



SPECIMEN REQUIREMENTS AND SHIPMENT

BLOOD SAMPLES

A) WHOLE BLOOD FOR ENZYME ANALYSIS

1. **Phosphorylase Kinase** assay:
 - a. **Sample volume: 1-5 mL** of whole blood in a **EDTA** lavender top tube.
 - b. Also send **1-5 mL of a control blood** (non-familial) with the patient sample.
 - c. The patient and control blood should be collected at approximately the same time.
2. **Pompe, Fabry, MPS I, MPS II, LAL-D, Gaucher, and Biotinidase DBS blood enzyme testing:**
 - a. **Sample volume: 0.5-5 mL** EDTA lavender top tube.
3. **CRIM analysis:**
 - a. **Sample volume: 3-5 mL** of whole blood
 - b. **Specimen type: BD Vacutainer CPT tube, or Acid Citrate Dextrose (ACD) yellow top tube, or EDTA lavender top tube.**

For all of these tests above, ship **whole blood** sample 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 lysosomal storage disease enzyme testing.

1. Specimen type: Dried Blood Spot (DBS) on Newborn Screening or Whatman 903 Protein Saver card.
2. Minimum sample requirement: 1 full circle
3. Ideally, these samples should be shipped within 24 hours of collection.
4. Ship at 4°C on a cold pack to maximize sample integrity for enzyme analysis.
5. DBS preparation instructions
 - 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/mm/yyyy format), sample collection time and test name.
- 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.
- k. Cards should be stored in the presence of a desiccant (silica gel bag) when possible, to minimize exposure to moisture.

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.
2. Flush the sample out of the biopsy needle onto an absorbent filter paper **immediately** to remove the water.
3. 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.
4. Store sample at -80°C and ship overnight on dry ice by express delivery.

MUSCLE SAMPLES

1. Minimum of 20 mg total weight (approximately 2 x 0.5 cm) is required.
2. After collection of the muscle biopsy, follow the instructions above in Liver Samples #3 for rapid freezing and preservation of the sample.
3. Store sample at -80°C and ship overnight on dry ice by express delivery.

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. Carefully wrap each flask with padding material to avoid breakage during transportation.
3. SHIP AT ROOM TEMPERATURE by overnight express delivery. Avoid freezing or overheating the cells. Stabilize the flasks in the container before shipping.

URINE SAMPLES FOR HEX₄ AND GLYCOSAMINOGLYCANS

1. Collect 1.0 mL (minimum 0.25 mL); no preservatives. No specific restrictions.
2. Store at -20°C and ship overnight on dry ice.

PLASMA SAMPLES FOR LYSO-GB₁ AND LYSO-GB₃

1. Collect 1.0 mL EDTA plasma (minimum 0.3 mL).
2. Store at -20°C and ship overnight on dry ice.

FOR ANY OTHER SAMPLE TYPE, PLEASE CONTACT THE LAB.