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 Assigned at Birth: ☐Female ☐Male ☐Intersex **Gender Identity (optional):** ☐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 variant testing).

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

Master Label