## 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 2</td>
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<tr>
<td></td>
<td>Muscle</td>
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<td>Liver</td>
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<td>Liver</td>
<td>82657</td>
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<td>Muscle</td>
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<tr>
<td>❌ GSD Type Ia (Von Gierke)*</td>
<td>Liver</td>
<td>82657</td>
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<td></td>
<td>Muscle</td>
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<tr>
<td>❌ GSD Type II (Pompe disease, acid maltase deficiency)*</td>
<td>Blood/Dried blood spot</td>
<td>82657</td>
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<td></td>
<td>Muscle</td>
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<tr>
<td></td>
<td>Fibroblast</td>
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<tr>
<td>❌ GSD Type III (Cori / Forbes)*</td>
<td>Liver</td>
<td>82657</td>
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<td>Muscle</td>
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<tr>
<td>❌ GSD Type IV (Andersen)*</td>
<td>Liver</td>
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<td>Muscle</td>
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<tr>
<td></td>
<td>Fibroblast</td>
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<tr>
<td>❌ GSD Type V (McArdle)*</td>
<td>Muscle</td>
<td>82657</td>
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<tr>
<td>❌ GSD VI (Hers)*</td>
<td>Muscle</td>
<td>82657</td>
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<tr>
<td>❌ GSD Type VII (Tarui)*</td>
<td>Muscle</td>
<td>82657</td>
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<tr>
<td>❌ GSD Type IX*</td>
<td>Liver</td>
<td>82657</td>
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<tr>
<td></td>
<td>Blood (RBC)</td>
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<td>Muscle</td>
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<td></td>
<td>Heart</td>
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<tr>
<td>❌ Fructose Metabolism Disorders</td>
<td>Liver</td>
<td>82657 x 2</td>
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<tr>
<td>❌ GAA CRIM Analysis</td>
<td>Fibroblast</td>
<td>88372 (both tissue types)</td>
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<tr>
<td></td>
<td>Blood (PBMC)</td>
<td>88233 (fibroblasts only)</td>
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<tr>
<td>❌ Biotinidase</td>
<td>Whole blood</td>
<td>82657</td>
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<tr>
<td></td>
<td>Dried blood spot</td>
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</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.
**Patient Information Form**

**Patient Name:** __________________________________________

**Patient DOB:** _______________ **Hospital ID:** _______________ **Sex:** M / F (please circle)

**Indication for Testing:** ______________________________________

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**Clinical Information:** Circle all that apply

**I. General Physical Abnormalities**

1. length cm   
2. weight kg   
3. head 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   
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:**

__________________________

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**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.*

Version date: 11-30-2022