



# Newborn Screening ACT Sheet

[FS]

## Sickle Cell Anemia (HbSS Disease or HbS/Beta Zero Thalassemia)

**Differential Diagnosis:** Homozygous sickle cell disease (Hb SS), sickle beta-zero thalassemia, or sickle hereditary persistence of fetal hemoglobin (S-HPFH).

**Condition Description:** A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin S in the absence of hemoglobin A. The hemoglobins are listed in order of the amount of hemoglobin present (F>S). This result is different from FAS which is consistent with sickle carrier.

### **You Should Take the Following Actions:**

- Contact the family to inform them of the screening result.
- Consult a specialist in hemoglobinopathies; refer if needed.
- Evaluate infant and assess for splenomegaly.
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Initiate daily penicillin VK (125mg po bid) prophylaxis and other treatment as recommended by the consultant.
- Educate parents/caregivers regarding the risk of sepsis, the need for urgent evaluation if fever of  $\geq 38.5^{\circ}\text{C}$  ( $101^{\circ}\text{F}$ ) or signs and symptoms of splenic sequestration.

**Diagnostic Evaluation:** Hemoglobin separation by electrophoresis, isoelectric focusing or HPLC showing FS pattern. Family or DNA studies may be used to confirm genotype. Sickledex is not appropriate for confirmation of diagnosis in infants.

**Clinical Considerations** – Newborn infants are usually well. Hemolytic anemia and vaso-occlusive complications develop during infancy or early childhood. Complications include life-threatening infection, splenic sequestration, pneumonia, acute chest syndrome, pain episodes, aplastic crisis, dactylitis, priapism and stroke. Comprehensive care including family education, immunizations, prophylactic penicillin and prompt treatment of acute illness reduces morbidity and mortality. S-HPFH is typically benign.

### **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 Care Paths and Protocols for Management of Acute and Chronic Complications**

**American Academy of Pediatrics**

**Sickle Cell Disease Association**

**Resources for Referral (Insert local, state, regional and national resources):**

**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.*

**State Resource Site** *(insert state newborn screening program website information)*

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Name  
URL  
Comments

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

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Name  
URL  
Comments

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Name  
URL  
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Comments

## APPENDIX: Resources with Full URL Addresses

### *Additional Information:*

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

**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 Care Paths and Protocols for Management of Acute and Chronic Complications**

[http://www.tdh.state.tx.us/newborn/sc\\_guide.htm](http://www.tdh.state.tx.us/newborn/sc_guide.htm)

**American Academy of Pediatrics** <http://pediatrics.aappublications.org/cgi/content/full/109/3/526>

**Sickle Cell Disease Association** <http://www.sicklecelldisease.org>

### *Resources for Referral (Insert local, state, regional and national resources):*

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

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

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