

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.
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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](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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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	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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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