

Positive Result:

Blood Spot Screen Result Notification



Absent/Reduced C0 Acylcarnitine (Free Carnitine)

Next Steps

This week, 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 tachycardia, reduced muscle tone, and 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

Screening result can be impacted by prematurity, specimen collection before 24 hours, and maternal carnitine deficiency.

Differential Diagnosis

Absent/reduced free carnitine is primarily associated with:

- Carnitine uptake defect (CUD) — Incidence is 1 in 100,000 (more common in Japanese population: 1 in 40,000)

Clinical Summary

Carnitine uptake defect (CUD) 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.

CUD has a variable presentation and age of onset. Symptoms often appear in infancy or early childhood with lethargy, cardiomyopathy, vomiting, hypoglycemia, and hypotonia. If untreated, symptoms can progress to heart failure, liver failure, seizures, coma, and sudden death.

Treatment consists of a lifelong diet of a low fat, high carbohydrate diet, carnitine supplementation, and avoidance of fasting. If treated early, children can have healthy growth and development.