

Positive Result:

Blood Spot Screen Result Notification



Hemoglobin S and C no A (hemoglobin SC 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

FSC (no A) is primarily associated with:

- Hemoglobin SC disease - More common in West African and African American populations

Clinical Summary

Hemoglobin SC disease is a disorder of the hemoglobin. Specific mutations 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. Hemoglobin SC disease is considered to be less severe than sickle cell disease.

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

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

Treatment can include hydroxurea, immunizations, pain management, and occasionally blood transfusions.