Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C16 and/or Elevated C18:1 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 signs of hepatomegaly, hypoglycemia, cardiac arrhythmia; arrange immediate referral if symptomatic and/or hypoglycemia is present.
- **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 C16 and/or elevated C18:1 acylcarnitines are primarily associated with:

- Carnitine palmitoyl transferase type II (CPT-II) deficiency — Incidence is unknown; rare
- Carnitine acylcarntitine translocase (CACT) deficiency — Incidence is unknown; very rare with ~30 cases reported

Clinical Summary

Both CPT-II and CACT deficiencies are fatty acid oxidation disorders. Fatty acid oxidation occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism.

CPT-II deficiency has three forms with variable symptoms and onset. Symptoms can range from muscle weakness, to cardiac arrhythmias, to multi-system organ failure. The age of onset can begin in the neonatal period to adulthood. Disease severity correlates with age of onset.

Symptoms of CACT deficiency include breathing problems, hepatomegaly, hypoketotic hypoglycemia, seizures, and cardiac arrhythmia.

Treatment for both CPT-II and CACT deficiency consists of a lifelong low fat, high carbohydrate diet and avoidance of fasting. Some specialists may prescribe supplements. Early treatment can be life-saving. Even with treatment, however, long-term complications can occur.

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