Newborn Screening ACT Sheet
[Increased Total Galactose with normal GALT]
Primary or Secondary Hypergalactosemia

Differential Diagnosis: Galactokinase (GALK) deficiency; UDP-galactose-4 epimerase deficiency; undefined increase.

Condition Description: Galactose comes from the lactose of breast milk or formula. Galactokinase deficiency is caused by a defect in conversion of galactose to galactose-1-phosphate. Epimerase deficiency limits the production of UDP-galactose, a co-substrate of GALT.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result.
- See and evaluate the infant. Check for reducing substance in urine.
- Consult/refer to a metabolic specialist to determine appropriate follow-up.
- If clinical evaluation is normal and urinary reducing substance is negative, collect and send repeat dried blood specimen to the newborn screening program.
- Report the findings to the newborn screening program.

Diagnostic Evaluation: The diagnosis of galactokinase and epimerase deficiencies is established by quantitation of the respective enzyme activity in erythrocytes.

Clinical Considerations: The neonate is usually normal. If GALK deficiency is untreated, cataracts develop. Treatment is withdrawal of milk. Epimerase deficiency is usually benign.

Additional Information:

Gene Reviews
Genetics Home Reference

Referral (local, state, regional and national):

Testing
Clinical Services
Find Genetic Services

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

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:

Gene Reviews
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=galactosemia

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=galactosemia

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2229?db=genetests&country=United%20States

Clinical Services
http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

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