

Table 2. Point Mutations Seen in Individuals with Friedreich Ataxia Who Are Compound Heterozygous

Exon/ Intron	Mutation	Nt Position	NT Change	Predicted Effect on Frataxin	Reference
Exon 1	M1L	1	ATG>CTG	Aberrant initiation of translation	Cossee et al 1999
Exon 1	M1T	2	ATG>ACG	Aberrant initiation of translation	Cossee et al 1999
Exon 1	2delT	2	ATG>A-G	Aberrant initiation of translation	Bidichandani et al, unpublished; Zhu et al 2002
Exon 1	M1I	3	ATG>ATT	Aberrant initiation of translation	Cossee, Campuzano et al 1997
Exon 1	M1I	3	ATG>ATA	Aberrant initiation of translation	Potter et al 2000
Exon 1	118delC	118	del C	Premature termination of translation	De Castro et al 2000
Exon 1	158delC	158	del C	Premature termination of translation	Cossee et al 1999
Exon 1	158insC	158	ins C	Premature termination of translation	Cossee et al 1999
Intron 1	Splice donor	+5	G>C	Aberrant splicing	McCormack et al 2000
Exon 2	202GTCA>TTG	202-205	GTCA>TTG	Premature termination of translation	Pook et al 2000
Exon 3	297insT	297	ins T	Premature termination of translation	De Castro et al 2000

Exon/ Intron	Mutation	Nt Position	NT Change	Predicted Effect on Frataxin	Reference
Exon 3	L106X	317	TTA>TGA	Premature termination of translation	Campuzano et al 1996
Exon 3	L106S	317	TTA>TCA	Missense	Bartolo et al 1998
Exon 3	317delT	317	del T	Premature termination of translation	Pook et al 2000
Exon 3	340del13	340-352	del of 13 bp	Premature termination of translation	Pook et al 2000
Exon 3	D122Y	364	GAC>TAC	Missense	Cossee et al 1999
Intron 3	Splice donor	384+1	AG>GG	Aberrant splicing	Doudney et al 1998
Intron 3	Splice acceptor	-2	AG>GG	Frameshift caused by aberrant splicing of exon 4	Campuzano et al 1996
Exon 4	G130V ¹	389	G130V 1	Missense; aberrant processing	Bidichandani et al 1997, Koutnikova et al 1998
Exon 4	Q148R	443	CAG>CGG	Missense	McDaniel et al 2003
Exon 4	I154Fd 1	460	ATC>TTC	Missense; aberrant processing	Campuzano et al 1996, Koutnikova et al 1998
Exon 4	W155X	465	TGG>TAG	Premature termination of translation	De Castro et al 2000
Exon 4	L156P	467	CTA>CCA	Missense	Cossee et al 1999
Intron 4	Splice donor	482+2	GT>GG	Frameshift caused by aberrant splicing of exon 4	Forrest et al 1998

Exon/ Intron	Mutation	Nt Position	NT Change	Predicted Effect on Frataxin	Reference
Intron 4	Splice donor	482+3	+3 del A	Frameshift caused by aberrant splicing of exon 4	Cossee et al 1999
Exon 5a	R165C ¹	493	CGT>TGT	Missense	Forrest et al 1998, McCormack et al 2000
Exon 5a	R165P	494	CGT>CCT	Missense	De Michele et al 2000
Exon 5a	W173G ¹	517	TGG>GGG	Missense	Cossee et al 1999
Exon 5a	L182F	544	CTC>TTC	Missense	Forrest et al 1998
Exon 5a	L182H	545	CTC>CAC	Missense	Cossee et al 1999
Exon 5a	H183R	548	CAT>CGT	Missense	Cossee et al 1999
Exon 5a	L198R	593	CTG>CGG	Missense	Al-Mahdawi et al 2000

1. Recurrent mutations