GeneReviews Title: Mitochondrial Neurogastrointestinal Encephalopathy Disease Table 2

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Note: The following information is provided by the author listed above and has not been reviewed by *GeneReviews* staff.

Table 3. *OFD1* Allelic Variants

Exon	Variant	Codon	Amino Acid Change	Reference [1]
Exon 6	c.647C>T	Codon 216	Ala>Val	
Exon 7	c.831G>A	Codon 277	Synonymous	
Exon 8	c.966T>C	Codon 322	Synonymous	Hagiwara et al [1991]
Exon 8	c.972T>C	Codon 324	Synonymous	Nishino et al [1999]
Exon 8	c.1069A>G	Codon 357	Ala>Thr	Nishino et al [1999]
Exon 9	c.1284T>A	Codon 428	Synonymous	Kocaefe et al [2003]
Exon 10	c.1393G>A	Codon 465	Ala>Thr	
Exon 10	c.1401C>T	Codon 467	Synonymous	
Exon 10	c.1412C>T	Codon 471	Ser>Leu	Hagiwara et al [1991]
Exon 10	c.1432C>T	Codon 478	Synonymous	

1. Polymorphisms in exons are from the Single Nucleotide Polymorphism database, NCBI, geneID: 1890 except those designated with references.

References

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