cys134Gly	400T>G	C134G	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM077278	TGT-TCT	Cys134Ser	401G>C	C134S	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM085405	TGC-AGC	Cys136Ser	406T>A	C136S	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM1312142	TGC-TGA	Cys136Term	408C>A	C136*	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM127279	TGT-CGT	Cys145Arg	433T>C	C145R	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM077274	TGT-TAT	Cys145Tyr	434G>A	C145Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM098636	CCT-TCT	Pro148Ser	442C>T	P148S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM040031	TGT-TCT	Cys154Ser	461G>C	C154S	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM055260	TGT-TGA	Cys154Term	462T>A	C154*	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM098637	TGT-GGT	Cys160Gly	478T>G	C160G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098638	TGT-TAT	Cys160Tyr	479G>A	C160Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM023914	AAT-AGT	Asn164Ser	491A>G	N164S	Marfan syndrome	Comeglio (2002) <i>Br J Ophthalmol</i> 86:1359	PubMed 12446365
FBN1	CM055251	CGA-TGA	Arg165Term	493C>T	R165*	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM950437	TGT-TTT	Cys166Phe	497G>T	C166F	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM040032	TGT-AGT	Cys166Ser	496T>A	C166S	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM970505	TGT-TCT	Cys166Ser	497G>C	C166S	Marfan syndrome	Collod-Beroud (1997) <i>Nucleic Acids Res</i> 25:147	PubMed 9016526
FBN1	CM119210	TGC-TGG	Cys168Trp	504C>G	C168W	Marfan syndrome	Millá (2012) <i>Eur J Ophthalmol</i> 22:667	PubMed 22034023
FBN1	CM040033	TAC-TAG	Tyr170Term	510C>G	Y170*	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM972798	CAG-TAG	Gln176Term	526C>T	Q176*	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM054702	TGT-CGT	Cys177Arg	529T>C	C177R	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM085416	TGT-AGT	Cys177Ser	529T>A	C177S	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM085427	TGT-TAT	Cys177Tyr	530G>A	C177Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM043995	CAG-TAG	Gln196Term	586C>T	Q196*	Marfan syndrome	Uyeda (2004) <i>J Hum Genet</i> 49:404	PubMed 15221638

FBN1	CM098510	TGC-TAC	Cys209Tyr	626G>A	C209Y	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM067404	TGT-TAT	Cys210Tyr	629G>A	C210Y	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM1312102	ACA-AGA	Thr212Arg	635C>G	T212R	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM094367	GGC-CGC	Gly214Arg	640G>C	G214R	Marfan syndrome	Voermans (2009) <i>Clin Genet</i> 76:25	PubMed 19659760
FBN1	CM003136	CGA-TGA	Arg215Term	643C>T	R215*	Marfan syndrome	Matsukawa (2000) <i>Hum Mutat</i> 17:71	PubMed 11139245
FBN1	CM940757	TGG-GGG	Trp217Gly	649T>G	W217G	Marfan syndrome	Karttunen (1994) <i>Am J Hum Genet</i> 55:1083	PubMed 7977366
FBN1	CM056960	TGG-TGA	Trp217Term	651G>A	W217*	Marfan syndrome	Baumgartner (2005) <i>Methods Inf Med</i> 44:487	PubMed 16342915
FBN1	CM098612	GAG-TAG	Glu222Term	664G>T	E222*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098613	CAG-TAG	Gln227Term	679C>T	Q227*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM088212	AAT-AAC	Asn238Asn	714T>C	N238N	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM983861	CGC-TGC	Arg240Cys	718C>T	R240C	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM090180	CGC-CAC	Arg240His	719G>A	R240H	Marfan syndrome	Van Dijk (2009) <i>Eur J Med Genet</i> 52:1	PubMed 19059503
FBN1	CM062712	CAG-TAG	Gln258Term	772C>T	Q258*	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM940758	GGG-AGG	Gly267Arg	799G>A	G267R	Marfan syndrome	Karttunen (1994) <i>Am J Hum Genet</i> 55:1083	PubMed 7977366
FBN1	CM132727	TGC-TGA	Cys271Term	813C>A	C271*	Marfan syndrome	Zhao (2013) <i>Mol Vis</i> 19:751	PubMed 23592911
FBN1	CM074792	CAA-TAA	Gln284Term	850C>T	Q284*	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM098622	TGC-TGA	Cys292Term	876C>A	C292*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM118002	TGT-TGA	Cys315Term	945T>A	C315*	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM100463	TAC-TAA	Tyr337Term	1011C>A	Y337*	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM067393	GGG-AGG	Gly343Arg	1027G>A	G343R	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM098623	TGC-TGA	Cys345Term	1035C>A	C345*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM054692	CAG-TAG	Gln348Term	1042C>T	Q348*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM074793	CAG-TAG	Gln351Term	1051C>T	Q351*	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM144424	ACC-AGC	Thr354Ser	1061C>G	T354S	Marfan syndrome	Wilson (2014) <i>Am J Med Genet A</i> 164:1209	PubMed 24664531
FBN1	CM042033	TGC-TAC	Cys359Tyr	1076G>A	C359Y	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM074801	TGC-CGC	Cys365Arg	1093T>C	C365R	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM145703	TGC-TGA	Cys365Term	1095C>A	C365*	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM015151	TGG-TGT	Trp366Cys	1098G>T	W366C	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM1312103	TGG-TTG	Trp366Leu	1097G>T	W366L	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM074855	TGG-TAG	Trp366Term	1097G>A	W366*	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM100464	TGG-TGA	Trp366Term	1098G>A	W366*	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM091206	GAG-CAG	Glu383Gln	1147G>C	E383Q	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM091205	GAG-AAG	Glu383Lys	1147G>A	E383K	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM020685	CGA-TGA	Arg429Term	1285C>T	R429*	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM972799	TAT-TAG	Tyr434Term	1302T>G	Y434*	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM054709	CGG-GGG	Arg439Gly	1315C>G	R439G	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM098614	GAG-TAG	Glu440Term	1318G>T	E440*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM022783	GTT-ATT	Val449Ile	1345G>A	V449I	Marfan syndrome	Rommel (2002) <i>Hum Mutat</i> 20:406	PubMed 12402346
FBN1	CM054690	CAG-CCG	Gln454Pro	1361A>C	Q454P	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM1312144	CAG-TAG	Gln454Term	1360C>T	Q454*	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM098642	TAT-TCT	Tyr458Ser	1373A>C	Y458S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098643	TGT-TCT	Cys460Ser	1379G>C	C460S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM118486	TGT-TAT	Cys460Tyr	1379G>A	C460Y	Marfan syndrome	Zadeh (2011) <i>Am J Med Genet A</i> 155:2661	PubMed 21932315

FBN1	CM098605	CAA-TAA	Gln461Term	1381C>T	Q461*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098611	TAC-TAA	Tyr472Term	1416C>A	Y472*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM000701	TGT-TTT	Cys474Phe	1421G>T	C474F	Marfan syndrome	Chikumi (2000) <i>J Hum Genet</i> 45:115	PubMed 10721679
FBN1	CM012598	TGT-TGG	Cys474Trp	1422T>G	C474W	Marfan syndrome	Comeglio (2001) <i>Hum Mutat</i> 18:251	PubMed 11524736
FBN1	CM118484	TGC-CGC	Cys476Arg	1426T>C	C476R	Marfan syndrome	Zadeh (2011) <i>Am J Med Genet A</i> 155:2661	PubMed 21932315
FBN1	CM940759	TGC-GGC	Cys476Gly	1426T>G	C476G	Marfan syndrome	Piersall (1994) <i>Hum Mol Genet</i> 3:1013	PubMed 7951214
FBN1	CM098606	AAA-TAA	Lys478Term	1432A>T	K478*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM056956	CGT-TGT	Arg485Cys	1453C>T	R485C	Marfan syndrome	Baumgartner (2005) <i>Methods Inf Med</i> 44:487	PubMed 16342915
FBN1	CM085417	TGT-CGT	Cys488Arg	1462T>C	C488R	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM042034	TGT-TTT	Cys488Phe	1463G>T	C488F	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM095758	TGT-TGA	Cys488Term	1464T>A	C488*	Marfan syndrome	Tang (2009) <i>Am J Med Genet A</i> 149A:2216	PubMed 19725129
FBN1	CM970506	GAT-TAT	Asp490Tyr	1468G>T	D490Y	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:415	PubMed 9401003
FBN1	CM1313965	TGT-TAT	Cys494Tyr	1481G>A	C494Y	Marfan syndrome	McInerney-Leo (2013) <i>Bonekey Rep</i> 2:456	PubMed 24501682
FBN1	CM992320	TGT-CGT	Cys499Arg	1495T>C	C499R	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM087769	TGT-AGT	Cys499Ser	1495T>A	C499S	Marfan syndrome	Li (2008) <i>Genet Test</i> 12:325	PubMed 18471089
FBN1	CM130602	TGT-TCT	Cys499Ser	1496G>C	C499S	Marfan syndrome	Wang (2013) <i>J Mol Med (Berl)</i> 91:37	PubMed 22772377
FBN1	CM117704	TGT-TAT	Cys499Tyr	1496G>A	C499Y	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM990586	TGT-TTT	Cys504Phe	1511G>T	C504F	Marfan syndrome	El-Aleem (1999) <i>Hum Mutat</i> 14:181	PubMed 10425041
FBN1	CM077275	TGT-TAT	Cys504Tyr	1511G>A	C504Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM054693	CGA-TGA	Arg516Term	1546C>T	R516*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM042035	TAT-TGT	Tyr519Cys	1556A>G	Y519C	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM1213072	TGC-TAC	Cys528Tyr	1583G>A	C528Y	Marfan syndrome	Winer (2012) <i>Endocr Pathol</i> 23:254	PubMed 22847364
FBN1	CM074166	CGA-TGA	Arg529Term	1585C>T	R529*	Marfan syndrome	Rand-Hendriksen (2007) <i>Am J Med Genet A</i> 143A:1968	PubMed 17663468
FBN1	CM098645	GAG-GGG	Glu533Gly	1598A>G	E533G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098646	TGT-CGT	Cys534Arg	1600T>C	C534R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM990587	TGT-TAT	Cys534Tyr	1601G>A	C534Y	Marfan syndrome	Kilpatrick (1999) <i>Hum Hered</i> 49:176	PubMed 10364683
FBN1	CM088217	CAG-TAG	Gln536Term	1606C>T	Q536*	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM1313966	TGC-TTC	Cys541Phe	1622G>T	C541F	Marfan syndrome	McInerney-Leo (2013) <i>Bonekey Rep</i> 2:456	PubMed 24501682
FBN1	CM055252	TGC-TAC	Cys541Tyr	1622G>A	C541Y	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM098647	GGA-CGA	Gly544Arg	1630G>C	G544R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM970507	CGC-TGC	Arg545Cys	1633C>T	R545C	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM074848	TGC-TGG	Cys546Trp	1638C>G	C546W	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM930242	AAC-ATC	Asn548Ile	1643A>T	N548I	Marfan syndrome	Dietz (1993) <i>Genomics</i> 17:468	PubMed 8406497
FBN1	CM145705	GCG-GTG	Ala559Val	1676C>T	A559V	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM013917	GGC-AGC	Gly560Ser	1678G>A	G560S	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM950438	CGA-TGA	Arg565Term	1693C>T	R565*	Marfan syndrome	Quan (1995) <i>Am J Hum Genet</i> 57S:A333	Source: Meeting abstract
FBN1	CM992321	TGT-CGT	Cys570Arg	1708T>C	C570R	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM117992	TGT-TCT	Cys570Ser	1709G>C	C570S	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM013918	TGT-TAT	Cys570Tyr	1709G>A	C570Y	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM020134	GAA-TAA	Glu571Term	1711G>T	E571*	Marfan syndrome	Korkko (2002) <i>J Med Genet</i> 39:34	PubMed 11826022
FBN1	CM100473	GAT-GGT	Asp574Gly	1721A>G	D574G	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM085422	TGC-TAC	Cys576Tyr	1727G>A	C576Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798

FBN1	CM085415	TGC-CGC	Cys582Arg	1744T>C	C582R	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM100093	GGA-CGA	Gly585Arg	1753G>C	G585R	Marfan syndrome	Khau Van Kien (2010) <i>Hum Mutat</i> 31:E1021	PubMed 19802897
FBN1	CM056957	GGA-GAA	Gly585Glu	1754G>A	G585E	Marfan syndrome	Baumgartner (2005) <i>Methods Inf Med</i> 44:487	PubMed 16342915
FBN1	CM970508	TGT-TAT	Cys587Tyr	1760G>A	C587Y	Marfan syndrome	Booms (1997) <i>Hum Genet</i> 100:195	PubMed 9254848
FBN1	CM013919	GGC-GAC	Gly592Asp	1775G>A	G592D	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM117993	TGT-GGT	Cys596Gly	1786T>G	C596G	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM021994	TGT-TAT	Cys596Tyr	1787G>A	C596Y	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM983862	TGC-TGG	Cys598Trp	1794C>G	C598W	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM117705	TGC-CGC	Cys611Arg	1831T>C	C611R	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM096437	GAC-AAC	Asp613Asn	1837G>A	D613N	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM972800	GAG-GGG	Glu616Gly	1847A>G	E616G	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM098649	GAG-AAG	Glu616Lys	1846G>A	E616K	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM117706	TGT-GGT	Cys617Gly	1849T>G	C617G	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM098650	TGC-GGC	Cys623Gly	1867T>G	C623G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM010883	TGC-TTC	Cys623Phe	1868G>T	C623F	Marfan syndrome	Toudjarska (2001) <i>Am J Med Genet</i> 99:294	PubMed 11251996
FBN1	CM940760	CGT-TGT	Arg627Cys	1879C>T	R627C	Marfan syndrome	Hayward (1994) <i>Hum Mol Genet</i> 3:373	PubMed 8004112
FBN1	CM127280	TGC-TAC	Cys628Tyr	1883G>A	C628Y	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM098651	AAC-AAA	Asn630Lys	1890C>A	N630K	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM023915	TCC-CCC	Ser634Pro	1900T>C	S634P	Marfan syndrome	Comeglio (2002) <i>Br J Ophthalmol</i> 86:1359	PubMed 12446365
FBN1	CM054705	TAC-TGC	Tyr635Cys	1904A>G	Y635C	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM095635	TAC-TAG	Tyr635Term	1905C>G	Y635*	Marfan syndrome	Magyar (2009) <i>Hum Mutat</i> 30:1355	PubMed 19618372
FBN1	CM054686	AGA-ATA	Arg636Ile	1907G>T	R636I	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM992322	TGT-CGT	Cys637Arg	1909T>C	C637R	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM098652	TGT-TTT	Cys637Phe	1910G>T	C637F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM117994	TGT-TGG	Cys637Trp	1911T>G	C637W	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM132728	TGT-TAT	Cys637Tyr	1910G>A	C637Y	Marfan syndrome	Zhao (2013) <i>Mol Vis</i> 19:751	PubMed 23592911
FBN1	CM077265	TGC-TAC	Cys639Tyr	1916G>A	C639Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM098653	CTG-CCG	Leu643Pro	1928T>C	L643P	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM118485	CGT-TGT	Arg650Cys	1948C>T	R650C	Marfan syndrome	Zadeh (2011) <i>Am J Med Genet A</i> 155:2661	PubMed 21932315
FBN1	CM1312096	TGT-CGT	Cys652Arg	1954T>C	C652R	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM040034	TGT-TCT	Cys652Ser	1955G>C	C652S	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM023916	TGT-TAT	Cys652Tyr	1955G>A	C652Y	Marfan syndrome	Comeglio (2002) <i>Br J Ophthalmol</i> 86:1359	PubMed 12446365
FBN1	CM021592	GAC-AAC	Asp654Asn	1960G>A	D654N	Marfan syndrome	Halliday (2002) <i>J Med Genet</i> 39:589	PubMed 12161601
FBN1	CM970509	TGC-CGC	Cys661Arg	1981T>C	C661R	Marfan syndrome	Collod-Beroud (1997) <i>Nucleic Acids Res</i> 25:147	PubMed 9016526
FBN1	CM098654	TGC-GGC	Cys661Gly	1981T>G	C661G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM021995	TGC-TAC	Cys661Tyr	1982G>A	C661Y	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM113531	TAC-TAG	Tyr665Term	1995C>G	Y665*	Marfan syndrome	Lebreiro (2011) <i>Rev Esp Cardiol</i> 64:151	PubMed 21194821
FBN1	CM021996	TCT-TAT	Ser681Tyr	2042C>A	S681Y	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM021997	TGC-CGC	Cys683Arg	2047T>C	C683R	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM100474	TGC-TAC	Cys683Tyr	2048G>A	C683Y	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM085428	TGT-TAT	Cys684Tyr	2051G>A	C684Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM021998	TGC-TGG	Cys685Trp	2055C>G	C685W	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM117707	TGC-TAC	Cys685Tyr	2054G>A	C685Y	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060

FBN1	CM098656	GCC-GAC	Ala686Asp	2057C>A	A686D	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM096438	GCC-ACC	Ala686Thr	2056G>A	A686T	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM054724	TGT-TGA	Cys699Term	2097T>A	C699*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM960626	GCG-ACG	Ala705Thr	2113G>A	A705T	Marfan syndrome	Ades (1996) <i>J Med Genet</i> 33:665	PubMed 8863159
FBN1	CM095636	CAG-TAG	Gln708Term	2122C>T	Q708*	Marfan syndrome	Magyar (2009) <i>Hum Mutat</i> 30:1355	PubMed 19618372
FBN1	CM960627	TGC-TAC	Cys711Tyr	2132G>A	C711Y	Marfan syndrome	Ades (1996) <i>J Med Genet</i> 33:665	PubMed 8863159
FBN1	CM085404	GGC-TGC	Gly721Cys	2161G>T	G721C	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM930243	GAT-GCT	Asp723Ala	2168A>C	D723A	Marfan syndrome	Dietz (1993) <i>Genomics</i> 17:468	PubMed 8406497
FBN1	CM021999	GAT-GTT	Asp723Val	2168A>T	D723V	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM117995	ATA-AGA	Ile724Arg	2171T>G	I724R	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM067400	ATA-GTA	Ile724Val	2170A>G	I724V	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM098657	GAA-GGA	Glu726Gly	2177A>G	E726G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM074856	TGT-TAT	Cys727Tyr	2180G>A	C727Y	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM022000	TGC-TTC	Cys734Phe	2201G>T	C734F	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM098658	TGT-CGT	Cys739Arg	2215T>C	C739R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM950439	TAT-TGT	Tyr746Cys	2237A>G	Y746C	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM145708	TGT-CGT	Cys748Arg	2242T>C	C748R	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM021593	TGT-TAT	Cys748Tyr	2243G>A	C748Y	Marfan syndrome	Halliday (2002) <i>J Med Genet</i> 39:589	PubMed 12161601
FBN1	CM940761	TGC-GGC	Cys750Gly	2248T>G	C750G	Marfan syndrome	Hayward (1994) <i>Hum Mol Genet</i> 3:373	PubMed 8004112
FBN1	CM098659	TGC-AGC	Cys750Ser	2248T>A	C750S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM055253	TGC-TGA	Cys750Term	2250C>A	C750*	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM042036	TAT-TGT	Tyr754Cys	2261A>G	Y754C	Marfan syndrome	Summers (2004) <i>Clin Genet</i> 65:66	PubMed 15032979
FBN1	CM077248	TGT-CGT	Cys769Arg	2305T>C	C769R	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM067389	TGT-AGT	Cys769Ser	2305T>A	C769S	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM098660	TGT-TAT	Cys769Tyr	2306G>A	C769Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM022001	TGT-GGT	Cys776Gly	2326T>G	C776G	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM098661	TGT-TTT	Cys776Phe	2327G>T	C776F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM127281	TGT-TGG	Cys776Trp	2328T>G	C776W	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM013920	TGT-TAT	Cys776Tyr	2327G>A	C776Y	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM013921	TGT-CGT	Cys781Arg	2341T>C	C781R	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM055256	TGT-TAT	Cys781Tyr	2342G>A	C781Y	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM098663	TGT-GGT	Cys790Gly	2368T>G	C790G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098664	TGT-TTT	Cys790Phe	2369G>T	C790F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM126517	TGT-AGT	Cys790Ser	2368T>A	C790S	Marfan syndrome	Micheal (2012) <i>Mol Vis</i> 18:1918	PubMed 22876116
FBN1	CM117709	TGT-TAT	Cys790Tyr	2369G>A	C790Y	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM054713	TGC-TGA	Cys792Term	2376C>A	C792*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM145730	AAA-TAA	Lys803Term	2407A>T	K803*	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM077280	TGT-TGG	Cys805Trp	2415T>G	C805W	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM054710	GAA-TAA	Glu810Term	2428G>T	E810*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM096439	TGC-TGA	Cys811Term	2433C>A	C811*	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM042037	TGC-TGG	Cys811Trp	2433C>G	C811W	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM117710	TGC-TAC	Cys811Tyr	2432G>A	C811Y	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM003137	TCA-TGA	Ser813Term	2438C>G	S813*	Marfan syndrome	Matsukawa (2000) <i>Hum Mutat</i> 17:71	PubMed 11139245

FBN1	CM085433	TGC-CGC	Cys816Arg	2446T>C	C816R	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM066807	TGC-GGC	Cys816Gly	2446T>G	C816G	Marfan syndrome	Lledo (2006) <i>Fertil Steril</i> 86:949	PubMed 17027361
FBN1	CM067402	TGC-TTC	Cys816Phe	2447G>T	C816F	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM983863	TGC-TCC	Cys816Ser	2447G>C	C816S	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM098625	TGC-TGA	Cys816Term	2448C>A	C816*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM100094	GGA-GAA	Gly819Glu	2456G>A	G819E	Marfan syndrome	Khau Van Kien (2010) <i>Hum Mutat</i> 31:E1021	PubMed 19802897
FBN1	CM098626	TGC-TGA	Cys821Term	2463C>A	C821*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM096440	TGC-TAC	Cys821Tyr	2462G>A	C821Y	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM077251	CCA-TCA	Pro825Ser	2473C>T	P825S	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM074871	TTT-TGT	Phe828Cys	2483T>G	F828C	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM098665	TGT-AGT	Cys830Ser	2488T>A	C830S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM065179	TGT-TAT	Cys830Tyr	2489G>A	C830Y	Marfan syndrome	Ganesh (2006) <i>Arch Ophthalmol</i> 124:205	PubMed 16476890
FBN1	CM096441	TGT-TTT	Cys832Phe	2495G>T	C832F	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM145732	TGT-TGG	Cys832Trp	2496T>G	C832W	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM972801	TGT-TAT	Cys832Tyr	2495G>A	C832Y	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM1311111	TGC-TAC	Cys845Tyr	2534G>A	C845Y	Marfan syndrome	Callier (2013) <i>Clin Genet</i> :	PubMed 23506379
FBN1	CM117711	TGC-AGC	Cys853Ser	2557T>A	C853S	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM077272	TGC-TGA	Cys853Term	2559C>A	C853*	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM042038	TGG-TGA	Trp854Term	2562G>A	W854*	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM098615	CAG-TAG	Gln855Term	2563C>T	Q855*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM972802	CGA-TGA	Arg861Term	2581C>T	R861*	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM930244	TGT-CGT	Cys862Arg	2584T>C	C862R	Marfan syndrome	Tynan (1993) <i>Hum Mol Genet</i> 2:1813	PubMed 8281141
FBN1	CM098668	TGC-CGC	Cys875Arg	2623T>C	C875R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM022784	GGT-AGT	Gly880Ser	2638G>A	G880S	Marfan syndrome	Rommel (2002) <i>Hum Mutat</i> 20:406	PubMed 12402346
FBN1	CM042039	GCG-GTG	Ala882Val	2645C>T	A882V	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM085414	GGA-GAA	Gly884Glu	2651G>A	G884E	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM138430	TGC-TAC	Cys887Tyr	2660G>A	C887Y	Marfan syndrome	Attanasio (2013) <i>Eur J Med Genet</i> 56:356	PubMed 23684891
FBN1	CM952168	TGC-CGC	Cys890Arg	2668T>C	C890R	Marfan syndrome	Kielty (1995) <i>J Cell Sci</i> 108:1317	PubMed 7622614
FBN1	CM054688	TGC-GGC	Cys890Gly	2668T>G	C890G	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM098669	TGT-TAT	Cys896Tyr	2687G>A	C896Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM022002	TGT-CGT	Cys908Arg	2722T>C	C908R	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM014705	TGT-TAT	Cys908Tyr	2723G>A	C908Y	Marfan syndrome	Judge (2001) <i>Am J Med Genet</i> 99:39	PubMed 11170092
FBN1	CM108648	GAT-AAT	Asp910Asn	2728G>A	D910N	Marfan syndrome	Hilhorst-Hofstee (2010) <i>Hum Mutat</i> 31:E1915	PubMed 20886638
FBN1	CM093610	GAT-CAT	Asp910His	2728G>C	D910H	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM013922	GAA-GGA	Glu913Gly	2738A>G	E913G	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM098670	TGT-AGT	Cys914Ser	2740T>A	C914S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM022003	TGT-GGT	Cys921Gly	2761T>G	C921G	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM093611	TGT-TGA	Cys921Term	2763T>A	C921*	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM950440	TGT-CGT	Cys926Arg	2776T>C	C926R	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM098671	TGT-GGT	Cys926Gly	2776T>G	C926G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM065180	TGT-TAT	Cys926Tyr	2777G>A	C926Y	Marfan syndrome	Ganesh (2006) <i>Arch Ophthalmol</i> 124:205	PubMed 16476890
FBN1	CM096443	ACT-CCT	Thr929Pro	2785A>C	T929P	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM067395	TGT-CGT	Cys950Arg	2848T>C	C950R	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931

FBN1	CM091207	CGC-CAC	Arg954His	2861G>A	R954H	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM074811	CGC-TGC	Arg974Cys	2920C>T	R974C	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM074813	CGC-CAC	Arg976His	2927G>A	R976H	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM100475	TGC-AGC	Cys980Ser	2938T>A	C980S	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM098672	TGC-TAC	Cys980Tyr	2939G>A	C980Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM062714	TGC-TCC	Cys981Ser	2942G>C	C981S	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM960628	GTC-ATC	Val984Ile	2950G>A	V984I	Marfan syndrome	Grau (1996) <i>Eur J Pediatr</i> 155:739	PubMed: No PubMed ID
FBN1	CM015152	GGG-AGG	Gly985Arg	2953G>A	G985R	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM990588	GGG-GAG	Gly985Glu	2954G>A	G985E	Marfan syndrome	Collod-Beroud (1999) <i>Am J Hum Genet</i> 65:917	PubMed 10441597
FBN1	CM145733	GCA-ACA	Ala986Thr	2956G>A	A986T	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM980726	GAG-TAG	Glu994Term	2980G>T	E994*	Marfan syndrome	Karttunen (1998) <i>Hum Mutat</i> S1:S34	PubMed 9452033
FBN1	CM983864	TGT-CGT	Cys996Arg	2986T>C	C996R	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM995300	ATG-GTG	Met998Val	2992A>G	M998V	Marfan syndrome	Yuan (1999) <i>Hum Mutat</i> 14:440	PubMed 10533071
FBN1	CM076184	TGT-CGT	Cys1008Arg	3022T>C	C1008R	Marfan syndrome	Qin (2007) <i>Neuro Endocrinol Lett</i> 28:629	PubMed 17984934
FBN1	CM085429	TGT-TAT	Cys1008Tyr	3023G>A	C1008Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM950441	GGA-AGA	Gly1013Arg	3037G>A	G1013R	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM983865	GGA-CGA	Gly1013Arg	3037G>C	G1013R	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM118911	GCC-CCC	Ala1015Pro	3043G>C	A1015P	Marfan syndrome	Lebreiro (2011) <i>Rev Port Cardiol</i> 30:649	PubMed 22005308
FBN1	CM062715	GCC-ACC	Ala1015Thr	3043G>A	A1015T	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM010035	ACA-GCA	Thr1020Ala	3058A>G	T1020A	Marfan syndrome	Tiecke (2001) <i>Eur J Hum Genet</i> 9:13	PubMed 11175294
FBN1	CM940762	AAG-AAC	Lys1023Asn	3069G>C	K1023N	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	CM106076	GAT-TAT	Asp1028Tyr	3082G>T	D1028Y	Marfan syndrome	Evangelisti (2010) <i>Eur J Med Genet</i> 53:299	PubMed 20538085
FBN1	CM014706	GAT-GTT	Asp1028Val	3083A>T	D1028V	Marfan syndrome	Ivan (2001) <i>Chin Med J (Engl)</i> 114:473	PubMed 11780406
FBN1	CM096444	GAG-TAG	Glu1031Term	3091G>T	E1031*	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM010036	TGC-TAC	Cys1032Tyr	3095G>A	C1032Y	Marfan syndrome	Ng (1999) <i>J Paediatr Child Health</i> 35:321	PubMed 10404462
FBN1	CM136187	CTC-TTC	Leu1038Phe	3112C>T	L1038F	Marfan syndrome	Classen (2013) <i>Hum Genet</i> 132:825	PubMed 23552953
FBN1	CM972803	TGC-TAC	Cys1039Tyr	3116G>A	C1039Y	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM062708	GGC-GAC	Gly1042Asp	3125G>A	G1042D	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM085409	GGC-AGC	Gly1042Ser	3124G>A	G1042S	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM970510	AAG-AGG	Lys1043Arg	3128A>G	K1043R	Marfan syndrome	Wang (1997) <i>Hum Mutat</i> 9:359	PubMed 9101298
FBN1	CM055254	TGC-TGA	Cys1044Term	3132C>A	C1044*	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM040035	TGC-TAC	Cys1044Tyr	3131G>A	C1044Y	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM100476	AAC-AGC	Asn1046Ser	3137A>G	N1046S	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM960629	ATT-ACT	Ile1048Thr	3143T>C	I1048T	Marfan syndrome	Lonnqvist (1996) <i>Genomics</i> 36:468	PubMed 8884270
FBN1	CM960630	TGC-CGC	Cys1053Arg	3157T>C	C1053R	Marfan syndrome	Putnam (1996) <i>Am J Med Genet</i> 62:233	PubMed 8882780
FBN1	CM960631	TGT-GGT	Cys1055Gly	3163T>G	C1055G	Marfan syndrome	Ades (1996) <i>J Med Genet</i> 33:665	PubMed 8863159
FBN1	CM056958	TGT-AGT	Cys1055Ser	3163T>A	C1055S	Marfan syndrome	Baumgartner (2005) <i>Methods Inf Med</i> 44:487	PubMed 16342915
FBN1	CM013923	TGT-TGG	Cys1055Trp	3165T>G	C1055W	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM013924	TGT-TAT	Cys1055Tyr	3164G>A	C1055Y	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM054699	GGC-GAC	Gly1058Asp	3173G>A	G1058D	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM098673	TCT-TGT	Ser1063Cys	3188C>G	S1063C	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM020686	GAC-GGC	Asp1070Gly	3209A>G	D1070G	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM960632	ATT-AGT	Ile1071Ser	3212T>G	I1071S	Marfan syndrome	Wang (1996) <i>J Med Genet</i> 33:760	PubMed 8880577

FBN1	CM960633	GAC-GGC	Asp1072Gly	3215A>G	D1072G	Marfan syndrome	Putnam (1996) <i>Am J Med Genet</i> 62:233	PubMed 8882780
FBN1	CM960634	GAA-GAT	Glu1073Asp	3219A>T	E1073D	Marfan syndrome	Wang (1996) <i>J Med Genet</i> 33:760	PubMed 8880577
FBN1	CM950442	GAA-AAA	Glu1073Lys	3217G>A	E1073K	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM950443	TGC-CGC	Cys1074Arg	3220T>C	C1074R	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	HM971939	TGC-TAC	Cys1074Tyr	3221G>A	C1074Y	Marfan syndrome	Comeglio (2001) <i>Hum Genet</i> 109:468	Source: Online submission
FBN1	CM972804	TCT-CCT	Ser1077Pro	3229T>C	S1077P	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM020687	TGT-GGT	Cys1081Gly	3241T>G	C1081G	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM078031	TGT-CGT	Cys1086Arg	3256T>C	C1086R	Marfan syndrome	Sutherell (2007) <i>Congenit Heart Dis</i> 2:342	PubMed 18377451
FBN1	CM950444	TGT-TGG	Cys1086Trp	3258T>G	C1086W	Marfan syndrome	Matthews (1995) <i>Am J Hum Genet</i> 57S:A339	Source: Meeting abstract
FBN1	CM992323	TGT-TAT	Cys1086Tyr	3257G>A	C1086Y	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM074790	AAC-ATC	Asn1088Ile	3263A>T	N1088I	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM117996	AAC-AGC	Asn1088Ser	3263A>G	N1088S	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM117712	CCT-TCT	Pro1090Ser	3268C>T	P1090S	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM077271	TGC-TGA	Cys1095Term	3285C>A	C1095*	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM127646	GAA-TAA	Glu1099Term	3295G>T	E1099*	Marfan syndrome	Cui (2012) <i>Orphanet J Rare Dis</i> 7:	PubMed 22913777
FBN1	CM042040	GGC-GTC	Gly1100Val	3299G>T	G1100V	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM013925	TAT-TGT	Tyr1101Cys	3302A>G	Y1101C	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM100465	GAA-TAA	Glu1102Term	3304G>T	E1102*	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM127282	TGC-TAC	Cys1111Tyr	3332G>A	C1111Y	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM972805	GAT-GGT	Asp1113Gly	3338A>G	D1113G	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM055248	GAT-GTT	Asp1113Val	3338A>T	D1113V	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM010038	GAT-GGT	Asp1115Gly	3344A>G	D1115G	Marfan syndrome	Tiecke (2001) <i>Eur J Hum Genet</i> 9:13	PubMed 11175294
FBN1	CM960635	TGT-GGT	Cys1117Gly	3349T>G	C1117G	Marfan syndrome	Putnam (1996) <i>Am J Med Genet</i> 62:233	PubMed 8882780
FBN1	CM930245	TGT-TAT	Cys1117Tyr	3350G>A	C1117Y	Marfan syndrome	Tynan (1993) <i>Hum Mol Genet</i> 2:1813	PubMed 8281141
FBN1	CM1312097	TGC-TTC	Cys1124Phe	3371G>T	C1124F	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM055245	CGA-TGA	Arg1125Term	3373C>T	R1125*	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM990589	TGC-TAC	Cys1129Tyr	3386G>A	C1129Y	Marfan syndrome	El-Aleem (1999) <i>Hum Mutat</i> 14:181	PubMed 10425041
FBN1	CM093612	CAT-CCT	His1130Pro	3389A>C	H1130P	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM098674	AAC-AAG	Asn1131Lys	3393C>G	N1131K	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM145735	AAC-AGC	Asn1131Ser	3392A>G	N1131S	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM970511	AAC-TAC	Asn1131Tyr	3391A>T	N1131Y	Marfan syndrome	Wang (1997) <i>Hum Mutat</i> 9:359	PubMed 9101298
FBN1	CM085419	TAC-TAG	Tyr1136Term	3408C>G	Y1136*	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM910154	CGC-CCC	Arg1137Pro	3410G>C	R1137P	Marfan syndrome	Dietz (1991) <i>Nature</i> 352:337	PubMed 1852208
FBN1	CM098675	TGT-GGT	Cys1138Gly	3412T>G	C1138G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM145736	TGT-TTT	Cys1138Phe	3413G>T	C1138F	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM098676	TGT-TCT	Cys1138Ser	3413G>C	C1138S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM074857	TGT-TAT	Cys1138Tyr	3413G>A	C1138Y	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM074851	TGC-TGA	Cys1140Term	3420C>A	C1140*	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM995301	CCG-CTG	Pro1141Leu	3422C>T	P1141L	Marfan syndrome	Yuan (1999) <i>Hum Mutat</i> 14:440	PubMed 10533071
FBN1	CM096445	GCG-GTG	Ala1152Val	3455C>T	A1152V	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM054719	TGT-TCT	Cys1153Ser	3458G>C	C1153S	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM960636	TGT-TAT	Cys1153Tyr	3458G>A	C1153Y	Marfan syndrome	Ades (1996) <i>J Med Genet</i> 33:665	PubMed 8863159

FBN1	CM960637	GAC-AAC	Asp1155Asn	3463G>A	D1155N	Marfan syndrome	Milewicz (1996) <i>Circulation</i> 94:2708	PubMed 8941093
FBN1	CM108649	GAC-GGC	Asp1155Gly	3464A>G	D1155G	Marfan syndrome	Hilhorst-Hofstee (2010) <i>Hum Mutat</i> 31:E1915	PubMed 20886638
FBN1	CM074817	GAA-GGA	Glu1158Gly	3473A>G	E1158G	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM980727	TGC-TAC	Cys1166Tyr	3497G>A	C1166Y	Marfan syndrome	Perez (1998) <i>Hum Mutat</i> 13:84	PubMed 10189222
FBN1	CM117997	AAT-AGT	Asn1168Ser	3503A>G	N1168S	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM940763	CGT-CAT	Arg1170His	3509G>A	R1170H	Marfan syndrome	Hayward (1994) <i>Mol Cell Probes</i> 8:325	PubMed 7870075
FBN1	CM992324	TGC-CGC	Cys1171Arg	3511T>C	C1171R	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM095637	TGC-TGA	Cys1171Term	3513C>A	C1171*	Marfan syndrome	Magyar (2009) <i>Hum Mutat</i> 30:1355	PubMed 19618372
FBN1	CM970512	TGC-TGG	Cys1171Trp	3513C>G	C1171W	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM970513	AAC-AAG	Asn1173Lys	3519C>G	N1173K	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM983866	ATA-ACA	Ile1175Thr	3524T>C	I1175T	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM067388	GGG-AGG	Gly1176Arg	3526G>A	G1176R	Marfan syndrome	Valiev (2006) <i>Mol Biol (Mosk)</i> 40:1021	PubMed 17209430
FBN1	CM098677	TAT-TGT	Tyr1178Cys	3533A>G	Y1178C	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098678	TGT-CGT	Cys1180Arg	3538T>C	C1180R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM980728	TGC-TCC	Cys1182Ser	3545G>C	C1182S	Marfan syndrome	Perez (1998) <i>Hum Mutat</i> 13:84	PubMed 10189222
FBN1	CM972857	TGC-TGG	Cys1182Trp	3546C>G	C1182W	Marfan syndrome	Pepe (1997) <i>J Mol Cell Cardiol</i> 29:1877	PubMed 9236141
FBN1	CM117713	GGC-GAC	Gly1185Asp	3554G>A	G1185D	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM020688	TAC-TGC	Tyr1186Cys	3557A>G	Y1186C	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM098679	TGT-TAT	Cys1195Tyr	3584G>A	C1195Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM130606	GAT-GCT	Asp1199Ala	3596A>C	D1199A	Marfan syndrome	Wang (2013) <i>J Mol Med (Berl)</i> 91:37	PubMed 22772377
FBN1	CM990590	GAA-GGA	Glu1200Gly	3599A>G	E1200G	Marfan syndrome	Rantamaki (1999) <i>Am J Hum Genet</i> 64:993	PubMed 10090884
FBN1	CM118003	TGC-TGA	Cys1201Term	3603C>A	C1201*	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM081261	TGC-TAC	Cys1201Tyr	3602G>A	C1201Y	Marfan syndrome	Derbent (2008) <i>Clin Dysmorphol</i> 17:127	PubMed 18388785
FBN1	CM054708	TAT-TGT	Tyr1219Cys	3656A>G	Y1219C	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM074815	TGT-CGT	Cys1223Arg	3667T>C	C1223R	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM940764	TGT-TAT	Cys1223Tyr	3668G>A	C1223Y	Marfan syndrome	Hewett (1994) <i>J Med Genet</i> 31:338	PubMed 8071963
FBN1	CM088213	CAG-CAT	Gln1224His	3672G>T	Q1224H	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM118912	GGA-TGA	Gly1226Term	3676G>T	G1226*	Marfan syndrome	Lebreiro (2011) <i>Rev Port Cardiol</i> 30:649	PubMed 22005308
FBN1	CM117624	TCA-CCA	Ser1235Pro	3703T>C	S1235P	Marfan syndrome	Meng (2011) <i>Mol Vis</i> 17:2421	PubMed 21976953
FBN1	CM055941	TGC-CGC	Cys1236Arg	3706T>C	C1236R	Marfan syndrome	ter Heide (2005) <i>Clin Dysmorphol</i> 14:81	PubMed 15770129
FBN1	CM995302	GAC-AAC	Asp1238Asn	3712G>A	D1238N	Marfan syndrome	Yuan (1999) <i>Hum Mutat</i> 14:440	PubMed 10533071
FBN1	CM010039	GAC-GGC	Asp1238Gly	3713A>G	D1238G	Marfan syndrome	Tiecke (2001) <i>Eur J Hum Genet</i> 9:13	PubMed 11175294
FBN1	CM098681	GAG-GGG	Glu1241Gly	3722A>G	E1241G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM940765	TGT-TAT	Cys1242Tyr	3725G>A	C1242Y	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	CM074816	TGT-CGT	Cys1249Arg	3745T>C	C1249R	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM950446	TGT-TCT	Cys1249Ser	3746G>C	C1249S	Marfan syndrome	Dietz (1995) <i>Hum Mol Genet</i> 4:1799	PubMed 8541880
FBN1	CM1314085	TGC-CGC	Cys1254Arg	3760T>C	C1254R	Marfan syndrome	Pees (2013) <i>Clin Genet</i> epub:epub	PubMed 24199744
FBN1	CM004133	TGC-AGC	Cys1254Ser	3760T>A	C1254S	Marfan syndrome	Oh (2000) <i>Pediatr Int</i> 42:488	PubMed 11059536
FBN1	CM972806	GAG-TAG	Glu1260Term	3778G>T	E1260*	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM054700	TAC-GAC	Tyr1261Asp	3781T>G	Y1261D	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM990591	TAC-TGC	Tyr1261Cys	3782A>G	Y1261C	Marfan syndrome	El-Aleem (1999) <i>Hum Mutat</i> 14:181	PubMed 10425041
FBN1	CM091208	TGC-CGC	Cys1263Arg	3787T>C	C1263R	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM980729	TGT-CGT	Cys1265Arg	3793T>C	C1265R	Marfan syndrome	Montgomery (1998) <i>Am J Hum Genet</i> 63:1703	PubMed 9837823

FBN1	CM098683	TGT-GGT	Cys1265Gly	3793T>G	C1265G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM085430	TGT-TAT	Cys1265Tyr	3794G>A	C1265Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM020689	TAT-TTT	Tyr1266Phe	3797A>T	Y1266F	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM054720	TGT-TCT	Cys1278Ser	3833G>C	C1278S	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM972807	AAT-AGT	Asn1282Ser	3845A>G	N1282S	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM091209	TGT-CGT	Cys1284Arg	3850T>C	C1284R	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM055250	TGT-GGT	Cys1284Gly	3850T>G	C1284G	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM117714	TGT-TAT	Cys1284Tyr	3851G>A	C1284Y	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM010040	AAA-GAA	Lys1300Glu	3898A>G	K1300E	Marfan syndrome	Tiecke (2001) <i>Eur J Hum Genet</i> 9:13	PubMed 11175294
FBN1	CM074858	TGT-TAT	Cys1307Tyr	3920G>A	C1307Y	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM099390	GGC-GAC	Gly1310Asp	3929G>A	G1310D	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM077262	GGC-GTC	Gly1310Val	3929G>T	G1310V	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM085410	TGT-CGT	Cys1320Arg	3958T>C	C1320R	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM1312098	TGT-TTT	Cys1320Phe	3959G>T	C1320F	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM972808	TGT-AGT	Cys1320Ser	3958T>A	C1320S	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM054723	TGT-TGA	Cys1320Term	3960T>A	C1320*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM077258	GAC-GGC	Asp1322Gly	3965A>G	D1322G	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM108650	GAC-CAC	Asp1322His	3964G>C	D1322H	Marfan syndrome	Hilhorst-Hofstee (2010) <i>Hum Mutat</i> 31:E1915	PubMed 20886638
FBN1	CM040036	GAA-CAA	Glu1325Gln	3973G>C	E1325Q	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM010041	GAA-TAA	Glu1325Term	3973G>T	E1325*	Marfan syndrome	Tiecke (2001) <i>Eur J Hum Genet</i> 9:13	PubMed 11175294
FBN1	CM992325	TGT-CGT	Cys1326Arg	3976T>C	C1326R	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM054698	TGT-AGT	Cys1333Ser	3997T>A	C1333S	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM983867	GCT-CCT	Ala1337Pro	4009G>C	A1337P	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM145760	TGT-TCT	Cys1339Ser	4016G>C	C1339S	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM013927	TGT-TAT	Cys1339Tyr	4016G>A	C1339Y	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM972809	AAT-AGT	Asn1341Ser	4022A>G	N1341S	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM098684	GCA-ACA	Ala1343Thr	4027G>A	A1343T	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM096446	GGA-AGA	Gly1344Arg	4030G>A	G1344R	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM118910	TTC-TGC	Phe1346Cys	4037T>G	F1346C	Marfan syndrome	Lebreiro (2011) <i>Rev Port Cardiol</i> 30:649	PubMed 22005308
FBN1	CM077281	TTC-TTG	Phe1346Leu	4038C>G	F1346L	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM074869	TTC-TCC	Phe1346Ser	4037T>C	F1346S	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM020135	TGT-TGA	Cys1348Term	4044T>A	C1348*	Marfan syndrome	Korkko (2002) <i>J Med Genet</i> 39:34	PubMed 11826022
FBN1	CM117720	TGC-TTC	Cys1350Phe	4049G>T	C1350F	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM098685	TGC-TAC	Cys1350Tyr	4049G>A	C1350Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098686	GGG-AGG	Gly1353Arg	4057G>A	G1353R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM992326	TGC-TAC	Cys1361Tyr	4082G>A	C1361Y	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM040037	GAA-AAA	Glu1366Lys	4096G>A	E1366K	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM043996	GAA-TAA	Glu1366Term	4096G>T	E1366*	Marfan syndrome	Uyeda (2004) <i>J Hum Genet</i> 49:404	PubMed 15221638
FBN1	CM062702	TGT-CGT	Cys1367Arg	4099T>C	C1367R	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM088214	TGC-GGC	Cys1374Gly	4120T>G	C1374G	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM040038	TGC-TCC	Cys1374Ser	4121G>C	C1374S	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM098687	TGC-TAC	Cys1374Tyr	4121G>A	C1374Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM077266	TGC-TAC	Cys1380Tyr	4139G>A	C1380Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385

FBN1	CM077247	AAT-AAA	Asn1382Lys	4146T>A	N1382K	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM950447	AAT-AGT	Asn1382Ser	4145A>G	N1382S	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM130607	ATG-TTG	Met1384Leu	4150A>T	M1384L	Marfan syndrome	Wang (2013) <i>J Mol Med (Berl)</i> 91:37	PubMed 22772377
FBN1	CM098689	TAC-TGC	Tyr1387Cys	4160A>G	Y1387C	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM040039	TGT-CGT	Cys1389Arg	4165T>C	C1389R	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM088215	TGC-TTC	Cys1391Phe	4172G>T	C1391F	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM054712	TGC-TGA	Cys1391Term	4173C>A	C1391*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM130608	GAT-AAT	Asp1398Asn	4192G>A	D1398N	Marfan syndrome	Wang (2013) <i>J Mol Med (Berl)</i> 91:37	PubMed 22772377
FBN1	CM054725	TGT-CGT	Cys1402Arg	4204T>C	C1402R	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM992327	TGT-TGG	Cys1402Trp	4206T>G	C1402W	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM012599	TGT-TAT	Cys1402Tyr	4205G>A	C1402Y	Marfan syndrome	Comeglio (2001) <i>Hum Mutat</i> 18:251	PubMed 11524736
FBN1	CM970514	GAC-TAC	Asp1404Tyr	4210G>T	D1404Y	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM088218	CTT-CGT	Leu1405Arg	4214T>G	L1405R	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM010042	GAT-GGT	Asp1406Gly	4217A>G	D1406G	Marfan syndrome	Tiecke (2001) <i>Eur J Hum Genet</i> 9:13	PubMed 11175294
FBN1	CM010043	TGC-TTC	Cys1408Phe	4223G>T	C1408F	Marfan syndrome	Tiecke (2001) <i>Eur J Hum Genet</i> 9:13	PubMed 11175294
FBN1	CM098690	TGC-TTC	Cys1420Phe	4259G>T	C1420F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM100477	TGC-TGG	Cys1420Trp	4260C>G	C1420W	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM004134	CTC-TTC	Leu1421Phe	4261C>T	L1421F	Marfan syndrome	Oh (2000) <i>Pediatr Int</i> 42:488	PubMed 11059536
FBN1	CM983868	CCA-GCA	Pro1424Ala	4270C>G	P1424A	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM054685	CCA-TCA	Pro1424Ser	4270C>T	P1424S	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM117998	TAC-TGC	Tyr1427Cys	4280A>G	Y1427C	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM983869	TGT-AGT	Cys1429Ser	4285T>A	C1429S	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM1312099	TGT-TAT	Cys1429Tyr	4286G>A	C1429Y	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM1312100	TGC-CGC	Cys1431Arg	4291T>C	C1431R	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM117722	TGC-TGG	Cys1431Trp	4293C>G	C1431W	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM117721	TGC-TAC	Cys1431Tyr	4292G>A	C1431Y	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM067398	TGC-GGC	Cys1450Gly	4348T>G	C1450G	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM091210	TGC-TAC	Cys1450Tyr	4349G>A	C1450Y	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM098691	TGT-TCT	Cys1456Ser	4367G>C	C1456S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM147406	TGC-CGC	Cys1461Arg	4381T>C	C1461R	Marfan syndrome	Li (2014) <i>Mol Vis</i> 20:1017	PubMed 25053872
FBN1	CM077255	TGT-CGT	Cys1470Arg	4408T>C	C1470R	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM020690	TGT-TAT	Cys1470Tyr	4409G>A	C1470Y	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM055242	GGC-AGC	Gly1475Ser	4423G>A	G1475S	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM098692	TAC-TGC	Tyr1476Cys	4427A>G	Y1476C	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM972810	GAA-TAA	Glu1477Term	4429G>T	E1477*	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM074814	TGC-CGC	Cys1485Arg	4453T>C	C1485R	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM077267	TGC-TAC	Cys1485Tyr	4454G>A	C1485Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM117723	GAT-GCT	Asp1487Ala	4460A>C	D1487A	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM117724	AAT-AAA	Asn1489Lys	4467T>A	N1489K	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM118913	TGC-TTC	Cys1491Phe	4472G>T	C1491F	Marfan syndrome	Lebreiro (2011) <i>Rev Port Cardiol</i> 30:649	PubMed 22005308
FBN1	CM091211	TGC-TAC	Cys1491Tyr	4472G>A	C1491Y	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM980730	TGC-TCC	Cys1497Ser	4490G>C	C1497S	Marfan syndrome	Perez (1998) <i>Hum Mutat</i> 13:84	PubMed 10189222
FBN1	CM062705	AGT-TGT	Ser1499Cys	4495A>T	S1499C	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936

FBN1	CM065181	TGT-TAT	Cys1502Tyr	4505G>A	C1502Y	Marfan syndrome	Ganesh (2006) <i>Arch Ophthalmol</i> 124:205	PubMed 16476890
FBN1	CM145766	TGT-CGT	Cys1511Arg	4531T>C	C1511R	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM940766	TGC-CGC	Cys1513Arg	4537T>C	C1513R	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	CM065178	TGC-TGG	Cys1513Trp	4539C>G	C1513W	Marfan syndrome	Ganesh (2006) <i>Arch Ophthalmol</i> 124:205	PubMed 16476890
FBN1	CM001687	CGA-TGA	Arg1523Term	4567C>T	R1523*	Marfan syndrome	Youil (2000) <i>Hum Mutat</i> 16:92	PubMed 10874320
FBN1	CM096448	TGT-TAT	Cys1526Tyr	4577G>A	C1526Y	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM074841	GAT-TAT	Asp1528Tyr	4582G>T	D1528Y	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM085426	TGC-TGA	Cys1534Term	4602C>A	C1534*	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM010044	CGA-TGA	Arg1539Term	4615C>T	R1539*	Marfan syndrome	Tiecke (2001) <i>Eur J Hum Genet</i> 9:13	PubMed 11175294
FBN1	CM993159	CGA-TGA	Arg1541Term	4621C>T	R1541*	Marfan syndrome	Halliday (1999) <i>Hum Genet</i> 105:587	PubMed 10647894
FBN1	CM145768	ACA-ATA	Thr1547Ile	4640C>T	T1547I	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM098693	TGC-TGG	Cys1563Trp	4689C>G	C1563W	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM054721	TGT-TTT	Cys1564Phe	4691G>T	C1564F	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM040040	TGT-TAT	Cys1564Tyr	4691G>A	C1564Y	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM118004	TGG-TAG	Trp1570Term	4709G>A	W1570*	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM055243	ATG-ACG	Met1576Thr	4727T>C	M1576T	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM940767	GTG-GGG	Val1580Gly	4739T>G	V1580G	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	CM013929	GAG-TAG	Glu1584Term	4750G>T	E1584*	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM127267	TAC-AAC	Tyr1585Asn	4753T>A	Y1585N	Marfan syndrome	Sheikhzadeh (2012) <i>Clin Genet</i> 82:240	PubMed 21883168
FBN1	CM930246	TGT-TTT	Cys1589Phe	4766G>T	C1589F	Marfan syndrome	Tynan (1993) <i>Hum Mol Genet</i> 2:1813	PubMed 8281141
FBN1	CM118005	GAA-TAA	Glu1593Term	4777G>T	E1593*	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM127283	GGT-AGT	Gly1594Ser	4780G>A	G1594S	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM062709	GGT-GTT	Gly1594Val	4781G>T	G1594V	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM972811	GAA-AAA	Glu1605Lys	4813G>A	E1605K	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM970515	TGC-GGC	Cys1610Gly	4828T>G	C1610G	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM096449	CAA-TAA	Gln1618Term	4852C>T	Q1618*	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM074803	TGT-CGT	Cys1622Arg	4864T>C	C1622R	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM098617	CAG-TAG	Gln1630Term	4888C>T	Q1630*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM054701	TGC-GGC	Cys1631Gly	4891T>G	C1631G	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM096453	CGC-CAC	Arg1632His	4895G>A	R1632H	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM067396	GAT-GGT	Asp1642Gly	4925A>G	D1642G	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM013931	CGA-TGA	Arg1644Term	4930C>T	R1644*	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM145769	TGT-TAT	Cys1646Tyr	4937G>A	C1646Y	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM145770	GAT-AAT	Asp1648Asn	4942G>A	D1648N	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM003138	TGT-TAT	Cys1652Tyr	4955G>A	C1652Y	Marfan syndrome	Matsukawa (2000) <i>Hum Mutat</i> 17:71	PubMed 11139245
FBN1	CM096454	TGT-TTT	Cys1658Phe	4973G>T	C1658F	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM100095	GGG-AGG	Gly1661Arg	4981G>A	G1661R	Marfan syndrome	Khau Van Kien (2010) <i>Hum Mutat</i> 31:E1021	PubMed 19802897
FBN1	CM920268	TGT-CGT	Cys1663Arg	4987T>C	C1663R	Marfan syndrome	Dietz (1992) <i>Hum Mutat</i> 1:366	PubMed 1301946
FBN1	CM062717	TGT-TTT	Cys1663Phe	4988G>T	C1663F	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM054716	TGT-TAT	Cys1663Tyr	4988G>A	C1663Y	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM972812	GTT-ATT	Val1667Ile	4999G>A	V1667I	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM992329	TGT-CGT	Cys1672Arg	5014T>C	C1672R	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM992328	TGT-TTT	Cys1672Phe	5015G>T	C1672F	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319

FBN1	CM127644	TGT-TCT	Cys1672Ser	5015G>C	C1672S	Marfan syndrome	Cui (2012) <i>Orphanet J Rare Dis</i> 7:	PubMed 22913777
FBN1	CM085431	TGT-TAT	Cys1672Tyr	5015G>A	C1672Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM085411	TGT-GGT	Cys1674Gly	5020T>G	C1674G	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM077276	TGT-TAT	Cys1674Tyr	5021G>A	C1674Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM117725	TGC-TGA	Cys1687Term	5061C>A	C1687*	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM138475	TGC-TAC	Cys1695Tyr	5084G>A	C1695Y	Marfan syndrome	Cecchi (2013) <i>Am J Med Genet A</i> 161:2305	PubMed 23897642
FBN1	CM098694	TGC-TAC	Cys1719Tyr	5156G>A	C1719Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM074843	TGC-TAC	Cys1720Tyr	5159G>A	C1720Y	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM983870	TGT-TAT	Cys1721Tyr	5162G>A	C1721Y	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM091212	GCG-CCG	Ala1728Pro	5182G>C	A1728P	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM055255	TGG-TGA	Trp1729Term	5187G>A	W1729*	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM085401	CAG-TAG	Gln1735Term	5203C>T	Q1735*	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM145771	CAA-TAA	Gln1751Term	5251C>T	Q1751*	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM067401	GAT-CAT	Asp1766His	5296G>C	D1766H	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM020691	GAG-AAG	Glu1769Lys	5305G>A	E1769K	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM040041	TGC-TTC	Cys1770Phe	5309G>T	C1770F	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99 Huang (2004) <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i> 21:562	PubMed 14695540 PubMed 15583982
FBN1	CM045613	TGC-TAC	Cys1770Tyr	5309G>A	C1770Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM077260	GAG-TAG	Glu1772Term	5314G>T	E1772*	Marfan syndrome	Khau Van Kien (2010) <i>Hum Mutat</i> 31:E1021	PubMed 19802897
FBN1	CM100096	GGA-GAA	Gly1780Glu	5339G>A	G1780E	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM096455	GTG-GCG	Val1781Ala	5342T>C	V1781A	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098695	TGT-TAT	Cys1782Tyr	5345G>A	C1782Y	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM013932	CGA-CCA	Arg1790Pro	5369G>C	R1790P	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM054694	CGA-TGA	Arg1790Term	5368C>T	R1790*	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM055244	TGT-CGT	Cys1791Arg	5371T>C	C1791R	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM077279	TGT-TTT	Cys1791Phe	5372G>T	C1791F	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM013933	TGT-TAT	Cys1791Tyr	5372G>A	C1791Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM077249	TGT-CGT	Cys1793Arg	5377T>C	C1793R	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM040042	TGT-TGG	Cys1793Trp	5379T>G	C1793W	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM074859	TGT-TAT	Cys1793Tyr	5378G>A	C1793Y	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM074823	GGA-GTA	Gly1796Val	5387G>T	G1796V	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM062703	AAG-TAG	Lys1802Term	5404A>T	K1802*	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM022004	TGT-TCT	Cys1806Ser	5417G>C	C1806S	Marfan syndrome	Rommel (2002) <i>Hum Mutat</i> 20:406	PubMed 12402346
FBN1	CM022785	TGT-TAT	Cys1806Tyr	5417G>A	C1806Y	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM074812	GAG-AAG	Glu1811Lys	5431G>A	E1811K	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM093613	TGT-CGT	Cys1812Arg	5434T>C	C1812R	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM100621	TGC-GGC	Cys1818Gly	5452T>G	C1818G	Marfan syndrome	Hayashi (2013) <i>J Dermatol Sci</i> 72:325	PubMed 23978319
FBN1	CM1310986	TGC-TGA	Cys1818Term	5454C>A	C1818*	Marfan syndrome	Perez (1998) <i>Hum Mutat</i> 13:84	PubMed 10189222
FBN1	CM980731	TGC-TAC	Cys1818Tyr	5453G>A	C1818Y	Marfan syndrome	Perez (1998) <i>Hum Mutat</i> 13:84	PubMed 10189222
FBN1	CM980732	GAA-TAA	Glu1823Term	5467G>T	E1823*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098696	TGC-TAC	Cys1824Tyr	5471G>A	C1824Y	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM093614	AAC-AGC	Asn1826Ser	5477A>G	N1826S	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM100478	ACT-CCT	Thr1827Pro	5479A>C	T1827P	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM074808	AGC-TGC	Ser1830Cys	5488A>T	S1830C	Marfan syndrome		

FBN1	CM983871	CGC-TGC	Arg1832Cys	5494C>T	R1832C	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM077256	TGT-CGT	Cys1833Arg	5497T>C	C1833R	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM1314086	TGT-GGT	Cys1833Gly	5497T>G	C1833G	Marfan syndrome	Pees (2013) <i>Clin Genet</i> epub:epub	PubMed 24199744
FBN1	CM990592	TGT-TCT	Cys1833Ser	5498G>C	C1833S	Marfan syndrome	El-Aleem (1999) <i>Hum Mutat</i> 14:181	PubMed 10425041
FBN1	CM074864	TGT-TTT	Cys1835Phe	5504G>T	C1835F	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM993160	TGT-TAT	Cys1835Tyr	5504G>A	C1835Y	Marfan syndrome	Halliday (1999) <i>Hum Genet</i> 105:587	PubMed 10647894
FBN1	CM960638	CCC-TCC	Pro1837Ser	5509C>T	P1837S	Marfan syndrome	Milewicz (1996) <i>Circulation</i> 94:2708	PubMed 8941093
FBN1	CM065177	GGC-TGC	Gly1838Cys	5512G>T	G1838C	Marfan syndrome	Ganesh (2006) <i>Arch Ophthalmol</i> 124:205	PubMed 16476890
FBN1	CM062710	TGC-CGC	Cys1847Arg	5539T>C	C1847R	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM074849	TGC-TGG	Cys1847Trp	5541C>G	C1847W	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM098608	CAA-TAA	Gln1854Term	5560C>T	Q1854*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM085406	TGC-CGC	Cys1860Arg	5578T>C	C1860R	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM098697	TGC-TTC	Cys1860Phe	5579G>T	C1860F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM085423	TGC-TAC	Cys1860Tyr	5579G>A	C1860Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM130603	TGC-CGC	Cys1865Arg	5593T>C	C1865R	Marfan syndrome	Wang (2013) <i>J Mol Med (Berl)</i> 91:37	PubMed 22772377
FBN1	CM096456	ACA-CCA	Thr1868Pro	5602A>C	T1868P	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM054711	TGC-TAC	Cys1876Tyr	5627G>A	C1876Y	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM074829	GGT-GAT	Gly1879Asp	5636G>A	G1879D	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM098698	GGT-TGT	Gly1879Cys	5635G>T	G1879C	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM055241	ACC-ATC	Thr1887Ile	5660C>T	T1887I	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM098699	TGC-TTC	Cys1889Phe	5666G>T	C1889F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM100479	TGC-TCC	Cys1889Ser	5666G>C	C1889S	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM108651	GAC-CAC	Asp1891His	5671G>C	D1891H	Marfan syndrome	Hilhorst-Hofstee (2010) <i>Hum Mutat</i> 31:E1915	PubMed 20886638
FBN1	CM970516	AAT-AAA	Asn1893Lys	5679T>A	N1893K	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM085421	GAA-AAA	Glu1894Lys	5680G>A	E1894K	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM004135	TGT-CGT	Cys1895Arg	5683T>C	C1895R	Marfan syndrome	Oh (2000) <i>Pediatr Int</i> 42:488	PubMed 11059536
FBN1	CM098700	TGT-TAT	Cys1895Tyr	5684G>A	C1895Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM096457	TGT-TTT	Cys1900Phe	5699G>T	C1900F	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM098701	TGT-TCT	Cys1900Ser	5699G>C	C1900S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM054717	TGT-TAT	Cys1900Tyr	5699G>A	C1900Y	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM127284	GGA-AGA	Gly1903Arg	5707G>A	G1903R	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM098702	TGC-CGC	Cys1905Arg	5713T>C	C1905R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM145779	AAC-AAG	Asn1907Lys	5721C>G	N1907K	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM022786	ACA-ATA	Thr1908Ile	5723C>T	T1908I	Marfan syndrome	Rommel (2002) <i>Hum Mutat</i> 20:406	PubMed 12402346
FBN1	CM098703	ATT-AGT	Ile1909Ser	5726T>G	I1909S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM013934	ATT-ACT	Ile1909Thr	5726T>C	I1909T	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM980733	GGT-GTT	Gly1910Val	5729G>T	G1910V	Marfan syndrome	Perez (1998) <i>Hum Mutat</i> 13:84	PubMed 10189222
FBN1	CM013935	CGC-AGC	Arg1915Ser	5743C>A	R1915S	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM098704	TGC-AGC	Cys1916Ser	5746T>A	C1916S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM145781	TGC-TAC	Cys1916Tyr	5747G>A	C1916Y	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM022787	GGT-GAT	Gly1919Asp	5756G>A	G1919D	Marfan syndrome	Rommel (2002) <i>Hum Mutat</i> 20:406	PubMed 12402346
FBN1	CM096458	GGT-GTT	Gly1919Val	5756G>T	G1919V	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM118914	AAT-GAT	Asn1926Asp	5776A>G	N1926D	Marfan syndrome	Lebreiro (2011) <i>Rev Port Cardiol</i> 30:649	PubMed 22005308

FBN1	CM950448	TGT-CGT	Cys1928Arg	5782T>C	C1928R	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM055247	TGT-GGT	Cys1928Gly	5782T>G	C1928G	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM090179	TGT-TCT	Cys1928Ser	5783G>C	C1928S	Marfan syndrome	Van Dijk (2009) <i>Eur J Med Genet</i> 52:1	PubMed 19059503
FBN1	CM055257	TGT-TAT	Cys1928Tyr	5783G>A	C1928Y	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM972813	GAT-AAT	Asp1930Asn	5788G>A	D1930N	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM127266	GAT-GGT	Asp1930Gly	5789A>G	D1930G	Marfan syndrome	Sheikhzadeh (2012) <i>Clin Genet</i> 82:240	PubMed 21883168
FBN1	CM096459	GAA-GTA	Glu1933Val	5798A>T	E1933V	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM085407	TGT-GGT	Cys1934Gly	5800T>G	C1934G	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM117726	TGT-AGT	Cys1934Ser	5800T>A	C1934S	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM042041	GGA-TGA	Gly1937Term	5809G>T	G1937*	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM054691	CAA-TAA	Gln1946Term	5836C>T	Q1946*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM145782	TGC-TAC	Cys1947Tyr	5840G>A	C1947Y	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM096460	TTC-TGC	Phe1954Cys	5861T>G	F1954C	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM067397	TGC-CGC	Cys1956Arg	5866T>C	C1956R	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM117999	TGC-TAC	Cys1958Tyr	5873G>A	C1958Y	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM013936	TGT-TAT	Cys1971Tyr	5912G>A	C1971Y	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM117727	GAA-GGA	Glu1976Gly	5927A>G	E1976G	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM077264	GAA-AAA	Glu1976Lys	5926G>A	E1976K	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM021596	TGT-CGT	Cys1977Arg	5929T>C	C1977R	Marfan syndrome	Halliday (2002) <i>J Med Genet</i> 39:589	PubMed 12161601
FBN1	CM085432	TGT-TGG	Cys1977Trp	5931T>G	C1977W	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM983872	TGT-TAT	Cys1977Tyr	5930G>A	C1977Y	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM117728	TGT-CGT	Cys1984Arg	5950T>C	C1984R	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM012600	GGT-CGT	Gly1987Arg	5959G>C	G1987R	Marfan syndrome	Comeglio (2001) <i>Hum Mutat</i> 18:251	PubMed 11524736
FBN1	CM098705	GGT-GTT	Gly1987Val	5960G>T	G1987V	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM022005	TGC-TAC	Cys1998Tyr	5993G>A	C1998Y	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM077270	TGC-TCC	Cys2000Ser	5999G>C	C2000S	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM127285	TGT-GGT	Cys2011Gly	6031T>G	C2011G	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM108652	GAT-TAT	Asp2013Tyr	6037G>T	D2013Y	Marfan syndrome	Hilhorst-Hofstee (2010) <i>Hum Mutat</i> 31:E1915	PubMed 20886638
FBN1	CM098706	GAG-AAG	Glu2016Lys	6046G>A	E2016K	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM972814	TGT-CGT	Cys2017Arg	6049T>C	C2017R	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM127268	GAA-AAA	Glu2019Lys	6055G>A	E2019K	Marfan syndrome	Sheikhzadeh (2012) <i>Clin Genet</i> 82:240	PubMed 21883168
FBN1	CM972815	ATT-ACT	Ile2023Thr	6068T>C	I2023T	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM098627	TGC-TGA	Cys2029Term	6087C>A	C2029*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098707	TGT-TTT	Cys2038Phe	6113G>T	C2038F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM055258	TGT-TAT	Cys2038Tyr	6113G>A	C2038Y	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM972816	CTG-GTG	Leu2039Val	6115C>G	L2039V	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM145784	TGT-TAT	Cys2040Tyr	6119G>A	C2040Y	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM992330	TGC-TTC	Cys2053Phe	6158G>T	C2053F	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM074852	TGC-TGA	Cys2053Term	6159C>A	C2053*	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM003139	CAA-CCA	Gln2054Pro	6161A>C	Q2054P	Marfan syndrome	Matsukawa (2000) <i>Hum Mutat</i> 17:71	PubMed 11139245
FBN1	CM013937	CAA-TAA	Gln2054Term	6160C>T	Q2054*	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM972817	CGA-CAA	Arg2057Gln	6170G>A	R2057Q	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM972818	CGA-TGA	Arg2057Term	6169C>T	R2057*	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652

FBN1	CM098708	TGT-CGT	Cys2061Arg	6181T>C	C2061R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM092467	TGT-TTT	Cys2061Phe	6182G>T	C2061F	Marfan syndrome	Zhao (2009) <i>Mol Vis</i> 15:826	PubMed 19390640
FBN1	CM085420	TAT-TAA	Tyr2062Term	6186T>A	Y2062*	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM085412	AAG-TAG	Lys2064Term	6190A>T	K2064*	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM062713	TCC-TGC	Ser2079Cys	6236C>G	S2079C	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM100466	CAG-TAG	Gln2081Term	6241C>T	Q2081*	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM098709	TGC-TTC	Cys2083Phe	6248G>T	C2083F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM127286	TGC-TCC	Cys2084Ser	6251G>C	C2084S	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM093615	TGC-TGG	Cys2084Trp	6252C>G	C2084W	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM085424	TGC-TAC	Cys2084Tyr	6251G>A	C2084Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM055246	TGT-CGT	Cys2085Arg	6253T>C	C2085R	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM118000	TGC-TTC	Cys2099Phe	6296G>T	C2099F	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM970517	TGC-TGG	Cys2099Trp	6297C>G	C2099W	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM098710	TGC-TAC	Cys2099Tyr	6296G>A	C2099Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098711	CCC-TCC	Pro2100Ser	6298C>T	P2100S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM020692	ACG-AAG	Thr2101Lys	6302C>A	T2101K	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM098618	CAG-TAG	Gln2109Term	6325C>T	Q2109*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM020136	TGT-CGT	Cys2111Arg	6331T>C	C2111R	Marfan syndrome	Korkko (2002) <i>J Med Genet</i> 39:34	PubMed 11826022
FBN1	CM970518	TGT-TAT	Cys2111Tyr	6332G>A	C2111Y	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM983873	TAT-TAA	Tyr2113Term	6339T>A	Y2113*	Marfan syndrome	Collod-Beroud (1998) <i>Nucleic Acids Res</i> 26:229	PubMed 9399842
FBN1	CM950449	TAT-TAG	Tyr2113Term	6339T>G	Y2113*	Marfan syndrome	Dietz (1993) <i>Science</i> 259:680	PubMed 8430317
FBN1	CM074799	ATC-ATG	Ile2118Met	6354C>G	I2118M	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM940768	GAT-GAA	Asp2127Glu	6381T>A	D2127E	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	CM003140	GAT-TAT	Asp2127Tyr	6379G>T	D2127Y	Marfan syndrome	Matsukawa (2000) <i>Hum Mutat</i> 17:71	PubMed 11139245
FBN1	CM106074	GAT-GTT	Asp2127Val	6380A>T	D2127V	Marfan syndrome	Evangelisti (2010) <i>Eur J Med Genet</i> 53:299	PubMed 20538085
FBN1	CM067392	GAA-AAA	Glu2130Lys	6388G>A	E2130K	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM100097	GGA-AGA	Gly2140Arg	6418G>A	G2140R	Marfan syndrome	Khau Van Kien (2010) <i>Hum Mutat</i> 31:E1021	PubMed 19802897
FBN1	CM100098	GGA-GAA	Gly2140Glu	6419G>A	G2140E	Marfan syndrome	Khau Van Kien (2010) <i>Hum Mutat</i> 31:E1021	PubMed 19802897
FBN1	CM990593	TGC-TAC	Cys2142Tyr	6425G>A	C2142Y	Marfan syndrome	El-Aleem (1999) <i>Hum Mutat</i> 14:181	PubMed 10425041
FBN1	CM1312104	ATC-AAC	Ile2143Asn	6428T>A	I2143N	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM074805	AAT-GAT	Asn2144Asp	6430A>G	N2144D	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM930247	AAT-AGT	Asn2144Ser	6431A>G	N2144S	Marfan syndrome	Hewett (1993) <i>Hum Mol Genet</i> 2:475	PubMed 8504310
FBN1	CM074833	ACA-CCA	Thr2145Pro	6433A>C	T2145P	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM145787	TAT-TGT	Tyr2149Cys	6446A>G	Y2149C	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM098712	TGC-CGC	Cys2151Arg	6451T>C	C2151R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM940769	TGC-TGG	Cys2151Trp	6453C>G	C2151W	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	CM012601	TGT-TAT	Cys2153Tyr	6458G>A	C2153Y	Marfan syndrome	Comeglio (2001) <i>Hum Mutat</i> 18:251	PubMed 11524736
FBN1	CM054687	GCA-CCA	Ala2160Pro	6478G>C	A2160P	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM117730	GAT-AAT	Asp2166Asn	6496G>A	D2166N	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM077259	GAT-GGT	Asp2168Gly	6503A>G	D2168G	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM025496	TGT-TTT	Cys2170Phe	6509G>T	C2170F	Marfan syndrome	Ng (2002) <i>Genome Res</i> 12:436	PubMed 11875032
FBN1	CM067390	TGT-AGT	Cys2170Ser	6508T>A	C2170S	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	BM1157862	GGC-AGC	Gly2173Ser	6517G>A	G2173S	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952

Accession	Gene	Variant	Protein	Position	RefSeq	Protein	Source	PubMed
FBN1	HM080072	GAA-TAA	Glu2189Term	6565G>T	E2189*	Marfan syndrome	Ellison (2009) <i>Hum Genet</i> 125:338	Source: Online submission
FBN1	CM040043	GAG-TAG	Glu2193Term	6577G>T	E2193*	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM116657	GAG-TAG	Glu2194Term	6580G>T	E2194*	Marfan syndrome	Díaz de Bustamante (2011) <i>Rev Esp Cardiol</i> epub:epub	PubMed 21840105
FBN1	CM013939	GGA-TGA	Gly2195Term	6583G>T	G2195*	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM145790	TTC-TCC	Phe2219Ser	6656T>C	F2219S	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM003141	CGA-TGA	Arg2220Term	6658C>T	R2220*	Marfan syndrome	Matsukawa (2000) <i>Hum Mutat</i> 17:71	PubMed 11139245
FBN1	CM992331	TGT-CGT	Cys2221Arg	6661T>C	C2221R	Marfan syndrome	Schrijver (1999) <i>Am J Hum Genet</i> 65:1007	PubMed 10486319
FBN1	CM022006	TGT-GGT	Cys2221Gly	6661T>G	C2221G	Marfan syndrome	Katzke (2002) <i>Hum Mutat</i> 20:197	PubMed 12203992
FBN1	CM054722	TGT-TTT	Cys2221Phe	6662G>T	C2221F	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM920269	TGT-TCT	Cys2221Ser	6662G>C	C2221S	Marfan syndrome	Dietz (1992) <i>Hum Mutat</i> 1:366	PubMed 1301946
FBN1	CM013940	AAC-CAC	Asn2223His	6667A>C	N2223H	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM127287	TGT-CGT	Cys2232Arg	6694T>C	C2232R	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM972858	TGT-TAT	Cys2232Tyr	6695G>A	C2232Y	Marfan syndrome	Pepe (1997) <i>J Mol Cell Cardiol</i> 29:1877	PubMed 9236141
FBN1	CM1314087	CCC-CGC	Pro2233Arg	6698C>G	P2233R	Marfan syndrome	Pees (2013) <i>Clin Genet</i> epub:epub	PubMed 24199744
FBN1	CM067394	GTG-ATG	Val2234Met	6700G>A	V2234M	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM098714	GAT-GGT	Asp2247Gly	6740A>G	D2247G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM067387	GAT-TAT	Asp2247Tyr	6739G>T	D2247Y	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM100480	GAT-GTT	Asp2247Val	6740A>T	D2247V	Marfan syndrome	Yoo (2010) <i>Clin Genet</i> 77:177	PubMed 19863550
FBN1	CM022788	TGT-CGT	Cys2251Arg	6751T>C	C2251R	Marfan syndrome	Rommel (2002) <i>Hum Mutat</i> 20:406	PubMed 12402346
FBN1	CM995303	TGT-AGT	Cys2251Ser	6751T>A	C2251S	Marfan syndrome	Yuan (1999) <i>Hum Mutat</i> 14:440	PubMed 10533071
FBN1	CM970519	TGT-CGT	Cys2258Arg	6772T>C	C2258R	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM993161	TGT-TAT	Cys2258Tyr	6773G>A	C2258Y	Marfan syndrome	Halliday (1999) <i>Hum Genet</i> 105:587	PubMed 10647894
FBN1	CM950450	CAA-TAA	Gln2262Term	6784C>T	Q2262*	Marfan syndrome	Nijbroek (1995) <i>Am J Hum Genet</i> 57:8	PubMed 7611299
FBN1	CM098715	TGC-TTC	Cys2265Phe	6794G>T	C2265F	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM056959	TGC-TAC	Cys2265Tyr	6794G>A	C2265Y	Marfan syndrome	Baumgartner (2005) <i>Methods Inf Med</i> 44:487	PubMed 16342915
FBN1	CM098716	AAC-ATC	Asn2267Ile	6800A>T	N2267I	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM077261	GGC-GAC	Gly2270Asp	6809G>A	G2270D	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM074850	TGC-TGG	Cys2274Trp	6822C>G	C2274W	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM094366	TGT-TAT	Cys2276Tyr	6827G>A	C2276Y	Marfan syndrome	Voermans (2009) <i>Clin Genet</i> 76:25	PubMed 19659760
FBN1	CM145793	CAG-TAG	Gln2281Term	6841C>T	Q2281*	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM970520	CGG-TGG	Arg2282Trp	6844C>T	R2282W	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM085402	CCT-ACT	Pro2284Thr	6850C>A	P2284T	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM098718	TGT-TAT	Cys2289Tyr	6866G>A	C2289Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM106075	GAT-AAT	Asp2291Asn	6871G>A	D2291N	Marfan syndrome	Evangelisti (2010) <i>Eur J Med Genet</i> 53:299	PubMed 20538085
FBN1	CM042042	GAA-GCA	Glu2294Ala	6881A>C	E2294A	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM094365	TGT-CGT	Cys2295Arg	6883T>C	C2295R	Marfan syndrome	Voermans (2009) <i>Clin Genet</i> 76:25	PubMed 19659760
FBN1	CM020693	TGT-TAT	Cys2295Tyr	6884G>A	C2295Y	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM145794	CAG-TAG	Gln2296Term	6886C>T	Q2296*	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM093616	AAG-TAG	Lys2298Term	6892A>T	K2298*	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM118909	TGT-TGA	Cys2302Term	6906T>A	C2302*	Marfan syndrome	Lebreiro (2011) <i>Rev Port Cardiol</i> 30:649	PubMed 22005308
FBN1	CM098719	TGC-CGC	Cys2307Arg	6919T>C	C2307R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM950451	TGC-TCC	Cys2307Ser	6920G>C	C2307S	Marfan syndrome	Dietz (1995) <i>Hum Mol Genet</i> 4:1799	PubMed 8541880

FBN1	CM145795	TGC-TAC	Cys2307Tyr	6920G>A	C2307Y	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM098720	TGT-CGT	Cys2316Arg	6946T>C	C2316R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM117731	TGT-CGT	Cys2318Arg	6952T>C	C2318R	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM098721	TGT-TAT	Cys2318Tyr	6953G>A	C2318Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM091213	TGC-TGA	Cys2331Term	6993C>A	C2331*	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM145796	TGC-TAC	Cys2331Tyr	6992G>A	C2331Y	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM022007	CGG-TGG	Arg2335Trp	7003C>T	R2335W	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM088216	TGC-CGC	Cys2339Arg	7015T>C	C2339R	Marfan syndrome	Rybczynski (2008) <i>Am J Med Genet A</i> 146A:3157	PubMed 19012347
FBN1	CM118001	TGC-GGC	Cys2339Gly	7015T>G	C2339G	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM022008	TGC-TAC	Cys2339Tyr	7016G>A	C2339Y	Marfan syndrome	Robinson (2002) <i>Hum Mutat</i> 20:153	PubMed 12203987
FBN1	CM067405	TGT-TAT	Cys2365Tyr	7094G>A	C2365Y	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM096463	TGG-CGG	Trp2371Arg	7111T>C	W2371R	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM117735	TGG-TAG	Trp2371Term	7112G>A	W2371*	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM098619	CAG-TAG	Gln2381Term	7141C>T	Q2381*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM055249	GCT-ACT	Ala2385Thr	7153G>A	A2385T	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494 Rand-Hendriksen (2007) <i>Am J Med Genet A</i> 143A:1968	PubMed 16222657 PubMed 17663468
FBN1	CM074168	TGT-AGT	Cys2390Ser	7168T>A	C2390S	Marfan syndrome	Halliday (1999) <i>Hum Genet</i> 105:587	PubMed 10647894
FBN1	CM993162	CGA-TGA	Arg2394Term	7180C>T	R2394*	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM013941	TGC-TAC	Cys2406Tyr	7217G>A	C2406Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098722	TGC-CGC	Cys2413Arg	7237T>C	C2413R	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM062706	CGA-CAA	Arg2414Gln	7241G>A	R2414Q	Marfan syndrome	Korkko (2002) <i>J Med Genet</i> 39:34	PubMed 11826022
FBN1	CM020137	CGA-TGA	Arg2414Term	7240C>T	R2414*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098609	TAT-TAA	Tyr2425Term	7275T>A	Y2425*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098723	TGT-CGT	Cys2429Arg	7285T>C	C2429R	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098724	TGT-GGT	Cys2442Gly	7324T>G	C2442G	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM117732	TGT-AGT	Cys2442Ser	7324T>A	C2442S	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM098628	TGT-TGA	Cys2442Term	7326T>A	C2442*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM040044	TGT-TGG	Cys2442Trp	7326T>G	C2442W	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM108653	GAT-GGT	Asp2444Gly	7331A>G	D2444G	Marfan syndrome	Hilhorst-Hofstee (2010) <i>Hum Mutat</i> 31:E1915	PubMed 20886638
FBN1	CM940770	GAG-AAG	Glu2447Lys	7339G>A	E2447K	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	CM054696	GAG-TAG	Glu2447Term	7339G>T	E2447*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM062711	TGC-CGC	Cys2448Arg	7342T>C	C2448R	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM091214	TGC-TCC	Cys2455Ser	7364G>C	C2455S	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM077268	TGC-TAC	Cys2455Tyr	7364G>A	C2455Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM077246	AAA-AGA	Lys2460Arg	7379A>G	K2460R	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM098725	AAC-ACC	Asn2461Thr	7382A>C	N2461T	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM021597	TAC-TAA	Tyr2466Term	7398C>A	Y2466*	Marfan syndrome	Halliday (2002) <i>J Med Genet</i> 39:589	PubMed 12161601
FBN1	CM021598	CAG-TAG	Gln2467Term	7399C>T	Q2467*	Marfan syndrome	Halliday (2002) <i>J Med Genet</i> 39:589	PubMed 12161601
FBN1	CM062716	TGC-TGA	Cys2470Term	7410C>A	C2470*	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM085425	TGC-TAC	Cys2470Tyr	7409G>A	C2470Y	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM098610	CAA-TAA	Gln2477Term	7429C>T	Q2477*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM014707	TGC-TAC	Cys2483Tyr	7448G>A	C2483Y	Marfan syndrome	Judge (2001) <i>Am J Med Genet</i> 99:39	PubMed 11170092
FBN1	CM970521	TGT-CGT	Cys2489Arg	7465T>C	C2489R	Marfan syndrome	Hayward (1997) <i>Hum Mutat</i> 10:280	PubMed 9338581
FBN1	CM020373	TGT-GGT	Cys2489Gly	7465T>G	C2489G	Marfan syndrome	Loeys (2002) <i>Prenat Diagn</i> 22:22	PubMed 11810645

FBN1	CM067406	TGT-TAT	Cys2489Tyr	7466G>A	C2489Y	Marfan syndrome	Valiev (2006) <i>Mol Biol (Mosk)</i> 40:1021	PubMed 17209430
FBN1	CM1312101	TGC-CGC	Cys2496Arg	7486T>C	C2496R	Marfan syndrome	Aalberts (2014) <i>Gene</i> 534:40	PubMed 24161884
FBN1	CM096464	TGC-TAC	Cys2496Tyr	7487G>A	C2496Y	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM054689	TGT-CGT	Cys2500Arg	7498T>C	C2500R	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM025899	TGT-AGT	Cys2500Ser	7498T>A	C2500S	Marfan syndrome	Ng (2002) <i>Genome Res</i> 12:436	PubMed 11875032
FBN1	CM054718	TGT-TAT	Cys2500Tyr	7499G>A	C2500Y	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM098726	TGC-TAC	Cys2509Tyr	7526G>A	C2509Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM940771	TGT-CGT	Cys2511Arg	7531T>C	C2511R	Marfan syndrome	Kainulainen (1994) <i>Nat Genet</i> 6:64	PubMed 8136837
FBN1	CM995304	TGT-TAT	Cys2511Tyr	7532G>A	C2511Y	Marfan syndrome	Yuan (1999) <i>Hum Mutat</i> 14:440	PubMed 10533071
FBN1	CM025900	GGA-AGA	Gly2514Arg	7540G>A	G2514R	Marfan syndrome	Ng (2002) <i>Genome Res</i> 12:436	PubMed 11875032
FBN1	CM077253	GGA-TGA	Gly2514Term	7540G>T	G2514*	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM074795	ACG-ATG	Thr2520Met	7559C>T	T2520M	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM000514	TGC-TCC	Cys2522Ser	7565G>C	C2522S	Marfan syndrome	Palz (2000) <i>Am J Med Genet</i> 91:212	PubMed 10756346
FBN1	CM093617	TGC-TAC	Cys2522Tyr	7565G>A	C2522Y	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM000515	TGC-TGA	Cys2535Term	7605C>A	C2535*	Marfan syndrome	Palz (2000) <i>Am J Med Genet</i> 91:212	PubMed 10756346
FBN1	CM054714	TGC-TGG	Cys2535Trp	7605C>G	C2535W	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM012602	GGG-AGG	Gly2536Arg	7606G>A	G2536R	Marfan syndrome	Comeglio (2001) <i>Hum Mutat</i> 18:251	PubMed 11524736
FBN1	CM074809	CAG-TAG	Gln2542Term	7624C>T	Q2542*	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM127288	CCT-TCT	Pro2545Ser	7633C>T	P2545S	Marfan syndrome	Robinson (2012) <i>Clin Genet</i> 82:223	PubMed 21895641
FBN1	CM1314088	TGT-TAT	Cys2550Tyr	7649G>A	C2550Y	Marfan syndrome	Pees (2013) <i>Clin Genet</i> epub:epub	PubMed 24199744
FBN1	CM127886	CAG-TAG	Gln2553Term	7657C>T	Q2553*	Marfan syndrome	Song (2012) <i>J Genet</i> 91:233	PubMed 22942097
FBN1	CM127269	CGG-CAG	Arg2554Gln	7661G>A	R2554Q	Marfan syndrome	Sheikhzadeh (2012) <i>Clin Genet</i> 82:240	PubMed 21883168
FBN1	CM074824	GGA-GTA	Gly2555Val	7664G>T	G2555V	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM077263	TCA-TAA	Ser2557Term	7670C>A	S2557*	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM085413	ACC-CCC	Thr2561Pro	7681A>C	T2561P	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM077254	GTG-ATG	Val2568Met	7702G>A	V2568M	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM054695	GAG-AAG	Glu2570Lys	7708G>A	E2570K	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM054703	TGT-CGT	Cys2571Arg	7711T>C	C2571R	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM074867	TGT-TGA	Cys2571Term	7713T>A	C2571*	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM138995	CGC-TGC	Arg2576Cys	7726C>T	R2576C	Marfan syndrome	Hogue (2013) <i>Clin Genet</i> 84:392	PubMed 23278365
FBN1	CM972859	TGC-CGC	Cys2577Arg	7729T>C	C2577R	Marfan syndrome	Pepe (1997) <i>J Mol Cell Cardiol</i> 29:1877	PubMed 9236141
FBN1	CM074844	TGC-TAC	Cys2577Tyr	7730G>A	C2577Y	Marfan syndrome	Comeglio (2007) <i>Hum Mutat</i> 28:928	PubMed 17657824
FBN1	CM013942	TGC-TTC	Cys2581Phe	7742G>T	C2581F	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM022009	TGC-TGG	Cys2581Trp	7743C>G	C2581W	Marfan syndrome	Korkko (2002) <i>J Med Genet</i> 39:34	PubMed 11826022
FBN1	CM077269	TGC-TAC	Cys2581Tyr	7742G>A	C2581Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM972820	ATT-ACT	Ile2585Thr	7754T>C	I2585T	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM098727	GGG-TGG	Gly2586Trp	7756G>T	G2586W	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM098728	TGC-TAC	Cys2590Tyr	7769G>A	C2590Y	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM054697	TGC-AGC	Cys2592Ser	7774T>A	C2592S	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM098729	GGC-AGC	Gly2595Ser	7783G>A	G2595S	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM095638	TAC-TAA	Tyr2600Term	7800C>A	Y2600*	Marfan syndrome	Magyar (2009) <i>Hum Mutat</i> 30:1355	PubMed 19618372
FBN1	CM056955	CAG-TAG	Gln2601Term	7801C>T	Q2601*	Marfan syndrome	Baumgartner (2005) <i>Methods Inf Med</i> 44:487	PubMed 16342915
FBN1	CM098629	TGG-TGA	Trp2602Term	7806G>A	W2602*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843

FBN1	CM091215	TGT-TGA	Cys2605Term	7815T>A	C2605*	Marfan syndrome	Söylen (2009) <i>Clin Genet</i> 75:265	PubMed 19159394
FBN1	CM055259	TGT-TAT	Cys2605Tyr	7814G>A	C2605Y	Marfan syndrome	Rommel (2005) <i>Hum Mutat</i> 26:529	PubMed 16220557
FBN1	CM108654	GAT-AAT	Asp2607Asn	7819G>A	D2607N	Marfan syndrome	Hilhorst-Hofstee (2010) <i>Hum Mutat</i> 31:E1915	PubMed 20886638
FBN1	CM972821	GAT-GGT	Asp2607Gly	7820A>G	D2607G	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM972822	GAA-AAA	Glu2610Lys	7828G>A	E2610K	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM077250	ATC-GTC	Ile2616Val	7846A>G	I2616V	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM098730	TGC-TGG	Cys2617Trp	7851C>G	C2617W	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM020694	GGA-AGA	Gly2618Arg	7852G>A	G2618R	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM1314089	TCC-GCC	Ser2621Ala	7861T>G	S2621A	Marfan syndrome	Pees (2013) <i>Clin Genet</i> epub:epub	PubMed 24199744
FBN1	CM042043	TGT-AGT	Cys2622Ser	7864T>A	C2622S	Marfan syndrome	Loeys (2004) <i>Hum Mutat</i> 24:140	PubMed 15241795
FBN1	CM970522	CAC-CCC	His2623Pro	7868A>C	H2623P	Marfan syndrome	Collod-Beroud (1997) <i>Nucleic Acids Res</i> 25:147	PubMed 9016526
FBN1	CM015153	AAC-AAG	Asn2624Lys	7872C>G	N2624K	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM000516	AAC-AGC	Asn2624Ser	7871A>G	N2624S	Marfan syndrome	Palz (2000) <i>Am J Med Genet</i> 91:212	PubMed 10756346
FBN1	CM098731	AAC-ACC	Asn2624Thr	7871A>C	N2624T	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM950452	GGG-AGG	Gly2627Arg	7879G>A	G2627R	Marfan syndrome	Dietz (1995) <i>Hum Mol Genet</i> 4:1799	PubMed 8541880
FBN1	CM054706	TAC-TGC	Tyr2629Cys	7886A>G	Y2629C	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM145808	TGC-TAC	Cys2631Tyr	7892G>A	C2631Y	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM141673	TGT-CGT	Cys2633Arg	7897T>C	C2633R	Marfan syndrome	Stevic (2014) <i>Biochem Genet</i> epub:epub	PubMed 24504995
FBN1	CM020695	TAT-AAT	Tyr2639Asn	7915T>A	Y2639N	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM020696	TAT-TGT	Tyr2639Cys	7916A>G	Y2639C	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM117733	TGC-CGC	Cys2646Arg	7936T>C	C2646R	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM020139	TGT-GGT	Cys2652Gly	7954T>G	C2652G	Marfan syndrome	Korkko (2002) <i>J Med Genet</i> 39:34	PubMed 11826022
FBN1	CM077277	TGT-TAT	Cys2652Tyr	7955G>A	C2652Y	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM098620	CAG-TAG	Gln2656Term	7966C>T	Q2656*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM095639	TGC-TGA	Cys2659Term	7977C>A	C2659*	Marfan syndrome	Magyar (2009) <i>Hum Mutat</i> 30:1355	PubMed 19618372
FBN1	CM062704	AGC-CGC	Ser2660Arg	7978A>C	S2660R	Marfan syndrome	Sakai (2006) <i>Am J Med Genet A</i> 140A:1719	PubMed 16835936
FBN1	CM043997	TGT-TCT	Cys2663Ser	7988G>C	C2663S	Marfan syndrome	Uyeda (2004) <i>J Hum Genet</i> 49:404	PubMed 15221638
FBN1	CM020697	GGC-GAC	Gly2668Asp	8003G>A	G2668D	Marfan syndrome	Matyas (2002) <i>Hum Mutat</i> 19:443	PubMed 11933199
FBN1	CM013943	GGC-TGC	Gly2668Cys	8002G>T	G2668C	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM098733	GGT-GTT	Gly2669Val	8006G>T	G2669V	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FBN1	CM930248	CGC-TGC	Arg2680Cys	8038C>T	R2680C	Marfan syndrome	Grossfield (1993) <i>Am J Hum Genet</i> 53S:A1167	Source: Meeting abstract
FBN1	CM077257	TGT-CGT	Cys2686Arg	8056T>C	C2686R	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM972823	TGT-TTT	Cys2686Phe	8057G>T	C2686F	Marfan syndrome	Liu (1997) <i>Genet Test</i> 1:237	PubMed 10464652
FBN1	CM145809	GGC-AGC	Gly2691Ser	8071G>A	G2691S	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM096465	CGA-CAA	Arg2694Gln	8081G>A	R2694Q	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM040045	CGA-TGA	Arg2694Term	8080C>T	R2694*	Marfan syndrome	Biggin (2004) <i>Hum Mutat</i> 23:99	PubMed 14695540
FBN1	CM093618	AAT-AGT	Asn2708Ser	8123A>G	N2708S	Marfan syndrome	Chung (2009) <i>Am J Med Genet A</i> 149A:1452	PubMed 19533785
FBN1	CM1314090	TCA-TGA	Ser2709Term	8126C>G	S2709*	Marfan syndrome	Pees (2013) <i>Clin Genet</i> epub:epub	PubMed 24199744
FBN1	CM013944	GAG-TAG	Glu2717Term	8149G>T	E2717*	Marfan syndrome	Loeys (2001) <i>Arch Intern Med</i> 161:2447	PubMed 11700157
FBN1	CM950453	CGG-TGG	Arg2726Trp	8176C>T	R2726W	Marfan syndrome	Milewicz (1995) <i>J Clin Invest</i> 95:2373	PubMed 7738200
FBN1	CM145811	AAT-AGT	Asn2740Ser	8219A>G	N2740S	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM077252	GAG-TAG	Glu2742Term	8224G>T	E2742*	Marfan syndrome	Howarth (2007) <i>Genet Test</i> 11:146	PubMed 17627385
FBN1	CM145812	TGG-TAG	Trp2756Term	8267G>A	W2756*	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577

FBN1	CM920270	TGG-TGA	Trp2756Term	8268G>A	W2756*	Marfan syndrome	Kainulainen (1992) <i>Proc Natl Acad Sci U S A</i> 89:5917	PubMed 1631074
FBN1	CM085408	AAG-TAG	Lys2774Term	8320A>T	K2774*	Marfan syndrome	Attanasio (2008) <i>Clin Genet</i> 74:39	PubMed 18435798
FBN1	CM940772	CGA-TGA	Arg2776Term	8326C>T	R2776*	Marfan syndrome	Hayward (1994) <i>Hum Mutat</i> 3:159	PubMed 7911051
FBN1	CM021599	CTC-CCC	Leu2780Pro	8339T>C	L2780P	Marfan syndrome	Halliday (2002) <i>J Med Genet</i> 39:589	PubMed 12161601
FBN1	CM067399	TAC-TGC	Tyr2793Cys	8378A>G	Y2793C	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM117734	TAC-CAC	Tyr2793His	8377T>C	Y2793H	Marfan syndrome	Baetens (2011) <i>Hum Mutat</i> 32:1053	PubMed 21542060
FBN1	CM054707	TAC-TAG	Tyr2793Term	8379C>G	Y2793*	Marfan syndrome	Arbustini (2005) <i>Hum Mutat</i> 26:494	PubMed 16222657
FBN1	CM067391	CAA-TAA	Gln2808Term	8422C>T	Q2808*	Marfan syndrome	Tjeldhorn (2006) <i>Genet Test</i> 10:258	PubMed 17253931
FBN1	CM145813	TCA-TGA	Ser2828Term	8483C>G	S2828*	Marfan syndrome	Lerner-Ellis (2014) <i>Mol Genet Metab</i> 112:171	PubMed 24793577
FBN1	CM096466	AGT-GGT	Ser2832Gly	8494A>G	S2832G	Marfan syndrome	Hung (2009) <i>Ann Hum Genet</i> 73:559	PubMed 19839986
FBN1	CM118006	GAA-TAA	Glu2841Term	8521G>T	E2841*	Marfan syndrome	Ogawa (2011) <i>Am J Cardiol</i> 108:1801	PubMed 21907952
FBN1	CM107867	TAT-TAG	Tyr2849Term	8547T>G	Y2849*	Marfan syndrome	Gao (2010) <i>Chin Med J (Engl)</i> 123:2874	PubMed 21034599
FBN1	CM098621	CAG-TAG	Gln2867Term	8599C>T	Q2867*	Marfan syndrome	Stheneur (2009) <i>Eur J Hum Genet</i> 17:1121	PubMed 19293843
FGFR2	CM950458	TCG-TGG	Ser252Trp	755C>G	S252W	Apert syndrome	Wilkie (1995) <i>Nat Genet</i> 9:165	PubMed 7719344
FGFR2	CM950459	CCT-CGT	Pro253Arg	758C>G	P253R	Apert syndrome	Wilkie (1995) <i>Nat Genet</i> 9:165	PubMed 7719344
FGFR2	CM960644	TAT-TGT	Tyr105Cys	314A>G	Y105C	Craniosynostosis	Pulleyn (1996) <i>Eur J Hum Genet</i> 4:283	PubMed 8946174
FGFR2	CM1313529	CCT-CTT	Pro253Leu	758C>T	P253L	Craniosynostosis	Roscioli (2013) <i>Am J Med Genet C Semin Med Genet</i> 163:259	PubMed 24127277
FGFR2	CM970528	TGG-TGT	Trp290Cys	870G>T	W290C	Craniosynostosis	Wilkie (1997) <i>Hum Mol Genet</i> 6:1647	PubMed 9300656
FGFR2	CM980752	GCC-TCC	Ala314Ser	940G>T	A314S	Craniosynostosis	Steinberger (1998) <i>Hum Genet</i> 102:145	PubMed 9521581
FGFR2	CM960646	GGG-GAG	Gly338Glu	1013G>A	G338E	Craniosynostosis	Pulleyn (1996) <i>Eur J Hum Genet</i> 4:283	PubMed 8946174
FGFR2	CM960650	TCC-TGC	Ser351Cys	1052C>G	S351C	Craniosynostosis	Pulleyn (1996) <i>Eur J Hum Genet</i> 4:283	PubMed 8946174
FGFR2	CM960654	GGG-AGG	Gly384Arg	1150G>A	G384R	Craniosynostosis	Pulleyn (1996) <i>Eur J Hum Genet</i> 4:283	PubMed 8946174
FGFR2	CM063988	AAT-ACT	Asn549Thr	1646A>C	N549T	Craniosynostosis	Wilkie (2006) <i>Am J Med Genet A</i> 140A:2631	PubMed 16838304
FGFR2	CM020143	AAG-AAT	Lys659Asn	1977G>T	K659N	Craniosynostosis	Kan (2002) <i>Am J Hum Genet</i> 70:472	PubMed 11781872
FGFR2	CM118255	GAA-AAA	Glu731Lys	2191G>A	E731K	Craniosynostosis	Park (2012) <i>J Cell Biochem</i> 113:457	PubMed 21928350
FGFR2	CM002016	GCC-TCC	Ala315Ser	943G>T	A315S	predisposition to	Johnson (2000) <i>Eur J Hum Genet</i> 8:571	PubMed 10951518
FGFR2	CM119685	TGC-CGC	Cys62Arg	184T>C	C62R	Crouzon syndrome	Sharma (2012) <i>Cleft Palate Craniofac J</i> 49:373	PubMed 22117175
FGFR2	CM970526	TCG-TTG	Ser252Leu	755C>T	S252L	Crouzon syndrome	Oldridge (1997) <i>Hum Mol Genet</i> 6:137	PubMed 9002682
FGFR2	CM036118	CAC-TAC	His254Tyr	760C>T	H254Y	Crouzon syndrome	Chun (2003) <i>Am J Med Genet A</i> 120A:470	PubMed 12884424
FGFR2	CM003312	CCG-CTG	Pro263Leu	788C>T	P263L	Crouzon syndrome	Kress (2000) <i>Cytogenet Cell Genet</i> 91:134	PubMed 11173845
FGFR2	CM1313530	GCC-CCC	Ala266Pro	796G>C	A266P	Crouzon syndrome	Roscioli (2013) <i>Am J Med Genet C Semin Med Genet</i> 163:259	PubMed 24127277
FGFR2	CM060998	TCC-TTC	Ser267Phe	800C>T	S267F	Crouzon syndrome	Lajeunie (2006) <i>Eur J Hum Genet</i> 14:289	PubMed 16418739
FGFR2	CM950460	TCC-CCC	Ser267Pro	799T>C	S267P	Crouzon syndrome	Oldridge (1995) <i>Hum Mol Genet</i> 4:1077	PubMed 7655462
FGFR2	CM980750	TTT-GTT	Phe276Val	826T>G	F276V	Crouzon syndrome	Steinberger (1998) <i>Hum Genet</i> 102:145	PubMed 9521581
FGFR2	CM950461	TGC-TTC	Cys278Phe	833G>T	C278F	Crouzon syndrome	Oldridge (1995) <i>Hum Mol Genet</i> 4:1077	PubMed 7655462
FGFR2	CM003313	TGC-TAC	Cys278Tyr	833G>A	C278Y	Crouzon syndrome	Kress (2000) <i>Cytogenet Cell Genet</i> 91:134	PubMed 11173845
FGFR2	CM013715	TAC-TGC	Tyr281Cys	842A>G	Y281C	Crouzon syndrome	Tsai (2001) <i>Pediatr Int</i> 43:263	PubMed 11380921
FGFR2	CM1313532	AGT-TGT	Ser282Cys	844A>T	S282C	Crouzon syndrome	Roscioli (2013) <i>Am J Med Genet C Semin Med Genet</i> 163:259	PubMed 24127277
FGFR2	CM060994	ATC-AAC	Ile288Asn	863T>A	I288N	Crouzon syndrome	Lajeunie (2006) <i>Eur J Hum Genet</i> 14:289	PubMed 16418739
FGFR2	CM003314	ATC-AGC	Ile288Ser	863T>G	I288S	Crouzon syndrome	Kress (2000) <i>Cytogenet Cell Genet</i> 91:134	PubMed 11173845

FGFR2	CM950462	CAG-CCG	Gln289Pro	866A>C	Q289P	Crouzon syndrome	Oldridge (1995) <i>Hum Mol Genet</i> 4:1077	PubMed 7655462
FGFR2	CM950463	TGG-CGG	Trp290Arg	868T>C	W290R	Crouzon syndrome	Oldridge (1995) <i>Hum Mol Genet</i> 4:1077	PubMed 7655462
FGFR2	CM950464	TGG-GGG	Trp290Gly	868T>G	W290G	Crouzon syndrome	Park (1995) <i>Hum Mol Genet</i> 4:1229 Roscioli (2013) <i>Am J Med Genet C Semin Med Genet</i> 163:259	PubMed 8528214 PubMed 24127277
FGFR2	CM1313533	TGG-TCG	Trp290Ser	869G>C	W290S	Crouzon syndrome	Steinberger (1997) <i>J Med Genet</i> 34:420	PubMed 9152842
FGFR2	CM970529	AAG-GAG	Lys292Glu	874A>G	K292E	Crouzon syndrome	Steinberger (1998) <i>Hum Genet</i> 102:145	PubMed 9521581
FGFR2	CM980751	TAC-TGC	Tyr301Cys	902A>G	Y301C	Crouzon syndrome	Lajeunie (2006) <i>Eur J Hum Genet</i> 14:289	PubMed 16418739
FGFR2	CM060996	TAC-TGC	Tyr308Cys	923A>G	Y308C	Crouzon syndrome	Jabs (1994) <i>Nat Genet</i> 8:275	PubMed 7874170
FGFR2	CM940777	TAT-TGT	Tyr328Cys	983A>G	Y328C	Crouzon syndrome	Steinberger (1996) <i>Hum Mutat</i> 8:386	PubMed 8956050
FGFR2	CM960645	AAT-ATT	Asn331Ile	992A>T	N331I	Crouzon syndrome	Stenirri (2007) <i>Clin Chem</i> 53:1767	PubMed 17693524
FGFR2	CM076185	GAC-GGC	Asp336Gly	1007A>G	D336G	Crouzon syndrome	Passos-Bueno (1998) <i>Am J Med Genet</i> 78:237	PubMed 9677057
FGFR2	CM980753	GCT-CCT	Ala337Pro	1009G>C	A337P	Crouzon syndrome	Gorry (1995) <i>Hum Mol Genet</i> 4:1387	PubMed 7581378
FGFR2	CM950466	GGG-CGG	Gly338Arg	1012G>C	G338R	Crouzon syndrome	Roscioli (2013) <i>Am J Med Genet C Semin Med Genet</i> 163:259	PubMed 24127277
FGFR2	CM1313534	TAT-AAT	Tyr340Asn	1018T>A	Y340N	Crouzon syndrome	Reardon (1994) <i>Nat Genet</i> 8:98	PubMed 7987400
FGFR2	CM940778	TAT-CAT	Tyr340His	1018T>C	Y340H	Crouzon syndrome	Lajeunie (2006) <i>Eur J Hum Genet</i> 14:289	PubMed 16418739
FGFR2	CM060997	TAT-TCT	Tyr340Ser	1019A>C	Y340S	Crouzon syndrome	Reardon (1994) <i>Nat Genet</i> 8:98	PubMed 7987400
FGFR2	CM940781	TGC-CGC	Cys342Arg	1024T>C	C342R	Crouzon syndrome	Reardon (1994) <i>Nat Genet</i> 8:98	PubMed 7987400
FGFR2	CM940780	TGC-AGC	Cys342Ser	1024T>A	C342S	Crouzon syndrome	Park (1995) <i>Hum Mol Genet</i> 4:1229	PubMed 8528214
FGFR2	CM950468	TGC-TGG	Cys342Trp	1026C>G	C342W	Crouzon syndrome	Reardon (1994) <i>Nat Genet</i> 8:98	PubMed 7987400
FGFR2	CM940779	TGC-TAC	Cys342Tyr	1025G>A	C342Y	Crouzon syndrome	Jabs (1994) <i>Nat Genet</i> 8:275	PubMed 7874170
FGFR2	CM940783	TCT-TGT	Ser347Cys	1040C>G	S347C	Crouzon syndrome	Reardon (1994) <i>Nat Genet</i> 8:98	PubMed 7987400
FGFR2	CM940784	TCT-TGT	Ser354Cys	1061C>G	S354C	Crouzon syndrome	Chun (2003) <i>Am J Med Genet A</i> 120A:470	PubMed 12884424
FGFR2	CM060999	TCT-TTT	Ser354Phe	1061C>T	S354F	Crouzon syndrome	Kress (2000) <i>Cytogenet Cell Genet</i> 91:134	PubMed 11173845
FGFR2	CM003315	TCT-TAT	Ser354Tyr	1061C>A	S354Y	Crouzon syndrome	Lajeunie (2006) <i>Eur J Hum Genet</i> 14:289	PubMed 16418739
FGFR2	CM061000	TTG-TCG	Leu357Ser	1070T>C	L357S	Crouzon syndrome	Everett (1999) <i>Cleft Palate Craniofac J</i> 36:533	PubMed 10574673
FGFR2	CM994121	GCG-TCG	Ala362Ser	1084G>T	A362S	Crouzon syndrome	Roscioli (2013) <i>Am J Med Genet C Semin Med Genet</i> 163:259	PubMed 24127277
FGFR2	CM1313536	AAG-GAG	Lys367Glu	1099A>G	K367E	Crouzon syndrome	Collet (2014) <i>Clin Genet</i> 85:598	PubMed 23808569
FGFR2	CM144764	TAC-AAC	Tyr381Asn	1141T>A	Y381N	Crouzon syndrome	de Ravel (2005) <i>Eur J Hum Genet</i> 13:503	PubMed 15523492
FGFR2	CM050617	AAA-GAA	Lys526Glu	1576A>G	K526E	Crouzon syndrome	Kan (2002) <i>Am J Hum Genet</i> 70:472	PubMed 11781872
FGFR2	CM020140	AAT-CAT	Asn549His	1645A>C	N549H	Crouzon syndrome	Suh (2013) <i>J Cell Biochem</i> epub:epub	PubMed 23913723
FGFR2	CM137634	TTG-TTC	Leu617Phe	1851G>C	L617F	Crouzon syndrome	Kan (2002) <i>Am J Hum Genet</i> 70:472	PubMed 11781872
FGFR2	CM020145	AGA-GGA	Arg678Gly	2032A>G	R678G	Crouzon syndrome	Zhang (2007) <i>Chin Med J (Engl)</i> 120:1017	PubMed 17624273
FGFR3	CM073067	AGC-TGC	Ser217Cys	649A>T	S217C	Achondroplasia	Heuertz (2006) <i>Eur J Hum Genet</i> 14:1240	PubMed 16912704
FGFR3	CM066079	AGT-TGT	Ser279Cys	835A>T	S279C	Achondroplasia	Chen (2001) <i>Prenat Diagn</i> 21:89	PubMed 11241532
FGFR3	CM010888	GGG-GAG	Gly346Glu	1037G>A	G346E	Achondroplasia	Ikegawa (1995) <i>Hum Genet</i> 96:309	PubMed 7649548
FGFR3	CM950472	GGC-TGC	Gly375Cys	1123G>T	G375C	Achondroplasia	Shiang (1994) <i>Cell</i> 78:335	PubMed 7913883
FGFR3	CM940785	GGG-AGG	Gly380Arg	1138G>A	G380R	Achondroplasia	Shiang (1994) <i>Cell</i> 78:335	PubMed 7913883
FGFR3	CM940786	GGG-CGG	Gly380Arg	1138G>C	G380R	Achondroplasia	Zhu (2003) <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i> 20:373	PubMed 14556186
FGFR3	CM034415	ACG-TCG	Thr394Ser	1180A>T	T394S	Achondroplasia	Bellus (1996) <i>Nat Genet</i> 14:174	PubMed 8841188
FGFR3	CM960655	CCG-CGG	Pro250Arg	749C>G	P250R	Craniosynostosis	Schindler (2002) <i>J Med Genet</i> 39:764	PubMed 12362036
FGFR3	CM022207	CCG-CTG	Pro250Leu	749C>T	P250L	Craniosynostosis		

FGFR3	CM1313538	CCC-ACC	Pro283Thr	847C>A	P283T	Craniosynostosis	Roscioli (2013) <i>Am J Med Genet C Semin Med Genet</i> 163:259	PubMed 24127277
FGFR3	CM119750	GCC-ACC	Ala334Thr	1000G>A	A334T	Craniosynostosis	Barroso (2011) <i>Am J Med Genet A</i> 155:3050	PubMed 22038757
FGFR3	CM950473	GCG-GAG	Ala391Glu	1172C>A	A391E	Crouzon syndrome	Meyers (1995) <i>Nat Genet</i> 11:462	PubMed 7493034
FGFR3	CM066084	TCG-TTG	Ser84Leu	251C>T	S84L	Hypochondroplasia	Heuertz (2006) <i>Eur J Hum Genet</i> 14:1240	PubMed 16912704
FGFR3	CM128685	CAG-CTG	Gln115Leu	344A>T	Q115L	Hypochondroplasia	Song (2012) <i>Am J Med Genet A</i> 158A:2456	PubMed 22903874
FGFR3	CM066080	CGC-TGC	Arg200Cys	598C>T	R200C	Hypochondroplasia	Heuertz (2006) <i>Eur J Hum Genet</i> 14:1240	PubMed 16912704
FGFR3	CM066078	AAC-CAC	Asn262His	784A>C	N262H	Hypochondroplasia	Heuertz (2006) <i>Eur J Hum Genet</i> 14:1240	PubMed 16912704
FGFR3	CM128684	ACG-ATG	Thr264Met	791C>T	T264M	Hypochondroplasia	Song (2012) <i>Am J Med Genet A</i> 158A:2456	PubMed 22903874
FGFR3	CM066081	GGC-TGC	Gly268Cys	802G>T	G268C	Hypochondroplasia	Heuertz (2006) <i>Eur J Hum Genet</i> 14:1240	PubMed 16912704
FGFR3	CM066083	TAC-TGC	Tyr278Cys	833A>G	Y278C	Hypochondroplasia	Heuertz (2006) <i>Eur J Hum Genet</i> 14:1240	PubMed 16912704
FGFR3	CM084803	GGC-TGC	Gly295Cys	883G>T	G295C	Hypochondroplasia	Baujat (2008) <i>Best Pract Res Clin Rheumatol</i> 22:3	PubMed 18328977
FGFR3	CM121343	CTC-GTC	Leu324Val	970C>G	L324V	Hypochondroplasia	Saito (2012) <i>Am J Med Genet A</i> 158A:630	PubMed 22302603
FGFR3	CM000015	AAC-ATC	Asn328Ile	983A>T	N328I	Hypochondroplasia	Winterpacht (2000) <i>Physiol Genomics</i> 2:9	PubMed 11015576
FGFR3	CM135371	GGC-TGC	Gly342Cys	1024G>T	G342C	Hypochondroplasia	Wang (2013) <i>Clin Chim Acta</i> 423:62	PubMed 23726269
FGFR3	CM084804	TCT-TGT	Ser351Cys	1052C>G	S351C	Hypochondroplasia	Baujat (2008) <i>Best Pract Res Clin Rheumatol</i> 22:3	PubMed 18328977
FGFR3	CM084805	GAG-AAG	Glu360Lys	1078G>A	E360K	Hypochondroplasia	Baujat (2008) <i>Best Pract Res Clin Rheumatol</i> 22:3	PubMed 18328977
FGFR3	CM066082	GTG-GAG	Val381Glu	1142T>A	V381E	Hypochondroplasia	Heuertz (2006) <i>Eur J Hum Genet</i> 14:1240	PubMed 16912704
FGFR3	CM084808	CAG-CGG	Gln485Arg	1454A>G	Q485R	Hypochondroplasia	Baujat (2008) <i>Best Pract Res Clin Rheumatol</i> 22:3	PubMed 18328977
FGFR3	CM970530	ATC-GTC	Ile538Val	1612A>G	I538V	Hypochondroplasia	Grigelioniene (1997) <i>Hum Mutat</i> 11:333	PubMed 10215410
FGFR3	CM950474	AAC-AAA	Asn540Lys	1620C>A	N540K	Hypochondroplasia	Bellus (1995) <i>Nat Genet</i> 10:357	PubMed 7670477
FGFR3	CM950475	AAC-AAG	Asn540Lys	1620C>G	N540K	Hypochondroplasia	Prinos (1995) <i>Hum Mol Genet</i> 4:2097	PubMed 8589686
FGFR3	CM000382	AAC-AGC	Asn540Ser	1619A>G	N540S	Hypochondroplasia	Mortier (2000) <i>J Med Genet</i> 37:220	PubMed 10777366
FGFR3	CM980754	AAC-ACC	Asn540Thr	1619A>C	N540T	Hypochondroplasia	Deutz-Terlouw (1998) <i>Hum Mutat</i> S1:S62	PubMed 9452043
FGFR3	CM002965	AAG-AAC	Lys650Asn	1950G>C	K650N	Hypochondroplasia	Bellus (2000) <i>Am J Hum Genet</i> 67:1411	PubMed 11055896
FGFR3	CM002966	AAG-AAT	Lys650Asn	1950G>T	K650N	Hypochondroplasia	Bellus (2000) <i>Am J Hum Genet</i> 67:1411	PubMed 11055896
FGFR3	CM002967	AAG-CAG	Lys650Gln	1948A>C	K650Q	Hypochondroplasia	Bellus (2000) <i>Am J Hum Genet</i> 67:1411	PubMed 11055896
HRAS	CM053284	GGC-GCC	Gly12Ala	35G>C	G12A	Costello syndrome	Aoki (2005) <i>Nat Genet</i> 37:1038	PubMed 16170316
HRAS	CM081305	GGC-GAC	Gly12Asp	35G>A	G12D	Costello syndrome	Lo (2008) <i>J Med Genet</i> 45:167	PubMed 18039947
HRAS	CM061797	GGC-TGC	Gly12Cys	34G>T	G12C	Costello syndrome	Hoornaert (2006) <i>J Med Genet</i> 43:406	PubMed 16155195
HRAS	CM053283	GGC-AGC	Gly12Ser	34G>A	G12S	Costello syndrome	Aoki (2005) <i>Nat Genet</i> 37:1038	PubMed 16170316
HRAS	CM123157	GGC-GTC	Gly12Val	35G>T	G12V	Costello syndrome	Burkitt-Wright (2012) <i>Am J Med Genet A</i> 158A:1102	PubMed 22495892
HRAS	CM053285	GGT-GAT	Gly13Asp	38G>A	G13D	Costello syndrome	Aoki (2005) <i>Nat Genet</i> 37:1038	PubMed 16170316
HRAS	CM060018	GGT-TGT	Gly13Cys	37G>T	G13C	Costello syndrome	Estep (2006) <i>Am J Med Genet A</i> 140A:8	PubMed 16372351
HRAS	CM081303	ACC-ATC	Thr58Ile	173C>T	T58I	Costello syndrome	Gripp (2008) <i>Am J Med Genet A</i> 146A:683	PubMed 18247425
HRAS	CM127689	TCT-TGT	Ser89Cys	266C>G	S89C	Costello syndrome	Gripp (2012) <i>Am J Med Genet A</i> 158A:2106	PubMed 22821884
HRAS	CM061796	AAG-AGG	Lys117Arg	350A>G	K117R	Costello syndrome	Hoornaert (2006) <i>J Med Genet</i> 43:406	PubMed 16155195
HRAS	CM070951	GCC-ACC	Ala146Thr	436G>A	A146T	Costello syndrome	Zampino (2007) <i>Hum Mutat</i> 28:265	PubMed 17054105
HRAS	CM081304	GCC-GTC	Ala146Val	437C>T	A146V	Costello syndrome	Gripp (2008) <i>Am J Med Genet A</i> 146A:683	PubMed 18247425
KRAS	CM076251	GGT-AGT	Gly12Ser	34G>A	G12S	syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
KRAS	CM070964	CAG-GAG	Gln22Glu	64C>G	Q22E	syndrome	Zenker (2007) <i>J Med Genet</i> 44:131	PubMed 17056636
KRAS	CM061081	CCA-CGA	Pro34Arg	101C>G	P34R	syndrome	Schubbert (2006) <i>Nat Genet</i> 38:331	PubMed 16474405
KRAS	CM061080	GGT-CGT	Gly60Arg	178G>C	G60R	syndrome	Niihori (2006) <i>Nat Genet</i> 38:294	PubMed 16474404

KRAS	CM1111357	TAC-CAC	Tyr71His	211T>C	Y71H	syndrome	Stark (2012) <i>Clin Genet</i> 81:590	PubMed 21797849
KRAS	CM1111358	AAG-GAG	Lys147Glu	439A>G	K147E	syndrome	Stark (2012) <i>Clin Genet</i> 81:590	PubMed 21797849
KRAS	CM070963	AAA-AAT	Lys5Asn	15A>T	K5N	Costello syndrome	Zenker (2007) <i>J Med Genet</i> 44:131	PubMed 17056636
KRAS	CM073168	AAA-GAA	Lys5Glu	13A>G	K5E	Costello syndrome	Bertola (2007) <i>J Hum Genet</i> 52:521	PubMed 17468812
KRAS	CM061082	GTA-ATA	Val14Ile	40G>A	V14I	Noonan syndrome	Schubbert (2006) <i>Nat Genet</i> 38:331	PubMed 16474405
KRAS	CM070966	CAG-CGG	Gln22Arg	65A>G	Q22R	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:131	PubMed 17056636
KRAS	CM106704	AAT-ATT	Asn26Ile	77A>T	N26I	Noonan syndrome	Leventopoulos (2010) <i>Clin Exp Rheumatol</i> 28:556	PubMed 20810036
KRAS	CM070967	CCA-CAA	Pro34Gln	101C>A	P34Q	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:131	PubMed 17056636
KRAS	CM070968	CCA-CTA	Pro34Leu	101C>T	P34L	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:131	PubMed 17056636
KRAS	CM070965	ATA-ATG	Ile36Met	108A>G	I36M	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:131	PubMed 17056636
KRAS	CM061079	ACA-ATA	Thr58Ile	173C>T	T58I	Noonan syndrome	Schubbert (2006) <i>Nat Genet</i> 38:331	PubMed 16474405
KRAS	CM092327	GGT-AGT	Gly60Ser	178G>A	G60S	Noonan syndrome	Kratz (2009) <i>Am J Med Genet A</i> 149A:1036	PubMed 19396835
KRAS	CM1313466	GGT-GTT	Gly60Val	179G>T	G60V	Noonan syndrome	Nosan (2013) <i>Croat Med J</i> 54:574	PubMed 24382853
KRAS	CM123160	ATG-CTG	Met72Leu	214A>C	M72L	Noonan syndrome	Brasil (2012) <i>Am J Med Genet A</i> 158A:1178	PubMed 22488932
KRAS	CM121363	AAT-AGT	Asn116Ser	347A>G	N116S	Noonan syndrome	Razzaque (2012) <i>Am J Med Genet A</i> 158A:524	PubMed 22302539
KRAS	CM061830	GTT-GGT	Val152Gly	455T>G	V152G	Noonan syndrome	Carta (2006) <i>Am J Hum Genet</i> 79:129	PubMed 16773572
KRAS	CM061083	GAT-GTT	Asp153Val	458A>T	D153V	Noonan syndrome	Schubbert (2006) <i>Nat Genet</i> 38:331	PubMed 16474405
KRAS	CM070969	TTC-ATC	Phe156Ile	466T>A	F156I	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:131	PubMed 17056636
KRAS	CM070970	TTC-TTG	Phe156Leu	468C>G	F156L	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:131	PubMed 17056636
LEPRE1	CM086850	TCG-TAG	Ser131Term	392C>A	S131*	Osteogenesis imperfecta, autosomal recessive	Baldrige (2008) <i>Hum Mutat</i> 29:1435	PubMed 18566967
LEPRE1	CM091982	CGA-TGA	Arg210Term	628C>T	R210*	Osteogenesis imperfecta, autosomal recessive	Willaert (2009) <i>J Med Genet</i> 46:233	PubMed 19088120
LEPRE1	CM112374	CGA-TGA	Arg214Term	640C>T	R214*	Osteogenesis imperfecta, autosomal recessive	Bell (2011) <i>Sci Transl Med</i> 3:65ra4	PubMed 21228398
LEPRE1	CM086848	TAC-TAA	Tyr255Term	765C>A	Y255*	Osteogenesis imperfecta, autosomal recessive	Baldrige (2008) <i>Hum Mutat</i> 29:1435	PubMed 18566967
LEPRE1	CM086849	TAC-TAG	Tyr311Term	933C>G	Y311*	Osteogenesis imperfecta, autosomal recessive	Baldrige (2008) <i>Hum Mutat</i> 29:1435	PubMed 18566967
LEPRE1	CM091983	CGA-TGA	Arg368Term	1102C>T	R368*	Osteogenesis imperfecta, autosomal recessive	Willaert (2009) <i>J Med Genet</i> 46:233	PubMed 19088120
LEPRE1	CM123316	CTG-CCG	Leu489Pro	1466T>C	L489P	Osteogenesis imperfecta, autosomal recessive	Zhang (2012) <i>J Bone Miner Metab</i> 30:69	PubMed 21667357
LEPRE1	CM070981	TAC-TAA	Tyr552Term	1656C>A	Y552*	Osteogenesis imperfecta, autosomal recessive	Cabral (2007) <i>Nat Genet</i> 39:359	PubMed 17277775
LEPRE1	CM086851	CGA-TGA	Arg681Term	2041C>T	R681*	Osteogenesis imperfecta, autosomal recessive	Baldrige (2008) <i>Hum Mutat</i> 29:1435	PubMed 18566967
MAP2K1	CM092997	CTT-TTT	Leu42Phe	124C>T	L42F	syndrome	Dentici (2009) <i>Eur J Hum Genet</i> 17:733	PubMed 19156172
MAP2K1	CM061105	TTT-TCT	Phe53Ser	158T>C	F53S	syndrome Cardio-facio-cutaneous	Rodriguez-Viciano (2006) <i>Science</i> 311:1287	PubMed 16439621
MAP2K1	CM147586	GTG-GGG	Val60Gly	179T>G	V60G	syndrome	Wong Ramsey (2014) <i>Am J Med Genet A</i> 164:2036	PubMed 24719372
MAP2K1	CM076269	GAC-AAC	Asp67Asn	199G>A	D67N	syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
MAP2K1	CM073191	CCG-CAG	Pro124Gln	371C>A	P124Q	syndrome	Gripp (2007) <i>Am J Med Genet A</i> 143A:1472	PubMed 17551924
MAP2K1	CM071852	CCG-CTG	Pro124Leu	371C>T	P124L	syndrome	Narumi (2007) <i>Am J Med Genet A</i> 143A:799	PubMed 17366577
MAP2K1	CM080433	GGC-GTC	Gly128Val	383G>T	G128V	syndrome	Schulz (2008) <i>Clin Genet</i> 73:62	PubMed 18042262

						Cardio-facio-cutaneous		
MAP2K1	CM086852	TAT-AAT	Tyr130Asn	388T>A	Y130N	syndrome	Rodriguez-Viciana (2008) <i>Methods Enzymol</i> 438:277	PubMed 18413255
MAP2K1	CM061104	TAT-TGT	Tyr130Cys	389A>G	Y130C	syndrome	Rodriguez-Viciana (2006) <i>Science</i> 311:1287	PubMed 16439621
MAP2K1	CM092998	TAT-CAT	Tyr130His	388T>C	Y130H	syndrome	Dentici (2009) <i>Eur J Hum Genet</i> 17:733	PubMed 19156172
MAP2K1	CM083720	GAG-CAG	Glu203Gln	607G>C	E203Q	syndrome	Nystrom (2008) <i>J Med Genet</i> 45:500	PubMed 18456719
MAP2K1	CM076271	ACC-CCC	Thr55Pro	163A>C	T55P	Costello syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
MAP2K1	CM076270	GAG-GGG	Glu44Gly	131A>G	E44G	Noonan syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
MAP2K1	CM137741	GCA-ACA	Ala347Thr	1039G>A	A347T	Noonan syndrome	Croonen (2013) <i>Eur J Hum Genet</i> 21:936	PubMed 23321623
MAP2K2	CM061106	TTT-TGT	Phe57Cys	170T>G	F57C	syndrome	Rodriguez-Viciana (2006) <i>Science</i> 311:1287	PubMed 16439621
						Cardio-facio-cutaneous		
MAP2K2	CM086853	TTT-ATT	Phe57Ile	169T>A	F57I	syndrome	Rodriguez-Viciana (2008) <i>Methods Enzymol</i> 438:277	PubMed 18413255
MAP2K2	CM083721	TTT-TTA	Phe57Leu	171T>A	F57L	syndrome	Nystrom (2008) <i>J Med Genet</i> 45:500	PubMed 18456719
MAP2K2	CM080435	TTT-GTT	Phe57Val	169T>G	F57V	syndrome	Schulz (2008) <i>Clin Genet</i> 73:62	PubMed 18042262
MAP2K2	CM071855	AAA-GAA	Lys61Glu	181A>G	K61E	syndrome	Narumi (2007) <i>Am J Med Genet A</i> 143A:799	PubMed 17366577
MAP2K2	CM076272	AAA-ACA	Lys61Thr	182A>C	K61T	syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
MAP2K2	CM076273	GCC-CCC	Ala62Pro	184G>C	A62P	syndrome	Nava (2007) <i>J Med Genet</i> 44:763	PubMed 17704260
MAP2K2	CM125907	GCG-ACG	Ala80Thr	238G>A	A80T	hypertrophic	Kaski (2012) <i>Circ Cardiovasc Genet</i> 5:317	PubMed 22589294
MAP2K2	CM071854	CCG-CGG	Pro128Arg	383C>G	P128R	syndrome	Narumi (2007) <i>Am J Med Genet A</i> 143A:799	PubMed 17366577
MAP2K2	CM101871	CCG-CAG	Pro128Gln	383C>A	P128Q	syndrome	Rauen (2010) <i>Am J Med Genet A</i> 152A:807	PubMed 20358587
MAP2K2	CM112525	GGC-GAC	Gly132Asp	395G>A	G132D	syndrome	Linden (2011) <i>Clin Dysmorphol</i> 20:86	PubMed 21178588
MAP2K2	CM071856	GGC-GTC	Gly132Val	395G>T	G132V	syndrome	Narumi (2007) <i>Am J Med Genet A</i> 143A:799	PubMed 17366577
						Cardio-facio-cutaneous		
MAP2K2	CM086854	TAC-TGC	Tyr134Cys	401A>G	Y134C	syndrome	Rodriguez-Viciana (2008) <i>Methods Enzymol</i> 438:277	PubMed 18413255
MAP2K2	CM080434	TAC-CAC	Tyr134His	400T>C	Y134H	syndrome	Schulz (2008) <i>Clin Genet</i> 73:62	PubMed 18042262
MAP2K2	CM071853	AAA-AGA	Lys273Arg	818A>G	K273R	syndrome	Narumi (2007) <i>Am J Med Genet A</i> 143A:799	PubMed 17366577
MECP2	CM058034	ATG-TTG	Met1Leu	1A>T	M1L	Rett syndrome	Gauthier (2005) <i>Can J Neurol Sci</i> 32:321	PubMed 16225173
MECP2	CM032595	GAA-CAA	Glu10Gln	28G>C	E10Q	Rett syndrome	Smeets (2003) <i>Am J Med Genet A</i> 122A:227	PubMed 12966523
MECP2	CM060329	GAA-TAA	Glu10Term	28G>T	E10*	Rett syndrome	Philippe (2006) <i>Eur J Med Genet</i> 49:9	PubMed 16473305
MECP2	CM023409	CAG-TAG	Gln16Term	46C>T	Q16*	Rett syndrome	RSRF (2002) <i>MECP2 LSDB entry</i> Unpublished:	Source: LSDB Source: Online
MECP2	HM971529	CAG-TAG	Gln19Term	55C>T	Q19*	Rett syndrome	Cook (2000) <i>Hum Genet</i> 106:258	submission
MECP2	CM010332	AAG-TAG	Lys22Term	64A>T	K22*	Rett syndrome	Vacca (2001) <i>J Mol Med</i> 78:648	PubMed 11269512
MECP2	CM057720	CAG-TAG	Gln47Term	139C>T	Q47*	Rett syndrome	Charman (2005) <i>Eur J Hum Genet</i> 13:1121	PubMed 16077736
MECP2	CM010333	TCA-TAA	Ser49Term	146C>A	S49*	Rett syndrome	Bourdon (2001) <i>Hum Genet</i> 108:43	PubMed 11214906
MECP2	CM110917	CAC-CAG	His51Gln	153C>G	H51Q	Rett syndrome	Khajuria (2011) <i>Brain Dev</i> :	PubMed 21300488
MECP2	CM035705	TCA-TGA	Ser65Term	194C>G	S65*	Rett syndrome	Friez (2003) <i>MECP2 LSDB entry</i> Unpublished:1732	Source: LSDB
MECP2	CM055984	TCA-TGA	Ser68Term	203C>G	S68*	Rett syndrome	Fukuda (2005) <i>Brain Dev</i> 27:211	PubMed 15737703
MECP2	CM001222	GAC-GAA	Asp97Glu	291C>A	D97E	Rett syndrome	Xiang (2000) <i>J Med Genet</i> 37:250	PubMed 10745042
MECP2	CM014800	GAC-TAC	Asp97Tyr	289G>T	D97Y	Rett syndrome	Hoffbuhr (2001) <i>Neurology</i> 56:1486	PubMed 11402105
MECP2	CM041798	CTG-CGG	Leu100Arg	299T>G	L100R	Rett syndrome	Schanen (2004) <i>Am J Med Genet</i> 126A:129	PubMed 15057977
MECP2	CM022424	CTG-GTG	Leu100Val	298C>G	L100V	Rett syndrome	Chae (2002) <i>J Child Neurol</i> 17:33	PubMed 11913567
MECP2	CM002055	CCT-CGT	Pro101Arg	302C>G	P101R	Rett syndrome	Hampson (2000) <i>J Med Genet</i> 37:610	PubMed 10991689
MECP2	CM000743	CCT-CAT	Pro101His	302C>A	P101H	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337
MECP2	CM000744	CCT-CTT	Pro101Leu	302C>T	P101L	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337

MECP2	CM010334	CCT-TCT	Pro101Ser	301C>T	P101S	Rett syndrome	Vacca (2001) <i>J Mol Med</i> 78:648	PubMed 11269512
MECP2	CM000745	CCT-ACT	Pro101Thr	301C>A	P101T	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337
MECP2	CM023410	TGG-CGG	Trp104Arg	310T>C	W104R	Rett syndrome	RSRF (2002) <i>MECP2 LSDB entry</i> Unpublished: No author given (2007) <i>MECP2 LSDB entry</i> Unpublished:2367	Source: LSDB
MECP2	CM076290	TGG-TAG	Trp104Term	311G>A	W104*	Rett syndrome	Unpublished:2367	Source: LSDB
MECP2	CM001223	CGG-CAG	Arg106Gln	317G>A	R106Q	Rett syndrome	Bienvenu (2000) <i>Hum Mol Genet</i> 9:1377	PubMed 10814719
MECP2	CM023812	CGG-GGG	Arg106Gly	316C>G	R106G	Rett syndrome	Bienvenu (2002) <i>Genet Test</i> 6:1	PubMed 12180070
MECP2	CM992175	CGG-TGG	Arg106Trp	316C>T	R106W	Rett syndrome	Amir (1999) <i>Nat Genet</i> 23:185	PubMed 10508514
MECP2	CM071859	CTT-CAT	Leu108His	323T>A	L108H	Rett syndrome	Zahorakova (2007) <i>J Hum Genet</i> 52:342	PubMed 17387578
MECP2	CM105545	CAA-TAA	Gln110Term	328C>T	Q110*	Rett syndrome	Temudo (2011) <i>Brain Dev</i> 33:69	PubMed 20116947
MECP2	CM010335	AGG-GGG	Arg111Gly	331A>G	R111G	Rett syndrome	Laccone (2001) <i>Hum Mutat</i> 17:183	PubMed 11241840
MECP2	CM096400	AAA-TAA	Lys112Term	334A>T	K112*	Rett syndrome	Lima (2009) <i>Arq Neuropsiquiatr</i> 67:577	PubMed 19722030
MECP2	CM091239	TCT-CCT	Ser113Pro	337T>C	S113P	Rett syndrome	Temudo (2009) <i>Brain Dev</i> 31:46	PubMed 18572337
MECP2	CM085533	CGC-TGC	Arg115Cys	343C>T	R115C	Rett syndrome	Bunyan (2008) <i>Genet Test</i> 12:373	PubMed 18652533
MECP2	CM014369	TAT-GAT	Tyr120Asp	358T>G	Y120D	Rett syndrome	Inui (2001) <i>Brain Dev</i> 23:212	PubMed 11376998
MECP2	CM070195	GTG-ATG	Val122Met	364G>A	V122M	Rett syndrome	Li (2007) <i>J Hum Genet</i> 52:38	PubMed 17089071
MECP2	CM002056	TTG-TTC	Leu124Phe	372G>C	L124F	Rett syndrome	Obata (2000) <i>J Med Genet</i> 37:608	PubMed 10991688
MECP2	CM014897	AAT-AGT	Asn126Ser	377A>G	N126S	Rett syndrome	Milunsky (2001) <i>Genet Test</i> 5:321	PubMed 11960578 Source: Online
MECP2	HM060005	AAT-TAT	Asn126Tyr	376A>T	N126Y	Rett syndrome	Conforti (2006) <i>Hum Genet</i> 119:676	submission
MECP2	CM010918	CCC-CTC	Pro127Leu	380C>T	P127L	Rett syndrome	Auranen (2001) <i>Neurology</i> 56:611	PubMed 11245712
MECP2	CM032596	CAG-CCG	Gln128Pro	383A>C	Q128P	Rett syndrome	Smeets (2003) <i>Am J Med Genet A</i> 122A:227	PubMed 12966523
MECP2	CM023411	CAG-TAG	Gln128Term	382C>T	Q128*	Rett syndrome	Charman (2005) <i>Eur J Hum Genet</i> 13:1121	PubMed 16077736
MECP2	CM070194	GGA-GTA	Gly129Val	386G>T	G129V	Rett syndrome	Li (2007) <i>J Hum Genet</i> 52:38	PubMed 17089071
MECP2	CM055983	GCC-GAC	Ala131Asp	392C>A	A131D	Rett syndrome	Fukuda (2005) <i>Brain Dev</i> 27:211	PubMed 15737703
MECP2	CM992176	CGC-TGC	Arg133Cys	397C>T	R133C	Rett syndrome	Amir (1999) <i>Nat Genet</i> 23:185	PubMed 10508514
MECP2	CM023813	CGC-GGC	Arg133Gly	397C>G	R133G	Rett syndrome	Bienvenu (2002) <i>Genet Test</i> 6:1	PubMed 12180070
MECP2	CM003766	CGC-CAC	Arg133His	398G>A	R133H	Rett syndrome	Dragich (2000) <i>Hum Mol Genet</i> 9:2365	PubMed 11005791
MECP2	CM001224	CGC-CTC	Arg133Leu	398G>T	R133L	Rett syndrome	De Bona (2000) <i>Eur J Hum Genet</i> 8:325	PubMed 10854091
MECP2	CM014898	CGC-CCC	Arg133Pro	398G>C	R133P	Rett syndrome	Milunsky (2001) <i>Genet Test</i> 5:321	PubMed 11960578
MECP2	CM000746	TCT-TGT	Ser134Cys	401C>G	S134C	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337
MECP2	CM055985	TCT-TTT	Ser134Phe	401C>T	S134F	Rett syndrome	Fukuda (2005) <i>Brain Dev</i> 27:211	PubMed 15737703
MECP2	CM045298	TCT-CCT	Ser134Pro	400T>C	S134P	Rett syndrome	Chae (2004) <i>J Child Neurol</i> 19:503	PubMed 15526954
MECP2	CM010336	AAA-GAA	Lys135Glu	403A>G	K135E	Rett syndrome	Laccone (2001) <i>Hum Mutat</i> 17:183	PubMed 11241840
MECP2	CM060330	TTG-TAG	Leu138Term	413T>A	L138*	Rett syndrome	Philippe (2006) <i>Eur J Med Genet</i> 49:9	PubMed 16473305
MECP2	CM003325	GCG-GTG	Ala140Val	419C>T	A140V	Rett syndrome	Orrico (2000) <i>FEBS Lett</i> 481:285	PubMed 11007980
MECP2	CM001225	TAC-TAG	Tyr141Term	423C>G	Y141*	Rett syndrome	De Bona (2000) <i>Eur J Hum Genet</i> 8:325	PubMed 10854091
MECP2	CM003454	AAG-TAG	Lys144Term	430A>T	K144*	Rett syndrome	Buyse (2000) <i>Am J Hum Genet</i> 67:1428	PubMed 11055898
MECP2	CM066123	GAC-GAG	Asp151Glu	453C>G	D151E	Rett syndrome	Petel-Galil (2006) <i>J Med Genet</i> 43:e56	PubMed 17142618
MECP2	CM023814	GAC-GGC	Asp151Gly	452A>G	D151G	Rett syndrome	Bienvenu (2002) <i>Genet Test</i> 6:1	PubMed 12180070
MECP2	CM000747	CCT-CGT	Pro152Arg	455C>G	P152R	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337
MECP2	CM023412	TTT-TGT	Phe155Cys	464T>G	F155C	Rett syndrome	BH (2002) <i>MECP2 LSDB entry</i> Unpublished:	Source: LSDB
MECP2	CM001226	TTT-ATT	Phe155Ile	463T>A	F155I	Rett syndrome	Xiang (2000) <i>J Med Genet</i> 37:250	PubMed 10745042
MECP2	CM992177	TTT-TCT	Phe155Ser	464T>C	F155S	Rett syndrome	Amir (1999) <i>Nat Genet</i> 23:185	PubMed 10508514

MECP2	CM055982	GAC-GCC	Asp156Ala	467A>C	D156A	Rett syndrome	Fukuda (2005) <i>Brain Dev</i> 27:211	PubMed 15737703
MECP2	CM001227	GAC-GAA	Asp156Glu	468C>A	D156E	Rett syndrome	Amir (2000) <i>Ann Neurol</i> 47:670	PubMed 10805343
MECP2	CM011798	GAC-GGC	Asp156Gly	467A>G	D156G	Rett syndrome	Trappe (2001) <i>Am J Hum Genet</i> 68:1093	PubMed 11309679
MECP2	CM078104	GAC-CAC	Asp156His	466G>C	D156H	Rett syndrome	Li (2007) <i>Zhonghua Er Ke Za Zhi</i> 45:579	PubMed 18021529
MECP2	CM058035	TTC-CTC	Phe157Leu	469T>C	F157L	Rett syndrome	Gauthier (2005) <i>Can J Neurol Sci</i> 32:321	PubMed 16225173
MECP2	CM078121	TTC-TTA	Phe157Leu	471C>A	F157L	Rett syndrome	Li (2007) <i>Zhonghua Er Ke Za Zhi</i> 45:579	PubMed 18021529
MECP2	CM003455	TTC-TTG	Phe157Leu	471C>G	F157L	Rett syndrome	Amir (2000) <i>Am J Med Genet</i> 97:147	PubMed 11180222
MECP2	CM010337	ACG-GCG	Thr158Ala	472A>G	T158A	Rett syndrome	Vacca (2001) <i>J Mol Med</i> 78:648	PubMed 11269512
MECP2	CM992178	ACG-ATG	Thr158Met	473C>T	T158M	Rett syndrome	Amir (1999) <i>Nat Genet</i> 23:185	PubMed 10508514
MECP2	CM023413	GGG-AGG	Gly161Arg	481G>A	G161R	Rett syndrome	RSRF (2002) <i>MECP2 LSDB entry</i> Unpublished:	Source: LSDB
MECP2	CM066124	GGG-GAG	Gly161Glu	482G>A	G161E	Rett syndrome	Kim (2006) <i>Exp Mol Med</i> 38:119	PubMed 16672765
MECP2	CM010338	GGG-TGG	Gly161Trp	481G>T	G161W	Rett syndrome	Bourdon (2001) <i>Hum Genet</i> 108:43	PubMed 11214906
MECP2	CM014899	GGG-GTG	Gly161Val	482G>T	G161V	Rett syndrome	Milunsky (2001) <i>Genet Test</i> 5:321	PubMed 11960578
MECP2	CM993353	CGA-TGA	Arg168Term	502C>T	R168*	Rett syndrome	Wan (1999) <i>Am J Hum Genet</i> 65:1520	PubMed 10577905
MECP2	CM001228	CAG-TAG	Gln170Term	508C>T	Q170*	Rett syndrome	Amir (2000) <i>Ann Neurol</i> 47:670	PubMed 10805343
MECP2	CM135744	CCT-CGT	Pro173Arg	518C>G	P173R	Rett syndrome	Chapleau (2013) <i>Am J Med Genet A</i> 161:1638	PubMed 23696494
MECP2	CM035703	AAG-TAG	Lys175Term	523A>T	K175*	Rett syndrome	Friez (2003) <i>MECP2 LSDB entry</i> Unpublished:1679	Source: LSDB
MECP2	CM023414	AAA-TAA	Lys177Term	529A>T	K177*	Rett syndrome	RSRF (2002) <i>MECP2 LSDB entry</i> Unpublished: Fendri-Kriaa (2011) <i>Biochem Biophys Res Commun</i> 409:270	Source: LSDB PubMed 21575601
MECP2	CM113963	CCC-TCC	Pro179Ser	535C>T	P179S	Rett syndrome		
MECP2	CM060326	AAA-TAA	Lys180Term	538A>T	K180*	Rett syndrome	Philippe (2006) <i>Eur J Med Genet</i> 49:9	PubMed 16473305
MECP2	CM1212523	AAA-TAA	Lys192Term	574A>T	K192*	Rett syndrome	Zvereff (2012) <i>Brain Dev</i> 34:750	PubMed 22277191
MECP2	CM023815	ACG-ATG	Thr197Met	590C>T	T197M	Rett syndrome	Bienvenu (2002) <i>Genet Test</i> 6:1	PubMed 12180070
MECP2	CM001229	AGA-TGA	Arg198Term	592A>T	R198*	Rett syndrome	Bienvenu (2000) <i>Hum Mol Genet</i> 9:1377	PubMed 10814719
MECP2	CM030693	AAG-TAG	Lys200Term	598A>T	K200*	Rett syndrome	Weaving (2003) <i>Am J Med Genet</i> 118A:103	PubMed 12655490
MECP2	CM023816	GCG-GTG	Ala201Val	602C>T	A201V	Rett syndrome	Bienvenu (2002) <i>Genet Test</i> 6:1	PubMed 12180070
MECP2	CM003456	TCA-TGA	Ser204Term	611C>G	S204*	Rett syndrome	Buyse (2000) <i>Am J Hum Genet</i> 67:1428	PubMed 11055898
MECP2	CM010339	GAG-TAG	Glu205Term	613G>T	E205*	Rett syndrome	Laccone (2001) <i>Hum Mutat</i> 17:183	PubMed 11241840
MECP2	CM010340	CAG-TAG	Gln208Term	622C>T	Q208*	Rett syndrome	Vacca (2001) <i>J Mol Med</i> 78:648	PubMed 11269512
MECP2	CM010341	AAA-ATA	Lys210Ile	629A>T	K210I	Rett syndrome	Laccone (2001) <i>Hum Mutat</i> 17:183	PubMed 11241840
MECP2	CM000748	CCT-CGT	Pro225Arg	674C>G	P225R	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337
MECP2	CM032953	CCT-CTT	Pro225Leu	674C>T	P225L	Rett syndrome	Moog (2003) <i>Eur J Paediatr Neurol</i> 7:5 No author given (2007) <i>MECP2 LSDB entry</i> Unpublished:2460	PubMed 12615169 Source: LSDB
MECP2	CM076289	GGT-GTT	Gly237Val	710G>T	G237V	Rett syndrome		
MECP2	CM003767	CAG-TAG	Gln244Term	730C>T	Q244*	Rett syndrome	Dragich (2000) <i>Hum Mol Genet</i> 9:2365	PubMed 11005791
MECP2	CM023415	CGC-TGC	Arg250Cys	748C>T	R250C	Rett syndrome	Das (2002) <i>MECP2 LSDB entry</i> Pers comm:	Source: LSDB
MECP2	CM035704	AAG-TAG	Lys254Term	760A>T	K254*	Rett syndrome	Friez (2003) <i>MECP2 LSDB entry</i> Unpublished:1790	Source: LSDB
MECP2	CM992179	CGA-TGA	Arg255Term	763C>T	R255*	Rett syndrome	Amir (1999) <i>Nat Genet</i> 23:185	PubMed 10508514
MECP2	CM001230	AAA-TAA	Lys256Term	766A>T	K256*	Rett syndrome	Amir (2000) <i>Ann Neurol</i> 47:670	PubMed 10805343
MECP2	CM014900	CAG-TAG	Gln262Term	784C>T	Q262*	Rett syndrome	Milunsky (2001) <i>Genet Test</i> 5:321	PubMed 11960578
MECP2	CM106874	AAG-TAG	Lys266Term	796A>T	K266*	Rett syndrome	Shapiro (2010) <i>Pediatr Res</i> 68:446	PubMed 20661168
MECP2	CM076287	AAA-TAA	Lys267Term	799A>T	K267*	Rett syndrome	Djarmati (2007) <i>Acta Neurol Scand</i> 116:413	PubMed 17986102

MECP2	CM014976	CGG-TGG	Arg268Trp	802C>T	R268W	Rett syndrome	Erlandson (2001) <i>Eur Child Adolesc Psychiatry</i> 10:117	PubMed 11469283
MECP2	CM000749	CGA-TGA	Arg270Term	808C>T	R270*	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337
MECP2	CM058036	CCG-TCG	Pro272Ser	814C>T	P272S	Rett syndrome	Gauthier (2005) <i>Can J Neurol Sci</i> 32:321	PubMed 16225173
MECP2	CM135743	GCC-ACC	Ala278Thr	832G>A	A278T	Rett syndrome	Chapleau (2013) <i>Am J Med Genet A</i> 161:1638	PubMed 23696494
MECP2	CM045299	GCT-GTT	Ala279Val	836C>T	A279V	Rett syndrome	Chae (2004) <i>J Child Neurol</i> 19:503	PubMed 15526954
MECP2	CM023416	AAA-AGA	Lys286Arg	857A>G	K286R	Rett syndrome	Das (2002) <i>MECP2 LSDB entry</i> Pers comm:	Source: LSDB
MECP2	CM057719	GTG-ATG	Val288Met	862G>A	V288M	Rett syndrome	Charman (2005) <i>Eur J Hum Genet</i> 13:1121 No author given (2007) <i>MECP2 LSDB entry</i> Unpublished:2105	PubMed 16077736 Source: LSDB
MECP2	CM076288	AAG-TAG	Lys289Term	865A>T	K289*	Rett syndrome	Zhang (2012) <i>Clin Genet</i> 82:526	PubMed 22182064
MECP2	CM1210654	GAG-TAG	Glu290Term	868G>T	E290*	Rett syndrome	Deutscher (2002) <i>Klin Padiatr</i> 214:291	PubMed 12235545
MECP2	CM023745	CGA-GGA	Arg294Gly	880C>G	R294G	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337
MECP2	CM000750	CGA-TGA	Arg294Term	880C>T	R294*	Rett syndrome	Nicolao (2001) <i>Hum Mutat</i> 18:132	PubMed 11462237
MECP2	CM012403	CAG-TAG	Gln297Term	889C>T	Q297*	Rett syndrome	Nicolao (2001) <i>Hum Mutat</i> 18:132	PubMed 11462237
MECP2	CM012404	GTA-ATA	Val300Ile	898G>A	V300I	Rett syndrome	Huppke (2000) <i>Hum Mol Genet</i> 9:1369	PubMed 10814718
MECP2	CM001231	CCC-GCC	Pro302Ala	904C>G	P302A	Rett syndrome	Bienvenu (2000) <i>Hum Mol Genet</i> 9:1377	PubMed 10814719
MECP2	CM001232	CCC-CGC	Pro302Arg	905C>G	P302R	Rett syndrome	Amano (2000) <i>J Hum Genet</i> 45:231	PubMed 10944854
MECP2	CM003457	CCC-CAC	Pro302His	905C>A	P302H	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119	PubMed 10767337
MECP2	CM000751	CCC-CTC	Pro302Leu	905C>T	P302L	Rett syndrome	Zahorakova (2007) <i>J Hum Genet</i> 52:342	PubMed 17387578
MECP2	CM071858	CCC-TCC	Pro302Ser	904C>T	P302S	Rett syndrome	Fukuda (2005) <i>Brain Dev</i> 27:211	PubMed 15737703
MECP2	CM055981	CCC-ACC	Pro302Thr	904C>A	P302T	Rett syndrome	Philippe (2006) <i>Eur J Med Genet</i> 49:9	PubMed 16473305
MECP2	CM060325	ATC-AGC	Ile303Ser	908T>G	I303S	Rett syndrome	Das (2002) <i>MECP2 LSDB entry</i> Pers comm:	Source: LSDB
MECP2	CM023417	AAG-AGG	Lys304Arg	911A>G	K304R	Rett syndrome	Charman (2005) <i>Eur J Hum Genet</i> 13:1121	PubMed 16077736
MECP2	CM023418	AAG-CAG	Lys304Gln	910A>C	K304Q	Rett syndrome	Philippe (2006) <i>Eur J Med Genet</i> 49:9	PubMed 16473305
MECP2	CM060327	AAG-GAG	Lys304Glu	910A>G	K304E	Rett syndrome	Dragich (2000) <i>Hum Mol Genet</i> 9:2365	PubMed 11005791
MECP2	CM003768	AAG-AGG	Lys305Arg	914A>G	K305R	Rett syndrome	Li (2007) <i>J Hum Genet</i> 52:38	PubMed 17089071
MECP2	CM070193	AAG-GAG	Lys305Glu	913A>G	K305E	Rett syndrome	Wan (1999) <i>Am J Hum Genet</i> 65:1520	PubMed 10577905
MECP2	CM993354	CGC-TGC	Arg306Cys	916C>T	R306C	Rett syndrome	Cheadle (2000) <i>Hum Mol Genet</i> 9:1119 No author given (2007) <i>MECP2 LSDB entry</i> Unpublished:2206	PubMed 10767337 Source: LSDB
MECP2	CM000752	CGC-CAC	Arg306His	917G>A	R306H	Rett syndrome	Shapiro (2010) <i>Pediatr Res</i> 68:446	PubMed 20661168
MECP2	CM076285	CGC-CTC	Arg306Leu	917G>T	R306L	Rett syndrome	Kim (2006) <i>Exp Mol Med</i> 38:119	PubMed 16672765
MECP2	CM106875	CGC-CCC	Arg306Pro	917G>C	R306P	Rett syndrome	RSRF (2002) <i>MECP2 LSDB entry</i> Unpublished:	Source: LSDB
MECP2	CM066121	ACG-ATG	Thr311Met	932C>T	T311M	Rett syndrome	Bienvenu (2000) <i>Hum Mol Genet</i> 9:1377	PubMed 10814719
MECP2	CM023419	GAA-GCA	Glu318Ala	953A>C	E318A	Rett syndrome	Huppke (2000) <i>Hum Mol Genet</i> 9:1369	PubMed 10814718
MECP2	CM001233	CCC-GCC	Pro322Ala	964C>G	P322A	Rett syndrome	Gauthier (2005) <i>Can J Neurol Sci</i> 32:321	PubMed 16225173
MECP2	CM001234	CCC-CTC	Pro322Leu	965C>T	P322L	Rett syndrome	Philippe (2006) <i>Eur J Med Genet</i> 49:9	PubMed 16473305
MECP2	CM058037	CCC-ACC	Pro322Thr	964C>A	P322T	Rett syndrome	Fendri-Kriaa (2012) <i>J Child Neurol</i> 27:564	PubMed 21940684
MECP2	CM060328	CTC-GTC	Leu328Val	982C>G	L328V	Rett syndrome	Laccone (2002) <i>J Med Genet</i> 39:586 No author given (2007) <i>MECP2 LSDB entry</i> Unpublished:2206	PubMed 12161600 Source: LSDB
MECP2	CM118477	CGG-GGG	Arg344Gly	1030C>G	R344G	Rett syndrome	No author given (2007) <i>MECP2 LSDB entry</i> Unpublished:2025	Source: LSDB
MECP2	CM024216	CGG-TGG	Arg344Trp	1030C>T	R344W	Rett syndrome	Fendri-Kriaa (2012) <i>J Child Neurol</i> 27:564	PubMed 21940684
MECP2	CM076284	CGC-CAC	Arg354His	1061G>A	R354H	Rett syndrome		
MECP2	CM076286	CGC-CTC	Arg354Leu	1061G>T	R354L	Rett syndrome		
MECP2	CM118476	AGC-AGA	Ser355Arg	1065C>A	S355R	Rett syndrome		

MECP2	CM070192	GCC-ACC	Ala358Thr	1072G>A	A358T	Rett syndrome	Li (2007) <i>J Hum Genet</i> 52:38	PubMed 17089071
MECP2	CM023817	TCC-CCC	Ser359Pro	1075T>C	S359P	Rett syndrome	Bienvenu (2002) <i>Genet Test</i> 6:1	PubMed 12180070
MECP2	CM003458	TCA-TAA	Ser360Term	1079C>A	S360*	Rett syndrome	Buyse (2000) <i>Am J Hum Genet</i> 67:1428	PubMed 11055898
MECP2	CM035701	AAG-TAG	Lys363Term	1087A>T	K363*	Rett syndrome	Friez (2003) <i>MECP2 LADB entry</i> Unpublished:1717	Source: LADB
MECP2	CM0910821	TCA-TGA	Ser373Term	1118C>G	S373*	Rett syndrome	Smeets (2009) <i>Am J Med Genet A</i> 149A:199	PubMed 19133691
MECP2	CM014901	GCC-GGC	Ala378Gly	1133C>G	A378G	Rett syndrome	Milunsky (2001) <i>Genet Test</i> 5:321	PubMed 11960578
MECP2	CM124038	CCA-CTA	Pro381Leu	1142C>T	P381L	Rett syndrome	Fendri-Kriaa (2012) <i>Gene</i> 502:163	PubMed 22561697
MECP2	CM014902	CCA-CTA	Pro388Leu	1163C>T	P388L	Rett syndrome	Milunsky (2001) <i>Genet Test</i> 5:321	PubMed 11960578
MECP2	CM030694	CCA-TCA	Pro388Ser	1162C>T	P388S	Rett syndrome	Conforti (2003) <i>Am J Med Genet</i> 117A:184 Vorsanova (2002) <i>Zh Nevrol Psikhiatr Im S S Korsakova</i> 102:23	PubMed 12567420
MECP2	CM025497	CCA-ACA	Pro388Thr	1162C>A	P388T	Rett syndrome		PubMed 12449561
MECP2	CM016262	GAG-AAG	Glu397Lys	1189G>A	E397K	Rett syndrome	Giunti (2001) <i>Brain Dev</i> 23S1:S242	PubMed 11738883
MECP2	CM0911290	GAG-TAG	Glu397Term	1189G>T	E397*	Rett syndrome	Raizis (2009) <i>N Z Med J</i> 122:21	PubMed 19652677
MECP2	CM003326	CAG-TAG	Gln406Term	1216C>T	Q406*	Rett syndrome	Meloni (2000) <i>Am J Hum Genet</i> 67:982	PubMed 10986043
MECP2	CM058038	CCC-TCC	Pro419Ser	1255C>T	P419S	Rett syndrome	Gauthier (2005) <i>Can J Neurol Sci</i> 32:321	PubMed 16225173
MECP2	CM024129	GAG-TAG	Glu455Term	1363G>T	E455*	Rett syndrome	Maiwald (2002) <i>Neurogenetics</i> 4:107	PubMed 12481990
MECP2	CM1212526	GTG-ATG	Val481Met	1441G>A	V481M	Rett syndrome	Zvereff (2012) <i>Brain Dev</i> 34:750 Erlandson (2001) <i>Eur Child Adolesc Psychiatry</i> 10:117	PubMed 22277191
MECP2	CM014977	TGA-CGA	Term487Arg	1459T>C	*487R	Rett syndrome		PubMed 11469283
MECP2	CM001235	TGA-TGC	Term487Cys	1461A>C	*487C	Rett syndrome	Bienvenu (2000) <i>Hum Mol Genet</i> 9:1377	PubMed 10814719
MECP2	CM096401	TGA-TTA	Term487Leu	1460G>T	*487L	Rett syndrome	Lima (2009) <i>Arq Neuropsiquiatr</i> 67:577	PubMed 19722030
MECP2	CM023421	TGA-TGG	Term487Trp	1461A>G	*487W	Rett syndrome	RSRF (2002) <i>MECP2 LADB entry</i> Unpublished:	Source: LADB
NRAS	CM112982	ATC-AAC	Ile24Asn	71T>A	I24N	Noonan syndrome	Runtuwene (2011) <i>Dis Model Mech</i> 4:393	PubMed 21263000
NRAS	CM128754	CCC-CTC	Pro34Leu	101C>T	P34L	Noonan syndrome	Denayer (2012) <i>Mol Syndromol</i> 3:34	PubMed 22855653
NRAS	CM100126	ACC-ATC	Thr50Ile	149C>T	T50I	Noonan syndrome	Cirstea (2010) <i>Nat Genet</i> 42:27	PubMed 19966803
NRAS	CM100127	GGA-GAA	Gly60Glu	179G>A	G60E	Noonan syndrome	Cirstea (2010) <i>Nat Genet</i> 42:27	PubMed 19966803
PKD1	CM146749	GGA-GGT	Gly109Gly	327A>T	G109G	Polycystic kidney disease	Gonzalez-Paredes (2014) <i>Gene</i> 546:243	PubMed 24907393
PKD1	CM143894	TGC-CGC	Cys522Arg	1564T>C	C522R	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143939	GCG-GTG	Ala1201Val	3602C>T	A1201V	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143896	GAG-TAG	Glu1884Term	5650G>T	E1884*	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143897	AAC-GAC	Asn2044Asp	6130A>G	N2044D	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143898	CAG-TAG	Gln2196Term	6586C>T	Q2196*	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143903	TGC-TAC	Cys2495Tyr	7484G>A	C2495Y	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143905	CTG-CGG	Leu2619Arg	7856T>G	L2619R	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143908	TGG-TAG	Trp3195Term	9584G>A	W3195*	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143940	GCA-ACA	Ala3240Thr	9718G>A	A3240T	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143930	CAG-TAG	Gln3441Term	10321C>T	Q3441*	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143934	TGG-TGA	Trp3724Term	11172G>A	W3724*	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM143935	CGG-GGG	Arg3750Gly	11248C>G	R3750G	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM146750	CGG-AGG	Arg3753Arg	11257C>A	R3753R	Polycystic kidney disease	Gonzalez-Paredes (2014) <i>Gene</i> 546:243	PubMed 24907393
PKD1	CM143936	CAG-TAG	Gln3962Term	11884C>T	Q3962*	Polycystic kidney disease	Obeidová (2014) <i>BMC Med Genet</i> 15:41	PubMed 24694054
PKD1	CM010371	CTG-CAG	Leu13Gln	38T>A	L13Q	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM108813	TGC-TGA	Cys37Term	111C>A	C37*	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670

PKD1	CM1210912	TGC-TGG	Cys51Trp	153C>G	C51W	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM010372	TCC-TTC	Ser75Phe	224C>T	S75F	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM074434	TCG-TAG	Ser91Term	272C>A	S91*	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM108786	GAT-GGT	Asp97Gly	290A>G	D97G	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM090554	AGC-ATC	Ser99Ile	296G>T	S99I	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	CM121779	GGA-AGA	Gly109Arg	325G>A	G109R	Polycystic kidney disease 1	Neumann (2012) <i>Int Urol Nephrol</i> 44:1753	PubMed 22367170
PKD1	CM121780	ATA-ACA	Ile120Thr	359T>C	I120T	Polycystic kidney disease 1	Neumann (2012) <i>Int Urol Nephrol</i> 44:1753	PubMed 22367170
PKD1	CM1210928	TGT-CGT	Cys129Arg	385T>C	C129R	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076407	CGA-TGA	Arg138Term	412C>T	R138*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM010373	TGG-TGT	Trp139Cys	417G>T	W139C	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1210929	TGT-TAT	Cys155Tyr	464G>A	C155Y	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM034111	CAG-CGG	Gln164Arg	491A>G	Q164R	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM1210930	TGC-TAC	Cys183Tyr	548G>A	C183Y	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM034112	TGC-GGC	Cys210Gly	628T>G	C210G	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM010374	TCG-TAG	Ser225Term	674C>A	S225*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM010375	CAG-TAG	Gln227Term	679C>T	Q227*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM076414	TGC-AGC	Cys230Ser	688T>A	C230S	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM022652	TGC-TGA	Cys230Term	690C>A	C230*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM1210851	CAG-TAG	Gln236Term	706C>T	Q236*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076431	TGT-TAT	Cys259Tyr	776G>A	C259Y	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM022653	CAG-TAG	Gln266Term	796C>T	Q266*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM074427	GCC-GAC	Ala271Asp	812C>A	A271D	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM119057	CAG-TAG	Gln288Term	862C>T	Q288*	Polycystic kidney disease 1	Bataille (2011) <i>BMC Nephrol</i> 12:57	PubMed 22008521
PKD1	CM085643	GAG-TAG	Glu313Term	937G>T	E313*	Polycystic kidney disease 1	Connor (2008) <i>Am J Transplant</i> 8:232	PubMed 17973957
PKD1	CM991042	CGC-CTC	Arg324Leu	971G>T	R324L	Polycystic kidney disease 1	Thomas (1999) <i>Am J Hum Genet</i> 65:39	PubMed 10364515
PKD1	CM076417	TAT-TGT	Tyr325Cys	974A>G	Y325C	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM076402	CAG-TAG	Gln351Term	1051C>T	Q351*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210852	GAG-TAG	Glu371Term	1111G>T	E371*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM020485	GGT-TGT	Gly381Cys	1141G>T	G381C	Polycystic kidney disease 1	McCluskey (2002) <i>Hum Mutat</i> 19:240	PubMed 11857740
PKD1	CM076392	GGT-AGT	Gly381Ser	1141G>A	G381S	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1312937	GGT-GTT	Gly381Val	1142G>T	G381V	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM013784	CGA-TGA	Arg400Term	1198C>T	R400*	Polycystic kidney disease 1	Peters (2001) <i>Eur J Hum Genet</i> 9:957	PubMed 11840199
PKD1	CM1312938	CCG-CTG	Pro404Leu	1211C>T	P404L	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM074431	TAC-TGC	Tyr420Cys	1259A>G	Y420C	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1312939	CGC-TGC	Arg421Cys	1261C>T	R421C	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM076406	GCG-GTG	Ala432Val	1295C>T	A432V	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM108787	TGT-CGT	Cys436Arg	1306T>C	C436R	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM076432	TGT-TAT	Cys436Tyr	1307G>A	C436Y	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM108792	GCC-CCC	Ala442Pro	1324G>C	A442P	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM076424	TGG-TGA	Trp467Term	1401G>A	W467*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210853	GAG-TAG	Glu506Term	1516G>T	E506*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM034113	TGC-CGC	Cys508Arg	1522T>C	C508R	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM1210854	TGC-TGA	Cys508Term	1524C>A	C508*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176

PKD1	CM1312940	GGG-AGG	Gly515Arg	1543G>A	G515R	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM1210931	CAC-GAC	His526Asp	1576C>G	H526D	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM074432	TAC-TGC	Tyr528Cys	1583A>G	Y528C	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210855	CAG-TAG	Gln533Term	1597C>T	Q533*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076380	CAG-TAG	Gln562Term	1684C>T	Q562*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM020486	CAG-TAG	Gln563Term	1687C>T	Q563*	Polycystic kidney disease 1	McCluskey (2002) <i>Hum Mutat</i> 19:240	PubMed 11857740
PKD1	CM1210932	GTA-GGA	Val577Gly	1730T>G	V577G	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM085642	CGT-TGT	Arg585Cys	1753C>T	R585C	Polycystic kidney disease 1	Tsakamoto (2008) <i>Clin Exp Nephrol</i> 12:82	PubMed 18175055
PKD1	CM076398	CTG-CGG	Leu605Arg	1814T>G	L605R	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM074421	CGG-TGG	Arg611Trp	1831C>T	R611W	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM057348	TCC-TTC	Ser629Phe	1886C>T	S629F	Polycystic kidney disease 1	Zhang (2005) <i>Nephron Exp Nephrol</i> 100:e63	PubMed 15775720
PKD1	CM1210856	GGA-TGA	Gly644Term	1930G>T	G644*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210933	GAG-AAG	Glu685Lys	2053G>A	E685K	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM034114	GTT-GAT	Val690Asp	2069T>A	V690D	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM1210857	CAG-TAG	Gln697Term	2089C>T	Q697*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210934	TAC-GAC	Tyr698Asp	2092T>G	Y698D	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM010376	CAG-TAG	Gln705Term	2113C>T	Q705*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1210858	CAG-TAG	Gln718Term	2152C>T	Q718*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM108793	CTG-CGG	Leu727Arg	2180T>G	L727R	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM076395	CTG-CCG	Leu727Pro	2180T>C	L727P	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM010377	TCA-TGA	Ser749Term	2246C>G	S749*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM991043	TTG-TCG	Leu845Ser	2534T>C	L845S	Polycystic kidney disease 1	Thomas (1999) <i>Am J Hum Genet</i> 65:39	PubMed 10364515
PKD1	CM108814	CAG-TAG	Gln848Term	2542C>T	Q848*	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM1210859	TGG-TAG	Trp861Term	2582G>A	W861*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076421	TGC-TGA	Cys874Term	2622C>A	C874*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210860	TAC-TAG	Tyr953Term	2859C>G	Y953*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1212765	CGG-TGG	Arg966Trp	2896C>T	R966W	Polycystic kidney disease 1	Lalaye (2012) <i>Genet Couns</i> 23:435	PubMed 23431742
PKD1	CM014073	TGG-CGG	Trp967Arg	2899T>C	W967R	Polycystic kidney disease 1	Phakdeekitcharoen (2001) <i>J Am Soc Nephrol</i> 12:955	PubMed 11316854
PKD1	CM1210861	TAT-TAG	Tyr986Term	2958T>G	Y986*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM021120	CAG-CAT	Gln987His	2961G>T	Q987H	Polycystic kidney disease 1	Burtey (2002) <i>J Med Genet</i> 39:422	PubMed 12070253
PKD1	CM053381	CAG-TAG	Gln1020Term	3058C>T	Q1020*	Polycystic kidney disease 1	Peltola (2005) <i>J Mol Med</i> 83:638	PubMed 15772804
PKD1	CM104475	GTG-ATG	Val1045Met	3133G>A	V1045M	Polycystic kidney disease 1	Vujic (2010) <i>J Am Soc Nephrol</i> 21:1097	PubMed 20558538
PKD1	CM074435	TCG-TTG	Ser1047Leu	3140C>T	S1047L	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210862	CAG-TAG	Gln1062Term	3184C>T	Q1062*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210863	TAC-TAG	Tyr1071Term	3213C>G	Y1071*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076420	TCG-TAG	Ser1081Term	3242C>A	S1081*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM076416	TAC-TAG	Tyr1095Term	3285C>G	Y1095*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM053382	CAG-TAG	Gln1117Term	3349C>T	Q1117*	Polycystic kidney disease 1	Peltola (2005) <i>J Mol Med</i> 83:638	PubMed 15772804
PKD1	CM1210864	TAC-TAG	Tyr1147Term	3441C>G	Y1147*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210865	TAC-TAG	Tyr1159Term	3477C>G	Y1159*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM034562	TGG-TGA	Trp1161Term	3483G>A	W1161*	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM003329	GGC-AGC	Gly1166Ser	3496G>A	G1166S	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM1210913	GTG-GGG	Val1206Gly	3617T>G	V1206G	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176

PKD1	CM1210866	GGA-TGA	Gly1214Term	3640G>T	G1214*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210935	ATC-AAC	Ile1241Asn	3722T>A	I1241N	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM090555	ACG-ATG	Thr1242Met	3725C>T	T1242M	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	CM1210867	TGG-TGA	Trp1243Term	3729G>A	W1243*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1111449	GTG-ATG	Val1274Met	3820G>A	V1274M	Polycystic kidney disease 1	Bergmann (2011) <i>J Am Soc Nephrol</i> 22:2047	PubMed 22034641
PKD1	CM057350	TGG-TGA	Trp1328Term	3984G>A	W1328*	Polycystic kidney disease 1	Zhang (2005) <i>Nephron Exp Nephrol</i> 100:e63	PubMed 15775720
PKD1	CM022654	CGG-TGG	Arg1340Trp	4018C>T	R1340W	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM074430	CGG-TGG	Arg1351Trp	4051C>T	R1351W	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210868	GAG-TAG	Glu1379Term	4135G>T	E1379*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076393	TAC-GAC	Tyr1412Asp	4234T>G	Y1412D	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM074418	CGA-TGA	Arg1436Term	4306C>T	R1436*	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM108815	TCA-TGA	Ser1457Term	4370C>G	S1457*	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM1210936	GTG-GGG	Val1460Gly	4379T>G	V1460G	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210869	CAG-TAG	Gln1463Term	4387C>T	Q1463*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM108816	CAG-TAG	Gln1483Term	4447C>T	Q1483*	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM076412	GGG-CGG	Gly1503Arg	4507G>C	G1503R	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210937	TAC-TGC	Tyr1517Cys	4550A>G	Y1517C	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210870	TAC-TAG	Tyr1517Term	4551C>G	Y1517*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM010378	GAG-TAG	Glu1537Term	4609G>T	E1537*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1210871	TGG-TGA	Trp1539Term	4617G>A	W1539*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM104476	ACG-ATG	Thr1570Met	4709C>T	T1570M	Polycystic kidney disease 1	Vujic (2010) <i>J Am Soc Nephrol</i> 21:1097	PubMed 20558538
PKD1	CM076430	TGG-TGA	Trp1582Term	4746G>A	W1582*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210872	TAC-TAG	Tyr1599Term	4797C>G	Y1599*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM119593	TCC-TTC	Ser1619Phe	4856C>T	S1619F	Polycystic kidney disease 1	Irazabal (2011) <i>Clin J Am Soc Nephrol</i> 6:1274	PubMed 21551026
PKD1	CM090559	CAG-TAG	Gln1630Term	4888C>T	Q1630*	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	CM022655	CAG-TAG	Gln1636Term	4906C>T	Q1636*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM1210873	CAG-TAG	Gln1651Term	4951C>T	Q1651*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM003330	CAG-TAG	Gln1653Term	4957C>T	Q1653*	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM034115	ACT-CCT	Thr1667Pro	4999A>C	T1667P	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM119058	TAC-TAA	Tyr1693Term	5079C>A	Y1693*	Polycystic kidney disease 1	Bataille (2011) <i>BMC Nephrol</i> 12:57	PubMed 22008521
PKD1	CM076403	CAG-TAG	Gln1696Term	5086C>T	Q1696*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM076422	TGG-TAG	Trp1707Term	5120G>A	W1707*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210914	CCG-CTG	Pro1727Leu	5180C>T	P1727L	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210874	GAG-TAG	Glu1764Term	5290G>T	E1764*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM022656	GAG-AAG	Glu1811Lys	5431G>A	E1811K	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM023748	CAG-TAG	Gln1828Term	5482C>T	Q1828*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM022657	TGG-TGA	Trp1837Term	5511G>A	W1837*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM074415	ACC-ATC	Thr1861Ile	5582C>T	T1861I	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210915	AAC-GAC	Asn1870Asp	5608A>G	N1870D	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076382	AAC-CAC	Asn1870His	5608A>C	N1870H	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM119060	AAC-AAG	Asn1870Lys	5610C>G	N1870K	Polycystic kidney disease 1	Bataille (2011) <i>BMC Nephrol</i> 12:57	PubMed 22008521
PKD1	CM971178	TGG-TGA	Trp1874Term	5622G>A	W1874*	Polycystic kidney disease 1	Roelfsema (1997) <i>Am J Hum Genet</i> 61:1044	PubMed 9345095
PKD1	CM991044	CAG-TAG	Gln1922Term	5764C>T	Q1922*	Polycystic kidney disease 1	Thomas (1999) <i>Am J Hum Genet</i> 65:39	PubMed 10364515

PKD1	CM003331	GTG-GAG	Val1956Glu	5867T>A	V1956E	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM003332	CAG-TAG	Gln1960Term	5878C>T	Q1960*	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM119059	CAG-TAG	Gln1975Term	5923C>T	Q1975*	Polycystic kidney disease 1	Bataille (2011) <i>BMC Nephrol</i> 12:57	PubMed 22008521
PKD1	CM076391	GGC-AGC	Gly1999Ser	5995G>A	G1999S	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM076411	GGC-GTC	Gly1999Val	5996G>T	G1999V	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210875	CAG-TAG	Gln2011Term	6031C>T	Q2011*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM021121	CAG-TAG	Gln2014Term	6040C>T	Q2014*	Polycystic kidney disease 1	Inoue (2002) <i>Hum Mutat</i> 19:622	PubMed 12007219
PKD1	CM003333	CAG-TAG	Gln2039Term	6115C>T	Q2039*	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM076409	GAG-TAG	Glu2056Term	6166G>T	E2056*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM121781	GCC-GGC	Ala2060Gly	6179C>G	A2060G	Polycystic kidney disease 1	Neumann (2012) <i>Int Urol Nephrol</i> 44:1753	PubMed 22367170
PKD1	CM076404	CAG-TAG	Gln2067Term	6199C>T	Q2067*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM021122	ACC-ATC	Thr2083Ile	6248C>T	T2083I	Polycystic kidney disease 1	Inoue (2002) <i>Hum Mutat</i> 19:622	PubMed 12007219
PKD1	CM022658	TAC-TGC	Tyr2092Cys	6275A>G	Y2092C	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM1210876	CAG-TAG	Gln2103Term	6307C>T	Q2103*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1312941	TAC-TGC	Tyr2114Cys	6341A>G	Y2114C	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM121077	TTC-TGC	Phe2132Cys	6395T>G	F2132C	Polycystic kidney disease 1	O'Brien (2012) <i>J Pediatr Gastroenterol Nutr</i> 54:83	PubMed 21694639
PKD1	CM022659	CAG-TAG	Gln2158Term	6472C>T	Q2158*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM010379	CGA-TGA	Arg2163Term	6487C>T	R2163*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM022660	TCA-TGA	Ser2164Term	6491C>G	S2164*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM074428	CGC-TGC	Arg2166Cys	6496C>T	R2166C	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210877	TAC-TAG	Tyr2181Term	6543C>G	Y2181*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM074417	CAG-CGG	Gln2182Arg	6545A>G	Q2182R	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM020487	TAC-GAC	Tyr2185Asp	6553T>G	Y2185D	Polycystic kidney disease 1	McCluskey (2002) <i>Hum Mutat</i> 19:240	PubMed 11857740
PKD1	CM1210938	TGG-CGG	Trp2187Arg	6559T>C	W2187R	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210878	TGG-TAG	Trp2187Term	6560G>A	W2187*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210879	TAT-TAA	Tyr2190Term	6570T>A	Y2190*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM034116	CGC-TGC	Arg2200Cys	6598C>T	R2200C	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM104474	CGG-TGG	Arg2220Trp	6658C>T	R2220W	Polycystic kidney disease 1	Vujic (2010) <i>J Am Soc Nephrol</i> 21:1097	PubMed 20558538
PKD1	CM010380	TGC-TGA	Cys2229Term	6687C>A	C2229*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM010381	CAG-TAG	Gln2243Term	6727C>T	Q2243*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM001302	ACG-ATG	Thr2250Met	6749C>T	T2250M	Polycystic kidney disease 1	Perrichot (2000) <i>Eur J Hum Genet</i> 8:353	PubMed 10854095
PKD1	CM1111444	CGC-TGC	Arg2255Cys	6763C>T	R2255C	Polycystic kidney disease 1	Bergmann (2011) <i>J Am Soc Nephrol</i> 22:2047	PubMed 22034641
PKD1	CM076418	TCA-TGA	Ser2264Term	6791C>G	S2264*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM074433	TAC-TAA	Tyr2265Term	6795C>A	Y2265*	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM020488	TCA-TGA	Ser2269Term	6806C>G	S2269*	Polycystic kidney disease 1	McCluskey (2002) <i>Hum Mutat</i> 19:240	PubMed 11857740
PKD1	CM076429	GGG-AGG	Gly2278Arg	6832G>A	G2278R	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210939	TAC-TGC	Tyr2282Cys	6845A>G	Y2282C	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076384	CAG-TAG	Gln2291Term	6871C>T	Q2291*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM076383	CAC-CCC	His2297Pro	6890A>C	H2297P	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210940	TGG-GGG	Trp2298Gly	6892T>G	W2298G	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210880	TGG-TAG	Trp2298Term	6893G>A	W2298*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076410	GAG-TAG	Glu2307Term	6919G>T	E2307*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM001303	CGG-TGG	Arg2329Trp	6985C>T	R2329W	Polycystic kidney disease 1	Perrichot (2000) <i>Eur J Hum Genet</i> 8:353	PubMed 10854095

PKD1	CM010382	TAC-GAC	Tyr2336Asp	7006T>G	Y2336D	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM001304	TAC-TAA	Tyr2336Term	7008C>A	Y2336*	Polycystic kidney disease 1	Perrichot (2000) <i>Eur J Hum Genet</i> 8:353	PubMed 10854095
PKD1	CM076425	TGG-TGA	Trp2343Term	7029G>A	W2343*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210881	GAG-TAG	Glu2349Term	7045G>T	E2349*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210882	CAG-TAG	Gln2354Term	7060C>T	Q2354*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM034117	TGT-CGT	Cys2370Arg	7108T>C	C2370R	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM076415	TGT-AGT	Cys2370Ser	7108T>A	C2370S	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM010383	TGT-TGA	Cys2370Term	7110T>A	C2370*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1210916	TCC-TGC	Ser2372Cys	7115C>G	S2372C	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM034118	TGC-TAC	Cys2373Tyr	7118G>A	C2373Y	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM022661	CAG-TAG	Gln2376Term	7126C>T	Q2376*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM1210917	TAC-TGC	Tyr2379Cys	7136A>G	Y2379C	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210883	TAC-TAG	Tyr2379Term	7137C>G	Y2379*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210884	TAC-TAG	Tyr2386Term	7158C>G	Y2386*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM108788	GGC-GAC	Gly2391Asp	7172G>A	G2391D	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM003334	CGA-TGA	Arg2402Term	7204C>T	R2402*	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM1210885	TGG-TGA	Trp2405Term	7215G>A	W2405*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM003335	CGT-TGT	Arg2408Cys	7222C>T	R2408C	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM090556	ACA-AAA	Thr2422Lys	7265C>A	T2422K	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	CM119592	TCC-TTC	Ser2423Phe	7268C>T	S2423F	Polycystic kidney disease 1	Irazabal (2011) <i>Clin J Am Soc Nephrol</i> 6:1274	PubMed 21551026
PKD1	CM003336	CGA-TGA	Arg2430Term	7288C>T	R2430*	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM108789	CGG-TGG	Arg2434Trp	7300C>T	R2434W	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM074423	CTC-CCC	Leu2448Pro	7343T>C	L2448P	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210941	GGC-GAC	Gly2459Asp	7376G>A	G2459D	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM012641	CCG-CTG	Pro2471Leu	7412C>T	P2471L	Polycystic kidney disease 1	Bouba (2001) <i>Eur J Hum Genet</i> 9:677	PubMed 11571556
PKD1	CM1210942	TGC-AGC	Cys2476Ser	7426T>A	C2476S	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239 Zhang (2006) <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i> 23:283	PubMed 22508176 PubMed 16767665
PKD1	CM068424	CGC-CAC	Arg2477His	7430G>A	R2477H	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210943	TGC-CGC	Cys2495Arg	7483T>C	C2495R	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM121782	TGG-TGT	Trp2498Cys	7494G>T	W2498C	Polycystic kidney disease 1	Neumann (2012) <i>Int Urol Nephrol</i> 44:1753	PubMed 22367170
PKD1	CM074429	CGC-TGC	Arg2516Cys	7546C>T	R2516C	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM012642	CAG-CTG	Gln2519Leu	7556A>T	Q2519L	Polycystic kidney disease 1	Bouba (2001) <i>Eur J Hum Genet</i> 9:677	PubMed 11571556
PKD1	CM1210886	CAG-TAG	Gln2519Term	7555C>T	Q2519*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076387	GAG-TAG	Glu2523Term	7567G>T	E2523*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210887	TAC-TAG	Tyr2528Term	7584C>G	Y2528*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM108790	CAC-TAC	His2546Tyr	7636C>T	H2546Y	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM074425	CAG-TAG	Gln2556Term	7666C>T	Q2556*	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM003337	CAG-TAG	Gln2558Term	7672C>T	Q2558*	Polycystic kidney disease 1	Phakdeekitcharoen (2000) <i>Kidney Int</i> 58:1400	PubMed 11012875
PKD1	CM108794	TCT-TGT	Ser2569Cys	7706C>G	S2569C	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM1210888	TAC-TAG	Tyr2611Term	7833C>G	Y2611*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM074424	CTG-CCG	Leu2619Pro	7856T>C	L2619P	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210889	TAC-TAA	Tyr2622Term	7866C>A	Y2622*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM119594	CAG-TAG	Gln2637Term	7909C>T	Q2637*	Polycystic kidney disease 1	Irazabal (2011) <i>Clin J Am Soc Nephrol</i> 6:1274	PubMed 21551026
PKD1	CM001305	CGA-TGA	Arg2639Term	7915C>T	R2639*	Polycystic kidney disease 1	Perrichot (2000) <i>Eur J Hum Genet</i> 8:353	PubMed 10854095

PKD1	CM074416	CGC-TGC	Arg2643Cys	7927C>T	R2643C	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM108795	ATC-ACC	Ile2646Thr	7937T>C	I2646T	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM012643	ACT-ATT	Thr2649Ile	7946C>T	T2649I	Polycystic kidney disease 1	Bouba (2001) <i>Eur J Hum Genet</i> 9:677	PubMed 11571556
PKD1	CM1210890	CAG-TAG	Gln2663Term	7987C>T	Q2663*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210891	CAG-TAG	Gln2670Term	8008C>T	Q2670*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM074426	CAG-TAG	Gln2686Term	8056C>T	Q2686*	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM014074	CTC-CGC	Leu2696Arg	8087T>G	L2696R	Polycystic kidney disease 1	Phakdeekitcharoen (2001) <i>J Am Soc Nephrol</i> 12:955	PubMed 11316854
PKD1	CM090560	CAG-TAG	Gln2699Term	8095C>T	Q2699*	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	CM1210892	CAG-TAG	Gln2735Term	8203C>T	Q2735*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM010384	GCC-GAC	Ala2752Asp	8255C>A	A2752D	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1111443	TAC-TAG	Tyr2753Term	8259C>G	Y2753*	Polycystic kidney disease 1	Bergmann (2011) <i>J Am Soc Nephrol</i> 22:2047	PubMed 22034641
PKD1	CM971181	CTC-GTC	Leu2763Val	8287C>G	L2763V	Polycystic kidney disease 1	Watnick (1997) <i>Hum Mol Genet</i> 6:1473	PubMed 9285784
PKD1	CM092156	CGC-TGC	Arg2765Cys	8293C>T	R2765C	Polycystic kidney disease 1	Rossetti (2009) <i>Kidney Int</i> 75:848	PubMed 19165178
PKD1	CM074419	CGC-TGC	Arg2767Cys	8299C>T	R2767C	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210918	CGC-CCC	Arg2767Pro	8300G>C	R2767P	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM010385	GTG-ATG	Val2768Met	8302G>A	V2768M	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1210919	AAC-ATC	Asn2770Ile	8309A>T	N2770I	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM010386	GAG-AAG	Glu2771Lys	8311G>A	E2771K	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1210920	CTG-CCG	Leu2774Pro	8321T>C	L2774P	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210893	CAG-TAG	Gln2784Term	8350C>T	Q2784*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM020489	GGC-GAC	Gly2785Asp	8354G>A	G2785D	Polycystic kidney disease 1	McCluskey (2002) <i>Hum Mutat</i> 19:240	PubMed 11857740
PKD1	CM971183	CGG-CAG	Arg2791Gln	8372G>A	R2791Q	Polycystic kidney disease 1	Watnick (1997) <i>Hum Mol Genet</i> 6:1473	PubMed 9285784
PKD1	CM034119	CCT-CTT	Pro2802Leu	8405C>T	P2802L	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM076386	CCC-CTC	Pro2809Leu	8426C>T	P2809L	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM074422	GAG-TAG	Glu2810Term	8428G>T	E2810*	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM010387	CTG-CCG	Leu2816Pro	8447T>C	L2816P	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1212764	CAG-TAG	Gln2824Term	8470C>T	Q2824*	Polycystic kidney disease 1	Laley (2012) <i>Genet Couns</i> 23:435	PubMed 23431742
PKD1	CM971184	ATC-ACC	Ile2826Thr	8477T>C	I2826T	Polycystic kidney disease 1	Watnick (1997) <i>Hum Mol Genet</i> 6:1473	PubMed 9285784
PKD1	CM074437	TTC-TCC	Phe2853Ser	8558T>C	F2853S	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210894	CAG-TAG	Gln2854Term	8560C>T	Q2854*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1312929	CAG-TAG	Gln2856Term	8566C>T	Q2856*	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM010388	GGC-AGC	Gly2858Ser	8572G>A	G2858S	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM076396	CTG-CCG	Leu2866Pro	8597T>C	L2866P	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210895	TCA-TAA	Ser2868Term	8603C>A	S2868*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM108796	AGC-AGG	Ser2889Arg	8667C>G	S2889R	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM971185	CAG-TAG	Gln2900Term	8698C>T	Q2900*	Polycystic kidney disease 1	Roelfsema (1997) <i>Am J Hum Genet</i> 61:1044	PubMed 9345095
PKD1	CM1111448	GGT-AGT	Gly2906Ser	8716G>A	G2906S	Polycystic kidney disease 1	Bergmann (2011) <i>J Am Soc Nephrol</i> 22:2047	PubMed 22034641
PKD1	CM002089	CAT-CCT	His2921Pro	8762A>C	H2921P	Polycystic kidney disease 1	Koptides (2000) <i>Hum Mutat</i> 16:176	PubMed 10923040
PKD1	CM1210896	TAC-TAG	Tyr2933Term	8799C>G	Y2933*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM108817	TAC-TAA	Tyr2941Term	8823C>A	Y2941*	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM003338	GAG-GAC	Glu2966Asp	8898G>C	E2966D	Polycystic kidney disease 1	Afzal (2000) <i>Genet Test</i> 4:365	PubMed 11216660
PKD1	CM021123	CAG-TAG	Gln2969Term	8905C>T	Q2969*	Polycystic kidney disease 1	Inoue (2002) <i>Hum Mutat</i> 19:622	PubMed 12007219
PKD1	CM1210944	ACC-ATC	Thr2977Ile	8930C>T	T2977I	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176

PKD1	CM014075	AGA-GGA	Arg2985Gly	8953A>G	R2985G	Polycystic kidney disease 1	Phakdeekitcharoen (2001) <i>J Am Soc Nephrol</i> 12:955	PubMed 11316854
PKD1	CM022662	TAC-TAG	Tyr2991Term	8973C>G	Y2991*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM971186	CTG-CCG	Leu2993Pro	8978T>C	L2993P	Polycystic kidney disease 1	Peral (1997) <i>Am J Hum Genet</i> 60:1399	PubMed 9199561
PKD1	CM1210921	CTC-CGC	Leu2995Arg	8984T>G	L2995R	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM014076	TGG-TAG	Trp3001Term	9002G>A	W3001*	Polycystic kidney disease 1	Phakdeekitcharoen (2001) <i>J Am Soc Nephrol</i> 12:955	PubMed 11316854
PKD1	CM076413	TCC-CCC	Ser3007Pro	9019T>C	S3007P	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM971187	GTG-CTG	Val3008Leu	9022G>C	V3008L	Polycystic kidney disease 1	Watnick (1997) <i>Hum Mol Genet</i> 6:1473	PubMed 9285784
PKD1	CM971188	CAG-CGG	Gln3016Arg	9047A>G	Q3016R	Polycystic kidney disease 1	Peral (1997) <i>Am J Hum Genet</i> 60:1399	PubMed 9199561
PKD1	CM971189	GAG-TAG	Glu3020Term	9058G>T	E3020*	Polycystic kidney disease 1	Peral (1997) <i>Am J Hum Genet</i> 60:1399	PubMed 9199561
PKD1	CM076381	ACC-ATC	Thr3036Ile	9107C>T	T3036I	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM014077	CGC-TGC	Arg3039Cys	9115C>T	R3039C	Polycystic kidney disease 1	Phakdeekitcharoen (2001) <i>J Am Soc Nephrol</i> 12:955	PubMed 11316854
PKD1	CM1210945	GGC-CGC	Gly3052Arg	9154G>C	G3052R	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1312942	GCC-ACC	Ala3053Thr	9157G>A	A3053T	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM1111439	AAC-AAG	Asn3074Lys	9222C>G	N3074K	Polycystic kidney disease 1	Bergmann (2011) <i>J Am Soc Nephrol</i> 22:2047	PubMed 22034641
PKD1	CM092155	CGG-TGG	Arg3105Trp	9313C>T	R3105W	Polycystic kidney disease 1	Rossetti (2009) <i>Kidney Int</i> 75:848	PubMed 19165178
PKD1	CM1210922	ACA-ATA	Thr3126Ile	9377C>T	T3126I	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM121783	GGT-GAT	Gly3133Asp	9398G>A	G3133D	Polycystic kidney disease 1	Neumann (2012) <i>Int Urol Nephrol</i> 44:1753	PubMed 22367170
PKD1	CM053380	GTG-ATG	Val3138Met	9412G>A	V3138M	Polycystic kidney disease 1	Peltola (2005) <i>J Mol Med</i> 83:638	PubMed 15772804
PKD1	CM1210923	GGG-GAG	Gly3144Glu	9431G>A	G3144E	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210946	GGG-TGG	Gly3144Trp	9430G>T	G3144W	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM108791	CTG-CCG	Leu3154Pro	9461T>C	L3154P	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM1111442	CGC-TGC	Arg3162Cys	9484C>T	R3162C	Polycystic kidney disease 1	Bergmann (2011) <i>J Am Soc Nephrol</i> 22:2047	PubMed 22034641
PKD1	CM1210947	CGC-CTC	Arg3162Leu	9485G>T	R3162L	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM022663	ATC-TTC	Ile3167Phe	9499A>T	I3167F	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM076433	TTC-TTG	Phe3168Leu	9504C>G	F3168L	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM076389	GCC-CCC	Ala3171Pro	9511G>C	A3171P	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM010389	TGG-TGA	Trp3180Term	9540G>A	W3180*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM1210924	AAG-AAC	Lys3181Asn	9543G>C	K3181N	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210897	CGA-TGA	Arg3183Term	9547C>T	R3183*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210898	TGG-TGA	Trp3185Term	9555G>A	W3185*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076401	CAC-TAC	His3186Tyr	9556C>T	H3186Y	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM092154	AAC-AGC	Asn3188Ser	9563A>G	N3188S	Polycystic kidney disease 1	Rossetti (2009) <i>Kidney Int</i> 75:848	PubMed 19165178
PKD1	CM1210925	CCT-CTT	Pro3193Leu	9578C>T	P3193L	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1312943	TGG-CGG	Trp3195Arg	9583T>C	W3195R	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM1210899	CAG-TAG	Gln3198Term	9592C>T	Q3198*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM001306	CAG-TAG	Gln3206Term	9616C>T	Q3206*	Polycystic kidney disease 1	Perrichot (2000) <i>Eur J Hum Genet</i> 8:353	PubMed 10854095
PKD1	CM076426	TGG-TGA	Trp3218Term	9654G>A	W3218*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM003339	CGC-CAC	Arg3247His	9740G>A	R3247H	Polycystic kidney disease 1	Afzal (2000) <i>Genet Test</i> 4:365	PubMed 11216660
PKD1	CM092153	CGC-TGC	Arg3277Cys	9829C>T	R3277C	Polycystic kidney disease 1	Rossetti (2009) <i>Kidney Int</i> 75:848	PubMed 19165178
PKD1	CM014078	GTT-ATT	Val3285Ile	9853G>A	V3285I	Polycystic kidney disease 1	Phakdeekitcharoen (2001) <i>J Am Soc Nephrol</i> 12:955	PubMed 11316854

PKD1	CM074414	AAC-AAA	Asn3295Lys	9885C>A	N3295K	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1210900	TGG-TAG	Trp3298Term	9893G>A	W3298*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM074436	GGC-CGC	Gly3326Arg	9976G>C	G3326R	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM1312944	CGG-CAG	Arg3348Gln	10043G>A	R3348Q	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM034120	CCG-CTG	Pro3355Leu	10064C>T	P3355L	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM1210901	CAG-TAG	Gln3362Term	10084C>T	Q3362*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM057346	GAC-AAC	Asp3368Asn	10102G>A	D3368N	Polycystic kidney disease 1	Zhang (2005) <i>Nephron Exp Nephrol</i> 100:e63	PubMed 15775720
PKD1	CM981543	GTG-ATG	Val3375Met	10123G>A	V3375M	Polycystic kidney disease 1	Koptides (1998) <i>Hum Genet</i> 103:709	PubMed 9921908
PKD1	CM003340	ACG-ATG	Thr3382Met	10145C>T	T3382M	Polycystic kidney disease 1	Afzal (2000) <i>Genet Test</i> 4:365	PubMed 11216660
PKD1	CM053383	TCA-TGA	Ser3384Term	10151C>G	S3384*	Polycystic kidney disease 1	Peltola (2005) <i>J Mol Med</i> 83:638	PubMed 15772804
PKD1	CM1210902	CAG-TAG	Gln3390Term	10168C>T	Q3390*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM010390	CAG-TAG	Gln3395Term	10183C>T	Q3395*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM076427	TGG-TGA	Trp3420Term	10260G>A	W3420*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1312945	CGG-TGG	Arg3439Trp	10315C>T	R3439W	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM076419	TCA-TGA	Ser3468Term	10403C>G	S3468*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM022664	CAG-TAG	Gln3474Term	10420C>T	Q3474*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM992200	CAG-TAG	Gln3475Term	10423C>T	Q3475*	Polycystic kidney disease 1	Perrichot (1999) <i>Hum Genet</i> 105:231	PubMed 10987650
PKD1	CM094919	CAA-TAA	Gln3488Term	10462C>T	Q3488*	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM013956	ACG-ATG	Thr3510Met	10529C>T	T3510M	Polycystic kidney disease 1	Mizoguchi (2001) <i>J Hum Genet</i> 46:511	PubMed 11558899
PKD1	CM971191	CTG-GTG	Leu3511Val	10531C>G	L3511V	Polycystic kidney disease 1	Peral (1997) <i>Am J Hum Genet</i> 60:1399	PubMed 9199561
PKD1	CM971192	CAG-TAG	Gln3514Term	10540C>T	Q3514*	Polycystic kidney disease 1	Peral (1997) <i>Am J Hum Genet</i> 60:1399	PubMed 9199561
PKD1	CM090561	CAG-TAG	Gln3530Term	10588C>T	Q3530*	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	CM057349	TCC-TTC	Ser3556Phe	10667C>T	S3556F	Polycystic kidney disease 1	Zhang (2005) <i>Nephron Exp Nephrol</i> 100:e63	PubMed 15775720
PKD1	CM014740	GGG-AGG	Gly3560Arg	10678G>A	G3560R	Polycystic kidney disease 1	Tsuchiya (2001) <i>Mutat Res</i> 458:77	PubMed 11691639
PKD1	CM1210903	TCA-TGA	Ser3573Term	10718C>G	S3573*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM053384	TGG-TGA	Trp3575Term	10725G>A	W3575*	Polycystic kidney disease 1	Peltola (2005) <i>J Mol Med</i> 83:638	PubMed 15772804
PKD1	CM1210904	TCA-TAA	Ser3599Term	10796C>A	S3599*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM021655	GGC-AGC	Gly3602Ser	10804G>A	G3602S	Polycystic kidney disease 1	Eo (2002) <i>Clin Genet</i> 62:169	PubMed 12220456
PKD1	CM109265	TGG-CGG	Trp3603Arg	10807T>C	W3603R	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	BM1135887	TGG-TAG	Trp3603Term	10808G>A	W3603*	Polycystic kidney disease 1	Irazabal (2011) <i>Clin J Am Soc Nephrol</i> 6:1274	PubMed 21551026
PKD1	CM076408	GAG-AAG	Glu3604Lys	10810G>A	E3604K	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM094920	CTG-CCG	Leu3617Pro	10850T>C	L3617P	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM961116	GAG-GAC	Glu3632Asp	10896G>C	E3632D	Polycystic kidney disease 1	Peral (1996) <i>Am J Hum Genet</i> 58:86	PubMed 8554072
PKD1	CM021656	CCC-CTC	Pro3649Leu	10946C>T	P3649L	Polycystic kidney disease 1	Ding (2002) <i>Nephrol Dial Transplant</i> 17:75	PubMed 11773467
PKD1	CM1210948	GGC-GAC	Gly3651Asp	10952G>A	G3651D	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM057347	GGC-AGC	Gly3651Ser	10951G>A	G3651S	Polycystic kidney disease 1	Zhang (2005) <i>Nephron Exp Nephrol</i> 100:e63	PubMed 15775720
PKD1	CM1210926	GCA-GTA	Ala3653Val	10958C>T	A3653V	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210949	GCC-GAC	Ala3657Asp	10970C>A	A3657D	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM971193	ATG-ACG	Met3678Thr	11033T>C	M3678T	Polycystic kidney disease 1	Turco (1997) <i>Hum Mutat</i> 10:164	PubMed 9259200
PKD1	CM119595	CTG-CAG	Leu3682Gln	11045T>A	L3682Q	Polycystic kidney disease 1	Irazabal (2011) <i>Clin J Am Soc Nephrol</i> 6:1274	PubMed 21551026
PKD1	CM034121	CTG-CCG	Leu3682Pro	11045T>C	L3682P	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM094921	TCA-TTA	Ser3693Leu	11078C>T	S3693L	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM094922	TGC-CGC	Cys3694Arg	11080T>C	C3694R	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598

PKD1	CM013957	TGC-TGA	Cys3694Term	11082C>A	C3694*	Polycystic kidney disease 1	Mizoguchi (2001) <i>J Hum Genet</i> 46:511	PubMed 11558899
PKD1	CM121784	TAC-TCC	Tyr3699Ser	11096A>C	Y3699S	Polycystic kidney disease 1	Neumann (2012) <i>Int Urol Nephrol</i> 44:1753	PubMed 22367170
PKD1	CM021657	CAA-TAA	Gln3702Term	11104C>T	Q3702*	Polycystic kidney disease 1	Eo (2002) <i>Clin Genet</i> 62:169	PubMed 12220456
PKD1	CM003044	CGG-CAG	Arg3719Gln	11156G>A	R3719Q	Polycystic kidney disease 1	Aguiari (2000) <i>Hum Mutat</i> 16:444	PubMed 11058904
PKD1	CM085996	TGG-CGG	Trp3726Arg	11176T>C	W3726R	Polycystic kidney disease 1	Reed (2008) <i>Am J Kidney Dis</i> 52:1042	PubMed 18640754
PKD1	CM090557	TGG-TCG	Trp3726Ser	11177G>C	W3726S	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	CM109266	CGG-CAG	Arg3750Gln	11249G>A	R3750Q	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM034122	CAG-CGG	Gln3751Arg	11252A>G	Q3751R	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM034123	CAG-TAG	Gln3751Term	11251C>T	Q3751*	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM076390	CGG-CAG	Arg3753Gln	11258G>A	R3753Q	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM003893	CGG-TGG	Arg3753Trp	11257C>T	R3753W	Polycystic kidney disease 1	Kim (2000) <i>Mutat Res</i> 432:39	PubMed 10729710
PKD1	CM022665	TAC-TAG	Tyr3781Term	11343C>G	Y3781*	Polycystic kidney disease 1	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD1	CM034563	TGG-TGA	Trp3785Term	11355G>A	W3785*	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM985521	TGG-TAG	Trp3795Term	11384G>A	W3795*	Polycystic kidney disease 1	Neophytou (1998) <i>Balkan J Med Genet</i> 1:149	PubMed: No PubMed ID
PKD1	CM094923	TGG-TAG	Trp3806Term	11417G>A	W3806*	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM108818	TGG-TAG	Trp3808Term	11423G>A	W3808*	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM003894	GAC-AAC	Asp3815Asn	11443G>A	D3815N	Polycystic kidney disease 1	Kim (2000) <i>Mutat Res</i> 432:39	PubMed 10729710
PKD1	CM961117	TAC-TAA	Tyr3819Term	11457C>A	Y3819*	Polycystic kidney disease 1	Peral (1996) <i>Hum Mol Genet</i> 5:539	PubMed 8845849
PKD1	CM001307	CAG-TAG	Gln3821Term	11461C>T	Q3821*	Polycystic kidney disease 1	Perrichot (2000) <i>Eur J Hum Genet</i> 8:353	PubMed 10854095
PKD1	CM1210950	CTG-CCG	Leu3823Pro	11468T>C	L3823P	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM1210905	GAG-TAG	Glu3828Term	11482G>T	E3828*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076399	CTG-CGG	Leu3834Arg	11501T>G	L3834R	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM961118	CAG-TAG	Gln3838Term	11512C>T	Q3838*	Polycystic kidney disease 1	Peral (1996) <i>Am J Hum Genet</i> 58:86	PubMed 8554072
PKD1	CM076423	TGG-TAG	Trp3842Term	11525G>A	W3842*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM003045	CTG-CCG	Leu3852Pro	11555T>C	L3852P	Polycystic kidney disease 1	Aguiari (2000) <i>Hum Mutat</i> 16:444	PubMed 11058904
PKD1	CM1210927	GAG-CAG	Glu3872Gln	11614G>C	E3872Q	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM090564	GAG-TAG	Glu3872Term	11614G>T	E3872*	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	HM070038	TCG-TAG	Ser3898Term	11693C>A	S3898*	Polycystic kidney disease 1	Li (2007) <i>Hum Genet</i> 122:413	Source: Online submission
PKD1	CM076400	CTG-CGG	Leu3908Arg	11723T>G	L3908R	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM094924	GAG-TAG	Glu3918Term	11752G>T	E3918*	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM076428	TGG-TGA	Trp3922Term	11766G>A	W3922*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM1210906	TGG-TAG	Trp3928Term	11783G>A	W3928*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM053379	GCC-CCC	Ala3954Pro	11860G>C	A3954P	Polycystic kidney disease 1	Peltola (2005) <i>J Mol Med</i> 83:638	PubMed 15772804
PKD1	CM076385	CAG-TAG	Gln3955Term	11863C>T	Q3955*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM108819	CAG-TAG	Gln3979Term	11935C>T	Q3979*	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM076405	CAG-TAG	Gln3982Term	11944C>T	Q3982*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM066172	CAG-TAG	Gln4004Term	12010C>T	Q4004*	Polycystic kidney disease 1	Gao (2006) <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i> 23:23	PubMed 16456780
PKD1	CM994745	CAG-TAG	Gln4005Term	12013C>T	Q4005*	Polycystic kidney disease 1	Iglesias (1999) <i>Medicina (B Aires)</i> 59:133	PubMed 10413889
PKD1	CM981544	CAG-TAG	Gln4011Term	12031C>T	Q4011*	Polycystic kidney disease 1	Daniells (1998) <i>Hum Genet</i> 102:216	PubMed 9521593
PKD1	CM1210907	TGG-TAG	Trp4012Term	12035G>A	W4012*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM971194	TGG-TGA	Trp4012Term	12036G>A	W4012*	Polycystic kidney disease 1	Roelfsema (1997) <i>Am J Hum Genet</i> 61:1044	PubMed 9345095

PKD1	CM094925	TTA-TGA	Leu4019Term	12056T>G	L4019*	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM961119	CGA-TGA	Arg4021Term	12061C>T	R4021*	Polycystic kidney disease 1	Rossetti (1996) <i>Am J Med Genet</i> 65:155	PubMed 8911610
PKD1	CM981545	GAG-TAG	Glu4025Term	12073G>T	E4025*	Polycystic kidney disease 1	Daniells (1998) <i>Hum Genet</i> 102:216	PubMed 9521593
PKD1	CM981546	GGC-GAC	Gly4032Asp	12095G>A	G4032D	Polycystic kidney disease 1	Daniells (1998) <i>Hum Genet</i> 102:216	PubMed 9521593
PKD1	CM010391	TAC-TAA	Tyr4040Term	12120C>A	Y4040*	Polycystic kidney disease 1	Rossetti (2001) <i>Am J Hum Genet</i> 68:46	PubMed 11115377
PKD1	CM950939	CAG-TAG	Gln4042Term	12124C>T	Q4042*	Polycystic kidney disease 1	Turco (1995) <i>Hum Mol Genet</i> 4:1331	PubMed 7581371
PKD1	CM034564	TGG-TAG	Trp4056Term	12167G>A	W4056*	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	HM971242	CAG-TAG	Gln4060Term	12178C>T	Q4060*	Polycystic kidney disease 1	Daniells (1998) <i>Hum Genet</i> 102:127	submission
PKD1	CM1210908	TGG-TAG	Trp4080Term	12239G>A	W4080*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM961120	TGT-TGA	Cys4087Term	12261T>A	C4087*	Polycystic kidney disease 1	Neophytou (1996) <i>Hum Genet</i> 98:437	PubMed 8792818
PKD1	CM1210909	TGG-TAG	Trp4096Term	12287G>A	W4096*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	HM971243	CAG-TAG	Gln4125Term	12373C>T	Q4125*	Polycystic kidney disease 1	Daniells (1998) <i>Hum Genet</i> 102:127	submission
PKD1	CM971195	TAC-TAG	Tyr4127Term	12381C>G	Y4127*	Polycystic kidney disease 1	Turco (1997) <i>Hum Mutat</i> 10:164	PubMed 9259200
PKD1	CM034124	GAG-TAG	Glu4128Term	12382G>T	E4128*	Polycystic kidney disease 1	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD1	CM1312930	GAG-TAG	Glu4131Term	12391G>T	E4131*	Polycystic kidney disease 1	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD1	CM992201	AGG-GGG	Arg4136Gly	12406A>G	R4136G	Polycystic kidney disease 1	Perrichot (1999) <i>Hum Genet</i> 105:231	PubMed 10987650
PKD1	CM076397	CTG-CCG	Leu4137Pro	12410T>C	L4137P	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM076394	CTC-CCC	Leu4139Pro	12416T>C	L4139P	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM992202	TGG-TGA	Trp4140Term	12420G>A	W4140*	Polycystic kidney disease 1	Perrichot (1999) <i>Hum Genet</i> 105:231	PubMed 10987650
PKD1	CM074420	CGC-TGC	Arg4150Cys	12448C>T	R4150C	Polycystic kidney disease 1	Garcia-Gonzalez (2007) <i>Mol Genet Metab</i> 92:160	PubMed 17574468
PKD1	CM992203	CGC-TGC	Arg4154Cys	12460C>T	R4154C	Polycystic kidney disease 1	Perrichot (1999) <i>Hum Genet</i> 105:231	PubMed 10987650
PKD1	CM090558	TTT-GTT	Phe4155Val	12463T>G	F4155V	Polycystic kidney disease 1	Tan (2009) <i>Hum Mutat</i> 30:264	PubMed 18837007
PKD1	CM1210910	TGT-TGA	Cys4208Term	12624T>A	C4208*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM094926	CAA-CAC	Gln4216His	12648A>C	Q4216H	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM1210911	CAA-TAA	Gln4216Term	12646C>T	Q4216*	Polycystic kidney disease 1	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD1	CM076388	GAG-TAG	Glu4220Term	12658G>T	E4220*	Polycystic kidney disease 1	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD1	CM993191	CAG-CCG	Gln4225Pro	12674A>C	Q4225P	Polycystic kidney disease 1	Badenas (1999) <i>Kidney Int</i> 55:1225	PubMed 10200984
PKD1	CM961121	CGA-TGA	Arg4228Term	12682C>T	R4228*	Polycystic kidney disease 1	Peral (1996) <i>Am J Hum Genet</i> 58:86	PubMed 8554072
PKD1	CM094927	CAG-TAG	Gln4231Term	12691C>T	Q4231*	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM003948	TAC-TAG	Tyr4237Term	12711C>G	Y4237*	Polycystic kidney disease 1	Perrichot (2000) <i>Hum Mutat</i> 15:582	PubMed 10862097
PKD1	CM108820	CAG-TAG	Gln4241Term	12721C>T	Q4241*	Polycystic kidney disease 1	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD1	CM094928	CAG-TAG	Gln4242Term	12724C>T	Q4242*	Polycystic kidney disease 1	Stekrova (2009) <i>BMC Med Genet</i> 10:78	PubMed 19686598
PKD1	CM021658	CCC-TCC	Pro4255Ser	12763C>T	P4255S	Polycystic kidney disease 1	Eo (2002) <i>Clin Genet</i> 62:169	PubMed 12220456
PKD1	CM993192	CGG-TGG	Arg4276Trp	12826C>T	R4276W	Polycystic kidney disease 1	Badenas (1999) <i>Kidney Int</i> 55:1225	PubMed 10200984
PKD1	CM1313864	GAT-GCT	Asp97Ala	290A>C	D97A	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM123995	AAC-GAC	Asn125Asp	373A>G	N125D	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	BM1366604	TGT-TTT	Cys129Phe	386G>T	C129F	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	BM1349690	TGT-AGT	Cys155Ser	463T>A	C155S	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259

PKD1	CM124001	TGC-TAC	Cys210Tyr	629G>A	C210Y	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1314577	TCG-TTG	Ser225Leu	674C>T	S225L	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1314578	TGT-TAT	Cys232Tyr	695G>A	C232Y	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1313865	CCG-CGG	Pro297Arg	890C>G	P297R	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313866	GTG-ATG	Val333Met	997G>A	V333M	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313867	TGC-TGA	Cys363Term	1089C>A	C363*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313868	CAG-TAG	Gln378Term	1132C>T	Q378*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM123999	TAC-TCC	Tyr420Ser	1259A>C	Y420S	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM124017	GTC-GCC	Val460Ala	1379T>C	V460A	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM124012	TCG-TTG	Ser471Leu	1412C>T	S471L	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1314579	GGG-TGG	Gly515Trp	1543G>T	G515W	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1313869	TCA-TGA	Ser523Term	1568C>G	S523*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	BM1377218	TCC-TGC	Ser629Cys	1886C>G	S629C	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313870	CAG-TAG	Gln663Term	1987C>T	Q663*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313871	GGG-GAG	Gly693Glu	2078G>A	G693E	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM124003	TAT-TGT	Tyr796Cys	2387A>G	Y796C	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313872	GTG-CTG	Val909Leu	2725G>C	V909L	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313873	GGC-AGC	Gly960Ser	2878G>A	G960S	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313874	CGG-CAG	Arg966Gln	2897G>A	R966Q	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313875	GTC-GAC	Val984Asp	2951T>A	V984D	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313876	AAG-TAG	Lys993Term	2977A>T	K993*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313877	CAG-TAG	Gln1116Term	3346C>T	Q1116*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313878	TCG-TAG	Ser1183Term	3548C>A	S1183*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM124021	AGC-AGA	Ser1278Arg	3834C>A	S1278R	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM141747	GGG-AGG	Gly1319Arg	3955G>A	G1319R	Polycystic kidney disease, autosomal dominant	Yang (2014) <i>Clin Chim Acta</i> epub:epub	PubMed 24582653

PKD1	CM124004	TGG-CGG	Trp1328Arg	3982T>C	W1328R	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313879	TAC-TAG	Tyr1370Term	4110C>G	Y1370*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM124005	TGC-TAC	Cys1375Tyr	4124G>A	C1375Y	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM123946	CAG-TAG	Gln1482Term	4444C>T	Q1482*	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM123990	GGG-AGG	Gly1503Arg	4507G>A	G1503R	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1110583	GTG-ATG	Val1604Met	4810G>A	V1604M	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM1313880	ATC-ACC	Ile1610Thr	4829T>C	I1610T	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM123971	CAG-TAG	Gln1621Term	4861C>T	Q1621*	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313881	GAG-GGG	Glu1633Gly	4898A>G	E1633G	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313882	GTT-ATT	Val1656Ile	4966G>A	V1656I	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM124000	GCC-GAC	Ala1699Asp	5096C>A	A1699D	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313883	CAG-TAG	Gln1903Term	5707C>T	Q1903*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1110561	CAG-TAG	Gln1908Term	5722C>T	Q1908*	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM124018	GGC-GCC	Gly1914Ala	5741G>C	G1914A	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1314580	TGC-TAC	Cys1979Tyr	5936G>A	C1979Y	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1314581	GGC-GAC	Gly1987Asp	5960G>A	G1987D	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1314582	CGC-TGC	Arg1995Cys	5983C>T	R1995C	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1314583	TCT-CCT	Ser2000Pro	5998T>C	S2000P	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1313884	TTT-TGT	Phe2096Cys	6287T>G	F2096C	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313886	TAC-TAA	Tyr2114Term	6342C>A	Y2114*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1314584	AAC-CAC	Asn2128His	6382A>C	N2128H	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1313885	CAG-CGG	Gln2136Arg	6407A>G	Q2136R	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1110562	CAG-TAG	Gln2142Term	6424C>T	Q2142*	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM1313887	AAC-GAC	Asn2167Asp	6499A>G	N2167D	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313888	CGG-TGG	Arg2215Trp	6643C>T	R2215W	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259

PKD1	CM1314586	CTG-CAG	Leu2216Gln	6647T>A	L2216Q	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM124022	CTG-CAG	Leu2417Gln	7250T>A	L2417Q	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM124019	CGG-CAG	Arg2434Gln	7301G>A	R2434Q	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM124016	CCC-CTC	Pro2467Leu	7400C>T	P2467L	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM124011	GGC-GAC	Gly2530Asp	7589G>A	G2530D	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM123947	CAG-TAG	Gln2602Term	7804C>T	Q2602*	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM124020	TCG-TGG	Ser2612Trp	7835C>G	S2612W	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313889	GGA-AGA	Gly2721Arg	8161G>A	G2721R	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1110563	TAT-TAA	Tyr2796Term	8388T>A	Y2796*	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM124015	CTG-CAG	Leu2816Gln	8447T>A	L2816Q	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1314587	ACC-ATC	Thr2846Ile	8537C>T	T2846I	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM124006	ATC-AGC	Ile2872Ser	8615T>G	I2872S	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313890	GTC-TTC	Val2944Phe	8830G>T	V2944F	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM123970	TCG-TAG	Ser2948Term	8843C>A	S2948*	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1314588	TTC-TGC	Phe2978Cys	8933T>G	F2978C	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1313891	TAC-TAA	Tyr2991Term	8973C>A	Y2991*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM123994	TGT-CGT	Cys3081Arg	9241T>C	C3081R	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313892	CAG-TAG	Gln3100Term	9298C>T	Q3100*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1110582	TGT-TTT	Cys3112Phe	9335G>T	C3112F	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM1313894	CAG-TAG	Gln3114Term	9340C>T	Q3114*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1314589	GAG-AAG	Glu3121Lys	9361G>A	E3121K	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1110581	CGG-TGG	Arg3130Trp	9388C>T	R3130W	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM1314590	TCA-TTA	Ser3132Leu	9395C>T	S3132L	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM124014	AGC-ATC	Ser3178Ile	9533G>T	S3178I	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313893	GTG-ATG	Val3179Met	9535G>A	V3179M	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259

PKD1	CM124013	GAC-GAG	Asp3187Glu	9561C>G	D3187E	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1314591	CCT-TCT	Pro3193Ser	9577C>T	P3193S	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1313895	TGG-TGA	Trp3195Term	9585G>A	W3195*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM123996	GAG-GTG	Glu3233Val	9698A>T	E3233V	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM123997	TGG-CGG	Trp3263Arg	9787T>C	W3263R	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1110580	AAC-AGC	Asn3295Ser	9884A>G	N3295S	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM123944	TGG-TGA	Trp3298Term	9894G>A	W3298*	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1314592	GGG-AGG	Gly3300Arg	9898G>A	G3300R	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1313896	GGC-GAC	Gly3326Asp	9977G>A	G3326D	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313897	TAC-CAC	Tyr3337His	10009T>C	Y3337H	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313898	AAG-CAG	Lys3350Gln	10048A>C	K3350Q	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313899	TGG-TAG	Trp3411Term	10232G>A	W3411*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM123993	GAG-AAG	Glu3415Lys	10243G>A	E3415K	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313900	CGC-CCC	Arg3662Pro	10985G>C	R3662P	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313901	TAC-TGC	Tyr3734Cys	11201A>G	Y3734C	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1314593	GTC-GAC	Val3735Asp	11204T>A	V3735D	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1314594	CAG-AAG	Gln3751Lys	11251C>A	Q3751K	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM124002	CGG-CTG	Arg3753Leu	11258G>T	R3753L	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD1	CM1313902	GAA-AAA	Glu3756Lys	11266G>A	E3756K	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1110579	CCT-CTT	Pro3762Leu	11285C>T	P3762L	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM1110565	TGG-TAG	Trp3785Term	11354G>A	W3785*	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD1	CM1313903	TCA-CCA	Ser3798Pro	11392T>C	S3798P	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM141151	GGC-GCC	Gly3818Ala	11453G>C	G3818A	Polycystic kidney disease, autosomal dominant	Paul (2014) <i>Kidney Int</i> 85:383	PubMed 23760289
PKD1	CM1313904	CGC-CTC	Arg3870Leu	11609G>T	R3870L	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313905	CGC-CAC	Arg3892His	11675G>A	R3892H	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259

PKD1	CM1314595	TCG-TGG	Ser3904Trp	11711C>G	S3904W	Polycystic kidney disease, autosomal dominant	Cornec-Le Gall (2013) <i>J Am Soc Nephrol</i> 24:1006	PubMed 23431072
PKD1	CM1313906	TGC-TTC	Cys3906Phe	11717G>T	C3906F	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM141748	TGG-TGA	Trp4122Term	12366G>A	W4122*	Polycystic kidney disease, autosomal dominant	Yang (2014) <i>Clin Chim Acta</i> epub:epub	PubMed 24582653
PKD1	CM1313907	CGC-CCC	Arg4154Pro	12461G>C	R4154P	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD1	CM1313908	CAG-TAG	Gln4225Term	12673C>T	Q4225*	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD2	CM044696	CAG-TAG	Gln49Term	145C>T	Q49*	Polycystic kidney disease 2	Stekrova (2004) <i>Nephrol Dial Transplant</i> 19:1116	PubMed 14993477
PKD2	CM076437	CAG-TAG	Gln85Term	253C>T	Q85*	Polycystic kidney disease 2	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD2	CM127156	GAG-TAG	Glu95Term	283G>T	E95*	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM065386	CGA-TGA	Arg157Term	469C>T	R157*	Polycystic kidney disease 2	Chung (2006) <i>Clin Genet</i> 70:502	PubMed 17100995
PKD2	CM020954	CAG-TAG	Gln160Term	478C>T	Q160*	Polycystic kidney disease 2	Reiterova (2002) <i>Hum Mutat</i> 19:573	PubMed 11968093
PKD2	CM004277	CGA-TGA	Arg186Term	556C>T	R186*	Polycystic kidney disease 2	Hateboer (2000) <i>Kidney Int</i> 57:1444	PubMed 10760080
PKD2	CM085644	TGG-TGA	Trp189Term	567G>A	W189*	Polycystic kidney disease 2	Zhao (2008) <i>Clin J Am Soc Nephrol</i> 3:146 Zhang (2004) <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i> 21:211	PubMed 18077784 PubMed 15192819
PKD2	CM044027	GCG-ACG	Ala190Thr	568G>A	A190T	Polycystic kidney disease 2	Rossetti (2003) <i>Lancet</i> 361:2196	PubMed 12842373
PKD2	CM034565	TGG-TAG	Trp201Term	602G>A	W201*	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM127157	CGA-TGA	Arg213Term	637C>T	R213*	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM119093	GAG-TAG	Glu214Term	640G>T	E214*	Polycystic kidney disease 2	Bataille (2011) <i>BMC Nephrol</i> 12:57	PubMed 22008521
PKD2	CM055478	TAC-TAA	Tyr227Term	681C>A	Y227*	Polycystic kidney disease 2	Chang (2005) <i>Ren Fail</i> 27:95	PubMed 15717641
PKD2	CM065387	GAG-TAG	Glu266Term	796G>T	E266*	Polycystic kidney disease 2	Chung (2006) <i>Clin Genet</i> 70:502	PubMed 17100995
PKD2	CM1210951	CAG-TAG	Gln300Term	898C>T	Q300*	Polycystic kidney disease 2	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD2	CM044695	CGA-CAA	Arg306Gln	917G>A	R306Q	Polycystic kidney disease 2	Stekrova (2004) <i>Nephrol Dial Transplant</i> 19:1116	PubMed 14993477
PKD2	CM971196	CGA-TGA	Arg306Term	916C>T	R306*	Polycystic kidney disease 2	Veldhuisen (1997) <i>Am J Hum Genet</i> 61:547	PubMed 9326320
PKD2	CM994226	CGA-TGA	Arg320Term	958C>T	R320*	Polycystic kidney disease 2	Reynolds (1999) <i>J Am Soc Nephrol</i> 10:2342	PubMed 10541293
PKD2	CM053385	CGG-CAG	Arg322Gln	965G>A	R322Q	Polycystic kidney disease 2	Peltola (2005) <i>J Mol Med</i> 83:638	PubMed 15772804
PKD2	CM020955	CGG-TGG	Arg322Trp	964C>T	R322W	Polycystic kidney disease 2	Reiterova (2002) <i>Hum Mutat</i> 19:573	PubMed 11968093
PKD2	CM076436	CGA-CAA	Arg325Gln	974G>A	R325Q	Polycystic kidney disease 2	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD2	CM076435	CGA-TGA	Arg325Term	973C>T	R325*	Polycystic kidney disease 2	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD2	CM993193	GCT-CCT	Ala356Pro	1066G>C	A356P	Polycystic kidney disease 2	Torra (1999) <i>Kidney Int</i> 56:28	PubMed 10411676
PKD2	CM065385	CGA-TGA	Arg361Term	1081C>T	R361*	Polycystic kidney disease 2	Chung (2006) <i>Clin Genet</i> 70:502	PubMed 17100995
PKD2	CM961122	TGG-TAG	Trp380Term	1139G>A	W380*	Polycystic kidney disease 2	Mochizuki (1996) <i>Science</i> 272:1339	PubMed 8650545
PKD2	CM1210952	TGG-TGA	Trp380Term	1140G>A	W380*	Polycystic kidney disease 2	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD2	CM108799	GCA-CCA	Ala384Pro	1150G>C	A384P	Polycystic kidney disease 2	Hoefele (2010) <i>Nephrol Dial Transplant</i> 26:2181	PubMed 21115670
PKD2	CM993512	TAT-TAA	Tyr386Term	1158T>A	Y386*	Polycystic kidney disease 2	Aguiari (1999) <i>Am J Kidney Dis</i> 33:880	PubMed 10213643
PKD2	CM1312935	GCT-GTT	Ala389Val	1166C>T	A389V	Polycystic kidney disease 2	Chang (2013) <i>J Hum Genet</i> 58:720 Zhang (2004) <i>Zhonghua Yi Xue Yi Chuan Xue Za Zhi</i> 21:211	PubMed 23985799 PubMed 15192819
PKD2	CM044028	GGC-AGC	Gly390Ser	1168G>A	G390S	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM127158	GGC-GTC	Gly390Val	1169G>T	G390V	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM1312936	TAT-GAT	Tyr392Asp	1174T>G	Y392D	Polycystic kidney disease 2	Chang (2013) <i>J Hum Genet</i> 58:720	PubMed 23985799
PKD2	CM961123	CAA-TAA	Gln405Term	1213C>T	Q405*	Polycystic kidney disease 2	Mochizuki (1996) <i>Science</i> 272:1339	PubMed 8650545
PKD2	CM971197	TGG-GGG	Trp414Gly	1240T>G	W414G	Polycystic kidney disease 2	Veldhuisen (1997) <i>Am J Hum Genet</i> 61:547	PubMed 9326320

PKD2	CM981549	CGA-TGA	Arg417Term	1249C>T	R417*	Polycystic kidney disease 2	Pei (1998) <i>J Am Soc Nephrol</i> 9:1853	PubMed 9773786
PKD2	CM065384	ACC-GCC	Thr419Ala	1255A>G	T419A	Polycystic kidney disease 2	Chung (2006) <i>Clin Genet</i> 70:502	PubMed 17100995
PKD2	CM044694	AGG-GGG	Arg420Gly	1258A>G	R420G	Polycystic kidney disease 2	Stekrova (2004) <i>Nephrol Dial Transplant</i> 19:1116	PubMed 14993477
PKD2	CM121801	AGG-AGT	Arg440Ser	1320G>T	R440S	Polycystic kidney disease 2	Neumann (2012) <i>Int Urol Nephrol</i> 44:1753	PubMed 22367170
PKD2	CM065388	TGG-TGA	Trp455Term	1365G>A	W455*	Polycystic kidney disease 2	Chung (2006) <i>Clin Genet</i> 70:502	PubMed 17100995
PKD2	CM971198	CGA-TGA	Arg464Term	1390C>T	R464*	Polycystic kidney disease 2	Viribay (1997) <i>Hum Genet</i> 101:229	PubMed 9402976
PKD2	CM080498	TTC-TGC	Phe482Cys	1445T>G	F482C	Polycystic kidney disease 2	Dedoussis (2008) <i>Eur J Clin Invest</i> 38:180	PubMed 18257781
PKD2	CM971199	GAA-TAA	Glu494Term	1480G>T	E494*	Polycystic kidney disease 2	Veldhuisen (1997) <i>Am J Hum Genet</i> 61:547	PubMed 9326320
PKD2	CM127159	TGG-TGA	Trp507Term	1521G>A	W507*	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM994227	GAT-GTT	Asp511Val	1532A>T	D511V	Polycystic kidney disease 2	Reynolds (1999) <i>J Am Soc Nephrol</i> 10:2342	PubMed 10541293
PKD2	CM034262	CAG-TAG	Gln537Term	1609C>T	Q537*	Polycystic kidney disease 2	Magistroni (2003) <i>J Am Soc Nephrol</i> 14:1164	PubMed 12707387
PKD2	CM971200	CAG-TAG	Gln555Term	1663C>T	Q555*	Polycystic kidney disease 2	Viribay (1997) <i>Hum Genet</i> 101:229	PubMed 9402976
PKD2	CM993194	CAG-TAG	Gln585Term	1753C>T	Q585*	Polycystic kidney disease 2	Torra (1999) <i>Kidney Int</i> 56:28	PubMed 10411676
PKD2	CM1210953	CGA-TGA	Arg592Term	1774C>T	R592*	Polycystic kidney disease 2	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD2	CM127160	CAG-TAG	Gln613Term	1837C>T	Q613*	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM1210954	TAC-TAA	Tyr616Term	1848C>A	Y616*	Polycystic kidney disease 2	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD2	CM1210955	CAA-TAA	Gln630Term	1888C>T	Q630*	Polycystic kidney disease 2	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD2	CM034263	TGT-CGT	Cys632Arg	1894T>C	C632R	Polycystic kidney disease 2	Magistroni (2003) <i>J Am Soc Nephrol</i> 14:1164	PubMed 12707387
PKD2	CM127161	TGT-TAT	Cys632Tyr	1895G>A	C632Y	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM119598	CGA-TGA	Arg654Term	1960C>T	R654*	Polycystic kidney disease 2	Irazabal (2011) <i>Clin J Am Soc Nephrol</i> 6:1274	PubMed 21551026
PKD2	CM1210958	TTT-TCT	Phe664Ser	1991T>C	F664S	Polycystic kidney disease 2	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD2	CM1210956	CAG-TAG	Gln693Term	2077C>T	Q693*	Polycystic kidney disease 2	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD2	CM961124	CGA-TGA	Arg742Term	2224C>T	R742*	Polycystic kidney disease 2	Mochizuki (1996) <i>Science</i> 272:1339	PubMed 8650545
PKD2	CM971201	TAC-TAA	Tyr762Term	2286C>A	Y762*	Polycystic kidney disease 2	Veldhuisen (1997) <i>Am J Hum Genet</i> 61:547	PubMed 9326320
PKD2	CM098191	ATG-CTG	Met800Leu	2398A>C	M800L	Polycystic kidney disease 2	Fraile Gómez (2009) <i>Nefrologia</i> 29:562	PubMed 19936001
PKD2	CM012179	CGA-TGA	Arg803Term	2407C>T	R803*	Polycystic kidney disease 2	Deltas (2001) <i>Hum Mutat</i> 18:13	PubMed 11438989
PKD2	CM076434	AGT-AAT	Ser804Asn	2411G>A	S804N	Polycystic kidney disease 2	Rossetti (2007) <i>J Am Soc Nephrol</i> 18:2143	PubMed 17582161
PKD2	CM034264	CGA-CAA	Arg807Gln	2420G>A	R807Q	Polycystic kidney disease 2	Magistroni (2003) <i>J Am Soc Nephrol</i> 14:1164	PubMed 12707387
PKD2	CM971202	CGA-TGA	Arg807Term	2419C>T	R807*	Polycystic kidney disease 2	Viribay (1997) <i>Hum Genet</i> 101:229	PubMed 9402976
PKD2	CM022666	GAG-TAG	Glu814Term	2440G>T	E814*	Polycystic kidney disease 2	Rossetti (2002) <i>Kidney Int</i> 61:1588	PubMed 11967008
PKD2	CM971203	GAA-TAA	Glu837Term	2509G>T	E837*	Polycystic kidney disease 2	Viribay (1997) <i>Hum Genet</i> 101:229	PubMed 9402976
PKD2	CM003771	CGA-TGA	Arg845Term	2533C>T	R845*	Polycystic kidney disease 2	Torra (2000) <i>Am J Kidney Dis</i> 36:728	PubMed 11007674
PKD2	CM127162	CTA-CCA	Leu867Pro	2600T>C	L867P	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM994295	CGA-TGA	Arg872Term	2614C>T	R872*	Polycystic kidney disease 2	Reynolds (1999) <i>J Am Soc Nephrol</i> 10:2342	PubMed 10541293
PKD2	CM127163	GAT-AAT	Asp919Asn	2755G>A	D919N	Polycystic kidney disease 2	Robinson (2012) <i>BMC Nephrol</i> 13:79	PubMed 22863349
PKD2	CM1210957	TGA-CGA	Term969Arg	2905T>C	*969R	Polycystic kidney disease 2	Audrézet (2012) <i>Hum Mutat</i> 33:1239	PubMed 22508176
PKD2	CM1313856	GTG-ATG	Val262Met	784G>A	V262M	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD2	CM124027	TAC-TAG	Tyr292Term	876C>G	Y292*	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD2	CM1110578	TAT-TGT	Tyr345Cys	1034A>G	Y345C	Polycystic kidney disease, autosomal dominant	Yu (2011) <i>BMC Med Genet</i> 12:164	PubMed 22185115
PKD2	CM1313857	GAA-GAC	Glu444Asp	1332A>C	E444D	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259

PKD2	CM124024	ACA-AAA	Thr448Lys	1343C>A	T448K	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD2	CM1111438	TTC-GTC	Phe482Val	1444T>G	F482V	Polycystic kidney disease, autosomal dominant	Bergmann (2011) <i>J Am Soc Nephrol</i> 22:2047	PubMed 22034641
PKD2	CM1313858	TAT-CAT	Tyr487His	1459T>C	Y487H	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD2	CM124035	TAC-TAG	Tyr836Term	2508C>G	Y836*	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD2	CM1313859	CGA-GGA	Arg872Gly	2614C>G	R872G	Polycystic kidney disease, autosomal dominant	Neumann (2013) <i>Nephrol Dial Transplant</i> 28:1472	PubMed 23300259
PKD2	CM124025	GAT-GGT	Asp886Gly	2657A>G	D886G	Polycystic kidney disease, autosomal dominant	Rossetti (2012) <i>J Am Soc Nephrol</i> 23:915	PubMed 22383692
PKD2	CM1110164	TTG-TGG	Leu656Trp	1967T>G	L656W	neonatal-onset	Losekoot (2012) <i>J Med Genet</i> 49:37	PubMed 22114106
PKHD1	CM032304	GCA-GTA	Ala17Val	50C>T	A17V	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM020490	ACG-ATG	Thr36Met	107C>T	T36M	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM051116	CGG-GGG	Arg92Gly	274C>G	R92G	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100442	CGG-TGG	Arg92Trp	274C>T	R92W	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM051117	GGG-AGG	Gly112Arg	334G>A	G112R	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051118	CGA-TGA	Arg124Term	370C>T	R124*	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051119	TTC-TCC	Phe129Ser	386T>C	F129S	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051120	CCC-CGC	Pro137Arg	410C>G	P137R	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM065394	TAT-TGT	Tyr143Cys	428A>G	Y143C	Polycystic kidney disease	Adeva (2006) <i>Medicine (Baltimore)</i> 85:1	PubMed 16523049
PKHD1	CM041053	TGG-TGA	Trp158Term	474G>A	W158*	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM054813	GAA-AAA	Glu218Lys	652G>A	E218K	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM020491	ATC-GTC	Ile222Val	664A>G	I222V	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM032305	GGC-AGC	Gly223Ser	667G>A	G223S	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM100563	CAG-GAG	Gln225Glu	673C>G	Q225E	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM100422	ATC-ACC	Ile246Thr	737T>C	I246T	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM054817	CTT-TTT	Leu252Phe	754C>T	L252F	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM020956	TTC-CTC	Phe253Leu	757T>C	F253L	Polycystic kidney disease	Onuchic (2002) <i>Am J Hum Genet</i> 70:1305	PubMed 11898128
PKHD1	CM100419	TAC-TGC	Tyr255Cys	764A>G	Y255C	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM051121	ATA-ATG	Ile261Met	783A>G	I261M	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051122	GGG-GTG	Gly269Val	806G>T	G269V	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM100427	GCA-GTA	Ala293Val	878C>T	A293V	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM041054	ATT-ACT	Ile307Thr	920T>C	I307T	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM034265	GGC-GTC	Gly326Val	977G>T	G326V	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM127370	AAT-GAT	Asn327Asp	979A>G	N327D	Polycystic kidney disease	Zhang (2012) <i>Chin Med J (Engl)</i> 125:2482	PubMed 22882926
PKHD1	CM100565	CGA-GGA	Arg328Gly	982C>G	R328G	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM032306	CGA-TGA	Arg328Term	982C>T	R328*	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM100558	CAG-TAG	Gln353Term	1057C>T	Q353*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM100562	TGG-TGA	Trp365Term	1095G>A	W365*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM051123	TTC-TTG	Phe372Leu	1116C>G	F372L	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM054804	CGG-TGG	Arg375Trp	1123C>T	R375W	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM100554	AAT-ATT	Asn387Ile	1160A>T	N387I	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM051124	GCA-ACA	Ala394Thr	1180G>A	A394T	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161

PKHD1	CM087492	TTG-TCG	Leu400Ser	1199T>C	L400S	Polycystic kidney disease	Harris (2008) <i>Rev Med Chil</i> 136:815	PubMed 18769842
PKHD1	CM051125	TGG-TGA	Trp404Term	1212G>A	W404*	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051126	GGT-CGT	Gly448Arg	1342G>C	G448R	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051127	GGG-GCG	Gly466Ala	1397G>C	G466A	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100417	GGG-GAG	Gly466Glu	1397G>A	G466E	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM051128	GGT-GAT	Gly470Asp	1409G>A	G470D	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM100407	GGT-GTT	Gly470Val	1409G>T	G470V	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM032307	ATT-AGT	Ile473Ser	1418T>G	I473S	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM041055	TAC-CAC	Tyr486His	1456T>C	Y486H	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM100421	TAC-TAA	Tyr486Term	1458C>A	Y486*	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM041056	CGA-TGA	Arg494Term	1480C>T	R494*	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM032308	CGA-CCA	Arg496Pro	1487G>C	R496P	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM032309	CGA-TGA	Arg496Term	1486C>T	R496*	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051129	CAG-CAC	Gln504His	1512G>C	Q504H	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100441	ATT-ACT	Ile539Thr	1616T>C	I539T	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM051130	TTA-TCA	Leu542Ser	1625T>C	L542S	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100548	CGA-TGA	Arg564Term	1690C>T	R564*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM100570	GGC-GTC	Gly584Val	1751G>T	G584V	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM051131	CGA-TGA	Arg592Term	1774C>T	R592*	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM100549	TAT-TAA	Tyr610Term	1830T>A	Y610*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM116253	AAG-AGG	Lys626Arg	1877A>G	K626R	Polycystic kidney disease	Jang (2011) <i>J Obstet Gynaecol Res</i> 37:1744	PubMed 21790888
PKHD1	CM051132	ATG-AAG	Met627Lys	1880T>A	M627K	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM041057	TGG-TAG	Trp646Term	1937G>A	W646*	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM032310	TGG-TGT	Trp656Cys	1968G>T	W656C	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051133	CCC-CGC	Pro676Arg	2027C>G	P676R	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100439	CAT-CCT	His686Pro	2057A>C	H686P	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM032311	GAT-AAT	Asp703Asn	2107G>A	D703N	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM100410	CCA-CGA	Pro724Arg	2171C>G	P724R	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM054802	AAT-AGT	Asn727Ser	2180A>G	N727S	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM032312	CCG-CTG	Pro739Leu	2216C>T	P739L	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM051134	CCG-CTG	Pro755Leu	2264C>T	P755L	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM032313	ATC-CTC	Ile757Leu	2269A>C	I757L	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM020957	CGC-CAC	Arg760His	2279G>A	R760H	Polycystic kidney disease	Onuchic (2002) <i>Am J Hum Genet</i> 70:1305	PubMed 11898128
PKHD1	CM051135	CGA-TGA	Arg781Term	2341C>T	R781*	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM032314	CCT-CTT	Pro805Leu	2414C>T	P805L	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM100555	TAC-TAA	Tyr844Term	2532C>A	Y844*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM100569	TGG-AGG	Trp848Arg	2542T>A	W848R	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM051136	GTC-TTC	Val865Phe	2593G>T	V865F	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034266	ACT-CCT	Thr899Pro	2695A>C	T899P	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM051137	CAT-CCT	His916Pro	2747A>C	H916P	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051138	CTC-CCC	Leu926Pro	2777T>C	L926P	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034267	TGG-TAG	Trp937Term	2810G>A	W937*	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM051139	ATT-AGT	Ile948Ser	2843T>G	I948S	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423

PKHD1	CM054810	GGA-AGA	Gly952Arg	2854G>A	G952R	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM054803	ACC-ATC	Thr979Ile	2936C>T	T979I	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM032315	ATG-AAG	Met997Lys	2990T>A	M997K	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM032316	GCG-GAG	Ala1030Glu	3089C>A	A1030E	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051140	GCG-GTG	Ala1030Val	3089C>T	A1030V	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100556	CGA-TGA	Arg1040Term	3118C>T	R1040*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM020958	GGC-AGC	Gly1122Ser	3364G>A	G1122S	Polycystic kidney disease	Onuchic (2002) <i>Am J Hum Genet</i> 70:1305	PubMed 11898128
PKHD1	CM032317	GGT-AGT	Gly1123Ser	3367G>A	G1123S	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051141	GAG-AAG	Glu1124Lys	3370G>A	E1124K	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051142	TAT-TGT	Tyr1136Cys	3407A>G	Y1136C	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051143	TCG-TTG	Ser1156Leu	3467C>T	S1156L	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051144	TGT-TAT	Cys1237Tyr	3710G>A	C1237Y	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM020492	TGT-TGG	Cys1249Trp	3747T>G	C1249W	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM100566	GTC-GAC	Val1271Asp	3812T>A	V1271D	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM051145	TCA-TAA	Ser1283Term	3848C>A	S1283*	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM100571	ACA-AAA	Thr1300Lys	3899C>A	T1300K	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM052341	GTG-ATG	Val1347Met	4039G>A	V1347M	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM052342	GTG-GCG	Val1380Ala	4139T>C	V1380A	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM032318	CCT-ACT	Pro1389Thr	4165C>A	P1389T	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM051146	TGT-TTT	Cys1402Phe	4205G>T	C1402F	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM020493	CTT-CGT	Leu1407Arg	4220T>G	L1407R	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM051147	TGT-AGT	Cys1431Ser	4291T>A	C1431S	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM054809	CAG-TAG	Gln1444Term	4330C>T	Q1444*	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM041058	TGT-TAT	Cys1472Tyr	4415G>A	C1472Y	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM032319	CCT-CTT	Pro1486Leu	4457C>T	P1486L	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM034268	AGC-ATC	Ser1584Ile	4751G>T	S1584I	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM054818	TGC-TAC	Cys1613Tyr	4838G>A	C1613Y	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM020959	CGG-TGG	Arg1624Trp	4870C>T	R1624W	Polycystic kidney disease	Onuchic (2002) <i>Am J Hum Genet</i> 70:1305	PubMed 11898128
PKHD1	CM090482	CCC-GCC	Pro1628Ala	4882C>G	P1628A	Polycystic kidney disease	Michel-Calemard (2009) <i>Clin Genet</i> 75:203	PubMed 19021639
PKHD1	CM054815	TAT-TGT	Tyr1652Cys	4955A>G	Y1652C	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM020494	TCT-TTT	Ser1664Phe	4991C>T	S1664F	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM051148	ATA-ACA	Ile1687Thr	5060T>C	I1687T	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM090481	TGT-TAT	Cys1692Tyr	5075G>A	C1692Y	Polycystic kidney disease	Michel-Calemard (2009) <i>Clin Genet</i> 75:203	PubMed 19021639
PKHD1	CM034269	CTT-TTT	Leu1709Phe	5125C>T	L1709F	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM100426	GGG-AGG	Gly1712Arg	5134G>A	G1712R	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM020495	GTG-ATG	Val1741Met	5221G>A	V1741M	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM051149	AAC-CAC	Asn1744His	5230A>C	N1744H	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051150	GGC-CGC	Gly1746Arg	5236G>C	G1746R	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051151	GGC-AGC	Gly1746Ser	5236G>A	G1746S	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051152	CGA-TGA	Arg1775Term	5323C>T	R1775*	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034270	ACA-ATA	Thr1781Ile	5342C>T	T1781I	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM051153	TTC-CTC	Phe1785Leu	5353T>C	F1785L	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051154	TGC-TTC	Cys1787Phe	5360G>T	C1787F	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423

PKHD1	CM034271	GTT-CTT	Val1789Leu	5365G>C	V1789L	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM100560	CCC-CTC	Pro1791Leu	5372C>T	P1791L	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM051155	TTG-TCG	Leu1796Ser	5387T>C	L1796S	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM052343	CGT-TGT	Arg1804Cys	5410C>T	R1804C	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM100567	TAT-TGT	Tyr1816Cys	5447A>G	Y1816C	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM100418	GTG-GGG	Val1817Gly	5450T>G	V1817G	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM100550	CAA-TAA	Gln1829Term	5485C>T	Q1829*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM032320	TCG-TTG	Ser1833Leu	5498C>T	S1833L	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM051156	TGG-TGT	Trp1834Cys	5502G>T	W1834C	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM032321	TAC-TGC	Tyr1838Cys	5513A>G	Y1838C	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM051157	TCG-TTG	Ser1862Leu	5585C>T	S1862L	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM032322	AGC-AAC	Ser1867Asn	5600G>A	S1867N	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM100561	AGC-GGC	Ser1867Gly	5599A>G	S1867G	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM100440	GTT-GGT	Val1875Gly	5624T>G	V1875G	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM054806	ATA-ACA	Ile1908Thr	5723T>C	I1908T	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM020496	CAG-CGG	Gln1917Arg	5750A>G	Q1917R	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM054808	CAG-CCG	Gln1917Pro	5750A>C	Q1917P	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM051158	CAG-CTG	Gln1923Leu	5768A>T	Q1923L	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100415	TGG-TTG	Trp1928Leu	5783G>T	W1928L	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM032323	GAT-GGT	Asp1942Gly	5825A>G	D1942G	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM051159	ACA-GCA	Thr1960Ala	5878A>G	T1960A	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034272	GGC-GAC	Gly1971Asp	5912G>A	G1971D	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM127371	GGA-AGA	Gly1979Arg	5935G>A	G1979R	Polycystic kidney disease	Zhang (2012) <i>Chin Med J (Engl)</i> 125:2482	PubMed 22882926
PKHD1	CM065390	ATC-ACC	Ile1988Thr	5963T>C	I1988T	Polycystic kidney disease	Adeva (2006) <i>Medicine (Baltimore)</i> 85:1	PubMed 16523049
PKHD1	CM020497	GAG-GGG	Glu1995Gly	5984A>G	E1995G	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM032324	ATT-ACT	Ile1998Thr	5993T>C	I1998T	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM100438	GCT-ACT	Ala2009Thr	6025G>A	A2009T	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM051160	AGT-TGT	Ser2016Cys	6046A>T	S2016C	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034273	GTG-CTG	Val2032Leu	6094G>C	V2032L	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM065393	AGG-GGG	Arg2033Gly	6097A>G	R2033G	Polycystic kidney disease	Adeva (2006) <i>Medicine (Baltimore)</i> 85:1	PubMed 16523049
PKHD1	CM041059	TGG-TAG	Trp2070Term	6209G>A	W2070*	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM051161	AGT-AAT	Ser2080Asn	6239G>A	S2080N	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100420	CTT-CGT	Leu2106Arg	6317T>G	L2106R	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM032325	CTG-CCG	Leu2134Pro	6401T>C	L2134P	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051162	ACC-CCC	Thr2140Pro	6418A>C	T2140P	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100425	GGG-GAG	Gly2210Glu	6629G>A	G2210E	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM065392	AGC-TGC	Ser2218Cys	6652A>T	S2218C	Polycystic kidney disease	Adeva (2006) <i>Medicine (Baltimore)</i> 85:1	PubMed 16523049
PKHD1	CM100424	GGA-CGA	Gly2224Arg	6670G>C	G2224R	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM052344	GGA-GAA	Gly2285Glu	6854G>A	G2285E	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM041060	ATC-TTC	Ile2303Phe	6907A>T	I2303F	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM020498	ATA-AAA	Ile2331Lys	6992T>A	I2331K	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM112375	TAC-TAA	Tyr2343Term	7029C>A	Y2343*	Polycystic kidney disease	Bell (2011) <i>Sci Transl Med</i> 3:65ra4	PubMed 21228398
PKHD1	CM100572	AGG-GGG	Arg2370Gly	7108A>G	R2370G	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839

PKHD1	CM100551	CAG-TAG	Gln2393Term	7177C>T	Q2393*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM100409	TGC-CGC	Cys2422Arg	7264T>C	C2422R	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM034274	TGC-GGC	Cys2422Gly	7264T>G	C2422G	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM051163	ATT-ACT	Ile2427Thr	7280T>C	I2427T	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100437	GAA-GTA	Glu2431Val	7292A>T	E2431V	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM034275	CAA-TAA	Gln2493Term	7477C>T	Q2493*	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM100408	CGT-TGT	Arg2573Cys	7717C>T	R2573C	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM100568	TAC-AAC	Tyr2638Asn	7912T>A	Y2638N	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM100552	TCA-TAA	Ser2639Term	7916C>A	S2639*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM034276	ACC-GCC	Thr2641Ala	7921A>G	T2641A	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM020499	CGA-TGA	Arg2671Term	8011C>T	R2671*	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM032326	TGT-TTT	Cys2688Phe	8063G>T	C2688F	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM051164	TGG-CGG	Trp2690Arg	8068T>C	W2690R	Polycystic kidney disease	Sgro (2004) <i>Ultrasound Obstet Gynecol</i> 23:73	PubMed 14971004
PKHD1	CM100557	TGG-GGG	Trp2736Gly	8206T>G	W2736G	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM032327	GAT-TAT	Asp2761Tyr	8281G>T	D2761Y	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM104211	GTC-GCC	Val2771Ala	8312T>C	V2771A	Polycystic kidney disease	Tinsa (2010) <i>Tunis Med</i> 88:68	PubMed 20415225
PKHD1	CM054814	GTC-GAC	Val2771Asp	8312T>A	V2771D	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM051165	GTC-ATC	Val2771Ile	8311G>A	V2771I	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034277	CTT-CCT	Leu2772Pro	8315T>C	L2772P	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM054820	GTG-TTG	Val2773Leu	8317G>T	V2773L	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM051166	AGA-AGC	Arg2795Ser	8385A>C	R2795S	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100413	GTT-GGT	Val2798Gly	8393T>G	V2798G	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM100412	TGC-CGC	Cys2803Arg	8407T>C	C2803R	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM100414	ATG-AAG	Met2804Lys	8411T>A	M2804K	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM051167	GGG-AGG	Gly2809Arg	8425G>A	G2809R	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051168	AGA-AAA	Arg2831Lys	8492G>A	R2831K	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051169	CGT-TGT	Arg2840Cys	8518C>T	R2840C	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM052345	ATG-GTG	Met2841Val	8521A>G	M2841V	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051170	ATT-ACT	Ile2851Thr	8552T>C	I2851T	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034278	AGT-GGT	Ser2861Gly	8581A>G	S2861G	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM034279	TAT-TGT	Tyr2863Cys	8588A>G	Y2863C	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM032328	ACA-AAA	Thr2869Lys	8606C>A	T2869K	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM052346	GTG-ATG	Val2913Met	8737G>A	V2913M	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM052347	CGG-TGG	Arg2930Trp	8788C>T	R2930W	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM100553	CGA-TGA	Arg2942Term	8824C>T	R2942*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM020500	ATA-ACA	Ile2957Thr	8870T>C	I2957T	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM041061	GAC-GGC	Asp2962Gly	8885A>G	D2962G	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM054807	TGT-CGT	Cys2965Arg	8893T>C	C2965R	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM051171	GGG-TGG	Gly2967Trp	8899G>T	G2967W	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100564	CGA-TGA	Arg2979Term	8935C>T	R2979*	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM034280	TCA-TTA	Ser2983Leu	8948C>T	S2983L	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM020501	TCT-TTT	Ser3018Phe	9053C>T	S3018F	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM051172	CAT-CCT	His3029Pro	9086A>C	H3029P	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423

PKHD1	CM034281	GTA-GGA	Val3036Gly	9107T>G	V3036G	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM100411	CAT-CGT	His3049Arg	9146A>G	H3049R	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM051173	ATA-ACA	Ile3051Thr	9152T>C	I3051T	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM052348	GCG-GTG	Ala3072Val	9215C>T	A3072V	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM1313819	GGA-GAA	Gly3080Glu	9239G>A	G3080E	Polycystic kidney disease	Brinkert (2013) <i>Transpl Int</i> 26:640	PubMed 23582048
PKHD1	CM032329	ATC-GTC	Ile3081Val	9241A>G	I3081V	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051174	GCA-ACA	Ala3097Thr	9289G>A	A3097T	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051175	TTT-TGT	Phe3104Cys	9311T>G	F3104C	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM032330	CGA-TGA	Arg3107Term	9319C>T	R3107*	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051176	CAT-CCT	His3124Pro	9371A>C	H3124P	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM041062	CAT-TAT	His3124Tyr	9370C>T	H3124Y	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM020960	GAC-TAC	Asp3139Tyr	9415G>T	D3139Y	Polycystic kidney disease	Onuchic (2002) <i>Am J Hum Genet</i> 70:1305	PubMed 11898128
PKHD1	CM100559	GAC-GGC	Asp3154Gly	9461A>G	D3154G	Polycystic kidney disease	Denamur (2010) <i>Kidney Int</i> 77:350	PubMed 19940839
PKHD1	CM051177	TAT-TGT	Tyr3155Cys	9464A>G	Y3155C	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034282	ATA-TTA	Ile3167Leu	9499A>T	I3167L	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM034283	AAT-GAT	Asn3175Asp	9523A>G	N3175D	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM034284	AAT-AGT	Asn3175Ser	9524A>G	N3175S	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM034056	ATT-ACT	Ile3177Thr	9530T>C	I3177T	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM051178	GGT-TGT	Gly3178Cys	9532G>T	G3178C	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM100423	GCC-ACC	Ala3207Thr	9619G>A	A3207T	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM051179	AGC-CGC	Ser3209Arg	9625A>C	S3209R	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM054816	TCA-TAA	Ser3228Term	9683C>A	S3228*	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM051180	CGA-CAA	Arg3240Gln	9719G>A	R3240Q	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051181	CGA-CTA	Arg3240Leu	9719G>T	R3240L	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM032331	CGA-TGA	Arg3240Term	9718C>T	R3240*	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051182	GGT-GTT	Gly3242Val	9725G>T	G3242V	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM054819	TGG-TGA	Trp3255Term	9765G>A	W3255*	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM052349	GTG-GCG	Val3263Ala	9788T>C	V3263A	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051183	TGC-TGA	Cys3287Term	9861C>A	C3287*	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM051184	AGC-ATC	Ser3289Ile	9866G>T	S3289I	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM032332	GAT-GTT	Asp3293Val	9878A>T	D3293V	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM051185	AGG-ACG	Arg3333Thr	9998G>C	R3333T	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM054821	TTA-TGA	Leu3344Term	10031T>G	L3344*	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM032333	TGT-CGT	Cys3346Arg	10036T>C	C3346R	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM020502	CAG-TAG	Gln3392Term	10174C>T	Q3392*	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM041063	CAG-TAG	Gln3407Term	10219C>T	Q3407*	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM054811	GAC-GTC	Asp3409Val	10226A>T	D3409V	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM052350	GTC-GAC	Val3440Asp	10319T>A	V3440D	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM032334	ATC-GTC	Ile3468Val	10402A>G	I3468V	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM032335	GTC-GGC	Val3471Gly	10412T>G	V3471G	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM032336	CGC-TGC	Arg3482Cys	10444C>T	R3482C	Polycystic kidney disease	Bergmann (2003) <i>J Am Soc Nephrol</i> 14:76	PubMed 12506140
PKHD1	CM032337	GAG-GTG	Glu3502Val	10505A>T	E3502V	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM032338	GAA-CAA	Glu3529Gln	10585G>C	E3529Q	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734

PKHD1	CM051186	TTG-TCG	Leu3543Ser	10628T>C	L3543S	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM020503	ATT-ACT	Ile3553Thr	10658T>C	I3553T	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM054805	ATA-AAA	Ile3555Lys	10664T>A	I3555K	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM052351	CGC-TGC	Arg3556Cys	10666C>T	R3556C	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051187	TCA-TGA	Ser3570Term	10709C>G	S3570*	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034285	TGG-TGA	Trp3576Term	10728G>A	W3576*	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM034286	CAG-TAG	Gln3589Term	10765C>T	Q3589*	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM051188	CCT-CTT	Pro3602Leu	10805C>T	P3602L	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM032339	TGC-TAC	Cys3622Tyr	10865G>A	C3622Y	Polycystic kidney disease	Rossetti (2003) <i>Kidney Int</i> 64:391	PubMed 12846734
PKHD1	CM065389	CGT-TGT	Arg3637Cys	10909C>T	R3637C	Polycystic kidney disease	Adeva (2006) <i>Medicine (Baltimore)</i> 85:1	PubMed 16523049
PKHD1	CM051189	ATG-ATA	Met3642Ile	10926G>A	M3642I	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051190	ATG-ACG	Met3642Thr	10925T>C	M3642T	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM034287	CGA-TGA	Arg3692Term	11074C>T	R3692*	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM051191	CCC-TCC	Pro3740Ser	11218C>T	P3740S	Polycystic kidney disease	Bergmann (2005) <i>Kidney Int</i> 67:829	PubMed 15698423
PKHD1	CM054812	CCA-ACA	Pro3762Thr	11284C>A	P3762T	Polycystic kidney disease	Losekoot (2005) <i>Hum Genet</i> 118:185	PubMed 16133180
PKHD1	CM041064	CGA-TGA	Arg3772Term	11314C>T	R3772*	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:487	PubMed 15108281
PKHD1	CM034288	CCA-TCA	Pro3783Ser	11347C>T	P3783S	Polycystic kidney disease	Furu (2003) <i>J Am Soc Nephrol</i> 14:2004	PubMed 12874454
PKHD1	CM051192	CGA-TGA	Arg3842Term	11524C>T	R3842*	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM051193	TGG-CGG	Trp3871Arg	11611T>C	W3871R	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM020504	TGG-TAG	Trp3871Term	11612G>A	W3871*	Polycystic kidney disease	Ward (2002) <i>Nat Genet</i> 30:259	PubMed 11919560
PKHD1	CM052352	CGC-TGC	Arg3913Cys	11737C>T	R3913C	Polycystic kidney disease	Sharp (2005) <i>J Med Genet</i> 42:336	PubMed 15805161
PKHD1	CM100416	CGC-TGC	Arg3957Cys	11869C>T	R3957C	Polycystic kidney disease	Gunay-Aygun (2010) <i>Mol Genet Metab</i> 99:160	PubMed 19914852
PKHD1	CM045947	TAC-TAG	Tyr4009Term	12027C>G	Y4009*	Polycystic kidney disease	Bergmann (2004) <i>Hum Mutat</i> 23:453	PubMed 15108277
PKHD1	CM065391	CCG-CTG	Pro4067Leu	12200C>T	P4067L	Polycystic kidney disease	Adeva (2006) <i>Medicine (Baltimore)</i> 85:1	PubMed 16523049
PTPN11	CM117756	CAG-CAC	Gln510His	1530G>C	Q510H	LEOPARD syndrome	Wakabayashi (2011) <i>Am J Med Genet A</i> 155:2529	PubMed 21910226
PTPN11	CM041069	TAT-TCT	Tyr279Ser	836A>C	Y279S	ML / LEOPARD syndrome	Sarkozy (2004) <i>J Med Genet</i> 41:e68	PubMed 15121796
PTPN11	CM096577	GCT-TCT	Ala461Ser	1381G>T	A461S	ML / LEOPARD syndrome	Osawa (2009) <i>Br J Dermatol</i> 161:1202	PubMed 19659470
PTPN11	CM043335	GCT-ACT	Ala461Thr	1381G>A	A461T	ML / LEOPARD syndrome	Yoshida (2004) <i>Am J Med Genet</i> 130A:432	PubMed 15389709
PTPN11	CM041070	GGC-GCC	Gly464Ala	1391G>C	G464A	ML / LEOPARD syndrome	Sarkozy (2004) <i>J Med Genet</i> 41:e68	PubMed 15121796
PTPN11	CM021672	ACG-ATG	Thr468Met	1403C>T	T468M	ML / LEOPARD syndrome	Digilio (2002) <i>Am J Hum Genet</i> 71:389	PubMed 12058348
PTPN11	CM074987	ACG-CCG	Thr468Pro	1402A>C	T468P	ML / LEOPARD syndrome	Seishima (2007) <i>Br J Dermatol</i> 157:1297	PubMed 17927788
PTPN11	CM041071	CGG-CTG	Arg498Leu	1493G>T	R498L	ML / LEOPARD syndrome	Sarkozy (2004) <i>J Med Genet</i> 41:e68	PubMed 15121796
PTPN11	CM041072	CGG-TGG	Arg498Trp	1492C>T	R498W	ML / LEOPARD syndrome	Sarkozy (2004) <i>J Med Genet</i> 41:e68	PubMed 15121796
PTPN11	CM031338	CAG-CCG	Gln506Pro	1517A>C	Q506P	ML / LEOPARD syndrome	Conti (2003) <i>Hum Mutat</i> 21:654	PubMed 14961557
PTPN11	CM043070	CAG-CCG	Gln510Pro	1529A>C	Q510P	ML / LEOPARD syndrome	Keren (2004) <i>J Med Genet</i> 41:e117	PubMed 15520399
PTPN11	CM032348	ACA-ATA	Thr2Ile	5C>T	T2I	Noonan syndrome	Sarkozy (2003) <i>J Med Genet</i> 40:704	PubMed 12960218
PTPN11	CM021125	ACA-GCA	Thr42Ala	124A>G	T42A	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM122798	ACC-ATC	Thr52Ile	155C>T	T52I	Noonan syndrome	Ezquieta (2012) <i>Rev Esp Cardiol</i> 65:447	PubMed 22465605
PTPN11	CM044250	AAC-GAC	Asn58Asp	172A>G	N58D	Noonan syndrome	Zenker (2004) <i>J Pediatr</i> 144:368	PubMed 15001945
PTPN11	CM060441	AAC-CAC	Asn58His	172A>C	N58H	Noonan syndrome	Limal (2006) <i>J Clin Endocrinol Metab</i> 91:300	PubMed 16263833
PTPN11	CM030492	AAC-AAG	Asn58Lys	174C>G	N58K	Noonan syndrome	Musante (2003) <i>Eur J Hum Genet</i> 11:201	PubMed 12634870
PTPN11	CM086895	ACT-GCT	Thr59Ala	175A>G	T59A	Noonan syndrome	Ko (2008) <i>J Hum Genet</i> 53:999	PubMed 19020799
PTPN11	CM021126	GGT-GCT	Gly60Ala	179G>C	G60A	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261

PTPN11	CM060445	GGT-TGT	Gly60Cys	178G>T	G60C	Noonan syndrome	Limal (2006) <i>J Clin Endocrinol Metab</i> 91:300	PubMed 16263833
PTPN11	CM122799	GGT-AGT	Gly60Ser	178G>A	G60S	Noonan syndrome	Ezquieta (2012) <i>Rev Esp Cardiol</i> 65:447	PubMed 22465605
PTPN11	CM098208	GAT-GCT	Asp61Ala	182A>C	D61A	Noonan syndrome	Chen (2009) <i>Lymphology</i> 42:134	PubMed 19927903
PTPN11	CM021127	GAT-AAT	Asp61Asn	181G>A	D61N	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM013415	GAT-GGT	Asp61Gly	182A>G	D61G	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM101143	GAT-CAT	Asp61His	181G>C	D61H	Noonan syndrome	Houweling (2010) <i>Prenat Diagn</i> 30:284	PubMed 20112233
PTPN11	CM060446	TAC-AAC	Tyr62Asn	184T>A	Y62N	Noonan syndrome	Tartaglia (2006) <i>Am J Hum Genet</i> 78:279	PubMed 16358218
PTPN11	CM021128	TAC-GAC	Tyr62Asp	184T>G	Y62D	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM013416	TAT-TGT	Tyr63Cys	188A>G	Y63C	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM030493	GAG-CAG	Glu69Gln	205G>C	E69Q	Noonan syndrome	Musante (2003) <i>Eur J Hum Genet</i> 11:201	PubMed 12634870
PTPN11	CM092721	GAG-GTG	Glu69Val	206A>T	E69V	Noonan syndrome	Pierpont (2009) <i>Genes Brain Behav</i> 8:275	PubMed 19077116
PTPN11	CM056378	TTT-ATT	Phe71Ile	211T>A	F71I	Noonan syndrome	Niihori (2005) <i>J Hum Genet</i> 50:192	PubMed 15834506
PTPN11	CM030494	TTT-CTT	Phe71Leu	211T>C	F71L	Noonan syndrome	Musante (2003) <i>Eur J Hum Genet</i> 11:201	PubMed 12634870
PTPN11	CM013417	GCC-GGC	Ala72Gly	215C>G	A72G	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM090463	GCC-CCC	Ala72Pro	214G>C	A72P	Noonan syndrome	Lee (2009) <i>Clin Genet</i> 75:190	PubMed 18759865
PTPN11	CM013418	GCC-TCC	Ala72Ser	214G>T	A72S	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM021129	ACT-ATT	Thr73Ile	218C>T	T73I	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM013419	GAG-GAC	Glu76Asp	228G>C	E76D	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM060442	GAG-GAT	Glu76Asp	228G>T	E76D	Noonan syndrome	Tartaglia (2006) <i>Am J Hum Genet</i> 78:279	PubMed 16358218
PTPN11	CM137734	GAG-GTG	Glu76Val	227A>T	E76V	Noonan syndrome	Croonen (2013) <i>Eur J Hum Genet</i> 21:936	PubMed 23321623
PTPN11	CM013420	CAG-CGG	Gln79Arg	236A>G	Q79R	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM032349	CAG-CCG	Gln79Pro	236A>C	Q79P	Noonan syndrome	Sarkozy (2003) <i>J Med Genet</i> 40:704	PubMed 12960218
PTPN11	CM021130	GAT-GCT	Asp106Ala	317A>C	D106A	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM1213017	GAT-GGT	Asp106Gly	317A>G	D106G	Noonan syndrome	Bertelloni (2013) <i>Hormones (Athens)</i> 12:86	PubMed 23624134
PTPN11	CM044251	GAA-GCA	Glu110Ala	329A>C	E110A	Noonan syndrome	Zenker (2004) <i>J Pediatr</i> 144:368	PubMed 15001945
PTPN11	CM122800	GAA-AAA	Glu110Lys	328G>A	E110K	Noonan syndrome	Ezquieta (2012) <i>Rev Esp Cardiol</i> 65:447	PubMed 22465605
PTPN11	CM021132	GAG-GAC	Glu139Asp	417G>C	E139D	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM021131	GAG-GAT	Glu139Asp	417G>T	E139D	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM137739	CCT-TCT	Pro144Ser	430C>T	P144S	Noonan syndrome	Croonen (2013) <i>Eur J Hum Genet</i> 21:936	PubMed 23321623
PTPN11	BM1244269	CGG-TGG	Arg186Trp	556C>T	R186W	Noonan syndrome	Bertelloni (2013) <i>Hormones (Athens)</i> 12:86	PubMed 23624134
PTPN11	CM140985	ATA-GTA	Ile221Val	661A>G	I221V	Noonan syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
PTPN11	CM030495	CAA-CGA	Gln256Arg	767A>G	Q256R	Noonan syndrome	Musante (2003) <i>Eur J Hum Genet</i> 11:201	PubMed 12634870
PTPN11	CM053388	CAA-AAA	Gln256Lys	766C>A	Q256K	Noonan syndrome	Binder (2005) <i>J Clin Endocrinol Metab</i> 90:5377	PubMed 15985475
PTPN11	CM097361	TGC-AGC	Cys259Ser	775T>A	C259S	Noonan syndrome	Lo (2009) <i>Clin Chim Acta</i> 409:75	PubMed 19737548
PTPN11	CM122720	CTT-CAT	Leu261His	782T>A	L261H	Noonan syndrome	Ferrero (2012) <i>Hum Mutat</i> 33:703	PubMed 22253195
PTPN11	CM122801	CTT-TTT	Leu261Phe	781C>T	L261F	Noonan syndrome	Ezquieta (2012) <i>Rev Esp Cardiol</i> 65:447	PubMed 22465605
PTPN11	CM122719	CTC-CGC	Leu262Arg	785T>G	L262R	Noonan syndrome	Ferrero (2012) <i>Hum Mutat</i> 33:703	PubMed 22253195
PTPN11	CM060444	GGT-TGT	Gly268Cys	802G>T	G268C	Noonan syndrome	Tartaglia (2006) <i>Am J Hum Genet</i> 78:279	PubMed 16358218
PTPN11	CM060443	GGT-AGT	Gly268Ser	802G>A	G268S	Noonan syndrome	Tartaglia (2006) <i>Am J Hum Genet</i> 78:279	PubMed 16358218
PTPN11	CM021133	TAT-TGT	Tyr279Cys	836A>G	Y279C	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM013421	ATC-GTC	Ile282Val	844A>G	I282V	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM060447	TTT-TGT	Phe285Cys	854T>G	F285C	Noonan syndrome	Tartaglia (2006) <i>Am J Hum Genet</i> 78:279	PubMed 16358218
PTPN11	CM086896	TTT-ATT	Phe285Ile	853T>A	F285I	Noonan syndrome	Ferrero (2008) <i>Eur J Med Genet</i> 51:566	PubMed 18678287

PTPN11	CM024733	TTT-CTT	Phe285Leu	853T>C	F285L	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM073286	TTT-TTG	Phe285Leu	855T>G	F285L	Noonan syndrome	Hung (2007) <i>J Formos Med Assoc</i> 106:169	PubMed 17339163
PTPN11	CM021134	TTT-TCT	Phe285Ser	854T>C	F285S	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM013422	AAT-GAT	Asn308Asp	922A>G	N308D	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM021135	AAT-AGT	Asn308Ser	923A>G	N308S	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM044252	AAT-ACT	Asn308Thr	923A>C	N308T	Noonan syndrome	Zenker (2004) <i>J Pediatr</i> 144:368	PubMed 15001945
PTPN11	CM021136	ATC-GTC	Ile309Val	925A>G	I309V	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM140986	GAC-TAC	Asp395Tyr	1183G>T	D395Y	Noonan syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
PTPN11	CM140987	TAT-CAT	Tyr396His	1186T>C	Y396H	Noonan syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
PTPN11	CM070248	GGG-GCG	Gly409Ala	1226G>C	G409A	Noonan syndrome	Zenker (2007) <i>Eur J Med Genet</i> 50:43	PubMed 17052965
PTPN11	CM043334	ACG-ATG	Thr411Met	1232C>T	T411M	Noonan syndrome	Bertola (2004) <i>Am J Med Genet</i> 130A:378	PubMed 15384080
PTPN11	CM140988	GTG-TTG	Val428Leu	1282G>T	V428L	Noonan syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
PTPN11	CM067454	CCC-CAC	Pro491His	1472C>A	P491H	Noonan syndrome	Bertola (2006) <i>Genet Test</i> 10:186	PubMed 17020470
PTPN11	CM053389	CCC-CTC	Pro491Leu	1472C>T	P491L	Noonan syndrome	Binder (2005) <i>J Clin Endocrinol Metab</i> 90:5377	PubMed 15985475
PTPN11	CM044253	CCC-TCC	Pro491Ser	1471C>T	P491S	Noonan syndrome	Zenker (2004) <i>J Pediatr</i> 144:368	PubMed 15001945
PTPN11	CM122802	CCC-ACC	Pro491Thr	1471C>A	P491T	Noonan syndrome	Ezquieta (2012) <i>Rev Esp Cardiol</i> 65:447	PubMed 22465605
PTPN11	CM021137	AGG-AAG	Arg501Lys	1502G>A	R501K	Noonan syndrome	Tartaglia (2002) <i>Am J Hum Genet</i> 70:1555	PubMed 11992261
PTPN11	CM055504	TCA-GCA	Ser502Ala	1504T>G	S502A	Noonan syndrome	Kratz (2005) <i>Blood</i> 106:2183	PubMed 15928039
PTPN11	CM044932	TCA-TTA	Ser502Leu	1505C>T	S502L	Noonan syndrome	Bertola (2006) <i>Genet Test</i> 10:186	PubMed 17020470
PTPN11	CM022450	TCA-ACA	Ser502Thr	1504T>A	S502T	Noonan syndrome	Maheshwari (2002) <i>Hum Mutat</i> 20:298	PubMed 12325025
PTPN11	CM060440	GGG-AGG	Gly503Arg	1507G>A	G503R	Noonan syndrome	Tartaglia (2006) <i>Am J Hum Genet</i> 78:279	PubMed 16358218
PTPN11	CM032350	GGG-CGG	Gly503Arg	1507G>C	G503R	Noonan syndrome	Sarkozy (2003) <i>J Med Genet</i> 40:704	PubMed 12960218
PTPN11	CM086897	GGG-GAG	Gly503Glu	1508G>A	G503E	Noonan syndrome	Ferrero (2008) <i>Eur J Med Genet</i> 51:566	PubMed 18678287
PTPN11	CM013423	ATG-GTG	Met504Val	1510A>G	M504V	Noonan syndrome	Tartaglia (2001) <i>Nat Genet</i> 29:465	PubMed 11704759
PTPN11	CM090464	ACA-AAA	Thr507Lys	1520C>A	T507K	Noonan syndrome	Lee (2009) <i>Clin Genet</i> 75:190	PubMed 18759865
PTPN11	CM052358	CAG-CGG	Gln510Arg	1529A>G	Q510R	Noonan syndrome	Bertola (2005) <i>Am J Med Genet</i> 136A:242	PubMed 15948193
PTPN11	CM055503	CAG-GAG	Gln510Glu	1528C>G	Q510E	Noonan syndrome	Takahashi (2005) <i>Eur J Pediatr</i> 164:497	PubMed 15889278
PTPN11	CM090465	ACG-ATG	Thr553Met	1658C>T	T553M	Noonan syndrome	Lee (2009) <i>Clin Genet</i> 75:190	PubMed 18759865
PTPN11	CM032351	CTC-TTC	Leu560Phe	1678C>T	L560F	Noonan syndrome	Sarkozy (2003) <i>J Med Genet</i> 40:704	PubMed 12960218
RAF1	CM1314432	CCT-TCT	Pro235Ser	703C>T	P235S	Noonan syndrome	Quaio (2013) <i>Clinics (Sao Paulo)</i> 68:1079	PubMed 24037001
RAF1	CM073295	AGG-AGT	Arg256Ser	768G>T	R256S	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM073301	TCG-TTG	Ser257Leu	770C>T	S257L	Noonan syndrome	Razzaque (2007) <i>Nat Genet</i> 39:1013	PubMed 17603482
RAF1	CM073300	TCC-TTC	Ser259Phe	776C>T	S259F	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM137735	TCC-CCC	Ser259Pro	775T>C	S259P	Noonan syndrome	Croonen (2013) <i>Eur J Hum Genet</i> 21:936	PubMed 23321623
RAF1	CM086899	TCC-ACC	Ser259Thr	775T>A	S259T	Noonan syndrome	Ko (2008) <i>J Hum Genet</i> 53:999	PubMed 19020799
RAF1	CM073288	ACA-AGA	Thr260Arg	779C>G	T260R	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM073292	CCT-GCT	Pro261Ala	781C>G	P261A	Noonan syndrome	Razzaque (2007) <i>Nat Genet</i> 39:1013	PubMed 17603482
RAF1	CM122806	CCT-CAT	Pro261His	782C>A	P261H	Noonan syndrome	Ezquieta (2012) <i>Rev Esp Cardiol</i> 65:447	PubMed 22465605
RAF1	CM073296	CCT-CTT	Pro261Leu	782C>T	P261L	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM073293	CCT-TCT	Pro261Ser	781C>T	P261S	Noonan syndrome	Razzaque (2007) <i>Nat Genet</i> 39:1013	PubMed 17603482
RAF1	CM116372	CCT-ACT	Pro261Thr	781C>A	P261T	Noonan syndrome	Lee (2011) <i>J Pediatr</i> 159:1029	PubMed 21784453
RAF1	CM140994	AAT-ATT	Asn262Ile	785A>T	N262I	Noonan syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
RAF1	CM073299	GTC-GCC	Val263Ala	788T>C	V263A	Noonan syndrome	Razzaque (2007) <i>Nat Genet</i> 39:1013	PubMed 17603482

RAF1	CM128773	GTC-GAC	Val263Asp	788T>A	V263D	Noonan syndrome	Schulz (2012) <i>Prenat Diagn</i> 32:1016	PubMed 22821648
RAF1	CM137738	GTG-GGG	Val312Gly	935T>G	V312G	Noonan syndrome	Croonen (2013) <i>Eur J Hum Genet</i> 21:936	PubMed 23321623
RAF1	CM122807	GAA-AAA	Glu478Lys	1432G>A	E478K	Noonan syndrome	Ezquieta (2012) <i>Rev Esp Cardiol</i> 65:447	PubMed 22465605
RAF1	CM073294	GAT-AAT	Asp486Asn	1456G>A	D486N	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM073298	GAT-GGT	Asp486Gly	1457A>G	D486G	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM073289	ACA-AGA	Thr491Arg	1472C>G	T491R	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM073291	ACA-ATA	Thr491Ile	1472C>T	T491I	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM073297	TCT-ACT	Ser612Thr	1834T>A	S612T	Noonan syndrome	Pandit (2007) <i>Nat Genet</i> 39:1007	PubMed 17603483
RAF1	CM073302	CTA-GTA	Leu613Val	1837C>G	L613V	Noonan syndrome	Razzaque (2007) <i>Nat Genet</i> 39:1013	PubMed 17603482
RAF1	CM101369	AGA-ATA	Arg191Ile	572G>T	R191I	Noonan syndrome, atypical	Kobayashi (2010) <i>Hum Mutat</i> 31:284	PubMed 20052757
SHH	CM041831	AGA-ACA	Arg6Thr	17G>C	R6T	Holoprosencephaly	Dubourg (2004) <i>Hum Mutat</i> 24:43	PubMed 15221788
SHH	CM005509	CTG-CCG	Leu17Pro	50T>C	L17P	Holoprosencephaly	Kato (2000) <i>Ann Neurol</i> 47:514	PubMed 10762164
SHH	CM126224	TGC-TGA	Cys19Term	57C>A	C19*	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM041832	TGC-TGA	Cys24Term	72C>A	C24*	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SHH	CM095847	CCG-CTG	Pro26Leu	77C>T	P26L	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM044942	GGC-GCC	Gly27Ala	80G>C	G27A	Holoprosencephaly	Hehr (2004) <i>Eur J Pediatr</i> 163:347	PubMed 15107988
SHH	CM961274	GGG-AGG	Gly31Arg	91G>A	G31R	Holoprosencephaly	Roessler (1996) <i>Nat Genet</i> 14:357	PubMed 8896572
SHH	CM095848	CTG-CCG	Leu39Pro	116T>C	L39P	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM136863	TAC-TAA	Tyr44Term	132C>A	Y44*	Holoprosencephaly	Lami (2013) <i>J Genet</i> 92:97	PubMed 23640411
SHH	CM095849	CAG-TAG	Gln46Term	136C>T	Q46*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM126225	GCC-ACC	Ala52Thr	154G>A	A52T	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM095850	GAG-AAG	Glu53Lys	157G>A	E53K	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM126226	GAG-TAG	Glu53Term	157G>T	E53*	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM122188	CGA-TGA	Arg72Term	214C>T	R72*	Holoprosencephaly	Roessler (2012) <i>Mol Genet Metab</i> 105:658	PubMed 22310223
SHH	CM095851	GAC-GTC	Asp83Val	248A>T	D83V	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095852	ATC-TTC	Ile84Phe	250A>T	I84F	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM992966	GAT-GTT	Asp88Val	263A>T	D88V	Holoprosencephaly	Nanni (1999) <i>Hum Mol Genet</i> 8:2479	PubMed 10556296
SHH	CM992216	CAG-CAC	Gln100His	300G>C	Q100H	Holoprosencephaly	Odent (1999) <i>Hum Mol Genet</i> 8:1683	PubMed 10441331
SHH	CM961275	CAG-TAG	Gln100Term	298C>T	Q100*	Holoprosencephaly	Roessler (1996) <i>Nat Genet</i> 14:357	PubMed 8896572
SHH	CM095853	TGT-CGT	Cys102Arg	304T>C	C102R	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM094633	TGT-TAT	Cys102Tyr	305G>A	C102Y	Holoprosencephaly	Bendavid (2009) <i>Hum Mutat</i> 30:1175	PubMed 19431187
SHH	CM961276	AAG-TAG	Lys105Term	313A>T	K105*	Holoprosencephaly	Roessler (1996) <i>Nat Genet</i> 14:357	PubMed 8896572
SHH	CM095854	TTG-TTT	Leu109Phe	327G>T	L109F	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM041833	GCC-GAC	Ala110Asp	329C>A	A110D	Holoprosencephaly	Dubourg (2004) <i>Hum Mutat</i> 24:43	PubMed 15221788
SHH	CM095855	GCC-ACC	Ala110Thr	328G>A	A110T	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM052914	ATC-AAC	Ile111Asn	332T>A	I111N	Holoprosencephaly	El-Jaick (2005) <i>Am J Med Genet</i> 136A:345	PubMed 15942952
SHH	CM122189	TCG-TAG	Ser112Term	335C>A	S112*	Holoprosencephaly	Roessler (2012) <i>Mol Genet Metab</i> 105:658	PubMed 22310223
SHH	CM992967	AAC-AAA	Asn115Lys	345C>A	N115K	Holoprosencephaly	Nanni (1999) <i>Hum Mol Genet</i> 8:2479	PubMed 10556296
SHH	CM961277	TGG-CGG	Trp117Arg	349T>C	W117R	Holoprosencephaly	Roessler (1996) <i>Nat Genet</i> 14:357	PubMed 8896572
SHH	CM961278	TGG-GGG	Trp117Gly	349T>G	W117G	Holoprosencephaly	Roessler (1996) <i>Nat Genet</i> 14:357	PubMed 8896572
SHH	CM095856	GGA-TGA	Gly119Term	355G>T	G119*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095857	GTG-ATG	Val124Met	370G>A	V124M	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM030953	TGG-TAG	Trp128Term	383G>A	W128*	Holoprosencephaly	Marini (2003) <i>Am J Med Genet</i> 117A:112	PubMed 12567406

SHH	CM1111553	TGG-TGA	Trp128Term	384G>A	W128*	Holoprosencephaly	Aguinaga (2011) <i>Case Rep Genet</i> 2011:	PubMed 23074678
SHH	CM041834	GAA-TAA	Glu130Term	388G>T	E130*	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SHH	CM095912	GAG-AAG	Glu136Lys	406G>A	E136K	Holoprosencephaly	Bertolacini (2009) <i>Brain Dev</i> :	PubMed 19398181
SHH	CM012472	CAC-CCC	His140Pro	419A>C	H140P	Holoprosencephaly	Orioli (2001) <i>Hum Genet</i> 109:1	PubMed 11479728
SHH	CM095858	GGC-GAC	Gly143Asp	428G>A	G143D	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM126229	CGC-CAC	Arg144His	431G>A	R144H	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM126228	CGC-CTC	Arg144Leu	431G>T	R144L	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM095859	CGC-CCC	Arg144Pro	431G>C	R144P	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095860	GAC-AAC	Asp147Asn	439G>A	D147N	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM041835	ACG-AGG	Thr150Arg	449C>G	T150R	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SHH	CM095861	ACG-AAG	Thr150Lys	449C>A	T150K	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095862	AGC-AGA	Ser156Arg	468C>A	S156R	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM118760	AAG-TAG	Lys157Term	469A>T	K157*	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SHH	CM991129	TAC-TAG	Tyr158Term	474C>G	Y158*	Holoprosencephaly	Odent (1999) <i>Hum Mol Genet</i> 8:1683	PubMed 10441331
SHH	CM126230	CTG-CAG	Leu161Gln	482T>A	L161Q	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM126231	GCG-GTG	Ala165Val	494C>T	A165V	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM095863	TTC-TGC	Phe170Cys	509T>G	F170C	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095864	GAC-CAC	Asp171His	511G>C	D171H	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095865	TAC-TAG	Tyr175Term	525C>G	Y175*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095866	TGC-CGC	Cys183Arg	547T>C	C183R	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM012473	TGC-TTC	Cys183Phe	548G>T	C183F	Holoprosencephaly	Orioli (2001) <i>Hum Genet</i> 109:1	PubMed 11479728
SHH	CM095867	TGC-TAC	Cys183Tyr	548G>A	C183Y	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095868	TCG-TTG	Ser184Leu	551C>T	S184L	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM991130	GAG-CAG	Glu188Gln	562G>C	E188Q	Holoprosencephaly	Odent (1999) <i>Hum Mol Genet</i> 8:1683	PubMed 10441331
SHH	CM095870	GGA-GAA	Gly196Glu	587G>A	G196E	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095871	GGC-GTC	Gly197Val	590G>T	G197V	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095872	TGC-AGC	Cys198Ser	592T>A	C198S	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095913	TGC-TAC	Cys198Tyr	593G>A	C198Y	Holoprosencephaly	Bertolacini (2009) <i>Brain Dev</i> :	PubMed 19398181
SHH	CM118761	GGC-GAC	Gly201Asp	602G>A	G201D	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SHH	CM992968	CAG-TAG	Gln209Term	625C>T	Q209*	Holoprosencephaly	Nanni (1999) <i>Hum Mol Genet</i> 8:2479	PubMed 10556296
SHH	CM066233	CTG-CCG	Leu218Pro	653T>C	L218P	Holoprosencephaly	Richieri-Costa (2006) <i>Am J Med Genet A</i> 140A:2587	PubMed 17001669
SHH	CM991131	GAC-AAC	Asp222Asn	664G>A	D222N	Holoprosencephaly	Odent (1999) <i>Hum Mol Genet</i> 8:1683	PubMed 10441331
SHH	CM971374	GTG-GAG	Val224Glu	671T>A	V224E	Holoprosencephaly	Roessler (1997) <i>Hum Mol Genet</i> 6:1847	PubMed 9302262
SHH	CM118762	CTG-CCG	Leu225Pro	674T>C	L225P	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SHH	CM971375	GCG-ACG	Ala226Thr	676G>A	A226T	Holoprosencephaly	Roessler (1997) <i>Hum Mol Genet</i> 6:1847	PubMed 9302262
SHH	CM095873	GGC-GTC	Gly231Val	692G>T	G231V	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095874	CGG-GGG	Arg232Gly	694C>G	R232G	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095875	CTC-CCC	Leu234Pro	701T>C	L234P	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM992969	AGC-AGA	Ser236Arg	708C>A	S236R	Holoprosencephaly	Nanni (1999) <i>Hum Mol Genet</i> 8:2479	PubMed 10556296
SHH	CM095876	AGC-AAC	Ser236Asn	707G>A	S236N	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM146116	ACT-CCT	Thr240Pro	718A>C	T240P	Holoprosencephaly	Savastano (2014) <i>Genet Mol Biol</i> 37:250	PubMed 24764759
SHH	CM118763	TTC-CTC	Phe241Leu	721T>C	F241L	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SHH	CM095878	TTC-TTG	Phe241Leu	723C>G	F241L	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532

SHH	CM095877	TTC-GTC	Phe241Val	721T>G	F241V	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095879	ATC-AAC	Ile255Asn	764T>A	I255N	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM992970	GAG-TAG	Glu256Term	766G>T	E256*	Holoprosencephaly	Nanni (1999) <i>Hum Mol Genet</i> 8:2479	PubMed 10556296
SHH	CM044941	ACC-ATC	Thr267Ile	800C>T	T267I	Holoprosencephaly	Hehr (2004) <i>Eur J Pediatr</i> 163:347	PubMed 15107988
SHH	CM041836	CTG-CCG	Leu271Pro	812T>C	L271P	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SHH	CM095880	GCG-GAG	Ala275Glu	824C>A	A275E	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM066234	TCG-TAG	Ser280Term	839C>A	S280*	Holoprosencephaly	Richieri-Costa (2006) <i>Am J Med Genet A</i> 140A:2587	PubMed 17001669
SHH	CM095881	TCG-TGG	Ser280Trp	839C>G	S280W	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM971376	GAG-TAG	Glu284Term	850G>T	E284*	Holoprosencephaly	Roessler (1997) <i>Hum Mol Genet</i> 6:1847	PubMed 9302262
SHH	CM992971	GGC-GAC	Gly290Asp	869G>A	G290D	Holoprosencephaly	Nanni (1999) <i>Hum Mol Genet</i> 8:2479	PubMed 10556296
SHH	CM095882	GGG-GCG	Gly296Ala	887G>C	G296A	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095884	CGC-TGC	Arg310Cys	928C>T	R310C	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095885	CGT-AGT	Arg321Ser	961C>A	R321S	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM041837	GTG-GCG	Val332Ala	995T>C	V332A	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SHH	CM095886	GAG-TAG	Glu339Term	1015G>T	E339*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095887	GCG-GTG	Ala346Val	1037C>T	A346V	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM094632	CCG-CGG	Pro347Arg	1040C>G	P347R	Holoprosencephaly	Bendavid (2009) <i>Hum Mutat</i> 30:1175	PubMed 19431187
SHH	CM041838	CCG-CAG	Pro347Gln	1040C>A	P347Q	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SHH	CM095888	CCG-CTG	Pro347Leu	1040C>T	P347L	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095889	CAG-TAG	Gln351Term	1051C>T	Q351*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM041839	ATT-ACT	Ile354Thr	1061T>C	I354T	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SHH	CM126232	AAC-AAA	Asn357Lys	1071C>A	N357K	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM076517	TCG-TTG	Ser362Leu	1085C>T	S362L	Holoprosencephaly	Peng (2007) <i>Fetal Diagn Ther</i> 22:389	PubMed 17556830
SHH	CM126233	TCG-TAG	Ser362Term	1085C>A	S362*	Holoprosencephaly	Solomon (2012) <i>J Med Genet</i> 49:473	PubMed 22791840
SHH	CM066235	TGC-TAC	Cys363Tyr	1088G>A	C363Y	Holoprosencephaly	Richieri-Costa (2006) <i>Am J Med Genet A</i> 140A:2587	PubMed 17001669
SHH	CM095890	TAC-TGC	Tyr364Cys	1091A>G	Y364C	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM044943	GCG-ACG	Ala373Thr	1117G>A	A373T	Holoprosencephaly	Hehr (2004) <i>Eur J Pediatr</i> 163:347	PubMed 15107988
SHH	CM095891	CAC-CGC	His374Arg	1121A>G	H374R	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095892	GCC-GAC	Ala376Asp	1127C>A	A376D	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095893	TTC-TCC	Phe377Ser	1130T>C	F377S	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM041840	CGC-CCC	Arg381Pro	1142G>C	R381P	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SHH	CM095894	CTG-CCG	Leu382Pro	1145T>C	L382P	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM971377	GCG-ACG	Ala383Thr	1147G>A	A383T	Holoprosencephaly	Roessler (1997) <i>Hum Mol Genet</i> 6:1847	PubMed 9302262
SHH	CM095895	GCG-ACG	Ala391Thr	1171G>A	A391T	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM095896	ACC-GCC	Thr416Ala	1246A>G	T416A	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM992972	CCG-GCG	Pro424Ala	1270C>G	P424A	Holoprosencephaly	Nanni (1999) <i>Hum Mol Genet</i> 8:2479	PubMed 10556296
SHH	CM095897	TAC-AAC	Tyr435Asn	1303T>A	Y435N	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SHH	CM992973	TCG-TTG	Ser436Leu	1307C>T	S436L	Holoprosencephaly	Nanni (1999) <i>Hum Mol Genet</i> 8:2479	PubMed 10556296
SHH	CM095898	GGC-CGC	Gly456Arg	1366G>C	G456R	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E921	PubMed 19603532
SIX3	CM1210361	GGC-CGC	Gly34Arg	100G>C	G34R	Holoprosencephaly	Solomon (2012) <i>Mol Syndromol</i> 3:140	PubMed 23112757
SIX3	CM086940	GGT-TGT	Gly37Cys	109G>T	G37C	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM064275	GGC-GAC	Gly69Asp	206G>A	G69D	Holoprosencephaly	Ribeiro (2006) <i>Am J Med Genet A</i> 140A:2577	PubMed 17001667

SIX3	CM064276	GCC-CCC	Ala72Pro	214G>C	A72P	Holoprosencephaly	Ribeiro (2006) <i>Am J Med Genet A</i> 140A:2577	PubMed 17001667
SIX3	CM118745	ATG-CTG	Met79Leu	235A>C	M79L	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SIX3	CM093337	ATG-GTG	Met79Val	235A>G	M79V	Holoprosencephaly	Lacbawan (2009) <i>J Med Genet</i> 46:389	PubMed 19346217
SIX3	CM122195	CAG-TAG	Gln91Term	271C>T	Q91*	Holoprosencephaly	Roessler (2012) <i>Mol Genet Metab</i> 105:658	PubMed 22310223
SIX3	CM041841	GTG-GGG	Val92Gly	275T>G	V92G	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SIX3	CM086944	GCC-GAC	Ala93Asp	278C>A	A93D	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM064274	GAC-GGC	Asp104Gly	311A>G	D104G	Holoprosencephaly	Ribeiro (2006) <i>Am J Med Genet A</i> 140A:2577	PubMed 17001667
SIX3	CM041842	ATC-GTC	Ile105Val	313A>G	I105V	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SIX3	CM118746	CGG-TGG	Arg107Trp	319C>T	R107W	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SIX3	CM092397	TGG-TGT	Trp113Cys	339G>T	W113C	Holoprosencephaly	Solomon (2009) <i>Am J Med Genet A</i> 149A:919	PubMed 19353631
SIX3	CM122196	TGG-TAG	Trp113Term	338G>A	W113*	Holoprosencephaly	Roessler (2012) <i>Mol Genet Metab</i> 105:658	PubMed 22310223
SIX3	CM086951	TGG-TGA	Trp113Term	339G>A	W113*	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM086949	TCG-TTG	Ser114Leu	341C>T	S114L	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM118747	TCG-TGG	Ser114Trp	341C>G	S114W	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SIX3	CM086934	GAG-TAG	Glu129Term	385G>T	E129*	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM086948	TCG-TAG	Ser130Term	389C>A	S130*	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM118748	GCG-ACG	Ala134Thr	400G>A	A134T	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SIX3	CM064273	CGC-CCC	Arg135Pro	404G>C	R135P	Holoprosencephaly	Ribeiro (2006) <i>Am J Med Genet A</i> 140A:2577	PubMed 17001667
SIX3	CM086946	GTC-GAC	Val138Asp	413T>A	V138D	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM086947	TTC-ATC	Phe157Ile	469T>A	F157I	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM102533	GCC-TCC	Ala167Ser	499G>T	A167S	Holoprosencephaly	Hehr (2010) <i>Hum Genet</i> 127:555	PubMed 20157829
SIX3	CM086945	GCG-GTG	Ala172Val	515C>T	A172V	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM041843	CAC-CCC	His173Pro	518A>C	H173P	Holoprosencephaly	Dubourg (2004) <i>Hum Mutat</i> 24:43	PubMed 15221788
SIX3	CM086941	TAC-CAC	Tyr174His	520T>C	Y174H	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM146117	TAC-TAA	Tyr174Term	522C>A	Y174*	Holoprosencephaly	Savastano (2014) <i>Genet Mol Biol</i> 37:250	PubMed 24764759
SIX3	CM041844	ACC-ATC	Thr202Ile	605C>T	T202I	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
SIX3	CM086935	GAG-TAG	Glu207Term	619G>T	E207*	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM118749	TGC-TAC	Cys212Tyr	635G>A	C212Y	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SIX3	CM086942	TTC-GTC	Phe213Val	637T>G	F213V	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM086937	CGG-CCG	Arg218Pro	653G>C	R218P	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM086950	CGG-TGG	Arg218Trp	652C>T	R218W	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM991132	CTA-GTA	Leu226Val	676C>G	L226V	Holoprosencephaly	Wallis (1999) <i>Nat Genet</i> 22:196	PubMed 10369266
SIX3	CM086932	CAG-CCG	Gln227Pro	680A>C	Q227P	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM041845	CCC-CGC	Pro231Arg	692C>G	P231R	Holoprosencephaly	Dubourg (2004) <i>Hum Mutat</i> 24:43	PubMed 15221788
SIX3	CM086943	CAG-TAG	Gln241Term	721C>T	Q241*	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM086939	GGC-TGC	Gly244Cys	730G>T	G244C	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM991133	GTA-GCA	Val250Ala	749T>C	V250A	Holoprosencephaly	Wallis (1999) <i>Nat Genet</i> 22:196	PubMed 10369266
SIX3	CM086952	TTT-TTA	Phe254Leu	762T>A	F254L	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM118750	TTT-GTT	Phe254Val	760T>G	F254V	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
SIX3	CM093346	CGG-GGG	Arg257Gly	769C>G	R257G	Holoprosencephaly	Lacbawan (2009) <i>J Med Genet</i> 46:389	PubMed 19346217
SIX3	CM991134	CGG-CCG	Arg257Pro	770G>C	R257P	Holoprosencephaly	Wallis (1999) <i>Nat Genet</i> 22:196	PubMed 10369266
SIX3	CM041846	CGG-TGG	Arg257Trp	769C>T	R257W	Holoprosencephaly	Dubourg (2004) <i>Hum Mutat</i> 24:43	PubMed 15221788
SIX3	CM086938	CGG-CTG	Arg258Leu	773G>T	R258L	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198

SIX3	CM116764	CGG-TGG	Arg258Trp	772C>T	R258W	Holoprosencephaly	Petracchi (2011) <i>Prenat Diagn</i> 31:887	PubMed 21706511
SIX3	CM086936	CGC-CAC	Arg262His	785G>A	R262H	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM093348	AGG-ATG	Arg269Met	806G>T	R269M	Holoprosencephaly	Lacbawan (2009) <i>J Med Genet</i> 46:389	PubMed 19346217
SIX3	CM086931	AGG-AGC	Arg269Ser	807G>C	R269S	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM093347	AGG-ACG	Arg269Thr	806G>C	R269T	Holoprosencephaly	Lacbawan (2009) <i>J Med Genet</i> 46:389	PubMed 19346217
SIX3	CM093350	GCC-CCC	Ala284Pro	850G>C	A284P	Holoprosencephaly	Lacbawan (2009) <i>J Med Genet</i> 46:389	PubMed 19346217
SIX3	CM086933	CCG-CTG	Pro297Leu	890C>T	P297L	Holoprosencephaly	Domene (2008) <i>Hum Mol Genet</i> 17:3919	PubMed 18791198
SIX3	CM093349	GAC-GAT	Asp315Asp	945C>T	D315D	Holoprosencephaly	Lacbawan (2009) <i>J Med Genet</i> 46:389	PubMed 19346217
SIX3	CM065464	ACC-ATC	Thr316Ile	947C>T	T316I	Holoprosencephaly	Ribeiro (2006) <i>Am J Med Genet A</i> 140A:2577	PubMed 17001667
SLC26A2	CM010430	CTG-CCG	Leu132Pro	395T>C	L132P	Achondrogenesis 1B	Rossi (2001) <i>Hum Mutat</i> 17:159	PubMed 11241838
SLC26A2	CM960496	CGA-TGA	Arg178Term	532C>T	R178*	Achondrogenesis 1B	Superti-Furga (1996) <i>Nat Genet</i> 12:100	PubMed 8528239
SLC26A2	CM960499	AAT-GAT	Asn425Asp	1273A>G	N425D	Achondrogenesis 1B	Superti-Furga (1996) <i>Nat Genet</i> 12:100	PubMed 8528239
SLC26A2	CM962525	CTT-CCT	Leu483Pro	1448T>C	L483P	Achondrogenesis 1B	Rossi (1996) <i>J Biol Chem</i> 271:18456	PubMed 8702490
SLC26A2	CM960500	GGA-GTA	Gly678Val	2033G>T	G678V	Achondrogenesis 1B	Superti-Furga (1996) <i>Nat Genet</i> 12:100	PubMed 8528239
SOS1	CM109458	ACA-GCA	Thr158Ala	472A>G	T158A	syndrome	Tumurkhuu (2010) <i>Pediatr Int</i> 52:557	PubMed 20030748
SOS1	CM116049	TTC-TGC	Phe78Cys	233T>G	F78C	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM100895	CCT-CGT	Pro102Arg	305C>G	P102R	Noonan syndrome	Denayer (2010) <i>Genes Chromosomes Cancer</i> 49:242	PubMed 19953625
SOS1	CM070271	GAA-AAA	Glu108Lys	322G>A	E108K	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM116033	CCT-CGT	Pro112Arg	335C>G	P112R	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM086966	AAG-GAG	Lys170Glu	508A>G	K170E	Noonan syndrome	Ko (2008) <i>J Hum Genet</i> 53:999	PubMed 19020799
SOS1	CM116034	ATA-ACA	Ile252Thr	755T>C	I252T	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM070270	ACA-AAA	Thr266Lys	797C>A	T266K	Noonan syndrome	Roberts (2007) <i>Nat Genet</i> 39:70	PubMed 17143285
SOS1	CM070275	ATG-AGG	Met269Arg	806T>G	M269R	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM074570	ATG-ACG	Met269Thr	806T>C	M269T	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:651	PubMed 17586837
SOS1	CM070284	GAT-TAT	Asp309Tyr	925G>T	D309Y	Noonan syndrome	Roberts (2007) <i>Nat Genet</i> 39:70	PubMed 17143285
SOS1	CM070283	TAT-TGT	Tyr337Cys	1010A>G	Y337C	Noonan syndrome	Roberts (2007) <i>Nat Genet</i> 39:70	PubMed 17143285
SOS1	CM123739	CCC-TCC	Pro340Ser	1018C>T	P340S	Noonan syndrome	Fahrner (2012) <i>Am J Med Genet A</i> 158A:1414	PubMed 22585553
SOS1	CM116035	ATG-GTG	Met422Val	1264A>G	M422V	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM116036	GAG-AAG	Glu424Lys	1270G>A	E424K	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM070287	TGG-CCG	Trp432Arg	1294T>C	W432R	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM070281	GAG-AAG	Glu433Lys	1297G>A	E433K	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM074572	GGA-AGA	Gly434Arg	1300G>A	G434R	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:651	PubMed 17586837
SOS1	CM070282	GGA-CGA	Gly434Arg	1300G>C	G434R	Noonan syndrome	Roberts (2007) <i>Nat Genet</i> 39:70	PubMed 17143285
SOS1	CM140989	ATT-AAT	Ile437Asn	1310T>A	I437N	Noonan syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
SOS1	CM116029	ATT-ACT	Ile437Thr	1310T>C	I437T	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM070286	TGT-TAT	Cys441Tyr	1322G>A	C441Y	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM105248	CAG-CCG	Gln477Arg	1430A>G	Q477R	Noonan syndrome	Longoni (2010) <i>Am J Med Genet A</i> 152A:2176	PubMed 20683980
SOS1	CM074571	CCA-CGA	Pro478Arg	1433C>G	P478R	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:651	PubMed 17586837
SOS1	CM116037	GGT-CGT	Gly482Arg	1444G>C	G482R	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM116039	CTT-CGT	Leu490Arg	1469T>G	L490R	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM105249	CGA-CAA	Arg497Gln	1490G>A	R497Q	Noonan syndrome	Longoni (2010) <i>Am J Med Genet A</i> 152A:2176	PubMed 20683980
SOS1	CM105345	AGT-AGG	Ser548Arg	1644T>G	S548R	Noonan syndrome	Hastings (2010) <i>Genes Chromosomes Cancer</i> 49:967	PubMed 20607846

SOS1	CM070280	AGT-CGT	Ser548Arg	1642A>C	S548R	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM116040	ACA-AAA	Thr549Lys	1646C>A	T549K	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM070279	CTG-CCG	Leu550Pro	1649T>C	L550P	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM070269	AGG-GGG	Arg552Gly	1654A>G	R552G	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM070273	AGG-AAG	Arg552Lys	1655G>A	R552K	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM116030	AGG-ATG	Arg552Met	1655G>T	R552M	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
SOS1	CM070274	AGG-AGC	Arg552Ser	1656G>C	R552S	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM078474	AGG-AGT	Arg552Ser	1656G>T	R552S	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:651	PubMed 17586837
SOS1	CM095735	AGG-ACG	Arg552Thr	1655G>C	R552T	Noonan syndrome	Beneteau (2009) <i>Eur J Hum Genet</i> 17:1216	PubMed 19352411
SOS1	CM122804	AGG-TGG	Arg552Trp	1654A>T	R552W	Noonan syndrome	Ezquieta (2012) <i>Rev Esp Cardiol</i> 65:447	PubMed 22465605
SOS1	CM074573	TTT-ATT	Phe623Ile	1867T>A	F623I	Noonan syndrome	Zenker (2007) <i>J Med Genet</i> 44:651	PubMed 17586837
SOS1	CM070277	CCA-CTA	Pro655Leu	1964C>T	P655L	Noonan syndrome	Roberts (2007) <i>Nat Genet</i> 39:70	PubMed 17143285
SOS1	CM070278	TAT-CAT	Tyr702His	2104T>C	Y702H	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM123741	GCA-ACA	Ala708Thr	2122G>A	A708T	Noonan syndrome	Fahrner (2012) <i>Am J Med Genet A</i> 158A:1414 Jongmans (2010) <i>Genes Chromosomes Cancer</i> 49:635	PubMed 22585553 PubMed 20461756
SOS1	CM102937	AAA-ATA	Lys728Ile	2183A>T	K728I	Noonan syndrome		PubMed 20461756
SOS1	CM070285	TGG-TTG	Trp729Leu	2186G>T	W729L	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM140998	ATC-AAC	Ile733Asn	2198T>A	I733N	Noonan syndrome	Lepri (2014) <i>BMC Med Genet</i> 15:	PubMed 24451042
SOS1	CM070276	ATC-TTC	Ile733Phe	2197A>T	I733F	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM070272	GAA-AAA	Glu846Lys	2536G>A	E846K	Noonan syndrome	Tartaglia (2007) <i>Nat Genet</i> 39:75	PubMed 17143282
SOS1	CM116032	CCA-CGA	Pro894Arg	2681C>G	P894R	Noonan syndrome	Lepri (2011) <i>Hum Mutat</i> 32:760	PubMed 21387466
TRIP11	CM139338	CGA-TGA	Arg253Term	757C>T	R253*	Achondrogenesis type 1A	Grigelioniene (2013) <i>Am J Med Genet A</i> 161:2554	PubMed 23956106
TRIP11	CM100903	CGA-TGA	Arg264Term	790C>T	R264*	Achondrogenesis type 1A	Smits (2010) <i>N Engl J Med</i> 362:206	PubMed 20089971
TRIP11	CM139339	CAG-TAG	Gln579Term	1735C>T	Q579*	Achondrogenesis type 1A	Grigelioniene (2013) <i>Am J Med Genet A</i> 161:2554	PubMed 23956106
TRIP11	CM100908	CGA-TGA	Arg1028Term	3082C>T	R1028*	Achondrogenesis type 1A	Smits (2010) <i>N Engl J Med</i> 362:206	PubMed 20089971
TRIP11	CM100904	CAG-TAG	Gln1160Term	3478C>T	Q1160*	Achondrogenesis type 1A	Smits (2010) <i>N Engl J Med</i> 362:206	PubMed 20089971
TRIP11	CM100905	CGA-TGA	Arg1167Term	3499C>T	R1167*	Achondrogenesis type 1A	Smits (2010) <i>N Engl J Med</i> 362:206	PubMed 20089971
ZIC2	CM041870	CAG-CCG	Gln36Pro	107A>C	Q36P	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
ZIC2	CM091687	GAC-AAC	Asp37Asn	109G>A	D37N	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM041871	CAG-TAG	Gln46Term	136C>T	Q46*	Holoprosencephaly	Dubourg (2004) <i>Hum Mutat</i> 24:43	PubMed 15221788
ZIC2	CM041872	GGA-TGA	Gly58Term	172G>T	G58*	Holoprosencephaly	Dubourg (2003) <i>Ann Biol Clin (Paris)</i> 61:679	PubMed 14711609
ZIC2	CM091688	CAG-TAG	Gln73Term	217C>T	Q73*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091689	GAC-AAC	Asp128Asn	382G>A	D128N	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM118768	CAC-TAC	His156Tyr	466C>T	H156Y	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
ZIC2	CM091690	GAG-TAG	Glu164Term	490G>T	E164*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM122194	CAG-TAG	Gln178Term	532C>T	Q178*	Holoprosencephaly	Roessler (2012) <i>Mol Genet Metab</i> 105:658	PubMed 22310223
ZIC2	CM091691	TAC-TAA	Tyr194Term	582C>A	Y194*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091692	CAG-TAG	Gln250Term	748C>T	Q250*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091693	TGG-TAG	Trp260Term	779G>A	W260*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091694	CAA-TAA	Gln265Term	793C>T	Q265*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091695	AGC-AAC	Ser272Asn	815G>A	S272N	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091697	CAC-CAG	His286Gln	858C>G	H286Q	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091696	CAC-CTC	His286Leu	857A>T	H286L	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091698	CAC-TAC	His286Tyr	856C>T	H286Y	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455

ZIC2	CM118769	CAC-AAC	His291Asn	871C>A	H291N	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
ZIC2	CM091699	CAC-TAC	His291Tyr	871C>T	H291Y	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091700	TGG-AGG	Trp304Arg	910T>A	W304R	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091701	TGG-TGA	Trp304Term	912G>A	W304*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM118770	TGT-TAT	Cys307Tyr	920G>A	C307Y	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
ZIC2	CM091702	GAG-TAG	Glu310Term	928G>T	E310*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091703	TTC-TGC	Phe314Cys	941T>G	F314C	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091705	CGC-CTC	Arg325Leu	974G>T	R325L	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091704	CGC-AGC	Arg325Ser	973C>A	R325S	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091706	CAC-TAC	His327Tyr	979C>T	H327Y	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM118771	GAG-GGG	Glu330Gly	989A>G	E330G	Holoprosencephaly	Mercier (2011) <i>J Med Genet</i> 48:752	PubMed 21940735
ZIC2	CM091707	TGC-TTC	Cys335Phe	1004G>T	C335F	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091708	AAG-TAG	Lys351Term	1051A>T	K351*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091709	CAG-TAG	Gln364Term	1090C>T	Q364*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091710	CGC-CCC	Arg373Pro	1118G>C	R373P	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091712	TAC-AAC	Tyr402Asn	1204T>A	Y402N	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091711	TAC-TAG	Tyr402Term	1206C>G	Y402*	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091713	ACG-AAG	Thr403Lys	1208C>A	T403K	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091714	CAC-CGC	His404Arg	1211A>G	H404R	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091715	CGG-TGG	Arg409Trp	1225C>T	R409W	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM091716	CAT-CAG	His415Gln	1245T>G	H415Q	Holoprosencephaly	Roessler (2009) <i>Hum Mutat</i> 30:E541	PubMed 19177455
ZIC2	CM105347	GGC-GAC	Gly483Asp	1448G>A	G483D	Holoprosencephaly	Paulussen (2010) <i>Eur J Hum Genet</i> 18:999	PubMed 20531442