Exon/Intron	Trivial Name	Codon	Nucleotide Change*	Codon Changed	Systematic(cDNA based)	Systematic (genomic based)	Other names	Type
2	M-1T	-1	CCA**T**GGC>CCA**C**GGC	Met>Thr	c.2T>C	g.4895T>C	M1T	transition
2	R3Op	3	CAC**C**GAT>CAC**T**GAT	Arg>Opal	c.10C>T	g.4903C>T	R3X, R3ter	transition
2	ΔA20	20	CCC[**A**]GAG>CCCGAG	Gln>Arg (fs)	c.62delA	g.4955delA (first base of deletion)	ΔA20	deletion
IVS2	IVS2-1G>A		ata**g**GTA>ata**a**GTA			g.5814G>A	c.113-1G>A	transition
3	IVS2-1Δ4	37-38	ata[**gGTA**]CCA>ataCCA			g.5813delGGTA	G12/Δ3;IVS2-delGGTA;Δ4IVS2-E3	deletion
3	R59op	59	TTC**C**GAG>TTC**T**GAG	Arg>Opal	c.178C>T	g.5880C>T	R59X;R59ter	transition
3	I73T	73	GC**A**TCGG>GCA**C**CGG	Ile>Thr	c.221T>C	g.5923T>C		transition
3	L83ΔC	83	C**C**T[**C**]TAC>C**C**TTAC	Leu>Leu(fs)	c.252delC	g.5954delC	c.250 delC	deletion
3	1123ins12	104	GGT[]GGG>GGT[**GGGGATCGTGGT**]GGG		c.314^315ins12	g.6016^6017insGGGGATCGTGGT	−12E3;V104+GIVV	insertion
IVS3	K107KΔ12	104-107del	GTG[GTGGGAATCA**AG**]gtt>GTGgtgggaatcaa**a**gtt		c.324G>A;delGTGGGAATCAAG(312-324)	g.6026G>A	g.1133G>A	transition
IVS3	IVS3-1G>A		aca**g**TTA>aca**a**TTA			g.7257G>A	c.325-1G>C, g.8180G>C,IVS3sas	transversion
4	Δ28E4	114-151	T**C**C[**TCTTGCAGGAACAAACAAAGAAACCACC**]ATT>T**C**CATT	Pro>Pro(fs)	c.345-372del28	g.7277^7305del	c.345_72del28	deletion
4	Q110Oc	110	GAC**C**AAG>GAC**T**AAG	Gln>Ochre	c.331C>T	g.7264C>T		transition
4	Δ4E4	118-120	GAAC[**AAAC**]AAAG>GAACAAAG		c.357delAAAC	g.7289delAAAC	Δ4, MDΔ4	deletion
4-5	ΔE4-E5		ccc[**tgt...TCC**]AGC>cccAGC			g.6594^8239del	F13/Δ4,5	deletion
5	C134R	134	CGC**T**GTG>CGC**C**GTG	Cys>Arg	c.403T>C	g.8162T>C	C135R	transition
5	W147R	147	AAG**T**GGC>AAG**C**GGC	Trp>Arg	c.442T>C	g.8201T>C	W148R	transition
5	A149P	149	CGT**G**CTG>CGT**C**CTG	Ala>Pro	c.448G>C	g.8212G>C	A150P	transversion
5	Y173Am	173	CTA**C**GCC>CTA**G**GCC	Tyr>Amber	c.522C>G	g.8281C>G	Y174X	transversion
5	A174D	174	ACG**C**CAG>ACG**A**CAG	Ala>Asp	c.524C>A	g.8283C>A	A175D	transversion
5	C177R	177	ATC**T**GTC>ATC**C**GTC	Cys>Arg	c.532T>C	g.8291T>C	p.C178R	transition
6-7	ΔE6-E7		atc[**tca...agc**]act>atcact			g.8989^10457del	G10/Δ6,7	deletion
IVS5	IVS5+1G>C		CAG**g**tgc>CAG**c**tgc			g.8300G>C	G>C,5'intron5;IVS5nt1	transversion
6	Δ6E6		GGA[**CTGGTA**]CCT>GGACCT	LeuValdel	c.547-552delCTGGTA	g.9149-9154delCTGGTA	Leu182-Val183del	deletion
6	P184R	184	TA**C**CTAT>TA**C**GTAT	Pro>Arg	c.554C>G	g.9156C>G	p.P184R	transversion
6	Y203Oc	203	GTA**T**GTT>GTA**A**GTT	Tyr>Ochre	c.612T>A	g.9214T>A	Y203X;Y203term	transversion
6	Y203Am	203	GTA**T**GTT>GTA**G**GTT	Tyr>Amber	c.612T>G	g.9214T>G	p.Y204X	transversion
IVS6	IVS6-2A>G		ttc**a**gGT>ttc**g**gGT			g.10152A>G		transition
IVS6	IVS6-1G>A		tca**g**GTC>tca**a**GTC			g.10153G>A	G>A,3'intron6;IVS6sas	transition
7	V221F	221	CAT**G**TTT>CAT**T**TTT	Val>Phe	c.664G>T	g.10193G>T	g.10236G>T; p.V222F	transversion
7	L228P	228	TGC**T**AAA>TGC**C**AAA	Leu>Pro	c.686T>C	g.10215T>C	g.10258T>C; p.L229P	transition
7	C239Op	239	CTG**C**ACC>CTG**A**ACC	Cys>Opal	c.720C>A	g.10249C>A	M240;C239X;C239ter	transversion
7	L256P	256	CTC**T**CCA>CTC**C**CCA	Leu>Pro	c.770T>C	g.10299T>C		transition
IVS7	IVS7+2T>A		TGg**t**aag>TGg**a**aag			g.10333T>A	c.799+2T>A	transversion
8	Δ2E8	280	GCC[**AC**]TCT>GCC**T**CT	Thr>Ser (fs)	c.841-842delAC	c.10780-10781delAC	c.841_842delAC	deletion
8	L283P	283	ACC**T**CAA>ACC**C**CAA	Leu>Pro	c.851T>C	g.10790T>C	p.L284P	transition
8	L288ΔC	288	AAC[**C**]TTT>AAC**T**TT	Leu>Phe (fs)	c.865delC	g.10804delC	L288delC	deletion
8	R303W	303	GGA**C**GGG>GGA**T**GGG	Arg>Trp	c.910C>T	g.10849C>T		transition
8	R303Q*	303	GA**C**GGGC>GA**C**AGGC	Arg>Gln	c.911G>A	g.10850G>A		transition
IVS8	Δ7+1	333-334	cta[**gGCTAAC**]TGCCA>ctaTG[**G**]CCA		[c.1000-1005delGGCTAAC;c.1007^1008insG]	[g.13875delGGCTAAC;g.13884^13885insG]		indel
9	N334K	334	TAA**C**TGC>TAA**G**TGC	Asn>Lys	c.1005C>G	g.13882C>G		transversion
9	A337V	337	AGG**C**GGC>AGG**T**GGC	Ala>Val	c.1013C>T	g.13890C>T		transition
9	Δ6+5	347-349	TTC[**TTCTGG**]GGC>TTC[**ACACT**]GGC	SerSerGly>Ser	[c.1043delTTCTGG;c.1043^1044insACACT	[g.13920delTTCTGG;g.13920^13921insACACT]	c.1044_1049delTTCTGGinsACACT	indel
			*(Uppercase denotes protein coding sequence, lowercase denotes non-coding sequence; change is in bold; any codons are underlined; all are given within a 7-base context)					

Consequence	**Validaton**	**Frequency**
missense	DNA only	private
nonsense	DNA only	private
frameshift	DNA only	0.002
splicing defect	DNA only	private
splicing defect	DNA only	private
nonsense	DNA only	0.02
missense	Protein expression only	private
frameshift	DNA only	private
insertion of 4 amino acids	DNA only	private
missense leading to splicing defect leading to deletion	RNA expression analysis	private
splicing defect	DNA only	private
frameshift	DNA only	private
nonsense	DNA only	private
frameshift	Activity measured in liver biopsy from homozygote	0.03
large deletion	DNA only	private
missense	Protein expressed, partial activity, unstable	private
missense	Protein expressed, partially active	private
missense	Protein expressed, partial activity, unstable, crystal structure done	0.57
nonsense	DNA only	private
missense	Protien expressed, unstable; Activity measured in liver biopsy from homozygote	0.13
missense	DNA only	private
large deletion	DNA only	private
splicing defect	DNA only	private
deletion of 2 amino acids	DNA only	private
missense	DNA only	private
nonsense	DNA only	private
nonsense	DNA only	private
splicing defect	DNA only	private
splicing defect	DNA only	private
missense	Protein expression only	private
missense	Protein expression only	private
nonsense	Protein expressed	private
missense	Protein expressed, partially active (mild)	0.01
splicing defect	DNA only	private
frameshift	DNA only	private
missense	DNA only	private
frameshift	DNA only	0.003
missense	Protein expressed, decreased activity	private
missense	*Protein expressed, active	private/poly?
splicing defect	RNA splicing analysis confirmation	private
missense	Protein expressed, partially active	0.05
missense	Protein expressed, mild defect, liver biopsy of homozygote showed decreased activity	0.01
frameshift	DNA only	private

Comments	Reference
initiation codon	Ali et al, 1993
CpG dinucleotide, opal stop codon	Ali et. al., 1994b
only in Italy	Santamaria, 1996
	Santer, 2005
3'-endIVS2 acc spl site, frameshift	Cross and Cox,1990
CpG dinucleotide, opal stop codon	Brooks and Tolan, 1994
Protein insoluble	Esposito, 2004
	Gruchota, 2006
	Pappas and Tolan, 1997 (#34)
	Sanchez, 2002
	Esposito, 2004
stop codon at aa 151	Santer, 2005
	Esposito, 2004
	Dazzo and Tolan, 1990
1646 bp deletion from IVS3 to part of E5	Cross and Cox,1990
	Brooks and Tolan, 1994
	Ali and Cox, 1995
Most common, most well studied	Cross et al, 1988
	Gruchota, 2006
	Cross et al, 1990a
	Santer, 2005
1469 bp deletion from IVS5 to IVS7	Cross and Cox,1990
	Ali et al, 1996
	Santamaria, 1999
	Sanchez-Gutierrez, 2002
ochre stop codon	Ali et al, 1993
	Santer, 2005
	Esposito, 2004
3'-end of IVS6 acc splice site	Ali et. al., 1994b
Protein insoluble	Esposito, 2004
Protein insoluble	Esposito, 2004
opal stop codon	Kajihara et al, 1990
	Ali et al, 1994a
	Santer, 2005
	Santer, 2005
	Santer, 2005
only in Sicily	Cross et al, 1990a
CpG dinucleotide	Santamaria, 1996
	Santamaria, 2000
3'-end of IVS8 acc splice site and codons333-334 deleted, plus G insertion at codon 335, expression of RNA showed splicing defect	Brooks et al, 1991
	Cross et al, 1990b
CpG dinucleotide, expression showed alterations in activity	Rellos et al, 1999
deletion of 6, insertion of 5, stop codon at 369	Santer, 2005