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 — Extremely rare; only three patients have been reported in medical literature to date

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 three cases reported in medical literature amongst millions of children screened, the true course of this disorder is unknown.

The three reported children with DE-RED presented prenatally or shortly after birth. All three had microcephaly, failure to thrive, hypotonia, mild dysmorphic features, and brain abnormalities including ventriculomegaly. Other symptoms found in two of the three reported children included epilepsy, vomiting, movement disorders, and optical alterations. Two children passed at a young age: one passed at four months of age after developing sepsis and respiratory acidosis, and one passed at five years from aspiration pneumonia after neurologic decline. The third child was alive at 10 years old.

Suggested treatment includes dietary lysine restriction, caloric support, provision of medium-chain fatty acids, and carnitine supplementation. Administration of additional supplements, such as pyridoxal phosphate, may also be considered.

DEPARTMENT OF HEALTH

Minnesota Newborn Screening Program | www.health.state.mn.us/newbornscreening
P.O. Box 64899, St. Paul, MN 55164
To obtain this information in a different format:
contact health.newbornscreening@state.mn.us
or call (800) 664-7772 ***interpreters available***

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