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Hearing Loss Panel Gene and Disease List

Disease	Genes
DFNA	ACTB, CCDC50, CEACAM16, COCH, CRYM, ICERE1, DIABLO, DIAPH 1, DSPP, EYA4,GJB3, GRHL2, HOMER2, KCNQ4, KITLG, MCM2, miR9 6, MYH14, MYH9, P2RX2, POU4F3, SLC17A8, SLC26A5, TBC1D24, TJ P2, TMC1,TNC, WFS1
DFNB	ACTG1, CABP2, CDC14A, CLDN14, ELMOD3, ESPN, ESRRB, GIPC3, GJB2, GJB6, GRXCR1, GRXCR2, HGF, ILDR1, KARS, CEMIP, LHFPL5, L OXHD1, LRTOMT, MARVELD2, MET, MSRB3, MYO15A, MYO3A, MY O6, OTOA, OTOF, OTOG, OTOGL, PDZD7(DFNB57), PJVK, PNPT1, PT PRQ, RDX, RIPOR2, S1PR2, SERPINB6, STRC, SYNE4, TECTA, TMIE, T MPRSS3, TPRN, TRIOBP
DFNX	AIFM1, POU3F4, PRPS1
Aminoglycoside-Associated Deafness Genes	MT-CO1, MT-TE, MT-RNR1
Mitochondria-Associated Hearing Loss Genes	MT-TD, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TQ, MT-TS1, MT-TS2
Other Deafness-Associated Genes	ECE1, EDNRA, ERCC2, ERCC3, FAS, GJB4, GSTP1, HAL, FGF3, FGFR3, GATA3, GJA1, JAG1, LHX3, NF2, MIR182, MIR183, MTAP, MYO1A, M YO1C, MYO1F, NDP, NR2F1, OTOR, PMP22, SLC4A11, SNAI2, SOX2, SPINK5, TBL1X, TCF21, TFCP2, TMPRSS5
Distal Renal Tube Acidsis and Progressive Deafness	ATP6V1B1
DDOD Syndrome	ATP6V1B2
Bjornstad Syndrome (BJS)	BCS1L

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Sensorineural Deafness with Mild Renal Dysfunction; Bartter Syndrome, Type 4a	BSND (DFNB73)
Biotinidase Deficiency	BTD
Male Infertility	CATSPER2
Usher Syndrome	MYO7A (USH1B/DFNB2/DFNA11), USH1C (DFNB18/PDZD7C), CDH23 (USH1D/DFNB12), PCDH15 (USH1F/DFNB23), USH1G (SANS), CIB2 (USH1J/DFNB48),USH2A (Usherin), ADGRV1 (USH2B/2C), WHRN (USH2D/DFNB31),CLRN1 (USH3A)
Stickler Syndrome	COL11A1 (STL2), COL11A2 (STL3/DFNA13/DFNB53), COL2A1 (STL1), COL9A2 (STL5), COL9A3
Alport Syndrome (X-linked)	COL4A3, COL4A4, COL4A5, COL4A6 (DFNX6)
Waardenburg Syndrome (WS)	EDNRB, EDN3, MITF, PAX3, SMPX, SOX10
Branchiootorenal Syndrome (BOR)	EYA1, SIX1(DFNA23), SIX5
Enlarged Vestibular Aqueduct (EVA)	FOXI1, KCNJ10, SLC26A4 (Pendred Syndrome)
X-linked Dominant Charcot-Marie-Tooth Disease	GJB1(Cx32), TIMM8A
Chudley-McCullough Syndrome	GPSM2
Jervell and Lange-Nielsen Syndrome, Deafness and LQT	KCNE1, KCNQ1
Refsum Disease	PEX7, PHYH