ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

*A nonprofit enterprise of the University of Utah and its Department of Pathology*

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**THIS IS NOT A TEST REQUEST FORM.**
**The information below is required to perform mitochondrial disorder testing.**
**Please fill out this form and submit it with the test request form or electronic packing list.**

# PATIENT HISTORY FOR MITOCHONDRIAL DISORDER TESTING

**Patient Name** _____ **Date of Birth** _____ **Sex** ☐ **F** ☐ **M**

**Physician** _____ **Physician Phone** _____

**Practice Specialty** _____ **Physician Fax** _____

**Genetic Counselor** _____ **Counselor Phone** _____

**Patient's ethnicity** (Check all that apply)

☐ African American ☐ Ashkenazi Jewish ☐ Asian ☐ Caucasian
☐ Hispanic ☐ Middle Eastern ☐ Native American ☐ Other

**Suspected Diagnosis:**

☐ Cardiomyopathy ☐ Kearns-Sayre ☐ LHON ☐ MERRF ☐ Pearson
☐ CPEO ☐ Leigh/Leigh-like ☐ MELAS ☐ NARP ☐ Other_____

**Is the patient SYMPTOMATIC?** ☐ No ☐ Yes (Check all that apply)

**Developmental:** ☐ MR/DD/IDD ☐ Autism/PDD ☐ Other_____
**Neurological:** ☐ Hypotonia ☐ Encephalopathy ☐ Seizures ☐ Other_____
**Neuromuscular:** ☐ Neuropathy ☐ Exercise intolerance ☐ Muscle weakness ☐ Other_____
**Sensory:** ☐ Hearing loss ☐ Retinitis Pigmentosa ☐ Other_____
**Cardiac:** ☐ Cardiomyopathy ☐ Other_____
**Endocrine** (Describe): _____
**Other Symptoms**: _____

**Laboratory Findings:**

**Muscle biopsy** ☐ Normal ☐ Abnormal **(If abnormal, what was found?)**
☐ Ragged red fibers ☐ COX deficiency ☐ Abnormal ultrastructure (EM)
☐ Abnormal histology ☐ Enlarged mitochondria/mitochondrial proliferation
☐ Abnormal respiratory enzymes ☐ Other_____

**Metabolic** ☐ Normal ☐ Abnormal **(If abnormal, what was found?)**
☐ Lactic acidosis ☐ Elevated pyruvate ☐ Other_____

**Has the patient undergone previous genetic testing for a mitochondrial disorder?** ☐ No ☐ Yes ☐ Unknown
If yes, please describe the test performed and results: _____

**Does the patient have a FAMILY HISTORY of a mitochondrial disorder?** ☐ No ☐ Yes ☐ Unknown
NAME OF THE DISORDER diagnosed in the symptomatic/affected relative(s): _____
RELATIONSHIP of the affected family member(s) to the patient: _____
Is the relative a: ☐ healthy carrier ☐ affected with the above disorder.
What are the variant(s) identified in the relative? _____
**A copy of the DNA test results for the affected/carrier relative MUST be provided.**

Check **the test you intend to order:**

☐ **2006054 Mitochondrial Disorders Panel (mtDNA Sequencing, Nuclear Genes by Sequencing and Deletion/Duplication)**

☐ **2006065 Mitochondrial Disorders (mtDNA) Sequencing**: Sequencing of the mitochondrial genome.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Targeted sequencing for a mitochondrial or nuclear gene variant previously identified in a family member; copy of relative's lab result is REQUIRED. Contact an ARUP genetic counselor prior to ordering.

**For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141**

**Master Label**