

Positive Result:

Blood Spot Screen Result Notification



Elevated C0 Acylcarnitine (Free Carnitine)

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 lethargy, hepatomegaly, and seizures; 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.

Differential Diagnosis

Elevated free carnitine is primarily associated with:

- Carnitine palmitoyl transferase type I (CPT-I) deficiency — Incidence is unknown; fewer than 50 individuals reported

Clinical Summary

CPT-I 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.

CPT-I deficiency has a variable presentation. Symptoms often appear in early childhood with lethargy, irritability, poor appetite, hypoketotic hypoglycemia, and hepatomegaly. If untreated, symptoms can progress to liver failure, seizures, coma, and sudden death.

Treatment consists of a lifelong low fat, high carbohydrate diet and avoidance of fasting. Some specialists may prescribe supplements. If treated early, children can have healthy growth and development.