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

ACT Sheet

Newborn Screening ACT Sheet

[FA + Low/Moderate Barts Hb] FAB2, FAB1

Alpha (α) Thalassemia: Silent carrier and alpha thalassemia trait

Differential Diagnosis: α-thalassemia silent carrier (1 α-globin gene deletion), α-thalassemia trait (2 α-globin gene deletions in *cis* or *trans*), non-deletion α-thalassemia (e.g. Hb Constant Spring) with or without deletion of other genes; Hb H disease, prematurity. Hb Barts may be present with a structural hemoglobin variant.

Condition Description: The α-thalassemias are inherited types of red blood cell disorders characterized by abnormal hemoglobin production. The number of dysfunctional α-globin genes correspond directly to the relative decrease in α-globin chain production, resulting in an excess of γ- and β- globin chains. The severity of each disorder depends on the number of α genes affected. Individuals who are silent carriers or have an α-thalassemia trait are clinically unaffected.

You Should Take the Following Actions:

- Inform the family of the screening result.
- Ascertain clinical status (newborns are expected to be asymptomatic).
- Evaluate the newborn (newborns are expected to be asymptomatic).
- Coordinate confirmatory diagnostic testing as recommended by a genetic counselor.
- Provide family with basic information about α 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 (FA +Barts). Quantitative assay, preferably high performance liquid chromatography (HPLC) is used to quantify the amount of hemoglobin Barts present. Complete blood count: the red cell count is usually elevated and mean corpuscular volume is characteristically low dependent on the type of α-thalassemia. Molecular genetic testing may be performed to definitively diagnose and to provide reproductive risk counseling for α-thalassemia trait or silent carriers.

Barts levels decrease rapidly after birth and vary significantly depending on time of collection and methodology. Diagnostic specificity varies widely between NBS programs. It is essential to work with the State Health Department, thalassemia specialist, and/or genetic counselor on a detailed interpretation and response.

Clinical Considerations: Individuals are asymptomatic with laboratory features that are normal or may resemble iron deficiency anemia with a significantly decreased MCV, slightly reduced hemoglobin, and elevated red blood cell count. Newborns with suspected mild forms of α-thalassemia such as α-thalassemia trait or silent carriers can be followed by primary care providers after referral for genetic counseling. No routine monitoring is necessary after diagnostics and genetic counseling. Iron deficiency should be documented before initiating iron supplementation. Parents should be reassured that the child will not have medical issues related to this disorder.

Additional Information:

How to Communicate Newborn Screening Results
Gene Reviews
Medline Plus
Condition Information for Families- HRSA Newborn Screening Clearinghouse
Cooley's Anemia Foundation
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/NBK1435/

Medline Plus
- https://medlineplus.gov/genetics/condition/alpha-thalassemia/

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

Cooley's Anemia Foundation
- https://www.thalassemia.org

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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