

# 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.