Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Argininosuccinic Acid

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 signs of hyperammonemia. Discuss other possible symptoms and when to contact you with concerns.

False Positives

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

Differential Diagnosis

Elevated argininosuccinic acid is primarily associated with:

- Argininosuccinic acidemia — Incidence of 1 in 70,000

Clinical Summary

Argininosuccinc acidemia (ASA) is caused by a defect in the enzyme responsible for breaking down nitrogen so that it does not build up as ammonia.

ASA can present acutely in the neonatal period. Early symptoms include hyperammonemia, poor feeding, vomiting, lethargy, ataxia, seizures, and coma. Occasionally, a child may have a less common, mild form of ASA where hyperammonemia occurs only during periods of illness or other stress.

Treatment can be life-saving and includes life-long 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.