

Specimen Requirements for Biochemical Genetics Testing

Sample Type	Code	Collection Container(s)	Collection and Processing Instructions	Shipping Instructions and Preferred Sample Condition
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 CQ, HO, & BB041	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, BB021, and XB040 Sodium heparin tube 3-5 ml (each): BCDGP EDTA tube 1-5 ml (each): HO, BB041, CQ 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. For test codes CQ & 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. Step 5: Repeat Steps 3 & 4 twice for a total of 3 washes or until the supernatant is clear. Step 6: Completely remove the supernatant and freeze the red cells at -20°C, preferably at -80°C. Ship frozen.	Ship frozen sample on dry ice with overnight delivery.
Serum	SRM	Clean container without additives		Fresh sample accepted if received within 24 hours of collection. If not, freeze and ship frozen on dry ice. NOT ACCEPTED ON SATURDAYS

Sample Type	Code	Collection Container(s)	Collection and Processing Instructions	Shipping Instructions and Preferred Sample Condition
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 & BHEX4.</i>)</p>	Ship frozen sample on dry ice with overnight delivery.
Whole Blood (Sodium heparin)	WBG	Sodium Heparin (Green Top)	<p>1 ml or more (each): ZF, BKRBD</p> <p>3-5 ml (each): BMPS6, BCQLC, GP, GK, LA, LQ, GT, LG, LR, LO, HC, DW, GR, LP, LM, LN, LB</p> <p>3-10 ml (each): BC</p> <p>5-10 ml (each): LS</p> <p>Infants and Young Children (<2 years of age): 5 ml Older Children and Adults: 7 ml</p> <p>Test code: GS</p> <p>Infants and Young Children (<2 years of age): 1 ml Older Children and Adults: 3-5 ml</p> <p>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</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>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): AG, CZ, TT, BNGLY, BCDGS, BCDGP</p> <p>3-5 ml (each): BM, 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>