

# Positive Result:

## Blood Spot Screen Result Notification



### Elevated C16-OH 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 for signs of feeding difficulties, lack of energy, hypoglycemia, hypotonia, and liver problems; 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

Screening result can be impacted by specimen collection before 24 hours and carnitine supplementation in the infant. Maternal acute fatty liver of pregnancy (AFLP) and HELLP (hemolysis, elevated liver enzymes, and low platelets) syndrome can also cause a false positive screening result.

#### Differential Diagnosis

Elevated C16-OH acylcarnitine, is primarily associated with:

- Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency — Incidence is unknown. In Finnish populations incidence may be as high as 1 in 62,000.
- Trifunctional protein (TFP) deficiency - Incidence is unknown.

#### Clinical Summary

LCHAD deficiency and TFP deficiency are both 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.

LCHAD deficiency and TFP deficiency usually appear in infancy and can be life-threatening. Symptoms include feeding difficulties, hypotonia, hypoglycemia, and hepatomegaly. Rhabdomyolysis may occur. They are also at risk for serious heart problems, breathing difficulties, coma, and sudden death. TFP deficiency has a broader spectrum of symptoms, including milder variations.

Treatment consists of a lifelong low fat, high carbohydrate diet and avoidance of fasting. Some specialists may prescribe carnitine supplementation. If treated early, children can have healthy growth and development. Even with treatment, however, long-term complications can occur.