

NEWBORN SCREENING ACT SHEET

Newborn Screening Result (FSC) Hemoglobin SC Disease (HbSC)

Differential Diagnosis: Hemoglobin SC Disease.

Condition Description: A red blood cell disorder characterized by the presence of fetal hemoglobin (F) and hemoglobins S and C in the absence of hemoglobin A. The hemoglobins are listed in order of the amount of hemoglobin present (F>S>C). 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 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 Newborn Screening Program.

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

CONFIRMATION OF DIAGNOSIS: Complete blood count (CBC). Hemoglobin separation by electrophoresis, isoelectric focusing, or high performance liquid chromatography (HPLC) showing FSC pattern. Family or DNA studies may be used to confirm genotype.

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. Hemoglobin SC Disease is generally not as severe as Hemoglobin SS Disease. Comprehensive care including family education, immunizations, prophylactic penicillin, and prompt treatment of acute illness reduces morbidity and mortality.

HEMOGLOBINOPATHY (SICKLE CELL) RESOURCE CENTERS:

Region 1

Children's Mercy Hospital
Kansas City, MO
Gerald Woods, M.D.
Pediatric Hematologist
Susie Sarcone, RN, CPNP
Clinical Nurse Coordinator
816-234-3265

Region 2

University Hospital and Clinics
Columbia, MO
Barbara Gruner, M.D.
Pediatric Hematologist
Elizabeth Gunier, RN
Clinical Nurse Coordinator
573-882-3961

Region 3

St. Louis Children's Hospital
St. Louis, MO
Monica Hulbert, M.D.
Pediatric Hematologist
Alison Towerman, RN, CPNP
Clinical Nurse Coordinator
314-286-1167

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.