MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for AudioloGene Hearing Loss Panel

The following applies to AHLP / AudioloGene Hearing Loss Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from January 2023 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, or genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ABHD12	NM_001042472.3	-	-
ACTG1	NM_001614.5	-	-
ADCY1	NM_021116.4	-	CNV analysis in exon 20 will not be performed
ADGRV1 (GPR98)	NM_032119.4	-	-
AIFM1	NM_004208.4	-	-
ALMS1	ENST00000264448.6	-	-
ARSG	NM_014960.5	-	-
ATP2B2	NM_001683.5	-	-
ATP2B2	NM_001001331.4	-	-
ATP6V1B1	NM_001692.4	-	-
ATP6V1B2	NM_001693.4	-	-
BCS1L	NM_004328.5	-	-
BSND	NM_057176.3	-	-
BTD	NM_000060.4	-	-
CABP2	NM_016366.3	-	-
CACNA1D	NM_000720.4	-	-
CATSPER[a]	NM_172095.4	MLPA is performed for exons 1, 2, 4, 7, and 12	Analysis for sequence variants will not be performed
CCDC50	NM_178335.3	-	-
CD164	NM_006016.6	-	-
CDC14A	NM_033312.2	-	-
CDH23	NM_022124.6	-	CNV analysis in exon 12 will not be performed
CEACAM16	NM_001039213.4	-	-
CEP250	NM_007186.6	-	-
CEP78	NM_001098802.3	-	CNV analysis in exon 10 will not be performed
CHD7	NM_017780.4	chr8:61757794 (c.5051-15T>A) chr8:61763034 (c.5405-18C>A) chr8:61763035 (c.5405-17G>A) chr8:61763039 (c.5405-13G>A)	-
CIB2	NM_006383.4	-	-
CISD2	NM_001008388.5	-	CNV analysis in exon 3 will not be performed
CLDN14	NM_144492.3	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
CLIC5	NM_001114086.2	-	-
CLIC5	NM_016929.5	-	-
CLPP	NM_006012.4	-	-
CLRN1	NM_174878.3	chr3:150660197 (c.254-649T>G)	-
COCH	NM_004086.3	-	-
COL11A1	NM_001854.4	-	-
COL11A2	NM_080680.3	-	-
COL2A1	NM_001844.5	-	-
COL4A3	NM_000091.5	-	-
COL4A4	NM_000092.5	-	-
COL4A5	NM_000495.5	-	-
COL4A5	NM_033380.3	-	-
COL4A6	NM_001847.4	-	CNV analysis in exon 1 will not be performed
COL9A1	NM_001851.5	-	-
COL9A2	NM_001852.4	-	-
COL9A3	NM_001853.4	-	-
CRYL1[b]	NM_015974.3	This gene is reported only in the context of GJB2	Analysis for sequence variants will not be performed
CRYM	NM_001888.5	-	-
DCDC2	NM_016356.5	-	-
DIABLO	NM_019887.6	-	-
DIAPH1	NM_005219.5	-	-
DIAPH3	NM_001042517.2	-	CNV analysis in exon 18 will not be performed
DMXL2	NM_001174116.2	-	-
DNMT1	NM_001130823.3	-	CNV analysis in exon 5 will not be performed
DSPP	NM_014208.3	-	Analyses for sequence variants and CNV in exon 5 will not be performed
EDN3	NM_207034.3	-	-
EDNRB	NM_000115.5	-	-
ELMOD3	NM_001135022.2	-	-
EPS8	NM_004447.6	-	-
EPS8L2	NM_022772.4	-	-
ESPN	NM_031475.3	-	CNV analysis in exon 7 will not be performed
ESRRB	NM_004452.3	-	-
EYA1	NM_000503.6	-	-
EYA4	NM_004100.5	-	-
FDXR	NM_024417.5	-	-
FGF3	NM_005247.4	-	-
FGFR2	NM_000141.4	-	-
FGFR3	NM_000142.4	-	-
FITM2	NM_001080472.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
FLNA	NM_001456.3	-	-
FOXC1	NM_001453.3	-	-
FOXI1	NM_012188.5	-	-
GATA3	NM_001002295.2	-	-
GIPC3	NM_133261.3	-	-
GJB2 (DFNB1) with GJB6 and CRYL1[b]	NM_004004.6	chr13:20763744 (c.-22-2A>C) chr13:20766920 (c.-23+2T>A) chr13:20766921 (c.-23+1G>A) chr13:20766922 (c.-23G>T)	-
GJB6[b]	NM_006783.4	-	Analysis for sequence variants will not be performed
GPSM2	NM_013296.5	-	-
GREB1L	NM_001142966.2	-	CNV analysis will not be performed
GRHL2	NM_024915.4	-	-
GRXCR1	NM_001080476.2	-	-
GRXCR2	NM_001080516.1	-	-
GSDME	NM_004403.3	chr7:24746008_24746010 (c.991-15_991-13del)	-
HARS2	NM_012208.4	-	-
HGF	NM_000601.6	-	-
HOMER2	NM_004839.4	-	-
HOXA2	NM_006735.4	-	-
HSD17B4	NM_000414.4	chr5:118837725 (c.1210-11C>G)	-
ILDR1	NM_001199799.2	-	-
KARS1	NM_001130089.1	-	-
KARS1	NM_005548.2	-	-
KCNE1	NM_000219.6	-	-
KCNJ10	NM_002241.5	-	-
KCNQ1	NM_000218.3	-	-
KCNQ4	NM_004700.4	-	-
KITLG	NM_000899.5	-	-
LARS2	NM_015340.4	-	-
LHFPL5	NM_182548.4	-	-
LMX1A	NM_177398.4	-	-
LOXHD1	NM_144612.6	-	-
LOXHD1	NM_001145472.3	-	CNV analysis in exon 24 will not be performed
LRP2	NM_004525.3	-	-
LRTOMT	NM_001145308.4	-	-
MAN2B1	NM_000528.4	-	-
MANBA	NM_005908.4	-	-
MARVELD2	NM_001038603.3	-	CNV analysis in exon 6 will not be performed
MCM2	NM_004526.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
MET	NM_001127500.3	-	-
MIR96	NR_029512.1	-	CNV analysis will not be performed
MITF	NM_000248.3	-	-
MPZL2	NM_005797.4	-	-
MSRB3	NM_198080.4	-	CNV analysis in exon 4 will not be performed
MSRB3	NM_001031679.3	-	CNV analysis in exon 5 will not be performed
MT-RNR1	NC_012920.1	ddPCR for the detection of m.1494C>T, m.1555A>G to 5% heteroplasmy	No additional variants are detected
MT-TS1	NC_012920.1	ddPCR for the detection m.7445A>G of to 5% heteroplasmy	No additional variants are detected
MYH14	NM_024729.3	-	-
MYH9	NM_002473.5	-	-
MYO15A	NM_016239.4	-	-
MYO3A	NM_017433.5	-	-
MYO6	NM_004999.4	-	-
MYO7A	NM_000260.4	-	-
NARS2	NM_024678.6	-	CNV analysis in exon 9 will not be performed
NDRG1	NM_006096.4	-	-
NF2	NM_000268.3	-	-
NLRP3	NM_004895.4	-	-
OPA1	NM_015560.2	-	-
OPA1	NM_130837.2	-	-
OSBPL2	NM_144498.3	-	-
OTOA	NM_144672.4	MLPA is performed for exons 2, 5, 7, 11, 16, and 17	Analysis for sequence variants in exons 23–28 will not be performed CNV analysis in exons 21–29 will not be performed
OTOF	NM_194248.3	-	-
OTOF	NM_001287489.2	-	-
OTOG	NM_001277269.2	-	-
OTOGL	NM_173591.3	-	CNV analysis in exon 31 will not be performed
P2RX2	NM_174873.3	-	-
PAX3	NM_181457.4	-	-
PCDH15	NM_033056.4	-	-
PDZD7	NM_001195263.2	-	-
PEX1	NM_000466.3	-	-
PEX10	NM_153818.1	-	-
PEX11B	NM_003846.3	-	-
PEX12	NM_000286.3	-	-
PEX13	NM_002618.4	-	-
PEX14	NM_004565.3	-	-
PEX16	NM_004813.3	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
PEX19	NM_002857.3	-	-
PEX2	NM_000318.3	-	-
PEX26	NM_017929.6	-	-
PEX3	NM_003630.3	-	-
PEX5	NM_001131025.1	-	-
PEX6	NM_000287.4	-	-
PEX7	NM_000288.4	-	-
PHYH	NM_006214.4	-	-
PJVK	NM_001042702.4	-	-
PLS1	NM_002670.3	-	-
PNPT1	NM_033109.5	-	-
POLR1B	NM_019014.6	-	-
POLR1C	NM_203290.4	-	-
POLR1D	NM_015972.4	-	-
POU3F4	NM_000307.5	-	-
POU4F3	NM_002700.3	-	-
PRPS1	NM_002764.4	-	-
PTPN11	NM_002834.4	-	-
PTPRQ	ENST00000266688.5	-	Analysis for sequence variants in exons 4–11 will not be performed CNV analysis in exons 1 and 4–50 will not be performed
RAI1	NM_030665.4	-	-
RDX	NM_002906.3	-	CNV analysis in exon 2 will not be performed
RIPOR2	NM_014722.5	-	CNV analysis in exon 6 will not be performed
RMND1	NM_017909.4	-	-
S1PR2	NM_004230.4	-	-
SALL1	NM_002968.3	-	-
SERAC1	NM_032861.4	-	CNV analysis in exon 3 will not be performed
SERPINB6	NM_004568.5	-	-
SIX1	NM_005982.4	-	-
SLC12A2	NM_001046.3	-	-
SLC17A8	NM_139319.3	-	-
SLC19A2	NM_006996.3	-	-
SLC22A4	NM_003059.3	-	-
SLC26A4	NM_000441.2	chr7:107301201 (c.-103T>C) chr7:107334836 (c.1264-12T>A)	CNV analysis in exon 18 will not be performed
SLC26A5	NM_198999.3	-	-
SLC29A3	NM_018344.6	-	CNV analysis in exon 1 will not be performed
SLC4A11	NM_032034.3	-	-
SLC52A2	NM_024531.5	-	-
SLC52A3	NM_033409.4	-	-

Targeted Genes and Methodology Details for AudioloGene Hearing Loss Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
SLITRK6	NM_032229.3	-	-
SMPX	NM_014332.3	-	-
SNAI2	NM_003068.5	-	-
SOX10	NM_006941.4	-	-
SPATA5	NM_145207.3	-	-
STRC[a]	NM_153700.2	MLPA is performed for exons 19, 23, 24 and 25	CNV analysis in exons 1–18 and 29 will not be performed CNV in exons 20–22 and 26–28 may not be detected or reported
SUCLA2	NM_003850.2	-	-
SYNE4	NM_001039876.3	-	-
TBC1D24	NM_001199107.2	-	-
TCOF1	NM_001135243.1	-	-
TECTA	NM_005422.2	-	-
TFAP2A	NM_001372066.1	-	-
TIMM8A	NM_004085.4	-	-
TJP2	NM_004817.4	-	-
TMC1	NM_138691.2	-	-
TMEM132E	NM_001304438.2	-	-
TMIE	NM_147196.2	-	-
TMPRSS3	NM_024022.3	-	-
TNC	NM_002160.4	-	-
TPRN	NM_001128228.3	-	-
TRIOBP	NM_001039141.3	-	Analyses for sequence variants and CNV will not be performed in a portion of exon 7 (chr22:38119652-38120851) CNV in exon 7 may not be detected or reported
TUBB4B	NM_006088.6	-	-
TWNK	NM_021830.5	-	-
USH1C	NM_005709.4	-	-
USH1C	NM_153676.4	-	-
USH1G	NM_173477.5	-	-
USH2A	NM_206933.3	chr1:216592035 (c.486-14G>A) chr1:216064540 (c.7595-2144A>G)	
WBP2	NM_012478.4	-	-
WFS1	NM_006005.3	-	-
WHRN	NM_015404.4	-	-

[a] Sequence variants in CATSPER2 are not detected. Deletions in CATSPER2 are only reported if a deletion in STRC is also identified. Additionally, duplications in the STRC / CATSPER2 region may not be reported as exact breakpoints cannot be determined. To date, duplications in this region have not been associated with hearing loss and/or male infertility.

[b] Large deletions adjacent to GJB2 have been associated with GJB2-mediated autosomal recessive hearing loss. In order to identify these pathogenic deletions, copy number changes in GJB6 and CRYL1 are assessed. As these deletions disrupt GJB2 function, these findings are only reported in the context of GJB2, and sequence variants in CRYL1 are not detected or reported.