



Children's Hospital Colorado

## Mitochondrial Laboratory

**Shipping Address:**

**\* Please use FedEx or UPS ONLY \***  
 Children's Hospital Colorado  
 Clinical Lab – Mitochondrial Lab  
 13123 E 16<sup>th</sup> 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:		External ID:
		AM/PM		
Tissue Source:		Tissue Type:		
<input type="checkbox"/> Biopsy	<input type="checkbox"/> Autopsy	<input type="checkbox"/> Heart Weight(mg):	<input type="checkbox"/> Liver Weight(mg):	<input type="checkbox"/> Skin
Collected ____ hrs after death		<input type="checkbox"/> Muscle Weight(mg):	<input type="checkbox"/> Fibroblast Mycoplasma Tested? <input type="checkbox"/> Yes* <input type="checkbox"/> No	
Specimen storage prior to shipment			Antibiotics linezolid or tigecyclin in last week? (for tissues)	
<input type="checkbox"/> Liquid Nitrogen	<input type="checkbox"/> -20°C	<input type="checkbox"/> -70°C	<input type="checkbox"/> Yes	<input type="checkbox"/> No <input type="checkbox"/> Unknown

Testing Information	
<input type="checkbox"/> Mitochondrial Respiratory Chain, Fibroblast (LAB7079)	<input type="checkbox"/> Mitochondrial Respiratory Chain, Tissue (LAB7086)
<input type="checkbox"/> Blue Native Electrophoresis, Fibroblast (LAB7078)	<input type="checkbox"/> Blue Native Electrophoresis, Tissue (LAB7087)
<input type="checkbox"/> Pyruvate Dehydrogenase, Fibroblast (LAB7085)	<input type="checkbox"/> Primary Fibroblast Culture, Skin Biopsy (LAB9109)
<input type="checkbox"/> Complex 1 Assembly Western Blot, Fibroblast (LAB9301)	<input type="checkbox"/> GDF15 and FGF21, Serum/Plasma (LAB10330)

Clinical Information			
<b>General</b> <input type="checkbox"/> Failure to thrive <input type="checkbox"/> Short stature <b>Brain</b> <input type="checkbox"/> Microcephaly <input type="checkbox"/> Encephalophaly <input type="checkbox"/> Seizures <input type="checkbox"/> Myoclonic <input type="checkbox"/> Infantile <input type="checkbox"/> Other <input type="checkbox"/> Chorea <input type="checkbox"/> Dystonia <input type="checkbox"/> Parkinson <input type="checkbox"/> Ataxia <input type="checkbox"/> Neurodegeneration <input type="checkbox"/> Stroke-like episodes <input type="checkbox"/> Central apnea <input type="checkbox"/> Leukodystrophy <input type="checkbox"/> Other:	<b>Endocrine</b> <input type="checkbox"/> Diabetes mellitus <input type="checkbox"/> Hypoparathyroidism <b>Kidney</b> <input type="checkbox"/> Renal Fanconi <input type="checkbox"/> Glomerulosclerosis <input type="checkbox"/> Proteinuria – nephritic syndrome <b>Liver</b> <input type="checkbox"/> Elevated transaminases <input type="checkbox"/> Fibrosis/steatosis <input type="checkbox"/> Liver insufficiency <input type="checkbox"/> Hypoglycemia <b>GI</b> <input type="checkbox"/> Pancreatitis <input type="checkbox"/> Pancreatic insufficiency <input type="checkbox"/> Pseudo-obstruction <input type="checkbox"/> Malabsorption <b>Hearing</b> <input type="checkbox"/> Nerve deafness <input type="checkbox"/> Hearing loss	<b>Laboratory studies</b> <input type="checkbox"/> Lactate: <input type="checkbox"/> Blood: mM <input type="checkbox"/> CSF: mM <input type="checkbox"/> Lactate/pyruvate ratio: <input type="checkbox"/> 3OHB/AcAc Ratio: <input type="checkbox"/> 3-methylglutaconic acid <input type="checkbox"/> Low total carnitine <input type="checkbox"/> Lactate on MRS <input type="checkbox"/> Elevated alanine: μM <input type="checkbox"/> Incr. Krebs cycle metabolites <input type="checkbox"/> Other: <b>Radiology</b> <input type="checkbox"/> Abnormal basal ganglia <input type="checkbox"/> Abnormal brain stem <input type="checkbox"/> Abnormal dentate nucleus <input type="checkbox"/> Brain atrophy <input type="checkbox"/> Cerebellar atrophy <input type="checkbox"/> Leukodystrophy <input type="checkbox"/> Other:	<b>Recognized Syndromes</b> <input type="checkbox"/> Leigh disease <input type="checkbox"/> Kearns-Sayre <input type="checkbox"/> MELAS <input type="checkbox"/> MERRF <input type="checkbox"/> NARP <input type="checkbox"/> MNGIE <input type="checkbox"/> Diabetes-deafness <input type="checkbox"/> Diabetes-retinitis pigmentosa <input type="checkbox"/> CPEO <b>Genetic Information</b> Gene Name _____  Variant _____

\*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.

**Additional collection and handling instructions on the back/2nd page of this requisition.**

Please contact our Client Services team with any questions: LabClientServices@childrenscolorado.org

# 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](mailto: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 container filled completely with media. Please indicate anatomic source/location of tissue biopsy on the requisition.

## **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  $-70^{\circ}\text{C}$  until shipment on dry ice. ( $-20^{\circ}\text{C}$  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 monopty 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.*