CYTOGENETIC STUDIES

ROUTINE AND HIGH RESOLUTION BLOOD

Collect 4-6 mls of peripheral blood in a green top sodium heparin tube. Please do not used 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 used 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

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

METABOLIC STUDIES

COLLECTION OF PLASMA OR SERUM
(For Analysis of Quantitative Amino Acids, and Biotinidase)

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^{o}C, and plasma removed and frozen at -20$\pm$5^{o}C. Frozen samples must be sent to the laboratory via courier or overnight express on dry ice.

COLLECTION OF PLASMA OR SERUM
(For Transferrin Isoelectric Focusing for Congenital Disorders of Glycosylation Screening)

Collect 1 to 3 ml of whole blood or .5 to 2 ml of plasma from a green top (lithium or sodium heparin) tube or 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^{o}C, and plasma removed and frozen at -20$\pm$5^{o}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-P Uridyltransferase 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, Organic Acids, and Thin Layer Chromatography Procedures).

Collect 3-10 ml of random catch urine in a clean container (**a minimum of 10 ml is required for mucopolysaccharide chromatography)**. Ideally, the urine should be refrigerated at 4$\pm$2 ^{o}C and delivered to the laboratory the same day as collection. If the sample must be stored for a longer period before delivery, it should be frozen at -20$\pm$5 ^{o}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 place 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$\pm$5^{o}C. Frozen samples must be sent to the laboratory via courier or overnight express on dry ice.

- **WE ACCEPT SPECIMENS M-F. They must be received in the lab by 4:30 pm.**
- **A completed lab requisition must accompany each sample.**
- **They can be shipped to: 1430 Tulane Ave Rm 5301 Attn: Biochemical Genetics, New Orleans, La 70112**

Hayward Genetics Center

Deliver specimens to: 1430 Tulane Ave.,
Room 5550 New Orleans, LA 70112
Phone: 504-988-5229: Fax 504-988-1763
Biochemical Genetics beeper: 504-501-6011
Molecular/Cytogenetics beeper: 504-501-6096

REPORTS TO:
_____
_____
_____
FAX TO: _____
PHONE#: _____
BILLING ADDRESS:
_____
_____
_____
_____

PATIENT LAST NAME FIRST NAME

DATE OF BIRTH □MALE □FEMALE HOSPITAL NAME//PATIENT#

DATE COLLECTED PHYSICIAN

DIAGNOSIS/CLINICAL INFORMATION/INDICATION FOR STUDY

CURRENT MEDICATIONS

SPECIMEN TYPE

Cytogenetics	Biochemical genetics	Molecular Genetics
□Blood	□Blood	□Blood
□Amniotic fluid	□Urine	□Other_____
□Fibroblasts	□Other_____	
□Products of Conception		
□Bone marrow (WBC Count: _____)		
□Leukemic blood (WBC Count: _____)		
□Tissue		
□Paraffin-embedded tissue		
□Other_____		

CYTOGENETICS

PRENATAL STUDIES (Circle One)
*Gestational Age_____ wks by LMP ultrasound
□Acetylcholinesterase
□Alpha-fetoprotein
□FISH for Aneuploidy -13,-18,-21,X,Y
□Chromosome analysis
□Other_____

PERIPHERAL BLOOD STUDIES
□Routine chromosome analysis
□High resolution chromosome analysis

CANCER CYTOGENETIC STUDIES
□Chromosome analysis

OTHER _____

MOLECULAR GENETICS

FLUORESCENT IN SITU HYBRIDIZATION (CANCER)

□AML Panel	□MDS Panel
□ALL Panel	□CLL Panel
□MM Panel	□NHL Panel

□Acute Promyelocytic Leukemia t(15;17)
□Chronic Myelogenous Leukemia (CML) t(9;22)
B-Cell Lymphoma
□Burkitt Lymphoma Panel MYC/IGH, IGL, IGK
□Double-hit Lymphoma C-MYC, IGH/BCL2, BCL6
□Follicular IGH/BCL2 t(14;18)
□Mantle Cell Lymphoma CCND1/IGH t(11:14)
□Non-Hodgkin Lymphoma (NHL) BCL6, ATM, IGH, p53
□XX/XY Sex Mismatch Transplant

OTHER FISH STUDY
Please specify_____

FLUORESCENT IN SITU HYBRIDIZATION (CONSTI.)

Syndrome	Structural Abberation
□Cri-du-chat	del(5)(p15)
□DiGeorge	del(22)(q11.2)
□Kallman	del(X)(p22.3)
□Miller Dieker	del(17)(p13.3)
□Prader-Willi/Angelman	del(15)(q11-q13)
□Smith-Magenis	del(17(p11.2)
□Williams	del(7)(q11.23)
□X-linked Ichthyosis	del(X)(p22.3)

OTHER FISH STUDY Please specify_____

MICROARRAY CGH (aCGH)
□Whole Genome aCGH

DNA-BASED STUDIES
□Fragile X
□Methylation (Prader-Willi/Angelman)
□Y-Chromosome Testing
□Galactosemia 2 mutation panel
□Cystic Fibrosis 46 mutations*(Ethnicity_____)
*Informed Consent required

□FLT3 mutation	□JAK2 mutation
□c-Kit	□MECP2 (Rett Syndrome)
□NPM1	□Connexin 26 Sequencing

OTHER DNA STUDY Please specify_____

BIOCHEMICAL GENETICS

QUANTITATIVE AMINO ACIDS
□Plasma □Urine □CSF

ORGANIC ACIDS
□Urine Organic acids (GC/MS)

THIN LAYER CHROMATOGRAPHY
□Mucopolysaccharides
□Mono and Di-Sugars

ENZYME STUDIES/ OTHER METABOLICS

Disease	Assay
□Biotinidase Deficiency	Biotinidase
□Galactosemia (RBCs)	Galactose-1-Phosphate Uridyltransferase(GALT)
□Galactosemia monitoring	Galactose-1-Phosphate(G-1-P)

□Galactosemia Screening Panel (GALT, G-1-P, Mutation panel)
□Congenital Disorders of Glycosylation Screening by Transferrin Isoelectric Focusing (plasma)