

Galactosemia

Action required

Contact metabolic specialist today.
See infant today.

Issues to discuss with metabolic specialist

- Laboratory evaluation of infant
 - § Should testing be performed by primary care or metabolic clinic
 - § Should feeding be changed before evaluation
 - § gal-1-P and GALT levels

False Positives

- Common
- Milder variants seen with partial GALT activity
- Screening result can be impacted by transfusions and environmental factors, such as heat
- High false positive in Minnesota Hmong population

Review with family

Family has **not** been notified of result by MDH.

After discussion with metabolic specialist, contact family to coordinate clinic visit, lab work, and referral to metabolic clinic. Infant may be symptomatic (i.e., hospitalized with hyperbilirubinemia) when family is contacted or at clinic visit.

Prompt follow-up and intervention are critical.

NICU issues

Newborn screens cannot be accurately interpreted after transfusion.

Clinical summary

Galactosemia is an autosomal recessive disorder that results from the defective activity of galactose-1-phosphate uridyl transferase (GALT) enzyme. Newborns are typically asymptomatic, but may present with hyperbilirubinemia or *E. coli* sepsis. If an infant is not screened and/or left untreated, symptoms begin to appear early in infancy, and can include poor feeding, failure to thrive, liver dysfunction, and mental retardation.

Affected children require life-long dietary restriction of galactose and lactose and monitoring by both primary care and specialty providers.

Incidence: ~ 1/30,000; affects all ethnic groups

Clinical expectations

If treated promptly, children with galactosemia can be expected to be clinically stable with near normal development.

Developmental delay can occur even with treatment.

Affected children should be monitored for dietary compliance and need for intervention.

Resources

GeneTests: www.genetests.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

ACT Sheets: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

MN Newborn Screening Program:
www.health.state.mn.us/newbornscreening