



Laboratory Service Manual

Lab Dept: Anatomic Pathology

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

General Information

Lab Order Codes: OTOFK

Synonyms: Hearing Loss Genes; OTOF; DFNB9

CPT Codes: 83891 x2 – Isolation or extraction of highly purified nucleic acid
83894 x2 – Separation by gel electrophoresis
83898 x2 – Amplification of patient nucleic acid, each nucleic acid sequence
83904 x4 – Mutation identification by sequencing, single segment, each segment

Test Includes: Carrier testing for OTOF.

Logistics

Test Indications: Testing of a relative for a specific known mutation (carrier testing).

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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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. Please include completed University of Iowa Request form . Informed consent is required.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	10 mL (Minimum 8 mL) blood
Processed Volume:	8 – 10 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:	DHPLC and sequencing
References:	Clinical Diagnostics Service, Molecular Otolaryngology Research Lab information sheet from the University of Iowa. (319) 335-7997
Updates:	7/29/2010: CPT updates