ARUP LABORATORIES

*A nonprofit enterprise of the University of Utah and its Department of Pathology*

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**THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.**

# GLYCOGEN STORAGE DISORDER TESTING PATIENT HISTORY FORM

**Patient Name:** _____ **Date of Birth:** _____ **Sex:** ☐ Female ☐ Male

**Ordering Provider:** _____ **Provider's Phone:** _____

**Practice Specialty:** _____ **Provider's Fax:** _____

**Genetic Counselor:** _____ **Counselor Phone:** _____

**Patient's Ethnicity/Ancestry** (check all that apply)
☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____
**List country of origin (if known):** _____

**Clinical diagnosis of glycogen storage disorder (GSD):** ☐ Confirmed ☐ Suspected ☐ Unknown

☐ **GSD type 0**
☐ liver ☐ muscle ☐ unknown
☐ **GSD type I (von Gierke disease)**
☐ Ia ☐ Ib ☐ unknown
☐ **GSD type II (Pompe disease)**
☐ infantile onset ☐ late onset
☐ **GSD type III (Forbes-Cori disease)**
☐ IIIa ☐ IIIb ☐ IIIc ☐ IIId
☐ unknown
☐ GSD type IV (Andersen disease)
☐ GSD type V (McArdle disease)
☐ GSD type VI (Hers disease)
☐ GSD type VII (Tarui disease)
☐ **GSD type IX**
**(phosphorylase kinase deficiency)**
☐ IXa ☐ IXb ☐ IXc ☐ IXd
☐ unknown
☐ GSD type X
☐ GSD type XI
(Fanconi-Bickel syndrome)
☐ GSD type XII
(aldolase A deficiency)
☐ GSD type XIII
☐ GSD type XV
☐ Other (please specify): _____

**Symptoms:** ☐ Yes ☐ No ☐ Unknown **Age of symptom onset:** _____

☐ Hepatomegaly ☐ Splenomegaly ☐ Jaundice ☐ Other symptom(s): _____
☐ Muscle weakness ☐ Failure to thrive/poor growth ☐ Liver cirrhosis
☐ Exercise intolerance ☐ Cardiomyopathy ☐ Rickets
☐ Muscle pain/stiffness/cramps ☐ Delayed puberty ☐ Cardiac arrhythmia

**Laboratory findings:**

| | | | | |
|---|---|---|---|---|
| Newborn Screen: | ☐ Normal | ☐ Abnormal; describe: _____ | ☐ Unknown | |
| Serum creatine kinase: | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Blood glucose: | | | ☐ Fasting | ☐ Not fasting |
| | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Cholesterol: | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Plasma acylcarnitines: | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Blood uric acid: | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Uric acid: | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Urine organic acids: | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Liver enzymes (AST/ALT): | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Blood lactate: | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Triglycerides: | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |
| Imaging studies (MRI/ultrasound) | ☐ Normal | ☐ Abnormal _____ | ☐ Unknown | ☐ Not performed |

☐ Other: _____

**Has the patient undergone previous DNA testing?** ☐ No ☐ Yes ☐ Unknown
If yes, describe the test(s) and results: _____

**Is there any relevant family history of GSD?** ☐ No ☐ Yes ☐ Unknown
If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:

**Has DNA testing been performed for the family member(s)?** ☐ No ☐ Yes ☐ Unknown
**If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).**

**Check the test you intend to order.**

☐ **3001627 Glycogen Storage Disorders:** Preferred molecular test for confirming a suspected clinical diagnosis of GSD.
☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a sequence variant previously identified in a family member; a copy of a relative's lab result is REQUIRED.
☐ **3003144 Deletion/Duplication Analysis by MLPA:** Tests for large deletion/duplication previously identified in a family member; a copy of a relative's lab report is REQUIRED.

**For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.**

**Master Label**