

# SICKLE CELL ANEMIA



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**By**

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## **INTRODUCTION:**

Sickle cell anemia is an inherited blood disorder that affects the production of hemoglobin in red blood cells. Hemoglobin is a protein in red blood cells that carries oxygen throughout the body.

People with sickle cell anemia have an abnormal form of hemoglobin, which causes abnormal production of red blood cells. These cells are different from normal size of RBC cell and can get stuck in small blood vessels, causing pain, organ damage, and other complications.

## **CAUSES/ETIOLOGY:**

- Genetic
- Sickle cell anemia is caused by a mutation in the HBB gene, which provides instructions for making a protein called beta-globin. The mutation causes the production of an abnormal form of hemoglobin called hemoglobin S. It causes red blood cells to become rigid and sickle-shaped, rather than flexible and round like healthy red blood cells.

## **PATHOPHYSIOLOGY:**

- Hemoglobin is a protein in red blood cells that carries oxygen from the lungs to the body's tissues and organs.
- Sickle cell anemia develops in the body due to a mutation in the HBB gene that provides instructions for making beta-globin.
- The mutation causes the production of an abnormal form of hemoglobin called hemoglobin S. Hemoglobin S causes red blood cells to become rigid and sickle-shaped, rather than flexible and round like healthy red blood cells.

- This sickling process can cause the cells to get stuck in small blood vessels, leading to reduced blood flow and oxygen supply to the body's tissues and organs.
- When sickle cells get stuck in small blood vessels, they can cause a blockage or a "sickle cell crisis". The symptoms of a sickle cell crisis can include pain, swelling, and inflammation, and can lead to organ damage, stroke, and other serious complications.

### **CLINICAL MENIFESTATION/SIGN & SYMPTOMS:**

- **Pain crisis:** This is the most common symptom of sickle cell anemia, characterized by severe and sudden pain in the chest, abdomen, bones, or joints. The pain can last for several hours or days and may require hospitalization.
- **Fatigue:** Due to the abnormal shape of the red blood cells, the body may not be able to transport enough oxygen, resulting in fatigue, weakness, and lethargy.
- **Shortness of breath:** The rigid and deformed red blood cells may also cause blockages in the blood vessels, leading to a shortage of oxygen in the lungs and shortness of breath.
- **Jaundice:** Sickle cell anemia can cause a buildup of bilirubin, a substance produced by the liver when it breaks down old red blood cells. This buildup can lead to yellowing of the skin and eyes, a condition known as jaundice.
- **Swelling of hands and feet:** The abnormal blood cells can also cause blockages in the small blood vessels in the hands and feet, leading to swelling and pain.
- **Frequent infections:** Sickle cell anemia can weaken the immune system, making individuals more susceptible to infections.

- **Delayed growth and development:** Children with sickle cell anemia may experience delayed growth and puberty due to the lack of oxygen supply to the body tissues.

## **DIAGNOSIS:**

- Physical examination
- Family history & Medical history.
- **Hemoglobin electrophoresis:** This test is used to identify the type of hemoglobin present in the blood. Hemoglobin is a protein in red blood cells that carries oxygen. Individuals with sickle cell anemia have abnormal hemoglobin called hemoglobin S.
- **Blood smear:** A blood sample is examined under a microscope to check for the presence of sickle-shaped red blood cells.
- **Complete blood count (CBC):** This test measures the number of red blood cells, white blood cells, and platelets in the blood. In sickle cell anemia, the red blood cell count is low, and the white blood cell count may be elevated due to frequent infections.
- **Solubility test:** A small amount of blood is mixed with a solution to determine if hemoglobin S is present.
- **DNA analysis:** This test is used to identify the specific genetic mutation responsible for sickle cell anemia. It can be used to confirm a diagnosis and to identify carriers of the disease.

## **PHARMACOLOGICAL MANAGEMENT:**

- **Hydroxyurea:** This drug is often used to reduce the frequency of painful crises and the need for blood transfusions in people with sickle cell anemia. It works by increasing the production of a type of hemoglobin that is less likely to sickle.

- ✚ Hydrea: This is a brand-name medication that contains hydroxyurea. It comes in the form of a capsule and is taken orally.
- ✚ Droxia: This is another brand-name medication that contains hydroxyurea. It is also taken orally in the form of a capsule.
- ✚ Siklos: This is a brand-name medication that contains hydroxyurea. It is available in the form of a tablet and is taken orally.
- ✚ Mylocel: This is another brand-name medication that contains hydroxyurea. It is also available in the form of a tablet and is taken orally.
- **L-glutamine:** This is a newer drug that has been approved for the treatment of sickle cell anemia. It is believed to work by increasing the production of a molecule called nitric oxide, which helps to relax blood vessels and improve blood flow.
- ✚ Endari: This is a brand-name medication that contains L-glutamine. It comes in the form of a powder that is mixed with water or other liquids and is taken orally.
- ✚ NutreStore: This is another brand-name medication that contains L-glutamine. It is also available in the form of a powder that is mixed with liquids and taken orally.
- **Pain medications:** Pain is a common symptom of sickle cell anemia, and over-the-counter pain medications like acetaminophen and ibuprofen can be used to manage mild to moderate pain. For more severe pain, opioids may be prescribed.

- **Antibiotics:** People with sickle cell anemia are at increased risk of developing infections, so antibiotics may be prescribed to prevent or treat infections.
- ✚ **Penicillin:** This is a common antibiotic that is used to prevent bacterial infections in children with sickle cell anemia. It is usually taken daily from infancy until the age of 5.
- ✚ **Macrolides:** This class of antibiotics includes drugs like azithromycin and clarithromycin, which can be used to treat respiratory infections in people with sickle cell anemia.
- ✚ **Cephalosporins:** This class of antibiotics includes drugs like cephalexin, ceftriaxone, and cefepime, which can be used to treat a wide range of bacterial infections.

## **NON PHARMACOLOGICAL MANAGEMENT/PREVENTION:**

- **Genetic counseling:** If you have a family history of sickle cell anemia or are at risk of being a carrier, genetic counseling can help you understand your risk and make informed decisions about family planning.
- **Prenatal testing:** Prenatal testing, such as chorionic villus sampling or amniocentesis, can be used to detect sickle cell anemia in a developing fetus. This can help parents make decisions about continuing the pregnancy and planning for the care of a child with sickle cell anemia.

- **Carrier screening:** If you are planning to have children, carrier screening can determine if you or your partner are carriers of the sickle cell gene. This can help you make informed decisions about family planning.
- **Newborn screening:** All newborns in the United States are screened for sickle cell anemia shortly after birth. Early detection can help ensure prompt treatment and management of the condition.
- **Avoiding high-risk situations:** People with sickle cell anemia are at increased risk of complications from certain situations, such as extreme temperatures, dehydration, and high altitudes. Avoiding these situations can help reduce the risk of complications.