

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)

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

Suspected Diagnosis:

- | | | | | |
|---|---|--------------------------------|--------------------------------|--------------------------------------|
| <input type="checkbox"/> Cardiomyopathy | <input type="checkbox"/> Kearns-Sayre | <input type="checkbox"/> LHON | <input type="checkbox"/> MERRF | <input type="checkbox"/> Pearson |
| <input type="checkbox"/> CPEO | <input type="checkbox"/> Leigh/Leigh-like | <input type="checkbox"/> MELAS | <input type="checkbox"/> NARP | <input type="checkbox"/> Other _____ |

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

- | | | | |
|---|---|--|--------------------------------------|
| Developmental: <input type="checkbox"/> MR/DD/IDD | <input type="checkbox"/> Autism/PDD | <input type="checkbox"/> Other _____ | |
| Neurological: <input type="checkbox"/> Hypotonia | <input type="checkbox"/> Encephalopathy | <input type="checkbox"/> Seizures | <input type="checkbox"/> Other _____ |
| Neuromuscular: <input type="checkbox"/> Neuropathy | <input type="checkbox"/> Exercise intolerance | <input type="checkbox"/> Muscle weakness | <input type="checkbox"/> Other _____ |
| 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: _____

Laboratory Findings:

- | | | |
|----------------------|---------------------------------|---|
| Muscle biopsy | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal (If abnormal, what was found?) |
| | | <input type="checkbox"/> Ragged red fibers
<input type="checkbox"/> Abnormal histology
<input type="checkbox"/> Abnormal respiratory enzymes |
| | | <input type="checkbox"/> COX deficiency
<input type="checkbox"/> Enlarged mitochondria/mitochondrial proliferation
<input type="checkbox"/> Other _____ |
| Metabolic | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal (If abnormal, what was found?) |
| | | <input type="checkbox"/> Lactic acidosis
<input type="checkbox"/> Elevated pyruvate
<input type="checkbox"/> 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