

Newborn Screening ACT Sheet

[FSA]

Hemoglobin S/Beta plus Thalassemia (HbSβ⁺ Disease)

Differential Diagnosis: Sickle beta plus thalassemia. The hemoglobins are listed in order (F>S>A) of the amount of hemoglobin present. This result is different from FAS which is consistent with sickle carrier (trait).

Condition Description: Individuals with sickle beta plus thalassemia, a form of sickle cell disease, are compound heterozygotes for the Hb S and beta-thalassemia mutations in the beta-globin genes.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result.
- Perform a physical exam on the infant and assess for splenomegaly.
- Obtain a blood sample for confirmatory testing and a complete blood count (CBC) with reticulocyte count.
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Initiate penicillin (PenVK 125mg po bid) prophylaxis.
- Educate parents/caretakers regarding the risk of sepsis and advise that infant be immediately evaluated if a fever of $\geq 38.5^{\circ}$ C (101° F) is present.
- Contact a specialist in hemoglobin disorders for consultation on diagnostic evaluation and management.

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

Clinical Considerations: Infants are usually normal at birth. Later potential clinical problems include mild to moderate hemolytic anemia, life-threatening infection, vaso-occlusive pain episodes, dactylitis, and chronic organ damage. Prompt treatment of infection and splenic sequestration is associated with decreased mortality in the first three years of life.

Additional Information:

[Grady Comprehensive Sickle Cell Center Management and Therapy of Sickle Cell Disease](#)
[Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Protocols for Management of Acute and Chronic Complications](#)
[American Academy of Pediatrics](#)
[Sickle Cell Disease Association of America](#)

Referral (local, state, regional and national):

[Testing](#)
 Clinical Services
[Comprehensive Sickle Cell Center Directory](#)
[Sickle Cell Information Center](#)
[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:

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

Management and Therapy of Sickle Cell Disease

<http://www.nhlbi.nih.gov/health/prof/blood/sickle/index.htm>

Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Protocols for Management of Acute and Chronic Complications

<http://www.dshs.state.tx.us/newborn/pdf/sedona02.pdf>

American Academy of Pediatrics

<http://pediatrics.aappublications.org/cgi/content/full/109/3/526>

Sickle Cell Disease Association of America

<http://www.sicklecelldisease.org/>

Referral (local, state, regional and national):

Testing

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2028?db=genetests&country=United%20States

Clinical Services

Comprehensive Sickle Cell Center Directory

http://www.scinfo.org/index.php?option=com_content&view=article&id=197&Itemid=34

Sickle Cell Information Center

<http://www.scinfo.org/>

Find Genetic Services

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

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