

Specimen requirements

Guidelines for collection and shipment



a Mount Sinai venture

Specimen guidelines

DNA Analysis (Molecular)

Analysis	Specimen Type	Specimen Requirements	Transport Temperature	Specimen Stability*	Turnaround Time [†]
Single gene, disease, or diagnostic gene panel (including targeted analyses and family studies)	Blood	Two 5-10 mL EDTA tubes (lavender top), or Two 5-10 mL ACD tubes (yellow top), or Minimum 1 mL per collect for pediatric samples	Room temperature	7 days	Carrier screening: 2 weeks Diagnostic testing: 4 weeks
	Saliva	Oragene-DNA saliva collection kit	Room temperature	60 days	Carrier screening: 2 weeks Diagnostic testing: 4 weeks
	Cheek swab [‡]	ORACollect for Pediatrics cheek swab kit	Room temperature	60 days	Carrier screening: 2 weeks Diagnostic testing: 4 weeks
Carrier screening with or without Tay Sachs enzyme [§]	Blood	Two 5-10 mL EDTA tubes (lavender top), and One 5-10 mL ACD tube (yellow top)	Room temperature	≤5 days	12-14 days
	Saliva	Oragene-DNA saliva collection kit	Room temperature	60 days	10-14 days
Whole genome sequencing or whole exome sequencing	Blood	Two 5-10 mL EDTA tubes (lavender top), or Two 5-10 mL ACD tubes (yellow top) for adults and Minimum 1 mL per collect for pediatric samples	Room temperature	7 days	12 weeks
	Blood	One 5-10 mL EDTA tube for the child and One 5-10 mL EDTA tube for each parent	Room temperature	7 days	10-14 day
Natalis	Saliva	Oragene-DNA saliva collection kits for child and for each parent	Room temperature	60 days	10-14 days
	Cheek swab	ORACollect for Pediatrics cheek swab kits for child and for each parent	Room temperature	60 days	10-14 days
	Blood	Two 10 mL Streck BCT tubes of maternal blood Patient must be at least 9 weeks gestation	Room temperature	5 days	1 week
Prenatal diagnostic testing	Chorionic villi	5-10 mg in conical tube with sterile saline or transport media	Room temperature	72 hours	14 days
	Amniotic fluid	10 mL in conical tube	Room temperature	72 hours	14 days
	Cultured cells	Two confluent T-25 flasks	Room temperature	72 hours	14 days

- One 5-10 mL EDTA tube (lavender top) of maternal blood is required for diagnostic testing, prenatal samples, maternal cell contamination (MCC) studies, and mutation confirmation (as needed). Paternal blood is required for diagnostic testing and mutation confirmation (as needed)
- Please contact our lab for all prenatal specimens
- Please contact our lab for alternate specimen types, such as semen, extracted DNA, products of conception, or other tissue types

Chromosome and FISH Analysis (Cytogenetic)

Specimen Type	Specimen Requirements	Transport Temperature	Specimen Stability*	Turnaround Time [†]	Notes
Blood	One 5-10 mL sodium heparin tube (green top)	Room temperature Do not freeze	72 hours	4-10 days	
Chorionic villi	5-10 mg in conical tube with transport media If >24 hours, place in cell culture media	Room temperature Do not freeze	24-48 hours	5-10 days	• Turnaround time is 24-48 hours for AneuVysion FISH (13, 18, 21, X, and Y)
Product of conception	In conical tube with transport media, or Other sterile screw top container with 50 mL transport media	Room temperature Do not freeze	24-48 hours	Please contact the lab	• Chromosome analysis is not available for formalin-fixed, paraffin-embedded specimens
Amniotic fluid	20 mL in conical tube	Room temperature Do not freeze	48-72 hours	7-10 days	• Turnaround time is 24-48 hours for AneuVysion FISH (13, 18, 21, X, and Y)

- Please contact our lab for alternate specimen types
- Oncology specimens are not accepted for cytogenetic testing
- Specimens received outside of the stability window will be processed. Please contact the lab if a delay is anticipated

*Stability from the time of specimen collection to testing.

[†]Expected turnaround time.

[‡]Not all testing is validated on cheek swabs.

[§]Tay-Sachs enzyme testing is not performed on saliva specimens.

ACD=Acid citrate dextrose; CGH=Comparative genomic hybridization; EDTA=Ethylenediaminetetraacetic acid; FISH=Fluorescent in situ hybridization

Biochemical Analyses

Specimen Type	Specimen Requirements	Transport Temperature	Specimen Stability*	Turnaround Time [†]	Notes
Blood	Two 5-10 mL EDTA tubes (lavender top), or Two 5-10 mL ACD tubes (yellow top), or Two 5-10 mL sodium heparin or lithium heparin tubes (green top), or Minimum 3 mL per collect for pediatric samples	Room temperature	72-96 hours	7 days	<ul style="list-style-type: none"> Sample must arrive at the lab within 72 hours following collection Sodium heparin or lithium heparin tube is required for Fabry enzyme testing
Plasma	Preferred: 0.5-1 mL frozen plasma from sodium heparin tube (green top) Accepted: 0.5-1 mL frozen plasma from EDTA tube (lavender top)	Frozen	2-8 weeks	3-5 days	<ul style="list-style-type: none"> For amino acids analysis, fasting or 2-4 hour postprandial sample is recommended For tamoxifen and porphyria metabolite analyses, specimen needs to be protected from light
Urine	5-10 mL in clean container without any preservatives	Frozen	Frozen: 6 months Refrigerated: 2 weeks	3-5 days	<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 porphyria metabolite analyses, specimen needs to be protected from light
Dried blood spots	Two completely saturated spots	Room temperature	60 days	3-5 days	
Cerebrospinal fluid (CSF)	0.5-1 mL	Frozen	Frozen: 2 weeks Refrigerated: 72 hours	3-5 days	<ul style="list-style-type: none"> 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

- For prenatal Tay-Sachs or Sandhoff enzyme testing, contact a Sema4 genetic counselor at [800-298-6470](tel:800-298-6470) in advance for sample requirements
- STAT tests for metabolic emergencies are available within 24 hours with prior approval from the lab

Array CGH Analysis (Chromosomal Microarray)

Specimen Type	Specimen Requirements	Transport Temperature	Specimen Stability*	Turnaround Time [†]	Notes
Blood	One 5-10 mL EDTA tube (lavender top), and One 5-10 mL sodium heparin tube (green top)	Room temperature	72 hours	5-14 days	
Chorionic villi	5-10 mg in conical tube with transport media If >24 hours, place in cell culture media	Room temperature	24-48 hours	5-14 days	
Product of conception	In conical tube with transport media, or Other sterile screw top container with 50 mL transport media	Room temperature	24-48 hours	5-14 days	
Amniotic fluid	20 mL in conical tube	Room temperature	48-72 hours	5-14 days	
Cultured cells	Two confluent T-25 flasks	Room temperature	—	5-14 days	

- One 5-10 mL EDTA tube (lavender top) of maternal blood is required to perform maternal cell contamination (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
- Please allow additional time for specimens that require culturing

Additional guidelines

Please include the following with every sample

- ✓ Completed and signed test requisition form
- ✓ Completed and signed informed consent
- ✓ Two patient identifiers on each specimen collection tube
- ✓ Billing or payment information, including a copy of the patient's insurance card (if applicable)
- ✓ Indication for testing
- ✓ Any prior test results relevant to testing
Information about the patient's family history and ethnic background



To ensure proper processing and test performance, please label all specimens and complete all paperwork.

Specimens may be rejected if

- ✗ There are any discrepancies between the specimen label and the requisition form
- ✗ There are missing identifiers, such as first name, last name, or date of birth
- ✗ An inappropriate specimen type is received, such as a specimen collected in an incorrect container
- ✗ The specimen collection container is broken, damaged, or expired
- ✗ The specimen collection container is leaking
- ✗ Specimen quality is otherwise compromised or contaminated

Additionally, specimens for noninvasive prenatal tests may be rejected if

- ✗ Specimen volume is less than 6 mL
- ✗ The specimen is older than 5 days
- ✗ The blood specimen is hemolyzed
- ✗ The specimen was not shipped at ambient temperature

All samples should be shipped same day or overnight to

Sema4
1428 Madison Avenue
Atran Building, Room 2-25
New York, NY 10029

Samples must arrive 9 AM – 7 PM ET (Monday to Friday) or 9 AM - 4 PM ET (Saturday).

For practices located in the New York City area, we work with local courier services to transport specimens to our lab for quick processing. To confirm that you are located within our service area or to arrange a pick up, please contact our lab at [800-298-6470](tel:800-298-6470).