

Positive Result:

Blood Spot Screen Result Notification



Hemoglobin S no A (sickle cell 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, sepsis, fever); 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

FS (no A) is primarily associated with:

- Sickle cell disease — More common in African American and West African populations

Other (less likely) disorders to consider:

- Sickle beta-zero thalassemia
- Sickle hereditary persistence of fetal hemoglobin

Clinical Summary

Sickle cell (SS) disease is a disorder of the hemoglobin. A specific mutation in the hemoglobin causes it to become sickle-shaped. As a result of this alteration, red blood cells are less functional causing anemia and other vaso-occlusive complications.

An affected neonate is likely to appear healthy, but has a risk for sepsis and pneumonia. Individuals with sickle cell disease are at risk for the following:

- Hemolytic anemia
- Infection
- Pain episodes
- Splenic sequestration
- Stroke
- Vaso-occlusive complications

Treatment can include hydroxurea, prophylactic antibiotics, immunizations, pain management, blood transfusions, and stem cell transplantation.