HHLP

AudioloGene Hereditary Hearing Loss Panel

Patient ID	Patient Name		Birth Date	Gender	Age
SA00756587	TESTINGHHLP, NORMAL		2010-10-10	F	10
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00756587	SA00756587	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		06 Nov 2020 00:00			

Result Summary

Negative

Result

MCR

No reportable variants were identified.

Interpretation

MCR

This result decreases the likelihood but does not rule out the involvement of the genes evaluated in this panel.

Individuals may have a pathogenic variant in one of the interrogated genes that is not detectable by the methods utilized. Additionally, the clinical phenotype that is observed in this individual and/or family may be due to a pathogenic variant or variants in another gene not targeted by this test.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

A genetic consultation may be of benefit.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292

MAYO CLINIC LABORATORIES

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Specimen

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WB Whole Blood

Method

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Evaluation of 160 genes associated with hereditary hearing loss:

ABHD12, ACTG1, ADCY1, ADGRV1 (GPR98), AIFM1, ALMS1, ATP6V1B1, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CATSPER2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CEP78, CHD7, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, CRYM, DCDC2, DFNA5 (GSDME), DIABLO, DIAPH1, DIAPH3, DNMT1, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FGF3, FGFR2, FLNA, FOXC1, FOXI1, GATA3, GIPC3, GJB2, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, HARS2, HGF, HOMER2, HOXA2, HSD17B4, ILDR1, JAG1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MCM2, MET, MIR96, MITF, MSRB3, MT-RNR1, MT-TS1, MYH14, MYH9, MYO3A, MYO6, MYO7A, MYO15A, NARS2, NF2, NLRP3, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX6, PHYH, PJVK (DFNB59), PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPN11, PTPRQ, RDX, RIPOR2 (FAM65B), S1PR2, SERPINB6, SIX1, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TCOF1, TECTA, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TWNK (C10orf2), USH1C, USH1G, USH2A, WBP2, WFS1, and WHRN

Next generation sequencing (NGS) and/or Sanger sequencing was performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed. NGS and/or a PCR-based quantitative method was performed to test for the presence of deletions and duplications in the genes analyzed.

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, POU3F4, PTPN11, PTPRQ, SLC22A4, SLC52A2, SOX10, TIMM8A, TPRN, and TRIOBP. There are regions in the DNMT1, DSPP, HOMER2, OTOA, 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.

Disclaimer

1 MCR

Clinical Correlations
If testing was performed because of a clinically significant family history it is often useful to first test an affected family member. Detection of a reportable variant(s) in an affected family member would allow for more informative testing of at risk individuals.

To discuss the availability of further testing options or for assistance in the interpretation of these results, Mayo Clinic Laboratory genetic counselors can be contacted at 1–800–533–1710.

Technical Limitations
Next generation sequencing may not detect all types of genomic variants. In rare cases, false negative or false positive results may occur. The depth of coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria or for failed regions will be noted. Absence of the mitochondrial variants MT-RNR1 m.1494C>T, MT-RNR1 m.1555A>G, or MT-TS1 m.7445A>G on the report does not rule out the presence of these variants below the limits of detection of this assay (<5% heteroplasmy). Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively amplified for sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences. Confirmation of select reportable variants was performed by alternate methodologies based on internal laboratory criteria.

Additionally, low level mosaic variants may not be detected.

This test is not designed to differentiate between somatic and

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germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

Reclassification of Variants Policy
See www.mayocliniclabs.com
(TEST ID HHLP) for information regarding the laboratory's policy for reclassification of variants.

Variant Evaluation
Variant curation is performed using published ACMG-AMP recommendations as a guideline. Other gene-specific guidelines may also be considered. Variants classified as benign or likely benign are not reported.

Results from in silico evaluation tools may change over time and should be interpreted with caution and professional clinical judgment.

Released By MCR

Nicole J. Boczek, Ph.D.

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Laboratory Notes

1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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