

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](#)

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.

LOCAL RESOURCES: Insert State newborn screening program web site links

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

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

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input 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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