Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Total Galactose (TGAL) with Normal Galactose-1-Phosphate Uridyltransferase (GALT)

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, like the Hmong.

Differential Diagnosis

Elevated TGAL with normal GALT is primarily associated with:

- Galactokinase (GALK) deficiency — Incidence unknown, but is probably less than 1 in 100,000
- Galactose epimerase (GALE) deficiency — Incidence unknown

Clinical Summary

Clinical summaries for each of the two disorders in the differential are provided below.

GALK deficiency is a milder type of galactosemia in which the body is unable to properly digest galactose. If left untreated, affected neonates develop cataracts. Treatment consists of lifetime avoidance of foods with galactose to prevent a toxic build-up of undigested sugars. Many children with GALK deficiency need to avoid all milk products.

GALE deficiency is a type of galactosemia in which the body is unable to properly digest galactose. There are three forms of GALE deficiency (generalized, peripheral, and intermediate) with variable presentation. Most individuals with GALE deficiency have a benign peripheral form and do not manifest disease. If treatment is recommended by the metabolic specialist, it would involve a galactose-restricted diet.