

Specimen Requirements

Please visit ambrygen.com/specimen-requirements for details about submitting blood spots, bone marrow and semen samples.

GENERAL SPECIMEN REQUIREMENTS					
Please label each specimen and Test Requisition Form with at least 2 unique identifiers specific to patient (full name and DOB/Medical Record Number) and collection date. If patient is deceased, please call Ambry prior to sample submission.					
SPECIMEN	BLOOD (WHOLE/FETAL*/CORD*)	SALIVA***	DNA	FETAL (AMNIO AND CVS)*/ CULTURED CELLS	SOLID TISSUE
CONTAINER	<p>In order of preference:</p> <ol style="list-style-type: none"> EDTA tube (purple) Citric acetate tube (yellow) or Potassium oxalate/Sodium fluoride (grey top) <p>Note: Blood samples not accepted for patients that have undergone allogenic transplant (e.g bone marrow or peripheral stem cell).**</p>	<p>1 Oragene™ self-collection container. 2 containers required for all pediatric saliva testing kits and adults having ExomeNext for family member analysis.</p> <p>Note: Saliva not accepted for all tests - please see "Special Collection Notes" below and website for details.</p>	<p>Sterile plastic tube</p>	<p>Cultured cells (CVS, amniocytes, products of conception, fibroblasts): 2 sterile T25 cell flasks or suitable alternative in sterile culture media</p> <p>Direct CVS tissue: see "Solid Tissue" section for details</p> <p>Note: Please maintain back-up cell culture. Tissue culturing services available through Genesis Laboratories for additional charge. See ambrygen.com/forms for requisition form.</p>	<p>Types include pathology specimens (e.g liver, muscle, heart) and direct CVS tissue</p> <p>Sterile plastic tube or flask filled with sterile culture media</p>
AMOUNT	<p>Adult: 3-5cc Pediatric: 2cc minimum Fetal/Cord blood: 2cc minimum</p> <p>Exceptions: <i>NGS panels and clinical exome sequencing 6-10cc (adult) and 5cc (pediatric) minimum. Chromosomal microarray for newborns: 180K oligo array 0.5-1cc minimum; SNP Array 1cc minimum.</i></p>	<p>1cc of freshly collected saliva in Oragene™ container using its instructions</p> <p>Note: Fill up to black line with 1cc saliva and close lid. Once lid is closed, it automatically adds 1cc of buffer for a total volume of 2cc.</p>	<p>5µg of DNA in TE (10mM Tris-Cl pH 8.0, 1mM EDTA); preferred 100µl at ~50ng/µl concentration for multi-gene/NGS panels. DNA OD 260/280 ratio (preferred 1.7-1.9). Please send agarose picture with high molecular weight genomic DNA, if available.</p> <p>Exceptions: <i>Single Site Analysis minimum 1-2µg DNA (~100ng/µl conc.)</i></p>	<p>Cultured cells: 2 T25 cell flasks (or suitable alternative) at 80% confluence</p> <p>Exception: <i>Clinical exome sequencing requires 3 T25 cell flasks of cultured fibroblasts at 80% confluence</i></p>	<p>Fresh tissue: CVS: 10mg cleaned villi; Pathology specimens: Minimum 20mg - prefer 50mg (minimum size 0.5cm cube, prefer 1-2cm cube)</p> <p>Frozen Tissue: Minimum size is 0.5cm cube, Prefer 1-2cm cube</p>
SPECIAL COLLECTION NOTES	<p>Transfusion patients: Wait at least 2 weeks after a packed cell/platelet transfusion, and at least 4 weeks after a whole blood transfusion prior to blood draw for testing.</p> <p>Chemotherapy patients: DNA quality may be affected if patient has received chemotherapy within the last 120 days. Clients will be contacted to provide additional specimen if DNA quality is insufficient.</p>	<p>Exceptions: Saliva not accepted for Proband ExomeNext/ExomeNext-Rapid</p> <p>PMEFirst, PMENext and CSTB dodecamer expansion</p> <p>Patients that have undergone allogenic transplant (e.g bone marrow or peripheral stem cell).**</p>	<p>DNA should not be submitted if it has undergone multiple freeze/thaw cycles.</p> <p>Transfusion patients: Wait at least 2 weeks after a packed cell/platelet transfusion, and at least 4 weeks after a whole blood transfusion prior to blood draw (to extract DNA) for testing.</p> <p>Chemotherapy patients: DNA quality may be affected if patient has received chemotherapy within the last 120 days. Clients will be contacted to provide additional specimen if DNA quality is insufficient.</p>		

SPECIMEN	BLOOD (WHOLE/FETAL*/CORD*)	SALIVA***	DNA	FETAL (AMNIO AND CVS)*/ CULTURED CELLS	SOLID TISSUE
STORAGE	Room temp at 15-30°C (short-term) Refrigerated at 2-8°C (long-term) DO NOT FREEZE	In sterile bag, room temp at 15-30°C	Room temp at 15-30°C (immediate) Refrigerated at 2-8°C (short-term) Frozen at -30°C (long-term)	Cultured cells: in tissue culture incubator	Fresh tissue: Sterile plastic tube or flask with sterile transport media (e.g tissue culture media) Frozen Tissue: At -80°C and ship on dry ice overnight delivery to arrive Monday-Friday
STABILITY (WHEN STORED PER ABOVE)	Stable for 7 days, refrigerated Stability may be affected if stored at room temp for more than 3 days. Please call Ambry if your sample is more than 7 days old.	Stable up to 1 year	Stable for 3-4 days, room temp Stable for at least 1 year, refrigerated Stable indefinitely, frozen	Stable for up to 72 hours, refrigerated	Fresh tissue: Shipped to arrive to lab within 48 hours of collection. Shipping on a cold pack in Styrofoam may be considered to minimize the risk for extreme temperature exposure. Specimens should not be exposed to temperatures less than -20°C or greater than 30°C. Frozen tissue: Shipped on dry ice overnight to arrive Monday-Friday. Frozen tissue can be used at any point for testing; however, specimens that have gone through freeze/thaw cycles are not acceptable. Specimens should be maintained at minimum -20°C during packaging and shipping.
SHIPPING	Whole blood: Ship ambient for 2-day delivery (preferred) using Ambry Sample Submission kit or similar. Fetal/Cord blood: Please call ahead and ship ambient for next-day delivery on Monday- Friday (required), avoiding arrival on holidays.	Ship room temp for 2-day delivery (preferred) using Ambry Sample Submission kit or similar. Samples may be exposed to temperatures between -20°C to 50°C during collection and transport.	Ship frozen on ice/dry ice (or with cold pack if not available) for next-day delivery (preferred) using Ambry Sample Submission kit or similar	Please call ahead and ship ambient for next-day delivery to arrive Monday-Friday (required) using Ambry Sample Submission kit or simila, avoiding arrival on a holiday.	Fresh Tissue: Room temp and shipped for overnight delivery (preferred using Ambry Sample Submission Kit or similar). Specimens should arrive at the lab within 48 hours of collection. Delivery Monday-Friday is preferred. Frozen Tissue: Flash freeze and ship frozen on dry ice with overnight delivery to arrive Monday-Friday

* Please call Ambry before sending fetal/cord blood specimens. Maternal cell contamination (MCC) analysis required for all fetal/cord blood tests. Please send a maternal sample (using a separate requisition). An additional charge applies.

** Patients with a history of active/recent hematological disease, (e.g. leukemia, lymphoma, or myelodysplastic syndrome) subject to additional review prior to accessioning. Blood specimens in patients with hematological disease may not be accepted in some cases. For these cases and those who have undergone allogenic transplant (e.g. bone marrow or peripheral stem cell transplant), cultured fibroblasts are preferred, though fresh/fresh frozen normal tissue (no fixatives) may be accepted in some cases. Please refer to "Cultured Cells" section for those specimen requirements. **If you are sending fresh/fresh frozen normal tissue samples, please call Ambry to discuss before sample submission.**

*** Please contact Ambry for additional containers.

GENERAL SHIPPING INSTRUCTIONS

Ship samples using the Ambry Sample Submission Kit or suitable packaging:

1. Place up to 5 blood tubes within the protective foam
2. Place entire kit into a Biohazard bag and seal
3. Place Test Requisition/Billing form and any other patient paperwork into the side pouch of the Biohazard bag
4. Place kit into the Diagnostic/Clinical Pak and seal
5. Adhere air/waybill to the Diagnostic/Clinical Pak and ship to address below

Sending more than one sample per Sample Submission Kit is permissible and preferred. Please enclose a completed Test Requisition/Billing Form with each sample. All Test Requisition and Consent form requirements must be met prior to processing. Informed consent (documented on Test Requisition Form or electronic order that a copy is on file) is required for New York clients. Incomplete forms will result in the delay of sample processing.

Note: Please do not use any contents beyond their printed expiration date. Do not transfer samples to other containers. Please contact Ambry for additional containers if needed.