Suggested Follow-up for Pompe Disease
(Decreased or absent Acid Alpha-Glucosidase)

Condition Description:

Pompe disease is a lysosomal storage disorder (LSD) caused by a defect in the acid alpha-glucosidase (*GAA*) gene. This results in glycogen accumulation, primarily in cardiac and skeletal muscle. There is wide variability in severity and age of onset.

Note: *Pompe disease is an autosomal recessive disorder. Pompe carriers and patients with pseudo-deficiency alleles may also be identified in the screening test.*

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Discuss the newborn screening results with the family.
- Provide the family with basic information about Pompe disease. The handout "*When Baby has an abnormal test for Pompe Disease*" may be used for this general purpose.
- Evaluate the newborn with attention to muscle weakness, hypotonia, feeding difficulties, and any clinical evidence of heart disease.
- Contact a pediatric metabolic specialist, if indicated.
- Report patient outcomes and final diagnosis to the SC DHEC Newborn Screening Program.

Diagnostic Evaluation:

Ancillary enzymology (CK, LDH, AST, ALT) determination, confirmatory α-glucosidase enzyme assay, urine hexose tetra-saccharide (HEX4), and assessment for cardiomyopathy (CXR, ECG, ECHO).

When patients have extremely low or undetectable enzyme activity, other laboratory studies may be required.

Clinical Considerations:

The clinical presentation of Pompe disease ranges from a rapidly progressive infantile form, which is uniformly fatal if untreated, to more slowly progressive later onset forms. All forms of the disorder are eventually associated with progressive muscle weakness and respiratory insufficiency. Cardiomyopathy is associated almost exclusively with the infantile form.

Enzyme replacement therapy (ERT) is available for all forms of Pompe disease and should only be given under the guidance of a metabolic specialist. ERT should be started as soon as possible for patients with the infantile form after evaluating cross-reacting immunologic material (CRIM) status and determining if immune modulation is appropriate.

Medications - Methotrexate, Rituximab and IVIG (intravenous immunoglobulin)

Internet Resources:

https://www.ncbi.nlm.nih.gov/books/NBK1261/
https://ghr.nlm.nih.gov/condition/pompe-disease