

Newborn Screening ACT Sheet

Pompe Disease

Condition Description: Pompe Disease is a lysosomal/glycogen storage disorder caused by deficiency of acid alpha-glucosidase resulting in accumulation of glycogen, primarily in cardiac and skeletal muscle. There is **wide** variability in severity and age of onset. Pompe ranges from a rapidly progressive infantile form to a slowly progressive later-onset form.

Please Take the Following Immediate Actions:

- ☐ **Immediately consult with pediatric metabolic specialist (See attached list).**
- ☐ **Contact family. Discuss with the family the newborn screening result. We provided a resource for this conversation: Next steps after a positive Newborn Screening for Pompe**
- ☐ **Immediate cardiac evaluation** including chest x-ray, electrocardiogram, and echocardiogram for cardiomyopathy.
- ☐ **Evaluate infant** for signs and symptoms including muscle weakness, respiratory problems, cardiomyopathy, and feeding difficulties. If abnormal, send to ER and call metabolic specialist
- ☐ **Order the following labs:** creatine kinase (CK) and leukocyte acid alpha- glucosidase enzyme assay- contact metabolic specialist if unable to collect/send immediately. Keep in mind that initiation of Enzyme Replacement Therapy (ERT) before two weeks of age improves motor outcomes in the first 2 years of life, even when compared to infants in whom treatment was initiated only 10 days later.
- ☐ **Collect repeat screen (between 7-14 days of life)** if the second has not been done.
- ☐ **FAX lab results to {SGUserFax}**

Confirmation of Diagnosis: Leukocyte acid alpha-glucosidase enzyme assay will show decreased GAA enzyme activity which is suggestive of Pompe Disease. However, this result alone does not exclude pseudodeficiency, which causes decreased enzyme levels without disease. Urine hexose tetrasaccharides (Hex4) may be elevated in Pompe disease. Molecular genetic testing can confirm the diagnosis and can predict the phenotype. Gestational age and weight may affect results for this disorder. **Lysosomal enzyme activities are not reliable for premature infants with a birthweight of <1500 grams.**

Clinical Expectations: The clinical presentation of Pompe Disease ranges from a rapidly progressive infantile form to more slowly progressive later-onset forms. All forms of the disorder are associated with progressive muscle weakness and respiratory insufficiency. Cardiomyopathy is associated almost exclusively with the infantile form and newborns should undergo an immediate cardiac evaluation including chest x-ray, electrocardiogram, and echocardiogram. Enzyme Replacement Therapy (ERT) is available for all forms and should be started under guidance of a specialist.

ERT should be initiated as soon as possible for patients with the infantile form after evaluating cross-reactive immunogenic material (CRIM) status and determining if immune modulation is required. For late onset forms, therapy should be initiated at the first signs of muscle weakness.



Additional Information:

How to Communicate Newborn Screening Results

<https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/resources/achdnc-communication-guide-newborn.pdf>

Pompe Disease -Gene Reviews

<https://www.ncbi.nlm.nih.gov/books/NBK1261/>

Medline Plus

<https://medlineplus.gov/genetics/condition/pompe-disease/>

Condition Info for Families-HRSA Newborn Screening

<https://newbornscreening.hrsa.gov/conditions/pompe-disease>

