



KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: HEMOGLOBINS F & S

CONDITION: 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).

METABOLIC 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 carrier.

ACTION TO BE TAKEN:

- ◆ Contact the family to inform them of the screening result.
- ◆ Contact a consultant in hemoglobinopathies; refer if needed.
- ◆ Evaluate infant and assess for splenomegaly.
- ◆ Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- ◆ Initiate penicillin VK prophylaxis and other treatment as recommended by consultant.
- ◆ Educate parents/caregivers regarding the risk of sepsis, the need for urgent evaluation if fever of $\geq 38.5^{\circ}\text{C}$ (101°F) or signs and symptoms of splenic sequestration.
- ◆ 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 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, 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.

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

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