Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C4-DC + C5-OH 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 signs of hypoglycemia, ketonuria, or metabolic acidosis; 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 signs, symptoms, and when to contact you with concerns.

False Positives

Screening result can be impacted by specimen collection before 24 hours. Furthermore, an infant born to a mother with asymptomatic 3-MCC deficiency may screen positive because of elevations of C5-OH from the mother.

Differential Diagnosis

Elevated C4-DC + C5-OH acylcarnitines are primarily associated with:

- 3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency — Incidence of 1 in 36,000

Other disorders to consider:

- 3-hydroxy-3-methylglutaryl (HMG)-CoA lyase deficiency
- 3-methylglutaconic aciduria (3MGA)

Clinical Summary

3-MCC deficiency is an organic acid disorder. People with 3-MCC deficiency lack an enzyme needed to break down leucine.

Infants are asymptomatic at birth. Symptoms develop in infancy or early childhood, and range from mild to life-threatening. Symptoms may include feeding difficulties, recurrent vomiting and diarrhea, lethargy, and hypotonia. If untreated, symptoms can progress to metabolic acidosis, seizures, liver failure, and coma resulting in brain damage or even death. Some people with 3-MCC deficiency never experience symptoms.

If treatment is indicated, it requires a lifelong protein restricted diet and supplements may be prescribed.

DEPARTMENT OF HEALTH

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