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

ACT Sheet

Newborn Screening ACT Sheet

[Elevated total galactose with normal GALT enzyme activity]

Primary or Secondary Hypergalactosemia

Differential Diagnosis: Galactokinase (GALK) deficiency; UDP-galactose-4 epimerase (GALE) deficiency, Galactose mutarotase (GALM) deficiency, congenital portosystemic shunt.

Condition Description: Galactosemia refers to a group of inherited disorders caused by an inability to metabolize galactose, a sugar found in lactose. Unlike classic galactosemia, which is caused by deficiency of galactose-1-phosphate uridyl transferase (GALT), this group of disorders cause galactosemia by other enzyme deficiencies. The signs, symptoms, and severity vary by specific diagnosis.

You Should Take the Following Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (vomiting, diarrhea, irritability, jaundice, lethargy).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (poor feeding, vomiting, lethargy).
- Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
- Provide family with basic information about galactosemia including dietary management.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Enzyme assays for GALK and GALE activity (RBC): Decreased enzyme activity is indicative of specific condition. Red blood cell assays are not valid following transfusion. Molecular genetic testing may be required to establish the diagnosis and to identify GALM deficiency.

Clinical Considerations: The neonate is usually asymptomatic, but if untreated can develop cataracts. GALE deficiency is usually benign but in rare cases may present with liver dysfunction, feeding difficulties, and failure to thrive. Lactose restriction should be implemented until GALE deficiency has been excluded by enzyme assay, molecular genetic analysis, galactose-1-phosphate levels in erythrocytes, and galactitol in plasma or urine. Galactokinase and galactose mutarotase deficiencies can cause cataracts but are unlikely to develop other long-term complications. The treatment for all these disorders is the avoidance of dairy products and other foods containing lactose and by the administration of soy-based formulas in infancy.

Additional Information:

How to Communicate Newborn Screening Results
Gene Reviews
Medline Plus
Condition Information for Families- HRSA Newborn Screening Clearinghouse
- Galactokinase deficiency
- Galactoepimerase deficiency

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

Find a Genetics Clinic Directory
Genetic Testing Registry

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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

Gene Reviews

- https://www.ncbi.nlm.nih.gov/books/NBK51671/

Medline Plus

- https://medlineplus.gov/genetics/condition/galactosemia/

Condition Information for Families-HRSA Newborn Screening Clearinghouse

- https://newbornscreening.hrsa.gov/conditions/galactokinase-deficiency
- https://newbornscreening.hrsa.gov/conditions/galactoepimerase-deficiency

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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