NEWBORN SCREENING ACT SHEET

Newborn Screening Result **(FSA)**
Hemoglobin S/Beta plus Thalassemia **(HbSβ+ 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 ≥ 38.5º C (101º 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

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