Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Absent/Reduced 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, hypotonia, tachypnea, or 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 signs of hyperammonemia. Discuss other symptoms and when to contact you with concerns.

False Positives

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

Differential Diagnosis

Absent/reduced citulline is primarily associated with:

- Proximal urea cycle disorders—combined incidence of 1 in 8,500

Clinical Summary

Proximal urea cycle disorders are caused by defects in the enzymes early in the cycle that are responsible for converting ammonia to urea. This results in hyperammonemia and reduced citrulline. The associated disorders include N-acetylglutamate synthetase (NAGS) deficiency, carbamoylphosphate synthestase (CPS) deficiency, and ornithine transcarbamoylase (OTC) deficiency.

NAGS, CPS, and OTC deficiencies can present acutely in the neonatal period. Early symptoms include hyperammonemia, failure to thrive, lethargy, and seizures. If untreated, hyperammonemia can lead to coma and death.

Treatment includes lifelong dietary restriction of protein. Ammonia scavenging drugs and supplements may be prescribed. Sometimes dialysis to lower ammonia levels is needed. Children with these disorders often receive liver transplantation.

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

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