

# Positive Result:

## Blood Spot Screen Result Notification



## Hemoglobin C no A (hemoglobin C 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

FC (no A) is primarily associated with:

- Hemoglobin C disease — More common in African American and West African populations

Other (less likely) disorders to consider:

- Hemoglobin C/beta zero thalassemia
- Hemoglobin C/hereditary persistence of fetal hemoglobin

### Clinical Summary

Hemoglobin C disease is a disorder of the hemoglobin. A specific mutation in the hemoglobin causes red blood cells to become less functional causing mild anemia and other vaso-occlusive complications.

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

- Gallstones
- Hemolytic anemia
- Splenic sequestration

Many children do not require any regular treatment for hemoglobin C disease.