### Specimen Requirements for Biochemical Genetics Testing

<table>
<thead>
<tr>
<th>Sample Type</th>
<th>Code</th>
<th>Collection Container(s)</th>
<th>Collection and Processing Instructions</th>
<th>Shipping Instructions and Preferred Sample Condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cerebral Spinal Fluid</td>
<td>CSF</td>
<td>Clean container without additives</td>
<td>In sterile container without additives. 1 ml. Freeze. Note: Blood contamination can interfere with test results.</td>
<td>Ship frozen sample on dry ice with overnight delivery.</td>
</tr>
<tr>
<td>Cultured Fibroblasts</td>
<td>CF</td>
<td>T25 or T75 flask</td>
<td>At least 1- T25 (or T75) with a confluency of 75-80%. Note: Cultures should be &lt; 3 weeks old.</td>
<td>Send overnight at room temperature. Do not refrigerate or freeze.</td>
</tr>
<tr>
<td>Dried Blood Spot</td>
<td>DBS</td>
<td>Filter paper</td>
<td>Peripheral blood from finger prick or heel stick spotted on filter paper, completely saturating the circle. Air-dry the sample.</td>
<td>Do not expose specimen to heat or direct sunlight. Keep the specimen dry. Ship sample at room temperature with overnight delivery.</td>
</tr>
<tr>
<td>Plasma</td>
<td>PLA</td>
<td>Sodium heparin (green top) or EDTA (purple top) - Only accepted for test codes: CQ, HO, and BB041.</td>
<td>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 BSTR. 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.</td>
<td>Ship frozen sample on dry ice with overnight delivery.</td>
</tr>
<tr>
<td>Red Blood Cells, Washed</td>
<td>RBC</td>
<td>Sodium heparin (green top)</td>
<td>Washed red blood cell preparation: <strong>Step 1</strong>: Centrifuge the whole blood at 3000rpm (1800rcf) for 5 minutes. <strong>Step 2</strong>: Remove plasma anduffy coat layer. <strong>Step 3</strong>: 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. <strong>Step 4</strong>: Centrifuge for 5 minutes at 2000 rpm and discard the supernatant.</td>
<td>Ship frozen sample on dry ice with overnight delivery.</td>
</tr>
</tbody>
</table>
### 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.

### Serum
| SRM | No additive (red top) or SST (serum separator tube - no additives) | Centrifuge to separate serum and freeze. Special Note: biotinidase deficiency (test code: BX) requires special collection and handling. Please see specific test page for detailed instructions prior to collecting sample. No Additive or SST serum 1-2 ml (each) for test codes: AG, CZ, TT, BNGLY, BCDGP, and BCDGS. No Additive or SST serum 3-5 ml (each) for test codes: BM and BX. | 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) | 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. |

### Supernatant (For pyruvate/pyruvic acid testing only)
- Blood: Code ZF
- CSF: Code ZE

| PCS | Sodium heparin (green top) Or CSF | 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. |
### 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.)*

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Container Type</th>
<th>Minimum Volume (each)</th>
<th>Test Codes</th>
<th>Handling Instructions</th>
</tr>
</thead>
</table>
| Whole Blood (Sodium heparin) | WBG | Sodium heparin (green top) | 1 ml or more (each) | ZF and BKRBD.  
3-5 ml (each) | BMP56, BCQLC, GP, GK, LA, LQ, GT, LG, LR, LO, HC, DW, GR, LP, LM, LN, and LB.  
3-10 ml (each) | BC.  
5-10 ml (each) | LS.  
Infants and young children (<2 years of age): 5 ml  
Older children and adults: 7 ml  
For test code: GS.  
Infants and young children (<2 years of age): 1 ml  
Older children and adults: 3-5 ml  
For test code: BOGLY.  
| 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 (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, and BCDGP.  
3-5 ml (each) | BM and BX.  
| Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.  
Not accepted on Saturdays. |