ABOUT THE POMPE TESTING PROGRAM:
Pompe disease (also known as Acid Maltase Deficiency or Glycogen Storage Disease, type II) is a progressive and often fatal neuromuscular disorder with symptoms that can mimic other metabolic myopathies. Making the correct diagnosis is a critical step toward optimizing your patient’s care.

The Duke Glycogen Storage Disease (GSD) Laboratory offers non-invasive and free testing through the Pompe Disease Dried Blood Spot (DBS) and GAA Sequencing Program. This program is supported through a grant provided by Genzyme Corporation.

HOW IS POMPE DISEASE DIAGNOSED?
• Pompe disease occurs as the result of deficient acid alpha glucosidase (GAA) activity, caused by a genetic mutation in the GAA gene.

• Blood-based GAA enzyme testing is an initial screening tool for Pompe disease. GAA enzyme testing can be performed using either a dried blood spot sample (DBS) or whole blood. Both are considered a very effective screening method. GAA enzyme activity levels in the normal range exclude the diagnosis of Pompe disease.

• If your patient’s GAA enzyme activity shows deficient or low-level activity, then full gene GAA sequencing is required in order to confirm the diagnosis of Pompe disease. Whole blood is required for full gene sequencing.

IS THERE A TEST KIT AVAILABLE?
Pompe disease test kits are available if needed. Test kits include materials for both whole blood and dried blood spot testing as well as pre-paid shipping label and cold-pak.

If you would like to have a test kit sent to you at no charge, please place your order online at: [https://lsdtesting.myregistrationp.com](https://lsdtesting.myregistrationp.com) or call toll free 877-873-2753.

AM I REQUIRED TO USE THE DUKE TEST KIT?
Providers may collect and send samples without using our Pompe Test Kit. **However – in order for the testing to be done at no cost, each sample MUST be accompanied by a signed, Pompe Disease Testing Program Requisition Form available from your local Genzyme representative.**
**HOW DO I TEST MY PATIENTS?**

There are two different testing methods available:

- **Whole blood:** This is the preferred sample type. Whole blood allows us to test both enzyme activity and full GAA gene sequencing for confirmation using the same, single blood sample.

**HOW LONG DOES IT TAKE TO GET RESULTS?**

- Results of enzyme testing will be provided via electronic fax within 10 days.

- If your patient’s sample is in the positive or indeterminate range for Pompe disease, the sample will be processed for full gene sequencing. Copies of the sequencing results will be provided via electronic mail within 2-3 weeks.

- If you need to have a copies of the results faxed to you, please contact the lab at 919-684-0338 or 919-684-2722 or gwen.dickerson@duke.edu; dbali@duke.edu

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