

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn
Screening Program



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

Next Steps

Today, you should take the following recommended actions:

- **Consult** with a metabolic specialist. Contact information for the metabolic specialists can be found on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms.
- **Evaluate** infant (jaundice, abnormal bleeding); arrange immediate referral if symptomatic.
- **Arrange** referral to a metabolic specialist for further diagnostic work-up.

If you have questions about the newborn screening result or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

Review with Family

Discuss this result with the family as MDH has **not** notified them. Share the follow-up plan with them. Educate family about signs, symptoms, and need for urgent treatment. If advised by metabolic specialist, stop breast or milk-based formula feedings immediately and initiate non-lactose (soy formula) feedings.

False Positives

Screening result can be impacted by transfusion and environmental factors such as heat and humidity.

Differential Diagnosis

Absent/reduced GALT is primarily associated with:

- Classic galactosemia — Incidence of 1 in 50,000

Other disorders to consider:

- Duarte galactosemia (milder variant)
- Glucose-6-phosphate dehydrogenase deficiency (G6PD)

Clinical Summary

Classic galactosemia is a disorder of galactose metabolism. It is caused by a deficiency in the enzyme, GALT. As a result of this deficiency, consumption of food containing lactose or galactose (including breast milk) causes toxicity.

An affected neonate can develop life-threatening complications within a few days of life. Complications include poor feeding, lethargy, jaundice, vomiting, and abnormal bleeding. Other serious complications include liver disease and *E. coli* sepsis, which can be fatal. Concern is heightened if the neonate has non-resolving hyperbilirubinemia.

Treatment for classic galactosemia requires the immediate and life-long exclusion of lactose and galactose from the diet. In infancy, this is often accomplished by switching to a soy-based formula. If treated early, liver failure, sepsis, and neonatal death are prevented. However, affected individuals remain at increased risk for developmental delays, speech problems, and abnormalities of motor function.