Sickle Cell Anemia

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SICKLE CELL ANEMIA

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.

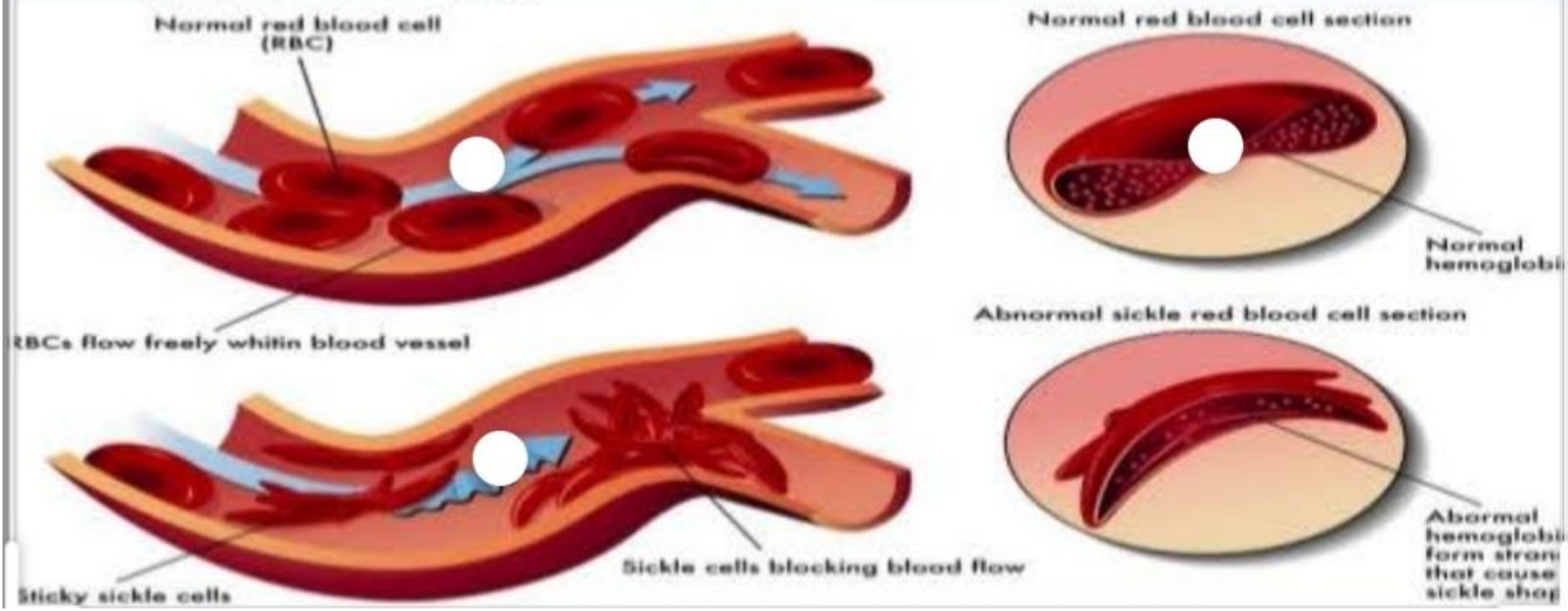
Sickle Cell Anemia

Sickle Cell anemia is an inherited red blood cell disorder. Normal red blood cells are round like doughnuts, and they move through small blood tubes in the body to deliver oxygen.

Sickle red blood cells become hard, sticky and shaped like sickles used to cut wheat. When these hard and pointed red cells go through the small blood tube, they clog the flow and break apart. This can cause pain, damage and a low blood count, or anemia.

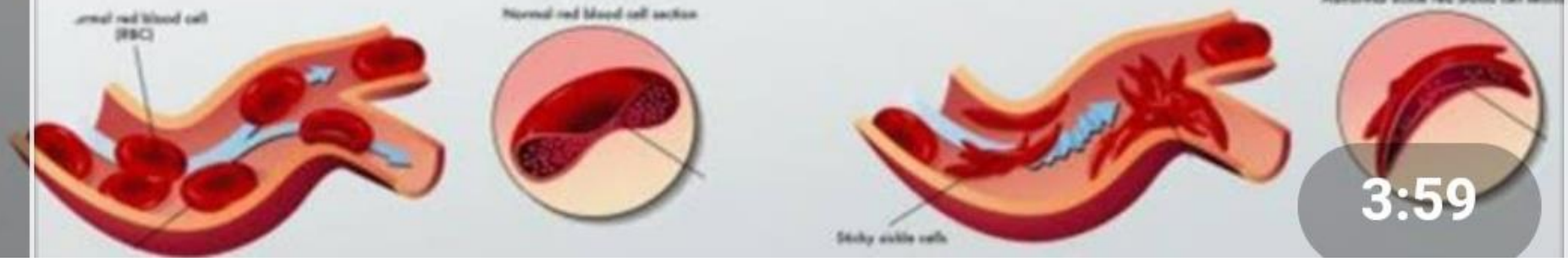
Sickle Cell Anemia

- 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.



Sickle cell anemia

normally oval or donut shaped blood cell instead resembles a sickle or crescent shape



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Epidemiology of sickle cell disease

- Incidence:
  - ✓ 6 to 9 million infants are born each year with sickle cell disease in Africa
  - ✓ Sickle cell disease occurs 1 in 600 African-American infants
  - ✓ 1 in 12 people in Africa have sickle cell trait

Prevalence (race)

- ✓ Highest in African, African-American, Mediterranean (Italian, Sicilian, Greek), Middle Eastern, East Indian, Caribbean, and Central or South American descent

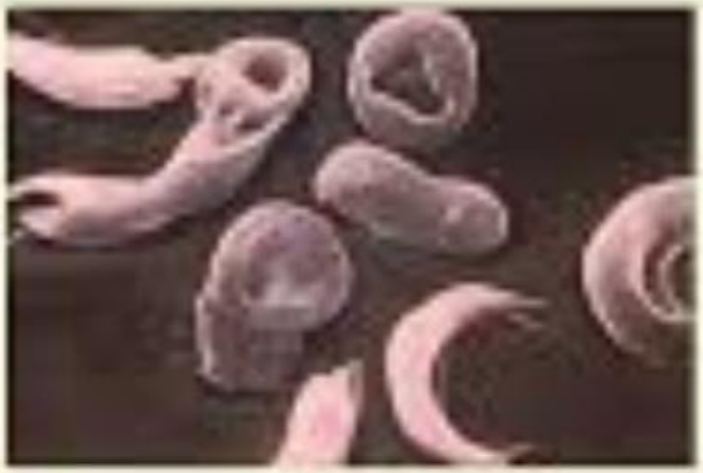
Age

- ✓ Affected patients characteristically are asymptomatic until approximately 4 to 6 months of age
- ✓ Median age at death is approximately 42 years for men and 48 years for women

Sex:

- ✓ Ratio of male to female is 1:1

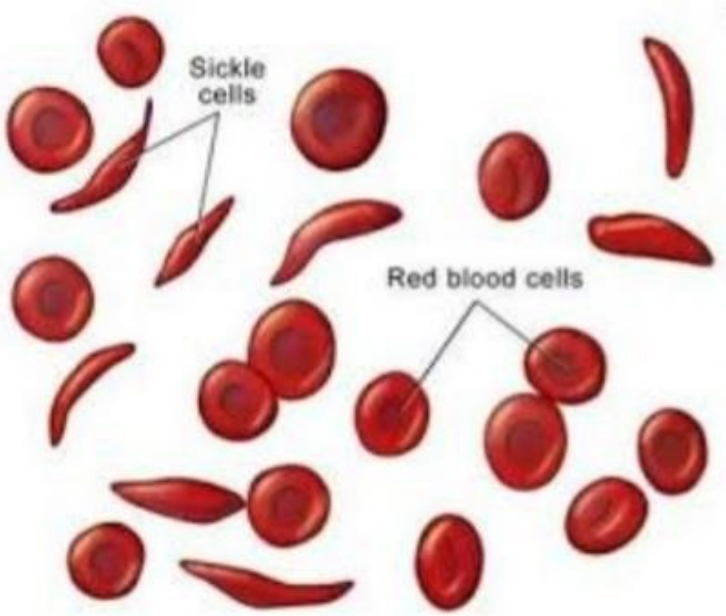
The cause of Sickle cell anemia



What can this cause?

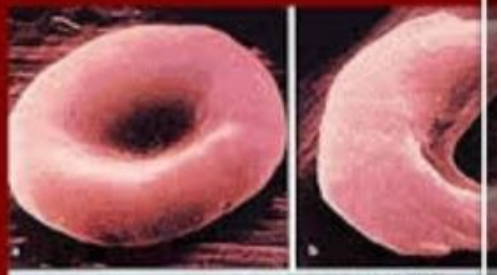
- **Sickle-Cell Anemia**: caused by a replacement of A by T on the 17th nucleotide for the beta chain of hemoglobin

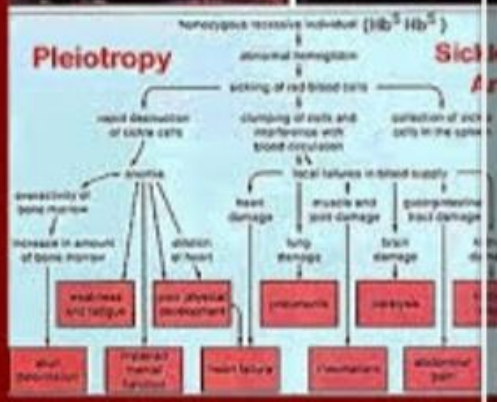
- RBC abnormal shape

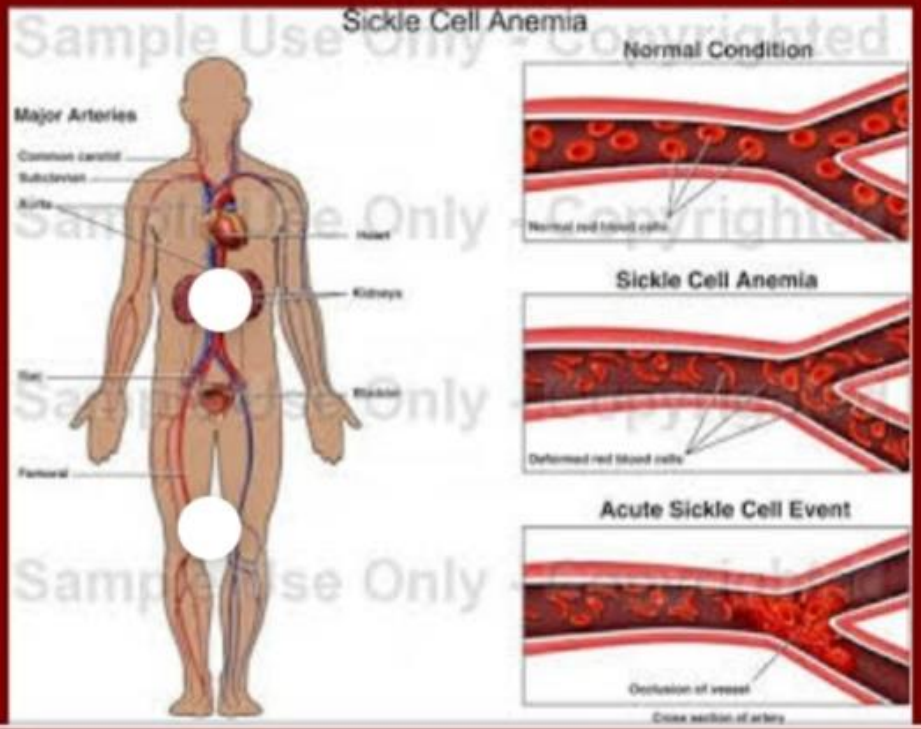


Sickle-Cell Anemia

- Caused by hemoglobin S that reduces O_2
- About 9% affected in U.S.
  - Homozygous recessive (ss)
- Heterozygous: (Ss) "carriers"
  - Can transmit gene to offspring







Pathophysiology

Deoxygenation

↓

polymerization of hemoglobin

↓

sickling of red cells

↓

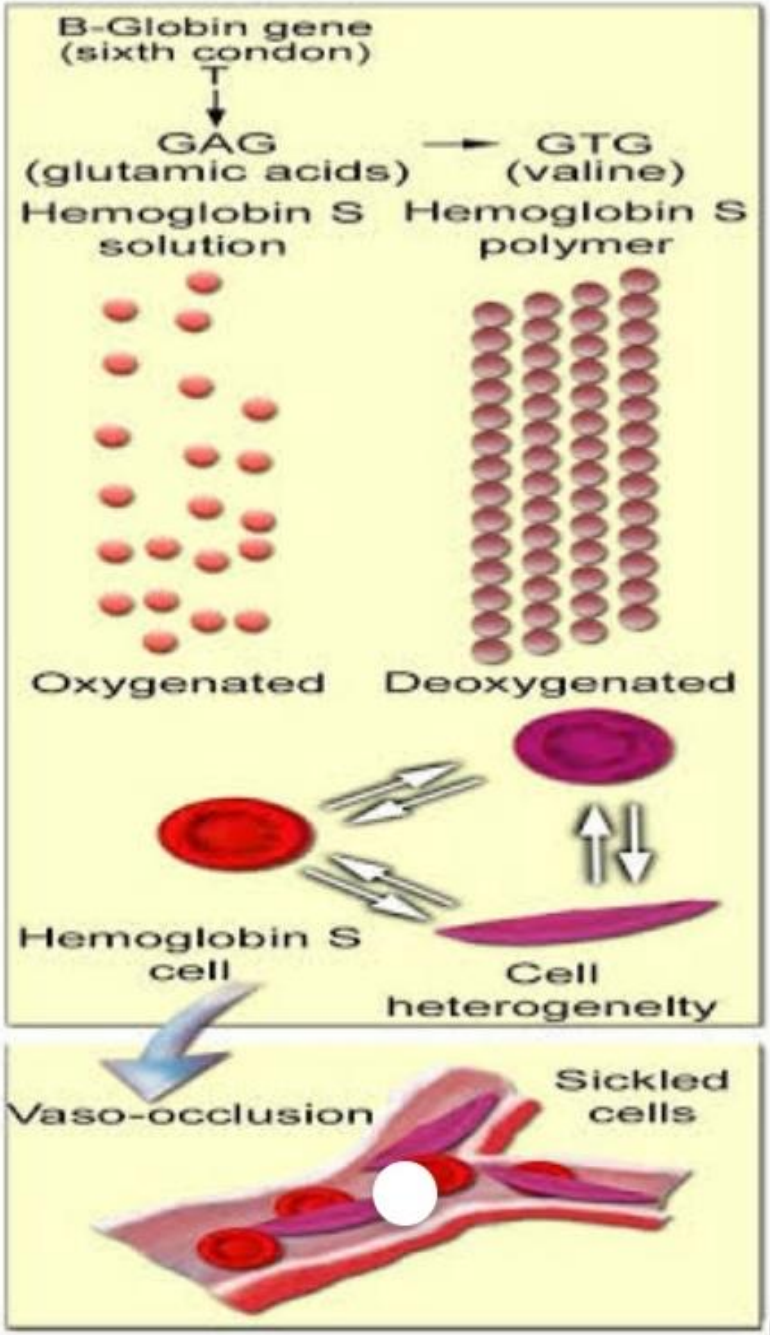
endothelial damage/activation

↓

RBC and leukocyte adhesion to endothelium, vasoconstriction

↓

vascular occlusion, organ ischemia and end-organ damage



Mechanism of Sickle cell formation

HbS

- Soluble, causes no problem when properly oxygenated
- Oxygen deficiency

↓

HbS- polymerizes form tactoid or fluids polymers

↓

Polymers realign → RBCs to deform

↓

Sickle shape 

↓

Sickling depends upon:

1. Decrease in oxygen
2. pH
3. Dehydration of patient

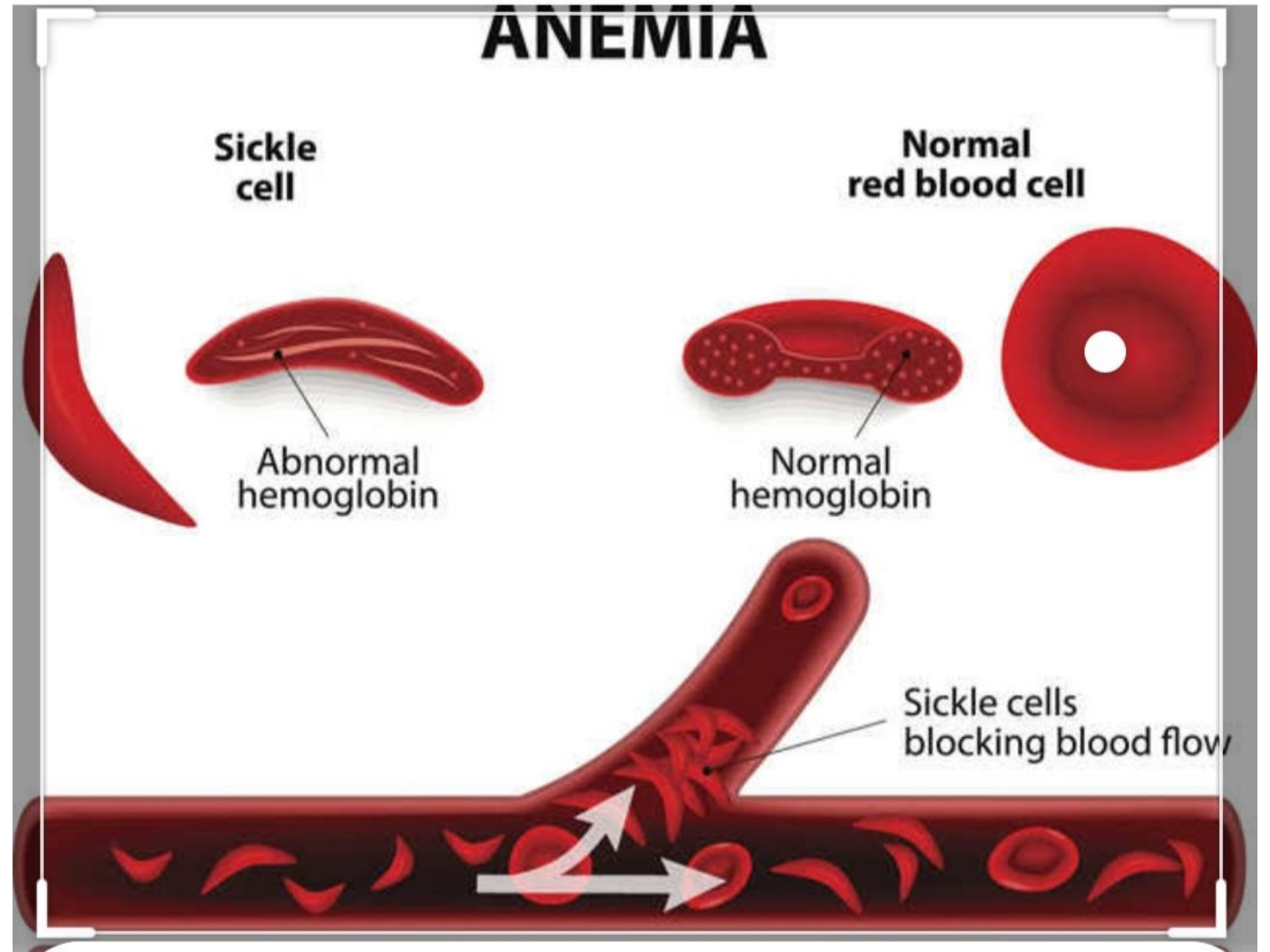
Sickle cell anemia signs and symptoms:

1. Anemia
2. Skin manifestation like leg ulcers
3. Arthritis
4. Renal disease
5. Eye involvement
6. Bone marrow infarcts
7. Autosplenectomy
8. Hapatomegaly
9. Gall bladder stones
10. Priapism
11. GI tract symptoms

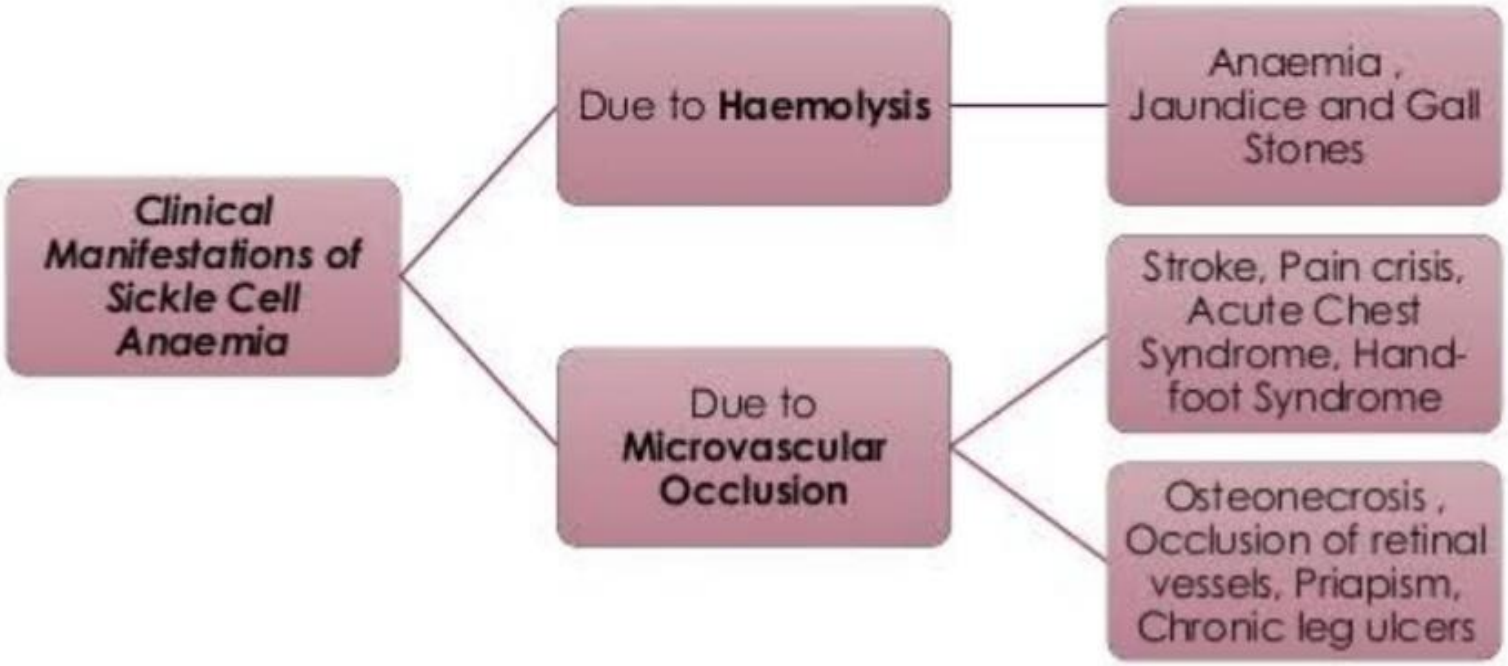
Sickle cell disease(SCD)- Clinical Manifestations

- **The presenting symptoms of SCD involve pain and anemia.**
- SCD usually manifests early in childhood. For the first 6 months of life, infants are protected largely by elevated levels of Hb F
- The disease is associated with **growth retardation, delayed sexual maturation**, and being underweight
- **Anemia** is universally present. It is chronic and hemolytic in nature and usually very well tolerated.
- Anemia may be complicated with Megaloblastic changes secondary to folate

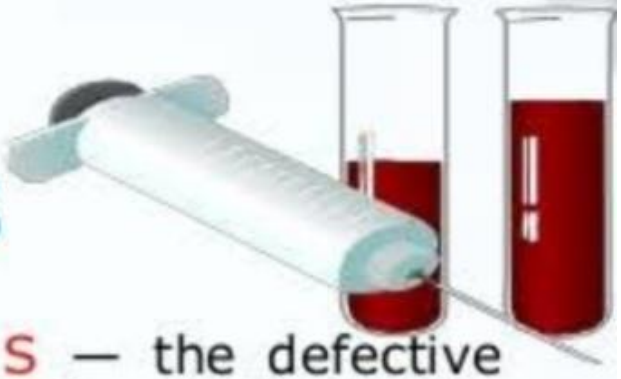
ANEMIA


Sickle cell
Normal red blood cell
Abnormal hemoglobin
Normal hemoglobin
Sickle cells blocking blood flow

CLINICAL MANIFESTATIONS OF SICKLE CELL ANAEMIA



Diagnosis

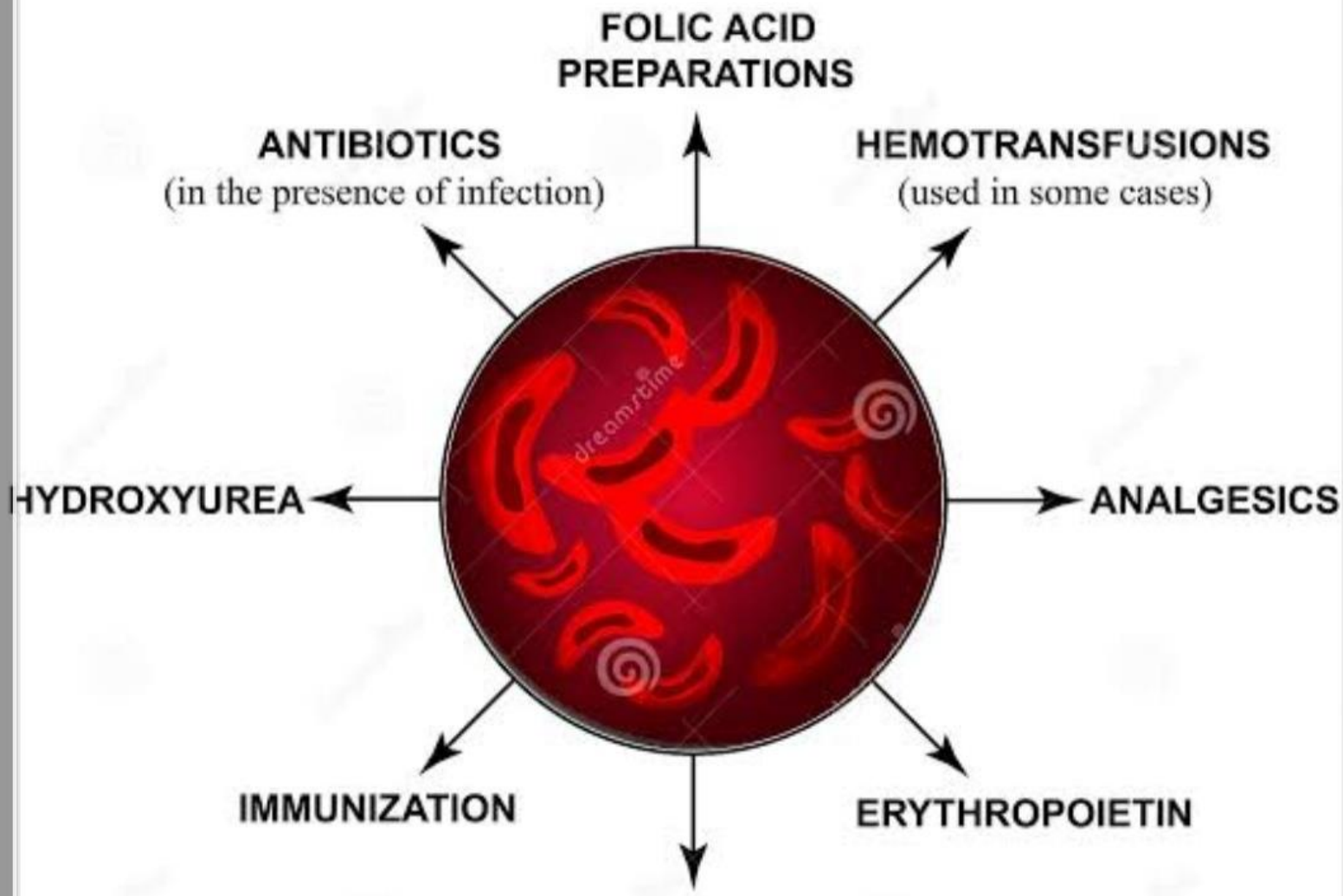


- A blood test can check for hemoglobin S — the defective form of hemoglobin that underlies sickle cell anemia. In the United States, this blood test is part of routine newborn screening done at the hospital. But older children and adults can be tested, too.
- In adults, a blood sample is drawn from 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.

Diagnosing sickle cell

- Sickle cell disease and the presence of the sickle cell trait can be diagnosed with blood tests or genetic tests.
- Sickle cell can be diagnosed at any age, but testing of newborns is preferred, so that treatment can start as early as possible.
- The disease can be diagnosed before birth by testing amniotic fluid or placental tissue.
- Parents can be tested for the trait using the same blood tests in order to determine if they are carriers.

TREATMENT OF SICKLE CELL ANEMIA



Treatment

Blood transfusions

- In a red blood cell transfusion, red blood cells are removed from a supply of donated blood, then given intravenously to a person with sickle cell anemia.
- Blood transfusions increase the number of normal red blood cells in circulation, helping to relieve anemia. In children with sickle cell anemia at high risk of stroke, regular blood transfusions can decrease the risk. Transfusions can also be used to treat other complications of sickle cell anemia, or they can be given to prevent complications.
- Blood transfusions carry some risk, including infection and excess iron buildup in your body. Because excess iron can damage your heart, liver and other organs, people who undergo regular transfusions might need treatment to reduce iron levels.

Treatment

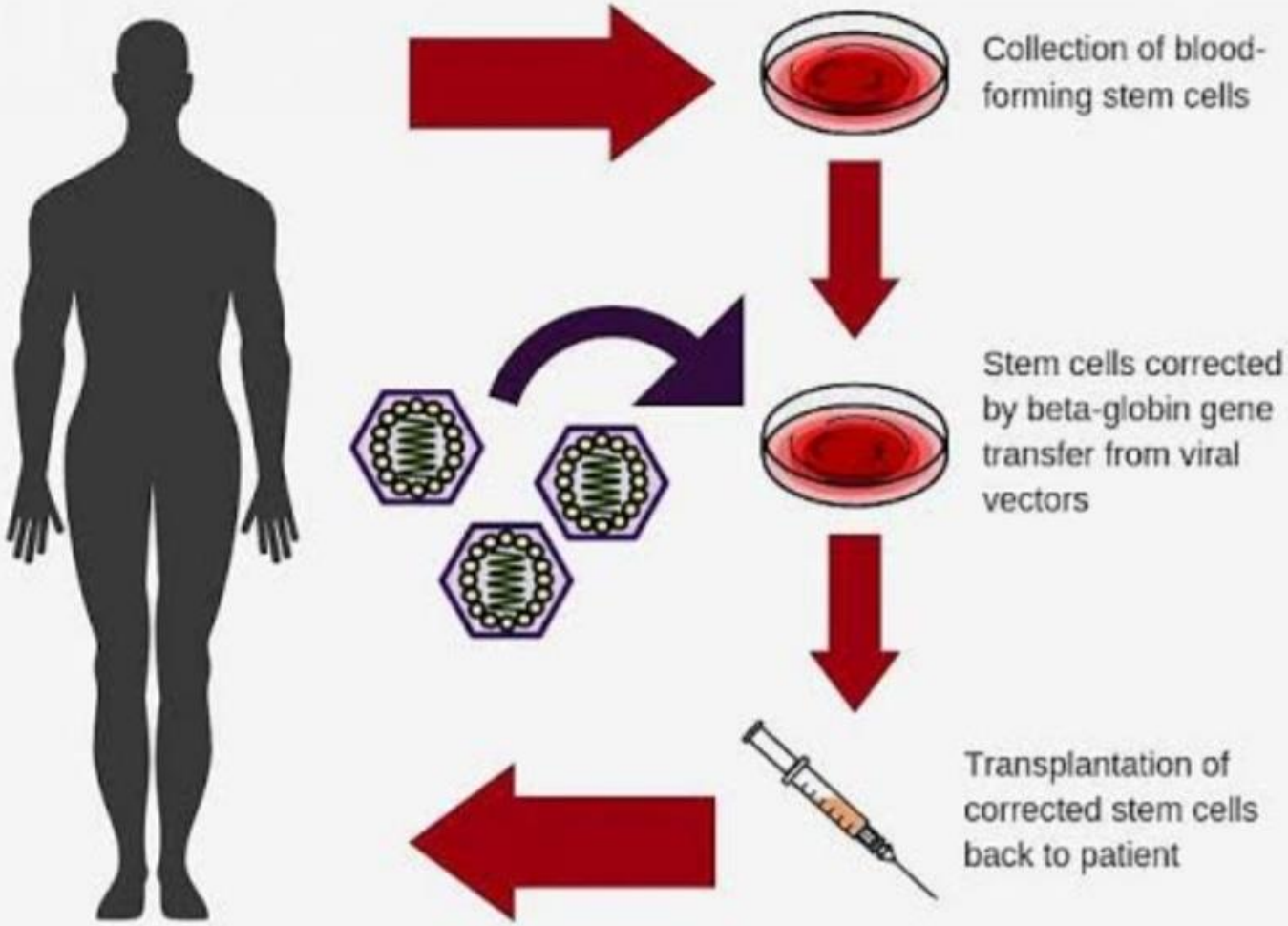
- Treatment is usually aimed at avoiding crises, relieving symptoms and preventing complications. Babies and children age 2 and younger with sickle cell anemia should make frequent visits to a doctor.
- Children older than 2 and adults with sickle cell anemia should see a doctor at least once a year, 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.

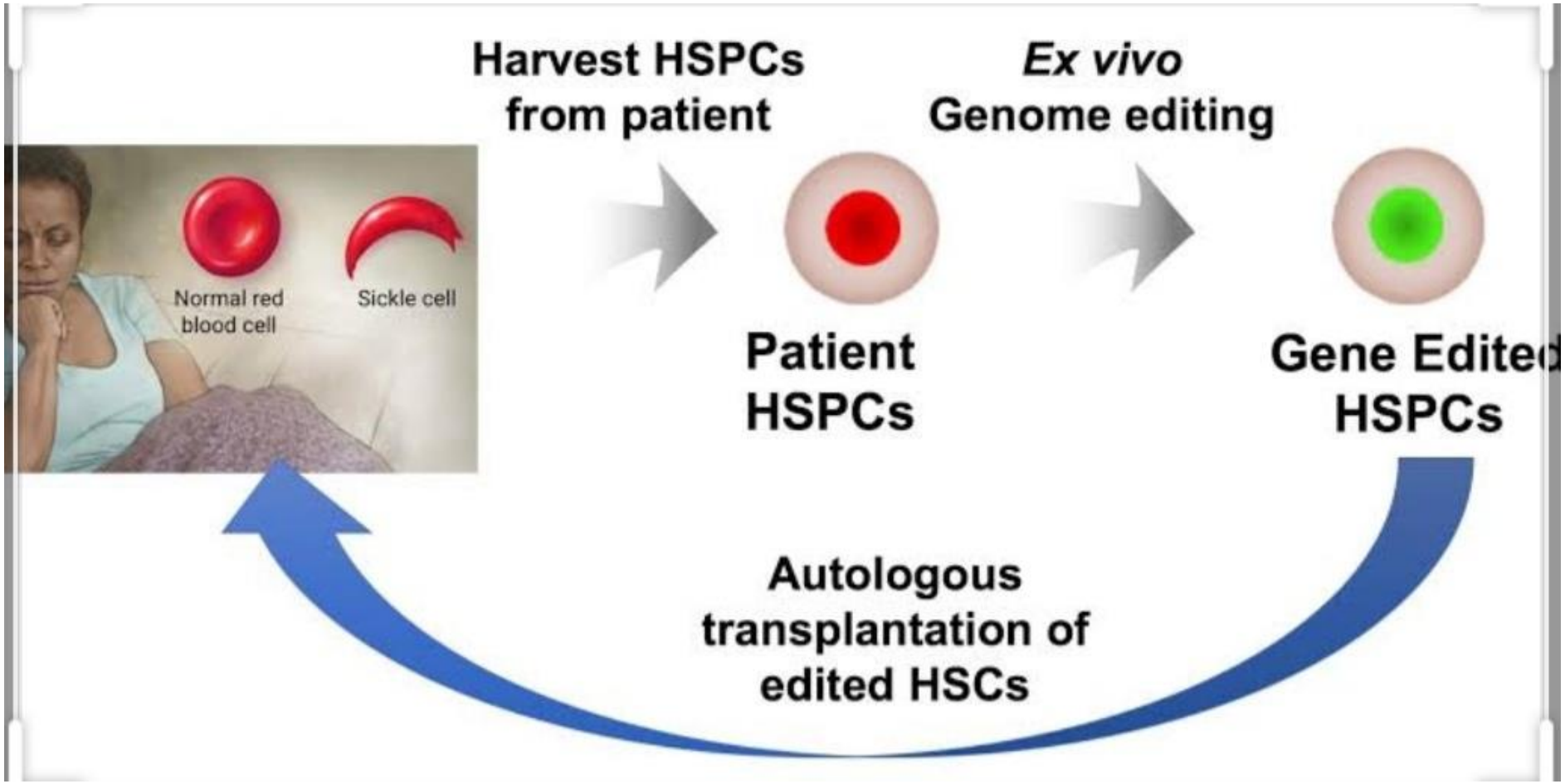
Explore treatment options.

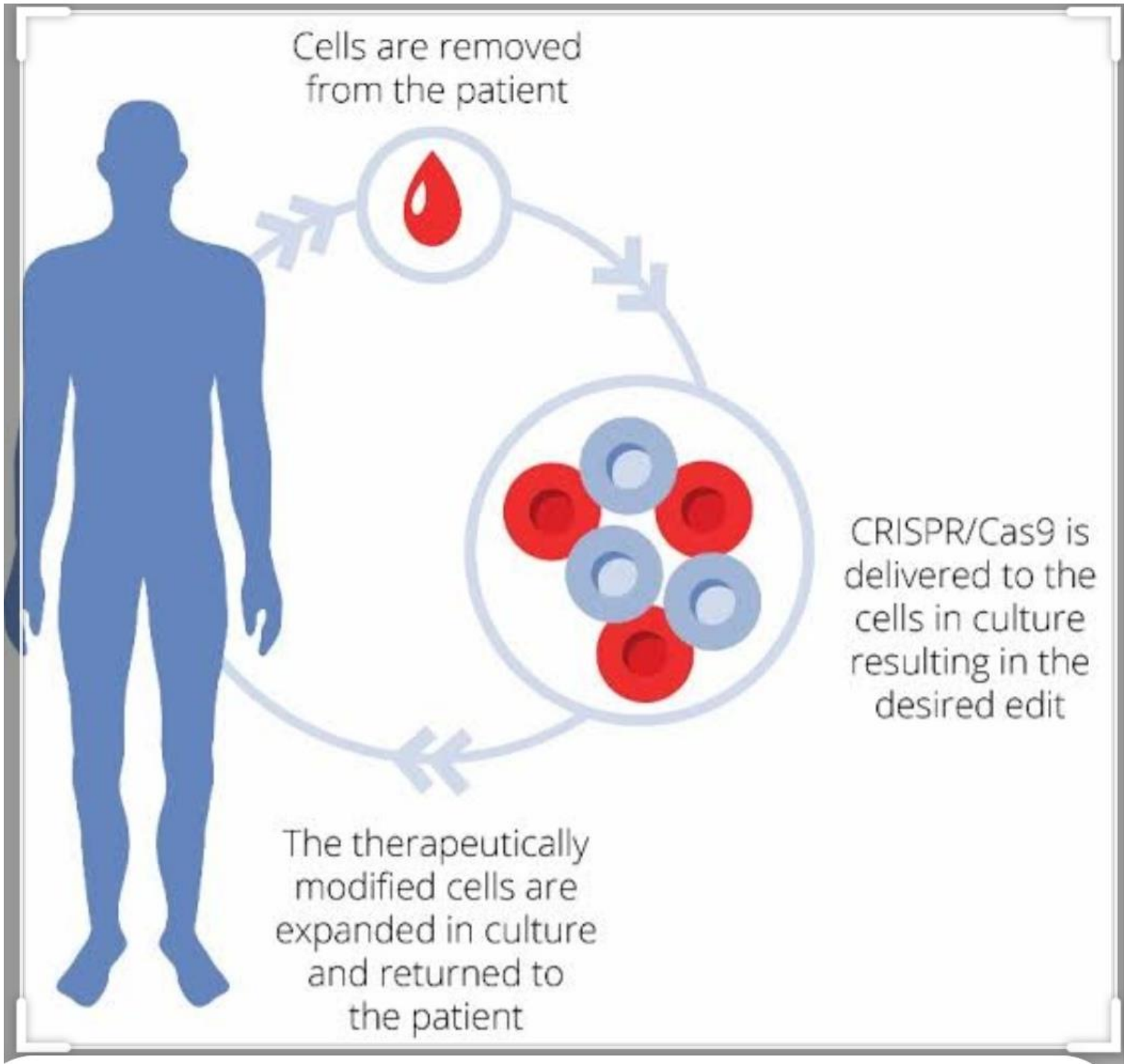
Compare benefits and risks.

Prepare for medical appointments.

Gene therapy for sickle cell disease




Harvest HSPCs
from patient
Ex vivo
Genome editing
Normal red
blood cell
Sickle cell
Patient
HSPCs
Gene Edited
HSPCs
Autologous
transplantation of
edited HSCs


Cells are removed from the patient
CRISPR/Cas9 is delivered to the cells in culture resulting in the desired edit
The therapeutically modified cells are expanded in culture and returned to the patient

Chronic Complications

- Anemia/Jaundice
- Brain Damage/Stroke
- Kidney failure
- Decreased lung function
- Eye disease (bleeding, retinal detachment)
- Leg ulcers
- Chronic pain management

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.

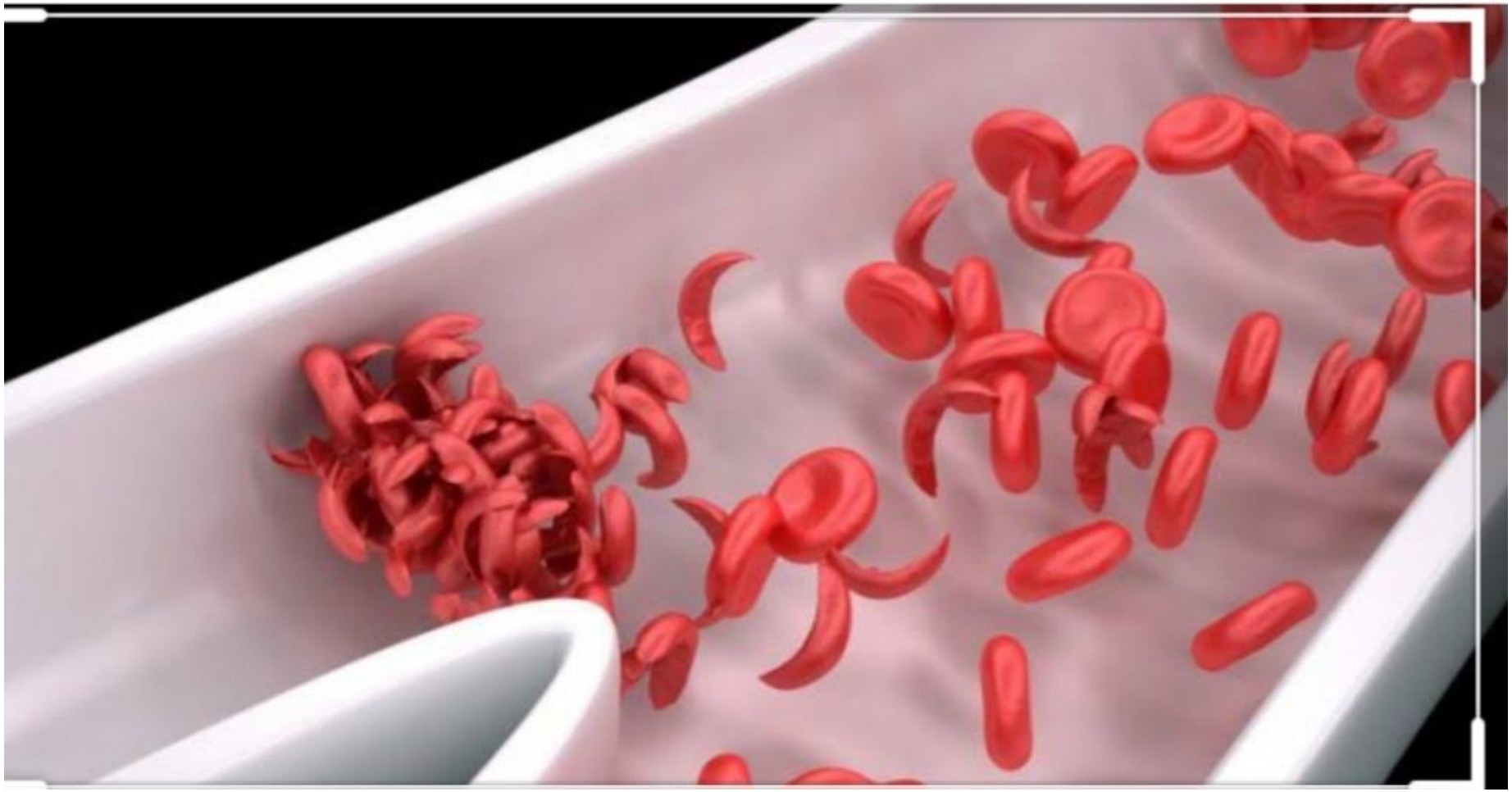
- **Pulmonary hypertension.** People with sickle cell anemia can develop high blood pressure in their lungs (pulmonary hypertension). This complication usually affects adults rather than children. Shortness of breath and fatigue are common symptoms of this condition, which can be fatal.

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.

Nursing Management

- Edu. of the family and child
- Early intervention for problems such as fever
- Administer antibiotics acc. To prescription
- Adequate hydration to prevent sickling
- Early recognition of resp. problems




thanks!