

Positive Result: Blood Spot Screen Result Notification

Minnesota Newborn
Screening Program



Absent/Reduced Acid Alpha-Glucosidase (GAA)

This screening result is suggestive of a biochemical diagnosis of Pompe disease. Molecular genetic testing is pending and MDH will provide the results to the metabolic specialist seeing your patient.

Next Steps

Today, you should take the following recommended actions:

- **Consult** with metabolic specialist. Contact information for the metabolic specialists can be found on the newborn screening report and on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms.
- **Evaluate** infant (hypotonia, feeding difficulties, evidence of cardiac failure); arrange immediate referral if symptomatic.
- **Arrange** referral to metabolic specialist for a comprehensive evaluation.

If you have questions about the newborn screening result or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

Review with Family

Discuss this result with the family as MDH has **not** notified them. Share your follow-up plan with them. Educate family about signs, symptoms, and when to contact you with concerns.

Differential Diagnosis

Absent/reduced GAA is primarily associated with:

- Pompe disease — Incidence of 1 in 28,000

Clinical Summary

Pompe disease is a lysosomal disorder caused by a deficiency in the enzyme acid alpha-glucosidase (GAA). As a result of this deficiency, glycogen accumulates—primarily in cardiac and skeletal muscle.

There are three types with wide variability: classic infantile-onset, non-classic infantile-onset, and late-onset. The classic infantile-onset is the most severe type and presents within a few months of birth. Infants develop profound weakness and cardiomyopathy, leading to death in the first year of life if untreated. The non-classic infantile-onset type usually appears by age one. It is characterized by delayed motor skills and progressive muscle weakness. The late-onset type is associated with progressive weakness and respiratory failure, with highly variable onset and progression.

Some individuals can have low enzyme activity without developing disease—referred to as “pseudodeficiency.”

Enzyme replacement therapy (ERT) is available and has been shown to significantly modify the course of the infantile-onset disease with early treatment producing better outcomes.