Newborn Screening ACT Sheet

[F]C

Hemoglobin CC Disease or Hemoglobin C/Beta Zero Thalassemia (HbC/β0 Disease)

**Differential Diagnosis:** Homozygous hemoglobin C, hemoglobin C/beta zero (β0) thalassemia, or hereditary persistence of fetal hemoglobin (Hb C/HPFH).

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

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**YOU SHOULD TAKE THE FOLLOWING ACTIONS:**

- Contact family to inform them of the screening result.
- Evaluate infant, assess for splenomegaly, and do complete blood count (CBC) and mean corpuscular volume (MCV).
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Consult a specialist in hemoglobin disorders; if patient is anemic for age, refer immediately.
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Report findings to newborn screening program.

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

**Clinical Considerations:** Infant is usually normal at birth. Hemoglobin CC is associated with a mild hemolytic anemia. Aplastic crises and gallstones may occur. Individuals with hemoglobin C/beta zero have a moderately severe anemia, splenomegaly, and may have bone changes. C-HPFH is clinically mild.

**Additional Information:**
- Hemoglobin Disorders (Grady Comprehensive Sickle Cell Center)
- Hemoglobin C
- Hemoglobin C Disease
- Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Care Paths and Protocols for Management of Acute and Chronic Complications

**Referral (local, state, regional and national):**
- Testing
- Clinical Services
  - Thalassemia Care Center Directory
  - Thalassemia Treatment Centers Directory
- Find Genetic Services
LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site (insert state newborn screening program website information)

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Local Resource Site (insert local and regional newborn screening website information)

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APPENDIX: Resources with Full URL Addresses

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  http://www.dshs.state.tx.us/newborn/pdf/sedona02.pdf

Referral (local, state, regional and national):
- Testing
- Clinical Services
  Thalassemia Care Center Directory
  http://www.cdc.gov/thalassemia/thal_center_list.htm
  Thalassemia Treatment Centers Directory

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx