Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C5 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 signs of metabolic acidosis; 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 when to contact you with concerns.

False Positives

Screening result can be impacted by specimen collection before 24 hours.

Differential Diagnosis

Elevated C5 acylcarnitine is primarily associated with:

- Isovaleric acidemia — Incidence of 1 in 250,000
- Short/branched chain acyl-CoA dehydrogenase (SBCAD) deficiency — Incidence of 1 in 230,000 (more common in Hmong populations: 1 in 500)

Clinical Summary

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

Isovaleric acidemia (IVA) is an organic acid disorder. People with IVA lack an enzyme needed to break down leucine. As a result, isovaleric acid accumulates, leading to the signs and symptoms of IVA. Symptoms range from very mild to life-threatening. In severe cases, symptoms can develop within a few days of life, including poor feeding, vomiting, seizures, and lethargy. Symptoms can progress to metabolic acidosis, coma, and possibly death. Affected children need a low protein diet.

SBCAD is an organic acid disorder. People with SBCAD lack an enzyme needed to break down isoleucine. Very few symptomatic patients have been reported. Symptomatic patients have presented with lethargy, poor feeding, hypoglycemia, and respiratory distress. Most infants found to have SBCAD by Minnesota newborn screening are of Hmong descent and remain asymptomatic. These individuals remain healthy with little or no treatment.

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REV 12/2018