



Newborn Screening ACT Sheet

[FSA]

Hemoglobin S/Beta⁺ - Thalassemia (HbSβ⁺ Disease)

Differential Diagnosis: Hemoglobin FSA pattern on newborn screen is highly suggestive of **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⁺ 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 with reticulocyte count.
- 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 $\geq 38.5^{\circ}\text{C}$ (101°F) is present.
- Contact a specialist in hemoglobinopathies for consultation on diagnostic evaluation and management.

Confirmation of Diagnosis: Hemoglobin separation by electrophoresis, isoelectric focusing or HPLC showing FSA. Family or DNA studies may be used to confirm genotype.

Clinical Expectations: Infants are usually normal at birth. Later potential clinical problems include mild 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:

(Click on the name to take you to the website. Complete URLs are listed in the Appendix)

[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):

[Comprehensive Sickle Cell Center Directory](#)

[Sickle Cell Information Center](#)

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines 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. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines.

APPENDIX: Resources with Full URL Addresses

Additional Information:

Grady Comprehensive Sickle Cell Center <http://scinfo.org/hemoglb.htm#SICKLE%20HEMOGLOBINS>
<http://www.scinfo.org/hemoglb.htm#BETA%20THALASSEMIAS>

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://sicklecelldisease.org/>

Referral (local, state, regional and national):

Comprehensive Sickle Cell Center Directory <http://www.rhofed.com/sickle/index.htm>

Sickle Cell Information Center <http://www.scinfo.org/clinics.htm>

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines 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. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines.