



The following applies to Hearing Loss Custom Gene 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 numbers 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
<i>ABHD12</i>	NM_001042472.3	-	-
<i>ACTG1</i>	NM_001614.5	-	-
<i>ADCY1</i>	NM_021116.4	-	CNV analysis in exon 20 will not be performed
<i>ADGRV1</i> ( <i>GPR98</i> )	NM_032119.4	-	-
<i>AIFM1</i>	NM_004208.4	-	-
<i>ALMS1</i>	ENST00000264448.6	-	-
<i>ARSG</i>	NM_014960.5	-	-
<i>ATP2B2</i>	NM_001683.5	-	-
<i>ATP2B2</i>	NM_001001331.4	-	-
<i>ATP6V1B1</i>	NM_001692.4	-	-
<i>ATP6V1B2</i>	NM_001693.4	-	-
<i>BCS1L</i>	NM_004328.5	-	-
<i>BSND</i>	NM_057176.3	-	-
<i>BTD</i>	NM_000060.4	-	-
<i>CABP2</i>	NM_016366.3	-	-
<i>CACNA1D</i>	NM_000720.4	-	-
<i>CATSPER<sup>a</sup></i>	NM_172095.4	MLPA is performed for exons 1, 2, 4, 7, and 12	Analysis for sequence variants will not be performed
<i>CCDC50</i>	NM_178335.3	-	-
<i>CD164</i>	NM_006016.6	-	-
<i>CDC14A</i>	NM_033312.2	-	-
<i>CDH23</i>	NM_022124.6	-	CNV analysis in exon 12 will not be performed
<i>CEACAM16</i>	NM_001039213.4	-	-
<i>CEP250</i>	NM_007186.6	-	-
<i>CEP78</i>	NM_001098802.3	-	CNV analysis in exon 10 will not be performed
<i>CHD7</i>	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)	-
<i>CIB2</i>	NM_006383.4	-	-
<i>CISD2</i>	NM_001008388.5	-	CNV analysis in exon 3 will not be performed

## Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CLDN14</i>	NM_144492.3	-	-
<i>CLIC5</i>	NM_001114086.2	-	-
<i>CLIC5</i>	NM_016929.5	-	-
<i>CLPP</i>	NM_006012.4	-	-
<i>CLRN1</i>	NM_174878.3	chr3:150660197 (c.254-649T>G)	-
<i>COCH</i>	NM_004086.3	-	-
<i>COL11A1</i>	NM_001854.4	-	-
<i>COL11A2</i>	NM_080680.3	-	-
<i>COL2A1</i>	NM_001844.5	-	-
<i>COL4A3</i>	NM_000091.5	-	-
<i>COL4A4</i>	NM_000092.5	-	-
<i>COL4A5</i>	NM_000495.5	-	-
<i>COL4A5</i>	NM_033380.3	-	-
<i>COL4A6</i>	NM_001847.4	-	CNV analysis in exon 1 will not be performed
<i>COL9A1</i>	NM_001851.5	-	-
<i>COL9A2</i>	NM_001852.4	-	-
<i>COL9A3</i>	NM_001853.4	-	-
<i>CRYL<sup>p</sup></i>	NM_015974.3	This gene is reported only in the context of GJB2	Analysis for sequence variants will not be performed
<i>CRYM</i>	NM_001888.5	-	-
<i>DCDC2</i>	NM_016356.5	-	-
<i>DIABLO</i>	NM_019887.6	-	-
<i>DIAPH1</i>	NM_005219.5	-	-
<i>DIAPH3</i>	NM_001042517.2	-	CNV analysis in exon 18 will not be performed
<i>DMXL2</i>	NM_001174116.2	-	-
<i>DNMT1</i>	NM_001130823.3	-	CNV analysis in exon 5 will not be performed
<i>DSPP</i>	NM_014208.3	-	Analyses for sequence variants and CNV in exon 5 will not be performed
<i>EDN3</i>	NM_207034.3	-	-
<i>EDNRB</i>	NM_000115.5	-	-
<i>ELMOD3</i>	NM_001135022.2	-	-
<i>EPS8</i>	NM_004447.6	-	-
<i>EPS8L2</i>	NM_022772.4	-	-
<i>ESPN</i>	NM_031475.3	-	CNV analysis in exon 7 will not be performed
<i>ESRRB</i>	NM_004452.3	-	-
<i>EYA1</i>	NM_000503.6	-	-
<i>EYA4</i>	NM_004100.5	-	-
<i>FDXR</i>	NM_024417.5	-	-
<i>FGF3</i>	NM_005247.4	-	-
<i>FGFR2</i>	NM_000141.4	-	-
<i>FGFR3</i>	NM_000142.4	-	-
<i>FITM2</i>	NM_001080472.4	-	-

# Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>FLNA</i>	NM_001456.3	-	-
<i>FOXC1</i>	NM_001453.3	-	-
<i>FOXI1</i>	NM_012188.5	-	-
<i>GATA3</i>	NM_001002295.2	-	-
<i>GIPC3</i>	NM_133261.3	-	-
<i>GJB2</i> ( <i>DFNB1</i> ) with <i>GJB6</i> and <i>CRYL1<sup>b</sup></i>	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)	-
<i>GJB6<sup>b</sup></i>	NM_006783.4	-	Analysis for sequence variants will not be performed
<i>GPSM2</i>	NM_013296.5	-	-
<i>GREB1L</i>	NM_001142966.2	-	CNV analysis will not be performed
<i>GRHL2</i>	NM_024915.4	-	-
<i>GRXCR1</i>	NM_001080476.2	-	-
<i>GRXCR2</i>	NM_001080516.1	-	-
<i>GSDME</i>	NM_004403.3	chr7:24746008_24746010 (c.991-15_991-13del)	-
<i>HARS2</i>	NM_012208.4	-	-
<i>HGF</i>	NM_000601.6	-	-
<i>HOMER2</i>	NM_004839.4	-	-
<i>HOXA2</i>	NM_006735.4	-	-
<i>HSD17B4</i>	NM_000414.4	chr5:118837725 (c.1210-11C>G)	-
<i>ILDR1</i>	NM_001199799.2	-	-
<i>KARS1</i>	NM_001130089.1	-	-
<i>KARS1</i>	NM_005548.2	-	-
<i>KCNE1</i>	NM_000219.6	-	-
<i>KCNJ10</i>	NM_002241.5	-	-
<i>KCNQ1</i>	NM_000218.3	-	-
<i>KCNQ4</i>	NM_004700.4	-	-
<i>KITLG</i>	NM_000899.5	-	-
<i>LARS2</i>	NM_015340.4	-	-
<i>LHFPL5</i>	NM_182548.4	-	-
<i>LMX1A</i>	NM_177398.4	-	-
<i>LOXHD1</i>	NM_144612.6	-	-
<i>LOXHD1</i>	NM_001145472.3	-	CNV analysis in exon 24 will not be performed
<i>LRP2</i>	NM_004525.3	-	-
<i>LRTOMT</i>	NM_001145308.4	-	-
<i>MAN2B1</i>	NM_000528.4	-	-
<i>MANBA</i>	NM_005908.4	-	-
<i>MARVELD2</i>	NM_001038603.3	-	CNV analysis in exon 6 will not be performed
<i>MCM2</i>	NM_004526.4	-	-
<i>MET</i>	NM_001127500.3	-	-
<i>MIR96</i>	NR_029512.1	-	CNV analysis will not be performed

# Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>MITF</i>	NM_000248.3	-	-
<i>MPZL2</i>	NM_005797.4	-	-
<i>MSRB3</i>	NM_198080.4	-	CNV analysis in exon 4 will not be performed
<i>MSRB3</i>	NM_001031679.3	-	CNV analysis in exon 5 will not be performed
<i>MT-RNR1</i>	NC_012920.1	ddPCR for the detection of m.1494C>T, m.1555A>G to 5% heteroplasmy	No additional variants are detected
<i>MT-TS1</i>	NC_012920.1	ddPCR for the detection m.7445A>G of to 5% heteroplasmy	No additional variants are detected
<i>MYH14</i>	NM_024729.3	-	-
<i>MYH9</i>	NM_002473.5	-	-
<i>MYO15A</i>	NM_016239.4	-	-
<i>MYO3A</i>	NM_017433.5	-	-
<i>MYO6</i>	NM_004999.4	-	-
<i>MYO7A</i>	NM_000260.4	-	-
<i>NARS2</i>	NM_024678.6	-	CNV analysis in exon 9 will not be performed
<i>NDRG1</i>	NM_006096.4	-	-
<i>NF2</i>	NM_000268.3	-	-
<i>NLRP3</i>	NM_004895.4	-	-
<i>OPA1</i>	NM_015560.2	-	-
<i>OPA1</i>	NM_130837.2	-	-
<i>OSBPL2</i>	NM_144498.3	-	-
<i>OTOA</i>	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
<i>OTOF</i>	NM_194248.3	-	-
<i>OTOF</i>	NM_001287489.2	-	-
<i>OTOG</i>	NM_001277269.2	-	-
<i>OTOGL</i>	NM_173591.3	-	CNV analysis in exon 31 will not be performed
<i>P2RX2</i>	NM_174873.3	-	-
<i>PAX3</i>	NM_181457.4	-	-
<i>PCDH15</i>	NM_033056.4	-	-
<i>PDZD7</i>	NM_001195263.2	-	-
<i>PEX1</i>	NM_000466.3	-	-
<i>PEX10</i>	NM_153818.1	-	-
<i>PEX11B</i>	NM_003846.3	-	-
<i>PEX12</i>	NM_000286.3	-	-
<i>PEX13</i>	NM_002618.4	-	-
<i>PEX14</i>	NM_004565.3	-	-
<i>PEX16</i>	NM_004813.3	-	-
<i>PEX19</i>	NM_002857.3	-	-
<i>PEX2</i>	NM_000318.3	-	-

## Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	-	-
SLITRK6	NM_032229.3	-	-
SMPX	NM_014332.3	-	-

# Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>SNAI2</i>	NM_003068.5	-	-
<i>SOX10</i>	NM_006941.4	-	-
<i>SPATA5</i>	NM_145207.3	-	-
<i>STRC</i> <sup>a</sup>	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
<i>SUCLA2</i>	NM_003850.2	-	-
<i>SYNE4</i>	NM_001039876.3	-	-
<i>TBC1D24</i>	NM_001199107.2	-	-
<i>TCOF1</i>	NM_001135243.1	-	-
<i>TECTA</i>	NM_005422.2	-	-
<i>TFAP2A</i>	NM_001372066.1	-	-
<i>TIMM8A</i>	NM_004085.4	-	-
<i>TJP2</i>	NM_004817.4	-	-
<i>TMC1</i>	NM_138691.2	-	-
<i>TMEM132E</i>	NM_001304438.2	-	-
<i>TMIE</i>	NM_147196.2	-	-
<i>TMPRSS3</i>	NM_024022.3	-	-
<i>TNC</i>	NM_002160.4	-	-
<i>TPRN</i>	NM_001128228.3	-	-
<i>TRIOBP</i>	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
<i>TUBB4B</i>	NM_006088.6	-	-
<i>TWINK</i>	NM_021830.5	-	-
<i>USH1C</i>	NM_005709.4	-	-
<i>USH1C</i>	NM_153676.4	-	-
<i>USH1G</i>	NM_173477.5	-	-
<i>USH2A</i>	NM_206933.3	chr1:216592035 (c.486-14G>A) chr1:216064540 (c.7595-2144A>G)	
<i>WBP2</i>	NM_012478.4	-	-
<i>WFS1</i>	NM_006005.3	-	-
<i>WHRN</i>	NM_015404.4	-	-

<sup>a</sup> 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. When *STRC* is ordered via Custom Gene Panel, Hereditary (CGPH), the lab will add *CATSPER2* to the order if the client has not already included this gene.

<sup>b</sup> 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 the *CRYL1* and *GJB6* genes are not detected or reported. When *GJB2* is ordered via Custom Gene Panel, Hereditary (CGPH), the lab will add *GJB6* and *CRYL1* to the order if the client has not already included these genes to appropriately assess the DFNB1 locus.

# Targeted Genes and Methodology Details for Hearing Loss Custom Gene Panel (continued)

## Available AudioloGene Hearing Loss Panels

Test ID	Test Name	Genes
AHLP	AudioloGene Hearing Loss Panel	<p> <i>ABHD12, ACTG1, ADCY1, ADGRV1 (GPR98), AIFM1, ALMS1, ARSG, ATP2B2, ATP6V1B1, ATP6V1B2, BCS1L, BSND, BTD, CABP2, CACNA1D, CATSPER2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYL1, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FDXR, FGF3, FGFR2, FGFR3, FITM2, FLNA, FOXC1, FOXI1, GATA3, GIPC3, GJB2 (DFNB1; with CRYL1 and GJB6), GJB6, GPSM2, GREB1L, GRHL2, GSDME, GRXCR1, GRXCR2, HARS2, HGF, HOMER2, HOXA2, HSD17B4, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LMX1A, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MCM2, MET, MIR96, MTF, MPZL2, MSRB3, MT-RNR1, MT-TS1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDRG1, NF2, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PJVK, PLS1, PNPT1, POLR1B, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPN11, PTPRQ, RAI1, RDX, RIPOR2, RMND1, S1PR2, SALL1, SERAC1, SERPINB6, SIX1, SLC12A2, SLC17A8, SLC19A2, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, SPATA5, STRC, SUCLA2, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TUBB4B, TWNK, USH1C, USH1G, USH2A, WBP2, WFS1, and WHRN</i> </p>