Title: Leber Hereditary Optic Neuropathy *GeneReview* Table 5
Authors: Yu-Wai-Man P, Chinnery PF
Updated: September 2013
Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 5. Pathogenic Variants in Mitochondrial DNA Associated with LHON

% of Mutated Alleles	Mitochondrial DNA Nucleotide Change	Gene	Predicted Protein Change	Reference Sequences [1]
90%	m.11778G>A	*MT-ND4*	p.Arg340His	AC_000021.2 NP_536852.1
	m.14484T>C	*MT-ND6*	p.Met64Val	AC_000021.2 NP_536854.1
	m.3460G>A	*MT-ND1*	p.Ala53Thr	AC_000021.2 NP_536843.1
10% [2]	m.3635G>A			
	m.3700G>A			
	m.3733G>A		p.Glu143Lys	
	m.4171C>A		p.Leu289Met	
	m.10663T>C	*MT-ND4L*	p.Val65Ala	AC_000021.2 NP_536851.1
	m.14459G>A	*MT-ND6*	p.Ala72Val	AC_000021.2 NP_536854.1
	m.14482C>A			
	m.14482C>G		p.Met64Ile	
	m.14495A>G		p.Leu60Ser	
	m.14568C>T			

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, 2007). Variants are named according to current nomenclature guidelines (www.hgvs.org).

1. AC_000021.2 is the mitochondrial DNA Cambridge reference sequence [Anderson et al 1982, Andrews et al 1999].

2. This represents the group of patients with a clinical diagnosis of LHON but who are not found to harbor one of the three most common mtDNA pathogenic variants. This group also includes those mtDNA variants thought to cause LHON, but which require further confirmation for pathogenicity. See Table 6.

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.