TEXAS Health and Human Services | Texas Department of State Health Services

Newborn Screening ACT Sheet

[FS] Hemoglobin SS Disease (Hb SS) or Hemoglobin S Beta Zero Thalassemia (Hb Sβ0Thalassemia)

Sickle Cell Disease

Differential Diagnosis

Hemoglobin FS pattern on newborn screen is highly suggestive of homozygous sickle cell disease (Hb SS), sickle beta-zero thalassemia (Hb Sβ0thal). Sickle hereditary persistence of fetal hemoglobin (Hb SHPFH) may be detected but is not a sickling condition. 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 cell trait.

Condition Description

A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin S in the absence of hemoglobin A. Individuals with Hemoglobin SS, a form of sickle cell disease, are homozygous for hemoglobin S mutation in both beta-globin genes. Individuals with sickle beta-zero thalassemia, a form of sickle cell disease, are heterozygotes for hemoglobin S and a beta thalassemia mutation that does not produce hemoglobin A in the beta globin gene.

Take the Following Actions

- Contact the family to inform them of the screening result;
- Consult with a pediatric hematologist (See attached list.);
- Perform physical exam on infant;
- Repeat newborn screen if second screen has not yet been done;
- Initiate daily penicillin prophylaxis and other treatment as recommended by the consultant;
- Educate parents/caregivers regarding the risk of sepsis, the need for urgent evaluation if fever of ≥ 101.5° F, or signs and symptoms of splenic sequestration; and
- Report findings to newborn screening program.

Diagnostic Evaluation

The Newborn screening program performs DNA studies to confirm S alleles. Sickle beta-zero thalassemia, or sickle hereditary persistence of fetal hemoglobin (Hb SHPFH), may not be detected by newborn screening DNA testing. Clinicians may choose to obtain further molecular diagnostic studies as indicated.

Clinical Considerations

Newborn infants are usually well. Hemolytic anemia and vaso-occlusive complications develop during infancy or early childhood. Potential 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.

Additional Information

Sickle Cell Disease Association of America
Centers for Disease Control and Prevention - Sickle Cell Disease
DSHS Sickle Cell Disease
National Institute of Health: Evidence-Based Management of Sickle Cell Disease
2019 sickle cell disease guidelines by the American Society of Hematology

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