ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

ACT Sheet

Newborn Screening ACT Sheet
[Hemoglobin FSC]
Hemoglobin S/C

Differential Diagnosis: None.

Condition Description: Hemoglobin S/C is an inherited type of red blood cell disorder characterized by abnormal hemoglobin production. It is due to genetic changes in the beta hemoglobin chain. Although asymptomatic at birth, symptoms begin as Hb F decreases and Hb S and Hb C predominate. The clinical course is highly variable, ranging from asymptomatic to infections, splenic sequestration, pain crises, acute chest syndrome, bone damage, retinopathy, and organ damage as the individual ages.

You Should Take the Following Actions:

- Inform the family of the newborn screening result.
- Ascertain clinical status (newborns are expected to be asymptomatic).
- Evaluate the newborn (assess for splenomegaly and send CBC).
- Administer prophylactic penicillin.
- Consult with a sickle cell specialist immediately with in-person follow up no later than 12 weeks of age.
- Coordinate confirmatory diagnostic testing and management as recommended by specialist.
- Provide family with basic information about Hemoglobin S/C including the need for urgent evaluation for fever ≥38.5 C (101 F), pallor, unexplained irritability, risks of sepsis, or other signs of illness.
- Refer for genetic counseling.
- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: The hemoglobins are listed in order of the amount of hemoglobin present on the newborn screen (F>S>C). Isoelectric focusing, high performance liquid chromatography (HPLC), or capillary zone electrophoresis: are used to confirm the newborn screening result. Complete blood count: the CBC, smear, and reticulocyte count may be normal at birth but over the first months of life demonstrate a worsening anemia, with an increasing reticulocyte count and sickle and other abnormal cells on smear. Molecular genetic testing may be used to confirm the diagnosis.

Clinical Considerations: Newborns with Hemoglobin S/C are generally asymptomatic. Hemolytic anemia and vaso-occlusive complications can develop during infancy or in early childhood. Although patients initially have a milder clinical course and have an increased life expectancy than those with sickle cell anemia (Hb S/S), symptoms can become more severe with age. Without appropriate treatment, complications may include life-threatening infection, splenic sequestration, pneumonia, acute chest syndrome, pain crises, aplastic crises, dactylitis, priapism and osteonecrosis. Comprehensive care including family education, a modified immunization schedule, prompt treatment of infection and of acute vaso-occlusive events, screening for early signs of organ damage, consideration of prophylactic penicillin and disease-modifying therapies reduces morbidity and mortality. Management should be done under the direction of a sickle cell specialist. Solubility testing (Sickledex) should not be used to confirm the diagnosis. Iron supplements should avoided unless iron deficiency is documented.

Additional Information:

How to Communicate Newborn Screening Results
Gene Reviews
Medline Plus
Sickle Cell Disease Association of America
Condition Information for Families- HRSA Newborn Screening Clearinghouse
Clinicaltrials.gov

Referral (local, state, regional, and national:

Find A Hematologist (Filter by Pediatric Hematology-Oncology)
Find a Genetics Clinic Directory
Genetic Testing Registry
National Alliance of Sickle Cell Centers

This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.

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Local Resources *(Insert Local Website Links)*

State Resource Site (Insert Website Information)

Name	
URL	
Comments	

Local Resource Site (Insert Website Information)

Name	
URL	
Comments	

Appendix *(Resources with Full URL Addresses)*

Additional Information

How to Communicate Newborn Screening Results
- https://bit.ly/NBSResultsHRSA

Gene Reviews
- https://www.ncbi.nlm.nih.gov/books/NBK1377/

Medline Plus
- https://medlineplus.gov/genetics/condition/sickle-cell-disease/

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

Condition Information for Families-HRSA Newborn Screening Clearinghouse
- https://newbornscreening.hrsa.gov/conditions/sc-disease

Clinicaltrials.gov
- https://clinicaltrials.gov/

Referral (local, state, regional and national)

Find A Hematologist (Filter by Pediatric Hematology-Oncology)
- https://www.hematology.org/education/patients/find-a-hematologist

Find a Genetics Clinic Directory
- https://clinics.acmg.net

Genetic Testing Registry
- https://www.ncbi.nlm.nih.gov/gtr/

National Alliance of Sickle Cell Centers
- https://sicklecellcenters.org/

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