Newborn Screening ACT Sheet –
Pompe Disease (Glycogen Storage Disease type II)

Condition Description: Pompe disease is a lysosomal storage disorder (LSD) caused by a defect in the acid alpha-glucosidase (*GAA*) gene, 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.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- IMMEDIATELY contact a pediatric metabolic specialist and evaluate the newborn with attention to muscle weakness, hypotonia, feeding difficulties, and clinical evidence of heart disease.
- Discuss with the family the newborn screening result and initiate referral to pediatric metabolic specialist.
- Provide the family with basic information about Pompe disease.
- Report final outcome to state newborn screening program.

Diagnostic Evaluation: Confirmatory alpha-glucosidase enzyme assay, ancillary enzymology, (CK, LDH, AST, ALT) determination, urine hexose tetrasaccharide (*Hex4*), and assessment for cardiomyopathy (CXR, ECG, ECHO). When patients have low enzyme activity, confirmatory *GAA* gene analysis and other laboratory studies may be required in consultation with the pediatric metabolic specialist.

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 material (CRM) status and determining if immune modulation is appropriate.

Additional Information:

GeneReviews
Genetics Home Reference
OMIM

Referral (local, state, regional and national):

Testing
Find Clinical Genetic Services
ACMG Clinic Directory

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LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

GeneReviews
http://www.ncbi.nlm.nih.gov/books/NBK1261/

Genetics Home Reference
http://ghr.nlm.nih.gov/condition/pompe-disease

OMIM
http://www.ncbi.nlm.nih.gov/omim/232300

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

Testing
https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=Pompe

Find Genetic Services
https://www.acmg.net/ACMG/Find_Genetic_Services/ACMG/ISGweb/FindaGeneticService.aspx?hkey=720856ab-a827-42fb-a788-b618b15079f9

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