



# Laboratory Service Manual

**Lab Dept:** Anatomic Pathology

**Test Name:** MELAS SYNDROME (MTTL1) KNOWN MUTATION

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## ***General Information***

**Lab Order Codes:** MELSK

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

**Test Includes:** Carrier testing for MTTL1.

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## ***Logistics***

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

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

**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.

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## ***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

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## ***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.