SickKids®

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Paediatric Laboratory Medicine

MITOCHONDRIAL TESTING

Referred-in Client Requisition

MITOCHONDRIAL LABORATORY

Room 3642, Atrium
555 University Ave
Toronto, ON, Canada, M5G 1X8

Tel: 416-813-7200
Fax: 416-813-5431

Patient Last Name:

First Name:

Birth date (YYYY-MM-DD):

Gender: ☐ *Male* ☐ *Female* ☐ *Unknown*

For Canada Only

Health Card #: *Version:*

Issuing Province:

Specimen Collection Information		
Date (YYYY-MM-DD)	**Time** (HH:MM)	**Collected by:**

Sample #: _____ **If fibroblasts, # of passages:** _____ **Date of Referral** (YYYY-MM-DD) _____

Referring Physician / Institution		
Name	**Address**	**Telephone**

Note: *DO NOT* submit specimens from patients with HIV+ve status. HIV+ve status interferes with testing.

SKIN FIBROBLAST, AMNIO/CVS TESTING

- ☐ Pyruvate Determination for L/P ratio
- ☐ Lactate Determination for L/P ratio
- ☐ Cytochrome Oxidase (Comp. IV)
- ☐ Succinate Cytochrome C Reductase (Comp. II+III)
- ☐ Pyruvate Dehydrogenase - Total
- ☐ Pyruvate Decarboxylase - E1
- ☐ Pyruvate Dehydrogenase - E2
- ☐ Pyruvate Dehydrogenase - E3
- ☐ Pyruvate Carboxylase (PC)
- ☐ Phosphoenolpyruvate carboxykinase (PEPCK)

- ☐ **Partial Screen:** All of the above tests with the exception of PC, PEPCK
- ☐ **Total screen**: All of the above tests

For in-patient: Skin biopsy collection medium can be obtained from Tissue Culture Laboratory (ext 202394) or Pathology (ext 205944). The sample MUST be sent to Tissue Culture Laboratory (Rm # 3225).

For testing on amniocytes: Provide at least 3 confluent 25 mL flasks of amniocytes with the same number of flasks of at least two different controls. Keep a backup flask growing. For controls use amniocytes/CVS from individuals approximately the same gestation and age, "discards" from testings for LATE MATERNAL AGE.

For testing on outpatients: Provide 2 x 25 mL flasks of cell culture. Cells will be cultured by Tissue Culture Laboratory for the duration of the tests.

***Note*:** All fibroblast and amnio specimens must be shipped at room temperature. For shipment of skin biopsies call Tissue Culture Laboratory at 416-813-7654 ext 202394.

- ☐ Skin fibroblast mitochondrial isolation (NADH: cytochrome c reductase (CI+III), CII+III, CIV, ATPase (CV), citrate synthase (CS))

Test requires **20 plates** (10 cm) for mitochondrial isolation with the same number of plates from a control cell line, and thus will delay testing and results.

BIOPSY TESTING ON FROZEN TISSUE (Total tissue homogenate: muscle, liver, heart, kidney, brain)

- ☐ Comp I+III, II+III, IV and CS

Provide about **50 mg** of tissue in a plastic cryovial snap frozen in liquid nitrogen.

Note: Specimen **should NOT** be immersed in isopentane or any other fluid before freezing. All frozen specimens must be shipped in a cryovial on plenty of **dry ice**. Ship early in the week by overnight courier. Specimens received thawed **CANNOT** be tested.

BIOPSY TESTING ON ENDOCARDIAL BIOPSY

- ☐ Comp I+III, II+III, IV and CS

Make arrangement with the lab at least **24 hrs prior to the procedure**. Provide **2-5 mg fresh specimen**.

Specimen should be transported in a small container **ON ICE**.

BIOPSY TESTING ON ISOLATED MUSCLE MITOCHONDRIA, FRESH TISSUE

- ☐ NADH: ubiquinone reductase (CI), CI+III, Succinate DCIP reductase (CII), CII+III, CIV, CV, CS

Make arrangement with the lab at least 24 hrs prior to the biopsy. Provide **250-300 mg of fresh muscle** for mitochondrial isolation. Specimen that weighs less than 200 mg will be snap frozen and processed as "frozen tissue"

All fresh biopsies should be transported in a plastic container **ON ICE**.

Please continue and complete the '**Clinical Information Sheet**' on page 2.

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Specimen Collection Information

Date (YYYY-MM-DD)	**Time** (HH:MM)	**Collected by:**

Sample #: _____ **If fibroblasts, # of passages:** _____ **Date of Referral** (YYYY-MM-DD) _____

Referring Physician / Institution

Name	**Address**	**Telephone**

Please complete and submit this form in conjunction with the "Mitochondrial Testing Requisition".

Clinical information (Please check):

Age at onset: _____

CNS	Ophthalmologic	Muscle	Cardiac	General
☐ Microcephaly	☐ Optic atrophy	☐ Myopathy	☐ Conduction abnormalities	☐ Failure to thrive
☐ Developmental Delay	☐ Leber's HON	☐ Hypotonia	☐ Cardiomyopathy	☐ Feeding problems
☐ Stroke-like episodes	☐ Pigmentary retinopathy	☐ Exercise intolerance	☐ Hypertrophic	☐ Lethargy
☐ Ataxia	☐ Cortical Blindness	☐ Ptosis	☐ Dilatative	☐ Dysmorphic facies
☐ Myoclonus	☐ Nystagmus		☐ Other	
☐ Dystonia	**Nerve**	**Relevant family history**		
☐ Sensorineural hearing loss	☐ Neuropathy			
☐ Seizures	☐ Axonal			
☐ Encephalopathy	☐ Demyelinating			
☐ Ophthalmoplegia	**Hepatic**			
☐ Leigh' s Disease	☐ Hepatic dysfunction			
☐ Basal Ganglia Calcification	☐ Hepatomegaly			
	Renal			
	☐ Renal tubular acidosis			

LABORATORY DATA (IF KNOWN):

☐ Serum lactate: _____	☐ CSF Lactate: _____	☐ EMG: _____	☐ ABR: _____
☐ ALT: _____	☐ AST: _____	☐ NCS: _____	☐ VEP: _____
☐ Alkaline phosphatase: _____	☐ BUN: _____	☐ CT: _____	☐ SSEP: _____
☐ Creatinine: _____	☐ Other: _____	☐ MRI: _____	

Past muscle or skin biopsy ☐ Yes ☐ No