Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C4 and Elevated C5 Acylcarnitines

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 'sweaty feet' odor, facial dysmorphism, and signs of lethargy; 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 need for infant to avoid fasting. 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 the infant.

Differential Diagnosis

Elevated C4 and C5, when together, is primarily associated with:

- Glutaric acidemia type II (GA-2), also called multiple acyl-CoA dehydrogenase deficiency (MADD)—Incidence is unknown.

Other disorder to consider:

- Ethylmalonic encephalopathy (EE)

Clinical Summary

GA-2 is a fatty acid oxidation disorder. Fatty acid oxidation occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism.

GA-2 is different for each child. Severe GA-2 often presents at birth with various birth defects, including brain malformations, hepatomegaly, dilated cardiomyopathy, kidney malformations, facial dysmorphism, and genital abnormalities. Some individuals present in childhood with a sudden metabolic crisis often triggered by illness or other stresses. The child becomes hypoglycemic, weak, feeds poorly, experiences vomiting, and has decreased activity. These metabolic crises can be life-threatening.

Treatment consists of a lifelong low fat, high carbohydrate diet and avoidance of fasting. Some specialists may prescribe riboflavin.