Sickle cell anemia

Abstract

Sickle cell anemia is an inherited blood disorder characterized primarily by chronic anemia and periodic episodes of pain. The underlying problem involves hemoglobin, a component of red blood cells. Hemoglobin molecules in each red blood cell carry oxygen from the lungs to body organs and tissues and bring carbon dioxide back to the lungs.

In sickle cell anemia, the hemoglobin is defective. After hemoglobin molecules give up their oxygen, some may cluster together and form long, rod-like structures. These structures cause red blood cells to become stiff and assume a sickle shape.

Unlike normal red cells, which are usually smooth and donut-shaped, sickle red cells cannot squeeze through small blood vessels. Instead, they stack up and cause blockages that deprive organs and tissues of oxygen-carrying blood.

“ Normal red blood cells live about 120 days in the bloodstream, but sickle red cells die after about 10 to 20 days. Because they cannot be replaced fast enough, the blood is chronically short of red blood cells, a condition called anemia”.

Sickle cell anemia affects millions throughout the world. “ It is particularly common among people whose ancestors come from Africa; South America, Cuba, Central America; Saudi Arabia; India; and Mediterranean countries such as Turkey, Greece, and Italy”.

Sickle cell anemia disease

The inherited haemoglobinopathies are a group of disorders that include thalassaemia and sickle-cell disease. “ These diseases are a major public health problem in the Mediterranean area, the Middle East, the Indian subcontinent, Asia, tropical Africa and the Caribbean. However, because of population flow, they are now widespread and occur in Europe and North and South America”. According to the World Health Organization, the approximate estimates of affected individuals indicate that 240 million people are heterozygous for these disorders and at least 200000 lethally affected homozygotes are born annually, approximately equally divided between sickle-cell anaemia and thalassaemia syndromes.

This research papers about the definition, causes, types, signs and symptoms, complications, diagnosis and treatment of sickle cell disease. Also, this paper talks about the general guidelines to keep the sickle cell patient healthy and recommendation that the patient and families should watch for it.

1-Definition

“ Sickle cell anemia (uh-NEE-me-uh) is a serious disease in which the body makes sickle-shaped red blood cells. “ Sickle-shaped” means that the red blood cells are shaped like a C. Normal red blood cells are disc-shaped and look like doughnuts without holes in the center. They move easily through your blood vessels. Red blood cells contain the protein hemoglobin (HEE-muh-glow-bin). This iron-rich protein gives blood its red color and carries oxygen from the lungs to the rest of the body. Sickle cells contain abnormal hemoglobin that causes the cells to have a sickle shape. Sickle-shaped cells

don’t move easily through your blood vessels. They’re stiff and sticky and tend to form groups and get stuck in the blood vessels. (Other cells also may play a role in this grouping process.)”

The groups of sickled cells block blood flow in the blood vessels that lead to the limbs and organs. Blocked blood vessels can cause pain, serious infections, and organ damage.

2- Causes

Sickle cell anemia is an autonomic recessive genetic disorder caused by a defect in the HBB gene, which codes for hemoglobin. “ The presence of two defective genes (SS) is needed for sickle cell anemia. If each parent carries one sickle hemoglobin gene (S) and one normal gene (A), each child has a 25% chance of inheriting two defective genes and having sickle cell anemia; a 25% chance of inheriting two normal genes and not having the disease; and a 50% chance of being an unaffected carrier like the parents.”

Two of the most common variations of the sickle cell gene are:

A- Sickle cell trait:

A person with the sickle cell trait is carrying the defective gene, but also has some normal hemoglobin. Individuals with sickle cell trait are usually without symptoms of the disease. Mild anemia may occur. Under intense stressful conditions, exhaustion, hypoxia (low oxygen), and/or severe infection, the sickling of the defective hemoglobin may occur and result in some complications associated with the sickle cell disease.

B- Sickle cell anemia:

A person with sickle cell anemia has most or all of the normal hemoglobin replaced with the sickle hemoglobin. It is the most common and most severe form of the sickle cell variations." These individuals suffer from a variety of complications due to the shape and thickness of the sickle cells. Due to the decreased number of hemoglobin cells circulating in the body, severe and chronic anemia is also a common characteristic".

3- Sign and Symptoms

The clinical course of sickle cell anemia does not follow a single pattern; some patients have mild symptoms, and some have very severe symptoms. The basic problem, however, is the same: the sickle-shaped red blood cells tend to get stuck in narrow blood vessels, blocking the flow of blood. These results in the following conditions:

A- Hand-foot syndrome:

When small blood vessels in hands or feet are blocked, pain and swelling can result, along with fever. This may be the first symptom of sickle cell anemia in infants.

Fatigue, paleness, and shortness of breath:

These are all symptoms of anemia or a shortage of red blood cells.

B- Pain that occurs suddenly in any body organ or joint:

A patient may experience pain wherever sickle blood cells block oxygen flow to tissues. " The frequency and amount of pain vary. Some patients have painful episodes (also called crises) less than once a year, and some have as many as 15 or more episodes in a year. Sometimes pain lasts only a few

hours; sometimes it lasts several weeks.” For severe continuous pain, the patient may be hospitalized and treated with painkillers and intravenous fluids. Pain is the principal symptom of sickle cell anemia in both children and adults.

C -Eye problems:

The retina, the film at the back of the eye that receives and processes visual images, can deteriorate when it does not get enough nourishment from circulating red blood cells. Damage to the retina can be serious enough to cause blindness.

D- Yellowing of skin and eyes:

These are signs of jaundice, resulting from rapid breakdown of red blood cells.

Delayed growth and puberty in children and often a slight build in adults: The slow rate of growth is caused by a shortage of red blood cells

4- Complications

A-Infections:

In general, both children and adults with sickle cell anemia are more liable to infections and have a harder time fighting them off. This is the result of spleen damage from sickle red cells, hence preventing the spleen from destroying bacteria in the blood. Also the bone marrow gets enlarged because of the increasing need to produce red blood cells. Infants and young children especially are suspectible to bacterial infections that can kill them in as little as 9 hours from onset of fever.

“ Pneumococcal infections used to be the principal cause of death in children with sickle cell anemia until physicians began routinely giving penicillin on a preventive basis to those who are diagnosed at birth or in early infancy”

B- Stroke:

Defective hemoglobin damages the walls of red blood cells, causing them to stick to blood vessel walls. The resulting narrowed or blocked small blood vessels in the brain can lead to serious, life-threatening strokes, primarily in children.

C-Acute chest syndrome:

Similar to pneumonia, this life-threatening complication is caused by infection or trapped sickle cells in the lung. It is characterized by chest pain, fever, and an abnormal chest X-ray.

5- Diagnosis

Early diagnosis of sickle cell anemia is critical so children who have the disease can receive proper treatment.

Blood test:

More than 40 states now perform a simple, inexpensive blood test for sickle cell disease on all newborn infants. This test is performed at the same time and from the same blood samples as other routine newborn-screening tests. Hemoglobin electrophoresis is the most widely used diagnostic test. If the test shows the presence of sickle hemoglobin, a second blood test is performed to confirm the diagnosis. These tests also tell whether or not the child carries the sickle cell trait.

6- Treatment

Although there is no cure for sickle cell anemia, doctors can do a great deal to help patients, and treatment is constantly being improved. Basic treatment of painful crises relies heavily on painkilling drugs and oral and intravenous fluids to reduce pain and prevent complications.

A- Blood Transfusions:

“ Transfusions correct anemia by increasing the number of normal red blood cells in circulation. They can also be used to treat spleen enlargement in children before the condition becomes life-threatening”. Regular transfusion therapy can help prevent recurring strokes in children at high risk.

B-Oral Antibiotics:

Giving oral penicillin twice a day beginning at 2 months and continuing until the child is at least 5 years old can prevent pneumococcal infection and early death. Recently, however, several new penicillin-resistant strains of pneumonia bacteria have been reported. Since vaccines for these bacteria are ineffective in young children, studies are being planned to test new vaccines.

C-Hydroxyurea

“ The first effective drug treatment for adults with severe sickle cell anemia was reported in early 1995, when a study conducted by the National Heart, Lung, and Blood Institute showed that daily doses of the anticancer drug hydroxyurea reduced the frequency of painful crises and acute chest syndrome”. Patients taking the drug needed less blood transfusions.

Regular health maintenance is critical for people with sickle cell anemia. Proper nutrition, good hygiene, bed rest, protection against infections, and avoidance of other stresses all are important in maintaining good health and preventing complications. Regular visits to a physician or clinic that provides comprehensive care are necessary to identify early changes in the patient's health and ensure immediate treatment.

Today, with good health care, many people with sickle cell anemia are in reasonably good health much of the time and living productive lives. In fact, in the past 30 years, the life expectancy of people with sickle cell anemia has increased.

What can be done to help prevent these complications?

Sickle cell patient should be under the care of a medical team that understands sickle cell disease. All newborn babies detected with sickle cell disease should be placed on daily penicillin to prevent serious infections. All of the childhood immunizations should be given in addition to the pneumococcal vaccine. Parents should know how to check for a high temperature because this signals the need for a quick medical checkup for serious infection.

The following are general guidelines to keep the sickle cell patient healthy:

1. Taking the vitamin folic acid (foliate) daily to help make new red cells
2. Daily penicillin until age six to prevent serious infection
3. Drinking plenty of water daily (8-10 glasses for adults)
4. Avoiding too hot or too cold temperatures
5. Avoiding over exertion and stress

6. Getting a sufficient amount of rest
7. Getting regular check-ups from knowledgeable health care providers

Patients and families should watch for the following conditions that need an immediate medical evaluation:

1. Fever
2. Chest pain
3. Shortness of Breath
4. Increasing tiredness
5. Abdominal swelling
6. Unusual headache
7. Any sudden weakness or loss of feeling
8. Pain that will not go away with home treatment
9. Pianism (painful erection that will not go down)
10. Sudden vision change.

Conclusion

To conclude, sickle cell anemia like other chronic life-threatening disease can cause this inherited to patient and family members joining where members share common experiences and problems can reveal this situation because it gets better understanding and management of the disease. There is no cure, therefore nurses should understand the actions that can prevent or relieve symptoms in order to meet the challenges of caring for patients with sickle cell disease and helping them to minimize its effect on their lives.

References

1. Brunner and Suddarth , Medical-Surgical Nursing , (2004), 10th edition
2. http://www. nhlbi. nih. gov/health/dci/Diseases/Sca/SCA_Summary. html

3. http://www. mayoclinic. com/health/sickle-cell-anemia/DS00324
4. http://www. nhlbi. nih. gov/health/dci/Diseases/Sca/SCA_WhatIs. html
5. http://www. kidshealth. org/teen/diseases_conditions/genetic/sickle_cell_anemia. html
6. http://www. medicinenet. com/sickle_cell/article. htm
7. Desai, D. V.; Hiren Dhanani (2004). “ Sickle Cell Disease: History And Origin”. The Internet Journal of Heamatology 1 (2). http://www. ispub. com/ostia/index. php? xmlFilePath= journals/ijhe/vol1n2/sickle. xml.
8. Pearson H (Aug 1977). “ Sickle cell anaemia and severe infections due to encapsulated bacteria” http://www. nlm. nih. gov/medlineplus/meningitis. html.
9. http://www. emro. who. int/Publications/EMHJ/0303/03. htm