

# Positive Result:

## Blood Spot Screen Result Notification



### Elevated Arginine

#### Next Steps

This week, 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, tachypnea, and signs of liver disease); arrange immediate referral if symptomatic. In most cases, the infant will be asymptomatic.
- **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 arginine is primarily associated with:

- Argininemia — Incidence of 1 in 300,000

#### Clinical Summary

Argininemia is caused by defects in the enzymes responsible for converting ammonia to urea resulting in hyperammonemia and elevated arginine.

A child with argininemia is usually asymptomatic in the neonatal period. Once dietary protein is introduced, mild to moderate hyperammonemia can occur. Symptoms typically present in early childhood around one to three years of age with muscle stiffness and spasticity, developmental delay, tremor, and ataxia. If untreated, symptoms can progress to include intellectual disabilities and seizures.

Treatment 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 intellectual disabilities or spasticity, may be difficult to prevent.