KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: HEMOGLOBINS F, S & C

CONDITION: HEMOGLOBIN SC DISEASE (HbSC)

***DIFFERENTIAL DIAGNOSIS**:* Hemoglobin SC disease most likely.

METABOLIC DESCRIPTION: A red blood cell disorder characterized by 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 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 treatment as recommended by consultant.
- Educate parents/caregivers regarding the risk of sepsis, the need for urgent evaluation if fever of ≥ 38.5° 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 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. Comprehensive care including family education, immunizations, prophylactic penicillin and prompt treatment of acute illness reduces morbidity and mortality.

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

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DISCLAIMER: These standards and 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. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines