



# Laboratory Service Manual

**Lab Dept:** Anatomic Pathology

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

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## **General Information**

**Lab Order Codes:** OTOF

**Synonyms:** Hearing Loss Genes; OTOF; DFNB9

**CPT Codes:** 83891 – Isolation or extraction of highly purified nucleic acid  
83894 x2 – Separation by gel electrophoresis  
83898 x38– Amplification of patient nucleic acid, each nucleic acid sequence  
83904 x38– Mutation identification by sequencing, single segment, each segment

**Test Includes:** Screening for OTOF; abnormal elution profiles are sequenced to determine the specific mutation. Exons carrying known SNPs are directly sequenced.

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## **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

**Referred to:** University of Iowa, Dr. Richard Smith

### **Phone Numbers:**

Minneapolis: 612-813-6280

Saint Paul: 651-220-6550



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|------------------------------|--|
| <b>Test Availability:</b>    | Monday – Thursday, 24 hours  |
| <b>Turnaround Time:</b>      | Approximately 3 months   |
| <b>Special Instructions:</b> | 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). <b>Do NOT send on Friday or Saturday.</b> |

Please include completed [University of Iowa Request form](#) with the patient or specimen to the laboratory. Informed consent is required.

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## ***Specimen***

|                             |  |
|-----------------------------|--|
| <b>Specimen Type:</b>       | Whole blood  |
| <b>Container:</b>           | Lavender top (EDTA) tube   |
| <b>Draw Volume:</b>         | 10 mL (Minimum: 8 mL) blood  |
| <b>Processed Volume:</b>    | 8 – 10 mL whole blood  |
| <b>Collection:</b>          | Routine venipuncture   |
| <b>Special Processing:</b>  | Send lavender tubes labeled with a minimum of Name, Date of Birth, and Sex. 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 <b>Room Temperature</b> Monday - Thursday. <b>Do Not send on Friday or Saturday.</b> Samples may be refrigerated if delivery is delayed. |
| <b>Patient Preparation:</b> | None   |
| <b>Sample Rejection:</b>    | Specimens older than 48 hours; mislabeled or unlabeled specimens   |

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## ***Interpretive***

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|-------------------------|--|
| <b>Reference Range:</b> | No abnormal allele variants detected   |
| <b>Methodology:</b>     | 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 SNP's are directly sequenced. |
| <b>References:</b>      | <a href="#">Clinical Diagnostics Service, Molecular Otolaryngology Research Lab</a> information sheet from the University of Iowa. June 2010<br>(319) 335-7997 Fax: (319) 353-5869   |
| <b>Updates:</b>         | 7/29/2010: CPT updates   |



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