Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Hemoglobin E no A (hemoglobin E disease)

Next Steps

This week you should take the following recommended actions:

- **Consult** with pediatric hematologist. Contact information for the pediatric hematologists can be found on the newborn screening report and on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms. It is unlikely infant will be symptomatic.
- **Evaluate** infant (splenomegaly); arrange emergency treatment if symptomatic.
- **Arrange** referral to pediatric hematologist for further diagnostic work-up. A sickle screen (e.g., sickledex or hemoglobin S solubility test) is NOT appropriate for diagnostic purposes.

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 your follow-up plan with them. Educate family about signs, symptoms, and when urgent treatment may be needed.

False Positives

Unlikely since the methodologies used in newborn screening are very accurate and specific.

Differential Diagnosis

FE (no A) is primarily associated with:

- Hemoglobin E disease — More common in SE Asian populations
- Hemoglobin E-beta thalassemia - More common in SE Asian populations

Clinical Summary

Hemoglobin E disease is a disorder of the hemoglobin. A specific mutation in the hemoglobin causes red blood cells to become less functional causing mild anemia.

An affected neonate is likely to appear healthy, but has a risk for mild anemia and minor complications. Individuals with hemoglobin E disease are at risk for the following:

- Hemolytic anemia
- Splenic sequestration

Many children do not require any regular treatment for hemoglobin E disease. Hemoglobin E-beta thalassemia is likely to require treatment which could include blood transfusion.