Known Mutations of the Electron Transfer Flavoprotein:Ubiquinone Oxidoreductase (ETF:QO) Gene:
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Location	Mutation	Base Change	Reference
Exon I	M1T	2T>C	Goodman et al, 2002
	A12P	34G>C	Goodman et al, unpublished, 2002
Intron 1	---	---	---
Exon II	Truncated protein	36del A	Olsen et al, 2003
	Truncated protein	45-46insC	Olsen et al, 2003
	F16C (Neutral)	47T>G	Goodman et al, 2002
	Truncated protein	51-52insT	Olsen et al, 2001
	P27S	79C>T	Goodman et al, unpublished, 2002
	I31T (Neutral)	92T>C	Goodman et al, 2002
	R41X	121C>T	Goodman et al, 2002
	Y49C	146A>G	Goodman et al, 2002
Intron 2	---	---	---
Exon III	S82P	244T>C	Goodman et al, 2002
	S82F	245C>T	Goodman et al, 2002
Intron 3	---	IVS3+3A>T	Olsen et al, 2004
Exon IV	L138R	413T>G	Goodman et al, 2002
	Truncated protein	427del7	Goodman et al, 2002
Intron 4	---	---	---
Exon V	W182X	545G>A	Goodman et al, 2002
Intron 5	---	---	---
Exon VI	D218N	652G>A	Goodman et al, 2002
	Q222P	665A>C	Goodman et al, 2002
Intron 6	---	---	---
Exon VII	G234R	700G>A	Goodman et al, unpublished, 2002
	L262F	786G>T	Goodman et al, 2002
	Q269L	806A>T	Olsen et al, 2001
Intron 7	---	---	---
Exon VIII	---	---	---
Intron 8	---	---	---
Exon IX	L334P	1001T>C	Goodman et al, 2002
	H346R	1037A>G	Goodman et al, 2002
	R358S	1074G>C	Olsen et al, 2003

Location	Mutation	Base Change	Reference
Intron 9	---	---	---
Exon X	---	---	---
Intron 10	---	---	---
Exon XI	R452K	1355G>A	Goodman et al, 2002
	P456L	1367C>T	Goodman et al, 2002
	Truncated protein	1392del2	Goodman et al, 2002
	G472R	1414G>A	Olsen et al, 2003
	L486P	1457T>C	Goodman et al, unpublished, 2002
Intron 11	---	---	---
Exon XII	Truncated protein	1623delT	Goodman et al, 2002
	P562L	1685C>T	Goodman et al, 2002
Intron 12	Skipping of exon 12	IVS12+1G>T	Goodman et al, 2002
Exon XIII	G546X	1726G>T	Goodman et al, unpublished, 2004
	G611E	1832G>A	Goodman et al, 2002

References:

Goodman SI, Binard RJ, Woontner MR, and Frerman FE: Glutaric acidemia type II: gene structure and mutations of the electron transfer flavoprotein:ubiquinone oxidoreductase (ETF:QO) gene. Molec Genet & Metab 77 (2002) 86-90.

Olsen RKJ, Andresen BS, Christensen E, Sunde L, Nielsen JP, and Gregersen N: Elucidation of the ETF/ETF-QO gene structures enables prenatal diagnosis of the mild form of multiple acyl-CoA dehydrogenation deficiency: DNA-based diagnosis in a pregnancy at risk. J Inherit Metab Dis 24 (2001) Suppl 1, 73 (Abst).

Olsen RK, Andresen BS, Christensen E, Bross P, Skovby F, and Gregersen N: Clear relationship between ETF/ETFDH genotype and phenotype in patients with multiple acyl-CoA dehydrogenation deficiency. Hum Mutat 22 (2003) 12-23.

Olsen RK, Pourfarzam M, Morris AA, Dias RC, Knudsen I, Andresen BS, Gregersen N, Olpin SE: Lipid-storage myopathy and respiratory insufficiency due to ETFQO mutations in a patient with late-onset multiple acyl-CoA dehydrogenation deficiency. J Inherit Metab Dis 27 (2004) 671-678.