

# 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

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## **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.*