BIOCHEMICAL GENETICS LABORATORY ● GLYCOGEN STORAGE DISEASE LABORATORY
DUKE UNIVERSITY HOSPITAL 801 CAPITOLA DRIVE SUITE 6 DURHAM NC 27713
PHONE: (919) 549-0445 ● FAX: (919) 549-0709 ● http://pediatrics.duke.edu/labservices

Glycogen Storage Diseases: Enzyme Assays
TEST REQUEST FORM

PATIENT NAME: _____

PATIENT DOB: _____ **HOSPITAL ID:** _____ **SEX: M / F** (please circle)

DATE AND TIME SAMPLE COLLECTED: _____ **FIRST SAMPLE / REPEAT** (please circle)

DATE SAMPLE SHIPPED: _____ **DATE AND TIME SAMPLE RECEIVED:** _____

Please check appropriate boxes		Acceptable Samples	CPT CODES
❑	**GSD Screen** - Includes: Glycogen content, Glucose-6-Phosphatase (liver only), Debranching Enzyme, Total Phosphorylase assay - Tests for: GSD types Ia (liver), IIIa (liver and muscle), IIIb (liver), V (muscle), and VI (liver)	❑Liver ❑Muscle	82657 x 4
❑	**Glycogen Content And Structure**	❑Liver ❑Muscle	82657
❑	**GSD Type Ia (Von Gierke)*** - Glucose-6-Phosphatase	❑Liver	82657
❑	**GSD Type II (Pompe disease, acid maltase deficiency)*** - Acid alpha-Glucosidase - See also the "Lysosomal Storage Disease Test Request Form" - Please provide clinical details by completing the "Pompe Disease Clinical Information" form	❑ Blood/Dried blood spot ❑Muscle ❑Fibroblast ❑Amniocytes ❑ Chorionic villi cultures	82657
❑	**GSD Type III (Cori / Forbes)*** - Debranching Enzyme	❑Liver ❑Muscle	82657
❑	**GSD Type IV (Andersen)*** - Branching Enzyme	❑Liver ❑Muscle ❑Fibroblast ❑Amniocytes ❑ Chorionic villi cultures	82657
❑	**GSD Type V (McArdle)*** - Muscle Phosphorylase	❑Muscle	82657
❑	**GSD VI (Hers)*** - Liver Phosphorylase	❑Liver	82657
❑	**GSD Type VII (Tarui)*** - Phosphofructokinase	❑Muscle	82657
❑	**GSD Type IX*** - Phosphorylase b Kinase	❑Liver ❑Blood (RBC) ❑Muscle ❑Heart	82657
❑	**Fructose Assay** - Fructose-1-Phosphatase, Fructose 1,6, diphosphatase & Fructose 1-Phosphate Aldolase B	❑Liver	82657 x 3
❑	**CRIM Analysis**	❑ Fibroblast	88372,88233

*Please note – Glycogen content and structure will be performed for all liver and muscle samples arriving in the lab for these assays. Therefore, the total cost will be the cost of the enzyme assay plus the cost of the glycogen content and structure analysis.

Version date: 10/04/2018

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PATIENT INFORMATION FORM

PATIENT NAME: _____

PATIENT DOB: _____ **HOSPITAL ID:** _____ **SEX: M / F** (please circle)

INDICATION FOR TESTING: _____

CLINICAL INFORMATION: Circle all that apply

I. GENERAL PHYSICAL ABNORMALITIES
1. length cm
2. weight kg
3. headcir cm
4. hepatomegaly
5. splenomegaly
6. cardiomegaly
7. skin xanthoma
8. strange smell

II. NEUROMUSCULAR ABNORMALITIES
1. mental retardation
2. muscle weakness
3. exercise intolerance
4. muscle cramping
5. muscle wasting
6. hypertonia
7. hypotonia
8. convulsions
9. lethargy/coma

III. GASTROINTESTINAL ABNORMALITIES
1. vomiting
2. diarrhea

IV. NEPHROLOGICAL ABNORMALITIES
1. creatine clearance
2. proteinuria
3. strange color/smell
4. _____

V. X-RAY ABNORMALITIES
1. delayed bone-age
2. _____

VI. IMMUNOLOGICAL ABNORMALITIES
1. recurrent infections
2. _____

VII. HEMATOLOGICAL ABNORMALITIES
1. anemia
2. neutropenia
3. thrombopenia
4. thrombo-embolic abnormalities
5. bleeding tendency

VIII. LABORATORY ABNORMALITIES
1. acidosis
2. hypoglycemia
3. abnormal liver function
4. ketosis
5. hyperammonemia
6. hyperlipidemia
7. hyperuricemia
8. hyperlactic acidemia
9. high CPK
10. _____

IX. BIOPSY - Glycogen

		Membrane Bound	Dispersed
1	liver	_____	_____
2	muscle	_____	_____

X. GENETICS
1. consanguinity
2. metabolic disease in family
3. pedigree if applicable
4. race

_____White _____ Black
_____Hispanic _____Asian

XI. MEDICATIONS:

_____

_____

XII. DIAGNOSIS:

_____

RESULTS ADDRESS:	***BILLING ADDRESS:**
Physician: _____	Attn: _____
Address: _____	Address: _____
_____	_____
_____	_____
TEL: _____ FAX: _____	TEL: _____ FAX: _____

***We do not bill patients or their insurance companies. You are responsible for charges incurred by tests ordered.**