FS (HbSS Disease or HbS/Beta Zero Thalassemia)

Sickle Cell Anemia

Differential Diagnosis: Homozygous sickle cell disease (Hb SS), sickle beta-zero thalassemia, or sickle hereditary persistence of fetal hemoglobin (S-HPFH).

Condition Description: A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin S in the absence of hemoglobin A. The hemoglobins are listed in order of the amount of hemoglobin present (F>S). This result is different from FAS, which is consistent with sickle carrier.

You Should Take the Following Actions

- Contact the family to inform them of the screening result.
- Refer to a pediatric hematologist. (See attached list.)
- Evaluate infant and assess for splenomegaly.
- Repeat newborn screen if second screen has not yet been done.
- Initiate daily penicillin VK (125mg po bid) prophylaxis and other treatment as recommended by the consultant.
- Educate parents/caregivers regarding the risk of sepsis, the need for urgent evaluation if fever of ≥ 101.5° F, or signs and symptoms of splenic sequestration.
- Report findings to newborn screening program.

Diagnostic Evaluation: The Newborn screening program does confirmatory DNA studies.

Clinical Considerations: Newborn infants are usually well. Hemolytic anemia and vaso-occlusive complications develop during infancy or early childhood. Complications include life-threatening infection, splenic sequestration, pneumonia, acute chest syndrome, pain episodes, aplastic crisis, dactylitis, priapism, and stroke. Comprehensive care, including family education, immunizations, prophylactic penicillin, and prompt treatment of acute illness, reduces morbidity and mortality. S-HPFH is typically benign.

Additional Information:
Sickle Cell Information Center:
https://scinfo.org/sickle-cell-disease

Sickle Cell Disease Association
http://www.sicklecelldisease.org/

National Heart Lung and Blood Institute
http://www.nhlbi.nih.gov/health/health-topics/topics/sca/

Cook Children’s Hospital
http://kidshealth.org/PageManager.jsp?dn=CookChildrensHospital&lic=403&cat_id=141&article_set=22658&ps=104

Utah Department of Health
http://health.utah.gov/newbornscreening/Disorders/HB/Hb_SS_Disease_SS/FactSheet_Provider_HbSS_En.pdf

Disclaimer: This information is adapted from American College of Medical Genetics website ACT sheets. 05/2015
Sickle Cell Anemia (Hb-SS)

What is Sickle Cell Anemia?
Sickle Cell Anemia (Hb-SS-Disease) is the most common Sickle Cell Disease. It clogs blood vessels and causes severe pain. It can also cause problems such as organ and tissue damage. Other common Sickle Cell Diseases are Hb-SC-Disease and sickle thalassemias.

What Causes Sickle Cell Anemia?
Sickle Cell Anemia is an inherited blood disease. It is very serious for infants and young children. A person with this life-long disease has abnormal red blood cells. Changes in these cells cause them to be shaped like a “sickle.” These red blood cells are very weak and sometimes rigid. They can get trapped in the blood vessels and harm blood flow. This can cause sudden pain anywhere in the body. It can also damage body tissues and organs over time.

What Problems Occur with Sickle Cell Anemia?
Many young children with sickle cell anemia are rarely sick. Sometimes serious problems happen that can cause death. These problems are:
- septicemia (infection of the blood)
- acute splenic sequestration (sudden enlargement of the spleen and rapid drop in the blood count)
- Other serious problems can include:
  - pneumonia
  - organ damage
  - painful erections in men
  - swelling of hands and feet
  - problems during pregnancy
  - chest pains and trouble breathing
  - blood in urine
  - fever
  - stroke
  - leg ulcers
  - infections
  - jaundice (yellow skin)
  - gallstones
  - anemia
  - pain
  - kidney failure

What is the Treatment for Sickle Cell Anemia?

Medication – Infants and young children with Sickle Cell Anemia sometimes get infections of the blood. Penicillin can prevent death from this. When given twice a day, penicillin can kill bacteria before they grow in the blood and cause very dangerous infections.

Fluids – It is important to drink plenty of fluids to keep blood vessels open.

Things to Remember
Children with sickle cell anemia can have periods of pain. This happens more in older children, but sometimes happens in babies. Usually the pain seems to be in the bone. Sometimes it is in the stomach. Such pain is not dangerous. It usually goes away after several hours or days.

Medications such as acetaminophen (such as Tylenol) or ibuprofen (such as Advil) often help the pain. Sometimes children won’t use the part of the body that hurts, even when it feels better. Don’t force your child to stand or walk. Children will be active again when they feel like it. Rest and drinking plenty of liquids can help the pain. A heating pad can also help. If a child’s pain isn’t better after taking medicine at home, he or she may need to be treated with stronger medicine in the hospital.
Sickle Beta Zero Thalassemia (Sβ₀-Thalassemia)

What is Sβ₀-Thalassemia?

Hemoglobin in the red blood cells carries oxygen to all parts of the body. With Sickle Beta Zero Thalassemia (Sβ₀-Thalassemia), the red blood cells have abnormal hemoglobin. This is called hemoglobin S (sickle hemoglobin). Also, there is a problem called thalassemia. This causes red blood cells to be small and pale.

What Causes Sβ₀-Thalassemia?

Sβ₀-Thalassemia is an inherited disease. Hemoglobin S is inherited from one parent. Thalassemia is inherited from the other parent. In Sβ₀-Thalassemia the red blood cells are not round. They are misshapen. Some may have a sickle (or banana) shape.

What Symptoms or Problems Occur with Sβ₀-Thalassemia?

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

The symptoms of Sβ₀-Thalassemia depend on the type and seriousness of the disease. Problems happen when different parts of the body don’t get enough oxygen. This occurs when there is low hemoglobin and not enough red blood cells in the blood (anemia).

Problems include:

- feeling tired and weak
- slightly enlarged spleen (an organ located on the upper left side of the stomach area)
- mild anemia
- periods of pain
- increased risk for infections

What is the Treatment for Sβ₀-Thalassemia?

Medications – Your child has more risk of getting infections. These can be infection in the brain, pneumonia, blood poisoning, and bone infection. Your child’s doctor will probably give him or her penicillin to help prevent infections. It is important that your child take the penicillin as directed. Any fever of 101.5°F is serious. Take your child to the doctor or clinic IMMEDIATELY if this happens.

Periods of pain can happen with Sβ₀-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. Pain can last for a few hours or up to a week or more. The amount of pain varies. Medicines help ease the pain.

Sometimes children with Sβ₀-Thalassemia have a slightly enlarged spleen. A bigger spleen usually doesn’t cause any problems.

Things to Remember

Children with Sβ₀-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).