

Specimen Requirements and Submission Guidelines

Analysis	Specimen Type	Recommended Volume and Container*	Transport Temperature	Specimen Stability**	Turnaround Time	
DNA (Molecular) Analysis	Blood	Any single disease/gene	Room Temperature	7 days	Approx. 14 days	
		Gene Panel			Approx. 4 weeks	
		WES/WGS			Approx. 12 weeks	
		Carrier Screening or other Gene Panel combined with Tay-Sachs enzyme			Approx. 12-14 days	
	Saliva	(unacceptable for Tay-Sachs enzyme analysis)	Oragene Saliva Collection (for molecular assays ONLY)			Test dependent
	Chorionic Villi	5-10 mg in conical tube with sterile saline OR transport media		Room Temperature	72 hours	Approx. 10-14 days
	Amniotic Fluid	10 ml in conical tube				
	Cultured Cells	2 confluent T-25 flasks				
<ul style="list-style-type: none"> • 5-10 ml EDTA (lavender-top) tube of maternal blood required to perform MCC studies on all prenatal samples • 5-10 ml EDTA (lavender-top) tube of paternal blood required for all prenatal samples where paternal genetic testing was performed outside of Sema4™. 						
Lysosomal Enzyme Analysis	Blood	2 ACD (yellow-top) OR EDTA (lavender-top) 5-10 ml tubes	Room Temperature	72 hours	Approx. 7 days	
	• Sample must arrive at the testing laboratory within 72 hrs after collection to ensure sample integrity					
Other Biochemical Analyses	Dried Blood Spots	2 completely saturated spots	Room Temperature	60 days	Approx. 3-5 days * STAT tests for metabolic emergencies available within 24 hr with prior approval from the laboratory	
	Plasma	0.5-1 ml	Frozen	72 hours		
	<ul style="list-style-type: none"> • For amino acids analysis, fasting or 2-4 hour postprandial sample is recommended • For Tamoxifen and Porphyrin metabolite analyses, specimen needs to be protected from light 					
	Cerebrospinal Fluid	0.5-1 ml	Frozen	72 hours		
	• Samples submitted for the evaluation of glycine encephalopathy should be accompanied by a simultaneously obtained plasma sample for calculation of CSF glycine/Plasma glycine ratio					
	Urine	5-10 ml in clean container without any preservatives	Frozen	72 hours		
	<ul style="list-style-type: none"> • For organic acids analysis, collect specimen during the time of acute illness as abnormal metabolite levels often decrease, sometimes to near normal concentrations, when the patient is well • For Porphyrin metabolite analyses, specimen needs to be protected from light • For prenatal Tay Sachs enzyme testing, contact our laboratory genetic counselor at 212-241-2537 for sample requirements 					
Chromosome and FISH (Cytogenetic) Analysis	Blood	5-10 ml in Sodium heparin (green-top) tube	Room Temperature	72 hours	Approx. 4-10 days *Aneuvysion FISH (13,18,21,X&Y) 6-48 hrs	
	CVS/POC	5-10 mg in conical tube with transport media OR cell culture media				
	Amniotic Fluid	20 ml in conical tube				
Array CGH Analysis (Chromosomal Microarray)	Blood	5-10 ml in EDTA (lavender-top) tubes AND 5-10 ml in Sodium heparin (green-top) tube	Room Temperature	72 hours	Approx. 5-14 days	
	CVS/POC	5-10 mg in conical tube with transport media OR cell culture media				
	Amniotic Fluid	10-15 ml in conical tube				
	Cultured Cells	2 confluent T-25 flasks				
	<ul style="list-style-type: none"> • 5-10 ml EDTA (lavender-top) tube of maternal blood required to perform MCC studies on all prenatal samples • Requested volume of prenatal specimens for Array CGH testing is IN ADDITION to necessary volume for other cytogenetic, biochemical or molecular analyses. 					

The following must be included with each sample:

- Completed and signed **Test Requisition** form.
- Completed and signed **Informed Consent** form.
- Billing information or payment (include copy of insurance card).
- Indication for testing, patient's family history, ethnic background and prior relevant test results.

▲ Samples must arrive between 9am and 7pm ET (Monday to Friday) or between 9am and 4pm ET (Saturday).

▲ Please call in advance for all prenatal Molecular and Biochemical specimens.

* Please contact us if you have questions about alternative acceptable specimen types for any test.

** Stability from the time of specimen collection to testing.

Sample transport:

- Call our lab at **800-298-6470** to schedule a sample pickup.
- Send via FedEx or courier for same day or overnight (morning) delivery to: **Sema4**
1428 Madison Avenue, Atran Building, Room 2-25
New York, NY 10029