MAYO CLINIC

1-800-533-1710

AudioloGene Hearing Loss Panel

AHLP

PATIENT NAME				ORDER NUMBER
TESTINGRNV, AHLP				**M514000381**
PATIENT ID SA00155875	DATE OF BIRTH 02/15/1970	AGE 52 Y	SEX Male	REQUESTED BY CLIENT CLIENT
COLLECTED 12/14/2022, 10:00 AM	RECEIVED 12/14/2022, 1:24 PM	REPORTED 12/14/2022, 3:59 PM		
The collected, received, and reported dates and times on the report are in the time zone of the performing location.				CLIENT ORDER NUMBER SA00155875
7028846 MCL RochesterCampus Rochester MN 55901				CLIENT MRN SA00155875

TEST DESCRIPTION

Evaluation of 200 genes associated with hereditary hearing loss

SPECIMEN

WB Whole Blood

RESULT SUMMARY

Pathogenic Variant Detected

RESULT

Gene (Transcript)	Variant	Zygosity	Classification
GJB2 NM_004004.5	c.35del p.Gly12Valfs*2 (p.G12Vfs*2) chr13(GRCh37):g.20763686	homozygous	PATHOGENIC

The following homozygous PATHOGENIC variant was detected:
GJB2 (NM_004004.5), chr13(GRCh37):g.20763686, c.35del, p.Gly12Valfs*2 (p.G12Vfs*2)

No additional reportable variants were detected within all other tested genes. See the Genes Analyzed section for a complete list of genes evaluated by this assay.

INTERPRETATION

GJB2 c.35del (p.G12Vfs*2), PATHOGENIC
The homozygous c.35del (p.G12Vfs*2) frameshift variant in the GJB2 gene (MIM:121011) is an established pathogenic variant (also described in the literature as c.35delG). The c.35del (p.G12Vfs*2) variant in the GJB2 gene has been associated with autosomal recessive nonsyndromic deafness (1). This result is supportive of a diagnosis of autosomal recessive nonsyndromic deafness for this individual. Clinical correlation is recommended.

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

Consultation with a genetics professional is recommended for interpretation of this result and to determine whether reproductive risk assessment and familial testing may be of benefit to this family. Genetic testing for family members is available by ordering FMTT / Familial Mutation, Targeted Testing for the specific variant(s) detected. Please contact the laboratory at 1-800-533-1710 or the online test catalog at www.mayocliniclabs.com for information about FMTT.

REFERENCES
1) Smith RJH, Jones MKN. Nonsyndromic Hearing Loss and Deafness, DFNB1. In: Adam MP, Ardinger HH, Pagon RA, et al., eds. GeneReviews. Seattle (WA): University of Washington, Seattle; September 28, 1998. (PMID 20301449)

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METHOD

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, as well as some other regions that have known pathogenic variants. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth >30X. Sensitivity is estimated at >99% for single nucleotide variants, >94% for delins up to 39 base pairs, >95% for deletions up to 75 base pairs and insertions up to 47 base pairs. NGS and/or a PCR-based quantitative method was performed to test for the presence of deletions and duplications in the genes analyzed. See the Genes Analyzed field for a list of genes tested.

There may be regions of genes that cannot be effectively evaluated 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. See www.mayocliniclabs.com AHLP for details regarding genes with regions not routinely covered.

GENES ANALYZED

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, DFNA5 (GSDME), DIABLO, DIAPH1, DIAPH3, DMXL2, DNMT, DSPP, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FDXR, FGF3, FGFR2, FGFR3, FITM2, FLNA, FOXC1, FOXI1, GATA3, GIPC3, GJB2, GJB6, GPSM2, GREB1L, GRHL2, GRXCR1, GRXCR2, HARS2, HGF, HOMER2, HOXA2, HSD17B4, ILDR1, KARS (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 (DFNB59), PLS1, PNPT1, POLR1B, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPN11, PTPRQ, RAI1, RDX, RIPOR2 (FAM65B), 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 (C10orf2), USH1C, USH1G, USH2A, WBP2, WFS1 and WHRN

DISCLAIMER

Clinical Correlations
An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

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

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

Genes may be added or removed based on updated clinical relevance. Please refer to the Targeted Genes and Methodology Details for the AudioloGene Hearing Loss Panel, Varies in the Special Instructions section of the Test Catalog for the most up to date list of genes included in this test.

Reclassification of Variants Policy
See www.mayocliniclabs.com (TEST ID AHLP) 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.

TEST CLASSIFICATION
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.

RELEASED BY

Nicole J. Boczek, Ph.D.

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Rochester MN 55901

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Code : MCR Laboratory : Mayo Clinic Laboratories - Rochester Main Campus Address : 200 FIRST STREET SW
Lab Director : WILLIAM G MORICE, II MD, PhD CLIA Certificate : 24D0404292 ROCHESTER MN 55905