Positive Result:
Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C4 Acylcarnitine

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. Although development of symptoms are unlikely, 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 carnitine supplementation in the infant.

Differential Diagnosis

Elevated C4 acylcarnitine is primarily associated with:

- Short-chain acyl-CoA dehydrogenase (SCAD) deficiency — Incidence of 1 in 40,000

Other disorders to consider:

- Isobutyryl-CoA dehydrogenase (IBD) deficiency

- Glutaric acidemia type II (GA-II)

- Ethylmalonic encephalopathy (EE)

Clinical Summary

SCAD deficiency is a fatty acid oxidation (FAO) 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.

SCAD deficiency can have a variable presentation. Most affected neonates are asymptomatic. An affected neonate; however, can become ill with vomiting, lethargy, seizures, and hypoketotic hypoglycemia.

Treatment consists primarily of avoidance of fasting and support during intercurrent illnesses. Some specialists may prescribe vitamin/cofactor supplementation.