Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Citrulline

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, lethargy, or tachypnea); 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 cituline is primarily associated with:

- Citrullinemia type I (CIT-I) — Incidence of 1 in 57,000
- Argininosuccinic acidemia — Incidence of 1 in 70,000

Other disorders to consider:

- Citrullinemia type II
- Pyruvate carboxylase deficiency

Clinical Summary

Citrullinemia type I (CIT-I) and argininosuccinc acidemia (ASA) are caused by defects in the enzymes responsible for converting ammonia to urea resulting in hyperammonemia and elevated citrulline.

CIT-I and ASA can present acutely in the neonatal period. Early symptoms include hyperammonemia, seizures, failure to thrive, lethargy, and coma.

Treatment includes lifelong dietary restriction of protein. Ammonia scavenging drugs and supplements may be prescribed.

Episodes of hyperammonemia requiring hospital admission may occur even with treatment. Long-term complications, such as brain damage, may be difficult to prevent.