Pompe Disease Dried Blood Spot Testing and GAA Sequencing Program Testing Requisition Form

Glycogen Storage Disease Laboratory, Pediatric Biochemical Genetics Laboratory
Duke University Hospital

PATIENT INFORMATION

Last Name _____ First Name _____ MI_____

DOB____/____/____ Gender: _____ Ethnicity of patient: ☐Caucasian/NW European ☐Hispanic
dd mmm yyyy male / female (Check all that apply) ☐Asian ☐African-American ☐Indian
☐Native American ☐Eastern European
☐Mediterranean Other_____

Required clinical information:

muscle weakness present: _____. If yes, please describe: _____
yes / no proximal / distal / limb girdle / LGMD, etc.

muscle wasting: _____ hypotonia:_____ exercise intolerance:_____ lower back pain: _____
yes / no yes / no yes / no yes / no

cardiomyopathy/cardiomegaly:_____ cardiac arrhythmia:_____ hepatomegaly:_____
yes / no / unknown yes / no / unknown yes / no / unknown

respiratory insufficiency: _____ If yes; _____
yes / no BiPap / CPap / mechanical ventilator

Has a muscle biopsy been performed? _____ If yes, please summarize results:_____
glycogen present, membrane bound, UNK

Family history of Pompe disease? _____ *If patient is part of a known Pompe family, please attach pedigree.
yes* / no / unknown

Other relevant clinical information: _____

SAMPLE INFORMATION

Sample requirements: **3-5mL whole blood in sodium-EDTA** (purple-top) tube.

Please indicate your testing preferences: ☐ DBS for GAA enzyme assay and GAA gene sequencing if indicated**

** If enzyme activity is decreased, GAA gene sequencing may be indicated to confirm diagnosis. **You must** indicate above if you wish for reflex GAA gene sequencing to be done if necessary.

Date sample collected: ____/_____/_____ Time sample collected: ____:____ AM PM
DD MMM YYYY

Sample should be shipped overnight with a cold pack. If unable to ship on the day sample is drawn, please keep at 4°C until able to be shipped. Ship samples Monday through Thursday only. **Do not ship samples on Fridays**; no weekend deliveries accepted. **Be sure to include this requisition form with your sample!**

Ship to: Glycogen Storage Disease (GSD) Laboratory
Biochemical Genetics Laboratories
Attn: Deeksha Bali, PhD – Pompe DBS Program
Duke Hospital
801 Capitola Drive, Suite 6
Durham, NC 27713

If questions please contact:

Deeksha Bali, PhD
Phone: 919-684-0025
Deeksha.Bali@duke.edu

or

Gwen Dickerson
Phone: 919-684-0338
gharmon@duke.edu

http://medgenetics.pediatrics.duke.edu

PHYSICIAN ORDERING TEST:

Name and Specialty: _____

Institution / address: _____

_____

City: _____ State: _____ Zip: _____

Phone: (_____)_____-_____ Fax: (_____)_____-_____

Email: _____

Duplicate report to: _____
Physician name & clinic

Phone: (_____) _____-_____

Fax: (_____) _____-_____

BILLING INFORMATION:

Are you an MDA-affiliated physician? ____ yes*
____ no

* If you are an MDA physician, you *must* provide billing info for your **local MDA office** below:

MDA Billing address:

Name: _____

Address: _____

_____

Phone: (_____) _____-_____

Fax: (_____) _____-_____