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.
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 ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

Suspected Diagnosis:

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

Does the patient have symptoms? ☐ No ☐ Yes (Check all that apply)

Developmental	Neurological	Neuromuscular	Sensory	Cardiac
☐ MR/DD/IDD	☐ Hypotonia	☐ Neuropathy	☐ Hearing loss	☐ Cardiomyopathy
☐ Autism/PDD	☐ Encephalopathy	☐ Exercise intolerance	☐ Retinitis Pigmentosa	☐ Other (describe below)
☐ Other (describe below)	☐ Seizures	☐ Muscle weakness	☐ Other (describe below)	
	☐ Other (describe below)	☐ Other (describe below)		

☐ Endocrine (describe): _____

☐ Other symptom(s): _____

_____

Laboratory Findings:

☐ Muscle biopsy: ☐ Normal ☐ Abnormal (If abnormal, what was found?)

☐ Abnormal histology ☐ Enlarged mitochondria/mitochondrial proliferation
☐ Abnormal respiratory enzymes ☐ Ragged red fibers
☐ Abnormal ultrastructure (EM) ☐ Other: _____
☐ COX deficiency

☐ 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

If yes, attach a pedigree or specify the relative's relationship to the patient. Describe the disorder. List the clinical findings and age of onset:

_____

_____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, attach a copy of the relative's DNA laboratory result. (REQUIRED for familial mutation testing.)

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 testing for a mitochondrial or nuclear gene sequence variant previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

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