ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

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 (β+) 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). Isoelectric focusing, high performance liquid chromatography (HPLC) or capillary electrophoresis: is used to confirm the newborn screening test result. Complete blood count: 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. Molecular genetic testing: may be required to identify the beta thalassemia variant.

Clinical Considerations: The specific β+ thalassemia variant determines the prognosis. Although asymptomatic at birth, individuals with HbC/β+ Thalassemia develop a variable degree of anemia and splenomegaly, depending on the specific β+ 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

Local Resources *(Insert Local Website Links)*

State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix *(Resources with Full URL Addresses)*

Additional Information

How to Communicate Newborn Screening Results
- https://bit.ly/NBSResultsHRSA

Gene Reviews
- https://www.ncbi.nlm.nih.gov/books/NBK1426/

Medline Plus
- Beta Thalassemia
  - https://medlineplus.gov/genetics/condition/beta-thalassemia/
- Heomglobin C Disease
  - https://medlineplus.gov/ency/article/000572.htm

Condition Information for Families-HRSA Newborn Screening Clearinghouse
- https://newbornscreening.hrsa.gov/conditions/s-beta-thalassemia

Clinicaltrials.gov
- https://clinicaltrials.gov/

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/