# Fact Sheet **Positive Result:**

**Blood Spot Screen Result Notification** 

Minnesota Newborn Screening Program

# Hemoglobin E no A (hemoglobin E disease)

## **Next Steps**

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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.



