Pompe Disease: Acid Alpha-Glucosidase Enzyme Activity, Leukocytes

**Condition Description**

Pompe disease, also called glycogen storage disease type II (GSD-II), is an autosomal recessive disorder due to a deficiency of the lysosomal enzyme acid alpha-1,4-glucosidase (abbreviated GAA). The function of the GAA enzyme, also known as acid maltase, is to breakdown glycogen in the lysosome. Absent or reduced GAA activity results in accumulation of glycogen within the lysosome, particularly in muscle cells. GSD-II is divided into two forms; an infantile form and a juvenile/adult onset form. In individuals with the infantile form of Pompe disease there is less than 1% of normal enzymatic activity, whereas in the juvenile/adult onset form there is some residual enzymatic activity. In Pompe disease, affected infants are severely hypotonic and have cardiomegaly. In addition, patients may have an enlarged tongue. The disease is usually fatal within the first year of life due cardiorespiratory failure. The clinical presentation in the juvenile/adult onset form (onset after 12 months of age) is much more variable than in the Infantile form of Pompe disease. In this later onset form of the disease, patients generally suffer from slowly progressive proximal muscle weakness with progressive respiratory insufficiency. Unlike the infantile form, in the later onset form there is usually not cardiomegaly or cardiomyopathy.

For questions about testing for Pompe disease, call EGL Genetics at 470-378-2200. For further clinical information about lysosomal storage diseases, including management and treatment, call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524.


**Reference:**
1) [www2.eur.nl/fgg/ch1/pompe](http://www2.eur.nl/fgg/ch1/pompe)

**Indications**

Confirmation of a clinical diagnosis of GSD II

**Methodology**

Flurometric Enzyme Assay using an artificial substrate (4-MU-a-glucopyronside) to evaluate acid alpha-glucosidase activity to confirm a diagnosis of GSD II (Pompe) disease.

**Detection**

- Dependent on level of clinical suspicion
- The amount of enzyme activity in blood or dried blood spots is reliable in diagnosing Pompe disease, but can not accurately predict or distinguish between the two subtypes of Pompe disease (infantile and juvenile/adult)
- This test will not detect carriers of this condition.

**Specimen Requirements**

**Type: Whole Blood**

Specimen Requirements:

In sodium heparin (green top) tube: 3-5 ml

Specimen Collection and Shipping: Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze.

**Related Tests**

Glycogen Storage Disease Type II (Pompe): Gene Sequencing (AN)
Known Mutation Analysis (KM) is available to test family members.