

ACT Sheet

Newborn Screening ACT Sheet

[Hemoglobin FAS] Hemoglobin A/S

Differential Diagnosis: Hemoglobin S/Beta Plus (β+) Thalassemia

Condition Description: Hemoglobin A/S is an inherited red blood cell disorder characterized by abnormal hemoglobin production. It is due to genetic changes in the beta hemoglobin chain. It is generally an asymptomatic genetic carrier state although adults may develop sickling and hypoxia at high altitude, severe dehydration, or extreme exertion.

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.
- Provide family with basic information about sickle cell trait and sickle cell disease.
- Initiate confirmatory diagnostic testing and management as recommended by genetic counselor.
- 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 (F>A>S). <u>Isoelectric focusing, high performance liquid chromatography (HPLC) or capillary zone electrophoresis</u> is used to confirm the newborn screening result. <u>Complete blood count:</u> the CBC, red blood cell count, and reticulocyte count are normal.

Clinical Considerations: Newborns are asymptomatic with normal blood counts and have a normal life expectancy. Older children and adults may develop hematuria. Splenic infarction may occur with exertion and dehydration at high altitude. An increased risk of sudden death has been reported associated with severe hypoxia, extreme physical exertion, and dehydration. In adults there is an increased risk of chronic kidney disease and thromboembolism. Solubility testing (Sickledex) should not be used to confirm the diagnosis. Iron supplements should be avoided unless iron deficiency is documented.

Additional Information:

How to Communicate Newborn Screening Results

Gene Reviews

Medline Plus

Condition Information for Families-HRSA Newborn Screening Clearinghouse

Sickle Cell Disease Association of America

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.



ACT Sheet

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.lv/NBSResultsHRSA

Gene Reviews

• https://www.ncbi.nlm.nih.gov/books/NBK1377/

Medline Plus

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

Condition Information for Families-HRSA Newborn Screening Clearinghouse

• https://newbornscreening.hrsa.gov/conditions/sickle-cell-trait

Sickle Cell Disease Association of America

• https://www.sicklecelldisease.org/

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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