Newborn Screening ACT Sheet
[F (Fetal Hemoglobin) Only]
Beta Thalassemia Major

Differential Diagnosis: Homozygous beta zero thalassemia (thalassemia major), hereditary persistence of fetal hemoglobin (HPFH), and prematurity.

Condition Description: A red blood cell disorder characterized by a lack of normal beta globin production and absence of Hb A (F [fetal Hb] only).

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, red blood count (RBC), and mean corpuscular volume (MCV).
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Consult a specialist in hemoglobin disorders; if patient is anemic for age, refer immediately.
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Report findings to newborn screening program.

Diagnostic Evaluation: Hemoglobin separation by electrophoresis, isoelectric focusing, or high performance liquid chromatography (HPLC), shows F-only pattern. DNA studies may be used to confirm genotype.

Clinical Considerations: Infants with this finding are usually normal. With beta-thalassemia, severe anemia may develop in the first few months of life. Complications include growth retardation, intercurrent infections, progressive hepatosplenomegaly, skeletal abnormalities, and severe iron overload. Comprehensive care including family education, immunizations, regular transfusions, and prompt treatment of acute illness reduces morbidity and mortality.

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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