ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

ACT Sheet

Newborn Screening ACT Sheet

[Pompe Disease (Glycogen Storage Disease Type II)]

Differential Diagnosis: None.

Condition Description: Pompe disease is a lysosomal 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 in age of onset.

You Should Take the Following IMMEDIATE Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (muscle weakness, respiratory insufficiency, feeding difficulties).
- Consult with pediatric metabolic specialist the same day.
- Evaluate the newborn (cardiomyopathy, hypotonia, respiratory status).
- Provide the family with basic information about Pompe disease and its management.

- Report the final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Leukocyte acid alpha-glucosidase enzyme assay: Decreased enzyme activity is suggestive of Pompe disease, but 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.

Clinical Considerations: 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 CXR, EKG, and echocardiogram. Enzyme replacement therapy (ERT) is available for both 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
Gene Reviews
Medline Plus
Condition Information for Families- HRSA Newborn Screening Clearinghouse

Referral (local, state, regional, and national):

Find a Genetics Clinic Directory
Genetic Testing Registry

 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957;#UH9MC30770; National Coordinating Center for the Regional Genetics Networks)

Local Resources *(Insert Local Website Links)*

State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix *(Resources with Full URL Addresses)*

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

Emergency Protocols (New England Consortium of Metabolic Programs)

Gene Reviews

- https://www.ncbi.nlm.nih.gov/books/NBK1261/

Medline Plus

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

Condition Information for Families-HRSA Newborn Screening Clearinghouse

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

Referral (local, state, regional and national)

Find a Genetics Clinic Directory

- https://clinics.acmg.net

Genetic Testing Registry

- https://www.ncbi.nlm.nih.gov/gtr/

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