

Newborn Screening ACT Sheet

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



ACMG website: bit.ly/3ZgZTEg

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 is characterized by diminished GALT enzyme activity.

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. The Duarte variant is considered a benign condition with no standard accepted management; 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](#)

This ACT sheet and algorithm is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this ACT sheet and algorithm is completely voluntary and does not necessarily assure a successful medical outcome. This ACT sheet and algorithm 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. In determining the propriety of any specific procedure or test, clinicians should apply their 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 ACT sheet and algorithm. Clinicians also are advised to take notice of the date this ACT sheet and algorithm was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures. Where individual authors are listed, the views expressed may not reflect those of authors' employers or affiliated institutions.

© American College of Medical Genetics and Genomics, 2026 Content Updated: January 2026

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

[Find a Genetics Clinic Directory](#)
[Genetic Testing Registry](#)

**January 2026: Update to the original
ACT Sheet:**

**Under Condition Description: The
Duarte variant is characterized by
diminished GALT enzyme activity.**

**Discontinue breast feeding was given a
separate line under Action Items.**