

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.
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 HO	Sodium heparin tube 1-2 ml (each): CN, AA, AR, BARST, BAAST, MQ, BNGLY, BSLOS, BCDGS, BOGLY, BSTRL Sodium heparin tube 2-5 ml(each): MW, BNBSF, and BB021 Sodium heparin tube 3-5 ml (each): BCDGP EDTA tube 1-5 ml (each): HO Sample should be collected while fasting or 2-4 hours postprandial. Centrifuge to separate plasma and freeze. Special Notes: For test code HO , separate plasma within 30 minutes of collection.	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	Ship frozen sample on dry ice with overnight delivery.

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			<p>normal saline (0.9% NaCl) with approximately 2 times the volume of the red cells, and invert the tube to mix.</p> <p>Step 4: Centrifuge for 5 minutes at 2000 rpm and discard the supernatant.</p> <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	Clean container without additives	<p>Spin down, transfer, and ship frozen.</p> <p>1-2 ml (each): AG, CZ, TT, BNGLY, BCDGS, BCDGP</p> <p>3-5 ml (each): BM and BX</p> <p>Special Note: Test code BX requires a control to be sent with sample for testing. Must be collected from an individual not related to the patient.</p>	Ship frozen sample on dry ice with overnight delivery.
Urine	URN	Clean container without additives	<p>Freeze sample.</p> <p>Fasting or first void sample is preferable.</p> <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. <i>(Does not apply to test codes BLSDS, GA, & OS.)</i></p>	Ship frozen sample on dry ice with overnight delivery.
Whole Blood (Sodium heparin)	WBG	Sodium Heparin (Green Top)	3-5 ml (each): BMPS6, GP, LQ, GT, LG, LR, LO, HC, DW, LP, LM, LN, LB	Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or

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			5-10 ml (each): LS Infants and Young Children (<2 years of age): 5 ml Older Children and Adults: 7 ml Test code: GS Infants and Young Children (<2 years of age): 1 ml Older Children and Adults: 3-5 ml Test code: BOGLY	freeze. NOT ACCEPTED ON SATURDAYS
Whole Blood (EDTA)	WBP	EDTA (Purple Top)	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	Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. NOT ACCEPTED ON SATURDAYS
Whole Blood (No additive)	WBR	No Additive (Red Top) or SST (Serum Separator Tube - no additives)	1-2 ml (each): AG, CZ, TT, BNGLY, BCDGS, BCDGP 3-5 ml (each): BM and BX Special Note: Test code BX requires a control to be sent with sample for testing. Must be collected from an individual not related to the patient.	Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. NOT ACCEPTED ON SATURDAYS