

Michael J. Palmieri Metabolic Laboratory  
Children's Hospital of Philadelphia

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. **CPT I & II (88233,82658,87109)** - See attachment.
  
6. **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.*
  
7. **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.**
  
8. **Fatty Acid Oxidation Analysis (CPT 89240,88233,87109)** – See attachment.
  
9. **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.*
  
10. **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.*
  
11. **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.*

**12. 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.*

**13. 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.*

**14. 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 – 1.0 ml in CSF tube. 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.*

**15. Lysosomal Enzymes (CPT 82657)**

WHITE BLOOD CELLS

Enzyme	Disease
β-galactosidase	GM1 gangliosidosis
alpha-galactosidase	Fabry
β-hexosaminidase	Tay-Sachs & variants
alpha-mannosidase	Mannosidosis
alpha-fucosidase	Fucosidosis
Arylsulfatase A	Metachromatic leukodystrophy
alpha-iduronidase	Hurler/Hurler-Scheie (additional charge if requested)
β-glucosidase	Gaucher
Sphingomyelinase	Niemann-Pick, Types A & B (special request)
β-glucuronidase	MPS VII

SERUM

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

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

**16. 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.*

**17. N-Glycan (CPT 83789, 84735)**

- 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.*

**18. Non Esterified Fatty Acids (CPT 82725)**

- a) 2 ml serum isolated from blood collected in a gold SST tube.  
*Specimens from reference labs should be shipped frozen on dry ice by overnight express carrier.*

**19. 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.*

**20. 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.*

**21. SCHAD (CPT 82657,88233,87109,)**

- a) 7-10 ml of blood collected in a sodium heparin/green top tube for isolation of white blood cells.  
b) skin fibroblast culture  
*Specimens from reference labs are to be sent at room temperature by overnight express carrier within 24 hours of draw.*

**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. Normal values will be included in the organic acid analysis, however, only if abnormal quantities of a particular compound(s) are found. Otherwise, the results will be reported as normal/abnormal.
3. Turnaround time for daily tests will be 5-7 working days. **Priority specimens** will be processed immediately and verbal results will be available within 24 hours. **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.

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