Sickle Cell Disease and Sickle Cell Trait

Sickle cell disease is an inherited blood disease most common in African Americans. Over time, serious complications can occur. Treatment focuses on preventing complications and keeping your child as healthy as possible. Sickle cell trait is different from sickle cell disease! People with this trait rarely have any health problems but may benefit from genetic counseling.

What is sickle cell disease?

Children with sickle cell disease have abnormal red blood cells with a "sickled" or crescent-moon shape. They have *anemia,* which means they have low amounts of hemoglobin, or red blood cells, in their blood.

These problems occur when your child inherits abnormal types of hemoglobin, which carries oxygen in the blood. The most common form of sickle cell disease is sickle cell anemia. Other abnormal hemoglobins can be inherited along with the "sickled" hemoglobin, causing other forms of sickle cell disease.

The sickle cells block small blood vessels, causing damage when blood cannot get to parts of the body. This leads to attacks of pain and other symptoms and complications. Sickle cell disease is a lifelong problem. It is more severe in some children than others.

What is sickle cell trait?

Sickle cell *trait* is much more common than sickle cell disease and generally does not cause any health problems. It occurs in 8% to 10% of African Americans. Children who are born with sickle cell trait live a normal lifespan and do not have sickle cell disease. They should receive regular medical attention, just like other children. Genetic counseling will help you to understand your family's risk of passing on sickle cell disease and sickle cell trait to future generations.

Complications are rare. People with sickle cell trait should make sure they stay well hydrated (drink lots of fluids) and avoid becoming overheated. Sickle cell trait can result in decreased ability of the kidneys to concentrate urine, which may lead to blood in the urine. There may be an increased risk of sudden death during exercise. None of these problems has been well studied. However, as mentioned, children with this trait rarely have such problems.

What causes sickle cell disease and sickle cell trait?

- Sickle cell disease is caused by abnormal (mutated) genes. To have sickle cell anemia, your child must inherit two sickle cell genes—one from each parent. If your child just inherits one sickle cell gene, he or she will have sickle cell trait (which rarely causes problems).
- If both parents have sickle cell trait, the risk of sickle cell anemia is 25% for each child. If one parent has sickle cell trait and the other has no abnormal hemoglobins, the chance of sickle cell trait is 50% in each child.
- Sickle cell disease is most common in African Americans, affecting about 1 in 625 newborns.

What kinds of health problems are caused by sickle cell disease?

- *Pain* is the most noticeable symptom:
  - Every child is different—some have more frequent and severe episodes of pain than others.
  - He or she may have occasional attacks of severe pain. These attacks, called *sickle cell crises,* may require hospital treatment. There is no way to predict how often your child will experience sickle cell crisis.
  - Pain usually occurs in the arms and legs in younger children, and in the head, chest, abdomen, or back in older children.
- *Hand-foot syndrome (*also called *acute sickle dactylitis)* may be the first sign of sickle cell disease. The hands and feet become painful and swollen. The blood vessels become blocked, causing damage to the bones.
- *Acute chest syndrome* is an occasional problem. Damage to the lungs occurs when blood vessels become blocked by sickle cells, causing attacks of chest pain. Acute chest syndrome often causes problems getting enough oxygen into the blood. In children, it often is seen along with infection of the lungs (pneumonia). This can be a very serious complication.
- *Anemia.* Low hemoglobin levels cause your child to become easily tired.
- *Infections.* Patients with sickle cell anemia are at increased risk of serious infections with bacteria.
- *Splenic sequestration* is an uncommon but serious complication. The spleen is an organ that filters blood, removes

damaged red blood cells and certain germs, and contains cells important for immune system function. It is located on the left side of the upper abdomen. It is usually under the ribs, but when enlarged it can be felt in the abdomen.

  - The spleen can fill up with blood and become quickly enlarged. This can cause your child to become anemic quickly and is a very serious situation. Your child will need a blood transfusion as soon as possible.

- *Strokes* occur in up to 10% of children with sickle cell disease. This happens when sickle cells interfere with blood flow to parts of the brain, causing damage to those areas.
- *Priapism.* This is an abnormal erection of the penis that can be painful and last a long time.
- *Aplastic crisis.* The body isn't making enough red blood cells. This causes your child to become very anemic. Aplastic crisis is usually caused by infection with a specific virus (Parvovirus).
- Over time, sickle cell disease can cause damage to many parts of the body, including the heart, kidneys, and eyes. It also may result in poor growth.

How is sickle cell disease diagnosed?

A simple blood test called *hemoglobin electrophoresis* determines whether your child has sickle cell disease or trait. Most states test all newborns at birth.

How is sickle cell disease treated?

Treatment focuses on preventing complications and treating your child as soon as possible when problems occur.

- *Infections.* Your child will receive vaccinations to help prevent infections. At least for the first 5 years, your child will take an antibiotic, such as penicillin.
  - When infections occur, they will be treated aggressively (quickly). This is because sickle cell patients have reduced ability to fight infections.
- *Pain crisis.* Pain crises are usually treated with strong pain medications, such as narcotics. Lots of fluids are needed to help prevent sickle cells from plugging blood vessels. Patients with severe pain crises often need hospital treatment.
- *Acute chest syndrome.* This is often treated with oxygen and blood transfusion.
- *Strokes.* Blood transfusion is needed to lower the number of sickle cells in the blood.
- *Priapism* (prolonged erections). If the problem doesn't clear up with pain medications and fluids, blood transfusion is needed.
- *Splenic sequestration.* This condition requires prompt blood transfusion.
- *Drug therapy.* Treatment with a drug called hydroxyurea, which decreases the amount of sickle hemoglobin in the blood, can be helpful for some patients.
- *Bone marrow transplantation* may cure the disease for some patients. However, there are many problems involved in performing this procedure. It usually requires a family member with "matched" bone marrow.

When should I call your office?

Call our office if any of the following occurs:

- Pain that seems like your child's "usual" pain but does not improve with medications and extra fluids.
- Fever of 101°F (38.5°C) or higher.
- Chest pain.
- Enlargement of the spleen. (Parents are taught how to feel the abdomen for an enlarged spleen.)

Sickle cell disease is a serious and complicated condition. Call your doctor's office if you have questions about this disease or about your child's treatment.

Please type your custom instructions and/or office contact information here.