

## 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.
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- 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](#)
- [Galactose epimerase deficiency](#)

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

[Find a Genetics Clinic Directory](#)

[Genetic Testing Registry](#)

This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource 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, 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 practice resource. Clinicians also are advised to take notice of the date this practice resource 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.

**Local Resources (Insert Local Website Links)**  
**State Resource Site (Insert Website Information)**

<b>Name</b>	
<b>URL</b>	
<b>Comments</b>	

**Local Resource Site (Insert Website Information)**

<b>Name</b>	
<b>URL</b>	
<b>Comments</b>	

**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/galactosemimerase-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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