

Specimen Requirements for Biochemical 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>
Cerebral Spinal Fluid	CSF	Clean container without additives	In sterile container without additives. 1 ml. Freeze. Note: Blood contamination can interfere with test results.	Ship frozen sample on dry ice with overnight delivery.
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.
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 sample at room temperature with overnight delivery.
Plasma	PLA	Sodium heparin (green top) or EDTA (purple top) - Only accepted for test codes: CQ, HO, and BB041.	Sample should be collected while fasting or 2-4 hours postprandial. Centrifuge to separate plasma and freeze. Sodium heparin plasma 1-2 ml (each) for test codes: CN, AA, AR, BARST, BAAST, MQ, BNGLY, BSLOS, BCDGS, BOGLY, and BSTRL. Sodium heparin plasma 2-5 ml (each) for test codes: MW, BNBSF, BB021, and XB040. Sodium heparin plasma 3-5 ml (each) for test code: BCDGP. EDTA plasma 1-5 ml (each) for test codes: HO, BB041, and CQ. Special Notes: For test code HO, separate plasma within 30 minutes of collection. For test codes CQ and BB041, sample must be protected from light.	Ship frozen sample on dry ice with overnight delivery.
Red Blood Cells, Washed	RBC	Sodium heparin (green top)	Washed red blood cell preparation: Step 1: Centrifuge the whole blood at 3000rpm (1800rcf) for 5 minutes. Step 2: Remove plasma and buffy coat layer. Step 3: Resuspend the red cells in normal saline (0.9% NaCl) with approximately 2 times the volume of the red cells, and invert the tube to mix. Step 4: Centrifuge for 5 minutes at 2000 rpm and discard the supernatant.	Ship frozen sample on dry ice with overnight delivery.

			<p>Step 5: Repeat Steps 3 & 4 twice for a total of 3 washes or until the supernatant is clear.</p> <p>Step 6: Completely remove the supernatant and freeze the red cells at -20°C, preferably at -80°C.</p> <p>Ship frozen.</p>	
Serum	SRM	No additive (red top) or SST (serum separator tube - no additives)	<p>Centrifuge to separate serum and freeze.</p> <p>Special Note: biotinidase deficiency (test code: BX) requires special collection and handling. Please see specific test page for detailed instructions prior to collecting sample.</p> <p>No Additive or SST serum 1-2 ml (each) for test codes: AG, CZ, TT, BNGLY, BCDGP, and BCDGS.</p> <p>No Additive or SST serum 3-5 ml (each) for test codes: BM and BX.</p>	Ship frozen sample on dry ice with overnight delivery.
Skin Biopsy	SK	Sterile container with EGL transport media or other sterile culture media (RPMI, Hanks Solution)	<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>	Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.
Supernatant (For pyruvate/pyruvic acid testing only) <ul style="list-style-type: none"> • Blood: Code ZF • CSF: Code ZE 	PCS	Sodium heparin (green top) Or CSF	<ol style="list-style-type: none"> 1. Collect specimen into a chilled sodium heparin tube. Pyruvate is stable for only 30 minutes. 2. Immediately after specimen collection add exactly 1 ml specimen to 2 mL of 7-8% perchloric acid in an uncoated tube. Mix well. 3. Let the sample stand for 10 minutes in ice bath. 4. Centrifuge at 1500xg in a refrigerated centrifuge for 10 minutes. 5. Transfer supernatant into a new clear uncoated tube, if supernatant is not clear, repeat centrifugation for another 10 minutes. 6. Transport clear supernatant into a new uncoated transport tube with patient identifiers, cap securely and keep frozen. 	Ship frozen sample on dry ice with overnight delivery.
Urine	URN	Clean container without additives	Freeze sample. Fasting or first void sample is preferable. Please check individual test codes for minimum specimen requirements.	Ship frozen sample on dry ice with overnight delivery.

			<p>Special Note: For diagnostic purposes, specimens should ideally be collected during time of acute illness as abnormal metabolite levels may decrease, sometimes to near normal concentrations, when patient is well. (Does not apply to test codes: BLSDS, GA, and OS.)</p>	
Whole Blood (Sodium heparin)	WBG	Sodium heparin (green top)	<p>1 ml or more (each) for test codes: ZF and BKRBD.</p> <p>3-5 ml (each) for test codes: BMPS6, BCQLC, GP, GK, LA, LQ, GT, LG, LR, LO, HC, DW, GR, LP, LM, LN, and LB.</p> <p>3-10 ml (each) for test code: BC.</p> <p>5-10 ml (each) for test code: LS.</p> <p>Infants and young children (<2 years of age): 5 ml Older children and adults: 7 ml For test code: GS.</p> <p>Infants and young children (<2 years of age): 1 ml Older children and adults: 3-5 ml For test code: BOGLY.</p>	<p>Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.</p> <p>Not accepted on Saturdays.</p>
Whole Blood (EDTA)	WBP	EDTA (purple top)	<p>Infants and young children (<2 years of age): 2-3 ml. Children > 2 years of age to 10 years old: 3-5 ml. Older children and adults: 5-10 ml.</p>	<p>Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.</p> <p>Not accepted on Saturdays.</p>
Whole Blood (No additive)	WBR	No additive (red top) or SST (serum separator tube - no additives)	<p>1-2 ml (each) for test codes: AG, CZ, TT, BNGLY, BCDGS, and BCDGP.</p> <p>3-5 ml (each) for test codes: BM and BX.</p>	<p>Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.</p> <p>Not accepted on Saturdays.</p>