Title: *OTOF*-Related Deafness *GeneReview* Table 3

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

Table 3. Selected *OTOF* Normal Allelic Variants

DNA Nucleotide Change	Protein Amino Acid Change (Alias [1])	Reference Sequences
c.158C>T	p.Ala53Val	NM_194248.1 NP_919224.1
c.244C>T [2]	p.Arg82Cys	
c.372A>G [2]	p.= [3] (Thr124Thr)	
c.945G>A [2]	p.= (Lys315Lys)	
c.1723G>A	p.Val575Met	
c.1926C>T	p.= (Asn642Asn)	
c.2022C>T	p.= (Asp674Asp)	
c.2025G>A	p.= (Glu675Glu)	
c.2317C>T [2]	p.Arg773Ser	
c.2464C>T	p.Arg822Trp	
c.2580C>G [2]	p.= (Val860Val)	
c.2736G>C [2]	p.= (Leu919Leu)	
c.3189G>A	p.= (Ala1063Ala)	
c.3247G>C	p.Ala1083Pro	
c.3470G>A	p.Arg1157Gln	
c.3966C>G	p.Asp1322Glu	
c.4677G>A	p.= (Val1559Val)	

DNA Nucleotide Change	Protein Amino Acid Change (Alias [1])	Reference Sequences
c.4767C>T	p.= (Arg1589Arg)	
c.4874G>A	p.Val1625Met	
c.4936C>T	p.Pro1646Ser	
c.5391C>T	p.= (Phe1797Phe)	
c.5655C>T	p.= (Arg1885Arg)	
c.5663G>A	p.Gly1888Asp	

See Quick Reference for an explanation of nomenclature. GeneReviews follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org).
1. Variant designation that does not conform to current naming conventions
2. Polymorphic allelic variations in bold have been described in both American and Spanish populations [Rodriguez-Ballesteros et al 2003, Varga et al 2003].
3. The designation p.= means that no amino acid change is expected.

References

Rodriguez-Ballesteros M, del Castillo FJ, Martin Y, Moreno-Pelayo MA, Morera C, Prieto F, Marco J, Morant A, Gallo-Teran J, Morales-Angulo C, Navas C, Trinidad G, Tapia MC, Moreno F, del Castillo I (2003) Auditory neuropathy in patients carrying mutations in the otoferlin gene (OTOF). Hum Mutat 22:451-6

Varga R, Kelley PM, Keats BJ, Starr A, Leal SM, Cohn E, Kimberling WJ (2003) Non-syndromic recessive auditory neuropathy is the result of mutations in the otoferlin (OTOF) gene. J Med Genet 40:45-50