

# Positive Result:

## Blood Spot Screen Result Notification



### Elevated C4 and Elevated C5 Acylcarnitines

#### 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 'sweaty feet' odor, facial dysmorphism, and signs of lethargy; 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 C4 and C5, when together, is primarily associated with:

- Glutaric acidemia type II (GA-2), also called multiple acyl-CoA dehydrogenase deficiency (MADD)—Incidence is unknown.

Other disorder to consider:

- Ethylmalonic encephalopathy (EE)

#### Clinical Summary

GA-2 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.

GA-2 is different for each child. Severe GA-2 often presents at birth with various birth defects, including brain malformations, hepatomegaly, dilated cardiomyopathy, kidney malformations, facial dysmorphism, and genital abnormalities. Some individuals present in childhood with a sudden metabolic crisis often triggered by illness or other stresses. The child becomes hypoglycemic, weak, feeds poorly, experiences vomiting, and has decreased activity. These metabolic crises can be life-threatening.

Treatment consists of a lifelong low fat, high carbohydrate diet and avoidance of fasting. Some specialists may prescribe riboflavin.