Newborn Screening ACT Sheet

FCA (HbC/Beta Plus Thalassemia)

HbC/β+Disease

Differential Diagnosis: Hb C beta plus thalassemia.

Condition Description: A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin C and hemoglobin A. The hemoglobins are listed in order of the amount of hemoglobin present (F>C>A).

You Should Take the Following Actions

- **Contact the family to inform them of the screening result.**
- **Evaluate infant, assess for splenomegaly, and do complete blood count (CBC), red blood count (RBC), and mean corpuscular volume (MCV).**
- **Contact a pediatric hematologist to determine need for further testing.**
- **Initiate timely confirmatory/diagnostic testing as recommended by consultant.**
- **Report findings to newborn screening program**

Diagnostic Evaluation: CBC, RBC, and MCV. Hemoglobin separation by electrophoresis, isoelectric focusing, or high performance liquid chromatography (HPLC), shows FCA pattern. DNA studies are used to confirm genotype.

Clinical Considerations: Infant is usually normal at birth. Individuals with HbC beta plus thalassemia may have a mild anemia and splenomegaly, depending on the specific β + thalassemia mutation. The clinical manifestations range from mild to moderate hemolytic anemia and splenomegaly resembling thalassemia intermedia in severe cases.

Additional Information:

Hemoglobin C Disease (Texas Department of State Health Services)
http://www.dshs.state.tx.us/newborn/hemo_c1.shtm

Utah Department of Health
http://health.utah.gov/newbornscreening/Disorders/HB/Hb_C_Disease_CC/FactSheet_Family_HbCC_En.pdf

Hemoglobin Disorders (Grady Comprehensive Sickle Cell Center)
http://scinfo.org/additional-online-books-and-articles/hemoglobins-what-the-results-mean

Hemoglobin C Disease
http://emedicine.medscape.com/article/200853-overview