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Location	Mutation	Base Change	Reference
Exon I	M1V	1A>G	Kolker et al, 2007
	---	11delG	Spector & Sharer, unpublished 2006
	---	89/90 delC	Mushimoto et al, 2011
Intron 1	IVS1+5G>T	---	Greenberg et al, 1995
Exon II	---	109-110delCA	Tsai et al, 2017
Intron 2	IVS2+1G>A	---	Venturoni et al, 2014
	IVS2-1G>C	---	Venturoni et al, 2014
	IVS2-2A>C	---	Zschocke et al, 2000
	IVS2-2A>T	---	Christensen et al, 2004
Exon III	---	146-149del4	Wen et al, 2012
	W50R	148T>C	Wang et al, 2014
	W50R	148T>A	Mosaeilhy et al, 2017
	W50X	149G>A	Spector et al, unpublished, 2015
	W50C	150G>C	Zschocke et al, 2000
	---	152-153insA	Goodman et al, 2005
	P53S	157C>T	Christensen et al, 2004
	P53Q	157C>A	Mosaeilhy et al, 2017
	---	158delC	Goodman et al, 1998
	---	174insG	Christensen et al, 2004
	E64D	192G>T	Goodman, unpublished 2001
	ΔL66	196-198delCTC	Scharer (2005)
	---	198delC	Spector et al, 2015
	F71S	212T>C	Busquets et al, 2000a
	R72G	214G>C	Lewis B, unpublished 2007
	R72L	215G>T	Mushimoto et al, 2011
	---	219delC	Zschocke et al, 2000
	C75G	223T>G	Christensen et al, 2004
	C75S	224G>C	Spector, unpublished, 2014
	Q76X	226C>T	Zschocke et al, 2000
	Q76P	227A>C	Abdul Wahab et al, 2016
	R78G	232A>G	Spector & Sharer, unpublished 2005
	R82P	245G>C	Liu et al, 2015
	A86G	257C>G	Christensen et al, 2004
	R88S	262C>A	Venturoni et al, 2014
	R88C	262C>T	Biery et al, 1996
	R88H	263G>A	Couce et al, 2013
	E90K	268G>A	Busquets et al, 2000a
Intron 3	IVS3+1G>A	---	Tang et al, 2000
	IVS3-2A>C	---	Goodman et al, 2005
Exon IV	H93L	278A>T	---
	R94L	281G>T	Schwartz et al, 1998
	R94Q	281G>A	Gupta et al, 2015
	M100I	300G>?	Spector, unpublished, 2012
	G101R	301G>A	Anikster et al, 1996
	G112X	334G>T	Zhang et al, 2016

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Intron 4	IVS4+2T>C	---	Mushimoto et al, 2011
	IVS4+5G>A	---	Goodman et al, 1998
Exon V	Y113H	337T>C	Zschocke et al, 2000
	C115Y	344G>A	Goodman et al, 1998
	G117R	349G>C	Christensen et al, 2004
	S119L	356C>T	Busquets et al, 2000a
	A122V	365C>T	Goodman et al, 1998
	G124E	371G>A	Wang et al, 2014
	R128G	382C>G	Goodman et al, 1998
	R128X	382C>T	Busquets et al, 2000a
	R128Q	383G>A	Zschocke et al, 2000
	---	387-388delGC	Busquets et al, 2000a
	E131V	392A>T	Abdul Wahab et al, 2016
	R132G	394C>G	Bahr et al, 2002
	R132Q	395G>A	Zschocke et al, 2000
	V133M	397G>A	Zschocke et al, 2000
	V133L	397G>T	Venturoni et al, 2014
	D134G	401A>G	Gupta et al, 2015
	G136C	406G>T	Wang et al, 2014
	Y137X	411C>G	Wang et al, 2014
	R138G	412A>G	Goodman et al, 1998
	R138K	413G>A	Zschocke et al, 2000
	S139L	416C>T	Goodman et al, 1998
	S139W	416C>G	Wang et al, 2014
	---	420del10	Wen et al, 2012
	---	423-424delGC	Christensen et al, 2004
	Q144P	431A>C	Kruthica-Vinod et al, 2017
	---	433delT	Kalkanoglu et al, 2003
	S146F	437C>T	Spector & Sharer, unpublished 2005
	S146Y	437C>A	Spector & Sharer, unpublished 2005
	V148I	442G>A	Schwartz et al, 1998
	I152M	456C>G	Kruthika-Vinod et al, 2017
	Y155H	463T>C	Busquets et al, 2000a
	Y155C	464G>A	Goodman, unpublished, 2001
	Q160X	478C>T	Park et al, 2010
	Q160R	479A>G	Goodman, unpublished, 2001
	R161W	481C>T	Busquets et al, 2000c
	R161Q	482G>A	Biery et al, 1996
	Q162R	485A>G	Radha Rama Devi et al, 2015
Intron 5	IVS5+1G>A	---	Zschocke et al, 2000
	IVS5-2A>G	---	Mahfoud et al, 2004
Exon VI	K170E	508A>G	Wen et al, 2012
	G171W	511G>T	Kolker et al, 2007
	L174P	523T>C	Goodman et al, unpublished, 2005
	C176R	526T>C	Zschocke et al, 2000
	G178R	532G>A	Biery et al, 1996
	G178E	533G>A	Goodman et al, unpublished, 2004
	L179P	536T>C	Tehrani S et al, 2011
	L179R	536T>G	Goodman et al, 1998
	E181Q	541G>C	Zschocke et al, 2000

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Exon VI	E181K	541G>A	Busquets et al, 2000a
	E181G	542A>G	Spector & Sharer, 2006
	G185R	553G>A	Goodman, unpublished, 2001
	G185A	554G>C	Zschocke et al, 2000
	dG185-S190	---	Bross et al, 2012
	S186C	556A>T	Mushimoto et al, 2011
	S186I	557G>T	Spector & Sharer, 2006
	M191T	572T>C	Schwartz et al, 1998
	T193_R194insH	---	Korman et al, 2007
	A195T	583G>A	Schwartz et al, 1998
Intron 6	IVS6-1G>A	---	Zschocke et al, 2000
		636-10_642dup	Badve et al, 2015
Exon VII	T214A	640A>G	Wang et al, 2014
	T214M	641C>T	Goodman, unpublished, 2001
	---	644-645insCTCG	Moseilhy et al, 2016
	S216L	647C>T	Goodman, unpublished, 2003
	P217L	650C>T	Zschocke et al, 2000
	A219T	655G>A	Tang et al, 2000
	D220N	658G>A	Spector et al, 2008
	D220Y	658G>T	Park et al, 2010
	L221P	662T>C	Gupta et al, 2015
	W225X	675G>A	Radha Rama Devi et al, 2016
	A226T	676G>A	Goodman, unpublished, 2001
	R227P	680G>C	Biery et al, 1996
	C228R	682T>C	Kolker et al, 2007
	C228F	683G>T	Spector & Sharer, unpublished 2006
	R234W	700C>T	Spector & Sharer, unpublished 2006
	R234Q	701G>A	Spector & Sharer, unpublished 2006
	F236L	706T>C	Goodman et al, 1998
	L238P	713T>C	Lin et al, 2002
	G244C	730G>T	Goodman, unpublished, 2001
	G244S	730G>A	Mushimoto et al, 2011
	P248L	743C>T	Zschocke et al, 2000
	Q251X	751C>T	Spector & Sharer, unpublished 2005
	S255L	764C>T	Busquets et al, 2000a
	S255W	764C>G	Kolker et al, 2007
	L256P	767T>C	Spector, 2008
	R257W	769C>T	Schwartz et al, 1998
	R257Q	770G>A	Schwartz et al, 1998
	S259P	775T>C	Zschocke et al, 2000
	S259L	776C>T	Christensen et al, 2004
	M263V	787A>G	Muhlhausen et al, 2003
	M266V	796A>G	Goodman et al, 1998
	M266T	797T>C	Wen et al, 2012
	---	802-803insG	Goodman, unpublished, 2002
	P278S	832C>T	Schwartz et al, 1998
	P278R	833C>T	Spector & Sharer, 2006
	---	848delT	Busquets et al, 2000a
	L283P	848T>C	Anikster et al, 1996
Intron 7	IVS7+1G>A	---	Goodman et al, 1998

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Exon VIII	P286S	856C>T	Radha Rama Devi et al, 2016
	N291K	873C>A	Tsai et al, 2017
	---	873delC	Wang et al, 2014
	A293T	877G>A	Biery et al, 1996
	R294W	880C>T	Schwartz et al, 1998
	R294P	881G>C	Gupta et al, 2015
	R294Q	881G>A	Christensen et al, 2004
	Y295H	883T>C	Goodman et al, 1995
	G296S	886G>A	Spector, unpublished, 2014
	A298V	893C>T	Spector & Sharer, unpublished 2005
	A298T	894G>A	Christensen et al, 2004
	V301M	901G>A	Wang et al, 2014
	V301A	902T>C	Spector & Sharer, unpublished 2006
	G303R	907G>A	Goodman, unpublished 2001
	A304T	910G>A	Busquets et al, 2000a
	S305L	914C>T	Anikster et al, 1996
	E306G	917A>G	Spector, 2008
	C308R	922T>C	Kim et al, 2014
	C308S	923G>C	Goodman et al, 1998
	L309W	926T>G	Goodman et al, 1998
	---	932insC	Spector, unpublished, 2014
	R313W	937C>T	Goodman et al, 1998
	R313Q	938G>A	Zschocke et al, 2000
	A316D	947C>A	Busquets et al, 2000a
Intron 8	---	---	---
Exon IX	A327T	979G>A	Wang et al, 2014
	N329S	986A>G	Korman et al, 2007
	Q333E	997C>G	Goodman et al, 1998
	Q333X	997C>T	Goodman et al, 2005
	K335Q	1003A>C	Christensen, unpublished, 2005
	M339V	1015A>G	Ikeda et al, 1998
	L340F	1018C>T	Kalkanoglu et al, 2003
	T341P	1021A>C	Korman et al, 2007
	T341I	1022C>T	Spector & Sharer, 2006
	L347F	1039C>T	Goodman, unpublished, 2002
	A349T	1045G>A	Schwartz et al, 1998
	Q352X	1054C>T	Mushimoto et al, 2011
	G354R	1060G>C	Goodman et al, 1998
	G354S	1060G>A	Schwartz et al, 1998
	G354A	1061G>C	Mushimoto et al, 2011
	R355C	1063C>T	Goodman et al, 1998
	R355H	1064G>A	Schwartz et al, 1998
	L356P	???	Spector, 2008
	K361E	1081A>G	Mushimoto et al, 2011
	---	1084del161	Christensen et al, 2004
Intron 9	---	---	---

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Exon X	A362D	1085C>A	Goodman et al, 2005, unpublished
	E365K	1093G>A	Biery et al, 1996
	L369P	1106T>C	Scharer, 2007, unpublished
	R372K	1115G>A	Busquets et al, 2000a
	C375R	1123T>C	Goodman et al, 1998
	C375Y	1124G>A	Goodman et al, 2002
	A382T	1144G>A	Goodman et al, 1998
	---	1144-1145delGC	Mushimoto et al, 2011
	R383C	1147C>T	Goodman et al, 1998
	R383H	1148G>A	Goodman et al, 1998
	A385V	1154C>T	Zschocke et al, 2000
	R386X	1156C>T	Biery et al, 1996
	R386G	1156C>G	Tang et al, 2000
	R386Q	1157G>A	Goodman et al, 1998
	R386P	1157G>C	Goodman et al, unpublished, 2005
	---	1160-1173del	Schwartz et al, 1998
	M388T	1163T>C	Muhlhausen et al, unpublished, 2004
	G390R	1168G>C	Anikster et al, 1996
	G390W	1168G>T	Abdul Wahab et al, 2016
	G390A	1169G>C	Goodman et al, 1998
	G390V	1169G>T	Busquets et al, 2000b
	---	1172-1173insT	Wang et al, 2014
	---	1173delG	Anikster et al, 1996
	---	1173-1174insG	Spector & Scharer, 2006, unpublished
	N392D	1174A>G	Goodman et al, 1998
	E397X	1189G>T	Mosaeilhy et al, 2017
	Y398C	1193A>G	Couce et al, 2013
	V400M	1198G>A	Biery et al, 1996
	---	1198-1199insT	Goodman, unpublished, 2003
	R402W	1204C>T	Biery et al, 1996
	R402Q	1205G>A	Goodman et al, 1998
	---	1207delG	Christensen et al, 2004
	H403Y	1207C>T	Wang et al, 2014
	H403R	1208A>G	Goodman et al, 1998
	---	1209delG	Christensen et al, 2004
	---	1209-1210insG	Busquets et al, 2000c
	M405V	1213A>G	Goodman, unpublished, 2004
	---	1213dupA	Lisyova et al, 2016
	N406S	1217A>G	Goodman, unpublished, 2003
	N406K	1218C>G	Goodman et al, 1998
	---	1219del15	Christensen et al, 2004
	L407V	1219C>G	Pierson et al, 2015
	L407P	1220T>C	Goodman et al, 1998
	A409T	1225G>A	Lisyova et al, 2016
	V410M	1228G>A	Kolker et al, 2007
	Y413D	1237T>G	Mushimoto et al, 2011
	Y413C	1238A>G	Gupta et al, 2015
	Y413X	1239C>A	Zschocke et al, 2000
	E414K	1240G>A	Biery et al, 1996
	E414X	1240G>T	Kruthika-Vinod et al, 2017
	E414A	1241A>C	Gupta et al, 2015

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Intron 10	IVS10+1G>C	---	Goodman et al, 1998
	IVS10-2A>C	---	Tang et al, 2000
	IVS10-2A>G	---	Goodman et al, unpublished, 2005
Exon XI	T416I	1247C>T	Anikster et al, 1996
	A421T	1261G>A	Goodman et al, 1998
	A421V (Amish)	1262C>T	Biery et al, 1996
	---	1282-1285ins71	Wang et al, 2014
	I428M	1284C>G	Mosaeilhy et al, 2017
	T429M	1286C>T	Schwartz et al, 1998
	A433E	1298C>A	Schwartz et al, 1998
	A433V	1298C>T	Busquets et al, 2000a
	F434L	1300T>C/1302C>A	Christensen, 2005
	X439W	1317A>G	Busquets et al, 2000b

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