



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 ₁ (Glucosylsphingosine)	UPLC-MS/MS	Plasma	82542
Lyso-Gb ₃ (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)



Glycogen Storage Disease and Enzymology Section Test Menu and CPT Codes			
	Test	Sample Types	CPT Codes
1	GSD Screen - Includes: Glycogen content, Glucose-6-Phosphatase (Liver only), Debranching Enzyme, Total Phosphorylase assay	Liver Muscle	82657 x 2
2	Glycogen Content And Structure	Liver Muscle	82657
3	GSD Type Ia (Von Gierke)* - Glucose-6-Phosphatase	Liver	82657
4	GSD Type II (Pompe disease, acid maltase deficiency)* - Acid alpha-Glucosidase	Blood/Dried blood spot Muscle Fibroblast	82657
5	GSD Type III (Cori / Forbes)* - Debranching Enzyme	Liver Muscle Other tissue	82657
6	GSD Type IV (Anderson)* - Branching Enzyme	Liver Muscle Fibroblast Other tissue	82657
7	GSD Type V (McArdle)* - Muscle Phosphorylase	Muscle	82657
8	GSD VI (Hers)* - Liver Phosphorylase	Liver	82657
9	GSD Type VII (Tauri)* - Phosphofructokinase	Muscle	82657
10	GSD Type IX* - Phosphorylase Kinase	Liver Blood (RBC) Muscle Heart	82657
11	Fructose Metabolism Disorders -Fructose 1,6, bisphosphatase -Aldolase B	Liver	82657 x 2
12	Fabry disease α -galactosidase A	Whole blood Dried blood spot	82657
13	Gaucher disease - Acid- β -glucosidase	Whole blood Dried blood spot	82657
14	LAL-D (Wolman disease / CESD) - Lysosomal acid lipase	Whole blood Dried blood spot	82657
15	MPS 1 - Alpha-L-iduronidase	Whole blood Dried blood spot	82657
16	MPS II -Iduronate-2-sulfatase	Whole blood Dried blood spot	82657
17	Cross Reactive Immunological Material (CRIM) Status for GSD Type II (Pompe disease, acid maltase deficiency)	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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MASS SPECTROMETRY SECTION ● 801 CAPITOLA DRIVE, SUITE 6, DURHAM, NC 27713
PHONE: (919) 549-0445 ● FAX: (919) 549-0709

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

Main Office Phone: (919) 549-0445; Fax: (919) 549-0709

Contact email: BCG_AllStaff@duke.edu

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

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