



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 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 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



Laboratory Service Manual

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 a completed University of Iowa [Request form](#) with the patient of specimen to the laboratory.

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. June 2010 (319) 335-7997 Fax: (319) 353-5869
Updates:	7/29/2010: CPT updates from University of Iowa.