MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for the Hereditary Hearing Loss Panel

Next-generation sequencing (NGS), Sanger sequencing, and/or Droplet Digital PCR (ddPCR) are performed to test for the presence of a variant in these genes. Additionally, NGS and/or Multiplex Ligation-Dependent Probe Amplification (MLPA) is used to test for the presence of large deletions and/or duplications. When appropriate, reported alterations detected by NGS are confirmed by an independent reference, such as Quantitative Polymerase Chain Reaction (qPCR) and/or Sanger sequencing. Additionally, regions of homology, high GC-rich content, and repetitive sequences may not provide accurate sequence, despite the use of alternate assays.[1]

Genomic Build: Reference transcripts based on build GRCh37 (hg19) interrogated by Hereditary Hearing Loss Panel.

Hereditary Hearing Loss Panel	
Gene	**RefSeq Transcript[2]**
ABHD12	NM_001042472
ACTG1	NM_001614
ADCY1	NM_021116
ADGRV1 (GPR98)	NM_032119
AIFM1	NM_004208
ALMS1	ENST00000264448
ATP6V1B1	NM_001692
BCS1L	NM_004328
BDP1	NM_018429
BSND	NM_057176
BTD	NM_000060
CABP2	NM_016366
CACNA1D	NM_000720
CATSPER2	NM_172095
CCDC50	NM_178335
CD164	NM_006016
CDC14A	NM_033312
CDH23	NM_022124
CEACAM16	NM_001039213
CEP78	NM_001098802
CHD7	NM_017780
CIB2	NM_006383
CISD2	NM_001008388
CLDN14	NM_144492
CLIC5	NM_001114086
CLPP	NM_006012
CLRN1	NM_174878
COCH	NM_004086
COL2A1	NM_001844
COL4A3	NM_000091

Hereditary Hearing Loss Panel	
Gene	**RefSeq Transcript[2]**
COL4A4	NM_000092
COL4A5	NM_000495
COL4A6	NM_001847
COL9A1	NM_001851
COL9A2	NM_001852
COL9A3	NM_001853
COL11A1	NM_001854
COL11A2	NM_080680
CRYM	NM_001888
DCDC2	NM_016356
DFNA5 (GSDME)	NM_004403
DIABLO	NM_019887
DIAPH1	NM_005219
DIAPH3	NM_001042517
DNMT1	NM_001130823
DSPP	NM_014208
EDN3	NM_207034
EDNRB	NM_000115
ELMOD3	NM_001329793
EPS8	NM_004447
EPS8L2	NM_022772
ESPN	NM_031475
ESRRB	NM_004452
EYA1	NM_000503
EYA4	NM_004100
FGF3	NM_005247
FGFR2	NM_000141
FLNA	NM_001456
FOXC1	NM_001453
FOXI1	NM_012188

Hereditary Hearing Loss Panel	
Gene	**RefSeq Transcript[2]**
GATA3	NM_001002295
GIPC3	NM_133261
GJB2	NM_004004
GJB6	NM_006783
GPSM2	NM_013296
GRHL2	NM_024915
GRXCR1	NM_001080476
GRXCR2	NM_001080516
HARS2	NM_012208
HGF	NM_000601
HOMER2	NM_004839
HOXA2	NM_006735
HSD17B4	NM_000414
ILDR1	NM_001199799
JAG1	NM_000214
KARS	NM_001130089
KCNE1	NM_000219
KCNJ10	NM_002241
KCNQ1	NM_000218
KCNQ4	NM_004700
KITLG	NM_000899
LARS2	NM_015340
LHFPL5	NM_182548
LOXHD1	NM_144612
LRTOMT	NM_001145308
MARVELD2	NM_001038603
MCM2	NM_004526
MET	NM_001127500
MIR96	NR_029512
MITF	NM_000248
MSRB3	NM_198080
MT-RNR1	NC_012920[3]
MT-TS1	NC_012920[3]
MYH14	NM_024729
MYH9	NM_002473
MYO15A	NM_016239
MYO3A	NM_017433
MYO6	NM_004999
MYO7A	NM_000260

Hereditary Hearing Loss Panel	
Gene	**RefSeq Transcript[2]**
NARS2	NM_024678
NF2	NM_000268
NLRP3	NM_004895
OPA1	NM_015560
OSBPL2	NM_144498
OTOA	NM_144672
OTOF	NM_194248
OTOG	NM_001277269
OTOGL	NM_173591
P2RX2	NM_174873
PAX3	NM_181457
PCDH15	NM_033056
PDZD7	NM_001195263
PEX1	NM_000466
PEX6	NM_000287
PHYH	NM_06214
PJVK (DFNB59)	NM_001042702
PNPT1	NM_033109
POLR1C	NM_203290
POLR1D	NM_015972
POU3F4	NM_000307
POU4F3	NM_002700
PRPS1	NM_002764
PTPN11	NM_002834
PTPRQ	ENST000266688
RDX	NM_002906
RIPOR2 (FAM65B)	NM_014722
S1PR2	NM_004230
SERPINB6	NM_004568
SIX1	NM_005982
SLC17A8	NM_139319
SLC22A4	NM_003059
SLC26A4	NM_000441
SLC26A5	NM_198999
SLC52A2	NM_024531
SLC52A3	NM_033409
SLITRK6	NM_032229
SMPX	NM_014332
SNAI2	NM_003068

Hereditary Hearing Loss Panel	
Gene	**RefSeq Transcript[2]**
SOX10	NM_006941
STRC	NM_153700
SYNE4	NM_001039876
TBC1D24	NM_001199107
TCOF1	NM_001135243
TECTA	NM_005422
TIMM8A	NM_004085
TJP2	NM_004817
TMC1	NM_138691
TMEM132E	NM_001304438
TMIE	NM_147196
TMPRSS3	NM_024022
TNC	NM_002160
TPRN	NM_001128228
TRIOBP	NM_001039141
TWNK (C10ORF2)	NM_021830
USH1C	NM_005709
USH1G	NM_173477
USH2A	NM_206933
WBP2	NM_012478
WFS1	NM_006005
WHRN	NM_015404

[1] Due to high homology with pseudogenes, or other regions in the genome, variant detection sensitivity may be decreased in the following genes: *ACTG1*, *ADCY1*, *CATSPER2*, *DNMT1*, *DSPP*, *ESPN*, *FOXC1*, *FOXI1*, *KARS*, *OTOA*, *PIEZO2*, *POU3F4*, *PTPN11*, *PTPRQ*, *SLC22A4*, *SLC52A2*, *SOX10*, *TIMM8A*, *TPRN*, and *TRIOBP*. There are regions in the *CATSPER2*, *DNMT1*, *DSPP*, *HOMER2*, *OTOA*, *PIEZO2*, *PTPRQ*, and *STRC* genes that cannot be effectively analyzed for the presence of large deletions and/or duplications as a result of technical limitations of the assay, including regions of homology, high GC-rich content, and repetitive sequences.

[2] Reference transcript numbers may have been updated due to database re-versioning. Refer to the patient report for the most updated gene transcript information. Always refer to final reports for the gene transcript information that is pertinent to each patient at the time of testing.

[3] Only m.1494C>T and m.1555A>G in MT-RNR1 and m.7445A>G in MT-TS1 are covered.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.