

NEWBORN SCREENING ACT SHEET

Newborn Screening Result (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 cell trait.

ACTION TO BE TAKEN:

- ✚ Contact a specialist in hemoglobinopathies for consultation or referral on diagnostic evaluation and management.
- ✚ Contact the family to inform them of the screening result.
- ✚ Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- ✚ Initiate penicillin VK prophylaxis and other treatment as recommended by consultant.
- ✚ Educate parents regarding the risk of sepsis, the need for urgent evaluation for fever of $\geq 38.5^{\circ}$ C (101° F) or signs and symptoms of splenic sequestration.
- ✚ Report findings to the Newborn Screening Program.

Pediatric specialists in hemoglobinopathies are available at the Hemoglobinopathy (Sickle Cell) Resource Centers below.

CONFIRMATION OF DIAGNOSIS: Complete blood count (CBC), mean corpuscular volume (MCV), and reticulocyte count. Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FS pattern. Family or DNA studies may be used to confirm genotype. Sickledex is not appropriate for confirmation of diagnosis in infants.

CLINICAL EXPECTATIONS: 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.

HEMOGLOBINOPATHY (SICKLE CELL) RESOURCE CENTERS:

Children's Mercy Hospital

Kansas City, MO 816-302-6808
Laurence N, M.D., Pediatric Hematologist
Susie Sarcone-Jones, RN, CPNP, Nurse Coordinator

Cardinal Glennon Children's Hospital

St. Louis, MO 314-268-4000
Leili Dolatshahi, M.D., Pediatric Hematologist
Abigail Sharamitaro, RN, MSN, CPNP, Nurse Coordinator

Children's Hospital – University of MO Health Care

Columbia, MO 573-882-3961
Alicia Bach, M.D., Pediatric Hematologist
Cara M. Hirner, RN CPN, Nurse Coordinator

St. Louis Children's Hospital

St. Louis, MO 314-454-6018
Monica Hulbert, M.D., Pediatric Hematologist
Alison Towerman, RN, CPNP, Nurse Coordinator

DISCLAIMER: These guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. These 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. Adherence to these guidelines does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the physician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Health care providers are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with these guidelines.