



## PREPARATION OF SAMPLES FOR SHIPMENT

### BLOOD SAMPLES

#### A) WHOLE BLOOD FOR ENZYME ANALYSIS

1. For **Phosphorylase Kinase** assay send 10 cc of whole blood in a purple-top EDTA tube, also send 10 cc of a control blood (not a family member) along with the patient sample. Try to draw patient and control at approximately the same time. For **Pompe disease** and **Fabry disease**, collect 3-5cc in an EDTA tube.
2. Blood must be sent on 4°C ice packs, by overnight courier, the **same day** it is drawn.

#### B) DRIED BLOOD SPOTS FOR ENZYME ANALYSIS

*Note: We prefer to receive whole blood rather than pre-dried blood spots for Pompe disease and Fabry disease testing.*

1. Blood on Newborn Screening Card/Guthrie card (Dried Blood Spot)  
Minimum volume – 0.5ml.
  - a. Warm the site of extraction for 3 to 5 minutes with a soft cloth, moistened with warm water.
  - b. Cleanse site with alcohol prep. Wipe DRY with sterile gauze pad to avoid contamination of blood with alcohol.
  - c. Puncture heel (newborn only) or finger with safety lancet following lancet instructions. Hold foot or finger such that the puncture faces down.
  - d. Wipe off first blood drop with sterile gauze pad. Allow another large hanging blood drop to form, massage the surrounding tissue gently if needed.
  - e. Lightly touch the blood drop on the center of the first circle on the Newborn Screening Card. Allow the blood to soak through and completely fill the circle. Blood drops should be large enough to fill a circle with a single application.
  - f. Apply blood to one side of the paper card only. Check that the blood spot is completely visible on the other side of the filter paper. Do not apply more than one drop in the same paper circle.
  - g. Repeat steps E and F until all circles on the card are filled.
  - h. Label the card with the patient name, patient ID#, sample collection date (dd/mmm/yyyy format), sample collection time and test name (GAA Activity Dried Blood Spot).
  - i. Dry cards on a clean, flat, non-absorbent surface 4-5 hours at room temperature. When dry, place the card into a small biohazard-labeled plastic bag and seal tightly.
  - j. If the card must be stored overnight, place biohazard bag in a refrigerator until shipment.
2. Ideally, these samples should be shipped within 24 hours of collection.

#### C) WHOLE BLOOD FOR DNA ANALYSIS

1. Collect 5cc blood in a purple top EDTA tube.
2. Send by overnight delivery at ambient temperature or at 4°C with cold packs.



### **LIVER SAMPLES**

1. If a percutaneous punch biopsy is the only option, please send at least two good passes measuring at least 0.5 cm each, or a minimum of 15 mg total weight. Flush the sample out of the biopsy needle onto an absorbent filter paper **immediately** to remove the water.
2. The fresh sample should be immediately placed onto tin foil, wrapped well and dropped in liquid nitrogen or dry ice. Transfer the frozen tissue to a small plastic (polypropylene) tube and quickly pack in a Styrofoam container filled with dry ice. Do **not** preserve the sample with any method other than freezing. Send the frozen sample in dry ice by overnight express delivery. (We have best results with Fed Ex).

### **MUSCLE SAMPLES**

1. Sample should be 2 cm x 0.5 cm
2. Freeze sample and ship as described for liver.

### **CULTURED SKIN FIBROBLASTS**

1. Send at least two **CONFLUENT** T25 flasks of patient cells along with two **CONFLUENT** T25 flasks of a control fibroblast cell culture.
2. SHIP AT ROOM TEMPERATURE by overnight express delivery. Avoid freezing or overheating the cells. Stabilize the flasks in the container before shipping.

### **PRENATAL SAMPLES**

#### **CHORIONIC VILLI CULTURES (CVS)**

1. Culture the sample and send two **CONFLUENT** T25 flasks. Also send 2 **CONFLUENT** T25 flasks from a control CVS cell culture.
2. SHIP AT ROOM TEMPERATURE by overnight express delivery. Avoid freezing or overheating the cells. Stabilize the flasks in the container before shipping.

#### **AMNIOCYTES**

1. Establish a culture and send two **CONFLUENT** T25 flasks. Also send 2 **CONFLUENT** T25 flasks from a control amniocyte cell culture.
2. SHIP AT ROOM TEMPERATURE by overnight express delivery. Avoid freezing or overheating the cells. Stabilize the flasks in the container before shipping.

### **URINE SAMPLES**

1. Collect 1.0 mL (minimum 0.25 mL); no preservatives. No specific restrictions.
2. Freeze immediately and ship on dry ice.