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 Registry**

## State and Other Resources

### State Newborn Screening Program

Newborn Genetic Screening, New Mexico Department of Health
877-890-4692, www.nmhealth.org/about/phd/fhb/cms/nbgs/

### Genetics/Metabolic Consultants

Michael Marble, MD
University of New Mexico Metabolic Clinic
505-272-6631, hsc.unm.edu/directory/marble-michael.html

### Information for Clinicians and Families

New Mexico Medical Home Portal (see also the Parents & Families section)
nm.medicalhomeportal.org/newborn/galactosemia

### Parent/Family Support

Galactosemia Foundation
www.galactosemia.org

## National Resources (with web addresses)

### Additional Information

How to Communicate Newborn Screening Results
www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf

Gene Reviews
www.ncbi.nlm.nih.gov/books/NBK1518/

Medline Plus
medlineplus.gov/genetics/condition/galactosemia/

Condition Information for Families-HRSA Newborn Screening Clearinghouse
newbornscreening.hrsa.gov/conditions/classic-galactosemia

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

Find a Genetics Clinic Directory
clinics.acmg.net

Genetic Testing Registry
www.ncbi.nlm.nih.gov/gtr/