Newborn Screening ACT Sheet
[FS]
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).

Condition 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.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result.
- Consult a specialist in hemoglobin disorders; refer if needed.
- Evaluate infant and assess for splenomegaly; do complete blood count (CBC) with mean corpuscular volume (MCV), and reticulocyte count.
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Initiate timely confirmatory/diagnostic testing as recommended by consultant.
- Initiate daily penicillin VK (125mg po bid) 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 ≥ 38.5° C (101° F) or signs and symptoms of splenic sequestration.

Diagnostic Evaluation: CBC, MCV, and reticulocyte count. Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FS pattern. DNA studies may be used to confirm genotype. Sickledex is not appropriate for confirmation of diagnosis in infants.

Clinical Considerations: 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. S-HPFH is typically benign.

Additional Information:

Grady Comprehensive Sickle Cell Center
Management and Therapy of Sickle Cell Disease
Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Protocols for Management of Acute and Chronic Complications
American Academy of Pediatrics
Sickle Cell Disease Association of America

Referral (local, state, regional and national):

Testing
Clinical Services
- Comprehensive Sickle Cell Center Directory
- Sickle Cell Information Center

Find Genetic Services

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	Quest To Cure Sickle Cell Foundation | Office for Children with Special Health Care Needs | Office of Newborn Screening
URL	www.questtocure.org | www.azdhs.gov/phs/owch/ocshcn | www.aznewborn.com
Comments	Quest to Cure Sickle Cell Foundation: (602) 200-0215 Office for Children With Special Health Care Needs: (602) 542-1860 Office of Newborn Screening: (602) 364-1409

Local Resource Site *(insert local and regional newborn screening website information)*

Name	Introduction to Sickle Cell Trait - Physician Training Module
URL	www.coloradosicklecellcenter.org/SickleCellTraitCourse/module1/index.htm
Comments	CONTRACTED SPECIALISTS PCH - Center for Cancer and Blood Disorders Meena Goyal-Khemka, MD; Christine Knoll, MD; Sanjay Shah, MD (602) 546-0920 UMC - Pediatrics Hematology & Oncology Brenda J. Wittman, MD (520) 626-4851

APPENDIX: Resources with Full URL Addresses

Additional Information:

Grady Comprehensive Sickle Cell Center
http://www.scinfo.org/index.php?option=com_content&view=article&id=218:hemoglobins-what-the-results-mean&catid=11&Itemid=21

Management and Therapy of Sickle Cell Disease
http://www.nhlbi.nih.gov/health/prof/blood/sickle/index.htm

Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Protocols for Management of Acute and Chronic Complications
http://www.dshs.state.tx.us/newborn/pdf/sedona02.pdf

American Academy of Pediatrics
http://pediatrics.aappublications.org/cgi/content/full/109/3/526

Sickle Cell Disease Association of America
http://www.sicklecelldisease.org/

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2028?db=genetests&country=United%20States

Clinical Services

Comprehensive Sickle Cell Center Directory
http://www.scinfo.org/index.php?option=com_content&view=article&id=197&Itemid=34

Sickle Cell Information Center
http://www.scinfo.org/

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx