

Specimen Requirements for Cytogenetics Genetics Testing

Sample Type	Code	Collection Container(s)	Collection and Processing Instructions	Shipping Instructions and Preferred Sample Condition
Amniotic Fluid	AF	15ml sterile conical centrifuge tube	20-30ml fluid, discarding the first 1-2 ml. Place in 2-3 specimen tubes.	Preferred gestational age >15 weeks. Must be received in a securely closed, sterile container. Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.
Cultured Amniocytes	CAF	T25 or T75 flask	At least 1- T25 (or T75) with a confluency of 75-80%. Note: Cultures should be < 3 weeks old. Accepted for test codes CMPRE and CMPRS only.	Send overnight at room temperature. Do not refrigerate or freeze.
Bone Marrow	BM	Sodium heparin (green top)	Collect 2-3 ml of bone marrow.	Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.
Chorionic Villi	CV	Sterile container with EGL transport media. If EGL media is not available, other sterile culture media can be used (RPMI, Hanks Solution).	≥ 15-30 mg chorionic villi/placental tissue using sterile technique. Place in sterile tube(s) with transport media. If ordering multiple tests, 30 mg is recommended.	Preferred gestational age > 10-12 weeks. Must be received in a securely closed, sterile container. Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.
Cultured Chorionic Villi	CCV	T25 or T75 flask	At least 1- T25 (or T75) with a confluency of 75-80%. Note: Cultures should be < 3 weeks old. Accepted for test codes CMPRE and CMPRS only.	Send overnight at room temperature. Do not refrigerate or freeze.
Cord Blood	CB	Please speak with a laboratory genetic counselor for these requests.	Please speak with a laboratory genetic counselor for these requests.	Please speak with a laboratory genetic counselor for these requests.
DNA, Isolated	DNA	Microtainer	Isolation using the Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.	Refrigerate until time of shipment in 100 ng/μL in TE buffer. Ship sample at room temperature with overnight delivery.
Dried Blood Spot	DBS	Filter paper	Peripheral blood from finger prick or heel stick spotted on filter paper, completely saturating the circle. Air-dry the sample.	Do not expose specimen to heat or direct sunlight. Keep the specimen dry. Ship

			<p>3 full spots are needed (Confirmation testing requires a peripheral blood sample to be sent as well - Client will be notified to send this sample when abnormal results are reported).</p>	<p>sample at room temperature with overnight delivery.</p>
Products of Conception	POC	<p>Sterile container with EGL transport media</p> <p>If EGL media is not available, other sterile culture media can be used (RPMI, Hanks Solution).</p>	<p>≥ 15-30 mg chorionic villi/placental tissue using sterile technique. Place in sterile tube(s) with transport media.</p> <p>Tissues fixed in formalin cannot be used.</p> <p>The laboratory cannot legally accept entire fetuses/stillbirths. Must be a part of an individual.</p>	<p>Must be received in securely closed, sterile container with sterile culture media.</p> <p>Refrigerate until time of shipment. Ship sample at room temperature for receipt at EGL within 24 hours of collection.</p> <p>If fetal demise occurred more than 48-72 hours prior to retrieval of tissue and delivery to the lab, fetal tissue is unlikely to grow. In this case, placenta is most likely to grow.</p>
Skin Biopsy	SK	<p>Sterile container with EGL transport media or other sterile culture media (RPMI, Hanks Solution).</p>	<p>Obtain 1-2 cm pieces of tissue from a central location rather than a distal location to enhance cell viability and growth. Place in sterile container with EGL transport media or other sterile culture media. Use sterile dissection (no prep) for internal tissue. Tissue fixed in formalin cannot be used. Refrigerate until time of shipment.</p> <p>In the absence of media, place in a sterile container with a small amount of sterile saline.</p>	<p>Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.</p>
Whole Blood (Sodium heparin)	WBG	<p>Sodium heparin (green top)</p> <p>Required for all tests</p>	<p>Infants and young children (<2 years of age): 2-3 ml.</p> <p>Children > 2 years of age to 10 years old: 3-5 ml.</p> <p>Older children and adults: 5-10 ml.</p> <p>Autopsy: 2-3 ml unclotted cord or cardiac blood.</p>	<p>Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.</p>
Whole Blood (EDTA)	WBP	<p>EDTA (purple top)</p> <p>Required for test codes: CMSNP, VA, XC020, and XC021 only.</p>	<p>Infants and young children (<2 years of age): 2-3 ml.</p> <p>Children >2 years of age to 10 years old: 3-5 ml.</p> <p>Older children and adults: 5-10 ml.</p> <p>Autopsy: 2-3 ml unclotted cord or cardiac blood.</p>	<p>Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.</p>