



Laboratory Service Manual

Lab Dept: Anatomic Pathology

Test Name: MELAS SYNDROME (MTTL1) MUTATION

General Information

Lab Order Codes: MELS

Synonyms: Hearing Loss Genes; MTTL1-A3243G Mutation; Diabetes-deafness syndrome

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 x2– Mutation identification by sequencing, single segment, each segment

Test Includes: Following sample amplification with oligonucleotide primers within the MTTL1 gene, a restriction digest is completed; digestion products are resolved by agarose gel electrophoresis. Presence of the A3243G mutation creates an additional Apal restriction site.

Logistics

Test Indications: This test is appropriate for a patient suspected of having maternally inherited diabetes mellitus and sensorineural hearing loss.

MTTL1 encodes the mitochondrial transfer RNA-Leucine protein. The A3243G mutation has been found in several families segregating maternally inherited diabetes mellitus and sensorineural hearing loss. The A3243G mutation also is found in MELAS. In population studies of diabetics, the A3243G mutation is found in a small percent of patients.

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



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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.