

## Specimen Collection Guide--Fibroblasts

### Applicable to:

- Acetyl-CoA:alpha-glucosaminide N-acetyltransferase
- Alpha-fucosidase
- Alpha-galactosidase
- Alpha-glucosidase
- Alpha-iduronidase
- Alpha-mannosidase
- Alpha-N-acetylgalactosaminidase
- Alpha-N-acetylglucosaminidase
- Alpha-neuraminidase
- Arylsulfatase A
- Arylsulfatase B
- Beta-glucosidase
- Beta-glucuronidase
- Beta-hexosaminidase
- Beta-mannosidase
- Galactocerebrosidase
- Heparan N-sulfate sulfatase
- Lysosomal acid lipase
- N-acetylgalactosamine-6-sulfate sulfatase
- Sphingomyelinase

### Minimum Specimen Requirements:

Fibroblasts - Two T25 flasks with ~80% confluent cells.

Biopsy - One 3 mm punch. Smaller specimens can be used with expected longer TAT.

### Storage/Transportation:

- **Fibroblasts**- Screw cap on tightly and seal with parafilm. Label flask with patients' name and date of birth. Ship overnight at room temperature
- **Tissue Biopsy**- Screw cap on tightly and seal with parafilm. Label tube with patient's name and date of birth. Wrap tubes in absorbent material and placed in a biohazard bag and sealed closed. Place in a small box containing absorbent material and tape closed.

**CELLS SHOULD BE MAINTAINED AT ROOM TEMPERATURE AT ALL TIMES**

Ship overnight at room temperature.

### Special Requirements

- Collection date is date of tissue biopsy.
- **Lysosomal Enzymes from Cultured Skin Fibroblasts:**  
Ship 2 x T25 flasks with ~80% confluent cells.  
Fill with media to top of flask. Screw cap on tightly and seal with parafilm.  
Label each flask clearly with patient name and date of birth (two identifiers are essential).
- **Lysosomal Enzymes from Tissue Biopsy:**  
Skin biopsy has to be performed under sterile conditions. Transfer skin biopsy immediately into individual sterile 15 mL screw cap tubes containing sterile tissue culture medium (Alpha MEM + 15% FBS + antibiotics). Top up the medium to fill the tube. Screw cap on tight and use parafilm to seal tube. Label each tube clearly with patient name and date of birth (two identifiers are essential). Wrap tubes in absorbent material and placed in a biohazard bag and sealed closed. If sending more than one biopsy, process each sample separately. If the specimen must be stored overnight prior to shipping, it can be left at room temperature in a sterile area.

## Specimen Collection Guide -- Leukocytes

### **Applicable to:**

- Alpha-fucosidase
- Alpha-galactosidase
- Alpha-iduronidase
- Alpha-mannosidase
- Alpha-N-acetylgalactosaminidase
- Alpha-N-acetylglucosaminidase
- Arylsulfatase A
- Beta-galactosidase
- Beta-glucosidase
- Beta-glucuronidase
- Beta-hexosaminidase (leukocytes preferred for pregnant females or females currently on birth control)
- Beta-mannosidase
- Galactocerebrosidase
- Lysosomal acid lipase
- N-acetylgalactosamine-6-sulfate sulfatase

### **Minimum Specimen Requirements**

- 5-6 mL whole blood collected in a Green Top Sodium or Lithium Heparin tube. DO NOT USE GEL-SEPARATOR TUBES.
- Several of the assays available may be performed on a single leukocyte pellet/blood

### **Storage/Transportation**

Ship whole blood sample at room temperature. Do not refrigerate or freeze.

Ship frozen isolated leukocytes on dry ice

### **Special Requirements**

- **Samples accepted by lab before 1:00p.m. on the day of collection, Monday to Friday (excluding statutory holidays)**
- Deliver to lab at room temperature.
- Consult the Laboratory if other arrangements are necessary.

## Specimen Collection Guide--Serum/Plasma

**Note: Most lysosomal enzymes are available on Leukocytes (preferred sample type) except for Hexosaminidase to rule out I-cell disease**

### Applicable to:

- Alpha-galactosidase (serum only)
- Alpha-N-Acetylglucosaminidase, (serum or plasma)

### Minimum Specimen Requirements

Serum: 2mL whole blood (Red Top Tube)

Plasma: 3-5mL whole blood Heparinized Green top tube (No gel-separator tubes)

### Storage/Transportation

Ship sample at room temperature. Do not refrigerate or freeze.

### Special Requirements

Specimen should be drawn early enough in the day to arrive in the Metabolic Diseases laboratory by 1:00 p.m. on day of collection, Monday to Friday except statutory holidays.  
Consult the laboratory if other arrangements are necessary.

## Specimen Collection Guide -- Tay-Sachs Carrier Testing-Enzyme Activity

### Minimum Specimen Requirements

- For all patients: 5mL red top (clotted) tube **AND** 5-7mL green top (lithium or sodium heparin, no gel-separator) tube **or** frozen prepared leukocytes
- Exception: Pregnant females require only 5-7mL green top (lithium or sodium heparin, no gel-separator) tube **or** frozen prepared leukocytes

### Storage/Transportation

Whole blood- Ship sample at room temperature. Do not refrigerate or freeze.

### Special Requirements

Please specify on the requisition if female patients are pregnant or on oral contraceptives. Complete the "Carrier Screening: Tay Sachs Enzyme Testing & Ashkenazi Jewish Molecular Panel" requisition.

## Tests Currently Unavailable

- Arylsulfatase B enzyme activity, WBC
- Iduronate sulfatase enzyme activity, Serum
- Iduronate sulfatase enzyme activity, Fibroblast
- N-acetylglucosamine-6-sulfate sulfatase enzyme activity, Fibroblasts (Sanfilippo syndrome type D)