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 ● https://clinlabs.duke.edu/bemical-

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	❑Whole blood ❑Dried blood spot ❑Muscle ❑Fibroblast	82657
❑	**Fabry disease** - α -galactosidase A	❑Whole blood ❑Dried blood spot	82657
❑	**Gaucher disease** - acid-β-glucosidase	❑Whole blood ❑Dried blood spot	82657
❑	**MPS 1** - alpha-L-iduronidase	❑Whole blood ❑Dried blood spot	82657
❑	**MPS II** - iduronate-2-sulfatase	❑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)**	❑Fibroblast ❑Whole blood	88372, 88233

Biomarker assays

Please check appropriate boxes		Acceptable Samples	CPT CODES
❑	**GSD Type II (Pompe disease, acid maltase deficiency)** - Hex_4 (glucose tetrasaccharide)	❑Urine	82542, 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: 05-05-2022

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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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