REQUISITION – Mitochondrial Genome Panel

Hamilton Regional Laboratory Medicine Program

Juravinski Hospital
Clinical Genetics Laboratory - Room H2-19A
711 Concession Street, Hamilton, ON L8V IC3 Phone:
(905) 521-2100 x76944 | Fax: (905) 521-7913
Email: moleculargenetics@hhsc.ca

Patient Information

Clear Form

***Name** (print):
Surname, First Name

***DOB** (DD/MM/YYYY):

***Sex**: ☐M ☐F ☐Other

***Health Card No.**:

**Mandatory Information. Specimen cannot be processed without this data.*

Note: Specimen collection is NOT completed at this lab. Please proceed to any community lab for blood draw.

Reports To:

*Ordering Physician: _____
*Address: _____
*Phone: _____ *Fax: _____
*Email: _____
*Authorized Signature: _____

Additional Copies To:

*Name: _____
*Address: _____
*Phone: _____
*Fax: _____
*Email: _____

Please see the HRLMP Laboratory Test Information Guide (LTIG) for complete sample requirements and test information:
https://ltig.hrlmp.ca/

SPECIMEN INFORMATION: *PLEASE VISIT THE LTIG WEBSITE FOR SAMPLE SHIPPING REQUIREMENTS*

Collection Date (DD/MM/YYYY): _____ Time of collection: _____

- ☐ **Peripheral blood** in EDTA (2 x 5ml >2 yrs/age, 2 x 3ml <2 yrs/age)
- ☐ **Fibroblast** (tissue culture dish 100 x 200mm)
- ☐ **Urine** (4ml of random urine, first catch, no preservatives)
- ☐ **Frozen Muscle** (30-50mg from muscle)
- ☐ **DNA** (minimum 5 µg, minimum [50ng/µl]).
- ☐ **Other** (contact Lab directly)

TEST REQUESTED: ☐ **Entire mtDNA sequenced by Next Generation Sequencing (NGS)** Please select from criteria below.

CLINICAL INFORMATION: Please check all that apply & provide relevant laboratory and/or pathology reports.

Suspected Disease:
[ ] Leigh disease
[ ] Primary lactic acidosis (in the absence of tissue hypoxia or multi-organ failure)
[ ] Chronic progressive external opthalmoplegia
[ ] Mitochondrial neuro-gastro-intestinal ophthalomyopathy (MNGIE)
[ ] Multiple symmetric lipomatosis (must include head and/or neck)
[ ] Pearson syndrome
[ ] Kearns-Sayres syndrome
[ ] Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS)
[ ] Myoclonic epilepsy with ragged-red fibers (MERFRF)
[ ] Neuropathy, ataxia, and retinis pigmentosa (NARF)
[ ] Leber's hereditary optic neuropathy (LHON)
[ ] Gentamicin-related sensorineural hearing loss

Pathological Features (attach report):
[ ] >2% ragged red fibers in skeletal muscle
[ ] 1-2% ragged red fibers, aged 30-50 years
[ ] Any ragged red fibers, aged <30 years
[ ] 2% subsarcolemma mitchondrial accumumlation, aged <16 years
[ ] >2% COX-negative fibers, aged <50 years
[ ] >5% COX-negative fibers, aged >50 years
[ ] Other suggestive pathology finding

Clinical Features:

A) Sign of highly oxidative tissue/organ:
* without other known etiology
[ ] CNS
[ ] Pigmentary retinopathy and/or optic atrophy
[ ] Peripheral neuropathy
[ ] Sensorineural hearing loss
[ ] Rhabdomyolysis
[ ] Fixed weakness of skeletal muscle
[ ] Cardiomyopathy and/or arrhythmias and/or conduction block
[ ] Proximal renal tubulopathy (Fanconi syndrome)
[ ] Type 2 diabetes mellitus
[ ] Sideronblastic anemia
[ ] GI tract: pseudoobstruction, hepatopathy
[ ] Unexplained failure to thrive or short-stature

B) AND one of the following:
[ ] Clinical progression with stepwise exacerbation of symptoms
[ ] Positive family history for any of A)
[ ] Lactic acidosis (non-acute illness)
[ ] Elevated alanine (plasma amino acids)
[ ] Elevated 3-methylglutaconic acid (urine organic acids)

Biochemical Features (attach report):
[ ] <30% activity of any RC complex in tissue or cell line (% of normal control mean relative to citrate synthase)
[ ] Persistent (>2 samples on separate days), unexplained hyperlactatemia (>3.0mmol/L in free-flowing sample drawn without tourniquet)
[ ] Increased lactate:pyruvate ratio (>25) in skin fibroblasts
[ ] Persistent (>2 samples) hyperalaninemia >500uM
[ ] Persistent (>2 samples) abnormal excretion of lactate, pyruvate, and/or TCA cycle intermediates in urine
[ ] Evidence of mtDNA depletion or multiple mtDNA deletions
[ ] Patient fibroblasts unable to grow with galactose as sole carbon source
[ ] Other suggestive biochemical finding

Other Relevant Clinical Information:
_____
_____
_____
_____

Continued on page 2.

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KNOWN FAMILIAL VARIANT: Please provide a pedigree and a proband report if not tested at HRLMP.

Proband Name: _____

Proband DOB (DD/MM/YY): _____

Proband Relationship to Patient:_____

Familial Variant Name or HRLMP Report #:_____

FAMILY PEDIGREE: Please clearly annotate all known symptoms and mutations.