Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Methionine

Next Steps

This week, you should take the following recommended actions:

- **Review** hospital birth records (if possible) to see if newborn received any amount of total parenteral nutrition (TPN).
- **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. 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 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, prematurity, and total parenteral nutrition (TPN).

Differential Diagnosis

Elevated methionine is primarily associated with:

- Hyperalimentation (e.g., TPN)
- Liver immaturity, dysfunction, or disease
- Classical homocystinuria — Incidence of 1 in 200,000

Other disorders to consider:

- Hypermethioninemia
- Methionine adenosyltransferase I/III (MAT I/III) deficiency
- Glycine n-methyltransferase (GNMT) deficiency
- Adenosylhomocysteine hyrolase deficiency

Clinical Summary

Classical homocystinuria is an amino acid disorder characterized by the inability to break down the amino acid, methionine, from protein leading to a toxic buildup.

Affected neonates are typically asymptomatic. If untreated, these children can develop intellectual disabilities, ectopia lentis, skeletal abnormalities, and thromboembolism.

Treatment requires a lifelong protein restricted diet. Medication and supplements may be prescribed. If treated early, most disease sequelae can be prevented. Even with treatment, some children still experience ophthalmic complications.