

Disease Name

Alternate Name(s)

Acronym

Disease Classification

Hemoglobin S-β Thalassemia Disease

Beta Thalassemia Sickle Disease

Hb β/S

Hemoglobinopathy

Infants whose hemoglobin does not produce enough beta protein have beta thalassemia. It is found in people of Mediterranean descent, such as Italians and Greeks, and is also found in the Arabian Peninsula, Iran, Africa, Southeast Asia and southern China. There are three types of beta thalassemia that also range from mild to severe in their effect on the body.

Thalassemia Minor or Thalassemia Trait. In this condition, the lack of beta protein is not great enough to cause problems in the normal functioning of the hemoglobin. A person with this condition simply carries the genetic **trait** for thalassemia and will usually experience no health problems other than a possible mild anemia.

Thalassemia Intermedia. In this condition the lack of beta protein in the hemoglobin is great enough to cause a moderately severe anemia and significant health problems, including bone deformities and enlargement of the spleen. However, there is a wide range in the clinical severity of this condition, and the borderline between thalassemia intermedia and the most severe form, thalassemia major, can be confusing. The deciding factor seems to be the amount of blood transfusions required by the patient. The more dependent the patient is on blood transfusions, the more likely he or she is to be classified as thalassemia major. Generally speaking, patients with thalassemia intermedia need blood transfusions to improve their quality of life, but not in order to survive.

Thalassemia Major or Cooley's Anemia. This is the most severe form of beta thalassemia in which the complete lack of beta protein in the hemoglobin causes a life-threatening anemia that requires regular blood transfusions and extensive ongoing medical care. These extensive, lifelong blood transfusions lead to iron-overload which must be treated with chelation therapy to prevent early death from organ failure.

Symptoms

Most children with thalassemia major appear healthy at birth, but during the first year or two of life they become pale, listless and fussy, and have a poor appetite. They grow slowly and often develop jaundice (yellowing of the skin).

Natural history without treatment

The spleen, liver, and heart soon become greatly enlarged. Bones become thin and brittle; face bones become distorted, and children with thalassemia often look alike. Heart failure and infection are the leading causes of death among children with untreated thalassemia major. Children with thalassemia intermedia may develop some of the same complications, although in most cases, the course of the disease is mild for the first two decades of life.

Treatment**Red Blood Cell Transfusion**

Because there is no natural way for the body to eliminate iron, the iron in the transfused blood cells builds up in a condition known as "iron overload" and becomes toxic to tissues and organs, particularly the liver and heart. Iron overload typically results in the patient's early death from organ failure.

Chelation Therapy: To help remove excess iron, patients undergo the difficult and painful infusion of a drug, Desferal. Reduced mortality and morbidity with appropriate penicillin prophylaxis.

Inheritance

Autosomal recessive

General population incidence

1:250,000

Genetests Linkwww.geneclinics.org**Support Group**

Cooley's Anemia Foundation

<http://www.cooleysanemia.org/>

Sickle Cell Information Center

<http://www.scinfo.org/>

Sickle Cell Disease Association of America, Inc.

<http://www.sicklecelldisease.org>

Newborn Screening ACT Sheet [FSA] Hemoglobin S/Beta plus Thalassemia (HbS β^+ Disease)

Differential Diagnosis: Hemoglobin FSA pattern on newborn screen is highly suggestive of sickle beta plus thalassemia. The hemoglobins are listed in order (F>S>A) of the amount of hemoglobin present. This result is different from FAS which is consistent with sickle carrier (trait).

Condition Description: Individuals with sickle beta plus thalassemia, a form of sickle cell disease, are compound heterozygotes for the Hb S and beta-thalassemia mutations in the beta-globin genes.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the screening result.
- Perform a physical exam on the infant and assess for splenomegaly.
- Obtain a blood sample for confirmatory testing and a complete blood count (CBC) with reticulocyte count.
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Initiate penicillin (PenVK 125mg po bid) prophylaxis.
- Educate parents/caretakers regarding the risk of sepsis and advise that infant be immediately evaluated if a fever of $\geq 38.5^\circ\text{C}$ (101°F) is present.
- Contact a specialist in hemoglobin disorders for consultation on diagnostic evaluation and management.

Diagnostic Evaluation: CBC. Hemoglobin separation by electrophoresis, isoelectric focusing or high performance liquid chromatography (HPLC) shows FSA. DNA studies may be used to confirm genotype.

Clinical Considerations: Infants are usually normal at birth. Later potential clinical problems include mild to moderate hemolytic anemia, life-threatening infection, vaso-occlusive pain episodes, dactylitis, and chronic organ damage. Prompt treatment of infection and splenic sequestration is associated with decreased mortality in the first three years of life.

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](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It 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. Adherence to this guideline does not necessarily ensure a successful medical outcome. 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 guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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