

Specimen Requirements for Molecular Genetics Testing

<i>Sample Type</i>	<i>Code</i>	<i>Collection Container(s)</i>	<i>Collection and Processing Instructions</i>	<i>Shipping Instructions and Preferred Sample Condition</i>	<i>Comments</i>
Amniotic Fluid	AF	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.	
Cultured Amniocytes	CAF	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.	
Bone Marrow	BM	EDTA (purple top) or ACD (yellow top)	Collect 2-3 ml of bone marrow.	Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.	
Cord Blood	CB	EDTA (purple top) or ACD (yellow top)	Collect 1 to 3 ml of fetal blood.	Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.	
Chorionic Villi	CV	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.	
Cultured Chorionic Villi	CCV	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.	
Cultured Fibroblasts	CF	T25 or T75 flask	At least 1-T25 (or T75) with a confluency of 75-80%. Note: Cultures should be < 3 weeks old.	Send overnight at room temperature. Do not refrigerate or freeze.	
DNA, Isolated	DNA	Microtainer	Next generation sequencing (NGS): 30µg. Sanger sequencing: 5-25µg (varies by size of gene. Please contact the laboratory for specific amounts). Non-sequencing Tests: 20 µg. Array-based Tests: 10 µg. Isolation using the	Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.	Research laboratories: DNA extracted in research laboratories is not acceptable. Only under exceptional circumstances (e.g. proband not available) will DNA extracted in a research laboratory be accepted for clinical testing. Additional testing (e.g. of other family members) may be required to confirm

			Qiagen™ Puregene or Perkin Elmer™ Chemagen kit for DNA extraction is recommended.		<p>results.</p> <p>Laboratories outside the United States: non-US laboratories, which are not subject to CLIA regulations, will be reviewed on a case-by-case basis. Please call to speak with a laboratory genetic counselor prior to submitting a DNA sample from any non-CLIA-certified laboratory.</p> <p>Special Note: All clinical tests have been validated using DNA isolated by the Chemagen Automated Extraction method or Qiagen™ Puregen kit. If extracted DNA is submitted, information regarding the method used for extraction should be sent along with the sample.</p>
Dried Blood Spot	DBS	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.	
Muscle Biopsy	MB	Sterile container	1-2 mm in length or > 100 mg is acceptable. Flash freeze sample upon collection using liquid nitrogen. If storage is required, store sample at -80°C or colder.	Ship frozen sample on dry ice with overnight delivery. Not accepted on Saturdays.	
Formalin-Fixed Paraffin Embedded (FFPE) Tissue	PE	Block or slides	Submit FFPE block and a H&E stained slide OR submit two 15µm scrolls and a H&E stained slide OR submit FFPE unstained slides (15µmX3) and a H&E stained slide.	Ship at ambient temperature (18-25°C/64-77°F) in an insulated container by overnight courier. Do not heat or freeze.	Full strength liquid formaldehyde fixed tissue is NOT acceptable.
Products of Conception	POC		This is not a preferred specimen. Please speak with a laboratory	Send overnight at room temperature. Do not refrigerate or freeze.	

			genetic counselor for these requests, as director approval is required. Specimen requirements will be similar to CYG requirements. Any deviations will be approved by a lab director.		
Saliva	SLV	Oragene™ Saliva Collection kit	Oragene™ Saliva Collection kit used according to manufacturer instructions. Please contact EGL for a saliva collection kit for patients that cannot provide a blood sample. Note: Whole blood is the preferred sample.	Please do not refrigerate or freeze saliva sample. Please store and ship at room temperature.	
Skin Biopsy	SK	Sterile container with EGL transport media or other sterile culture media (RPMI, Hanks Solution)	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. In the absence of media, place in a sterile container with a small amount of sterile saline.	Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.	
Tissue Biopsy	TB	Sterile container	1-2 mm in length or > 100 mg is acceptable. Flash freeze sample upon collection using liquid nitrogen. If storage is required, store sample at -80°C	Ship frozen sample on dry ice with overnight delivery. Not accepted on Saturdays.	

			or colder.		
Whole Blood (Sodium heparin) Exception - May not be accepted for: MD, HY, MM021 (females only), MXLI1 (females only), AK, and CM080	WBG	Sodium heparin (green top)	Infants (<2 years): 2-3 ml. Children (>2 years): 3-5 ml. Older children and adults: 5-10 ml. Special notes: <ul style="list-style-type: none"> • Clotted or hemolyzed samples are not accepted. • Southern blot analysis: minimum 3 ml blood. 	Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.	
Whole Blood (EDTA) Preferred sample type	WBP	EDTA (purple top)	Infants (<2 years): 2-3 ml. Children (>2 years to 10 years old): 3-5 ml. Older children and adults: 5-10 ml. Special notes: <ul style="list-style-type: none"> • Clotted or hemolyzed samples are not accepted. • Southern blot analysis: minimum 3 ml blood. 	Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.	
Whole Blood (ACD)	WBY	ACD (yellow top)	Infants (<2 years): 2-3 ml. Children (>2 years to 10 years old): 3-5 ml. Older children and adults: 5-10 ml. Special notes: <ul style="list-style-type: none"> • Clotted or hemolyzed samples are not accepted. • Southern blot analysis: minimum 3 ml blood. 	Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.	