**Lab Dept:** Anatomic Pathology  
**Test Name:** MITOCHONDRIAL (MTDNA) DNA POINT MUTATIONS AND DELETION SCREEN

### General Information

**Lab Order Codes:** MDNA  
**Synonyms:** Advanced mtDNA Point Mutations and Deletions; mtDNA Deletion Syndromes Screen  
**CPT Codes:** 81401 x5 – Molecular Pathology procedure, Level 2  
281479 x10 – Molecular Pathology, Unlisted  
**Test Includes:** This test includes 36 common mitochondrial DNA mutations in 17 mitochondrial genes (Screens for 36 common point mutations and deletions in MELAS, MERRF, NARP, Leigh Syndrome, LHON, Cardiomyopathy, Deafness and/or Diabetes, Pearson Syndrome, and Kearns-Sayre Syndrome):  
MT-RNR1, MT-TV, MT-TL1, MT-ND1, MT-TI, MT-CO1/MT-TS1 precursor, MT-TK, MT-ATP6, MT-TG, MT-ND3, MT-ND4, MT-TH, MT-TS2/MT-TL2, MT-TL2, MT-ND5, MT-ND6, MT-TE

### Logistics

**Test Indications:** Multisystemic disorders involving neuromuscular, neurosensory, CNS, cardiac, renal, haptic, GI, Immune, endocrine, etc, systems. Family history of mitochondrial disorders. Matrilinear family members with known mitochondrial DNA mutations. Confirmation of mitochondrial disease. Asymptomatic carrier testing.  
In general, this test can detect mtDNA mutations in at least 85% of patients with primary mtDNA defect.  
**Lab Testing Section:** Anatomic Pathology - Sendouts  
**Referred to:** Baylor College of Medicine, Department of Molecular and Human Genetics (Test: 2010)  
**Phone Numbers:** MIN Lab: 612-813-6280  
STP Lab: 651-220-6550  
**Test Availability:** Daily 24 hours, set up once a week  
**Turnaround Time:** 28 days
**Special Instructions:** The provider must obtain a parent signature for the Baylor Consent Form and complete a Baylor Genetics Request Form. These forms must accompany the patient or specimen to the laboratory. Testing and subsequent release of results will be delayed if this information is not collected and submitted with the specimen.

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**Specimen**

**Specimen Type:** Whole blood

**Container:** Lavender top (EDTA) tube

**Draw Volume:** Children: 3-5 mL blood  
Infants: 3 mL blood  
Adults: 3-5 mL blood

**Processed Volume:** Same as Draw Volume

**Collection:**  
- Routine venipuncture  
- Send Baylor Genetics Request and Consent Forms with a parent/guardian signature and the clinical history completed by the provider with the specimen to the lab.

**Special Processing:** Lab Staff: Do Not freeze blood, refrigerate. Ship blood at room temperature and within 24 hours of collection. A mitochondrial diagnostics check list and test request form must accompany the specimen.

**Patient Preparation:** None

**Sample Rejection:** Frozen samples; mislabeled or unlabeled specimens

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**Interpretive**

**Reference Range:** No mutation detected

**Critical Values:** N/A

**Limitations:** N/A

**Methodology:** Massively Parallel Sequencing (MPS)

**References:**  
Baylor College of Medicine June 2014  
(800) 411-4363  Fax (713) 798-2787
**Updates:**

1/5/2005: Moved from Athena Diagnostic Laboratories to Georgetown. Note changes in draw volume and test includes sections.

5/18/2005: Moved from Georgetown University to Baylor College of Medicine. Note changes in CPT coding, specimen collection volumes, requisition/paperwork requirements, and methodology. Single deletion testing has been expanded to include maternally inherited diabetes/hearing loss.

1/26/2012: CPT update.