

University of Colorado Denver (UCD)

**BIOCHEMICAL GENETICS
LABORATORY**

Department of Pediatrics

**RC-1 North Rm. P18-4403C
12800 East 19th Avenue
Aurora, CO 80045**

Phone: (303)724-3826

Fax: (303)724-3827

Web: www.uchsc.edu/bglab

PATIENT NAME:

DATE OF BIRTH:

FEMALE MALE

MEDICAL RECORD NUMBER:

INSTITUTION SAMPLE #:

REQUESTING PHYSICIAN:

PHONE:

FAX:

BILLING HOSPITAL NAME:

ADDRESS:

PHONE:

FAX:

COLLECTION DATE:

TIME:

Please use only private shipping companies (UPS, FedEx, DHL, etc.) for all parcels containing specimens.

PLEASE NOTE: THIS LABORATORY DOES NOT ACCEPT DELIVERIES ON WEEKENDS OR HOLIDAYS.

For letters mailed via the United States Postal Service, please replace 'RC-1 North Rm. P18-4403C' with 'Mail Stop 8313.'

Please check test required (below left) and complete all specimen and clinical information (below right and second page)

Mitochondrial respiratory chain enzyme assays

Tissue type:

Muscle weight: _____ mg

Liver weight: _____ mg

Tissue source:

Biopsy

Autopsy (collected ___ hours after death)

Specimen storage prior to shipment:

Liquid nitrogen

-70° C

-20° C

Additional Comments:

Tissue requirements: tissue must be snap frozen, stored at -70° Celsius, and shipped overnight on dry ice. Autopsied samples must be obtained less than 4 hours after death.

- **Muscle tissue:** Minimum volume required is 60 mg.
- **Liver tissue:** Minimum volume required is 20 mg. Samples > 60 mg will be performed with a regular assay. Samples of between 20 and 60 mg will require a microassay. Please note that the reliability of the microassay is less robust than that of the regular assay.

**PLEASE NOTE: REFERRAL SOURCE IS RESPONSIBLE FOR PAYMENT.
WE DO NOT BILL PATIENTS OR THEIR INSURANCE COMPANIES.**

FOR INTERNAL USE ONLY

Date Received: _____

Patient sample #: _____

CLINICAL INFORMATION

Please check all that apply

PATIENT NAME:

General

- failure to thrive
- short stature

Brain

- microcephaly
- encephalophaly
- seizures:
 - myclonic seizures
 - infantile
 - other seizures
- chorea
- dystonia
- parkinson
- ataxia
- neurodegenration
- stroke-like episodes
- central apnea
- other: _____

Eye

- optic atrophy
- retinitis pigmentosa / retinal dystrophy
- cataracts

Hearing

- nerve deafness
- hearing loss
- other

Liver

- elevated transaminases
- fibrosis/steatosis
- liver insufficiency
- hypoglycemia

Additional comments:

Print physician name: _____

Physician signature: _____

Kidney

- renal Fanconi
- glomerulosclerosis
- proteinuria – nephritic syndrome

Endocrine

- diabetes mellitus
- hypoparathyroidism

GI

- pancreatitis
- pancreatic insufficiency
- pseudo-obstruction
- malabsorption

Heart

- cardiomyopathy:
 - dilated
 - hypertrophic
- Conduction defect
- Arrhythmias

Skin

- hyperpigmentation

Muscle

- myopathy
- early fatigue
- abnormal exercise test
- rhabdomyolysis
- elevated CK: _____ U/L
- Biopsy:
 - ragged red fibers
 - abnormal e.m.

Laboratory Studies

- Lactate:
 - Blood: _____ mM
 - CSF: _____ mM
 - lactate/pyruvate ratio: _____
- 3OHB/AcAc ratio: _____
- 3-methylglucatonic acid
- low total carnitine
- lactate on MRS
- elevated alanine: _____ μM
- incr. Krebs cycle metabolites
- other: _____

Radiology

- abnormal basal ganglia
- abnormal brain stem
- abnormal nucl. dentate
- brain atrophy
- cerebellar atrophy
- leukodystrophy
- other: _____

Recognized Syndromes

- Leigh disease
- Progressive external ophthalmoplegia (CPEO)
- Kearns-Sayre
- MELAS
- MERRF
- NARP
- MNGIE
- Diabetes-deafness
- Diabetes- retinitis pigmentosa

FOR INTERNAL USE ONLY

Date Received: _____ Patient sample #: _____