

# ACT Sheet

# Newborn Screening ACT Sheet [Hemoglobin FSA] Hemoglobin S/Beta Plus Thalassemia (Hb S/β<sup>+</sup> Thalassemia)

Differential Diagnosis: Sickle Cell Trait.

**Condition Description:** Hemoglobin S/Beta Plus ( $\beta^+$ ) Thalassemia 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 HbF decreases and Hb S predominates. The clinical course is highly variable, ranging from asymptomatic to life-threatening infections, acute chest syndrome, splenic sequestion, organ damage, and pain crises; the phenotype is determined by the amount of Hb A present.

# You Should Take the Following Actions:

- Inform the family of the newborn screening result.
- Ascertain clinical status (newborns are expected to be asymptomatic).
- Administer prophylactic penicillin.
- Evaluate the newborn (assess for splenomegaly and send CBC).
- Consult with sickle cell specialist immediately with in person follow up by 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/Beta Plus (β<sup>+</sup>) Thalassemia including the need for urgent evaluation if fever of ≥ 38.5°C (101°F) or splenic sequestration.
- 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>A). <u>Isoelectric focusing, high performance liquid chromatography (HPLC) or capillary zone</u> <u>electrophoresis</u> is used to confirm the newborn screening result. <u>Complete blood count</u>: the CBC, smear, mean corpuscular volume and reticulocyte count may be normal at birth but over the first few months of life demonstrate a worsening microcytic anemia, with an increasing reticulocyte count and sickle cells on smear. <u>Molecular genetic</u> testing is required to characterize the beta thalassemia variant.

**Clinical Considerations:** Newborns with Hemoglobin  $S/\beta^+$  Thalassemia are generally asymptomatic. Hemolytic anemia and vaso-occlusive complications can develop during infancy or in early childhood. Without appropriate treatment, complications include life-threatening infection, splenic sequestration, pneumonia, acute chest syndrome, pain episodes, aplastic crises, dactylitis, priapism, and osteonecrosis. Comprehensive care including family education, a modified immunization schedule, prompt treatment of infections and of vaso-occlusive events, screening for early signs of organ damage, and consideration of prophylactic penicillin and other disease-modifying interventions, reduces morbidity and mortality. Patients with HbS/ $\beta^+$  Thalassemia often have a clinical course that is similar to, but is less severe than those with sickle cell anemia (Hb S/S) and have a longer life expectancy. Their phenotype is determined by the thalassemia variant. Monitoring depends on the specific diagnosis and 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 be avoided unless iron deficiency is documented.

# Additional Information:

How to Communicate Newborn Screening Results Gene Reviews (Beta Thalassemia | Sickle Cell Disease) Medline Plus (Beta Thalassemia | Sickle Cell Disease) 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.

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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

- <u>https://bit.ly/NBSResultsHRSA</u>
- Gene Reviews
  - <u>https://www.ncbi.nlm.nih.gov/books/NBK1426/</u>
  - <u>https://www.ncbi.nlm.nih.gov/books/NBK1377/</u>

### Medline Plus

- https://medlineplus.gov/genetics/condition/beta-thalassemia/
- https://medlineplus.gov/genetics/condition/sickle-cell-disease/
- Condition Information for Families-HRSA Newborn Screening Clearinghouse
- https://newbornscreening.hrsa.gov/conditions/s-beta-thalassemia
- Sickle Cell Disease Association of America
- <u>https://www.sicklecelldisease.org/</u>

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

• <u>https://www.ncbi.nlm.nih.gov/gtr/</u>

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

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