



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).

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);
- Repeat newborn screen if second screen has not yet been done;
- Contact a pediatric hematologist to determine need for further testing;
- Initiate timely confirmatory/diagnostic testing as recommended by consultant; and
- 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

[DSHS Hemoglobin C Disease](#)

[Medscape – Hemoglobin C Disease](#)

[Utah Department of Health & Human Services - Hemoglobin C, D, and E Disorders](#)