



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: HEMOGLOBINS F, S & A

CONDITION: HEMOGLOBIN S/BETA⁺ - THALASSEMIA
(HbSβ⁺ DISEASE)

DIFFERENTIAL DIAGNOSIS: Hemoglobin FSA pattern on newborn screen is highly suggestive of sickle beta plus thalassemia. The hemoglobins are listed in order (F>S>A) of the amount of hemoglobin present. This result is different than FAS which is consistent with sickle carrier (trait).

METABOLIC DESCRIPTION: Individuals with sickle beta⁺ 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 the family to inform them of the screening result.
- ◆ Perform a physical exam on the infant and assess for splenomegaly.
- ◆ Obtain a blood sample for confirmatory testing and a complete blood count with reticulocyte count.
- ◆ Contact a consultant in hemoglobinopathies for diagnostic evaluation and management.
- ◆ Initiate penicillin (Pen VK) prophylaxis and other treatment as recommended by consultant.
- ◆ Educate parents/caregivers regarding the risk of sepsis and advise that infant be immediately evaluated if fever of $\geq 38.5^{\circ}\text{C}$ (101°F) is present.
- ◆ Follow-up at six months of age.
- ◆ Report findings to newborn screening program.

CONFIRMATION OF DIAGNOSIS: Hemoglobin separation by electrophoresis, isoelectric focusing or 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. Prompt treatment of infection and splenic sequestration is associated with decreased mortality in the first three years of life.

REPORTING: Report diagnostic result to family and Kansas NBS program.

CONSULTANTS:

Dr. Thomas Loew
Pediatric Hematologist
KU Dept. of Pediatric Hematology
Kansas City, KS
Office: 913-588-6340

Dr. Youman Othman
Pediatric Hematologist
Cotton – O’Neil Clinic
Topeka, KS
Office: 785-270-4980

Dr. Rajoo Thapa
Pediatric Hematologist
Pediatric Faculty Clinic
Wichita, KS
Office: 316-962-2682