



Duke Hospital Biochemical Genetics Lab

Mass Spectrometry Section Test Menu				
	Test	Method	Sample Types	CPT Codes
1	Acylcarnitine Profile	MS/MS	Plasma, Serum, Dried Blood Spots or Whole Blood	82017
2	Amino Acid Profile	Ion-exchange HPLC	Plasma, Serum, Urine or CSF	82139 (P,S,C) 82139,82570 (U)
3	Carnitine Levels	MS/MS	Plasma, Serum or Urine	82379 (P,S) 82379,82570 (U)
4	Carnitine Levels & Acylcarnitine Profile COMBINED	MS/MS	Plasma or Serum	82017,82379
5	Creatine / Guanidinoacetate Assay	LC-MS/MS	Urine (preferred) and Plasma or Serum	82542,82540,82570 (U) 82542,82540,82565 (P)
6	Lyso-globotriaosylceramide (Lyso-Gb ₃)	UPLC-MS/MS	Plasma	82542
7	Methylmalonic Acid	UPLC-MS/MS	Plasma or Serum	83921
8	Mucopolysaccharide (KS)	UPLC-MS/MS	Urine	Contact Lab
9	Mucopolysaccharide (CS, DS, HS)	UPLC-MS/MS	Urine	Contact Lab
10	Mucopolysaccharide Screen (KS, CS, DS, HS)	UPLC-MS/MS	Urine	Contact Lab
11	Organic Acids (Qualitative)	GC-MS	Urine	83919,82570
12	S-Sulfocysteine	UPLC-MS/MS	Urine	82131,82570
13	Urine Hex ₄ (Glucose tetrasaccharide, Glc ₄)	UPLC-MS/MS	Urine	82570, 82542
14	Vitamin A & E, Serum	HPLC	Serum	84446 (E) 84590 (A)
15	Vitamin A only, Serum	HPLC	Serum	84590 (A)
16	Vitamin E only, Serum	HPLC	Serum	84446 (E)



Glycogen and Lysosomal Storage Disease Enzymology Test Menu			
	Test	Sample Type	CPT Codes
1	GSD Screen <ul style="list-style-type: none">Includes: Glycogen content, Glucose-6-Phosphatase (liver only), Debranching Enzyme, Total Phosphorylase assayTests for: GSD types Ia (liver), IIIa (liver and muscle), IIIb (liver), V (muscle), and VI (liver)	Liver, Muscle	82657 x 4
2	Glycogen Content And Structure	Liver, Muscle	82657
3	GSD Type Ia (Von Gierke) <ul style="list-style-type: none">Glucose-6-Phosphatase	Liver	82657
4	GSD Type II (Pompe disease, acid maltase deficiency) <ul style="list-style-type: none">Acid alpha-Glucosidase	Whole blood, Dried blood spot, Muscle, Fibroblasts, Amniocytes, Chorionic Villi Cultures	82657
5	GSD Type II (Pompe disease, acid maltase deficiency) <ul style="list-style-type: none">Cross Reactive Immunological Material (CRIM) status	Fibroblasts	88372, 88233
6	GSD Type III (Cori / Forbes) <ul style="list-style-type: none">Debranching Enzyme	Liver, Muscle	82657
7	GSD Type IV (Andersen) <ul style="list-style-type: none">Branching Enzyme	Liver, Muscle, Fibroblasts, Amniocytes, Chorionic Villi Cultures	82657
8	GSD Type V (McArdle) <ul style="list-style-type: none">Muscle Phosphorylase	Muscle	82657
9	GSD VI (Hers)* <ul style="list-style-type: none">Liver Phosphorylase	Liver	82657
10	GSD Type VII (Tarui) <ul style="list-style-type: none">Phosphofructokinase	Muscle	82657
11	GSD Type IX <ul style="list-style-type: none">Phosphorylase b Kinase	Liver, Whole blood, Muscle, Heart	82657
12	Fructose Assay <ul style="list-style-type: none">Fructose-1-Phosphatase, Fructose 1,6, diphosphatase & Fructose 1-Phosphate Aldolase B	Liver	82657 x 3
13	Fabry Disease <ul style="list-style-type: none">Alpha-Galactosidase A	Whole blood, Dried blood spot	80500, 82657, 84165
14	Gaucher disease <ul style="list-style-type: none">Acid-β-glucosidase	Whole blood, Dried blood spot	82657
15	LAL-D (Wolman disease / CESD) <ul style="list-style-type: none">lysosomal acid lipase	Whole blood, Dried blood spot	82657
16	MPS I <ul style="list-style-type: none">alpha-L-iduronidase	Whole blood, Dried blood spot	82657
17	MPS II <ul style="list-style-type: none">iduronate-2-sulfatase	Whole blood, Dried blood spot	82657

METHODOLOGY FOR ENZYME ASSAYS IS BASED ON SPECTROPHOTOMETRY, EXCEPT #4, 13-16 WHICH ARE PERFORMED USING FLUOROMETRY