DUKE UNIVERSITY HEALTH SYSTEM ● BIOCHEMICAL GENETICS LABORATORY ● GLYCOGEN STORAGE DISEASE LABORATORY ● 801 CAPITOLA DRIVE SUITE 6, DURHAM, NC 27713 ● PHONE: (919) 549-0445 ● FAX: (919) 549-0709

# LYSOSOMAL STORAGE DISEASES 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:** _____

**Enzyme assays**

| Please check appropriate boxes | | Acceptable Samples | CPT CODES |
|---|---|---|---|
| ❑ | **GSD Type II (Pompe disease, acid maltase deficiency)** - Acid α-glucosidase deficiency | ❑Whole blood ❑Dried blood spot ❑Muscle ❑Fibroblast | 82657 |
| ❑ | **Fabry disease** α -Galactosidase A deficiency | ❑Whole blood ❑Dried blood spot | 82657 |
| ❑ | **Gaucher disease** Acid-β-glucosidase deficiency | ❑Whole blood ❑Dried blood spot | 82657 |
| ❑ | **MPS 1** Alpha-L-iduronidase deficiency | ❑Whole blood ❑Dried blood spot | 82657 |
| ❑ | **MPS II** Iduronate-2-sulfatase deficiency | ❑Whole blood ❑Dried blood spot | 82657 |
| ❑ | **LAL-D (Wolman Disease)** Acid lipase deficiency | ❑Whole blood ❑Dried blood spot | 82657 |

**Cross Reactive Immunological Material (CRIM) status**

| Please check appropriate boxes | | Acceptable Samples | CPT CODES |
|---|---|---|---|
| ❑ | **GSD Type II (Pompe disease, acid maltase deficiency)** Acid alpha-glucosidase CRIM | ❑Fibroblast ❑Blood (PBMC) | 88372, 88233 |

**Biomarker assays**

| Please check appropriate boxes | | Acceptable Samples | CPT CODES |
|---|---|---|---|
| ❑ | **GSD Type II (Pompe disease, acid maltase deficiency)** - $Hex_4$ (glucose tetrasaccharide, $Glc_4$) | ❑Urine | 82542, 82570 |
| ❑ | **Fabry disease** Lyso-$Gb_3$ (lyso-globotriaosylceramide) | ❑Plasma (EDTA) | 82542 |
| ❑ | **Gaucher disease** Lyso-Gb1 (glucosylsphingosine) | ❑Plasma (EDTA) | 82542 |
| ❑ | **MPS I and II** Mucopolysaccharides CS, DS, HS (glycosaminoglycans: chondroitin sulfate, dermatan sulfate, heparan sulfate by LC-MS/MS) | ❑Urine | 83864 x 2<br>82570 |

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

| **RESULTS ADDRESS:** | ***BILLING ADDRESS:** |
|---|---|
| Physician: _____ | Attn: _____ |
| Address: _____ | Address: _____ |
| _____ | _____ |
| _____ | _____ |
| TEL: _____ FAX: _____ | TEL: _____ FAX: _____ |

Version date: 11-30-2022

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# DISEASE: CLINICAL INFORMATION FORM

**PATIENT NAME:** _____

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

## INDICATION FOR TESTING:

_____

_____

## CLINICAL INFORMATION (please check/circle all that apply)

### LABORATORY ABNORMALITIES:
CPK (value / date):_____
AST (SGOT): _____
ALT (SGPT):_____

### HEMATOLOGY:
Date of any Transfusions _____

### NEUROMUSCULAR ABNORMALITIES:
____ Proximal muscle weakness
____ Exercise intolerance
____ Muscle Wasting
____ Hypotonia
____ Lower back pain
____ Use of ambulatory assistance (circle) cane / walker / wheelchair
____ Age muscle weakness first noted

### CARDIAC ABNORMALITIES:
____ Cardiomyopathy
____ Cardiomegaly
____ Cardiac Arrhythmias

### PATHOLOGY:
Muscle biopsy: _____
Date of biopsy: _____
Location of biopsy: _____
Pertinent findings: _____
Glycogen: ___ Membrane /Lysosomal Bound

### RESPIRATORY NEEDS :
____ Oxygen use via nasal canula
____ Non invasive vent (BiPap, CPap): Day / Night / Both
____ Ventilator required: Day / Night / Both

### FAMILY / MEDICAL HISTORY:
____ Consanguinity
____ Family history of metabolic disease**
Race_____
**Please attach a pedigree if applicable

### TREATMENT:
____ On ERT
CRIM status: Positive/Negative
Frequency of infusion: Weekly / Biweekly
Dose: _____mg/kg/week
_____ Immunomodulation
Regimen:_____

| RESULTS ADDRESS: | *BILLING ADDRESS: |
|---|---|
| Physician: _____ | Attn: _____ |
| Address: _____ | Address: _____ |
| _____ | _____ |
| _____ | _____ |
| TEL: _____FAX:_____ | TEL: _____FAX:_____ |

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