
Lab Dept: **Anatomic Pathology**

Test Name: **GAA ENZYME ACTIVITY**

General Information

Lab Order Codes: GAA

Synonyms: Acid Maltase Activity (acid α -glucosidase, GAA) for Pompe Disease; GAA enzyme activity assays; GSD Type II (Pompe disease, acid maltase deficiency)

CPT Codes: 82657 – Enzyme Activity in blood cell, not elsewhere specified, each specimen

Test Includes: Testing includes Acid Maltase Activity (acid α -glucosidase, GAA)

Logistics

Test Indications: Patients with clinical symptoms consist with Pompe disease or deficient GAA enzyme activity as well as individuals with a family history of Pompe disease.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Duke University Molecular Diagnostics Laboratory

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Restricted Draw Time: Draw Monday thru Thursday before 2pm

Turnaround Time: Results within 14 days

Special Instructions: Restricted draw times. See [Test Availability](#).
<http://pediatrics.duke.edu/divisions/medical-genetics> click on the link & select Glycogen storage disease Lab link on the right under Laboratory services. Click on Test Request Form and then Pompe Disease Test Request Form.

Specimen

Specimen Type: Whole blood

Container: Lavender (EDTA) top tube

Draw Volume:	3 mL (Minimum: 1 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not centrifuge. Forward unprocessed peripheral blood promptly to Duke laboratory at ambient temperatures. Storage greater than 24 hours should be refrigerated.
Patient Preparation:	None
Sample Rejection:	Sample not received by reference lab within 48 hours of draw; mislabeled or unlabeled specimens; frozen specimens; specimens other than EDTA whole blood

Interpretive

Reference Range:	>9.9 pmol/punch/hour An interpretive report will be provided.
Critical Values:	N/A
Limitations:	N/A
Methodology:	GAA enzyme activity and glycogen content measured directly in tissue homogenates and compared with the established positive and negative controls.
References:	Duke University Molecular Diagnostics Laboratory January 2018 Phone: 919-684-2698 Fax: 919-688-5424