

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** _____

Genetic Counselor _____ **Counselor Phone** (____) _____

Patient's Ethnicity (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African-American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

Suspected diagnosis: Cardiomyopathy CPEO Kearns-Sayre Leigh/Leigh-like LHON
 MELAS MERRF NARP Pearson Other _____

Is the patient SYMPTOMATIC? No Yes If yes, check all symptoms that apply

- | | | |
|---|---|--|
| Developmental: <input type="checkbox"/> MR/DD | <input type="checkbox"/> Autism/PDD | <input type="checkbox"/> Other _____ |
| Neurological: <input type="checkbox"/> Hypotonia | <input type="checkbox"/> Encephalopathy | <input type="checkbox"/> Seizures |
| Neuromuscular: <input type="checkbox"/> Neuropathy | <input type="checkbox"/> Exercise intolerance | <input type="checkbox"/> Muscle weakness |
| Sensory: <input type="checkbox"/> Hearing loss | <input type="checkbox"/> Retinitis Pigmentosa | <input type="checkbox"/> Other _____ |
| Cardiac: <input type="checkbox"/> Cardiomyopathy | | <input type="checkbox"/> Other _____ |

Endocrine: Describe: _____
Other Symptoms: _____

LAB RESULTS

Muscle biopsy: Normal Abnormal **If abnormal, what was found?**

- | | | | |
|--|---|--|---|
| <input type="checkbox"/> Ragged red fibers | <input type="checkbox"/> COX deficiency | <input type="checkbox"/> Anormal ultrastructure (EM) | <input type="checkbox"/> Abnormal histology |
| <input type="checkbox"/> Enlarged mitochondria/mitochondrial proliferation | <input type="checkbox"/> Abnormal respiratory enzymes | <input type="checkbox"/> 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 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.

Circle 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 a genetic counselor at (800) 242-2787, ext. 2141

Master Label