

## **CYTOGENETIC STUDIES**

### **ROUTINE AND HIGH RESOLUTION BLOOD**

Collect 4-6 mls of peripheral blood in a green top sodium heparin tube. Please do not use lithium heparin or ammonium heparin as these can be toxic to the cells. Transport the blood at room temperature for delivery to the lab within 24-48 hours. In winter and summer months, protect the specimen from extremes of temperature.

### **AMNIOTIC FLUID**

Optimally 20-30 ccs of amniotic fluid should be submitted in 2-3 sterile tubes. Transport at room temperature for delivery within 24 hours. In winter and summer months, protect the specimen from extremes of temperature.

### **BONE MARROW/LEUKEMIC BLOOD**

Collect 3-5 mls of bone marrow or 4-6 mls of peripheral blood in a green top sodium heparin tube. Please do not use lithium heparin or ammonium heparin as these can be toxic to the cells. Transport the blood at room temperature for delivery to the lab within 24-48 hours. In winter and summer months, protect the specimen from extremes of temperature.

### **SOLID TISSUE/POC/SKIN BIOPSY**

Place specimen in sterile vial of transport media, which can be obtained from our center. Alternatively, the specimen can be placed in sterile saline, although cell growth may be compromised. Same day delivery at room temperature or within 24 hours delivery at refrigerated temperature. Do not freeze.

### **FISH STUDIES**

Can be performed on each of the samples above.

## **MOLECULAR (DNA) STUDIES**

### **PERIPHERAL BLOOD**

Collect 1-3 ml for newborns or 3-5 ml for adults of venous blood in a purple top (EDTA) using sterile techniques and deliver at room temperature. The sample should be delivered within 24-48 hours.

### **BONE MARROW/LEUKEMIC BLOOD**

Collect 3-5 mls of bone marrow or 4-6 mls of peripheral blood in a green top sodium heparin tube. Please do not use lithium heparin or ammonium heparin as these can be toxic to the cells. Transport the blood at room temperature for delivery to the lab within 24-48 hours. In winter and summer months, protect the specimen from extremes of temperature.

# METABOLIC STUDIES

## **COLLECTION OF PLASMA**

**(For analysis of amino acids, biotinidase and transferrin isoelectric focusing)**

Collect 1 to 3 ml of whole blood or .5 to 2 ml of plasma from a green top (lithium or sodium heparin) tube by venipuncture. The specimen should be placed on ice within one hour and should be received in the laboratory within eight hours of collection. If samples must be stored for a longer period before delivery then the blood should be centrifuged at 1,700 rpm for 5 minutes at 2-10°C, and plasma removed and frozen at -20±5°C. Frozen samples must be sent to the laboratory via courier or overnight express on dry ice.

## **COLLECTION OF SERUM**

**(For analysis of amino acids and transferrin isoelectric focusing)**

Collect 1 to 3 ml of whole blood or .5 to 2 ml of plasma from a red top (no treatment) tube by venipuncture. The specimen should be placed on ice within one hour and should be received in the laboratory within eight hours of collection. If samples must be stored for a longer period before delivery then the blood should be centrifuged at 1,700 rpm for 5 minutes at 2-10°C, and plasma removed and frozen at -20±5°C. Frozen samples must be sent to the laboratory via courier or overnight express on dry ice.

## **COLLECTION OF WHOLE BLOOD FOR RED CELL ASSAYS**

**(For galactose-1-phosphate uridylyltransferase and galactose-1-phosphate)**

Collect 3-7 ml of lithium or sodium heparinized whole blood via venipuncture. If not delivered within 2 hours after being drawn, refrigerate and deliver to the laboratory on wet ice. Do not allow specimen to freeze. The blood should be delivered via courier or overnight express within 24 hours of being drawn. If this is not possible, the lab should be contacted prior to collection.

## **COLLECTION OF URINE**

**(For analysis of amino acids and organic acids)**

Collect 3-10 ml of random catch urine in a clean container. Ideally, the urine should be refrigerated at 4±2 °C and delivered to the laboratory the same day as collection. If the sample must be stored for a longer period before delivery, it should be frozen at -20±5 °C. Frozen samples must be sent to the laboratory via courier or overnight express on dry ice.

## **COLLECTION OF CEREBROSPINAL FLUID**

**(For analysis of amino acids)**

Collect 0.5 to 1 ml of cerebrospinal fluid in a clean tube. It should be drawn within an hour of the plasma or serum amino acid. Sample should be placed on ice and delivered to the laboratory within 24 hours after collection. If the sample must be stored for longer than 12 hours, it should be frozen at -20±5°C. Frozen samples must be sent to the laboratory via courier or overnight express on dry ice.

- **We accept routine samples M-F during normal university operating hours. Contact the laboratory director for consideration of off-hour specimens.**
- **A completed lab requisition must accompany each sample.**
- **Samples can be shipped to: 1430 Tulane Ave Rm 5301 Attn: Biochemical Genetics, New Orleans, La 70112**