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

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

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

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

Name	
URL	
Comments	

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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