## SERVICE SCHEDULE & GUIDELINES FOR SAMPLE SHIPMENT

### ACYL-CARNITINE PROFILE: PLASMA/SERUM AND DRIED BLOOD SPOT (DBS)

**Amount of sample:**
- Heparinized (green top) plasma or serum: 0.5 mL (absolute minimum: 0.2 mL)
- Whole blood: 1.0 mL (absolute minimum: 0.5 mL)
- Dried blood spot (DBS) card: fill at least 2 spots; allow to air dry for 4 hours

**Special Note:** Heparinized plasma or DBS are the samples of preference

**Shipping:**
- Plasma or serum: Ship frozen on dry ice
- Whole blood: Ship at 4°C on wet ice or ice pack (not frozen)
- DBS: Mail in envelope at ambient temperature

**CPT Code:** 82017

**Method:** Tandem Mass Spectrometry (MS/MS)

**Test Schedule:** Tuesday and Friday, or as needed

### AMINO ACIDS: PLASMA/SERUM AND CSF

**Amount of sample:**
- Heparinized (green top) plasma or serum: 0.5 mL (absolute minimum: 0.2 mL)
- Whole blood: 1 to 3 mL (absolute minimum: 0.5 mL)
- CSF: 0.5 mL (absolute minimum: 0.2 mL)

**Shipping:**
- Plasma, serum or CSF: Ship frozen on dry ice
- Whole blood: Ship at 4°C on wet ice or ice pack (not frozen)

**CPT Code:** 82139

**Method:** Ion-exchange HPLC

**Test Schedule:** Daily, Monday through Friday

### AMINO ACIDS: URINE

**Amount of sample:** 5 mL (no preservatives); (absolute minimum: 1 mL)

**Shipping:** Ship frozen on dry ice

**CPT Codes:** 82139, 82570

**Method:** Ion-exchange HPLC

**Test Schedule:** Daily, Monday through Friday

### CARNITINE LEVELS (FREE & TOTAL): PLASMA/SERUM

**Amount of sample:** Heparinized (green top) plasma or serum: 0.5 mL; (absolute minimum: 0.2 mL)

**Shipping:**
- Plasma or serum: Ship frozen on dry ice
- Whole blood: Ship at 4°C on wet ice or ice pack (not frozen)

**CPT Codes:** 82379

**Method:** Tandem Mass Spectrometry (MS/MS)

**Test Schedule:** Tuesday, Wednesday and Friday

### CARNITINE LEVELS (FREE & TOTAL): URINE

**Amount of sample:** 5 mL (no preservatives); (absolute minimum: 0.6 mL)

**Shipping:** Ship frozen on dry ice

**CPT Code:** 82379, 82570

**Method:** Tandem Mass Spectrometry (MS/MS)

**Test Schedule:** Once a week
## Combination Carnitine Levels / Acylcarnitine Profile: Plasma/Serum

<table>
<thead>
<tr>
<th>Amount of sample</th>
<th>Heparinized (green top) plasma or serum: 0.75 mL; (absolute minimum: 0.5 mL)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Shipping</td>
<td>Plasma or serum: Ship frozen on dry ice</td>
</tr>
<tr>
<td></td>
<td>Whole blood: Ship at 4°C on wet ice or ice pack (not frozen)</td>
</tr>
<tr>
<td>CPT Codes</td>
<td>82017, 82379</td>
</tr>
<tr>
<td>Method</td>
<td>Tandem Mass Spectrometry (MS/MS)</td>
</tr>
<tr>
<td>Test Schedule*</td>
<td>See above for acylcarnitine and plasma carnitine schedule</td>
</tr>
</tbody>
</table>

## Creatine/Guanidinoacetate Assay: Plasma/Serum, Urine

<table>
<thead>
<tr>
<th>Amount of sample</th>
<th>Urine: 1.0 mL (no preservatives); (absolute minimum: 0.25 mL)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Plasma (heparinized or EDTA) or serum: 0.5 mL (absolute minimum: 0.25 mL)</td>
</tr>
<tr>
<td>Shipping</td>
<td>Ship frozen on dry ice</td>
</tr>
<tr>
<td>CPT Codes</td>
<td>Urine: 82542, 82540, 82570</td>
</tr>
<tr>
<td></td>
<td>Plasma: 82542, 82540, 82565</td>
</tr>
<tr>
<td>Method</td>
<td>Liquid Chromatography - Tandem Mass Spectrometry (LC-MS/MS)</td>
</tr>
<tr>
<td>Test Schedule*</td>
<td>Once a week</td>
</tr>
</tbody>
</table>

## Hexose Tetrasccharide (GLC4): Urine or Dried Urine Spot

<table>
<thead>
<tr>
<th>Amount of sample</th>
<th>Urine: 1.0 mL (no preservatives); (absolute minimum: 0.25 mL)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Plasma frozen on dry ice</td>
</tr>
<tr>
<td>Storage and handling</td>
<td>Urine should be stored at -20°C and shipped frozen on dry ice. Sample may be</td>
</tr>
<tr>
<td></td>
<td>kept at room temperature for up to 4 hours. After this time, keep sample cold at</td>
</tr>
<tr>
<td></td>
<td>4°C for up to 24 hours. Freeze if stored for longer than 24 hours before shipping.</td>
</tr>
<tr>
<td></td>
<td>If transportation to lab is same day as collection, (local hospitals only) the sample</td>
</tr>
<tr>
<td></td>
<td>must be transported at 4°C on an ice pack or frozen on dry ice.</td>
</tr>
<tr>
<td></td>
<td>For dried urine spots, please contact the laboratory for specific instruction.</td>
</tr>
<tr>
<td>CPT Codes</td>
<td>Urine: 82542, 82570</td>
</tr>
<tr>
<td>Method</td>
<td>Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)</td>
</tr>
<tr>
<td>Test Schedule*</td>
<td>Once a week</td>
</tr>
</tbody>
</table>

## Lyso-GB₁ (Glucosylsphingosine) Gaucher Biomarker: Plasma

<table>
<thead>
<tr>
<th>Amount of sample</th>
<th>Plasma (EDTA): 1.0 mL; (absolute minimum: 0.30 mL)**</th>
</tr>
</thead>
<tbody>
<tr>
<td>Shipping</td>
<td>Ship frozen on dry ice</td>
</tr>
<tr>
<td>CPT Codes</td>
<td>82542</td>
</tr>
<tr>
<td>Method</td>
<td>Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)</td>
</tr>
<tr>
<td>Test Schedule*</td>
<td>Once a month</td>
</tr>
<tr>
<td><strong>If sending whole blood for combined glucocerebroside enzyme analysis and lyso-GB₁ testing minimum volume needed is 2 mL whole blood, and ideally 4 mL whole blood.</strong></td>
<td></td>
</tr>
</tbody>
</table>

## Lyso-GB₃ (Lyso-Globotriaosylceramide) Fabry Biomarker: Plasma

<table>
<thead>
<tr>
<th>Amount of sample</th>
<th>Plasma (EDTA): 1.0 mL; (absolute minimum: 0.30 mL)**</th>
</tr>
</thead>
<tbody>
<tr>
<td>Shipping</td>
<td>Ship frozen on dry ice</td>
</tr>
<tr>
<td>CPT Codes</td>
<td>82542</td>
</tr>
<tr>
<td>Method</td>
<td>Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)</td>
</tr>
<tr>
<td>Test Schedule*</td>
<td>Once a month</td>
</tr>
<tr>
<td><strong>If sending whole blood for combined alpha-Gal A enzyme, gene sequencing and lyso-Gb₃ testing minimum volume needed is 2 mL whole blood, and ideally 4 mL whole blood.</strong></td>
<td></td>
</tr>
</tbody>
</table>
METHYLMALONIC ACID: PLASMA OR SERUM

Amount of sample: Plasma (heparinized or EDTA) or serum: 0.5 mL; (absolute minimum 0.2 mL)
Shipping: Ship frozen on dry ice
CPT Codes: 83921
Method: Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)
Test Schedule*: As Needed

MUCOPOLYSACCHARIDES CS, DS, HS: URINE

Amount of sample: 1.0 mL (no preservatives); (absolute minimum: 0.50 mL)
Shipping: Freeze within 4 hours of collection and ship frozen on dry ice
CPT Codes: 83864x2, 82570
Method: Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)
Test Schedule*: Bi-weekly

MUCOPOLYSACCHARIDE KS: URINE

Amount of sample: 1.0 mL (no preservatives); (absolute minimum: 0.50 mL)
Shipping: Freeze within 4 hours of collection and ship frozen on dry ice
CPT Codes: 83864, 82570
Method: Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)
Test Schedule*: Bi-weekly

MUCOPOLYSACCHARIDES SCREEN CS, DS, HS, KS: URINE

Amount of sample: 1.0 mL (no preservatives); (absolute minimum: 0.50 mL)
Shipping: Freeze within 4 hours of collection and ship frozen on dry ice
CPT Codes: 83864x3, 82570
Method: Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)
Test Schedule*: Bi-weekly

ORGANIC ACIDS: URINE

Amount of sample: 5 mL (no preservatives); (absolute minimum: 1.0 mL)
Shipping: Ship frozen on dry ice
CPT Codes: 83919, 82570
Method: Capillary Column Gas Chromatography - Mass Spectrometry (GC-MS)
Test Schedule*: Daily, Monday through Friday

SULFOCYSTEINE: URINE

Amount of sample: 1.0 mL (no preservatives); (absolute minimum: 0.25 mL)
Shipping: Ship frozen on dry ice
CPT Codes: 82131, 82570
Method: Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)
Test Schedule*: Twice a week (as needed)
## VITAMIN A AND E: SERUM

<table>
<thead>
<tr>
<th>Description</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amount of sample</td>
<td>Serum (red top tube) only: 1.0 mL; (absolute minimum: 0.5 mL)</td>
</tr>
<tr>
<td>Shipping</td>
<td>Ship frozen on dry ice</td>
</tr>
<tr>
<td>Storage and handling</td>
<td>Separate serum, protect from light and freeze as soon as possible and within 2 hours of collection. Store in an amber vial or a vial covered with aluminum foil.</td>
</tr>
<tr>
<td>CPT Codes</td>
<td>84446 (Vitamin E); 84590 (Vitamin A)</td>
</tr>
<tr>
<td>Method</td>
<td>Quantitative High Performance Liquid chromatography (HPLC) with UV detection</td>
</tr>
<tr>
<td>Test Schedule*</td>
<td>As Needed</td>
</tr>
</tbody>
</table>

*Test Schedule* indicates when the test should be performed as needed.
ADDITIONAL LABORATORY INFORMATION:

- *Test schedule is approximate and subject to change at any time.
- If results are required urgently, please contact the laboratory to discuss a possible faster turnaround time with the laboratory director.
- Preliminary results can be made available as a verbal report and FAX on request. Final reports are mailed after interpretation and review of all tests requested on a patient. ONLY DEFINITE DIAGNOSES ARE REPORTED PROMPTLY BY TELEPHONE AND FAX.
- Please provide telephone and fax number of referring physician.
- Please label all samples with name, date, and time using INDELIBLE INK, and place samples in a plastic bag (gummed labels fall off samples shipped on dry ice).
- Please ship samples only MONDAY – THURSDAY. No Saturday deliveries. The lab is closed on Saturdays and Sundays, and on major holidays.
- Please use only guaranteed OVERNIGHT COURIERS (U.S. Postal Service may take 3-4 days to arrive, samples will be ruined).

SHIPPING AND MAILING ADDRESS:
DUHS Biochemical Genetics Laboratory, 801 Capitola Drive, Suite 6, Durham, NC 27713
Main Office Phone: (919) 549-0445; Fax: (919) 549-0709
https://clinlabs.duke.edu/biochemical-genetics-laboratory

CERTIFICATION NUMBERS:
College of American Pathologist (CAP): 43190-36
Joint Commission (JCAHO): 006360390
Health Care Financing Admin. (HCFA or CLIA): 34D0674833

FEDERAL TAX ID NUMBER: 56-2070036 (Duke University Health System)

LABORATORY DIRECTORS:
Dwight Koeberl, MD, PhD, FACMG Medical Director 919-681-9919
Deeksha Bali, PhD, FACMG Co-Director 919-684-0025
Sarah Young, PhD, FACMG Co-Director 919-684-4259
Ashlee Stiles, PhD, FACMG Co-Director 919-684-0073

GENERAL INQUIRIES:
Jack Bowen Administrative Assistant 919-684-0189
Stephanie Terry Administrative Assistant 919-684-2969

GLYCOGEN STORAGE DISEASE (GSD) LAB:
For information on testing performed by the Duke Enzyme and Glycogen Storage Disease Section of DUHS Biochemical Genetics Lab please call the main laboratory number, (919) 549-0445, and select option #2 for the GSD laboratory.

https://testcatalog-test.dhe.duke.edu/?lab=123060043