



How to respond to hemoglobin variant on a newborn screen

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There are many hemoglobin variants. Some are silent, causing no signs or symptoms; others affect the functionality and/or stability of the hemoglobin molecule. Examples of clinically significant hemoglobin variants include hemoglobin S, hemoglobin C, hemoglobin O-Arab and hemoglobin E. In a homozygous state or in combination with other abnormal hemoglobins, disease may result from the presence of these hemoglobin variants.

Variants that tend to be silent would include hemoglobin J or hemoglobin Raleigh. There are also some gamma globin chain variants that are closely related to fetal hemoglobin which disappear with age, typically within the first few months of life.

In the majority of cases, hemoglobin variants are not clinically significant in carrier status (heterozygous status). For hemoglobin variant traits that are associated with clinical findings, most will have no clinical significance until after the age of 6 months due to the predominance of hemoglobin F until that time.

If you have an infant with an unidentified hemoglobin variant trait on a newborn screen, we recommend the following:

At age 6-9 months, perform a hemoglobin electrophoresis to determine if the hemoglobin variant trait has persisted and can be identified.

- If the hemoglobin variant trait is no longer apparent on hemoglobin electrophoresis, then no further testing or follow-up is necessary.
- If the hemoglobin variant trait has persisted and is identified, Cook Children's Hematology and Oncology Center staff can assist you to determine if it is clinically significant and whether any hematology evaluation is necessary. Hematology evaluation would not be necessary for clinically benign hemoglobin variants.

Hematology consultations and programs

Sickle cell disease and hemoglobin S trait

Cook Children's Hematology and Oncology Center offers a sickle cell disease program. In addition to seeing sickle cell patients, we also offer genetic counseling for children with hemoglobin S trait.

Alpha thalassemia trait

For some with alpha thalassemia trait, as indicated by presence of hemoglobin Barts on a newborn screen, we offer in-office consultations. A mutation analysis would be recommended. Children of Southeast Asian descent particularly may be at risk for having offspring with alpha thalassemia or hydrops if they have a two gene deletion alpha thalassemia trait.

Referral

You may refer children with sickle cell trait, thalassemia trait or other clinically significant hemoglobin variants at any time without the need for repeat testing prior to referral. Preferably, we would see these children after the age of 6 months.

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