

Newborn Screening ACT Sheet [FCA] Hemoglobin C/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) for Hb, red blood count (RBC), and mean corpuscular volume (MCV).
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Consult a specialist in hemoglobin disorders, refer if needed.
- 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 may be used to confirm genotype.

Clinical Considerations: Infant is usually normal at birth. Individuals with HbC beta plus thalassemia have a variable degree of 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 Disorders \(Grady Comprehensive Sickle Cell Center\)](#)

[Thalassemias](#)

[Genetics Home Reference](#)

[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](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

Hemoglobin Disorders (Grady Comprehensive Sickle Cell Center)

http://www.scinfo.org/index.php?option=com_content&view=article&id=218:hemoglobins-what-the-results-mean&catid=11&Itemid=21

Thalassemias

<http://kidshealth.org/parent/medical/heart/thalassemias.html#>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=betathalassemia>

Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Care Paths and Protocols for Management of Acute and Chronic Complications.

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

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2017?db=genetests&country=United%20States

Clinical Services

Thalassemia Care Center Directory

http://www.cdc.gov/ncbddd/hbd/thal_center_list.htm

Thalassemia Treatment Centers Directory

http://www.thalassemia.org/index.php?option=com_content&view=article&id=154:thalassemia-treatment-centers&catid=39:about-thalassemia&Itemid=27

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

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