Neurology

Hearing Loss

Hearing Loss Advanced Sequencing and CNV Evaluation

The American College of Medical Genetics and Genomics (ACMG) recommends that clinicians consider next-generation sequencing when evaluating the causes of hearing loss.[3]

Genetic testing from Athena Diagnostics can help uncover the etiology of hearing loss

When it comes to genetic hearing loss, over 6,000 causative variants have been identified in more than 100 genes, underscoring the substantial impact an all-encompassing, multi-gene panel may have in aiding timely diagnosis.[1]

Many hearing loss genes have overlapping presentations that are hard to differentiate, making test selection difficult.[2] Athena's next-generation sequencing (NGS) panel provides clinicians unique insights to help address the problem.

For complex interpretations, Athena Diagnostics MDs, PhDs, scientists, and genetic counselors assess the pathogenicity of variants of uncertain clinical significance, offering you a comprehensive, objective assessment that can help you help your patients.

Athena Diagnostics delivers a comprehensive testing solution that streamlines the path to diagnosis

The **Hearing Loss Advanced Sequencing and CNV Evaluation** is a comprehensive and up-to-date hearing loss panel. Our panel looks for sequence variations and copy number variants (CNVs) within the respective genes to provide physicians with clear insight to the etiology of the hearing loss.

Built on a foundation of decades of clinical testing, available literature, and the latest ACMG guidelines, the panel tests 183 genes and the DFNB1 locus, that are linked to hearing loss.

When used in conjunction with other clinical testing, the **Hearing Loss Advanced Sequencing and CNV Evaluation** can have benefits including:

- For newborns who fail hearing screen:
  - to explicitly determine the genetic etiology
  - to initiate an early intervention program and other medical management
- For congenitally deaf patients to understand the cause of their hearing loss and determining appropriate interventions/treatments
- For relatives of individuals with congenital deafness to determine carrier status and risk of recurrence
- For patients considering the use of aminoglycoside antibiotics with reason to suspect they may carry aminoglycoside-sensitive mutations
- For relatives of patients with an aminoglycoside-caused hearing loss condition

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Genes Tested in the Hearing Loss Advanced Sequencing and CNV Evaluation

A broad genetic testing panel can often shorten the diagnostic course and provide critical information for the physician and the patient. The **Hearing Loss Advanced Sequencing and CNV Evaluation** harnesses the power of sequencing and CNV analysis to provide a clear picture by testing for 183 genes and the DFNB1 locus[a]:

ABHD12
ACTB
ACTG1
ADCY1
ADGRV1 (GPR98)
AIFM1
ALMS1
ANKH
ATP2B2 (PMCA2)
ATP6V1B1
ATP6V1B2
BCS1L
BDP1
BSND
CABP2
CACNA1D
CCDC50
CD151
CD164
CDC14A
CDH23
CEACAM16
CEMIP
CHD7
CHSY1
CIB2
CISD2
CLDN14
CLIC5
CLPP
CLRN1
COCH
COL11A1
COL11A2
COL2A1
COL4A3
COL4A4
COL4A5
COL4A6
COL9A1
COL9A2
COL9A3
CRYM
DCDC2
DFNA5
DFNB1 locus[a]
DFNB59 (PJVK)
DIABLO
DIAPH1
DIAPH3
DLX5
DNMT1
DSPP
EDN3
EDNRA
EDNRB
ELMOD3
EPS8
EPS8L2
ERCC2
ERCC3
ESPN
ESRRB
EYA1
EYA4
FGF3
FGFR1
FGFR2
FGFR3
FOXI1
GATA3
GIPC3
GJB2
GJB3
GJB6[a]
GPSM2
GRHL2
GRXCR1
GRXCR2
HARS2
HGF
HOMER2
HOXB1
HSD17B4
ILDR1
KARS
KCNE1
KCNJ10
KCNQ1
KCNQ4
LARS2
LHFPL5
LOXHD1
LRTOMT
MANBA
MARVELD2
MCM2
MET
MIR96
MITF
MSRB3
MT-CO1[b]
MT-CO2[b]
MT-ND1[b]
MT-RNR1[b]
MT-TH[b]
MT-TI[b]
MT-TK[b]
MT-TL1[b]
MT-TQ[b]
MT-TS1[b]
MT-TS2[b]
MYH14
MYH9
MYO15A
MYO3A
MYO6
MYO7A
NARS2
NDP
NLRP3
NR2F1
OPA1
OSBPL2
OTOA
OTOF
OTOG
OTOGL
P2RX2
PAX3
PCDH15
PDZD7
PEX1
PEX6
PMP22
PNPT1
POLR1C
POLR1D
POU3F4
POU4F3
PRPS1
PTPRQ
RDX
RIPOR2
(FAM65B)
ROR1
S1PR2
SALL1[b]
SEMA3E
SERPINB6
SIX1
SIX5
SLC12A1
SLC17A8
SLC19A2
SLC22A4
SLC26A4
SLC26A5
SLC4A11
SLITRK6
SMPX
SNAI2
SOX10
STRC
SYNE4
TBC1D24
TBX1
TCOF1
TECTA
TFAP2A
TIMM8A
TJP2
TMC1
TMEM132E
TMIE
TMPRSS3
TNC
TPRN
TRIOBP
TSPEAR
USH1C
USH1G
USH2A
WFS1
WHRN

[a] CNV analysis only [b] Sequencing only

Test ordering information

Test Code	Test Name	CPT Codes	Turnaround Time
3029	Hearing Loss Advanced Sequencing and CNV Evaluation	81430(1), 81431(1)	28 – 35 days

Specimen Requirements: Whole blood, 8 mL (6mL minimum); Pediatric (0 – 3 years): 2 mL (1 mL minimum). Lavender Top Tube
Specimen Stability: Ambient: 10 days; Refrigerated: 10 days; Frozen: N/A
Special Transport Requirements: N/A

Additional hearing loss tests

Test Code	Test Name	CPT Codes	Turnaround Time
329	Connexin Related Deafness Evaluation Includes Connexin 30 DNA test and Connexin 26 DNA Sequencing Test	81252(1), 81254(1)	14 – 21 days
319	Connexin 30 (*GJB6*) DNA Test	81254(1)	14 – 21 days
321	Connexin 26 (*GJB2*) DNA Sequencing Test	81252(1)	14 – 21 days

The CPT codes provided are based on AMA guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party.

Please contact one of our genetic counselors regarding specific acceptance policies and specimen requirements for prenatal testing at 800-394-4493.

Athena Diagnostics offers a comprehensive genetic test menu for hearing loss. Customers in the U.S. and Canada please call toll free **1-800-394-4493** or visit us online at **AthenaDiagnostics.com/HearingLoss**.

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1 Shearer AE, Hildebrand MS, Smith RJH. Hereditary hearing loss and deafness overview. *GeneReviews*. 1999 (Updated July 2017). https://www.ncbi.nlm.nih.gov/books/NBK1434/. Accessed October 10, 2017.
2 Alford RL, Arnos KS, Fox M, et al. American College of Medical Genetics and Genomics guideline for the clinical evaluation and etiologic diagnosis of hearing loss. *Genet Med*. 2014:16(4):347-355. doi:10.1038/gim.2014.2.
3 Levenson D. New testing guidelines for hearing loss support next-generation sequencing: testing method may help determine genetic causes of hearing loss among patients whose phenotypes are not easily distinguished clinically. *Am J Med Genet*. 164A(7): vii–viii. doi:10.1002/ajmg.a.36643.

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