

Cytogenetics Laboratory Sample Requirements

Test	Specimen Type	Specimen Volume	Collection Tube	Estimated Turnaround Time	Delivery Window	Unacceptable Specimen
Chromosome Karyotype Chromosome Analysis	PB-Newborn	1-2 ml	Dark Green-top Tube (Sodium Heparin)	Preliminary Result: 3 days Final Report: 7 days	*Up to 48 hours after collection. *But ASAP. *PB, LB, and BM should be stored at room temperature. *POC, CVS, AF, and Biopsy. should be refrigerated and shipped with ice packs.	Incorrect tube type, Leaking tube, Unlabeled tube, Improper labeled tube
	PB	3-5 ml	Dark Green-top Tube (Sodium Heparin)	7-10 days		
	PB/LB-Heme	3-5 ml	Dark Green-top Tube (Sodium Heparin)	5-7 days		
	BM	3-5 ml	Dark Green-top Tube (Sodium Heparin)	5-7 days		
	POC	0.3-1 cm ³ <u>Preferred Specimen Type:</u> -Placenta contains Chorionic Villi -Fetal biopsy under arm or inner thigh	Sterile centrifuge tube with balanced salt solution	9-14 days		Fixed (formalin, formaldehyde or alcohol) or frozen specimen
	CVS	0.3-1 cm ³	Sterile centrifuge tube with balanced salt solution	9-14 days		
	AF	20 ml	Sterile centrifuge tube	9-14 days		
	Biopsy	0.3-1 cm ³	Sterile centrifuge tube with balanced salt solution	9-14 days		
FISH	PB-Newborn	1-2 ml	Dark Green-top Tube (Sodium Heparin)	Direct FISH: 1-2 days Regular: 3-5 days	*Up to 48 hours after collection. *But ASAP. *PB, LB, and BM should be stored at room temperature. *POC, CVS, AF, and Biopsy. should be refrigerated and shipped with ice packs.	Incorrect tube type, Leaking tube, Unlabeled tube, Improper labeled tube
	PB	3-5 ml	Dark Green-top Tube (Sodium Heparin)	Direct FISH:1-2 days Regular: 5-7 days		
	PB/LB-Heme	3-5 ml	Dark Green-top Tube (Sodium Heparin)	STAT: 1-3 days Regular: 3-5 days		
	BM	3-5 ml	Dark Green-top Tube (Sodium Heparin)	STAT: 1-3 days Regular: 3-5 days		Fixed (formalin, formaldehyde or alcohol) or frozen specimen
	FFPE	1 slide per FISH probe, plus 1 control tonsil slide	N/A	5-7 days		
	POC	0.3-1 cm ³ <u>Preferred Specimen Type:</u> -Placenta contains Chorionic Villi -Fetus-biopsy under arm or inner thigh	Sterile centrifuge tube with balanced salt solution	9-14 days		
	CVS	0.3-1 cm ³	Sterile centrifuge tube with balanced salt solution	Direct FISH: 1-2 days		
	AF	10-15 ml	Sterile centrifuge tube	Direct FISH: 1-2 days		
	Biopsy	0.3-1 cm ³	Sterile centrifuge tube with balanced salt solution	9-14 days		

Note:

- In some of the cases, extra time should be allowed for additional confirmation.
- Direct FISH only analyzes interphase cells, which is suitable for detecting aneuploidy. However, it is not recommended for diagnosing microdeletion or microduplication disorders, which need FISH analyzing on metaphase cells
- If unacceptable specimen is received, contact the lab director.
- Turnaround Time: The number of days from the date of pickup of a specimen to the date of result released
- Abbreviation: BM-bone marrow, PB-peripheral blood, LB-leukemic blood, AF-amniotic fluid, CVS-chorionic villus sampling, POC-products of conception, FISH-fluorescent in situ hybridization, FFPE-formalin-fixed paraffin-embedded

Molecular Lab Sample Requirements

Category	Test	Specimen Type	Specimen Volume	Collection Tubes	Estimated Turnaround Time		Delivery Window
Constitutional	CGH	Peripheral blood (PB) Product of conceptions (POC)	PB: 3-5 ml (Infant PB: 1-2 ml) POC: 5cm x 5cm x 5cm	Lavender/Purple-top (EDTA) tube; Tissue or cells in culture medium, PBS or saline.	Non-Newborn Samples: 14-21 Days	Newborn Samples: 7 Days	PB and BM: up to 48 hours after collection. POC: up to 36 hours after collection, but ASAP.
	Fragile X	Peripheral blood (PB)	PB: 3-5 ml (Infant PB: 1-2ml)	Lavender/purple-top (EDTA) tube			
	Methylation						
Cancer	NGS assays (NGSHM & NGSHL)	Peripheral blood (PB) Bone marrow (BM)	PB: 3-5 ml BM: 2-4 ml	Lavender/purple-top (EDTA) tube	14-21 Days		Samples should be refrigerated before send out. Ice packs can be used in the shipping package.
	FLT3				7 Days		
	JAK2				7 Days		
	NPM1				7 Days		

NOTE:

- All specimens will be checked for qualification for the test. Samples with QC failures will not be used for the test. If repetitive QC failure was experienced from all the received tubes, a new collection of samples will be requested. If a sample is only available less than required volume but still possible to have a qualified nucleic acid extraction, offer an option to accept the order for sample QC, but remind a higher chance of test cancellation due to insufficient sample for the ordering staff to decide. If further question is needed, please contact the lab director.
- The listed turnaround times are estimated on a smooth process from sample delivery to test report. Extensive time may be needed if anything happens during this period.
- Please note that patients undergoing chemotherapy will have a reduced white blood cell count, and therefore lower DNA yields. If possible, please retrieve at least double the recommended sample to ensure enough DNA is available for testing.

METABOLIC STUDIES

COLLECTION OF PLASMA

(For analysis of amino acids)

Collect 1 to 3 ml of whole blood or .5 to 2 ml of plasma from a green top (lithium or sodium heparin) tube. Centrifuge the blood and separate the plasma from the cells as soon as possible after collection. Avoid disturbing the buffy coat, which contains the white blood cells. Put the plasma into a tube without additives, such as a red-top or white top tube. Freeze the plasma below -20°C. Transport the plasma frozen.

COLLECTION OF WHOLE BLOOD

(For analysis of amino acids – the laboratory will separate the plasma)

Collect 1-3 ml of whole blood in a sodium or lithium heparin tube (green top). Transport the blood at room temperature for delivery to the lab within 24 hours of collection. Protect the specimen from extremes of temperature.

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 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 below -20°C. Transport the urine frozen.

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 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 a longer period of time, it should be frozen at below -20°C. Transport the specimen CSF specimen frozen.

- **A completed lab requisition form must accompany each sample**
- **Routine samples are accepted Monday through Friday from 8:30 am to 5 pm.**
- **Samples can be shipped to:**
 - 1430 Tulane Ave Rm 5301**
 - Attn: Biochemical Genetics Laboratory**
 - New Orleans, LA 70112**
- **Off-hours, weekend, and holiday samples may be delivered by courier to the monitored dropbox located outside of the Biochemical Genetics Laboratory located inside the Tulane School of Medicine at:**
 - 1430 Tulane Ave Rm 5301**
 - New Orleans, LA 70112**
- **Please contact the laboratory director for shipment of weekend and holiday samples.**