



Newborn Screening ACT Sheet

[FSA]

Hemoglobin S/Beta⁺ Thalassemia (HbSβ⁺ 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⁺ 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.
- Repeat newborn screen if second screen has not yet been done.
- 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 $\geq 101.5^{\circ}$ F is present.
- Contact a specialist in hemoglobinopathies for consultation on diagnostic evaluation and management. (See attached list.)
- Report findings to newborn screening program.

Confirmation of Diagnosis: Hemoglobin separation by electrophoresis, isoelectric focusing, or HPLC showing FSA. Family or DNA studies may be used to confirm genotype.

Clinical Expectations: Infants are usually normal at birth. Later potential clinical problems include mild 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 <http://scinfo.org/hemoglb.htm#SICKLE%20HEMOGLOBINS> <http://www.scinfo.org/hemoglb.htm#BETA%20THALASSEMIAS>

Referral (local, state, regional and national): Comprehensive Sickle Cell Center Directory <http://www.rhofed.com/sickle/index.htm>

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

Sickle Cell Information Center <http://www.scinfo.org/clinics.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://sicklecelldisease.org/>



Newborn Screening FACT Sheet

Sickle Beta Plus Thalassemia (S β^+ -Thalassemia)

What is S β^+ -Thalassemia?

Sickle Beta Plus Thalassemia (S β^+ thalassemia) is a “mild” form of sickle cell disease. Your child’s red blood cells have only a small amount of the normal hemoglobin called hemoglobin A. They also have abnormal hemoglobin called hemoglobin S (sickle hemoglobin). The red blood cells have another problem called beta plus thalassemia. This causes red blood cells to be small and pale.

What Causes S β^+ -Thalassemia?

S β^+ thalassemia is an inherited condition. Hemoglobin S comes from one parent. Beta plus thalassemia comes from the other parent. Instead of appearing round (like donuts), your child’s red blood cells are somewhat small, pale and misshapen.

What Symptoms or Problems Occur with S β^+ -Thalassemia?

(Symptoms are something out of the ordinary that a parent notices.)

Periods of pain can happen with S β^+ thalassemia. The red blood cells are rigid and stiff. Sometimes they “clog up” the small blood vessels in the bones and other parts of the body. This can cause pain because not enough oxygen can get into the bones and other parts of the body. The pain usually happens in the back, stomach, arms, and legs. There may be swelling. The pain can last for a few hours or up to a week or more. The amount of pain varies. Sometimes children with S β^+ thalassemia have a slightly enlarged spleen (an organ located on the upper left side of the stomach area). A bigger spleen usually doesn’t cause any problems.

What is the Treatment for S β^+ -Thalassemia?

Medication – There are medicines to help ease the pain. Taking medication such as acetaminophen (Tylenol) or ibuprofen (Advil) usually helps the pain.

Fluids – Your child has more risk of getting infections, especially pneumonia. The abnormal red blood cells “clog up” the lungs. This increases the risk of infection there. Your child will need to drink plenty of fluids to keep blood vessels open.

Things to Remember

Children with S β^+ -thalassemia can have normal lives and life spans. Don’t think of your child as “sick.” You should treat him or her normally. Your child will need to see the doctor for regular checkups and vaccinations. He or she will also need to make several visits a year to see a hematologist (a doctor who is a blood specialist).