

### **ACT Sheet**

### **Newborn Screening ACT Sheet**

## [Hemoglobin FCA]

# Hemoglobin C/Beta Plus Thalassemia (HbC/β+ Thalassemia)

Differential Diagnosis: Hb C trait.

Condition Description: Hemoglobin C/Beta Plus ( $\beta$ +) Thalassemia is an inherited type of red blood cell disorder characterized by abnormal hemoglobin production. It is due to genetic changes in the beta hemoglobin chain. Although asymptomatic at birth, a microcytic anemia develops within the first year of life. Clinical manifestations vary from a mild microcytic anemia to a moderate hemolytic anemia with splenomegaly.

### You Should Take the Following Actions:

- Inform the family of the newborn screening result.
- Ascertain clinical status (newborns are expected to be asymptomatic).
- Evaluate the newborn (newborns are expected to be asymptomatic and have a normal clinical exam. If significant signs or symptoms are identified, it is likely related to a different underlying disorder).
- Consult with pediatric hematologist with expertise in hemoglobin disorders within the first week of life with follow up typically recommended by 4 months of age (it is unusual to have symptoms before 3-6 months of life).
- Coordinate confirmatory diagnostic testing and management as recommended by specialist.
- Provide family with basic information about Hemoglobin C and Beta Plus (β+) Thalassemia.
- Refer for genetic counseling.
- Report final diagnostic outcome to newborn screening program.

**Diagnostic Evaluation:** The hemoglobins are listed in order of the amount of hemoglobin present on the newborn screen (F>C>A). <u>Isoelectric focusing, high performance liquid chromatography (HPLC) or capillary electrophoresis:</u> is used to confirm the newborn screening test result. <u>Complete blood count:</u> the CBC, reticulocyte count (with consideration of a smear) may be normal in infancy but a microcytic anemia develops within the first year of life. <u>Molecular genetic testing:</u> may be required to identify the beta thalassemia variant.

Clinical Considerations: The specific  $\beta$ + thalassemia variant determines the prognosis. Although asymptomatic at birth, individuals with HbC/ $\beta$ + Thalassemia develop a variable degree of anemia and splenomegaly, depending on the specific  $\beta$ + thalassemia variant. The typical clinical course includes a mild microcytic anemia and possible splenomegaly, but treatment is seldom required. Iron supplements should be avoided unless iron deficiency is documented.

#### Additional Information:

How to Communicate Newborn Screening Results
Gene Reviews
Medline Plus (Beta Thalassemia | Hemoglobin C Disease)
Clinicaltrials.gov

### Referral (local, state, regional, and national:

Find A Hematologist (Filter by Pediatric Hematology-Oncology)
Find a Genetics Clinic Directory
Genetic Testing Registry

This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource 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. 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 practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.



### **ACT Sheet**

### **Local Resources** (Insert Local Website Links)

State Resource Site (Insert Website Information)

Name	
URL	
Comments	

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Name	
URL	
Comments	

### **Appendix** (Resources with Full URL Addresses)

#### **Additional Information**

How to Communicate Newborn Screening Results

https://bit.ly/NBSResultsHRSA

Gene Reviews

• <a href="https://www.ncbi.nlm.nih.gov/books/NBK1426/">https://www.ncbi.nlm.nih.gov/books/NBK1426/</a>

Medline Plus

- Beta Thalassemia
  - o https://medlineplus.gov/genetics/condition/beta-thalassemia/
- Heomglobin C Disease
  - o <a href="https://medlineplus.gov/ency/article/000572.htm">https://medlineplus.gov/ency/article/000572.htm</a>

Condition Information for Families-HRSA Newborn Screening Clearinghouse

• https://newbornscreening.hrsa.gov/conditions/s-beta-thalassemia

Clinicaltrials.gov

• <a href="https://clinicaltrials.gov/">https://clinicaltrials.gov/</a>

#### Referral (local, state, regional and national)

Find A Hematologist (Filter by Pediatric Hematology-Oncology)

• https://www.hematology.org/education/patients/find-a-hematologist

Find a Genetics Clinic Directory

https://clinics.acmg.net

Genetic Testing Registry

• https://www.ncbi.nlm.nih.gov/gtr/

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