

# NEWBORN SCREENING ACT SHEET

## Newborn Screening Result (FSA) Hemoglobin S/Beta plus Thalassemia (HbS $\beta$ + Disease)

**Differential Diagnosis:** 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 cell trait.

**Condition Description:** Individuals with sickle beta plus thalassemia, a form of sickle cell disease, are compound heterozygotes for the Hb S and beta-thalassemia mutations in the beta-globin genes.

### ACTION TO BE TAKEN:

- ✚ Contact a specialist in hemoglobinopathies for consultation on diagnostic evaluation and management.
- ✚ Contact the family to inform them of the screening result.
- ✚ Obtain a blood sample for confirmatory testing and a complete blood count (CBC) with reticulocyte count.
- ✚ Initiate treatment as recommended by consultant.
- ✚ Educate parents/caregivers regarding the risk of sepsis and advise that infant be immediately evaluated if a fever of  $\geq 38.5^{\circ}$  C ( $101^{\circ}$  F) is present.
- ✚ Report findings to Newborn Screening Program.

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

**CONFIRMATION OF DIAGNOSIS:** Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) showing FSA pattern. 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. Sickle Beta Plus Thalassemia is generally not as severe as Hemoglobin SS Disease. Prompt treatment of infection and splenic sequestration is associated with decreased mortality in the first three years of life.

### HEMOGLOBINOPATHY (SICKLE CELL) RESOURCE CENTERS:

#### Children's Mercy Hospital

Kansas City, MO 816-302-6808  
Laurence Noisette, M.D., Pediatric Hematologist  
Susie Sarcone-Jones, RN, 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

#### Cardinal Glennon Children's Hospital

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