Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Leucine, Isoleucine, and Other Branched-Chain Amino Acids

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, lethargy, tachypnea, alternating hypertonia/hypotonia, and seizures); 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 hyperammonemia. Discuss signs, symptoms, and when to contact you with concerns.

False Positives

Screening result can be impacted by specimen collection before 24 hours.

Differential Diagnosis

Elevated leucine, isoleucine, and other branched-chain amino acids are primarily associated with:

- Maple syrup urine disease (MSUD) — Incidence of 1 in 185,000 (incidence of 1 in 380 in the Old Order Mennonite populations; also more common in people of French-Canadian ancestry and Ashkenazi Jewish ancestry)

Other disorders to consider:

- Hydroxyprolinemia (probably benign)

Clinical Summary

In MSUD, leucine, isoleucine, and other branched chain amino acids cannot be metabolized further than their alpha-ketoacid derivatives. This leads to toxic accumulations. The condition gets its name from the distinctive maple syrup smell from an affected infant's urine.

The most common and severe form of MSUD has a neonatal onset. MSUD presents with poor feeding, vomiting, lethargy, alternating hypertonia/hypotonia, and delayed development. If untreated, symptoms can progress to include seizures, coma, cerebral edema, and death.

Treatment includes lifelong dietary restriction of protein. Thiamine supplements are helpful for some children with MSUD and may be prescribed.

Some children may still develop symptoms of MSUD even with treatment.

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