BIOCHEMICAL GENETICS LABORATORY ● GLYCOGEN STORAGE DISEASE LABORATORY
DUKE UNIVERSITY HOSPITAL 801 CAPITOLA DRIVE SUITE 6 DURHAM NC 27713
PHONE: (919) 549-0445 ● FAX: (919) 549-0709 ● http://pediatrics.duke.edu/labservices

DNA SEQUENCING TEST REQUEST FORM

PATIENT NAME: _____

PATIENT DOB: _____ **HOSPITAL ID:** _____ **SEX: M / F** (please circle)

DATE AND TIME SAMPLE COLLECTED: _____

DATE SAMPLE SHIPPED: _____ **DATE AND TIME SAMPLE RECEIVED:** _____

Please check appropriate boxes		Sample type	CPT CODES
❑	**GSD type 1a (Von Gierke disease)** - Glucose-6-phosphatase (*G6PC*) gene - Full gene sequencing	❑ Blood ❑ Amniocytes	83891, 81404
❑	**GSD type 1b** - Glucose-6-phosphate translocase (*SLC37A4*) gene - Full gene sequencing	❑ Blood ❑ Amniocytes	83891, 81404
❑	**GSD II (Pompe disease, acid maltase deficiency)** - Acid alpha-glucosidase (*GAA*) gene - Full gene sequencing	❑ Blood ❑ Fibroblast ❑ Amniocytes ❑ Chorionic Villi	81406
❑	**GSD III (Cori Disease, Forbes Disease)** - Glycogen debranching enzyme (*AGL*) gene - Full gene sequencing	❑ Blood ❑ Fibroblast ❑ Amniocytes ❑ Chorionic Villi	81407
❑	**GSD IV (glycogen branching enzyme deficiency)** - Glycogen branching enzyme (*GBE1*) gene - Full gene sequencing	❑ Blood ❑ Fibroblast ❑ Amniocytes ❑ Chorionic Villi	83891, 81406
❑	**GSD V (McArdle disease)** - Muscle glycogen phosphorylase (*PYGM*) gene - Full gene sequencing	❑ Blood	83891, 81406
❑	**GSD IX (phosphorylase kinase deficiency)** ❑ *PHKA2* full gene sequencing ❑ *PHKG2* full gene sequencing	❑ Blood ❑ Amniocytes ❑ Chorionic Villi	*PHKA2*: 83891, 81407 *PHKG2*: 81405
❑	**Fabry disease** - alpha-galactosidase (*GLA*) Gene - Full gene sequencing	❑ Blood ❑ Amniocytes	81405
❑	**MPSI disease** - Iduronidase (IDUA) - Full gene sequencing	❑ Blood ❑ Amniocytes	81405
❑	**GSD PANEL sequencing (25 gene panel)** **Targeted mutation testing:** **Single EXON** **Two EXONS** ***Please send documentation**	❑ Blood	Single exon: 81403 (for all genes) Two exons: 81404 (for all genes) **Phone: 919-684-2698**

Sequencing analyses are performed by the Duke Molecular Diagnostics laboratory.
Phone: 919-684-2698
Samples can be sent to the GSD Laboratory and will be forwarded.

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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:
_____

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.**