

**PANEL NMG 18 HEARING LOSS**

**Syndromic Hearing Loss**

COL4A5	Alport syndrome
COL4A3	Alport syndrome, autosomal dominant/recessive
COL4A4	Alport syndrome, autosomal recessive
EYA1	Branchiootic syndrome 1
SIX5	Branchiootorenal syndrome 2
SIX1	Branchiootic syndrome 3
ATP1A3	CAPOS syndrome
CHD7	CHARGE syndrome
SEMA3E	
GPSM2	Chudley-McCullough syndrome
NIPBL	Cornelia de Lange syndrome
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss
PEX1	Heimler syndrome
KCNQ1	Jervell and Lange-Nielsen syndrome
KCNE1	
GDF6	
FGFR3	Klippel-Feil syndrome , autosomal dominant
NDP	Muenke syndrome
FOXI1	Norrie disease
SLC26A4	
HSD17B4	Perrault syndrome 1
HARS2	Perrault syndrome 2
CLPP	Perrault syndrome 3
LARS2	Perrault syndrome 4
KCNJ10	SESAME syndrome
COL2A1	Stickler syndrome, type I
COL11A1	Stickler syndrome, type II
COL9A1	Stickler syndrome, type IV
COL9A2	Stickler syndrome, type V
TCOF1	Treacher Collins syndrome 1
POLR1D	Treacher Collins syndrome 2
POLR1C	Treacher Collins syndrome 3
MYO7A	Usher syndrome, type 1B
USH1C	Usher syndrome, type 1C
CDH23	Usher syndrome, type 1D
PCDH15	Usher syndrome, type 1F/1D
USH1G	Usher syndrome, type 1G
USH2A	Usher syndrome, type 2A
DFNB31	Usher syndrome type 2B
GPR98	Usher syndrome, type 2C
CLRN1	Usher syndrome, type 3A
HARS	Usher syndrome type 3B
PDZD7	Usher syndrome, type IIC
CIB2	Usher syndrome, type IJ
VHL	von Hippel-Lindau syndrome
PAX3	Waardenburg syndrome, type 1/ type 3
MITF	Waardenburg syndrome, type 2A
SNAI2	Waardenburg syndrome, type 2D
SOX10	Waardenburg syndrome, type 2E / 4C
EDNRB	Waardenburg syndrome, type 4A
EDN3	Waardenburg syndrome, type 4B
COL11A2	Weissenbacher-Zweymuller syndrome
CISD2	Wolfram syndrome
WFS1	Wolfram-like syndrome, autosomal dominant

**NON-Syndromic Hearing Loss**

**Autosomal Dominant**

DIAPH1	Deafness, autosomal dominant 1
KCNQ4	Deafness, autosomal dominant 2A
GJB3	Deafness, autosomal dominant 2B Deafness, autosomal dominant, with peripheral neuropathy Deafness, autosomal recessive
GJB2	Deafness, autosomal dominant 3A Deafness, autosomal recessive 1A
GJB6	Deafness, autosomal dominant 3B Deafness, autosomal recessive 1B
MYH14	Deafness, autosomal dominant 4A
CEACAM16	Deafness, autosomal dominant 4B

DFNA5	Deafness, autosomal dominant 5
WFS1	Deafness, autosomal dominant 6 Deafness, autosomal dominant 14 Deafness, autosomal dominant 38
TECTA	Deafness, autosomal dominant 8 Deafness, autosomal recessive 21
COCH	Deafness, autosomal dominant 9
EYA4	Deafness, autosomal dominant 10
MYO7A	Deafness, autosomal dominant 11 Deafness, autosomal recessive 2
COL11A2	Deafness, autosomal dominant 13/53
POU4F3	Deafness, autosomal dominant 15
MYH9	Deafness, autosomal dominant 17
ACTG1	Deafness, autosomal dominant 20/26
MYO6	Deafness, autosomal dominant 22 / 37
SIX1	Deafness, autosomal dominant 23
SLC17A8	Deafness, autosomal dominant 25
GRHL2	Deafness, autosomal dominant 28
TMC1	Deafness, autosomal dominant 36 Deafness, autosomal recessive 7
CRYM	Deafness, autosomal dominant 40
P2RX2	Deafness, autosomal dominant 41
CCDC50	Deafness, autosomal dominant 44
TJP2	Deafness, autosomal dominant 51
TNC	Deafness, autosomal dominant 56
DIABLO	Deafness, autosomal dominant 64
TBC1D24	Deafness, autosomal dominant 65 Deafness, autosomal recessive 86
OSBPL2	Deafness, autosomal dominant 67
HOMER2	Deafness, autosomal dominant 68
<b>Autosomal Recessive</b>	
MYO15A	Deafness, autosomal recessive 3
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct (EVA)
TMIE	Deafness, autosomal recessive 6
TMPRSS3	Deafness, autosomal recessive 8/10
OTOF	Deafness, autosomal recessive 9
CDH23	Deafness, autosomal recessive 12
GIPC3	Deafness, autosomal recessive 15
STRC	Deafness, autosomal recessive 16
USH1C	Deafness, autosomal recessive 18A
OTOG	Deafness, autosomal recessive 18B
OTOA	Deafness, autosomal recessive 22
PCDH15	Deafness, autosomal recessive 23
RDX	Deafness, autosomal recessive 24
GRXCR1	Deafness, autosomal recessive 25
TRIOBP	Deafness, autosomal recessive 28
CLDN14	Deafness, autosomal recessive 29
MYO3A	Deafness, autosomal recessive 30
DFNB31	Deafness, autosomal recessive 31
ESRRB	Deafness, autosomal recessive 35
ESPN	Deafness, autosomal recessive 36
HGF	Deafness, autosomal recessive 39
ILDR1	Deafness, autosomal recessive 42
ADCY1	Deafness, autosomal recessive 44
CIB2	Deafness, autosomal recessive 48
MARVELD2	Deafness, autosomal recessive 49
BDP1	Deafness, autosomal recessive 49
DFNB59	Deafness, autosomal recessive 59
SLC26A5	Deafness, autosomal recessive 61
LRTOMT	Deafness, autosomal recessive 63
DCDC2	Deafness, autosomal recessive 66
LHFPL5	Deafness, autosomal recessive 67
PNPT1	Deafness, autosomal recessive 70
MSRB3	Deafness, autosomal recessive 74
SYNE4	Deafness, autosomal recessive 76
LOXHD1	Deafness, autosomal recessive 77
TPRN	Deafness, autosomal recessive 79
PTPRQ	Deafness, autosomal recessive 84A
OTOGL	Deafness, autosomal recessive 84B

ELMOD3	Deafness, autosomal recessive 88
KARS	Deafness, autosomal recessive 89
SERPINB6	Deafness, autosomal recessive 91
CABP2	Deafness, autosomal recessive 93
MET	Deafness, autosomal recessive 97
TSPEAR	Deafness, autosomal recessive 98
GRXCR2	Deafness, autosomal recessive 101
EPS8	Deafness, autosomal recessive 102
CLIC5	Deafness, autosomal recessive 103
FAM65B	Deafness, autosomal recessive 104
TMEM132E	Autosomal-recessive nonsyndromic hearing loss
<b>X-linked</b>	
PRPS1	Deafness, X-linked 1
TIMM8A	Deafness, X-linked 1, progressive
POU3F4	Deafness, X-linked 2
DFNX3	Deafness, X-linked 3
SMPX	Deafness, X-linked 4
COL4A6	Deafness, X-linked 6
SLITRK6	Deafness and myopia
<b>Mitochondrial</b>	
BCS1L	Mitochondrial complex III deficiency, nuclear type 1
NARS2	Combined oxidative phosphorylation deficiency
<b>Related to other disorders</b>	
BSND	Sensorineural deafness with mild renal dysfunction
ATP6V1B1	Renal tubular acidosis with deafness
ATP6VOA4	Renal tubular acidosis, distal, autosomal recessive with late-onset hearing loss
DNAJC3	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant
SLC33A1	Congenital cataracts combined with hearing loss, and neurodegeneration
ANKH	Craniometaphyseal dysplasia
SPATA5	Epilepsy, with hearing loss, and mental retardation
OPA1	Optic atrophy with deafness
SDHD	Paraganglioma with sensorineural hearing loss
ABHD12	Polyneuropathy with hearing loss, ataxia, retinitis pigmentosa, and cataract