

Positive Result:

Blood Spot Screen Result Notification



Elevation of C4 Acylcarnitine

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.
- **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 signs, symptoms, and when to contact you with concerns.

False Positives

Screening result can be impacted by carnitine supplementation in the infant.

Differential Diagnosis

Elevated C4 acylcarnitine is primarily associated with:

- Isobutyryl-CoA dehydrogenase (IBD) deficiency- very rare, exact incidence is unknown

Other disorders to consider:

- Short-chain acyl-CoA dehydrogenase (SCAD) deficiency
- Glutaric acidemia type II (GA-II)
- Ethylmalonic encephalopathy (EE)

Clinical Summary

IBD deficiency is a condition that disrupts the breakdown of the amino acid valine. This may lead to reduced energy production and cause the features of IBD deficiency in some people. People with IBD deficiency also have high levels of a compound called C4-acylcarnitine.

Most children with IBD deficiency are asymptomatic. In very rare cases, a child may have symptoms such as hypotonia, developmental delays, failure to thrive, anemia, cardiomyopathy, and seizures. However, because IBD deficiency is rare and usually does not cause symptoms, the true course and significance of this disorder is unknown.

Individuals with IBD deficiency may not require any treatment, especially if they are asymptomatic. A metabolic specialist may recommend dietary changes, avoidance of fasting, and/or supplements.