

## Newborn Screening ACT Sheet

### [FC]

## Hemoglobin CC Disease or Hemoglobin C/Beta Zero Thalassemia (HbC/ $\beta^0$ Disease)

**Differential Diagnosis:** Homozygous hemoglobin C, hemoglobin C/beta zero ( $\beta^0$ ) 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 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](#)

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

Hemoglobin C

<http://www.doh.wa.gov/EHSPHL/PHL/newborn/pubs/Ceng.pdf>

Hemoglobin C Disease

<http://emedicine.medscape.com/article/200853-overview>

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](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](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](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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