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

ACT Sheet

Newborn Screening ACT Sheet

[Hemoglobin FC]

Hemoglobin C/C or Hemoglobin C/Beta Zero Thalassemia (Hb C/C or HbC/β^0 Thalassemia)

Differential Diagnosis: Homozygous Hemoglobin C; Hemoglobin C/Beta Zero (β^0) Thalassemia, Hemoglobin C/Beta Plus (β^+) Thalassemia.

Condition Description: Hemoglobin C/C or Hemoglobin C/β^0 Thalassemia are inherited types of red blood cell disorders characterized by abnormal hemoglobin production. They are due to genetic changes in the beta hemoglobin chain. Although asymptomatic at birth, both lead to a microcytic anemia within the first year of life. HbC/β^0 Thalassemia has a variable clinical course ranging from mild hemolytic anemia and splenomegaly to a moderate hemolytic anemia with jaundice, splenomegaly, and scleral icterus that may require intermittent transfusions. HbC/C is typically asymptomatic.

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 between 2-4 months of age (it is unusual to have symptoms before 2-4 months of life).
- Coordinate confirmatory diagnostic testing and management as recommended by specialist.
- Provide the family with basic information about Hemoglobin C/C or Hemoglobin C/Beta Zero (β^0) 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). 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) are often normal at birth but may demonstrate a microcytic anemia within the first year of life. Molecular genetic testing: is required to confirm the diagnosis and to distinguish homozygous Hb C/C from HbC/β^0 Thalassemia.

Clinical Considerations: Infants are usually asymptomatic. Hemoglobin C/C is associated with a mild microcytic hemolytic anemia and splenomegaly. Individuals with Hb C/β^0 Thalassemia have a moderately severe anemia and splenomegaly, and in some cases may require intermittent transfusions and develop iron overload. Iron supplements should be avoided unless iron deficiency is documented.

Additional Information:

How to Communicate Newborn Screening Results
GeneReviews
Medline Plus
Condition Information for Families- HRSA Newborn Screening Clearinghouse
Clinicaltrials.gov

Referral (local, state, regional, and national:

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

 (Funded in part through MCHB/HRSA/HHS grant #U22MC03957;#UH9MC30770; National Coordinating Center for the Regional Genetics Networks*)*

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
- https://medlineplus.gov/genetics/condition/beta-thalassemia/

Condition Information for Families-HRSA Newborn Screening Clearinghouse
- https://newbornscreening.hrsa.gov/conditions/various-other-hemoglobinopathies

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/

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