

Positive Result:

Blood Spot Screen Result Notification



Elevated C8 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 (hypotonia, hepatomegaly); 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

Rare. Some studies suggest a possible impact on screening results from carnitine supplementation and seizure medication like valproate in the infant.

Differential Diagnosis

Elevated C8 acylcarnitine is primarily associated with:

- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency — Incidence of 1 in 17,000

Clinical Summary

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

Affected newborns are usually asymptomatic. The disease can present acutely in the neonate with vomiting, lethargy, metabolic acidosis, hepatomegaly, and hypoketotic hypoglycemia. Affected children are also at risk for seizures, breathing difficulties, liver problems, brain damage, coma, and sudden death. Symptoms are typically triggered by fasting or illness. MCAD deficiency is associated with high mortality unless treated promptly.

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.