Qure
Center for Complete Care
Blood Diseases, Cancer, Stem Cell Transplantation

Sickle cell disease

What is hemoglobin S?

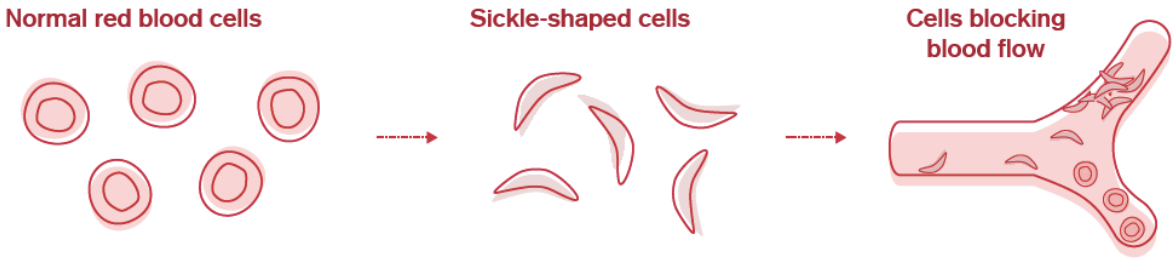
Hemoglobin is the part of red blood cells that carries oxygen to all parts of the body. The usual type of hemoglobin is called hemoglobin A. Genes that we inherit from each of our parents determine what type of hemoglobin we have. Hemoglobin S is a hemoglobin protein that is different from the common Hemoglobin A, and shows up separately on special testing.

There are different forms of sickle cell disease depending on the hemoglobin genes inherited:

- Sickle cell anemia, which includes sickle cell disease type SS and type sickle beta zero ($S\beta^0$) thalassemia
- Sickle cell disease type SC
- Sickle cell disease type sickle beta plus ($S\beta+$) thalassemia
- Sickle cell disease type SD, SE, and other sickle cell disease variants

What is sickle cell disease?

Sickle cell disease is an inherited blood disease that causes problems with red blood cells. Red blood cells circulate in the blood and contain hemoglobin, which carries oxygen to all parts of the body, and normal red blood cells are shaped like a disc. In sickle cell disease, the abnormal hemoglobin (hemoglobin S) sticks together in low oxygen situations deep in the body, causing the red cells to assume a curved shape like a sickle (a farming curved cutting tool, *daranti*). Sickle cells are less flexible and block the flow of in blood vessels causing problems.



What are the signs and symptoms of sickle cell?

Symptoms of sickle cell disease can vary, and a few patients may have only mild anemia and intermittent problems. However, many will have anemia sufficient to cause tiredness. In addition when sickle cells block blood vessels this can lead to:

- Pain crisis: pain in the chest, stomach, or bones
- Acute chest syndrome: shortness of breath which can be life threatening
- Stroke: weakness of one half of the body which can be permanent
- Kidney damage: which can eventually lead to kidney failure
- Retinal damage: which can eventually lead to vision loss
- Destruction of the spleen: which makes patients more prone to certain kinds of infection (e.g. pneumococcal)

Infection, fever and stress can cause destruction of more red blood cells than usual, causing the hemoglobin level to drop further and with jaundice of the skin and whites of the eyes. Since red blood cells are produced in the bone marrow, sometimes a viral infection (e.g. parvovirus) can cause the bone marrow to stop cell production temporarily causing a drop in hemoglobin with lack of reticulocytes in the blood. This is called an "aplastic crisis" and may last for several days before red blood cell production will resume, during which anemia may be severe enough to require a red blood cell transfusion.

How is sickle cell diagnosed?

The history and physical findings, and anemia with sickle cells in the peripheral blood smear suggest the diagnosis. Often the first indication is an acute crisis e.g. splenic sequestration crisis where blood is trapped in the spleen with a sudden anemia. Confirmatory tests are:

- Sickle cell test: in which a patient's red blood cells are placed in a special solution and undergo sickling (SickleDex). Unfortunately this is a screening test and less reliable.
- Hemoglobin electrophoresis test: in which the patient's red cells are subjected to an electric field and hemoglobin S shows up differently from hemoglobin A. This test is confirmatory.

How is sickle cell disease treated?

Treatment consists of daily folic acid (vitamin) supplementation. With destruction of the spleen, there is an increased risk for infection, and for this reason, penicillin is given twice a day for the rest of your child's life. It is very important that your child receive normal childhood immunizations and a few special immunizations (pneumococcal and meningococcal immunizations) to decrease the risk of infection.

There is a specific medication called hydroxyurea ("hi-drox-ee-ure-EE-a") that can help adults and children have fewer pain crises and live longer. Hydroxyurea makes the red blood cells bigger and helps them stay rounder and more flexible, by increasing a special kind of hemoglobin called hemoglobin F. Experts recommend hydroxyurea for patients with sickle cell disease type SS or type sickle beta zero ($S\beta^0$) thalassemia. Hydroxyurea has been used to treat sickle cell disease since the 1980s, and in 2017 the FDA approved it to treat children. Hydroxyurea treatment leads to fewer pain crises, acute chest syndrome, blood transfusions or hospital stays, and can prevent or slow down organ damage. Many people with sickle cell disease have taken hydroxyurea safely for over 20 years, and there's no evidence it causes cancer in people with sickle cell disease.

Some people who take hydroxyurea may experience mild side effects:

- Thinning hair or mild hair loss
- Fingernail beds that turn darker
- Nausea (feeling sick to your stomach)

Very rarely, hydroxyurea can cause more serious side effects. But most people with sickle cell disease who take hydroxyurea don't have any serious side effects. If you have any new symptoms after you start taking hydroxyurea, tell your doctor.

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Experts are still learning about how hydroxyurea affects the ability to have a healthy baby, so women who are planning to get pregnant, should talk to the doctor. Hydroxyurea can also lower the sperm count, but can reduce painful erections that are a complication of sickle cell disease.

Most people take hydroxyurea pills once a day. You'll need to get your blood cell counts checked regularly, usually at least once a month, when you take hydroxyurea. Typically the blood counts will show an increase in hemoglobin level (Hb) and red cell size, a decrease in white cell (neutrophil) counts, and sometimes a decrease in platelet counts. If the cell counts drop too low your dose may need to be adjusted.

It can be hard to remember to take hydroxyurea every day. But you can mark your calendar every day after you take it, use a pill box with a section for each day of the week, or set an alarm on your cell phone that goes off every day.

When should I call my doctor?

Call the clinic right away if:

- Your child has pain or weakness
- you notice that your child's skin is more pale than usual or he or she seems more tired.
- your child's skin and whites of the eyes are more yellow than usual
- your child has a fever of >100.5° F