Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C5-DC Acylcarnitine

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 for macrocephaly, metabolic acidosis, and dystonia; arrange immediate referral if symptomatic. In most cases, the infant will be asymptomatic.
- **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. Discuss signs, symptoms, and need for urgent treatment if infant becomes mildly ill.

False Positives

Screening result can be impacted by specimen collection before 24 hours and carnitine supplementation in infant. Furthermore, an infant born to a mother with asymptomatic GA-I may screen positive because of elevations of C5-DC from the mother.

Differential Diagnosis

Elevated C5-DC acylcarnitine is primarily associated with:

- Glutaric acidemia (GA-I) - incidence of 1 in 30,000 to 40,000 individuals (incidence of 1 in 300 in Amish communities and the Ojibwa population of Canada).

Clinical Summary

Glutaric acidemia type I (GA-I) is an organic acid disorder. GA-I is highly variable, even within families, with regard to onset of disease and symptoms. Infants with GA-I often present with macrocephaly. Additional symptoms can develop in the first year of life, including metabolic ketoacidosis, failure to thrive, and sudden onset of dystonia and athetosis due to irreversible striatal damage.

Affected individuals have a very high risk for neurologic problems before age five, so early and aggressive treatment is necessary.

Treatment for GA-I consists of a low-protein diet and IV fluids during illness. Some specialists may prescribe supplements.

Even with treatment, however, long-term complications can occur.