

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 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 |

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|-------------------------------------|-------------|--|---|--|
| 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> |