

Bill to: Client/Submitter

Submitting institution is responsible or ament



Children's Hospital Colorado

Mitochondrial Laboratory**Shipping Address:***** Please use FedEx or UPS ONLY ***Children's Hospital Colorado
Clinical Lab – Mitochondrial Lab
13123 E 16th Ave, Room B0200
Aurora, CO 80045

Submitting Institution Information		Patient Information		
Institution:		Patient's Last Name	First	Middle
Address:		DOB:	Sex:	Gender:
City:	State:	Zip:	Diagnosis:	
Phone:	Fax:	Ordering Provider (Last, First, Middle Initial):		
Contact Name:		NPI:	Phone:	
Phone:	Email:	Email:	Fax:	

By submitting this Requisition Form to CHCO, you are acknowledging and agree to our standard Terms and Conditions and agree to pay CHCO the rates associated with our standard fee schedule in effect on the day the specimen is received.

Required Specimen Information

Date Collected:	Time Collected:	AM/PM	External ID:
Tissue Source:		Tissue Type:	
Biopsy	Autopsy	Heart Weight(mg):	Liver Weight(mg):
Collected _____ hrs after death		Muscle Weight(mg):	Fibroblast Mycoplasma Tested? Yes* No
Specimen storage prior to shipment		Antibiotics linezolid or tigecycline in last week? (for tissues)	
Liquid Nitrogen	-20°C	-70°C	Yes No Unknown

Testing Information

Fibroblasts	Tissue
Mitochondrial Respiratory Chain, Fibroblast (LAB7079)	Mitochondrial Respiratory Chain, Tissue (LAB7086)
Blue Native Electrophoresis, Fibroblast (LAB7078)	Blue Native Electrophoresis, Tissue (LAB7087)
Pyruvate Dehydrogenase, Fibroblast (LAB7085)	
Other Sources	
Complex 1 Assembly Western Blot, Fibroblast (LAB9301)	
Complex V Hydrolysis Enzyme Assay (LAB10561)	Primary Fibroblast Culture, Skin Biopsy (LAB9109)
Complex V ATP Hydrolysis + Mito. Respiratory Chain, Fibroblast (LAB10592)	GDF15 and FGF21, Serum/Plasma (LAB10330)

Clinical Information

General Failure to thrive Short stature Brain Microcephaly Encephalopathy Seizures Myoclonic Infantile Other Chorea Dystonia Parkinson Ataxia Neurodegeneration Stroke-like episodes Central apnea Leukodystrophy Other:	Endocrine Diabetes mellitus Hypoparathyroidism Kidney Renal Fanconi Glomerulosclerosis Proteinuria – nephritic syndrome Liver Elevated transaminases Fibrosis/steatosis Liver insufficiency Hypoglycemia GI Pancreatitis Pancreatic insufficiency Pseudo-obstruction Malabsorption Hearing Nerve deafness Hearing loss	Laboratory studies Lactate: Blood: mM CSF: mM Lactate/pyruvate ratio: 3OHB/ACAc Ratio: 3-methylglutaconic acid Low total carnitine Lactate on MRS Elevated alanine: μ M Incr. Krebs cycle metabolites Other: Radiology Abnormal basal ganglia Abnormal brain stem Abnormal dentate nucleus Brain atrophy Cerebellar atrophy Leukodystrophy Other:	Recognized Syndromes Leigh disease Kearns-Sayre MELAS MERRF NARP MNGIE Diabetes-deafness Diabetes-retinitis pigmentosa CPEO Genetic Information Gene Name <hr/> Variant <hr/>
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If Mycoplasma was tested, SEND REPORT*FOR ALL MITOCHONDRIAL ASSAY SPECIMENS:**

Please enclose both the completed requisition and clinical information forms for mitochondrial enzyme assays. Direct contact information for the requesting physician is of importance to allow direct notification as needed, so please provide the clinician's phone number. Incomplete paperwork can result in delayed processing.

Instructions for Collecting, Handling & Shipping Specimens

FOR ALL MITOCHONDRIAL ASSAY SPECIMENS:

Please enclose both the completed requisition and clinical information forms for mitochondrial enzyme assays. Direct contact information for the requesting physician is of importance to allow direct notification as needed, so please provide the clinician's phone number. Incomplete paperwork can result in delayed processing.

Primary contact for mitochondrial enzyme assays:

Marisa Friederich, PhD

Phone: 720-777-0528 | Email: Marisa.Friederich@ChildrensColorado.org

Fibroblast for Respiratory Chain Enzyme Assays, BNPage with In-Gel Activity Staining Assay, complex I assembly assay, and Pyruvate Dehydrogenase Enzyme Assay:

- Please send two T-25 flasks containing confluent fibroblasts (with plug-seal caps, filled completely with media) in a Styrofoam box at room temperature. Make sure to avoid any freezing during transportation.
- Fibroblasts must be mycoplasma free. Please provide documentation of mycoplasma testing results. We will perform mycoplasma testing if no result is provided.
- Skin Biopsies should be collected using an aseptic technique and the skin punch should be placed in a leak proof tube filled completely with tissue culture media (RPMI, DMEM, MEM, etc). Please indicate anatomic source/location of tissue biopsy on the requisition. Ship tube overnight in a Styrofoam box either at room temperature or at 4°C.

Mitochondrial Respiratory Chain Enzyme Assays (muscle, liver, and heart):

- Tissue must be **frozen immediately** in liquid nitrogen or on dry ice, within at most a few minutes no longer (do not delay freezing until after wound closure as this will result in a degraded sample).
- Tissue cannot be placed in any preservative including OCT.
- Tissue must be stored in a microvial at -70° C until shipment on dry ice. (- 20°C is not sufficient)
- If possible, weigh biopsy before placing in the container. If the specimen was not weighed before it was frozen, DO NOT attempt to weigh a frozen sample as the tissues degrade very easily. Instead, ship the sample to the Mitochondrial Diagnostic Laboratory without a weight. Lab personnel will weigh the sample.
- Postmortem samples must be obtained within 2 hours of death, but as soon as possible as interpretation will become more difficult with time.

Specimen Requirements for Tissue Types:

- **Muscle Tissue:** Minimum of 60 mg required
 - Muscle biopsies must be obtained without the use of electrocautery and must be completely processed within 5 to 10 minutes of being excised. Muscle biopsies should be preferably about 120 mg in size.
- **Liver Tissue:** Minimum 20 mg required. Samples of <60 mg will require microassay, which is less robust.
 - Liver samples can be obtained as a wedge biopsy or as a needle biopsy. It is important to ensure that sufficient liver tissue is obtained. If using a needle biopsy, obtain two (2) needle biopsies 14 Gauche Bard monoptery instrument 2 cm length.
- **Heart Tissue:** Minimum 20 mg required
 - Heart biopsies must be obtained without the use of electrocautery and must be completely processed within 5 to 10 minutes of being excised. Heart biopsies should be at least 20 mg in size.

BNPage with In-Gel Activity Staining (muscle, liver, and heart)

Instructions for obtaining samples are the same as detailed for Respiratory Chain Enzyme Analysis

Specimen Requirements for Tissue Types:

- **Muscle:** 100 mg required. If ordering both Respiratory Chain Enzyme Assays and BNPage Analysis, minimum requirement is 150mg.
- **Liver:** 50 mg required. If ordering both Respiratory Chain Enzyme Assays and BNPage Analysis, minimum requirement is 80 to 120 mg.
- **Heart:** 50 mg required. If ordering both Respiratory Chain Enzyme Assays and BNPage Analysis, minimum requirement is 50 to 100 mg.

For all assays, samples smaller than minimal size may result in the lab not being able to process the sample or in reduced accuracy and reliability of the assay.