

# Sickle cell anemia case study



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A 20-year-old Africa- America woman visits her physical complaining of episodes of extreme pain and discomfort in her legs and lower back. She has been experiencing these recurrent episodes, accompanied by extreme fatigue, since she was a child. On physical examination, she appears jaundiced and has a hematocrit of 23% and a hemoglobin level of 7g/dL. She reports she has family members who experienced the same symptom.

Sickle cell anemia (sickle cell disease) is a disorder of the blood caused by an inherited abnormal hemoglobin (an oxygen-carrying protein within the red blood cells). The abnormal hemoglobin causes distorted (sickled) red blood cells. The sickled red blood cells are fragile and prone to rupture. When the number of red blood cells decreases from rupture (hemolysis), anemia is the result. This condition is referred to as sickle cell anemia. The irregular sickled cells can also block blood vessels causing tissue and organ damage and pain.

Sickle cell anemia is one of the most common inherited blood anemias. The disease primarily affects Africans and African Americans. It is estimated that in the United States, some 50, 000 African Americans are afflicted with the most severe form of sickle cell anemia. Overall, current estimates are that

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one in 1, 875 U. S. African American is affected with sickle cell anemia. Sickle cell anemia is caused by a point mutation in the  $\beta^2$ -globin chain of haemoglobin, causing the hydrophilic amino acid glutamic acid to be replaced with the hydrophobic amino acid valine at the sixth position. The  $\beta^2$ -globin gene is found on the short arm of chromosome 11. The association of two wild-type  $\beta^2$ -globin subunits with two mutant  $\beta^2$ -globin subunits forms haemoglobin S (HbS). Under low-oxygen conditions (being at high altitude, for example), the absence of a polar amino acid at position six of the  $\beta^2$ -globin chain promotes the non-covalent polymerisation (aggregation) of haemoglobin, which distorts red blood cells into a sickle shape and decreases their elasticity.

The loss of red blood cell elasticity is central to the pathophysiology of sickle-cell disease. Normal red blood cells are quite elastic, which allows the cells to deform to pass through capillaries. In sickle-cell disease, low-oxygen tension promotes red blood cell sickling and repeated episodes of sickling damage the cell membrane and decrease the cell's elasticity. These cells fail to return to normal shape when normal oxygen tension is restored. As a consequence, these rigid blood cells are unable to deform as they pass through narrow capillaries, leading to vessel occlusion and ischaemia.

The actual anaemia of the illness is caused by haemolysis, the destruction of the red cells inside the spleen, because of their misshape. Although the bone marrow attempts to compensate by creating new red cells, it does not match the rate of destruction.[17] Healthy red blood cells typically live 90-120 days, but sickle cells only survive 10-20 days.[18]

Normally, humans have Haemoglobin A, which consists of two alpha and two beta chains, Haemoglobin A2, which consists of two alpha and two delta chains and Haemoglobin F, consisting of two alpha and two gamma chains in their bodies. Of these, Haemoglobin A makes up around 96-97% of the normal haemoglobin in humans.

In normal Haemoglobin A, glutamic acid is on the 6th position of the beta chain, while in sickle-cell disease, this glutamic acid is replaced by valine leading to the formation of sickle cells. This happens due to a one point mutation. This leads to polymerization of the two beta chains and therefore their appearance as puzzle pieces (or lock and key); which means they fit into each other forming a longitudinal polymer that would lead to the cell becoming deformed and very rigid leading to vessel occlusion. This process of polymerization can be activated by infections, hypoxia, acidosis, physical exercise, vasoocclusion due to cold as well as hypertonic dehydration.

## **Diagnosis**

Sickle cell anemia is diagnosed through blood test, testing for hemoglobin S (the defective form of hemoglobin descriptive of the disease), the presence of other abnormal hemoglobin variants, evaluating status and number of erythrocytes, and/or determination of one or more altered hemoglobin gene copies. In the United States, this blood test is part of routine screening for newborns done in the hospital. However, older children and adults can be tested also. In adults, the blood sample is extracted from a vein in the arm. In young children and babies, blood is taken from a finger or heel. The testing itself is typically performed on a smear of blood utilizing a special

low-oxygen preparation, known as sickle prep. Other prep tests can be utilized, including but not limited to solubility tests. [4, 6]

Another screening testing is the Hb S solubility test. In this procedure, a chemical is added to the blood sample which reduces the amount of its oxygen carrying capacity. In individuals carrying even one sickle gene, some hemoglobin S will be present. The reduced amount of oxygen will cause S-related polymers to form and affected erythrocytes will sickle. This test, in essence, detects for the presence of Hb S alone. However, this exam should not be performed on infants until age six months, as babies with sickle cell will not produce significant amounts of Hb S until several months after birth. [6]

To confirm the diagnosis, DNA analysis can be utilized. This exam is used to detect alterations and mutations in the genes producing hemoglobin components. DNA analysis reveals one copy or two copies of the hemoglobin S gene, or copies of different hemoglobin variants. DNA analysis can be performed on the developing fetus in fourteen to sixteen weeks gestations via amniocentesis or through chorionic villus sampling.[6]

## **Treatment**

Treatment of sickle cell anemia is done by blocking the red blood cells from stacking together.

the health professional maintenance helps the patients to begin with early diagnosis of the disorder, preferably during the newborn period. Penicillin prophylaxis, vaccination against pneumococcus bacteria, and folic acid supplementation is standard. [2]

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Treatment of sickle cell complications includes , vitamin supplementation, intravenous fluids, blood transfusion, supplemental oxygen, surgery (splenectomy) and psychosocial support. Management is best accomplished via multidisciplinary program of care. [2, 3, 5]

Blood transfusions benefit by reducing recurring pain crises, risk of stroke, and other complications. Blood transfusions increase the amount of normocytic erythrocytes in circulation, helping relieve the anemic state. However, since erythrocytes contain iron and the body does not possess a natural process for its elimination, patients can accumulate iron in the blood. Thus, possible iron toxicity must be closely screened and methods to remove excess must be executed. Excess iron is removed artificially through administration of the drug Deferasirox (Exjade ®) orally in patients two years of age and older. If this is not checked, the excessive iron can accumulate in the heart, liver, and various other organs causing organ damage. [3]

Other treatments for this disease include finding a substance that prevents erythrocytes from sickling without producing deleterious effects to other body areas. The medication hydroxyurea has been found to reduce the frequency and severity of pain, acute chest syndrome, and decrease the need for blood transfusions in adult patients. Droxia ® (prescription name brand formulation of hydroxyurea) was approved by the Food and Drug Administration in 1998 and is currently available for adult patients. Studies are currently being conducted to determine the proper dosage in pediatric patients. However, there is concern in this medication that chronic usage may facilitate tumor growth or leukemia in certain individuals. [5]

Other pharmacological therapies include antibiotics and pain relievers.

Regarding antibiotics and aforementioned earlier, children benefit from penicillin at age two months and usually continue medication until age five. Prophylactic antibiotic treatment helps in preventing pneumonia. For adults, prophylactic antibiotic treatment can aide in fighting certain infections that they would normally fight provided they had normal erythrocytes. [2, 3, 5]

Non-pharmacotherapy treatments are bone marrow transplant and supplemental oxygen therapy. Bone marrow transplant procedure offers the only potential cure for sickle cell anemia. Replacing the system with unaffected bone marrow aids the body in producing normocytic erythrocytes. However, finding a donor provides difficult even with the advent of registries. Also, the extraction of bone marrow possesses serious risk, including death. After the procedure, a mandatory lengthy hospital stay is required. In the hospital and upon leaving, the patient will be administered medications to help prevent rejection of the donated marrow. The procedure is currently only used for those possessing serious symptoms and problems with sickle cell anemia. Regarding supplemental oxygen therapy, this proves beneficial by forcefully increasing oxygen content in the blood via oxygen mask. Supplemental oxygen can be helpful in acute chest syndrome or sickle cell crisis.[2, 3, 5]

Surgical treatment involve splenectomy to remove a possible damaged spleen from the sickle cells or eye surgery for vision problems associated with sickled cell damage.

New sickle cell treatments on the horizon include gene therapy, the pharmacological treatments of butyric acid, clotrimazole, nitric oxide, and nicosan. [3]

Since sickle cell anemia is caused by a defective gene, researchers speculate that insertion of a normal gene into bone marrow of people with sickle cell anemia will result in the production of normal hemoglobin. Another gene therapy possibility is “ turning off” the defective gene, while reactivating another gene responsible for production of fetal hemoglobin (a type of hemoglobin found in newborns) that prevent sickle cells from forming. [3]

Butyric acid, normally utilized as a food additive, may increase the amount of fetal hemoglobin in the blood in some patients. [3]

Clotrimazole, the over-the-counter antifungal medication may help prevent loss of water from erythrocytes, possibly reducing the number of sickle cells formed. [3]

Nitric oxide is decreased in sickle cell anemia, a gas that normal causing vasodilatation. Administration of this agent would prevent the sticking of sickled cells to one another.[3]

Nicosan, an herbal treatment in early trials in the United States, is currently being used to prevent sickle crisis in Nigeria (West Africa). [3]

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