Pompe Disease

Dried Blood Spot Testing and GAA Sequencing Program

About the Pompe Testing Program

Pompe disease (also known as Acid Maltase Deficiency) is a progressive and often fatal neuromuscular disorder with symptoms that can mimic other metabolic myopathies. Making the diagnosis is an important 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) testing and GAA Sequencing Program. This program is supported through a grant provided by Genzyme Corporation.

How do I test my patients?

- Collect 3 – 5 mL whole blood in an EDTA (purple-top) tube.
- Complete the requisition form (available from your local Genzyme representative).
- Send the sample to the address provided on the requisition form within 24 hours of collection:
  - If sample is collected on a Friday, please store at 4°C through the weekend and ship sample on the following Monday with a cold-pak enclosed.
  - The Pompe Disease Dried Blood Spot Testing and GAA Sequencing Program does not cover the costs associated with obtaining and shipping the sample.
- If a patient's GAA enzyme activity testing on blood sample is found to be low, the ordering physician will be notified via e-mail or phone call to recommend follow up testing through GAA full gene sequencing. If a patient's DBS tests negative, you will receive results via the mail within 10 days.

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Why does the test requisition form ask me to indicate my testing preferences?

Blood-based GAA enzyme testing is an initial screening test for Pompe disease. Dried blood spot (DBS) samples testing positive for GAA deficiency must be confirmed using GAA full gene sequencing for definitive diagnosis.

Initial testing performed on the patient's blood sample will be DBS based GAA enzyme activity measurement. Samples testing in the normal range in enzyme activity exclude the diagnosis of Pompe disease.

If your patient's GAA enzyme activity test shows deficiency or low-level activity, follow-up testing through GAA full gene sequencing is recommended for confirmation of diagnosis. There is no need to send an additional sample; sequencing can be performed using blood from the original sample.

If you would like GAA gene sequencing to be done as a reflective test for confirmation of diagnosis, **please indicate this by ordering both tests on the requisition form**.

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The Duke University GSD Laboratory specializes in the enzymatic and molecular diagnosis of glycogen storage disorders, disorders of fructose metabolism, and lysosomal storage diseases. The laboratory is certified by the College of American Pathologists (CAP) and Clinical Laboratory Improvement Amendments (CLIA) and is staffed by highly trained, licensed professionals and laboratory personnel. The latest advances in diagnostic technologies are utilized to provide physicians with an extensive menu of biochemical, enzyme and molecular tests. The laboratories work closely with the Division's board-certified medical geneticists and genetic counselors to ensure timely interpretation of laboratory results for health care professionals and their patients.

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