

SPECIMEN SHIPPING INSTRUCTIONS  
HUMAN GENETICS PROGRAM/HAYWARD GENETICS CENTER  
REVISED JANUARY, 1999

**METABOLIC STUDIES**

The clinical information provided should include patient name, date of birth, referring physician's name and address, date and time obtained, medication, and dietary information.

**CEREBRAL SPINAL FLUID (CSF):** For a CSF/plasma glycine ratio, the CSF and the plasma should be drawn at the same time. Obtain 0.5-1 ml of CSF in a sterile container. CSF should not be contaminated with blood. It should be frozen and sent to the lab on dry ice if not received by us the same day it is drawn.

**PLASMA:** For amino acids and carnitine obtain 1-3 ml blood in a sodium heparinized tube (green top). Keep the blood refrigerated. If the specimen cannot be delivered within 24 hours, the plasma should be separated, frozen, and sent to the laboratory on dry ice. For plasma phenylalanine and tyrosine a smaller quantity of plasma is required for this assay, thus 0.5 ml of blood obtained from a finger or heel stick into heparinized microfuge tubes are acceptable. Please note that excessive squeezing of the finger or heel results in hemolyzed specimens which are unacceptable.

**URINE:** For urine amino acids, organic acids, carnitine, and thin layer chromatography obtain 5-10 ml urine (15-20 ml for mucopolysaccharides) in a clean container. The container need not be sterile but should be new and not previously used for storage. Refrigerate sample if not delivered immediately. If not delivered within 24 hours, the sample should be frozen. The specimen can be kept a room temperature for several hours during transport. If transport will take longer than several hours, please send on dry ice.

**ENZYME STUDIES**

**SERUM:** For Iduronate Sulfatase obtain 2-3 ml blood in a red top tube. The sample should be refrigerated and delivered within 6 hours. If the specimen cannot be submitted within 6 hours, the serum should be separated, frozen, and sent to the laboratory on dry ice.

**PLASMA:** For  $\alpha$ -Hexosaminidase (except for women who are pregnant or on birth control pills) and Biotinidase obtain 2-3 ml blood in a green top tube. The sample should be refrigerated and delivered to the laboratory within 6 hours. If the specimen cannot be delivered within 6 hours, the plasma should be separated, frozen, and sent to the laboratory on dry ice.

**WHITE BLOOD CELL PELLET:** For all lysosomal enzymes (except Iduronate Sulfatase) obtain a minimum of 10-12 ml blood in a heparinized tube (large green top or 4-5 small green top tubes). The sample should be delivered to the laboratory on the day it is obtained (before 4 PM). Please call the laboratory if it cannot be delivered on the same day.

**GALACTOSE-1-PHOSPHATE URIDYLTRANSFERASE:** Heparinized blood in a green top tube (1-2 ml) may be submitted for this assay; the specimen should be kept refrigerated (not frozen) until delivered to our laboratory. Due to the marked instability of this enzyme, the assay must be performed within 24-36 hours after the specimen is obtained. Please contact our laboratory before submitting the specimen so that arrangements can be made.

\*STAT determination on the above studies within 24 hours is available upon confirmation with the laboratory.

**CYTOGENETICS STUDIES**

**METAPHASE (ROUTINE) AND LEUKEMIC BLOOD STUDIES:** Sterile heparinized vials are available upon request, and should be refrigerated until the day they are to be used. If vials are not available, a sterile tube with sodium heparin may be used (such as a small green top vacutainer tube). Collect 3-5 ml of venous blood using sterile technique, place in vial and deliver container at room temperature. The sample should be delivered within 24 to 36 hours.

**PROMETAPHASE AND CHROMOSOMAL BREAKAGE STUDIES:** Collect 4-6 ml of venous blood and submit as described above. If possible, the sample should be sent on Monday, Thursday or Friday. Receiving specimens in the laboratory on these days enables the optimal synchronization of the cultures.

**PROMETAPHASE AND FRAGILE X (MOLECULAR) STUDIES:** Collect 4-6 ml of venous blood in a sodium heparinized vial and 3-5 ml of venous blood in a purple top tube (EDTA). If possible, the sample should be sent on Monday, Thursday or Friday. Receiving specimens in the laboratory on these days enables the optimal synchronization of the cultures.

**AMNIOTIC FLUID:** Optimally, 20-30 ml of amniotic fluid should be submitted, although growth has been achieved with a smaller amount. Sterile technique is essential. Sterile tubes should be used. Dark glass is unnecessary. All fluids should be sent at room temperature and received in the laboratory on the same day obtained. If this is not possible, overnight delivery should be used.

**BONE MARROW:** Obtain 1½-2 ml of marrow should be added to the heparinized vials and delivered in the same fashion as venous blood.

**SOLID TISSUE:** Sterile vials containing media can be obtained from our Center. If not available, the specimen may be placed in sterile saline, although successful growth might be diminished. Deliver at refrigerated temperature (not frozen) within 24 hours.

**CHORIONIC VILLI SAMPLE:** CVS is received by previous notification. Special transporting media is available from our laboratory.

**FISH STUDIES**

Collect 3-5 ml of venous blood in a sodium heparinized vial using sterile technique, place in vial and deliver container at room temperature. The sample should be delivered within 24 to 36 hours.

**MOLECULAR (DNA) STUDIES**

Collect 3-5 ml of venous blood in a purple top tube (EDTA) using sterile technique, place in vial and deliver container at room temperature. The sample should be delivered within 24 to 36 hours.