Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Phenylalanine

Next Steps

Within one business day, 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.
- **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 need for infant to avoid protein. Discuss signs, symptoms, and when to contact you with concerns.

False Positives

Screening result can be impacted by specimen collection before 24 hours and total parenteral nutrition (TPN).

Differential Diagnosis

Elevated phenylalanine is primarily associated with:

- Phenylalanine hydroxylase deficiency
  - ▹ Phenylketonuria (PKU) — Incidence of 1 in 12,000
  - ▹ Hyperphenylalaninemia (H-PHE) — Incidence is unknown, but is less common than PKU

Other disorders to consider:

- Transient hyperphenylalaninemia
- Biopterin defects — Incidence of 1 in 1 million (more common in individuals from Saudi Arabia, Taiwan, China, and Turkey)

Clinical Summary

Phenylalanine hydroxylase (PAH) deficiency is a disease where the body is unable to break down the amino acid, phenylalanine, from protein leading to a toxic buildup. There are two main categories based on the levels of phenylalanine found in the blood.

The most severe form is called PKU. Neonates affected with PKU appear normal initially, however over months they will begin to show irritability, posturing, increased deep tendon reflexes, have a mousy odor, and vomiting. If untreated, they will develop microcephaly, intellectual disabilities, seizures, autistic-like features, and white matter disease. Treatment consists primarily of a lifelong protein restricted diet. Medication and supplements may be prescribed. If treated early, symptoms can be prevented.

The less severe form is called H-PHE. Most people with H-PHE experience mild or no symptoms. Treatment is typically not needed, but may consist of a protein restricted diet.