Newborn Screening ACT Sheet

Various Hemoglobinopathies

Condition Description: Hemoglobinopathies are inherited conditions that may alter the shape or amount of red blood cells in the body. There are many different hemoglobinopathies and while some can be severe and need treatment, others may not cause any clinical symptoms or manifestations and do not require intervention.

You Should Take the Following Actions

- **Contact the family to inform them of the screening result.**
- **Evaluate infant for increased sleeping, tiredness, shortness of breath, cold hands or feet, and pale skin.**
- **Review the baby's medical history and family history for jaundice, cyanosis, anemia and splenomegaly.**
- **Contact a pediatric hematologist to determine need for further testing.**
- **Report findings to the newborn screening program.**

Diagnostic Evaluation: Hemoglobin separation by electrophoresis, isoelectric focusing (IEF), or high performance liquid chromatography (HPLC). CBC and MCV may be recommended for further testing.

Clinical Considerations: Infants usually appear normal at birth. Severe hemoglobinopathies rarely manifest as hemolytic anemia, jaundice or cyanosis. Treatment may include fluids, pain medications and in some cases, blood transfusions. However, if infant is asymptomatic and has normal growth and development, there are no indications for further testing. Comprehensive care, including immunizations, regular checkups, family education and prompt treatment of acute illness, reduces morbidity and mortality.

Additional Information:

Hemoglobinopathies - Baby's First Test
http://www.babysfirsttest.org//newborn-screening/conditions/hemoglobinopathies

Sickle Cell Information Center: Variant Algorithm
https://scinfo.org/care-paths-and-protocols-children-adolescents/unidentified-hemoglobin-variants

Hemoglobinopathies Information - Texas Department of State Health Services
http://www.dshs.state.tx.us/newborn/sickle.shtm