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:** ________________

<table>
<thead>
<tr>
<th>Please check appropriate boxes</th>
<th>Acceptable Samples</th>
<th>CPT CODES</th>
</tr>
</thead>
<tbody>
<tr>
<td>GSD Screen</td>
<td>Liver</td>
<td>82657 x 4</td>
</tr>
<tr>
<td></td>
<td>Muscle</td>
<td></td>
</tr>
<tr>
<td>- 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)</td>
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<tr>
<td>Glycogen Content And Structure</td>
<td>Liver</td>
<td>82657</td>
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<tr>
<td></td>
<td>Muscle</td>
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<tr>
<td>GSD Type Ia (Von Gierke)*</td>
<td>Liver</td>
<td>82657</td>
</tr>
<tr>
<td>- Glucose-6-Phosphatase</td>
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<tr>
<td>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</td>
<td>Blood/Dried blood spot</td>
<td>82657</td>
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<tr>
<td></td>
<td>Muscle</td>
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<tr>
<td></td>
<td>Fibroblast</td>
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<td></td>
<td>Amniocytes</td>
<td></td>
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<td></td>
<td>Chorionic villi cultures</td>
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<tr>
<td>GSD Type III (Cori / Forbes)*</td>
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<tr>
<td>- Debranching Enzyme</td>
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<tr>
<td>GSD Type IV (Andersen)*</td>
<td>Liver</td>
<td>82657</td>
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<tr>
<td>- Branching Enzyme</td>
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<td>Muscle</td>
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<td></td>
<td>Chorionic villi cultures</td>
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<td>GSD Type V (McArdle)*</td>
<td>Muscle</td>
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<tr>
<td>- Muscle Phosphorylase</td>
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<tr>
<td>GSD VI (Hers)*</td>
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<tr>
<td>- Liver Phosphorylase</td>
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<tr>
<td>GSD Type VII (Tarui)*</td>
<td>Muscle</td>
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<tr>
<td>- Phosphofructokinase</td>
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<tr>
<td>GSD Type IX*</td>
<td>Liver</td>
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<tr>
<td>- Phosphorylase b Kinase</td>
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<tr>
<td>Fructose Assay</td>
<td>Blood (RBC)</td>
<td>82657</td>
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<tr>
<td>- Fructose-1-Phosphatase, Fructose 1,6, diphosphatase &amp; Fructose 1-Phosphate Aldolase B</td>
<td>Muscle</td>
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<td>Heart</td>
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<tr>
<td>CRIM Analysis</td>
<td>Fibroblast</td>
<td>88372,88233</td>
</tr>
</tbody>
</table>

*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
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

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

VI. IMMUNOLOGICAL ABNORMALITIES
1. recurrent infections

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

IX. BIOPSY - Glycogen
1. Membrane Bound
2. Dispersed

III. GASTROINTESTINAL ABNORMALITIES
1. liver
2. muscle

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

XI. MEDICATIONS:

XII. DIAGNOSIS:

RESULTS ADDRESS:
Physician: ____________________________________________
Address: ____________________________________________
________________________________________________________________________
________________________________________________________________________
TEL: ___________  FAX: ___________

*BILLING ADDRESS:
Attn: ____________________________________________
Address: ____________________________________________
________________________________________________________________________
________________________________________________________________________
TEL: ___________  FAX: ___________

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