

Specimen Requirements

- 1. Acylcarnitines (CPT 82017)**
 - a) 1 ml plasma isolated from blood collected in a sodium heparin/green top tube.
Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.
- 2. Amino Acid Quantitation (CPT 82139)**
 - a) Plasma: 1 ml isolated from blood collected in a sodium heparin/green top tube.
 - b) CSF: 1 ml
 - c) Urine: random specimen, minimum of 5-10 ml
Specimens from outside clients should be sent frozen on dry ice by overnight express carrier.
- 3. Carbohydrate Deficient Transferrin (CPT 82373)**

1 ml plasma isolated from blood collected in a sodium heparin/green top tube.
Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.
- 4. Carnitine (total & free) (CPT 84311)**
 - a) 1 ml plasma isolated from blood collected in a sodium heparin/green top tube.
 - b) Urine: random specimen, minimum of 5-10 ml
Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.
- 5. Disaccharidase Analysis (CPT 82491)**
 - a) Duodenal biopsy, 5-7 mg tissue
Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.
- 6. Epimerase [Uridine Diphosphate 4-Epimerase (CPT 82657)]**
 - a) 5 ml of blood collected in a sodium heparin/green top tube.
*Specimens from reference labs should be shipped room temperature by overnight express carrier. **Samples are to be sent within 24 hours of draw.***
- 7. Free Homocystine [Amino Acid Quant -- Homocystinuria (CPT 82139)]**
 - a) 1 ml of plasma. Blood is drawn into a sodium heparin/green top tube, chilled, centrifuged at 5°C and the plasma should be immediately frozen.
Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier within 24 hours of draw.
- 8. Galactitol Analysis (CPT 82491)**
 - a) Random urine specimen, minimum of 5-10 ml
Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.
- 9. Galactokinase (CPT 82759)**
 - a) 5 ml of blood collected in a sodium heparin/green top tube.
*Specimens from reference labs should be shipped room temperature by overnight express carrier. **Samples are to be sent within 24 hours of draw.***
- 10. Galactose-1-Phosphate (CPT 84378)**
 - a) 5 ml of blood collected in a sodium heparin/green top tube.
*Specimens from reference labs should be shipped room temperature by overnight express carrier. **Samples are to be sent within 24 hours of draw.***
- 11. Galactosemia Quantitation [Galactose-1-Phosphate Uridyltransferase (CPT 82775)]**
 - a) 5 ml of blood collected in a sodium heparin/green top tube.
*Specimens from reference labs should be shipped room temperature by overnight express carrier. **Samples are to be sent within 24 hours of draw.***
- 12. Lactate/Pyruvate Analysis (CPT 83605/84210)**
 - a) Collect 1.0 ml of blood without the use of a tourniquet and add to 2.0 ml of ice-cold 8% perchloric acid. Mix thoroughly by repeated inversion and centrifuge at 200 x g for 10 minutes. At 5°C. Measure total volume, if necessary. Collect supernate and freeze.
 - b) CSF – Add 1.0 ml of CSF to 2.0 ml of ice-cold 8% perchloric acid. Mix thoroughly by repeated inversion and centrifuge at 200 x g for 10 minutes. At 5°C. Measure total volume, if necessary. Collect supernate and freeze. If tap is bloody, collect sample as noted above for blood.
Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.

**13. Lysosomal Enzymes (CPT 82657)
WHITE BLOOD CELLS**

ENZYME

β-galactosidase
alpha-galactosidase
β-hexosaminidase
alpha-mannosidase
alpha-fucosidase
Arylsulfatase A
alpha-iduronidase
β-glucosidase
Sphingomyelinase
β-glucuronidase

DISEASE

GM1 gangliosidosis
Fabry
Tay-Sachs & variants
Mannosidosis
Fucosidosis
Metachromatic leukodystrophy
Hurler/Hurler-Scheie (additional charge if requested)
Gaucher
Niemann-Pick, Types A & B (special request)
MPS VII

SERUM

β-galactosidase
β-hexosaminidase alpha-
fucosidase β-glucuronidase

c) 7-10 ml of blood collected in a sodium heparin/green top tube for isolation of white blood cells.

d) 2 ml of blood collected in a red top (serum) tube.

Specimens from reference labs are to be sent at room temperature by overnight express carrier within 24 hours of draw.

14. Methylmalonic Acid (CPT 83921)

a) 1 ml plasma isolated from blood collected in a sodium heparin/green top tube.

Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.

15. N-glycan Analysis (CPT 83789/84375)

a) 1 ml plasma isolated from blood collected in a sodium heparin/green top tube.

b) *Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.*

16. Organic Acid Quantitation (CPT 83918)

a) Random urine sample, minimum of 5-10 ml.

Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.

17. Orotic Acid (CPT 83918)

a) Random urine specimen, 5-10 ml.

Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.

18. Ketone Bodies Panel (CPT 82010)

a) Patient prep: Preferred non-fasted

b) Preferred: Serum Separator Tube (SST/Gold Top)

c) Acceptable: Serum Red Top

d) Plasma Sodium Heparin (NaHep/Green Top)

e) Preferred Volume: 2.0 mL serum/plasma

f) Minimum Volume: 0.1 mL serum/plasma

g) *Stability and transport: Serum/plasma must be separated within 2 hours of collection when arrived at room temperature and within 6 hours of collection when kept on ice. Separated serum/plasma should be kept frozen at -80°C and transported on dry ice to the laboratory.*

19. Oxidative Phosphorylation Fibroblast (CPT 86257, 88233, 87109, 84157)

a) CHOP Patients: skin biopsy to be sent to DGD for culture

b) Outreach Clients: Cultured skin fibroblasts (less than 10 passage preferred), send 2 T25 fibroblast flasks.

c) *During off hours, send sample to Central Laboratory Services. Send copy of paperwork to Metabolic for processing. If sending from outside CHOP, you must call 215-590-3394 to schedule testing. Upon notification of test acceptance, please submit 2 T25 flasks of cultured fibroblasts by overnight courier on the scheduled date.*

ADDENDUM

1. All reports will have an interpretation.
2. Reports on quantitative tests will be accompanied by normal values established in our laboratory for the appropriate age group (when applicable). Otherwise, the results will be reported as normal/abnormal.
3. Turnaround time for each test are available in the online test menu. **Priority specimens** will be processed immediately and verbal results will be available within 1 business day. **The laboratory must be notified 24 hours in advance of such priority samples.**
4. Consultations with the Metabolic Staff to discuss results will be available upon request.

**Send to: Michael J. Palmieri Metabolic Laboratory
CHOP/Main Bldg/Room 5NW55
3401 Civic Center Blvd Philadelphia, PA 19104
Phone: (215) 590-3394
FAX: (215) 590-1998**