**Positive Result:**
Blood Spot Screen Result Notification

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**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.

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**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.