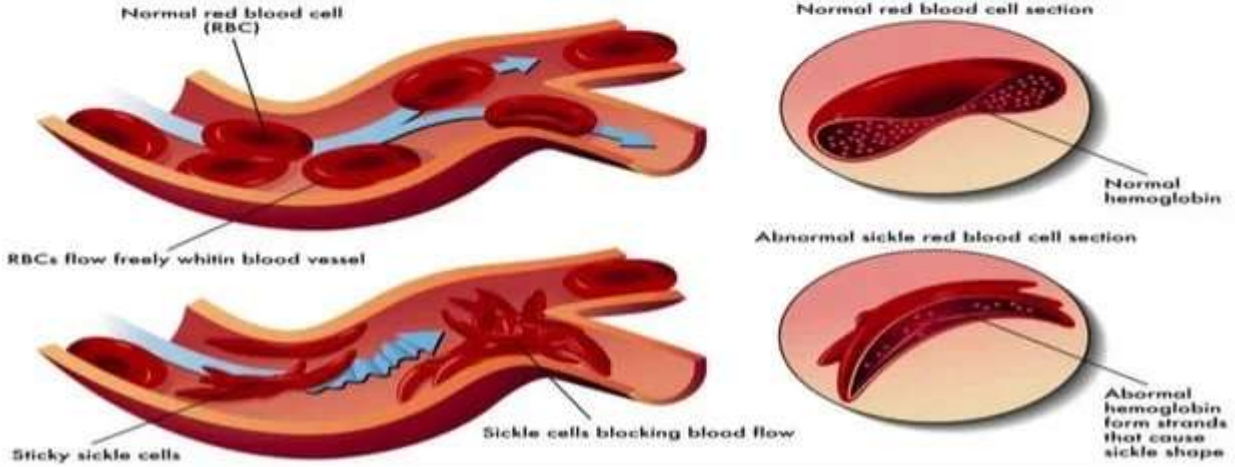
Sickle Cell Anemia

Sickle Cell Anemia

- Is an inherited form of anemia — a condition in which there aren't enough healthy red blood cells to carry adequate oxygen throughout your body.
- Normally, your red blood cells are flexible and round, moving easily through your blood vessels.

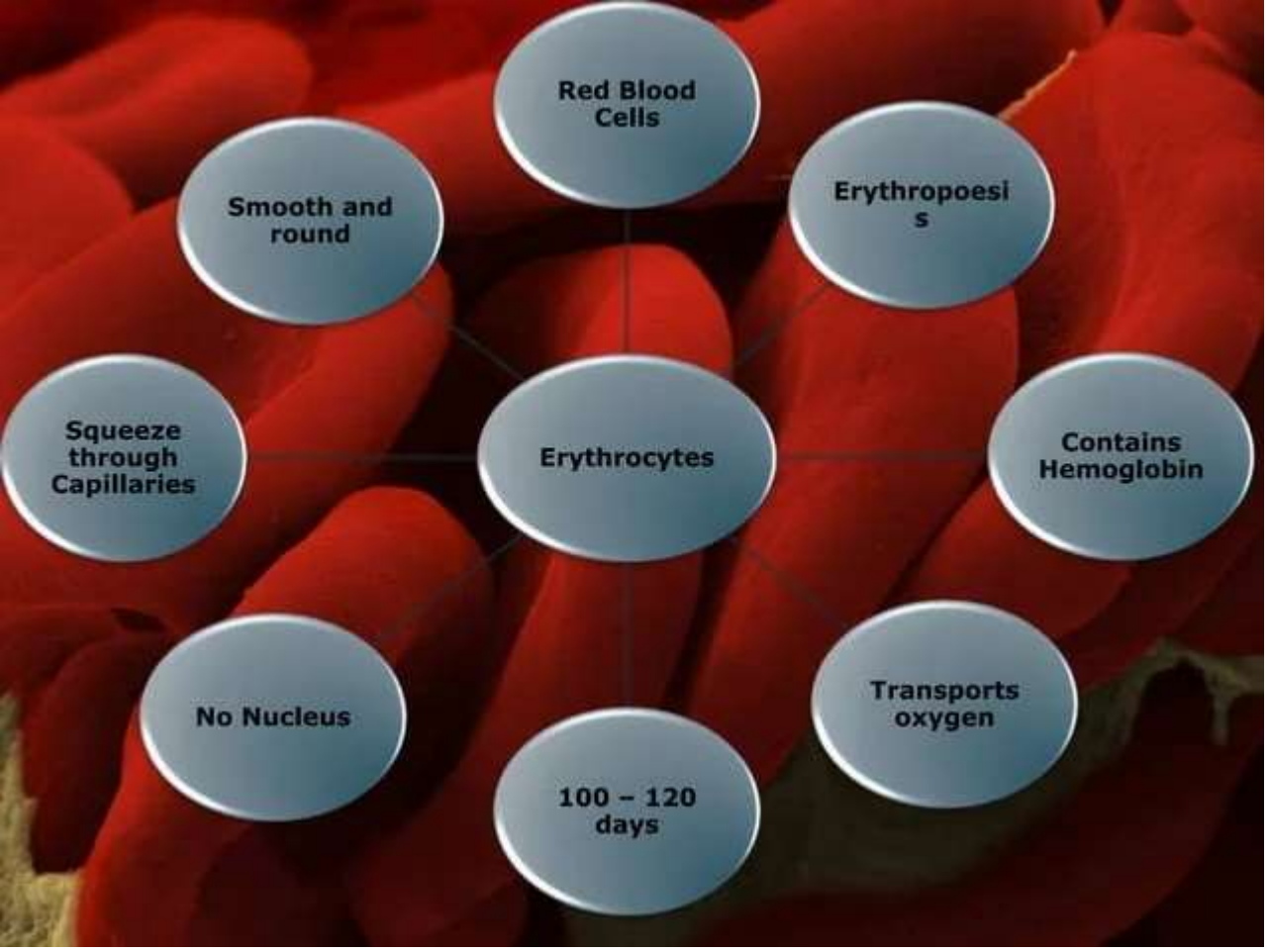
Sickle Cell Anemia

- In sickle cell anemia, the red blood cells become rigid and sticky and are shaped like sickles or crescent moons.
- These irregularly shaped cells can get stuck in small blood vessels, which can slow or block blood flow and oxygen to parts of the body.



Definition

- Sickle Cell disease: is a genetic disorder that affects erythrocytes (RBC) causing them to become sickle or crescent shaped.
- The effects of this condition due to an abnormality of the hemoglobin molecules found in erythrocytes.

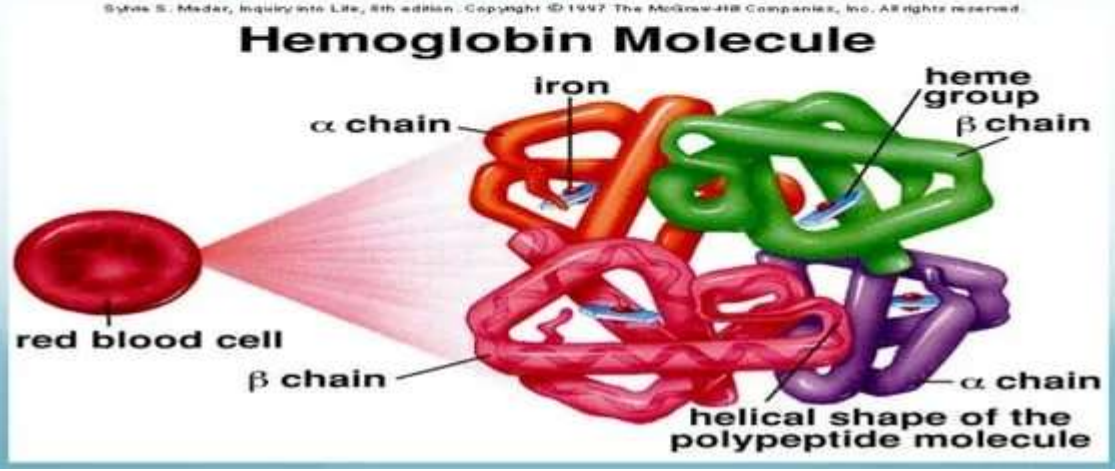

Red Blood Cells
Smooth and round
Erythropoesis
Squeeze through Capillaries
Erythrocytes
Contains Hemoglobin
No Nucleus
Transports oxygen
100 – 120 days

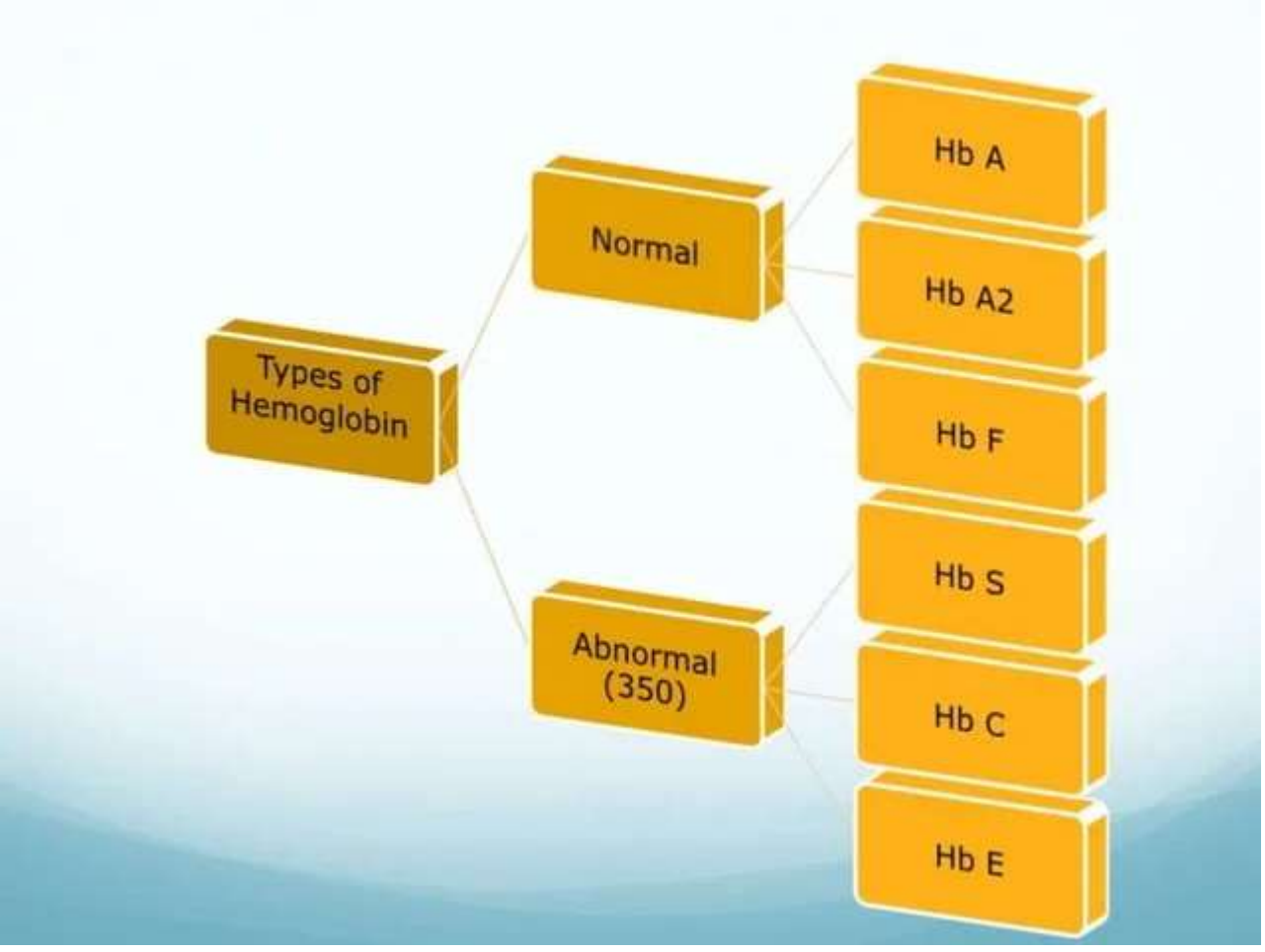
Hemoglobin

- The oxygen-carrying pigment and predominant protein in the red blood cells.
- Hemoglobin forms an unstable, reversible bond with oxygen.
- Oxyhemoglobin: Oxygenated (bright red).
- Deoxyhemoglobin: Reduced (purple-blue).

Hemoglobin

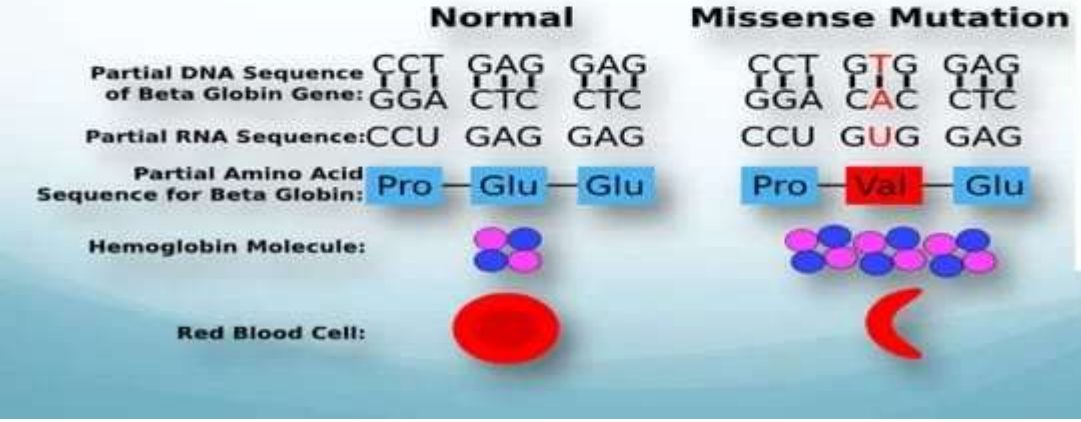
- Each hemoglobin molecule is made up of four heme groups surrounding a globin group.
- Heme contains iron and gives a red color to the molecule.
- Globin consists of two linked pairs of polypeptide chains.

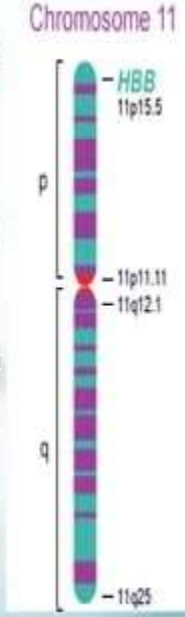



Types of Hemoglobin
Normal
Abnormal (350)
Hb A
Hb A2
Hb F
Hb S
Hb C
Hb E

Genetics of SCD

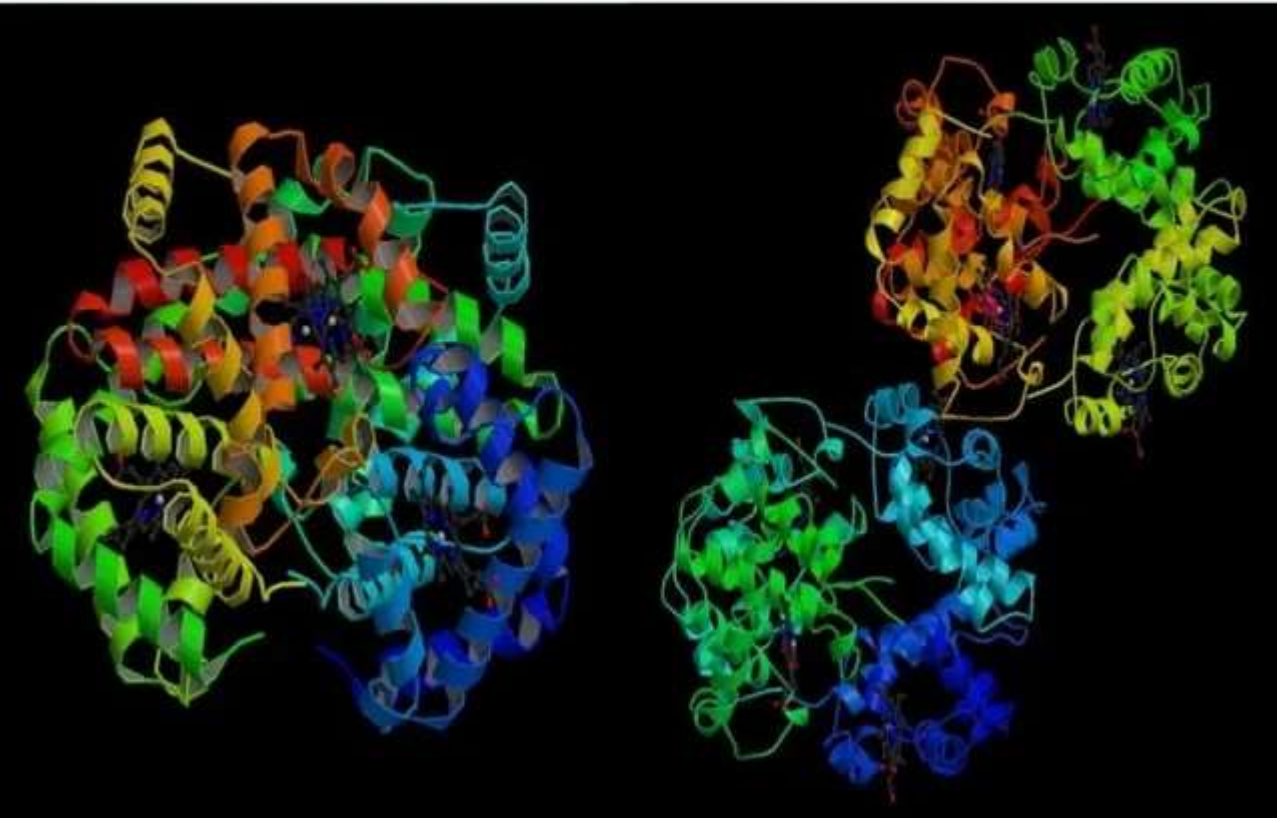
- The change in cell structure arises from a change in the structure of hemoglobin.
- A single change in an amino acid causes hemoglobin to aggregate.





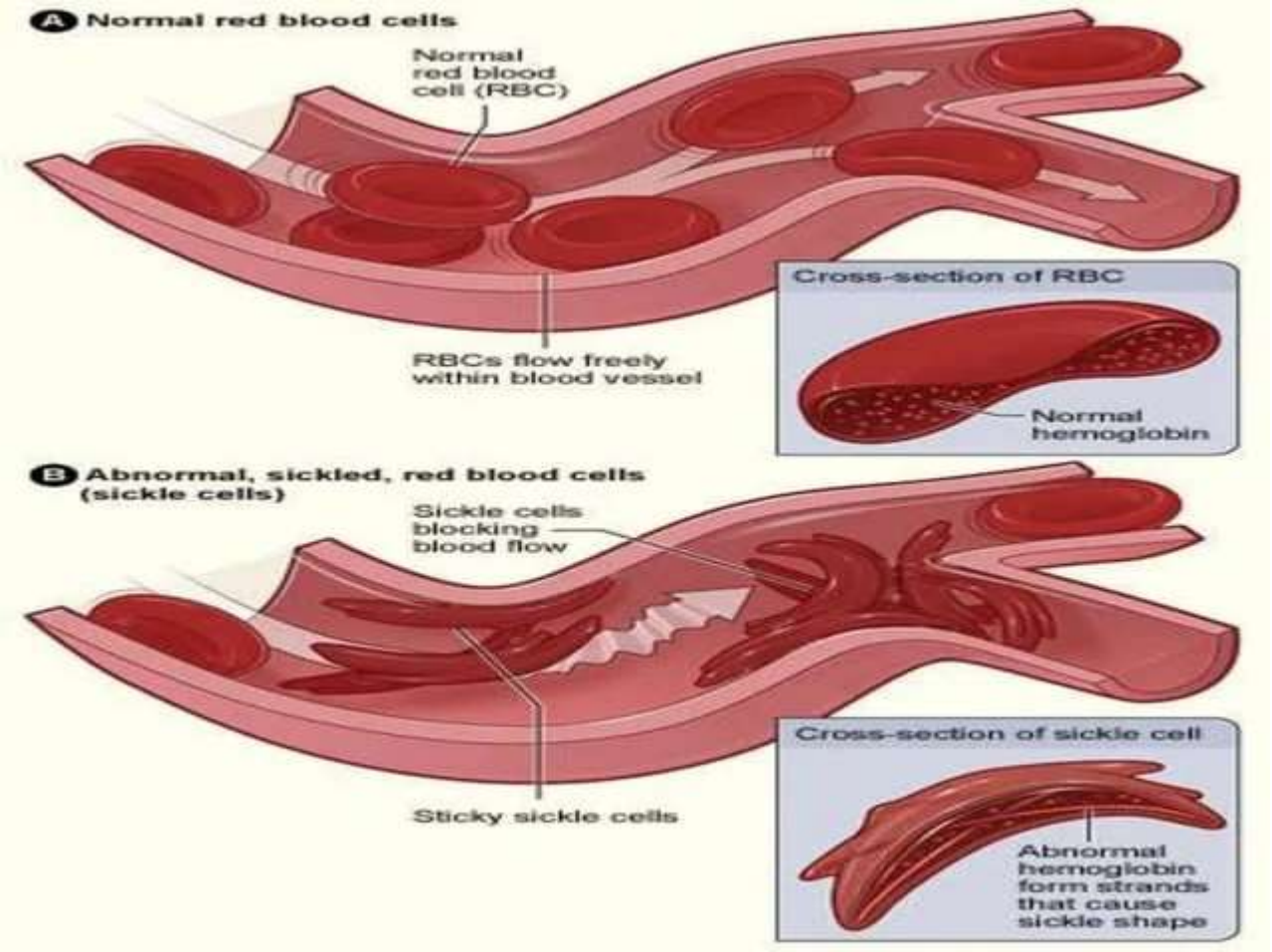
Hemoglobin A

Hemoglobin S



Mechanism

- When sickle hemoglobin (HbS) gives up its oxygen to the tissues, HbS sticks together
  - Forms long rods form inside RBC
  - RBC become rigid, inflexible, and sickle-shaped
  - Unable to squeeze through small blood vessels, instead blocks small blood vessels
  - Less oxygen to tissues of body
- RBCs containing HbS have a shorter lifespan
  - Normally 20 days
  - Chronic state of anemia


A Normal red blood cells
Normal red blood cell (RBC)
RBCs flow freely within blood vessel
Cross-section of RBC
Normal hemoglobin
B Abnormal, sickled, red blood cells (sickle cells)
Sickle cells blocking blood flow
Sticky sickle cells
Cross-section of sickle cell
Abnormal hemoglobin form strands that cause sickle shape

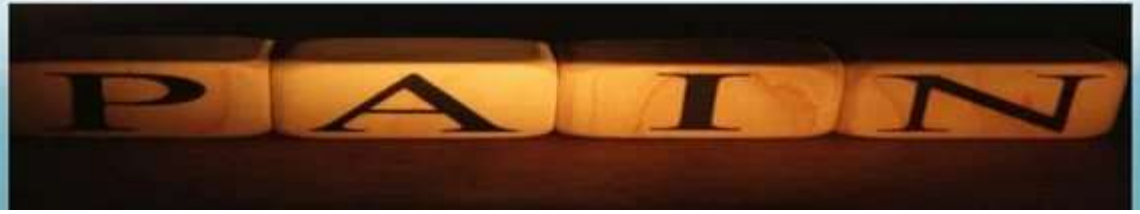
Symptoms

They vary from person to person and change over time, include:

- **Anemia.** Sickle cells break apart easily and die, leaving you without enough red blood cells. Red blood cells usually live for about 120 days before they need to be replaced. But sickle cells usually die in 10 to 20 days, leaving a shortage of red blood cells (anemia). Without enough red blood cells, your body can't get the oxygen it needs to feel energized, causing fatigue.

Symptoms

- **Episodes of pain.** Periodic episodes of pain, called crises, are a major symptom of sickle cell anemia.
- Pain develops when sickle-shaped red blood cells block blood flow through tiny blood vessels to your chest, abdomen and joints. Pain can also occur in your bones.
- The pain varies in intensity and can last for a few hours to a few weeks. Some people have only a few pain episodes. Others have a dozen or more crises a year. If a crisis is severe enough, you might need to be hospitalized. Some adolescents and adults with sickle cell anemia also have chronic pain, which can result from bone and joint damage, ulcers and other causes.



Symptoms

- **Painful swelling of hands and feet.** The swelling is caused by sickle-shaped red blood cells blocking blood flow to the hands and feet.
- **Frequent infections.** Sickle cells can damage an organ that fights infection (spleen), leaving you more vulnerable to infections. Doctors commonly give infants and children with sickle cell anemia vaccinations and antibiotics to prevent potentially life-threatening infections, such as pneumonia.

Symptoms

- **Delayed growth.** Red blood cells provide your body with the oxygen and nutrients you need for growth. A shortage of healthy red blood cells can slow growth in infants and children and delay puberty in teenagers.
- **Vision problems.** Tiny blood vessels that supply your eyes may become plugged with sickle cells. This can damage the retina — the portion of the eye that processes visual images, leading to vision problems.

Complications

Sickle cell anemia can lead to a host of complications, including:

- **Stroke.** A stroke can occur if sickle cells block blood flow to an area of your brain. Signs of stroke include seizures, weakness or numbness of your arms and legs, sudden speech difficulties, and loss of consciousness. If your baby or child has any of these signs and symptoms, seek medical treatment immediately. A stroke can be fatal.

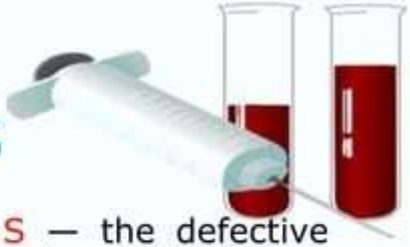
- **Acute chest syndrome.** This life-threatening complication causes chest pain, fever and difficulty breathing. Acute chest syndrome can be caused by a lung infection or by sickle cells blocking blood vessels in your lungs. It might require emergency medical treatment with antibiotics and other treatments.

- **Pulmonary hypertension.** People with sickle cell anemia can develop high blood pressure in their lungs (pulmonary hypertension). This complication usually affects adults rather than children. Shortness of breath and fatigue are common symptoms of this condition, which can be fatal.

Complications

- **Organ damage.** Sickle cells that block blood flow through blood vessels immediately deprive the affected organ of blood and oxygen. In sickle cell anemia, blood is also chronically low on oxygen. Chronic deprivation of oxygen-rich blood can damage nerves and organs in your body, including your kidneys, liver and spleen. Organ damage can be fatal.
- **Blindness.** Sickle cells can block tiny blood vessels that supply your eyes. Over time, this can damage the portion of the eye that processes visual images (retina) and lead to blindness.
- **Leg ulcers.** Sickle cell anemia can cause open sores, called ulcers, on your legs.
- **Gallstones.** The breakdown of red blood cells produces a substance called bilirubin. A high level of bilirubin in your body can lead to gallstones.

Diagnosis



- A blood test can check for hemoglobin S — the defective form of hemoglobin that underlies sickle cell anemia. In the United States, this blood test is part of routine newborn screening done at the hospital. But older children and adults can be tested, too.
- In adults, a blood sample is drawn from a vein in the arm. In young children and babies, the blood sample is usually collected from a finger or heel.
- If the screening test is negative, there is no sickle cell gene present.
- If the screening test is positive, further tests will be done to determine whether one or two sickle cell genes are present.
- Check for a low red blood cell count (anemia) will be done.

Treatment

- Treatment is usually aimed at avoiding crises, relieving symptoms and preventing complications. Babies and children age 2 and younger with sickle cell anemia should make frequent visits to a doctor.
- Children older than 2 and adults with sickle cell anemia should see a doctor at least once a year, according to the Centers for Disease Control and Prevention.
- Treatments might include medications to reduce pain and prevent complications, and blood transfusions, as well as a bone marrow transplant.

Explore treatment options.

Compare benefits and risks.

Prepare for medical appointments.

Medications

- **Antibiotics.** Children with sickle cell anemia may begin taking the antibiotic penicillin when they're about 2 months old and continue taking it until they're at least 5 years old.
- Doing so helps prevent infections, such as pneumonia, which can be life-threatening to an infant or child with sickle cell anemia.
- As an adult, if you've had your spleen removed or had pneumonia, you might need to take penicillin throughout your life.

Medications

- **Pain-relieving medications.** To relieve pain during a sickle cell crisis.
- **Hydroxyurea (Droxia, Hydrea).** When taken daily, hydroxyurea reduces the frequency of painful crises and might reduce the need for blood transfusions and hospitalizations.
- Hydroxyurea seems to work by stimulating