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 ophthalmyopathy (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.