

# Positive Result:

## Blood Spot Screen Result Notification



### Elevated C3 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.
- **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 symptomatic.

#### False Positives

Screening result can be impacted by specimen collection before 24 hours, carnitine supplementation, hyperbilirubinemia, and maternal B12 deficiency.

#### Differential Diagnosis

Elevated C3 acylcarnitine is primarily associated with:

- B12 deficiency (including maternal B12 deficiency)
- Propionic acidemia — Incidence of 1 in 35,000
- Methylmalonic acidemia — Incidence of 1 in 50,000

Other disorders to consider:

- Cobalamin defects

#### Clinical Summary

Clinical summaries for each of the two disorders in the primary differential are provided below.

Propionic acidemia (PA) is an organic acid disorder. Symptoms can develop within a few days of life, including poor feeding, vomiting, hypotonia, and lethargy. Symptoms can progress to tachypnea, seizures, and coma. Can be fatal without intervention.

Methylmalonic acidemia (MMA) is an organic acid disorder. Symptoms appear in early infancy and can include: vomiting, dehydration, hypotonia, developmental delay, lethargy, hepatomegaly, and failure to thrive. Can be fatal without intervention.

Treatment for both PA and MMA consists of limiting specific dietary proteins. Treatment also includes avoidance of fasting and support during illness. Some specialists may prescribe supplements to help break down fats and proteins. Early treatment can be life-saving. Even with treatment, however, long-term complications can occur.