

ROBERT GUTHRIE BIOCHEMICAL & MOLECULAR GENETICS LABORATORY**Georgirene D. Vladutiu, Ph.D., Director**

THE BUFFALO GENERAL HOSPITAL

KALEIDA HEALTH

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SPECIMEN REQUIREMENTS
FOR BIOCHEMICAL AND
MOLECULAR TESTING
Page 1 of 1***Please call laboratory office before shipping any specimens: (716) 859-7741
(See Collection, Shipping, and Handling Instructions for details)***

Test Category	Specimen Requirements	Shipping Instructions	Additional Information
Lysosomal Storage Diseases, General (Including Tay-Sachs disease carrier testing)	10 mL heparinized whole blood (1 green top tube + 1 control specimen from an unrelated individual)	Ship at room temperature to arrive in laboratory within 24 hrs of collection	5 mL whole blood acceptable for analysis in infants
Amino Acid Analysis, General	1.0 mL plasma (separated within 30 min of collection) 1-5 mL urine 0.5 mL cerebrospinal fluid	Ship overnight frozen on dry ice	Complete amino acid profile
Amino Acid Analysis, Special	4-5 drops of whole blood spotted on newborn screening filter paper card; air dried	Ship by regular mail double enveloped and labeled	Phenylalanine & tyrosine monitoring is provided for PKU patients
Metabolic Muscle Disease Profiles (Mitochondrial Myopathy Profile or Myoglobinuria Profile or Glycogen Storage Disease Profile)	200 mgs. cardiac, skeletal muscle, or liver tissue	Ship overnight frozen on dry ice	50 mg. minimum for individual test requests; (also, Modified Mitochondrial Myopathy Screen requires 50 mgs. total)
Lipid Myopathies: Carnitine	50 mgs. skeletal or cardiac muscle, or 1 mL plasma	Ship overnight frozen on dry ice	Quantitation of total, free and esterified carnitine
Lipid Myopathies: Carnitine palmitoyl-transferase (CPT) II	100 mgs. skeletal muscle	Shipped frozen overnight on dry ice	Quantitation of CPT enzyme activity. Citrate synthase activity is quantified as a mitochondrial marker to use in a ratio analysis with CPT
Mutation analysis (CPT II deficiency; myophosphorylase deficiency; myoadenylate deaminase deficiency)	10 mL EDTA blood (preferred) or 100 mgs. skeletal muscle; pelleted cultured fibroblasts; or lymphoblasts; or isolated DNA acceptable	Blood shipped at room temperature; muscle, cell pellets, or isolated DNA shipped frozen on dry ice (All specimens shipped by overnight carrier)	Analysis of the common mutations causing the disorders described

Coenzyme Q10 Analysis	100 mgs frozen skeletal muscle	Ship overnight frozen on dry ice	Quantitation of Coenzyme Q10 for the diagnosis if primary or secondary deficiency
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