Supp. Table 1. Pathogenicity prediction programs and scores for the novel missense variants identified in our cohort.

Gene	Variant	Protein change	PolyPhen-2[(1)]	PolyPhen-2 score	SIFT[(2)]	SIFT score	Mutation taster[(3)]	Mutation taster probability
ADGRV1	c.1055C>T	p.(Pro352Leu)	Probably damaging	1	Deleterious	0	Disease causing	1
CDH23	c.6083A>C	p.(Asp2028Ala)	Probably damaging	0.999	Deleterious	0	Polymorphism	1
CDH23	c.6202A>C	p.(Thr2068Pro)	Benign	0.251	Deleterious	0.04	Disease causing	1
CLCNKA	c.1985G>T	p.(Gly662Val)	Probably damaging	1	Deleterious	0	Disease causing	1
GPSM2	c.138C>A	p.(Phe46Leu)	Benign	0.007	Deleterious	0.02	Disease causing	1
GRXCR1	c.655G>A	p.(Glu219Lys)	Benign	0.104	Deleterious	0	Disease causing	1
LHFPL5	c.452G>T	p.(Gly151Val)	Probably damaging	1	Deleterious	0	Disease causing	1
MYO7A	c.20G>T	p.(Gly7Val)	Probably damaging	1	Deleterious	0	Disease causing	1
MYO7A	c.247C>A	p.(Arg83Ser)	Probably damaging	1	Deleterious	0	Disease causing	1
MYO7A	c.3364C>A[#]	p.(Leu1122Ile)	Benign	0.015	Tolerated	0.72	Disease causing	0.998
MYO7A	c.3590T>C	p.(Leu1197Pro)	Probably damaging	1	Deleterious	0	Disease causing	1
MYO7A	c.3728C>G	p.(Pro1243Arg)	Probably damaging	0.992	Deleterious	0	Disease causing	1
MYO7A	c.4505A>G	p.(Asp1502Gly)	Possibly damaging	0.908	Deleterious	0	Disease causing	1
MYO7A	c.5345G>C	p.(Gly1782Ala)	Probably damaging	0.999	Deleterious	0	Disease causing	1
MYO7A	c.5522C>G	p.(Thr1841Arg)	Probably damaging	0.972	Deleterious	0	Disease causing	1
OTOF	c.1550T>C	p.(Leu517Pro)	Probably damaging	0.998	Deleterious	0	Disease causing	1
OTOF	c.1904T>A	p.(Val635Asp)	Possibly damaging	0.904	Deleterious	0	Disease causing	1
OTOF	c.3514C>T	p.(Arg1172Trp)	Probably damaging	1	Deleterious	0	Disease causing	1
OTOF	c.5714G>T	p.(Gly1905Val)	Probably damaging	0.997	Deleterious	0	Disease causing	1
PCDH15	c.788C>A	p.(Pro263Gln)	Probably damaging	1	Deleterious	0	Disease causing	1
POU4F3	c.374C>T	p.(Pro125Leu)	Benign	0.021	Deleterious	0.03	Disease causing	1
SLC26A4	c.317C>T	p.(Ala106Val)	Probably damaging	0.969	Deleterious	0	Disease causing	1
TECTA	c.1774G>A	p.(Val592Met)	Possibly damaging	0.482	Tolerated	0.17	Disease causing	0.998
TMC1	c.1259G>A	p.(Cys420Tyr)	Possibly damaging	0.812	Deleterious	0	Disease causing	1
TMC1	c.1728C>G	p.(Asn576Lys)	Possibly damaging	0.797	Deleterious	0.02	Disease causing	1

(1) Polyphen-2, http://genetics.bwh.harvard.edu/pph2/bgi.shtml, cut-off: <0.10 FPR (probably damaging)

(2) SIFT(from PROVEAN), http://provean.jcvi.org/genome_submit_2.php?species=human, cut-off: <0.05 (damaging)

(3) MutationTaster, http://www.mutationtaster.org/, cut-off: 1 = high confidence of prediction

(#) This variant was found damaging by FATHMM, http://fathmm.biocompute.org.uk/fathmm-xf/

Supp. Table S2. Pathogenicity prediction programs scores for the novel splice-site variants identified in our cohort.

Gene	Variant	dbscSNV1.1 ADA SCORE[(1)]	dbscSNV1.1 RF SCORE[(1)]	HSF3.0 prediction[(2)]
EPS8	c.205-8A>G	0.9993	0.934	Activation of an intronic cryptic acceptor site
MSRB3	c.412-1G>A	1	0.938	Broken WT Acceptor Site
MYO7A	c.5326+3A>G	not available	not available	Broken WT Donor Site, activation of an intronic cryptic donor site
MYO7A	c.5856+5G>C	0.9992	0.914	Broken WT Donor Site
OTOF	c.4799+1G>A	not available	not available	Broken WT Donor Site
PTPRQ	c.6739-1G>A	not available	not available	WT site broken, activation of an intronic cryptic acceptor site
SLC26A4	c.1264-3C>G	0.9999	0.966	Broken WT Acceptor Site
TECTA	c.6162+5 G>A	0.99996671	0.996	Broken WT Donor Site
TMC1	c.1224+2T>C	0.9703	0.602	Broken WT Donor Site
TMPRSS3	c.783-1G>T	0.9999	0.942	not available

(1)>0.6 is predicted splice altering, (2) http://www.umd.be/HSF3/

Supp. Table S3. Novel and reported variants in this study of known genes associated with autosomal recessive hearing loss segregating in compound heterozygosity.

Family	Gene	Allele	Allele frequency			Gene	Allele	Allele frequency			Reference
			Pak	1000G	ExAC			Pak	1000G	ExAC	
PKDF055	*CDH23*	c.2968G>A, p.(Asp990Asn)	-	-	-	*CDH23*	c.6049+1G>A, Splice error	-	-	-	Bork, et al., 2001
PKDF936	*GJB2*	c.71G>A, p.(Trp24*)	-	0.0004	0.0006	*GJB2*	c.231C>T, p.(Trp77*)	-	0.0000	0.0001	Santos, et al., 2005b
DEM4582	*GJB2*	c.-23+1G>A, Splice error	-	-	2E-05	*GJB2*	c.71G>A, p.(Trp24*)	-	0.0004	0.0006	Denoyelle, et al., 1997;Kelsell, et al., 1997
PKDF019A	***MYO7A***	**c.20G>T, p.(Gly7Val)**	0/170	-	1E-05	*MYO7A*	c.1849T>C, p.(Ser617Pro)	0/160	-	5E-05	This study; Carss, et al., 2017
PKDF173	*MYO7A*	c.93C>A**,** p.(Cys31*)	-	0.017	5E-05	***MYO7A***	**c.3728C>G, p.(Pro1243Arg)**	0/164	-	-	Weston, et al., 1996; this study
PKDF312	*MYO7A*	c.397dupC, p.(His133Profs*7)	-	-	0.0006	*MYO7A*	c.3136dupC, p.(Leu1046Profs*9)	-	-	-	Riazuddin, et al., 2008
DEM4652(†)	*MYO7A*	c.1258A>T, p.(Lys420*)	-	-	4E-05	***MYO7A***	**c.4505A>G, p.(Asp1502Gly)**	0/454	-	2E-05	Le Quesne Stabej, et al., 2012;this study
DEM4190	*MYO7A*	c.1258A>T, p.(Lys420*)	-	-	4E-05	*MYO7A*	c.4838delA, p.(Asp1613Valfs*32)	-	-	-	Le Quesne Stabej, et al., 2012;Riazuddin, et al., 2008
DEM4561	***MYO7A***	**c.3364C>A, p.(Leu1122Ile)**	-	-	0.0001506	***MYO7A***	**c.5345G>C, p.(Gly1782Ala)**	-	-	4E-05	This study
DEM4281, DEM4692	*MYO7A*	c.3502C>T, p.(Arg1168Trp)	-	0.0002	0.0001	*MYO7A*	c.4838delA, p.(Asp1613Valfs*32)	-	-	-	Jaijo, et al., 2006;Riazuddin, et al., 2008
PKDF1693	*MYO15A*	c.4528C>T, p.(Gln1510*)	-	-	8E-06	***MYO15A***	**c.4570C>T, p.(Gln1524*)**	-	-	-	Rehman, et al., 2015;This study
DEM4149B	*SLC26A4*	c.71G>T, p.(Arg24Leu)	-	-	-	*SLC26A4*	c.716T>A, p.(Val239Asp)	-	0.0002	0.0002	Prasad, et al., 2004;Park, et al., 2003
DEM4220	*SLC26A4*	c.716T>A, p.(Val239Asp)	-	0.0002	0.0002	*SLC26A4*	c.1337A>G, p.(Gln446Arg)	-	-	8E-05	Park, et al., 2003;Reardon, et al., 2000
PKDF384b	*SLC26A4*	c.1226G>A, p.(Arg409His)	-	-	0.0001	*SLC26A4*	c.1229C>T, p.(Thr410Met)	-	0.0002	0.0002	Van Hauwe, et al., 1998;Reardon, et al., 2000
PKDF1452	***TECTA***	**c. 2736 C>A, p.(Cys912*)**	-	-	-	***TECTA***	**c.6162+5 G>A Splice error**	-	-	-	This study
PKSR13b	***TMC1***	**c.1143C>G, p.(Tyr381*)**	0/166	-	-	*TMC1*	c.1810C>T, p.(Arg604*)	-	-	8E-06	This study;Sirmaci, et al., 2009
HL006	***TMC1***	**c.1259G>A, p.(Cys420Tyr)**	0/125	-	-	*TMC1*	c.1788C>A, p.(Ser596Arg)	0/125	-	-	This study;Imtiaz, et al., 2016

Novel mutations are in bold font. (†) segregates another known mutation *MYO7A* (c.3502C>T, p.Arg1168Trp) (Jaijo, et al., 2006). Pak: Pakistani controls, 1000G: 1000 Genomes Project database, ExAc: Exome Aggregation Consortium database. (-) indicates that the variant is absent presently from the database. (*) indicates stop codon.

Supp. Table S4. Novel and reported mutant variants in known genes associated with autosomal recessive hearing loss in our Pakistani cohort.

Gene	Accession number	Studies	Nb of families	Variant	Protein change
ADCY1†	NM_021116.2	Santos-Cortez, et al., 2014	1	c.3112C>T	p.(Arg1038*)
ADGRV1	NM_032119.3	this study	1	**c.1055C>T**	**p.(Pro352Leu)**
BSND	NM_057176.2	Riazuddin, et al., 2009	1	c.10G>T	p.(Glu4*)
BSND	NM_057176.2	Riazuddin, et al., 2009; Naz, et al., 2017; this study	15	c.35T>C	p.(Ile12Thr)
CABP2	NM_016366.2	this study	1	**c.637+1G>T**	**Splice error**
CDH23†	NM_022124.5	Schultz, et al., 2011	2	c.778G>A	p.(Ala260Thr)
CDH23†	NM_022124.5	this study	1	**c.2866G>A**	**p.(Glu956Lys)**
CDH23†	NM_022124.5	Schultz, et al., 2011; this study	3	c.2968G>A	p.(Asp990Asn)
CDH23†	NM_022124.5	this study	1	**c.3481C>T**	**p.(Arg1161*)**
CDH23†	NM_022124.5	this study	1	**c.3880C>T**	**p.(Gln1294*)**
CDH23†	NM_022124.5	Schultz, et al., 2011	1	c.4267G>A	p.(Asp1423Asn)
CDH23†	NM_022124.5	Schultz, et al., 2011	1	c.4688T>C	p.(Leu1563Pro)
CDH23†	AF312024	Astuto, et al., 2002	2	c.4756G>C	p.(Ala1586Pro)
CDH23†	NM_022124.5	Schultz, et al., 2011	1	c.4829G>T	p.(Gly1610Val)
CDH23†	NM_022124.5	this study	1	**c.4892C>T**	**p.(Ala1631Val)**
CDH23†	NM_022124.5	this study	1	**c.5149T>C**	**p.(Cys1717Arg)**
CDH23†	NM_022124.5	Bork, et al., 2001; this study	2	c.6049+1G>A	Splice error
CDH23†	NM_022124.5	this study	1	**c.6050 -9G>A**	**Splice error**
CDH23†	NM_022124.5	this study	1	**c.6083A>C**	**p.(Asp2028Ala)**
CDH23†	NM_022124.5	Bork, et al., 2001; Schultz, et al., 2011; this study	5	c.6133G>A	p.(Asp2045Asn)
CDH23†	NM_022124.5	this study	1	**c.6202A>C**	**p.(Thr2068Pro)**
CDH23†	NM_022124.5	Bork, et al., 2001; Schultz, et al., 2011, this study	3	c.6604G>A	p.(Asp2202Asn)
CDH23†	NM_022124.5	Naz, et al., 2017, this study	2	c.7814A>G	p.(Asn2605Ser)
CDH23†	NM_022124.5	Schultz, et al., 2011, this study	2	c.7987_7989delTTC	p.(Phe2664del)
CDH23†	NM_022124.5	Schultz, et al., 2011	1	c.8113A>G	p.(Met2705Val)
CDH23†	NM_022124.5	this study	1	**c.8351_8352insCGAT**	**p.(Leu2785Aspfs*43)**
CDH23†	NM_022124.5	Schultz, et al., 2011; Shahzad, et al., 2013	2	c.8530C>T	p.(Pro2844Ser)
CDH23†	NM_022124.5	Schultz, et al., 2011	1	c.8531C>T	p.(Pro2844Leu)

CDH23†	AY010111	Bork, et al., 2001	1	IVS23+1G→A	Splice error
CDH23†	AY010111	Bork, et al., 2001	1		p.(Asp188Asn)
CDH23†	AY010111	Bork, et al., 2001	1		p.(Gln492*)
CIB2†	NM_006383.3	Riazuddin, et al., 2012	1	c.192G>C	p.(Glu64Asp)
CIB2†	NM_006383.3	Riazuddin, et al., 2012; Rehman, et al., 2015; Seco, et al., 2016; this study	71	c.272T>C	p.(Phe91Ser)
CIB2†	NM_006383.3	Riazuddin, et al., 2012; this study	3	c.297C>G	p.(Cys99Trp)
CLCNKA	NM_004070.3	this study	1	**c.1985G>T**	**p.(Gly662Val)**
CLDN14†	NM_001146077.1	Lee, et al., 2012a	1	c.167G>A	p.(Trp56*)
CLDN14†	NM_001146077.1	Lee, et al., 2012a	1	c.242G>A	p.(Arg81His)
CLDN14†	NM_012130	Wilcox, et al., 2001; Lee, et al., 2012a; Faridi, et al., 2017; Naz, et al., 2017; this study	10	c.254T>A	p.(Val85Asp)
CLDN14†	NM_012130	Wilcox, et al., 2001	1	c.398delT	p.(Met133Argfs*24)
CLDN14†	NM_001146077.1	Lee, et al., 2012a	1	c.694G>A	p.(Gly232Arg)
EDNRB	NM_000115.3	this study	1	**c.553G>A**	**p.(Val185Met)**
ELMOD3†	NM_032213.4	Jaworek, et al., 2013	1	c.794T>C	p.(Leu265Ser)
EPS8	NM_004447.5	this study	1	**c.205-8A>G**	**Splice error**
ESPN†	NM_031475.2	Naz, et al., 2004	1	c.1988_1991delAGAG	p.(Lys663Thrfs*2)
ESPN †	NM_031475.2	Naz, et al., 2017	1	c.2019dupG	p.(Leu674Alafs*72)
ESPN†	NM_031475.2	Naz, et al., 2004	1	c.2470_2473del	p.(Ser824Argfs*22)
ESPN†	NM_031475.2	this study	1	**c.2496_2496delC**	**p.(Tyr832*)**
ESRRB†	NM_004452.3	Collin, et al., 2008	1	c.329C>T	p.(Ala110Val)
ESRRB†	NM_004452.3	this study	1	**c.536G>A**	**p.(Arg179His)**
ESRRB†	NM_004452.3	Collin, et al., 2008	1	c.959T>C	p.(Leu320Pro)
ESRRB†	NM_004452.3	Lee, et al., 2011b	1	c.1018_1020delGAG	p.(Glu340del)
ESRRB†	NM_004452.3	Collin, et al., 2008	1	c.1024G>T	p.(Val342Leu)
ESRRB†	NM_004452.3	Collin, et al., 2008	1	c.1040T<C	p.(Leu347Pro)
GAB1†	NM_207123	Yousaf, et al., 2018a	1	c.347G>A	p.(Gly116Glu)
GIPC3†	NM_133261.2	Rehman, et al., 2011	1	c.264G>A	p.(Met88Ile)
GIPC3†	NM_133261.2	Rehman, et al., 2011	1	c.281G>A	p.(Gly94Asp)
GIPC3†	NM_133261.2	Rehman, et al., 2011	1	c.565C>T	p.(Gly46Arg)
GIPC3†	NM_133261.2	Rehman, et al., 2011; Naz, et al., 2017; this study	5	c.662C>T	p.(Thr221Ile)

GIPC3[†]	NM_133261.2	Rehman, et al., 2011	1	c.685dupG	p.(Ala229Glyfs*10)
GIPC3[†]	NM_133261.2	Rehman, et al., 2011	1	c.767G>A	p.(Gly256Asp)
GJB2	NM_004004.5	Rehman, et al., 2015	1	131-kb deletion	131-kb deletion
GJB2	NM_004004.5	this study	2	**c.-23+1G>A**	**Splice error**
GJB2	NM_004004.5	Santos, et al., 2005b; Rehman, et al., 2015; this study	10	c.35delG	p.(Gly12Valfs*2)
GJB2	NM_004004.5	Santos, et al., 2005b; Rehman, et al., 2015; Naz, et al., 2017; this study	36	c.71G>A	p.(Trp24*)
GJB2	NM_004004.5	Santos, et al., 2005b	1	c.95G>A	p.(Arg32His)
GJB2	NM_004004.5	Santos, et al., 2005b	1	c.167delT	p.(Leu56Argfs*26)
GJB2	NM_004004.5	this study	1	**c.223C>T**	**p.(Arg75Trp)**
GJB2	NM_004004.5	this study	2	**c.224G>A**	**p.(Arg75Gln)**
GJB2	NM_004004.5	Santos, et al., 2005b; Rehman, et al., 2015; Naz, et al., 2017; this study	28	c.231G>A	p.(Trp77*)
GJB2	NM_004004.5	this study	1	**c.355G>T**	**p.(Glu119*)**
GJB2	NM_004004.5	Santos, et al., 2005b; Naz, et al., 2017	2	c.358_360 del	p.(Glu120del)
GJB2	NM_004004.5	this study	2	**c.457G>A**	**p.(Val153Ile)**
GPSM2	NM_001321039.1	this study	1	**c.138C>A**	**p.(Phe46Leu)**
GRXCR1[†]	NM_001080476.2	Schraders, et al., 2010; this study	2	c.229C>T	p.(Gln77*)
GRXCR1[†]	NM_001080476.2	Schraders, et al., 2010	1	c.412C>T	p.(Arg138Cys)
GRXCR1[†]	NM_001080476.2	this study	1	**c.655G>A**	**p.(Glu219Lys)**
GRXCR2[†]	NM_001080516.1	Naz, et al., 2017	1	c.714dupT	p.(Gly239Trpfs*74)
HGF[†]	NM_000601.4	Schultz, et al., 2009; Rehman, et al., 2015; Naz, et al., 2017; this study	66	c.482 +1986_88delTGA	ND
HGF[†]	NM_000601.4	Schultz, et al., 2009; this study	3	c.482+1991_2000delG ATGATGAAA	ND
HGF[†]	NM_000601.4	Schultz, et al., 2009	1	c.495G>A	p.(Ser165Ser)
ILDR1	NM_001199799.1	Borck, et al., 2011; this study	2	c.3G>A	p.(Met1Ile)
ILDR1	NM_001199799.1	Borck, et al., 2011	1	c.290G>A	p.(Arg97Gln)
ILDR1	NM_001199799.1	Borck, et al., 2011	1	c.411delG	p.(Trp137Cysfs*25)
ILDR1	NM_001199799.1	Borck, et al., 2011	1	c.499+1G>A	ND
ILDR1	NM_175924.3	this study	1	**c.900delG**	**p.(Thr301Profs*20)**

ILDR1	NM_175924.3	this study	1	**c.1005_1005delG**	**p.(Glu335Aspfs*30)**
ILDR1	NM_175924.3	this study	1	**c.1029_1030delTT**	**p.(Trp344Glyfs*17)**
ILDR1	NM_001199799.1	Borck, et al., 2011	2	c.1032delG	p.(Thr345Profs*20)
ILDR1	NM_001199799.1	Borck, et al., 2011	1	c.1135G>T	p.(Glu379*)
ILDR1	NM_001199799.1	Borck, et al., 2011	1	c.1180delG	p.(Glu394Serfs*15)
ILDR1	NM_001199799.1	this study	1	**c.1384C>T**	**p.(Arg462*)**
ILDR1	NM_001199799.1	Borck, et al., 2011	1	c.1358G>A	p.(Arg453Gln)
ILDR1	NM_001199799.1	Borck, et al., 2011	1	c.1387C>T	p.(Arg463Cys)
KARS†	NM_001130089.1	Santos-Cortez, et al., 2013	2	c.571T>C	p.(Tyr173His)
KARS†	NM_001130089.1	Santos-Cortez, et al., 2013	1	c.1129G>A	p.(Asp377Asn)
LHFPL5†	NM_182548.3	Shabbir, et al., 2006; Rehman, et al., 2015; this study	4	c.250delC	p.(Leu84*)
LHFPL5†	NM_182548.3	this study	1	**c.452G>T**	**p.(Gly151Val)**
LRTOMT†	EU627069	Ahmed, et al., 2008a	1	c.328G>A	p.(Glu110Lys)
MARVELD2†	NM_001038603.2	Nayak, et al., 2015	1	8,095 bp deletion	p.(Cys395-Gln501del)
MARVELD2†	DQ682656.1	Riazuddin, et al., 2006a	2	IVS4+2delTGAG	Splice error
MARVELD2†	NM_001038603.2	this study	1	**c.1138C>T**	**p.(Gln380*)**
MARVELD2†	DQ682656.1	Riazuddin, et al., 2006a; Nayak, et al., 2015	2	c.1183-1G>A	Splice error
MARVELD2†	NM_001038603.2	this study	1	**c.1223_1224insA**	**p.(Glu408Argfs*4)**
MARVELD2†	NM_001038603.2	Chishti, et al., 2008	1	c.1295+1G>A	Splice error
MARVELD2†	NM_001038603.2	Riazuddin, et al., 2006a; Chishti, et al., 2008; Nayak, et al., 2015; this study	12	c.1295+2T>C	Splice error
MARVELD2†	DQ682656.1	Riazuddin, et al., 2006a	1	c.1498C>T	p.(Arg500*)
MET†	NM_000245.2	Mujtaba, et al., 2015	1	c.2521T>G	p.(Phe841Val)
MSRB3†	NM_001031679.2	Ahmed, et al., 2011	2	c.55C>T	p.(Arg19*)
MSRB3†	NM_198080.3	Ahmed, et al., 2011; this study	11	c.265T>G	p.(Cys89Gly)
MSRB3†	NM_001031679.2	this study	1	**c.412-1G>A**	**Splice error**
MYO6†	NM_004999.3	Ahmed, et al., 2003a	1	c.36dupT	p.(Thr13Tyrfs*20)
MYO6†	NM_004999.3	Ahmed, et al., 2003a	1	c.647A>T	p.(Glu216Val)
MYO6†	NM_004999.3	Naz, et al., 2017	1	c.1729_1741del	p.(Phe577Ilefs*28)
MYO6†	NM_004999.3	Ahmed, et al., 2003a	1	c.3496C>T	p.(Arg1166*)
MYO7A	NM_000260.3	this study	1	**c.20G>T**	**p.(Gly7Val)**
MYO7A	NM_000260.3	this study	1	**c.93C>A**	**p.(Cys31*)**
MYO7A	NM_000260.3	this study	1	**c.247C>A**	**p.(Arg83Ser)**

MYO7A	NM_000260.3	Riazuddin, et al., 2008	1	c.252C>G	p.(Asn84Lys)
MYO7A	NM_000260.3	Riazuddin, et al., 2008; Rehman, et al., 2015; this study	3	c.397dupC	p.(His133Profs*7)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.398A>C	p.(His133Pro)
MYO7A	NM_000260.3	Riazuddin, et al., 2008	1	c.471-1G>A	Frame shift
MYO7A	NM_000260.3	Riazuddin, et al., 2008; Shahzad, et al., 2013; this study	6	c.496delG	p.(Glu166Argfs*5)
MYO7A	NM_000260.3	Riazuddin, et al., 2008	2	c.640G>A	p.(Gly214Arg)
MYO7A	NM_000260.3	Shahzad, et al., 2013	3	c.721C>T	p.(Arg241Cys)
MYO7A	NM_000260.3	Shahzad, et al., 2013; this study	4	c.722G>A	p.(Arg241His)
MYO7A	NM_000260.3	Riazuddin, et al., 2008; this study	2	c.977T>A	p.(Leu326Gln)
MYO7A	NM_000260.3	Shahzad, et al., 2013; Naz, et al., 2017; this study	4	c.1183C>T	p.(Arg395Cys)
MYO7A	NM_000260.3	Shahzad, et al., 2013; this study	6	c.1258A>T	p.(Lys420*)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.1300G>C	p.(Gly434Arg)
MYO7A	NM_000260.3	Riazuddin, et al., 2008	2	c.1309G>A	p.(Asp437Asn)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.1355T>C	p.(Leu452Pro)
MYO7A	NM_000260.3	Riazuddin, et al., 2008; this study	2	c.1591C>T	p.(Gln531*)
MYO7A	NM_000260.3	this study	1	**c.1849T>C**	**p.(Ser617Pro)**
MYO7A	NM_000260.3	Riazuddin, et al., 2008	1	c.1935+1G>A	Splice error
MYO7A	NM_000260.3	this study	1	**c.2339delG**	**p.(Gly780Valfs*10)**
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.2367+1G>A	ND
MYO7A	NM_000260.3	Riazuddin, et al., 2008; Shahzad, et al., 2013	1	c.2476G>A	p.(Ala826Thr)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.2513G>A	p.(Trp838*)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.2695-9A>G	ND
MYO7A	AY821853.1	Riazuddin, et al., 2008	1	c.2914C>T	p.(Arg972*)
MYO7A	NM_000260.3	Riazuddin, et al., 2008; this study	2	c.3136dupC	p.(Leu1046Profs*9)
MYO7A	NM_000260.3	this study	1	**c.3364C>A**	**p.(Leu1122Ile)**
MYO7A	NM_000260.3	this study	4	**c.3502C>T**	**p.(Arg1168Trp)**
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.3504-1G>C	Splice error
MYO7A	NM_000260.3	Riazuddin, et al., 2008; this study	3	c.3508G>A	p.(Glu1170Lys)
MYO7A	NM_000260.3	this study	1	**c.3590T>C**	**p.(Leu1197Pro)**

MYO7A	NM_000260.3	Riazuddin, et al., 2008	1	c.3631delT	p.(Tyr1211fs*21)
MYO7A	NM_000260.3	this study	1	**c.3728C>G**	**p.(Pro1243Arg)**
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.3904delT	p.(Tyr1302Thrfs*97)
MYO7A	NM_000260.3	this study	1	**c.4505A>G**	**p.(Asp1502Gly)**
MYO7A	NM_000260.3	Riazuddin, et al., 2008; Shahzad, et al., 2013; this study	8	c.4838delA	p.(Asp1613Valfs*32)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.4911_4912insAT	p.(Asp1638Metfs*8)
MYO7A	NM_000260.3	Riazuddin, et al., 2008	1	c.5146_5148delGAG	p.(Glu1716 del)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.5168G>C	p.(Arg1723Thr)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.5177C>T	p.(Pro1726Leu)
MYO7A	NM_000260.3	this study	1	**c.5326+3A>G**	**Splice error**
MYO7A	NM_000260.3	this study	1	**c.5345G>C**	**p.(Gly1782Ala)**
MYO7A	NM_000260.3	Riazuddin, et al., 2008	1	c.5366+1G>A	Splice error
MYO7A	NM_000260.3	this study	1	**c.5434G>A**	**p.(Glu1812Lys)**
MYO7A	NM_000260.3	this study	1	**c.5522C>G**	**p.(Thr1841Arg)**
MYO7A	NM_000260.3	this study	1	**c.5660C>T**	**p.(Pro1887Leu)**
MYO7A	NM_000260.3	this study	1	**c.5856+5G>C**	**Splice error**
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.5899C>T	p.(Arg1967*)
MYO7A	NM_000260.3	Riazuddin, et al., 2008	2	c.5944G>A	p.(Gly1982Arg)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.6354+1G>A	Splice error
MYO7A	NM_000260.3	Shahzad, et al., 2013; Naz, et al., 2017	2	c.6354G>C	p.(Lys2118Asn)
MYO7A	NM_000260.3	Shahzad, et al., 2013	1	c.6487G>A	p.(Gly2163Ser)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.742C>G	p.(Arg248Gly)
MYO15A	NM_016239.3	Naz, et al., 2017	1	c.1185dupC	p.(Glu396Argfs*36)
MYO15A	NM_016239.3	Naz, et al., 2017	1	c.1657delC	p.(Arg553Glyfs*76)
MYO15A	NM_016239.3	Naz, et al., 2017; this study	2	c.2456C>A	p.(Ser819*)
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.3313G>T	p.(Glu1105*)
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.3336delG	p.(Arg1113Valfs*12)
MYO15A	NM_016239.3	Liburd, et al., 2001; Rehman, et al., 2016	2	c.3685C>T	p.(Gln1229*)
MYO15A	NM_016239.3	Liburd, et al., 2001	1	c.3756+1G>T	Splice error
MYO15A	NM_016239.3	this study	1	**c.3505C>T**	**p.(Arg1169*)**
MYO15A	NM_016239.3	Nal, et al., 2007; Naz, et al., 2017	2	c.3866+1G>A	Splice error
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.3932T>C	p.(Ile1311Thr)

MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.3944G>A	p.(Gly1315Glu)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.4108C>T	p.(Arg1370Cys)
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.4176C>A	p.(Tyr1392*)
MYO15A	NM_016239.3	Rehman, et al., 2016; this study	2	c.4528C>T	p.(Gln1510*)
MYO15A	NM_016239.3	this study	1	**c.4570C>T**	**p.(Gln1524*)**
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.4669A>G	p.(Lys1557Glu)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.4780G>C	p.(Asp1594His)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.4898T>C	p.(Ile1633Thr)
MYO15A	NM_016239.3	this study	1	**c.5005C>T**	**p.(Gln1669*)**
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.5117_5118delGCinsTT	p.(Gly1706Val)
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.5189T>C	p.(Leu1730Pro)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.5335delC	p.(Leu1779Trpfs*18)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.5809C>T	p.(Arg1937Cys)
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.6061C>T	p.(Gln2021*)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.6146C>A	p.(Pro2049His)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.6178-2A>G	Splice error
MYO15A	NM_016239.3	Nal, et al., 2007; Rehman, et al., 2016	2	c.6589C>T	p.(Gln2197*)
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.6731G>A	p.(Gly2244Glu)
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.6796G>A	p.(Val2266Met)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.6944delG	p.(Gly2315Glufs*102)
MYO15A	NM_016239.3	Liburd, et al., 2001	1	c.8148G>T	p.(Gln2716His)
MYO15A	NM_016239.3	Naz, et al., 2017	1	c.8158G>A	p.(Asp2720Asn)
MYO15A	NM_016239.3	Nal, et al., 2007	4	c.8158G>C	p.(Asp2720His)
MYO15A	NM_016239.3	Rehman, et al., 2016; this study	2	c.8224+3A>G	Splice error
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.8821_8822insTG	p.(Gly2941Valfs*94)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.9229+2T>C	Splice error
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.9478C>T	p.(Leu3160Phe)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.10208_10209delAGinsACCAGGCCCGTGCAGCTC	p.(Gln3403delinsProThrArgProValGlnLeu)
MYO15A	NM_016239.3	Nal, et al., 2007	1	c.10474C>T	p.(Gln3492*)
MYO15A	NM_016239.3	Rehman, et al., 2016	1	c.10572dupC	p.(Ser3525Glnfs*79)
NARS2[†]	NM_024678.5	Simon, et al., 2015	1	c.637G>T	p.(Val213Phe)
OTOA	NM_170664.2	this study	1	**c.1148C>T**	**p.(Ala383Val)**

OTOA	NM_144672.3	Lee, et al., 2013; this study	2	c.1352G>A	p.(Gly451Asp)
OTOA	NM_144672.3	Lee, et al., 2013	2	c.1879C>T	p.(Pro627Ser)
OTOF	NM_194248.2	Choi, et al., 2009	1	c.1103_1104delinsC	p.(Gly368Alafs*2)
OTOF	NM_194248.2	Choi, et al., 2009	1	c.1273C>T	p.(Arg425*)
OTOF	NM_194248.2	this study	2	**c.1550T>C**	**p.(Leu517Pro)**
OTOF	NM_194248.2	Choi, et al., 2009	1	c.1607G>A	p.(Trp536*)
OTOF	NM_194248.2	Choi, et al., 2009	1	c.1718T>G	p.(Leu573Arg)
OTOF	NM_194248.2	this study	1	**c.1904T>A**	**p.(Val635Asp)**
OTOF	NM_194248.2	Choi, et al., 2009; this study	3	c.2122C>T	p.(Arg708*)
OTOF	NM_194248.2	Choi, et al., 2009	1	c.2295_2297del	p.(Glu766del)
OTOF	NM_004802.3	Choi, et al., 2009; this study	2	c.2508C>A	p.(Tyr836*)
OTOF	NM_004802.3	Choi, et al., 2009; this study	3	c.2896G>A	p.(Glu966Lys)
OTOF	NM_194248.2	Naz, et al., 2017; this study	2	c.2965_2967del	p.(Phe989del)
OTOF	NM_194248.2	Choi, et al., 2009	1	c.3269C>A	p.(Ala1090Glu)
OTOF	NM_194248.2	Naz, et al., 2017	1	c.3289-1G>T	Splice error
OTOF	NM_194248.2	this study	1	**c.3376dupA**	**p.(Ile1126Asnfs*51)**
OTOF	NM_194323.2	this study	1	**c.3514C>T**	**p.(Arg1172Trp)**
OTOF	NM_194248.2	this study	1	**c.4799+1G>A**	**Splice error**
OTOF	NM_194248.2	Naz, et al., 2017	1	c.4805G>T	p.(Gly1602Val)
OTOF	NM_194248.2	Choi, et al., 2009	1	c.5567G>A	p.(Arg1856Gln)
OTOF	NM_194248.2	this study	1	**c.5714G>T**	**p.(Gly1905Val)**
OTOF	NM_194248.2	Choi, et al., 2009	1	c.5815C>T	p.(Arg1939Trp)
OTOG	NM_001277269.1	this study	1	**c.7235delG**	**p.(Cys2412Serfs*150)**
PCDH15[†]	NM_001142771.1	Ahmed, et al., 2001; Ahmed, et al., 2008b	2	c.7C>T	p.(Arg3*)
PCDH15[†]	NM_001142771.1	Ahmed, et al., 2003b; Ahmed, et al., 2008b; this study	3	c.400C>G	p.(Arg134Gly)
PCDH15[†]	NM_001142771.1	Ahmed, et al., 2008b	1	c.533A>G	p.(Asp178Gly)
PCDH15[†]	NM_001142771.1	Ahmed, et al., 2003b	1	c.785G>A	p.(Gly262Asp)
PCDH15[†]	NM_001142772.1	this study	1	**c.788C>A**	**p.(Pro263Gln)**
PCDH15[†]	NM_001142771.1	this study	1	**c.1737C>A**	**p.(Tyr584*)**
PCDH15[†]	NM_001142771.1	Ahmed, et al., 2003b	1	c.1927C>T	p.(Arg643*)
PCDH15[†]	NM_001142771.1	Ahmed, et al., 2008b	1	c.1940C>G	p.(Ser647*)
PCDH15[†]	NM_001142771.1	Ahmed, et al., 2008b	1	c.2052C>A	p.(Tyr684*)
PCDH15[†]	NM_001142771.1	Ahmed, et al., 2008b	1	c.2483delT	p.(Glu829Lysfs*13)

PCDH15†	NM_001142771.1	Ahmed, et al., 2008b	1	c.3717+1G>T	Splice error
PCDH15†	NM_001142771.1	Ahmed, et al., 2001	1	c.3718-2A>6	Splice error
PCDH15†	NM_001142771.1	Ahmed, et al., 2008b	1	c.4257delA	p.(Leu1419Phefs*99)
PCDH15†	NM_001142772.1	this study	1	**deletion of exons 14 and 15**	**N/A**
PJVK	NM_001042702.3	this study	1	**c.158C>G**	**p.(Ser53*)**
PJVK	NM_001042702.3	this study	1	**c.162_172del**	**p.(Pro55Glufs*23)**
PJVK	NM_001042702.3	this study	1	**c.406C>T**	**p.(Arg136*)**
PJVK	NM_001042702.3	this study	1	**c.908_910delACA**	**p.(Asn303del)**
PJVK	NM_001042702.3	Naz, et al., 2017	1	c.1028G>C	p.(Cys343Ser)
POU3F4	NM_000307.4	Rehman, et al., 2015	1	3.1-Mb deletion	3.1-Mb deletion
POU3F4	NM_000307.4	this study	1	**c.478C>T**	**p.(Gln160*)**
POU4F3	NM_002700.2	this study	1	**c.374C>T**	**p.(Pro125Leu)**
PPIP5K2†	NM_001276277	Yousaf, et al., 2018b	2	c.2510G>A	p.(Arg837His)
PTPRQ	NM_001145026.1	Naz, et al., 2017	1	c.189delC	p.(Glu65Lysfs*95)
PTPRQ	NM_001145026.1	this study	1	**c.5158_5159delAT**	**p.(Ile1720Glnfs*7)**
PTPRQ	NM_001145026.1	this study	1	**c.6739-1G>A**	**Splice error**
RDX†	NM_001260493.1	Lee, et al., 2011a	1	c.1076_1079delTTAA	p.(Ile359Lysfs*6)
RDX†	NM_001260493.1	Khan, et al., 2007	1	c.1405dupG	p.(Ala469Glyfs*19)
RDX†	NM_001260493.1	Khan, et al., 2007	1	c.1732G>A	p.(Asp578Asn)
S1PR2†	NM_004230.3	Santos-Cortez, et al., 2016	1	c.323G>C	p.(Arg108Pro)
S1PR2†	NM_004230.3	Santos-Cortez, et al., 2016	1	c.419A>G	p.(Tyr140Cys)
SLC26A4	NM_000441.1	this study	1	**c.42delC**	**p.(Glu15Serfs*51)**
SLC26A4	NM_000441.1	Anwar, et al., 2009; this study	5	c.71G>T	p.(Arg24Leu)
SLC26A4	NM_000441.1	this study	1	**c.154A>T**	**p.(Lys52*)**
SLC26A4	NM_000441.1	Anwar, et al., 2009; Naz, et al., 2017	2	c.170C>A	p.(Ser57*)
SLC26A4	NM_000441.1	Anwar, et al., 2009	5	c.269C>T	p.(Ser90Leu)
SLC26A4	NM_000441.1	Anwar, et al., 2009	1	c.304+2T>C	p.(Met103Lysfs*4)
SLC26A4	NM_000441.1	this study	1	**c.317C>T**	**p.(Ala106Val)**
SLC26A4	NM_000441.1	this study	1	**c.413T>A**	**p.(Val138Asp)**
SLC26A4	NM_000441.1	Anwar, et al., 2009; this study	2	c.416G>T	p.(Gly139Val)
SLC26A4	NM_000441.1	Anwar, et al., 2009; Rehman, et al., 2015	2	c.691G>A	p.(Val231Met)

SLC26A4	NM_000441.1	Anwar, et al., 2009; Shahzad, et al., 2013; Rehman, et al., 2015; Naz, et al., 2017; this study	47	c.716T>A	p.(Val239Asp)
SLC26A4	NM_000441.1	this study	3	**c.919-2A>G**	**Splice error**
SLC26A4	NM_000441.1	Naz, et al., 2017	1	c.965dupA	p.(Asn322Lysfs*8)
SLC26A4	NM_000441.1	this study	1	**c.1001G>T**	**p.(Gly334Val)**
SLC26A4	NM_000441.1	Anwar, et al., 2009	1	c.1115C>T	p.(Ala372Val)
SLC26A4	NM_000441.1	this study	1	**c.1198delT**	**p.(Cys400Valfs*32)**
SLC26A4	NM_000441.1	this study	1	**c.1226G>A**	**p.(Arg409His)**
SLC26A4	NM_000441.1	this study	3	**c.1226G>C**	**p.(Arg409Pro)**
SLC26A4	NM_000441.1	this study	2	**c.1229C>T**	**p.(Thr410Met)**
SLC26A4	NM_000441.1	this study	1	**c.1238A>G**	**p.(Gln413Arg)**
SLC26A4	NM_000441.1	this study	1	**c.1264-3C>G**	**Splice error**
SLC26A4	NM_000441.1	Anwar, et al., 2009	1	c.1264-477_2090-4927del11202	
SLC26A4	NM_000441.1	Anwar, et al., 2009; Rehman, et al., 2015; Naz, et al., 2017; this study	24	c.1337A>G	p.(Gln446Arg)
SLC26A4	AAC51873	Anwar, et al., 2009	1	c.1341+3A>C	p.(Gly439Valfs*19)
SLC26A4	AAC51873	Anwar, et al., 2009	1	c.1363A>T	p.(Ile455Phe)
SLC26A4	NM_000441.1	Anwar, et al., 2009; Naz, et al., 2017	4	c.1667A>G	p.(Tyr556Cys)
SLC26A4	NM_000441.1	Anwar, et al., 2009	1	c.1692_1693insA	p.(Cys565Metfs*9)
SLC26A4	NM_000441.1	this study	6	**c.2106delG**	**p.(Lys702Asnfs*19)**
SLC26A4	NM_000441.1	Anwar, et al., 2009	2	c.2145G>T	p.(Lys715Asn)
SLC26A4	AAC51873	Anwar, et al., 2009	1	c.-23177_c.164+1027del24368ins7	
TBC1D24[†]	NM_001199107.1	Rehman, et al., 2014	3	c.208G>T	p.(Asp70Tyr)
TBC1D24[†]	NM_001199107.1	Rehman, et al., 2014	1	c.878G>C	p.(Arg293Pro)
TECTA	NM_005422.2	Naz, et al., 2017	1	c.64+2T>C	Splice error
TECTA	NM_005422.2	this study	1	**c.840_841insT**	**p.(Val281Cysfs*11)**
TECTA	NM_005422.2	this study	1	**c.1247_1248delGG**	**p.(Gly416Aspfs*24)**
TECTA	NM_005422.2	this study	1	**c.1774G>A**	**p.(Val592Met)**
TECTA	NM_005422.2	this study	1	**c. 2736 C>A**	**p.(Cys912*)**
TECTA	NM_005422 .2	Naz, et al., 2003	1	c.6037delG	p.(Glu2013Argfs*6)
TECTA	NM_005422.2	this study	1	**c.6162+5 G>A**	**Splice error**

TMC1[†]	NM_138691.2	Kurima, et al., 2002	1	27-kb deletion encompassing exons 4 and 5	ND
TMC1[†]	NM_138691.2	Kitajiri, et al., 2007	1	c.16+1G>T	Splice error
TMC1[†]	NM_138691.2	Kurima, et al., 2002; Kitajiri, et al., 2007; Imtiaz, et al., 2016; this study	13	c.100C>T	p.(Arg34*)
TMC1[†]	NM_138691.2	Kurima, et al., 2002	1	c.298del	p.(Arg100Aspfs*3)
TMC1[†]	NM_138691.2	Kurima, et al., 2002; Santos, et al., 2005a; Rehman, et al., 2015	2	c.536-8T>A	Splice error
TMC1[†]	NM_138691.2	Imtiaz, et al., 2016; this study	2	c.596A>T	p.(Asn199Ile)
TMC1[†]	NM_138691.2	Santos, et al., 2005a; Rehman, et al., 2015	1	c.830A>G	p.(Tyr277Cys)
TMC1[†]	NM_138691.2	Kurima, et al., 2002	1	c.884+1G>A	Splice error
TMC1[†]	NM_138691.2	this study	1	**c.945G>A**	**p.(Trp315*)**
TMC1[†]	NM_138691.2	Santos, et al., 2005a; Rehman, et al., 2015; this study	4	c.1114G>A	p.(Val372Met)
TMC1[†]	NM_138691.2	this study	1	**c.1143C>G**	**p.(Tyr381*)**
TMC1[†]	NM_138691.2	Imtiaz, et al., 2016	1	c.1166G>A	p.(Arg389Gln)
TMC1[†]	NM_138691.2	this study	1	**c.1209G>A**	**p.(Trp403*)**
TMC1[†]	NM_138691.2	this study	1	**c.1224+2T>C**	**Splice error**
TMC1[†]	NM_138691.2	this study	1	**c.1259G>A**	**p.(Cys420Tyr)**
TMC1[†]	NM_138691.2	Santos, et al., 2005a; Rehman, et al., 2015; this study	2	c.1333C>T	p.(Arg445Cys)
TMC1[†]	NM_138691.2	Santos, et al., 2005a; Rehman, et al., 2015	1	c.1334G>A	p.(Arg445His)
TMC1[†]	NM_138691.2	this study	1	**c.1363T>C**	**p.(Tyr455His)**
TMC1[†]	NM_138691.2	Imtiaz, et al., 2016	1	c.1404+1G>T	Splice error
TMC1[†]	NM_138691.2	Kurima, et al., 2002	1	c.1534C>T	p.(Arg512*)
TMC1[†]	NM_138691.2	Kitajiri, et al., 2007	1	c.1541C>T	p.(Pro514Leu)
TMC1[†]	NM_138691.2	Kitajiri, et al., 2007; this study	3	c.1543T>C	p.(Cys515Arg)
TMC1[†]	NM_138691.2	this study	1	**c.1728C>G**	**p.(Asn576Lys)**
TMC1[†]	NM_138691.2	this study	1	**c. 1753_1754insA**	**p.(Asn407Lysfs*2)**
TMC1[†]	NM_138691.2	Imtiaz, et al., 2016; this study	2	c.1788C>A	p.(Ser596Arg)
TMC1[†]	NM_138691.2	this study	1	**c.1810C>T**	**p.(Arg604*)**
TMC1[†]	NM_138691.2	Kurima, et al., 2002	1	c.1960A>G	p.(Met654Val)

TMC1[†]	NM_138691.2	Santos, et al., 2005a; Kitajiri, et al., 2007; Rehman, et al., 2015	4	c.2004T>G	p.(Ser668Arg)
TMC1[†]	NM_138691.2	Santos, et al., 2005a; Rehman, et al., 2015; this study	4	c.2035G>A	p.(Glu679Lys)
TMIE[†]	NM_147196.2	Santos, et al., 2006	1	c.92A>G	p.(Glu31Gly)
TMIE[†]	NM_147196.2	Naz, et al., 2002	1	c.94-2_98delAGCCCAGinsC	ND
TMIE[†]	NM_147196.2	Naz, et al., 2002; Santos, et al., 2006; this study	4	c.241C>T	p.(Arg81Cys)
TMIE[†]	NM_147196.2	this study	1	**c.250C>T**	**p.(Arg84Trp)**
TMIE[†]	NM_147196.2	Naz, et al., 2002	1	c.274C>T	p.(Arg92Trp)
TMPRSS3[†]	NM_024022 .2	Ahmed, et al., 2004; Lee, et al., 2012b; Naz, et al., 2017	4	c.208delC	p.(His70Thrfs*19)
TMPRSS3[†]	NM_024022.2	this study	2	**c.271C>T**	**p.(Arg91*)**
TMPRSS3[†]	NM_024022.2	Lee, et al., 2012b	1	c.310G>A	p.(Glu104Lys)
TMPRSS3[†]	NM_024022.2	Lee, et al., 2012b	1	c.310G>T	p.(Glu104*)
TMPRSS3[†]	NM_024022 .2	Naz, et al., 2017; this study	3	c.323–6G>A	Splice error
TMPRSS3[†]	NM_024022 .2	Ben-Yosef, et al., 2001	1	c.325C>T	p.(Arg109Trp)
TMPRSS3[†]	NM_024022 .2	Ben-Yosef, et al., 2001; Ahmed, et al., 2004; this study	3	c.581G>T	p.(Cys194Phe)
TMPRSS3[†]	NM_024022.2	Lee, et al., 2012b; this study	3	c.767C>T	p.(Ala256Val)
TMPRSS3[†]	NM_024022.2	this study	1	**c.783-1G>T**	**Splice error**
TMPRSS3[†]	NM_024022 .2	Ben-Yosef, et al., 2001; Ahmed, et al., 2004; Lee, et al., 2012b; Naz, et al., 2017; this study	18	c.1219T>C	p.(Cys407Arg)
TMPRSS3[†]	NM_024022.2	Lee, et al., 2012b	1	c.1273T>C	p.(Cys425Arg)
TPRN[†]	NM_001128228.1	Rehman, et al., 2010	2	c.42_52del	p.(Gly15Alafs*150)
TPRN[†]	NM_001128228.1	Rehman, et al., 2010; Naz, et al., 2017	1	c.44_54dup	p.(Leu19Glyfs*374)
TPRN[†]	NM_001128228.1	Rehman, et al., 2010; this study	5	c.1056G>A	p.(Trp352*)
TPRN[†]	NM_001128228.1	Rehman, et al., 2010; Naz, et al., 2017	1	c.1244delC	p.(Pro415Argfs*67)
TRIOBP[†]	NM_001039141.2	Riazuddin, et al., 2006b	1	c.1193_1195delAAC	p.(Gln398del)
TRIOBP[†]	NM_001039141.2	Riazuddin, et al., 2006b	1	c.2362C>T	p.(Arg788*)
TRIOBP[†]	NM_001039141.2	Naz, et al., 2017	1	c.2968C>T	p.(Arg990*)
TRIOBP[†]	NM_001039141.2	Riazuddin, et al., 2006b	1	c.3202C>T	p.(Arg1068*)
TRIOBP[†]	NM_001039141.2	this study	1	**c.3460_3461delCT**	**p.(Leu1154Alafs*29)**

TRIOBP†	NM_001039141.2	this study	1	**c.3634_3646delCTGATCCCCCAAG**	**p.(Leu1212Cysfs*22)**
USH1C	NM_005709.3	Ahmed, et al., 2002; this study	3	c.238dup	p.(Arg80Profs*69)
USH1C	NM_005709.3	this study	1	**c.463C>T**	**p.(Arg155*)**
USH1C	NM_005709.3	Ahmed, et al., 2002	1	c.496+1G>A	Splice error
USH1C	NM_005709.3	Naz, et al., 2017	1	c.605dupC	p.(Gly203Trpfs*47)
USH1C	NM_005709.3	Ahmed, et al., 2002	1	c.674+2T>G	Splice error
USH1G	NM_173477.4	Naz, et al., 2017	1	c.163_164+13del	ND
USH1G	NM_173477.4	this study	1	**c.511G>T**	**p.(Glu171*)**
USH1G	NM_173477.4	this study	1	**c.812delC**	**p.(Pro271Argfs*52)**
USH2A	NM_206933.2	this study	1	**c.3661C>T**	**p.(Gln1221*)**
WHRN	NM_015404.3	this study	1	**c.2388_2389delCG**	**p.(Asn796Lysfs*46)**

Variants identified for the first time in our extended cohort are given in bold. (†) indicates genes associated with non-syndromic autosomal recessive hearing loss that were discovered through studies in Pakistani pedigrees. (*) indicates stop codon.

Supp. Table S5. American College of Medical Genetics (ACMG) classification of the new variants identified in our study.

Gene	Variant	Protein change	ACMG classification
ADGRV1	c.1055C>T	p.(Pro352Leu)	Likely pathogenic
CDH23	c.6083A>C	p.(Asp2028Ala)	Likely pathogenic
CDH23	c.6202A>C	p.(Thr2068Pro)	Likely pathogenic
CDH23	c.8351_8352insCGAT	p.(Leu2785Aspfs*43)	Pathogenic
CLCNKA	c.1985G>T	p.(Gly662Val)	Likely pathogenic
EPS8	c.205-8A>G	Splice error	Pathogenic
ESPN	c.2496_2496delC	p.(Tyr832*)	Pathogenic
GJB2	c.355G>T	p.(Glu119*)	Pathogenic
GPSM2	c.138C>A	p.(Phe46Leu)	Likely pathogenic
GRXCR1	c.655G>A	p.(Glu219Lys)	Likely pathogenic
ILDR1	c.900delG	p.(Thr301Profs*20)	Pathogenic
ILDR1	c.1005_1005delG	p.(Glu335Aspfs*30)	Pathogenic
ILDR1	c.1029_1030delTT	p.(Trp344Glyfs*17)	Pathogenic
ILDR1	c.1384C>T	p.(Arg462*)	Pathogenic
LHFPL5	c.452 G>T	p.(Gly151Val)	Likely pathogenic
MARVELD2	c.1223_1224insA	p.(Glu408Argfs*4)	Pathogenic
MSRB3	c.412-1G>A	Splice error	Pathogenic
MYO7A	c.20G>T	p.(Gly7Val)	Likely pathogenic
MYO7A	c.247C>A	p.(Arg83Ser)	Likely pathogenic
MYO7A	c.2339delG	p.(Gly780Valfs*10)	Pathogenic
MYO7A	c.3364C>A	p.(Leu1122Ile)	Likely pathogenic
MYO7A	c.3590T>C	p.(Leu1197Pro)	Likely pathogenic
MYO7A	c.3728C>G	p.(Pro1243Arg)	Likely pathogenic
MYO7A	c.4505A>G	p.(Asp1502Gly)	Likely pathogenic

MYO7A	c.5326+3A>G	Splice error	Pathogenic
MYO7A	c.5345G>C	p.(Gly1782Ala)	Likely pathogenic
MYO7A	c.5522C>G	p.(Thr1841Arg)	Likely pathogenic
MYO7A	c.5856+5G>C	Splice error	Pathogenic
MYO15A	c.3505C>T	p.(Arg1169*)	Pathogenic
MYO15A	c.4570C>T	p.(Gln1524*)	Pathogenic
MYO15A	c.5005C>T	p.(Gln1669*)	Pathogenic
OTOF	c.1550T>C	p.(Leu517Pro)	Likely pathogenic
OTOF	c.1904T>A	p.(Val635Asp)	Likely pathogenic
OTOF	c.3376dupA	p.(Ile1126Asnfs*51)	Pathogenic
OTOF	c.3514C>T	p.(Arg1172Trp)	Likely pathogenic
OTOF	c.4799+1G>A	Splice error	Pathogenic
OTOF	c.5714G>T	p.(Gly1905Val)	Likely pathogenic
OTOG	c.7235delG	p.(Arg2412Hisfs*77)	Pathogenic
PCDH15	deletion of exons 14 and 15		Pathogenic
PCDH15	c.788C>A	p.(Pro263Gln)	Likely pathogenic
PJVK	c.158C>G	p.(Ser53*)	Pathogenic
PJVK	c.162_172del	p.(Pro55Glufs*23)	Pathogenic
PJVK	c.406C>T	p.(Arg136*)	Pathogenic
PJVK	c.908_910delACA	p.(Asn303del)	Pathogenic
POU3F4	c.478C>T	p.(Gln160*)	Pathogenic
POU4F3	c.374C>T	p.(Pro125Leu)	Likely pathogenic
PTPRQ	c.5158_5159delAT	p.(Ile1720Glnfs*7)	Pathogenic
PTPRQ	c.6739-1G>A	Splice error	Pathogenic
SLC26A4	c.42delC	p.(Glu15Serfs*51)	Pathogenic
SLC26A4	c.154A>T	p.(Lys52*)	Pathogenic
SLC26A4	c.317C>T	p.(Ala106Val)	Likely pathogenic
SLC26A4	c.1264-3C>G	Splice error	Pathogenic
TECTA	c.840_841insT	p.(Val281Cysfs*11)	Pathogenic

TECTA	c.1247_1248delGG	p.(Gly416Aspfs*24)	Pathogenic
TECTA	c.1774G>A	p.(Val592Met)	Likely pathogenic
TECTA	c.2736 C>A	p.(Cys912*)	Pathogenic
TECTA	c.6162+5G>A	Splice error	Pathogenic
TMC1	c.945G>A	p.(Trp315*)	Pathogenic
TMC1	c.1143C>G	p.(Tyr381*)	Pathogenic
TMC1	c.1209G>A	p.(Trp403+)	Pathogenic
TMC1	c.1220dup	p.(Asn407Lysfs*2)	Pathogenic
TMC1	c.1224+2T>C	Splice error	Pathogenic
TMC1	c.1259G>A	p.(Cys420Tyr)	Likely pathogenic
TMC1	c.1728C>G	p.(Asn576Lys)	Likely pathogenic
TMPRSS3	c.783-1G>T	Splice error	Pathogenic
TRIOBP	c.3460_3461delCT	p.(Leu1154Alafs*29)	Pathogenic
TRIOBP	c.3634_3646delCTGATCCCCCAA	p.(Leu1212Cysfs*22)	Pathogenic
USH1G	c.511G>T	p.(Glu171*)	Pathogenic
USH1G	c.812delC	p.(Pro271Argfs*52)	Pathogenic
USH2A	c.3661C>T	p.(Gln1221*)	Pathogenic
WHRN	c.2388_2389delCG	p.(Asn796Lysfs*46)	Pathogenic

The classification of the new variants was done according to the ACMG guidelines (Richards, et al., 2015).

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