

ACT SHEET FOR POSITIVE NEWBORN SCREENING RESULT (FAS) SICKLE CELL TRAIT (Hb AS)

Disease Category: Hemoglobinopathy

Meaning of the Screening result: Hemoglobin FAS pattern on newborn screen is highly suggestive of sickle trait. However, IEF (the test used for screening) does not quantitate results. Without quantitation those newborns with A and S hemoglobins where the S hemoglobin is nearly as much as the A hemoglobin, could be at risk of a thalassemia.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result and to offer education and counseling.
- Reassure the family that infants do not have clinical problems related to the carrier state for hemoglobin S.
- Order confirmatory testing (hemoglobin electrophoresis).
- Encourage parents to seek genetic counseling and testing.
- Report findings to Nebraska Newborn Screening Program.

Pediatric specialists in hemoglobinopathies are available at Children's Hospital (402) 955-3950 and UNMC/Nebraska Medical Center (402) 559-7257.

Condition Description: Individuals with sickle cell trait are carriers of the gene for hemoglobin S. There are no other laboratory abnormalities associated with this genotype.

Clinical Expectations: Prognosis is for a normal life expectancy. Carriers are at risk for having children affected with sickle cell disease. Older children and adults may have hematuria. Rarely, splenic infarction and sudden death may occur with severe hypoxia and extreme dehydration. An increased risk of venous thromboembolism, especially pulmonary embolism, has also been reported.

Confirmation of Diagnosis: Diagnosis is confirmed by hemoglobin electrophoresis and parental or DNA studies as indicated. (However, if on confirmation a thalassemia or co-existent thalassemia is identified, it is important to refer these patients to pediatric hematology for further work-up, to determine clinical significance).

Additional information:

- Acute and Chronic Complications - www.tdh.state.tx.us/newborn/sc_guide.htm
- Grady Comprehensive Sickle Cell Center Web Site - www.scinfo.org
- Management and Therapy of Sickle Cell Disease - www.nhlbi.nih.gov/health/prof/blood/sickle/index.htm
- Sickle Cell Disease Association - www.sicklecelldisease.org
- Hemoglobinopathy Podcasts <http://feeds.feedburner.com/Pediatricsbytes>

