



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 number variants 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^a</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
<i>CLDN14</i>	NM_144492.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<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>CRYL1^b</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	-	-
<i>FLNA</i>	NM_001456.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<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^b</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^b</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
<i>MITF</i>	NM_000248.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<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	-	-
<i>PEX26</i>	NM_017929.6	-	-
<i>PEX3</i>	NM_003630.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>PEX5</i>	NM_001131025.1	-	-
<i>PEX6</i>	NM_000287.4	-	-
<i>PEX7</i>	NM_000288.4	-	-
<i>PHYH</i>	NM_006214.4	-	-
<i>PJVK</i>	NM_001042702.4	-	-
<i>PLS1</i>	NM_002670.3	-	-
<i>PNPT1</i>	NM_033109.5	-	-
<i>POLR1B</i>	NM_019014.6	-	-
<i>POLR1C</i>	NM_203290.4	-	-
<i>POLR1D</i>	NM_015972.4	-	-
<i>POU3F4</i>	NM_000307.5	-	-
<i>POU4F3</i>	NM_002700.3	-	-
<i>PRPS1</i>	NM_002764.4	-	-
<i>PTPN11</i>	NM_002834.4	-	-
<i>PTPRQ</i>	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
<i>RAI1</i>	NM_030665.4	-	-
<i>RDX</i>	NM_002906.3	-	CNV analysis in exon 2 will not be performed
<i>RIPOR2</i>	NM_014722.5	-	CNV analysis in exon 6 will not be performed
<i>RMND1</i>	NM_017909.4	-	-
<i>S1PR2</i>	NM_004230.4	-	-
<i>SALL1</i>	NM_002968.3	-	-
<i>SERAC1</i>	NM_032861.4	-	CNV analysis in exon 3 will not be performed
<i>SERPINB6</i>	NM_004568.5	-	-
<i>SIX1</i>	NM_005982.4	-	-
<i>SLC12A2</i>	NM_001046.3	-	-
<i>SLC17A8</i>	NM_139319.3	-	-
<i>SLC19A2</i>	NM_006996.3	-	-
<i>SLC22A4</i>	NM_003059.3	-	-
<i>SLC26A4</i>	NM_000441.2	chr7:107301201 (c.-103T>C) chr7:107334836 (c.1264-12T>A)	CNV analysis in exon 18 will not be performed
<i>SLC26A5</i>	NM_198999.3	-	-
<i>SLC29A3</i>	NM_018344.6	-	CNV analysis in exon 1 will not be performed
<i>SLC4A11</i>	NM_032034.3	-	-
<i>SLC52A2</i>	NM_024531.5	-	-
<i>SLC52A3</i>	NM_033409.4	-	-
<i>SLITRK6</i>	NM_032229.3	-	-
<i>SMPX</i>	NM_014332.3	-	-
<i>SNAI2</i>	NM_003068.5	-	-
<i>SOX10</i>	NM_006941.4	-	-
<i>SPATA5</i>	NM_145207.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>STRC</i> ^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
<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>TWNK</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	-	-

^aSequence 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.

^bLarge 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, MITF, 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>