MAYO CLINIC LABORATORIES

Newborn Screening Act Sheet

Pompe Disease: Decreased Acid Alpha-Glucosidase

Condition Description: Pompe disease is a lysosomal storage disorder (LSD) caused by a defect in acid alpha-glucosidase (GAA), resulting in glycogen accumulation primarily in cardiac and skeletal muscle. There is wide variability in severity and age of onset. Pompe disease is an autosomal recessive disorder.

Medical Emergency-Take the following actions:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, muscle weakness, respiratory concerns).
- Consult with genetic or metabolic specialist.
- Evaluate the newborn for signs of muscle and/or heart disease (hypotonia, generalized muscle weakness, feeding difficulties, respiratory distress). If any sign is present or infant is ill, transport to hospital for further evaluation and treatment in consultation with metabolic specialist.
- Initiate timely confirmatory/diagnostic testing and management, as recommended by specialist.
- Provide family with basic information about Pompe disease.

Diagnostic Evaluation: Confirmatory GAA enzyme assay, muscle enzymes (CK, LDH, AST, ALT), urine glucotetrasaccharides, and, if clinically indicated, assessment for cardiomyopathy (ECG, ECHO). When patients have low enzyme activity, *GAA* gene analysis and other laboratory studies may be required in consultation with the pediatric metabolic specialist.

Clinical Expectations: The clinical presentation of Pompe disease ranges from a rapidly progressive infantile variant, which is uniformly lethal if untreated, to a more slowly progressive late-onset variant. All disease variants are eventually associated with progressive muscle weakness and respiratory insufficiency. Cardiomyopathy is associated almost exclusively with the infantile form. Pompe disease is caused by mutations in the *GAA* gene and has an estimated incidence of approximately 1 in 15,000 live births. Enzyme replacement therapy (ERT) is available for all variants and should be started as soon as possible for patients with the infantile variant and at the first signs of muscle weakness in the later onset variants. ERT administration is highly complicated and should only be given under the guidance of a specialist with expertise in LSD.

Additional Information

Genetics Home Reference

Genetic Testing Registry

Baby's First Test

Mayo Clinic Laboratories Testing

PD2T / Pompe Disease Second Tier Newborn Screening, Blood Spot

HEX4 / Glucotetrasaccharides, Urine

GAAZ / Pompe Disease, Full Gene Analysis