



Laboratory Service Manual

Lab Dept: Anatomic Pathology

Test Name: NONSYNDROMIC HEARING LOSS (OTOF-DFNB9)
MUTATION

General Information

Lab Order Codes: OTOF

Synonyms: Hearing Loss Genes; OTOF; DFNB9

CPT Codes: 83891 – Isolation or extraction of highly purified nucleic acid
83894 – Separation by gel electrophoresis
83898 x38– Amplification of patient nucleic acid, each nucleic acid sequence
83903 x8 – Mutation screening, by physical properties, single segment, each
83904 x48– Mutation identification by sequencing, single segment, each segment

Test Includes: Screening for OTOF is performed by DHPLC and sequencing. Oligonucleotide primers have been designed to amplify each exon. Amplified samples are run on the DHPLC; abnormal elution profiles are sequenced to determine the specific mutation. Exons carrying known SNPs are directly sequenced.

Logistics

Test Indications: This test is appropriate for any person with congenital hearing impairment and the diagnosis of auditory neuropathy, if the family history is consistent with autosomal recessive inheritance. It is also appropriate for persons with congenital hearing impairment and a family history consistent with autosomal recessive inheritance if GJB2 mutation screening is negative.

OTOF is a gene of 48 exons that encodes a transmembrane protein of 1230 amino acids called otoferlin (OMIM#: *603681). Otoferlin is expressed in multiple cell types within the cochlea. More than 24 mutations in OTOF have been reported and are associated with autosomal recessive nonsyndromic sensorineural hearing loss. Often, otoacoustic emissions (OAEs) are present while pure tone audiometry or auditory brain stem responses (ABRs) are grossly abnormal, features characteristic of auditory neuropathy.

Lab Testing Section: Anatomic Pathology - Sendouts



Laboratory Service Manual

Referred to: University of Iowa, Dr. Richard Smith

Phone Numbers:

Minneapolis: 612-813-6280

Saint Paul: 651-220-6550

Test Availability: Monday – Thursday, 24 hours

Turnaround Time: 3 months

Special Instructions: Include clinical data, specifically audiograms, CT/MRI reports, and pedigree information. Include name, e-mail, and telephone number of contact person (physician or genetic counselor). **Do NOT send on Friday or Saturday.**

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 10.0 mL (Minimum 8.0 mL) blood

Processed Volume: 8.0 – 10.0 mL whole blood

Collection: Routine venipuncture

Special Processing: Send lavender tubes labeled with a minimum of Name, Date of Birth, and Age. Include clinical data, specifically audiograms, CT/MRI reports, and pedigree information. Include name, e-mail, and telephone number of contact person (physician or genetic counselor). Mail overnight delivery at **Room Temperature** Monday - Thursday. **Do Not send on Friday or Saturday.** Samples may be refrigerated if delivery is delayed.

Patient Preparation: None

Sample Rejection: Specimens older than 48 hours; mislabeled or unlabeled specimens

Interpretive

Reference Range: No abnormal allele variants detected

Methodology: Amplified with oligonucleotide primers; restriction digest; agarose gel, if necessary

References: [Clinical Diagnostics Service, Molecular Otolaryngology Research Lab](#) information sheet from the University of Iowa.



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