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

ACT Sheet

Newborn Screening ACT Sheet

[Absent/Reduced Galactose-1-Phosphate Uridyltransferase (GALT)]

Classic Galactosemia

Differential Diagnosis: Duarte variant galactosemia.

Condition Description: Galactosemia refers to a group of disorders which are caused by an inability to metabolize galactose, a sugar found in lactose. Classic galactosemia results from an inherited deficiency of the galactose-1-phosphate uridyltransferase (GALT) enzyme, causing elevated galactose and galactose-1-phosphate. If treatment is not initiated early, life threatening complications can occur. The Duarte variant refers to a diminished ability to break down galactose in childhood.

You Should Take the Following IMMEDIATE Actions:

- Inform family of the newborn screening result
- Ascertain clinical status (poor feeding, vomiting, lethargy, jaundice). Discontinue breast feeding and/or cow's milk formulas and initiate non-lactose based feedings with a soy formula.
- Consult with pediatric metabolic specialist the same day.
- Evaluate the newborn (jaundice, poor feeding, vomiting, lethargy, bulging fontanel, and bleeding). If any of these findings are present or if the newborn is ill, transfer to a hospital for further treatment in consultation with the metabolic specialist.
- Initiate confirmatory/diagnostic testing and management, as recommended by specialist.
- Provide family with basic information about classic galactosemia, including dietary management.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Red blood cell GALT activity: Complete or near-complete deficiency in classic galactosemia; partial reduction of normal activity with the Duarte variant. Red blood cell galactose-1-phosphate (gal-1-P): Elevated in both classic and the Duarte variant in patients consuming lactose. Red blood cell assays are not valid following transfusion. Molecular genetic testing may be required to confirm the diagnosis.

Clinical Considerations: Classic galactosemia presents in the first few days of life and may be fatal without treatment. Signs include poor feeding, vomiting, jaundice and may include lethargy and/or bleeding. Neonatal *E. coli* sepsis can occur and is often fatal. The treatment is the avoidance of dairy products and other foods containing galactose, and the administration of soy-based formulas. Symptomatic neonates will require emergency supportive measures. Considered a benign condition, there is no standard accepted management for the Duarte variant; some practitioners restrict high galactose foods in early childhood.

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 Regisstry

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/NBK1518/

Medline Plus

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

Condition Information for Families-HRSA Newborn Screening Clearinghouse

- https://newbornscreening.hrsa.gov/conditions/classic-galactosemia

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/