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:	81465 – Whole mitochondrial genome large deletion analysis panel
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 (Baylor 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**.

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 blood collection ● 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

Interpretive

Reference Range:	No mutation detected
Critical Values:	N/A
Limitations:	N/A
Methodology:	Massively Parallel Sequencing (MPS)
References:	Baylor College of Medicine April 2018 (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. 6/17/2014: Method change, replaced original test 3000.