

Disease Name	Sickle Cell Disease
Alternate Name(s)	Sickle Cell Anemia
Acronym	Hb S/S
Disease Classification	Hemoglobinopathy
Symptom onset	May be asymptomatic.
Symptoms	Any sign of illness in an infant with sickling disease is a potential medical emergency. Acute and chronic tissue injury can occur when sickled cells cause vascular occlusion. Sickling diseases can cause severe pain anywhere in the body, but most often in the hands, arms, chest, legs and feet. Complications may include, but are not limited to, the following: sepsis, acute chest syndrome, hand-and-foot syndrome, splenic sequestration crisis, aplastic crisis, stroke and painful episodes.
Natural history without treatment	Infants with sickle cell disease are vulnerable to serious bacterial infections that can be life threatening.
Natural history with treatment	Reduced mortality and morbidity with penicillin prophylaxis.
Treatment	The National Institutes of Health clinical guidelines for management of sickle cell disease state, "Penicillin prophylaxis should begin by 2 months of age for infants with suspected sickle cell anemia, whether or not the definitive diagnosis has been established." Antibiotic therapy should continue until at least 5 years of age.
Inheritance	Autosomal recessive
General population incidence	1:500 for African American and Hispanic American.
OMIM Link	http://www.ncbi.nlm.nih.gov/omim/253260
Genetests Link	www.geneclinics.org
Support Group	Sickle Cell Information Center http://www.scinfo.org/ Sickle Cell Disease Association of America, Inc. http://www.sicklecelldisease.org

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 hemoglobin disorders; refer if needed.
- Evaluate infant and assess for splenomegaly; do complete blood count (CBC) with mean corpuscular volume (MCV), and reticulocyte count.
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- 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°F) or signs and symptoms of splenic sequestration.

Diagnostic Evaluation: CBC, MCV, and reticulocyte count. Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FS pattern. 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 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](#)

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.

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