

Positive Result:

Blood Spot Screen Result Notification



Elevated C14:1 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.
- **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. Because symptoms may present acutely in the neonatal period, educate family about need for infant to avoid fasting and explain signs, symptoms, and need for urgent treatment with IV glucose if infant becomes even mildly ill.

False Positives

Screening result can be impacted by carnitine supplementation in the infant.

Differential Diagnosis

Elevated C14:1 acylcarnitine is primarily associated with:

- Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency — Incidence of 1 in 30,000

Clinical Summary

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

Newborns with VLCAD deficiency may present acutely and is associated with high mortality unless treated promptly. If untreated, symptoms can include hepatomegaly, cardiomyopathy, arrhythmias, lethargy, hypoketotic hypoglycemia, failure to thrive, and seizures. Milder variants exist.

Treatment requires a lifelong low fat, high carbohydrate diet and avoidance of fasting. Some specialists may prescribe carnitine supplementation. If treated early, health complications can be prevented.