Table S1: The ACMG/AMP criteria used for each variant in Subgroup 1

Gene	Variant	ACMG Criteria	Classification	Reference
SIN3A NM_001145358.2	c.3418C>T, p.(Arg1140*)	PVS1, PM2_Sup, PM6	P	This study
SOX10 NM_006941.4	c.698-4_698-2delinsATG, p.?	PVS1_Strong, PM2_Sup, PM6	LP	This study
	c.376_377insG, p.(Tyr126*)	PVS1, PM2_Sup, PM6	LP	This study
SETD5 NM_001080517.3	c.1333C>T, p.(Arg445*)	PVS1, PM2_Sup, PM6	P	(Grozeva et al. 2014)
	c.2182del, p.(Asp728Ilefs*9)	PVS1, PM2_Sup, PM6	P	This study
ANKRD11 NM_013275	c.6836_6837del p.(Val2279Glyfs*16)	PVS1, PM2_Sup, PS4_Sup	P	(Bestetti et al. 2022; Loberti et al. 2022; Peluso et al. 2023; Wang et al. 2021)
SMARCA4 NM_001387283.1	c.761G>T, p.(Gly254Val)	PM2_Sup, PP3_Sup (AlphaMissense: 0.254, CADD: 33, REVEL: 0.594)	VUS	This study
RPS6KA3 NM_004586.3	c.1741A>G, p.(Thr581Ala)	PM2_Sup, PM6, PP3_Mod, (AlphaMissense: 0.9939, CADD: 28.5, REVEL: 0.663), PP4	LP	(Zhou et al. 2022)
PTPN11 NM_002834.3	c.5C>T, p.(Thr2Ile)	PM2_Sup, PM6, PP3_Sup (AlphaMissense: 0.9335, CADD: 31, REVEL: 0.213), PS4_Mod	LP	(Leach et al. 2019)
BRAF NM_004333.6	c.2204G>A, p.(Arg735Gln)	PM2_Sup, PP3_Sup (AlphaMissense: 0.9661, CADD: 33, REVEL: 0.476)	VUS	This study
POGZ NM_015100.4	c.2569A>G, p.(Arg857Gly)	PS2, PM2_Sup, PP3 (SpliceAI Δ score: 0.83 (Acceptor loss), 0.90 (Donor loss))	LP	This study
KARS1 NM_001130089.2	c.223del, p.(Gln75Serfs*2)	PVS1, PM2_Sup PP4	P	(Cappuccio et al. 2021)
	c.1028T>C, p.(Val343Ala)	PM2_Sup, PM3, PP3_Mod (AlphaMissense: 0.9179, CADD: 28.2, REVEL: 0.876), PP4	LP	This study

	c.379T>C, p.(Phe127Leu)	PM2_Sup, PM3_Mod, PP3_Sup (AlphaMissense: 0.992, CADD: 27.4, REVEL: 0.570), PS4_Sup	LP	(Lin et al. 2021; Ardissone et al. 2018; AlAbdi et al. 2023)
	c.889C>G, p.(Pro297Ala)	PM2_Sup, PM3_Sup, PP3_Mod (AlphaMissense: 0.841, CADD: 29, REVEL: 0.957)	VUS	(Lin et al. 2021)
	c.1124A>G, p.(Tyr375Cys)	PM2_Sup, PM3_Mod, PP3_Mod (CADD: 31, REVEL: 0.914)	VUS	(Lin et al. 2021)
HSD17B4 NM_000414.4	c.394C>T, p.(Arg132Trp)	PM2_Sup, PM3, PP3_Sup (CADD:26.1, REVEL: 0.874), PS4_Mod	LP	(Ferdinandusse et al. 2006)
TWNK NM_021830.3	c.874C>A, p.(Pro292Thr)	PM2_Sup, PP3_Sup (AlphaMissense: 0.1908, CADD: 21.9, REVEL: 0.729)	VUS	(Jamali et al. 2019)
CASK NM_003688.3	c.173-2331_278+1542del	PVS1, PM2_Sup, PM6	P	This study
BBS1 NM_024649.4	c.1642del, p.(Leu548Trpfs*31)	PVS1_Strong, PM2_Sup, PP4	LP	(Beales et al. 2003)
FRAS1 NM_025074.6	c.9607G>T, p.(Asp3203Tyr)	PM2_Sup, PP3_Mod (AlphaMissense: 0.706, CADD: 31, REVEL: 0.920), PP4	VUS	This study
GALC NM_000153.4	c.328+1G>T, p.?	PVS1, PM2_Sup, PM3_Sup	P	(Capalbo et al. 2019)
PEX1 NM_000466.2	c.3116C>T, p.(Thr1039Ile)	PM2_Sup, PP3_Sup (AlphaMissense: 0.8202, CADD: 26.7, REVEL: 0.879)	VUS	This study
CACNA1D NM_000720.3	c.1580G>A, p.(Arg527His)	PM2_Sup, PP3_Sup (AlphaMissense: 0.3043, CADD: 25.8, REVEL: 0.694)	VUS	This study
LRP2 NM_004525.2	c.1342-2A>T, p.?	PVS1, PM2_Sup, PP4	P	This study
ELOVL4 NM_022726.3	c.424A>G, p.(Thr142Ala)	PM2_Sup, PP3_Mod (AlphaMissense: 0.9926, CADD: 29.7, REVEL: 0.838)	VUS	(Alabdulrazzaq et al. 2023)
AAAS NM_015665.5	c.398_399+2del. p.(Leu134Glnfs*8)	PVS1, PM2_Sup, PM3_Sup	P	(Yildirim et al. 2023)
SGSH NM_000199.5	c.364G>A, p.(Gly122Arg)	PS3, PM1, PM2_Sup, PP3_Sup (AlphaMissense: 0.9781, CADD: 25.7, REVEL: 0.938)	LP	(Sidhu et al. 2014)

KMT2D NM_003482.4	c.993dup, p.(Glu332Argfs*10)	PVS1, PM2_Sup, PM6	P	This study
FGF3 NM_005247.4	c.166C>T, p.(Leu56Phe)	PM2_Sup, PP3_Sup (AlphaMissense: 0.9468, CADD: 26.7, REVEL: 0.788)	VUS	(Doll et al. 2020)

Abbreviations: Sup, supporting; Mod, moderate; P, pathogenic; LP, likely pathogenic; VUS, variant of unknown significance

Table S2: The ACMG/AMP criteria used for each variant in Subgroup 2

Gene	**Variant**	**ACMG Criteria**	**Classification**	**Reference**
GATA3 NM_001002295.2	c.1059delA, p.(Arg353Serfs*3)	PVS1, PM2_Sup, PM6	P	This study
POLR1D NM_015972.3	c.161C>A, p.(Ser54Tyr)	PM1, PM2_Sup, PP3_Mod (AlphaMissense: 0.9562, CADD: 29.1 REVEL: 0.793)	VUS	This study
USH2A NM_206933.4	c.11864G>A, p.(Trp3955*)	PVS1, PM2_Sup, PM3_Strong	P	(van Wijk et al. 2004)
	c.653T>A, p.(Val218Glu)	PM2_Sup, PM3_Strong, PP3 (AlphaMissense: 0.8241, CADD: 25.5, REVEL: 0.858)	LP	(Besnard et al. 2014)
	c.2299del, p.(Glu757Serfs*21)	PVS1, PM2_Sup, PM3_Strong	P	(Aller et al. 2010)
	c.920_923dup, p.(His308Glnfs*16)	PVS1, PM2_Sup, PM3_ Strong	P	(Corton et al. 2013)
	c.7595-2144A>G, p.?	PM2_Sup, PM3_Very Strong, PS3_Sup	P	(Vache et al. 2012)
	Exon 14 del	PVS1, PM2_Sup, PM3_Mod	P	(Glockle et al. 2014)
	Exon 44 del	PVS1_Mod, PM2_Sup, PM3_Mod, PP4	LP	(Glockle et al. 2014)
	c.848+1G>T, p.?	PVS1, PM2_Mod	LP	(Mansard et al. 2021)
ADGRV1 NM_032119.3	c.12982G>T, p.(Glu4328*)	PVS1, PM2_Sup, PP4	P	(Turro et al. 2020)
	c.4458T>G, p.(Tyr1486*)	PVS1, PM2_Sup, PP4	P	This study
MYO7A NM_000260.3	c.4361T>G, p.(Val1454Gly)	PS1, PM2_Sup, PM3_Sup	LP	This study
	c.6231dup, p.(Lys2078Argfs*50)	PVS1, PM2_Sup, PM3_Sup	P	(Neuhaus et al. 2017)
	c.577A>C, p.(Thr193Pro)	PM2_Sup, PM3_Sup	VUS	(Sloan-Heggen et al. 2015)
	c.1190C>A, p.(Ala397Asp)	PS1, PM2_Sup, PM3_Sup, PP3_Sup (AlphaMissense: 0.9955, CADD: 26.5, REVEL: 0.942)	LP	(Watanabe et al. 2008)

CDH23 NM_022124.5	c.2349C>A, p.(Tyr783*)	PVS1, PM2_Sup, PM3_Sup	P	This study
	c.9040del, p.(Val3014*)	PVS1, PM2_Sup, PM3_Sup	P	This study
	Exons 4-6 del	PVS1, PM2_Sup, PM3_Sup	P	(Weisschuh et al. 2020; Lin et al. 2024)
	c.6050-70G>A, p.?	PM2_Sup, PP3	VUS	This Study
USH1C NM_153676.3	c.388-1G>C, p.?	PVS1, PS1, PM2_Sup	P	This study
SOX10 NM_006941.3	c.373C>T, (p.Gln125*)	PVS1, PM2_Sup, PM6	P	(Wakabayashi et al. 2021)
	c.137del, p.(Pro46Argfs*63)	PVS1, PM2_Sup	LP	This study
	c.378C>G, p.(Tyr126*)	PVS1, PM2_Sup, PS4_Sup	P	(Li et al. 2021)
	c.230del, p.(Ser77Thrfs*32)	PVS1, PM2_Sup, PM6	P	This study
EDN3 NM_207034.2	c.472C>T, p.(Arg158Cys)	PM2_Sup, PP3_Sup (AlphaMissense: 0.8773, CADD: 25, REVEL: 0.68), PP4	VUS	This study
HSD17B4 NM_000414.4	c.338A>T, p.(Asp113Val)	PM2_Sup, PP3_Sup (AlphaMissense: 0.2127, CADD: 26.6, REVEL: 0.796)	VUS	This study
LARS2 *NM_015340.4*	c.1565C>A, p.(Thr522Asn)	PS3_Sup, PM2_Sup, PM3_Strong_ PP3_Sup (AlphaMissense: 0.829, CADD: 24.9, REVEL: 0.844)	LP	(Demain et al. 2017)
OPA1 NM_130837.2	c.1819T>C, p.(Phe607Leu)	PM2, PM6, PP3_Mod (AlphaMissense: 0.9994, CADD: 29.4, REVEL: 0.86)	LP	This study
SLC29A3 NM_018344.5	c.1309G>A, p.(Gly437Arg)	PM1, PM2_Sup, PM3, PP3_Sup (AlphaMissense: 0.9613, CADD: 26.9, REVEL: 0.69)	LP	(El-Bassyouni, Thomas, and Tosson 2020)
	c.610+1G>A, p.?	PVS1, PS1, PM2_Sup	P	(Rezaie et al. 2024)
	c.1087C>T, (p.Arg363Trp)	PM2_Sup, PM3_Mod, PM5, PP3_Sup (AlphaMissense: 0.6113, CADD: 27, REVEL: 0.829)	LP	(Bloom et al. 2017)

COL4A3 NM_000091.4	c.172G>A, p.(Gly58Ser)	PM2_Sup, PP3_Sup (AlphaMissense: 0.2845, CADD: 25.4, REVEL :0.934)	VUS	(Moriniere et al. 2014)
	c.4347_4353del, p.(Arg1450Valfs*77)	PVS1, PM2_Sup, PS4_Sup, PM3_Sup	P	(Storey et al. 2013; Ding et al. 1995)
COL4A3 NM_000091.4	c.4862C>T, p.(Thr1621Met)	PM2_Sup, PP3_Sup (CADD: 26.2, REVEL: 0.801)	VUS	This study
COL4A3 NM_000091.4	c.3882+5G>A	PS1, PS4_Sup, PM2_Sup, PP3	LP	(Daga et al. 2018; Zhang et al. 2012)
RAF1 NM_002880	c.1922C>T, p.(Thr641Met)	PM2_Sup, PS4_Sup, PP3 (CADD: 25.6, REVEL: 0.713)	VUS	(Dhandapany et al. 2014)
KCNQ1 NM_000218.2	c.912G>A, p.(Trp304*)	PVS1, PM2_Sup	LP	This study
	c.514G>A, p.(Val172Met)	PS2_Sup, PM2_Sup	VUS	(Bdier et al. 2017)
	c.877C>T p.(Arg293Cys)	PS2_Sup, PM2_Sup	VUS	(Bdier et al. 2017)
	c.683+1G>A, p.?	PVS1, PM2_Sup	LP	(e and e 2019)
	c.1265dup, p.(Phe423Valfs*40)	PVS1, PS4_Sup, PM2_Sup	P	(Akgun-Dogan et al. 2022; Abolhassani et al. 2024)
BSND NM_057176.3	c.64G>A, p.(Gly22Ser)	PM2_Sup, PP3_Sup (AlphaMissense: 0.868, CADD: 27.2, REVEL: 0.783)	VUS	This study
SLC19A2 NM_006996.2	c.697C>T, p.(Gln233*)	PVS1, PM1, PM2_Sup	P	(Mohsen-Pour et al. 2022)
LHX3 NM_014564.5	c.331G>A, p.(Ala111Thr)	PM2_Sup, PP3_Sup (AlphaMissense: 0.9817, CADD: 27.9, REVEL: 0.899)	VUS	This study
	c.353T>G, p.(Leu118Arg)	PM2_Sup, PP3_Mod (AlphaMissense: 0.9724, CADD: 29.6, REVEL: 0.91)	VUS	This study
COL9A3 NM_001853.3	c.355del, p.(Leu119Serfs*10)	PVS1, PM2_Sup	P	(Rad et al. 2022)
PAX3 NM_181458.4	c.808C>T, p.(Arg270Cys)	PS1, PS4_Mod, PM2_Sup, PP3_Mod (AlphaMissense: 0.9996, CADD:32, REVEL: 0.935)	P	This study
EYA1 NM_000503.6	c.639+98_760del, p.(Asp214Profs*112)	PVS1, PM2_Sup, PP4	P	This study

Abbreviations: Sup, supporting; Mod, moderate; P, pathogenic; LP, likely pathogenic; VUS, variant of unknown significance

Table S3: The ACMG/AMP criteria used for each variant in Subgroup 3

Gene	Variant	ACMG Criteria	Classification	Phenotypes in the literature	Reference
SLC26A4 NM_000441.1	c.1001+1G>A, p.?	PVS1, PM2_Sup, PM3_Strong	P	Pendred, HL with enlarged vestibular aqueduct, NSHL	(Nakano et al. 2022) (de Moraes et al. 2013) (Mikkelsen, Tranebjaerg, and Mey 2019)
	c.1334T>G, p.(Leu445Trp)	PM2_Sup, PM3_ Very Strong, PP3_Mod (AlphaMissense: 0.9933, CADD: 29.3, REVEL: 0.965)	P	Pendred, HL with enlarged vestibular aqueduct, NSHL	(Smits et al. 2022) (Cengiz et al. 2017) (Rebeh et al. 2010; Ladsous et al. 2014)
	c.919-2A>G, p.?	PVS1, PM2, PM3_Strong	P	Pendred, HL with enlarged vestibular aqueduct	(Zhao et al. 2024) (Nakano et al. 2022; Li et al. 2012) (Cengiz et al. 2017) (Yang et al. 2005)
	c.2027T>A, p.(Leu676Gln)	PM2_Sup, PM3_ Very Strong, PP3_Sup (AlphaMissense: 0.8935, CADD: 26.3, REVEL: 0.733)	P	Pendred, HL with enlarged vestibular aqueduct	(Yoon et al. 2008) (Liu et al. 2021) (Lu et al. 2022) (Baldyga et al. 2023)
	c.578C>T, p.(The193Ile)	PM2_Sup, PS3_Sup, PM3_ Strong, PP3_Sup (AlphaMissense: 0.9082, CADD: 23, REVEL: 0.854)	LP	HL with enlarged vestibular aqueduct, NSHL	(de Moraes et al. 2016) (Cengiz et al. 2017)
	c.1238A>G, p.(Gln413Arg)	PS1, PM2_Sup, PS3_Sup, PM3_ Strong, PP3_Sup (AlphaMissense: 0.8859, CADD: 26.7, REVEL: 0.949)	P	Pendred, HL with enlarged vestibular aqueduct, NSHL	(de Moraes et al. 2016) (Kuhnen et al. 2014) (Huang et al. 2011) (Wang et al. 2014)
	c.716T>A, p.(Val239Asp)	PM2_Sup, PM3_Very Strong, PP3_Sup (AlphaMissense: 0.9612, CADD: 27, REVEL: 0.935)	P	Pendred, NSHL	(Walsh et al. 2006) (Soh et al. 2015) (Azadegan-Dehkordi et al. 2018)
	c.170C>A, p.(Ser57*)	PVS1, PM2_Sup, PM3_Mod	P	HL with enlarged vestibular aqueduct	(Kinoglu et al. 2020)
	c.212T>A, p.(Ile71Asn)	PM2_Sup, PM3_Mod, PP3_Sup (AlphaMissense: 0.7472, CADD: 25.2, REVEL: 0.646)	VUS	Pendred	(Said et al. 2022)
	c.415+2T>C, p.?	PVS1, PM2_Sup, PM3_Mod	P	HL with enlarged vestibular aqueduct	(Yuan et al. 2012) (Tian et al. 2021)
	c.704A>G, p.(Gln235Arg)	PM2_Sup, PM3_ Strong, PP3_Sup (AlphaMissense: 0.9444, CADD: 26.3, REVEL: 0.969)	LP	Pendred	(Tesolin et al. 2021)

SLC26A4 NM_000441.1	c.1223C>A, p.(Ser408Tyr)	PM2_Sup, PP3_Mod_PM5 (AlphaMissense: 0.9937, CADD: 29.5, REVEL: 0.957)	VUS	This study	This study
	c.2106del, p.(Lys702Asnfs*19)	PVS1, PM2_Sup, PM3_Sup	P	HL with enlarged vestibular aqueduct	(Yazdanpanahi et al. 2012) (Zhao et al. 2019)
	c.845G>A, p.(Cys282Tyr)	PM2_Sup, PM3_ Strong, PP3_Sup (AlphaMissense: 0.9444, CADD:26.3, REVEL: 0.969)	LP	HL with enlarged vestibular aqueduct	(Kinoglu et al. 2020) (Baldyga et al. 2023)
	c.1028_1029insTCAG, p.(Ser344Glnfs*34)	PVS1, PS1, PM2_Sup	P	This study	This study
	c.1001G>T, p.(Gly334Val)	PS1, PM2_Sup, PM3_ Strong, PP3_Sup (AlphaMissense: 0.7329, CADD: 35, REVEL: 0.961, SpliceAI Δ scores: Donor Loss: 0.46, Donor Gain: 0.64)	P	HL with enlarged vestibular aqueduct	(Cengiz et al. 2017)
	c.1234G>T, p.(Val412Phe)	PM2_Sup, PM3_Mod, PP3_Mod (AlphaMissense: 0.8251, CADD:29.7,REVEL: 0.937)	LP	HL no more comments	(Azaiez et al. 2018)
	c.1198del, p.(Cys400Valfs*32)	PVS1, PM2_Sup, PM3_Sup	P	HL with enlarged vestibular aqueduct	(Cengiz et al. 2017)
CDH23 NM_022124.5	c.2398-1G>T, p.?	PVS1, PM2_Sup, PM3_Sup	P	Retinal disease	(Ellingford et al. 2016)
USH1C NM_153676	c.463C>T, p.(Arg155*)	PVS1, PM2_Sup, PM3_Sup	P	Usher syndrome	(Huang et al. 2015) (Bonnet et al. 2016) (Hanany, Rivolta, and Sharon 2020)

Abbreviations: Sup, supporting; Mod, moderate; P, pathogenic; LP, likely pathogenic; VUS, variant of unknown significance; HL, hearing loss; NSHL, non-syndromic hearing loss

Table S4: Sequencing methods and analysis summaries

Patient ID	Center	Method	Data analysis and filtering strategy
Patient 1-22, 57, 59	Würzburg	WES	Koparir et al., 2024 (Koparir et al. 2024)
Patient 23-29	Würzburg	WGS	Bengl et al., 2025 (Bengl et al. 2025)
Patient 32-38	Istanbul	WES	Anitha et al., 2024 (Anitha et al. 2024)
Patient 39-56,58	Tübingen/Göttingen	WES	Redfield et al., 2024 (Redfield et al. 2024)
Patient 60-75	Würzburg	WES	Bahena et al., 2022 (Bahena et al. 2022)
Patient 76-104	Tübingen/Göttingen	WES	Redfield et al.,2022 (Redfield et al. 2024)
Patient 105-108	Tübingen	MIP	Reurink et al., 2021 (Reurink et al. 2021)
Patient 109,110	Tübingen	WGS	Falb et al., 2023 (Falb et al. 2023)
Patient 111-114	Tübingen/Göttingen	Sanger sequencing	Shadab et al., 2025 (Shadab et al. 2025)

WES, whole-exome sequencing; WGS, whole-genome sequencing; MIP, Molecular inversion probe; NA, not available.

Table S5. Molecular inversion probe 89 gene panel content

Gene name	Gene name	Gene name	Gene name	Gene name	Gene name
ACTG1	*DCDC2*	*GRM8*	*MYH9*	*POU3F4*	*TECTA*
ADCY1	*DIABLO*	*GRXCR1*	*MYO15A*	*POU4F3*	*TJP2*
BDP1	*DIAPH1*	*GRXCR2*	*MYO3A*	*PRPS1*	*TMC1*
BSND	*ELMOD3*	*GSDME (DFNA5)*	*MYO6*	*PTPRQ*	*TMEM132E*
CABP2	*EPS8*	*HGF*	*MYO7A*	*RDX*	*TMIE*
CCDC50	*ESPN*	*ILDR1*	*NAT2*	*RIPOR2 (FAM65B)*	*TMPRSS3*
CDH23	*ESRRB*	*KARS*	*OSBPL2*	*SERPINB6*	*TNC*
CEACAM16	*EYA4*	*KCNQ4*	*OTOA*	*SIX1*	*TPRN*
CIB2	*GIPC3*	*LHFPL5*	*OTOF*	*SLC17A8*	*TRIOBP*
CLDN14	*GJB2*	*LOXHD1*	*OTOG*	*SLC26A4*	*TSPEAR*
CLIC5	*GJB3*	*LRTOMT*	*OTOGL*	*SLC26A5*	*USH1C*
COCH	*GJB6*	*MARVELD2*	*P2RX2*	*SMPX*	*USH1G*
COL11A2	*GPSM2*	*MIR96*	*PCDH15*	*STRC*	*WHRN (DFNB31)*
COL4A6	*GRHL2*	*MSRB3*	*PJVK (DFNB59)*	*SYNE4*	*WFS1*
CRYM	*GRM7*	*MYH14*	*PNPT1*	*TBC1D24*	

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