

Newborn Screening ACT Sheet

[FA + Barts Hb]

Alpha (α) Thalassemia (phenotype varies with % Barts Hb)

Differential Diagnosis: Hemoglobin A/Barts, alpha thalassemia carrier, hemoglobin H disease, alpha thalassemia major.

Condition Description: A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin A, as well as one or more alpha globin mutations (resulting in hemoglobin Barts).

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) for Hb, and mean corpuscular volume (MCV).
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Consult a specialist in hemoglobin disorders; refer if needed.
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Report findings to newborn screening program.

Diagnostic Evaluation: CBC and MCV. Hemoglobin separation by electrophoresis, isoelectric focusing (IEF), or high performance liquid chromatography (HPLC), shows FA+Barts pattern. DNA studies may be used to confirm genotype.

Clinical Considerations: Severity depends on the number of the four alpha globin genes that are lost. Presence of less than 25% Hb Barts indicates loss of one (silent carrier) or two (alpha thalassemia trait) genes. Individuals are asymptomatic with laboratory features that are normal or may resemble iron deficiency anemia. Hemoglobin Barts above 25% in the newborn indicates a hemoglobin H disease, a clinically significant form of alpha thalassemia, is likely. Deletion or dysfunction of 3 of the 4 alpha globin genes manifests as Hb H disease. Hb H disease is characterized by splenomegaly and anemia that may require intermittent transfusions. Absence of all four alpha globin genes results in hydrops fetalis and is usually fatal, in utero or shortly after birth.

Additional Information:

[Hemoglobin Disorders \(Grady Comprehensive Sickle Cell Center\)](#)

[Thalassemias](#)

[Genetics Home Reference](#)

[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 style="width: 85%;" type="text"/>
URL	<input style="width: 85%;" type="text"/>
Comments	<input style="width: 85%; height: 40px;" type="text"/>

Local Resource Site (insert local and regional newborn screening website information)

Name	<input style="width: 85%;" type="text"/>
URL	<input style="width: 85%;" type="text"/>
Comments	<input style="width: 85%; height: 40px;" 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

Thalassemias

<http://kidshealth.org/parent/medical/heart/thalassemias.html#>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=betathalassemia>

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

Clinical Services

Thalassemia Care Center Directory

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

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

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