Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C10:2 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.
- **Evaluate** infant (poor feeding, vomiting, sepsis); 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. Discuss signs, symptoms, and when to contact you with concerns.

False Positives

Screening result can be impacted by specimen collection before 24 hours and carnitine supplementation in the infant.

Differential Diagnosis

Elevated C10:2 acylcarnitine is primarily associated with:

- 2, 4-Dienoyl-CoA reductase deficiency — Rare; only two patients reported

Clinical Summary

2, 4-Dienoyl-CoA reductase deficiency (DE-RED) is caused by a defect in the enzyme necessary for the degradation of certain fats.

With only two cases reported amongst millions of children screened, the true course of this disorder is unknown.

Case 1: The child was born with a small body habitus and microcephalic. The infant was readmitted on her second day of life for sepsis, hypotonia, poor feeding, and intermittent vomiting. Despite treatment, she developed respiratory acidosis and died at four months of age.

Case 2: The child presented with failure to thrive, microcephaly, cental hypotonia, and mild dysmorphic features. His clinical course included significant neurological decline, progressive encephalopathy, dystonia, renal tubular acidosis, intermittent lactic acidosis, and presence of leukodystrophy and basal ganglia lesions on MRI. He died at age 5 years from aspiration pneumonia.

Suggested treatment includes dietary lysine restriction, caloric support, provision of medium-chain fatty acids, and carnitine supplementation.