



Laboratory Service Manual

Lab Dept: Anatomic Pathology

Test Name: AMINOGLYCOSIDE-INDUCED DEAFNESS
(MTRNR1) KNOWN MUTATION

General Information

Lab Order Codes: AMNDK

Synonyms: Hearing Loss Genes; MTRNR; C494T Mutation; A1555G Mutations

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

Test Includes: Carrier testing for MTRNR1.

Logistics

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

MTRNR1 encodes the mitochondrial 12S ribosomal RNA protein. Hearing loss as a result of aminoglycoside exposure (OMIM# 58000) has been reported to involve at least two mutations in this gene, C1494T and A1555G. Damage to the inner ear is caused by reactive oxygen species, which provide a common pathway not only for aminoglycoside toxicity, but also for cisplatin toxicity and noise-induced hearing loss. Both mutations are screened.

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

Test Availability: Monday – Thursday, 24 hours

Turnaround Time: 3 months



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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: DHPLC and sequencing

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