



## Duke University Health System Biochemical Genetics Lab

Mass Spectrometry Section Test Menu and CPT Codes			
Test	Method	Sample Types	CPT Codes
Acylcarnitine Profile	MS/MS	Plasma, serum, dried blood spots, or whole blood	82017
Amino Acid Profile	Ion-exchange HPLC	Plasma, serum, CSF	82139
Amino Acid Profile	Ion-exchange HPLC	Urine	82139,82570
Amino Acid Profile	LC-MS/MS	Dried blood spots	82139
Carnitine Levels (Free & Total)	MS/MS	Plasma, serum or urine	82379 (P,S) 82379,82570 (U)
Combination Carnitine Levels & Acylcarnitine Profile	MS/MS	Plasma or serum	82017,82379
Guanidinoacetate and Creatine	LC-MS/MS	Urine (preferred) and plasma or serum	82542,82540,82570 (U) 82542,82540,82565 (P)
Hex4, Urine (Glucose tetrasaccharide, Glc4)	UPLC-MS/MS	Urine only	82542, 82570
Lyso-Gb <sub>1</sub> (Glucosylsphingosine)	UPLC-MS/MS	Plasma	82542
Lyso-Gb <sub>3</sub> (Lyso-globotriaosylceramide)	UPLC-MS/MS	Plasma	82542
Methylmalonic Acid	UPLC-MS/MS	Plasma or serum	83921
Mucopolysaccharides CS, DS, HS	UPLC-MS/MS	Urine only	83864x2, 82570
Mucopolysaccharide KS	UPLC-MS/MS	Urine only	83864, 82570
Mucopolysaccharides Screen CS, DS, HS, KS	UPLC-MS/MS	Urine only	83864x3, 82570
Organic Acids (Qualitative)	GC-MS	Urine only	83919,82570
S-Sulfocysteine	UPLC-MS/MS	Urine only	82131,82570
Vitamin A & E, Serum	HPLC	Serum only	84446 (E) 84590 (A)
Vitamin A only, Serum	HPLC	Serum only	84590 (A)
Vitamin E only, Serum	HPLC	Serum only	84446 (E)



<b>Glycogen Storage Disease and Enzymology Section Test Menu and CPT Codes</b>			
	<b>Test</b>	<b>Sample Types</b>	<b>CPT Codes</b>
1	<b>GSD Screen</b> - Includes: Glycogen content, Glucose-6-Phosphatase (Liver only), Debranching Enzyme, Total Phosphorylase assay	Liver Muscle	82657 x 2
2	<b>Glycogen Content And Structure</b>	Liver Muscle	82657
3	<b>GSD Type Ia (Von Gierke)*</b> - Glucose-6-Phosphatase	Liver	82657
4	<b>GSD Type II (Pompe disease, acid maltase deficiency)*</b> - Acid alpha-Glucosidase	Blood/Dried blood spot Muscle Fibroblast	82657
5	<b>GSD Type III (Cori / Forbes)*</b> - Debranching Enzyme	Liver  Muscle Other tissue	82657
6	<b>GSD Type IV (Anderson)*</b> - Branching Enzyme	Liver Muscle Fibroblast Other tissue	82657
7	<b>GSD Type V (McArdle)*</b> - Muscle Phosphorylase	Muscle	82657
8	<b>GSD VI (Hers)*</b> - Liver Phosphorylase	Liver	82657
9	<b>GSD Type VII (Tauri)*</b> - Phosphofructokinase	Muscle	82657
10	<b>GSD Type IX*</b> - Phosphorylase Kinase	Liver Blood (RBC) Muscle Heart	82657
11	<b>Fructose Metabolism Disorders</b> -Fructose 1,6, bisphosphatase -Aldolase B	Liver	82657 x 2
12	<b>Fabry disease</b> $\alpha$ -galactosidase A	Whole blood Dried blood spot	82657
13	<b>Gaucher disease</b> - Acid- $\beta$ -glucosidase	Whole blood Dried blood spot	82657
14	<b>LAL-D (Wolman disease / CESD)</b> - Lysosomal acid lipase	Whole blood Dried blood spot	82657
15	<b>MPS 1</b> - Alpha-L-iduronidase	Whole blood Dried blood spot	82657
16	<b>MPS II</b> -Iduronate-2-sulfatase	Whole blood Dried blood spot	82657
17	<b>Cross Reactive Immunological Material (CRIM) Status for GSD Type II (Pompe disease, acid maltase deficiency)</b>	Fibroblast Blood (PBMC)	88372, 88233 88372

METHODOLOGY FOR ENZYME ASSAYS IS BASED ON SPECTROPHOTOMETRY, EXCEPT #4, 12-16 WHICH ARE PERFORMED USING FLUOROMETRY



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For sample requirements see: <https://testcatalog.duke.edu/?lab=123060043>

**Shipping and Mailing Address Monday to Friday, 8 am to 5 pm:**

DUHS Biochemical Genetics Laboratory, 801 Capitola Drive, Suite 6, Durham, NC 27713

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Contact email: [BCG\\_AllStaff@duke.edu](mailto:BCG_AllStaff@duke.edu)

<https://clinlabs.duke.edu/biochemical-genetics-laboratory>

**Shipping address for sending urgent samples outside of regular lab hours, Saturday and Sunday**

DUHS Central Automated Laboratory (DCAL), Attention: Specimen Management

Hudson Bldg., DCAL, Rm 1520, 2351 Erwin Rd, Durham, NC 27705

Contact Number: (919) 681-7786, Hours of Operation (24/7)

**Note: Sending urgent samples outside of regular hours requires prior discussion with and approval by the laboratory directors. Please contact the Biochemical Genetics lab at: [BCG\\_AllStaff@duke.edu](mailto:BCG_AllStaff@duke.edu)**