Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Total Galactose (TGAL)

Next Steps

This week, 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.
- **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.

False Positives

Isolated TGAL elevations have been shown to be more common in certain ethnic groups, including the Hmong.

Differential Diagnosis

Elevated TGAL is primarily associated with:

- Duarte galactosemia — Incidence of 1 in 4,000

Other disorders to consider:

- Galactokinase (GALK) deficiency
- Galactose epimerase (GALE) deficiency
- Classic galactosemia

Clinical Summary

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 and life-threatening health complications within a few days of life.

Duarte galactosemia (DG) results from a partial deficiency of the GALT enzyme and is a milder variant of the disease. Individuals with DG are usually asymptomatic. Historically, there have been conflicting reports about the neurodevelopmental outcomes related to DG. However, a recent, larger study found that children with DG did not have increased risk for developmental challenges, regardless of the type of milk consumed in infancy.

Currently, there is no consensus agreement as to whether to treat individuals with DG with a galactose-restriction. Recommendations regarding treatment should be discussed with specialists and may be determined on a case-by-case basis.