ARUP LABORATORIES

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_____ /_____/_____ Gender [ ] F [ ] M

Physician _____ Physician Phone (_____)_____Practice Specialty_____

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 [ ] 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: [ ] MR/DD [ ] 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: _____

LAB RESULTS
Muscle biopsy: [ ] Normal [ ] Abnormal **If abnormal, what was found?**
[ ] Ragged red fibers [ ] COX deficiency [ ] Anormal ultrastructure (EM) [ ] Abnormal histology
[ ] Enlarged mitochondria/mitocondrial 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 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 mutations 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 by Sequencing and Deletion/Duplication, 121 Nuclear Genes by Sequencing, 119 Nuclear Genes by Deletion/Duplication): Next generation sequencing and deletion/duplication analysis of all 37 mitochondrial genes and nuclear genes associated with mitochondrial disorders.

2006065 Mitochondrial Disorders (mtDNA) Sequencing: Next generation sequencing of all 37 genes 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

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