

Positive Result:

Blood Spot Screen Result Notification



Elevated C4-OH Acylcarnitine

Next Steps

Within one business day, 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. 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-OH acylcarnitine is primarily associated with:

- Medium/short-chain hydroxyacyl-CoA dehydrogenase (M/SCHAD) deficiency — Incidence is unknown

Clinical Summary

M/SCHAD deficiency is a fatty acid oxidation 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.

M/SCHAD deficiency has a variable presentation. Most children never have symptoms. Symptomatic children can present with vomiting, lethargy, hypoglycemia, and hyperinsulinism. They are also at risk for seizures, life-threatening heart and breathing problems, coma, and sudden death.

Treatment consists primarily of avoidance of fasting. Sometimes a low fat, high carbohydrate diet is recommended under the guidance of a dietician familiar with M/SCHAD deficiency. Some specialists may prescribe carnitine supplementation and medication. If treated early, children can have healthy growth and development.