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:	Daily, 24 hours
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. Specimens collected on Friday or over the weekend should be stored refrigerated and sent on Monday morning or the next business day if a holiday lands on a Monday.
Patient Preparation:	None
Sample Rejection:	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 March 2020 Phone: 919-684-2698 Fax: 919-688-5424
Updates:	3/30/2020: Removed specimen collection restrictions.