Title: *OTOF*-Related Deafness *GeneReview* Table 4

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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 4. Selected *OTOF* Pathologic Allelic Variants

DNA Nucleotide Change (Alias [1])	Protein Amino Acid Change (Alias [1])	Reference	Reference Sequences
c.709C>T	p.Arg237X	Houseman et al [2001]	NM_194248.1 NP_919224.1
c.766-2A>G (IVS8-2A>G)	--	Yasunaga et al [2000]	
c.765G>C	p.Gln255His	Rodriguez-Ballesteros et al [2008]	
c.766-2T>A (IVS9-2T>A)		Zadro et al [2010]	
c.1103_1104delinsC	p.Gly368AlafsX2	Choi et al [2009]	
c.1180dupG	p.Glu394GlyfsX6	Rodriguez-Ballesteros et al [2008]	
c.1194 T>A	p.Asp398Glu	Wang et al [2010]	
c.1236delC	p.Glu413AsnfsX9	Rodríguez-Ballesteros et al [2008]	
c.1273C>T	p.Arg425X	Choi et al [2009]	
c.1469C>A	p.Pro490Gln	Mirghomizadeh et al [2002]	
c.1544T>C	p.Ile515Thr	Mirghomizadeh et al [2002]	
c.1552-1567del16		Romanos et al [1999]	
c.1601delC	p.Pro534GlnfsX4	Rodríguez-Ballesteros et al [2008]	
c.1607G>A	p.Trp536X	Choi et al [2009]	
c.1609delG	p.Val537X	Santarelli et al [2009]	
c.1651delG	p.Glu551SerfsX5	Varga et al [2003]	
c.1718T>G	p.Leu573Arg	Choi et al [2009]	
c.1740delC	p.Ser581ProfsX40	Wang et al [2010]	
c.1780G>A	p.Glu594Lys	Wang et al [2010]	
c.1841G>A	p.Gly614Glu	Romanos et al [2009]	

DNA Nucleotide Change (Alias [1])	Protein Amino Acid Change (Alias [1])	Reference	Reference Sequences
c.1886dupA (1886_1887insA)	p.Pro630AlafsX5 (Lys629fs)	Varga et al [2006]	
c.1912-2C>T (IVS17-2C>T		Zadro et al [2010]	
c.1966delC	p.Arg656GlyfsX10	Santarelli et al [2009]	
c.2122C>T	p.Arg708X	Rodriguez-Ballesteros et al [2003]	
c.2214+1G>T (IVS18+1G>T)	--	Varga et al [2006]	
c.2239G>T	p.Glu747X	Rodriguez-Ballesteros et al [2008]	
c.2295_2297delG	p.Glu766SerfsX	Choi et al [2009]	
c.2316C>A	p.Cys772X	Zadro et al [2010]	
c.2348delG	p.Gly783AlafsX17	Varga et al [2006]	
c.2381G>A	p.Arg794His	Varga et al [2006]	
c.2485C>T	p.Gln829X	Migliosi et al [2002], Rodriguez-Ballesteros et al [2003]	
c.2649C>A	p.Cys883X	Rodriguez-Ballesteros et al [2008]	
c.2684_2685delGG	p.Gly895GlufsX106	Rodriguez-Ballesteros et al [2008]	
c.2732_2735dupAGCT	p.Tyr913AlafsX90	Rodriguez-Ballesteros et al [2008], Santarelli et al [2009]	
c.2887C>T	p.Arg963X	Hutchin et al [2005]	
c.2891C>A	p.Ala964Glu	Rodriguez-Ballesteros et al [2008], Santarelli et al [2009]	
c.2905_2923delinsCTCCGAGCGCA	p.Ala969LeufsX30	Rodriguez-Ballesteros et al [2008]	
c. 2991+1G>A (IVS24+1G>A)	--	Adato et al [2000]	
c.2975_2976delAG (c.2975_2978delAG)	p.Gln994ValfsX6	Wang et al [2010]	
c.3032T>C	p.Leu1011Pro	Tekin et al [2005]	
c.3239G>C	p.Arg1080Pro	Romanos et al [2009]	
c.3269C>A	p.Ala1090Glu	Choi et al [2009]	

DNA Nucleotide Change (Alias [1])	Protein Amino Acid Change (Alias [1])	Reference	Reference Sequences
c.3400C>T	p.Arg1134X	Rodriguez-Ballesteros et al [2008]	
c.3413T>C	p.Leu1138Pro	Rodriguez-Ballesteros et al [2008]	
c.3571-2A>C (IVS28-2A>C)	--	Varga et al [2006]	
c.3704-3720delACCGCTCGGCCCCCAG	STOP 20 aa downstream (T1264X)	Zadro et al [2010]	
c.4157C>T	p.Arg577X	Shalin et al [2010]	
c.4227+1G>T		Rodriguez-Ballesteros et al [2008]	
c.4275G>A	p.Trp1425X	Rodriguez-Ballesteros et al [2003]	
c.4351G>T	p.Gly1451X	Rodriguez-Ballesteros et al [2008]	
c.4500+2T>G (IVS36+2T>G)	--	Rodriguez-Ballesteros et al [2003]	
c.4483C>T	p.Arg1495X	Rodriguez-Ballesteros et al [2008]	
c.4491T>A	p.Tyr1497X	Yasunaga et al [1999]	
c.4500+2T>G (IVS36+2T>G)	--	Rodriguez-Ballesteros et al [2003]	
c.4559G>A	p.Arg1520Gln	Rouillon et al [2006]	
c.4809C>A	p.Tyr1603X	Choi et al [2009]	
c.4819C>T	p.Arg1607Trp	Wang et al [2010]	
c.4960+1G>C (IVS39+1G>C)	--	Varga et al [2003]	
c.5011dupT	p.Trp1671LeufsX73	Rodriguez-Ballesteros et al [2008]	
c.5098G>C	p.Glu1700Gln	Chiu et al [2010]	
c.5197G>A	p.Glu1733Lys	Choi et al [2009]	
c.5384T>G	p.Phe1795Cys	Rodriguez-Ballesteros et al [2008], Santarelli et al [2009], Zadro et al [2010]	
c.5410_5412delGAG	p.Glu1804del	Marlin et al [2010]	
c.5473C>G	p.Pro1825Ala	Migliosi et al [2002]	

DNA Nucleotide Change (Alias [1])	Protein Amino Acid Change (Alias [1])	Reference	Reference Sequences
c.5567G>A	p.Arg1856Gln	Choi et al [2009]	
c.5712+1G>A (IVS44+1G>A)	--	Rouillon et al [2006]	
c.5800dupC	p.Leu1934ProfsX251	Rodriguez-Ballesteros et al [2008]	
c.5800dupC	p.Leu1934ProfsX185*	Rodriguez-Ballesteros et al [2008]	
c.5815C>T	p.Arg1939Trp	Choi et al [2009]	
c.5816G>A	p.Arg1939Gln	Varga et al [2003]	
c.5860_5862delATC [2]	p.Ile1954del	Rodriguez-Ballesteros et al [2003]	
c.5960C>G	p.Pro1987Arg	Varga et al [2003]	

Zadro et al [2010]: large deletion defined by snps rs_4491689 and rs_13004993 (52.8 to 61.6 kb)
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. This mutation is present the cochlear-specific isoform and occurs in exon 48. Both brain and cochlea have alternatively spliced isoforms that differ in that exon 47 is expressed in brain and translation is terminated with this exon. In cochlea, exon 47 is skipped and translation terminates with exon 48 [Varga et al 2003].

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