



## SERVICE SCHEDULE & GUIDELINES FOR SAMPLE SHIPMENT

### ACYLCARNITINE 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

Amount of sample: Heparinized (green top) plasma or serum: 0.75 mL; (absolute minimum: 0.5 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: 82017, 82379  
Method: Tandem Mass Spectrometry (MS/MS)  
Test Schedule\*: See above for acylcarnitine and plasma carnitine schedule

### CREATINE/GUANIDINOACETATE ASSAY: PLASMA/SERUM, URINE

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

### HEXOSE TETRASACCHARIDE (GLC4): URINE OR DRIED URINE SPOT

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

### LYSO-GB<sub>1</sub> (GLUCOSYLSPHINGOSINE) GAUCHER BIOMARKER: PLASMA

Amount of sample: Plasma (EDTA): 1.0 mL; (absolute minimum: 0.30 mL)\*\*  
Shipping: Ship frozen on dry ice  
CPT Codes: 82542  
Method: Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)  
Test Schedule\*: Once a month

\*\*If sending whole blood for combined glucocerebrosidase enzyme analysis and lyso-Gb<sub>1</sub> testing minimum volume needed is 2 mL whole blood, and ideally 4 mL whole blood.

### LYSO-GB<sub>3</sub> (LYSO-GLOBOTRIAOSYLCERAMIDE) FABRY BIOMARKER: PLASMA

Amount of sample: Plasma (EDTA): 1.0 mL; (absolute minimum: 0.30 mL)\*\*  
Shipping: Ship frozen on dry ice  
CPT Codes: 82542  
Method: Ultra performance liquid chromatography - Tandem Mass Spectrometry (UPLC-MS/MS)  
Test Schedule\*: Once a month

\*\*If sending whole blood for combined alpha-Gal A enzyme, gene sequencing and lyso-Gb<sub>3</sub> testing minimum volume needed is 2 mL whole blood, and ideally 4 mL whole blood.



### 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

Amount of sample: Serum (red top tube) only: 1.0 mL; (absolute minimum: 0.5 mL)  
Shipping: Ship frozen on dry ice  
Storage and handling: 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.  
CPT Codes: 84446 (Vitamin E); 84590 (Vitamin A)  
Method: Quantitative High Performance Liquid chromatography (HPLC) with UV detection  
Test Schedule\*: 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>