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 ≥ 38.5° 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

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:

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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