

Title: Leber Hereditary Optic Neuropathy *GeneReview* Table 6

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

**Table 6. Putative Pathogenic Variants in Mitochondrial DNA Associated with LHON**

Mitochondrial DNA Nucleotide Change	Gene	Predicted Protein Change	Reference Sequences <sup>1</sup>
m.3376G>A	MT-ND1		
m.3697G>A	MT-ND1		
m.4025C>T	MT-ND1		
m.4160T>C	MT-ND1		
m.4640C>A	MT-ND2		
m.5244G>A	MT-ND2		
m.10237T>C	MT-ND3		
m.11696G>A	MT-ND4		
m.11253T>C	MT-ND4		
m.12811T>C	MT-ND5		
m.12848C>T	MT-ND5		
m.13637A>G	MT-ND5		
m.13730G>A	MT-ND5		
m.14325T>C	MT-ND6		
m.14568C>T	MT-ND6		
m.14729G>A	MT-ND6		
m.14498C>T	MT-ND6		
m.14596A>T	MT-ND6		
m.9101T>C	MT-ATP6	p.Ile192Thr	<a href="#">AC 000021.2</a> <a href="#">NP 536848.1</a>
m.9804G>A	MT-CO3	p.Ala100Thr	<a href="#">AC 000021.2</a> <a href="#">NP 536849.1</a>
m.14831G>A	MT-CYB	p.Ala29Thr	<a href="#">AC 000021.2</a> <a href="#">NP 536855.1</a>

See [Quick Reference](#) for an explanation of nomenclature. (Note: The mitochondrial genetic code varies from the genomic genetic code given in the Quick Reference. For the genetic code, gene structure, and other features of the mitochondrial genome see MITOMAP: A Human Mitochondrial Genome Database at [www.mitomap.org](http://www.mitomap.org), 2007). Variants are named according to current nomenclature guidelines ([www.hgvs.org](http://www.hgvs.org)).

The variants in these genes are thought to be pathogenic in LHON but require further confirmation, as they have been identified only in single affected individuals or a single family ([www.mitomap.org](http://www.mitomap.org)).

1. [AC 000021.2](#) is the mitochondrial DNA Cambridge reference sequence [Anderson et al 1982, Andrews et al 1999].

## References

Anderson S, de Bruijn MH, Coulson AR, Eperon IC, Sanger F, Young IG. Complete sequence of bovine mitochondrial DNA. Conserved features of the mammalian mitochondrial genome. *J Mol Biol* 1982;156:683-717.

Andrews RM, Kubacka I, Chinnery PF, Lightowlers RN, Turnbull DM, Howell N. Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. *Nat Genet* 1999;23:147.