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**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
<input type="checkbox"/> <b>GSD Screen</b> - 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)	<input type="checkbox"/> Liver <input type="checkbox"/> Muscle	82657 x 4
<input type="checkbox"/> <b>Glycogen Content And Structure</b>	<input type="checkbox"/> Liver <input type="checkbox"/> Muscle	82657
<input type="checkbox"/> <b>GSD Type Ia (Von Gierke)*</b> - Glucose-6-Phosphatase	<input type="checkbox"/> Liver	82657
<input type="checkbox"/> <b>GSD Type II (Pompe disease, acid maltase deficiency)*</b> - Acid alpha-Glucosidase - See also the “Lysosomal Storage Disease Test Request Form” - Please provide clinical details by completing the “Pompe Disease Clinical Information” form	<input type="checkbox"/> Blood/Dried blood spot <input type="checkbox"/> Muscle <input type="checkbox"/> Fibroblast	82657
<input type="checkbox"/> <b>GSD Type III (Cori / Forbes)*</b> - Debranching Enzyme	<input type="checkbox"/> Liver <input type="checkbox"/> Muscle	82657
<input type="checkbox"/> <b>GSD Type IV (Andersen)*</b> - Branching Enzyme	<input type="checkbox"/> Liver <input type="checkbox"/> Muscle <input type="checkbox"/> Fibroblast	82657
<input type="checkbox"/> <b>GSD Type V (McArdle)*</b> - Muscle Phosphorylase	<input type="checkbox"/> Muscle	82657
<input type="checkbox"/> <b>GSD VI (Hers)*</b> - Liver Phosphorylase	<input type="checkbox"/> Liver	82657
<input type="checkbox"/> <b>GSD Type VII (Tarui)*</b> - Phosphofructokinase	<input type="checkbox"/> Muscle	82657
<input type="checkbox"/> <b>GSD Type IX*</b> - Phosphorylase b Kinase	<input type="checkbox"/> Liver <input type="checkbox"/> Blood (RBC) <input type="checkbox"/> Muscle <input type="checkbox"/> Heart	82657
<input type="checkbox"/> <b>Fructose Assay</b> - Fructose-1-Phosphatase, Fructose 1,6, diphosphatase & Fructose 1-Phosphate Aldolase B	<input type="checkbox"/> Liver	82657 x 3
<input type="checkbox"/> <b>CRIM Analysis</b>	<input type="checkbox"/> Fibroblast <input type="checkbox"/> Blood (RBC)	88372,88233
<input type="checkbox"/> <b>Biotinidase</b>	<input type="checkbox"/> Whole blood <input type="checkbox"/> Dried blood spot	82657

\*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.

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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:

\_\_\_\_\_

<b>RESULTS ADDRESS:</b> Physician: _____ Address: _____ _____ TEL: _____ FAX: _____	<b>*BILLING ADDRESS:</b> Attn: _____ Address: _____ _____ TEL: _____ FAX: _____
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\*We do not bill patients or their insurance companies. You are responsible for charges incurred by tests ordered.