Pathophysiology of sickel cell disease

Health & Medicine, Nursing

Sickle Cell Anemia of the of the Sickle Cell Anemia Introduction I chose to research on sickle cell anemia because my sister has the disease. As a child, my whole life changed as I had to “ live” for her. I stayed by her side rubbing her joints as I watched her cry in excruciating pain and this always put me in a state of empathy. For the life she has lived, her days of no pain are very countable, and my one greatest fear is that she may never make it to the age of 40 because of the long term effects and prognosis of the disease.

Sickle cell anemia is a non treatable hereditary problem which has an effect on the potential to produce healthy red blood cells. It results from homozygosity for a mutation in the beta globin gene and is transmitted by autosomal recessive inheritance (Passarge, 2007). A single nucleotide polymorphism (SNP) occurs in the beta globin chain, which results in the substitution of the amino acid residue glutamine at position 6 for valine. An individual may have one such abnormal gene and another normal gene and is said to be heterozygous. When a couple that is heterozygous for HbS give birth to children, there is a likelihood of one or more of the children inheriting these HbS genes from each of the parents. When this happens, the child has both HbS genes and is said to be homozygous for HbS. This is when a child suffers from sickle cell disease.

This disorder has potentially serious complications and early medication (prior to the development of symptoms) can advance morbidity and mortality. It affects hemoglobin, a protein that is part of red blood cells, which is responsible for oxygen transport. It is characterized by the presence of sickle cells in the bloodstream, which is also known as erythrocyte sickling. This causes difficulties in circulating red blood cells as the blood

vessels get clogged and cause symptoms such as pain in the extremities (Gordon, 2000).

This disorder develops within the first 2 years, and the symptoms can be traced back to two main phenomena which are hemolysis and clots. Hemolysis causes chronic anemia, jaundice, lack of growth and sexual maturation. The vascular occlusions cause crisis musculoskeletal pain, increase susceptibility to bacterial infections (S. pneumonia, K. pneumonia, Homophiles influenza, etc.), cerebral infarction, and renal impairment.

Discussion

Pathophysiological Implication

As a consequence of the mutation, when the hemoglobin is deoxygenated, it spontaneously undergoes polymerization to form a crystalline gel. Each polymer is composed of longitudinal beams 14 of deoxy-Hb which are arranged to form a tactoid body, insoluble and rigid cylindrical structure because these polymers, breaks the cytoskeleton of the erythrocyte, adopting this form of sickle cell trait (Maakaron, 2012).

Although the sickling phenomenon is reversible, between 5 and 50% of sickle erythrocytes fail to regain its original shape, being eliminated by the mononuclear phagocyte system. Furthermore, erythrocytes have a large decrease altered corpuscular volume and a large increase in the concentration of hemoglobin. This is because the deoxy-Hb induces alterations of the erythrocyte membrane (modification of the distribution and composition of the phospholipids in the bilayer) which are translated by a deep dehydration. Additionally, sickle exhibits a strong tendency to adhere to vascular endothelium favoring the formation of micro thrombi and

peripheral vascular occlusions (Maakaron, 2012).

Interestingly hemoglobin S (HbS) may interact with other forms of hemoglobin, particularly fetal hemoglobin (HbF). The presence of hemoglobin thus reduces the degree of polymerization of HbS, which explains that the sickle is not present during the neonatal period or hereditary persistence of fetal hemoglobin (Maakaron, 2012).

Symptoms

Sickle cell anemia indications do not follow a single pattern. It has been observed that some people have mild indications, and some have very severe indications. However, the fundamental predicament is the same where the red blood cells which are sickle shaped block the narrow vessels carrying blood disturbing the flow of blood resulting in a number of conditions. Anemia is the key feature of sickle cell disease, which produces a mandatory high cardiac output that may lead to cardiomegaly and failure of the heart. Occlusive pathology affects various regions of the body including the kidneys, lungs, spleen, the liver, and the brain.

The first signs of sickle-cell anemia in infants usually are swelling and tenderness of the hands or feet, fatigue, distortion of limbs, and sometimes failure to walk. This symptom is a result of blockage of red blood capillaries of small bones in hands and feet and impaired blood flow. Red blood cells fall out of the liquid part of the blood in the capillaries and get deposited as a precipitate. Accumulation of red blood cells gradually resolves itself, but as long as it does not need a doctor to alleviate pain and to detect potential co morbidity (Explore Sickle Cell Anemia, 2012).

Parents ought to seek medical help immediately if their child has any of

these symptoms. If the infection in the blood starts early, it is recommended that one starts taking antibiotics to avoid fatal complications. After 5 years of age, when a child is to develop appropriate natural antibodies to the bacteria, the likelihood of fatal bacterial infection is greatly reduced. The problem of school age children with sickle cell disease is usually sporadic blockage of red blood capillaries of large bones. Most of these episodes are relatively mild; there is only slight aching pain in the bones.

Adolescents with sickle cell disease experience anxiety and concern because their physical development is usually delayed by 2-3 years. These teens are usually shorter than their classmates and are often teased for delay in sexual development. However, over time puberty still occurs, and studies show that women with sickle cell disease can carry out their reproductive function normally. Adults with sickle cell disease may show symptoms of chronic (long-term or permanent) clogging of the capillaries of the lungs and kidneys and may develop chronic pulmonary or renal failure. These two complications lead to the early death of some patients with sickle cell disease (Sickle cell anemia, 2011).

Diagnosis of the Disease

Although the symptoms of sickle cell disease rarely occur before 3 months of age, the diagnosis can be made at any age. The fetus of sickle cell disease can be detected by amniocentesis (taking fetal treatment with a needle introduced into the uterus through the abdominal wall). The amniotic fluid contains fetal cells, and the cells contain genes responsible for the production of hemoglobin. Therefore, one can directly examine the genes of the fetus (Schnog, 2004). This testing is very important to prevent fatal

complications that are common in children with sickle cell disease is in the first years of life (Explore Sickle Cell Anemia, 2012).

Treatment and Prevention of Sickle Cell Anemia

Treatment for sickle cell disease does not exist. However, the overall improvement of health services presents the opportunity for longer and healthier lives of children with this disease. This currently focuses on nutrition, extension measures, and prevention of infections. It is important to maintain contact with a primary care physician who is familiar with early signs of complications arising from sickle-cell anemia (Sickle cell anemia, 2011).

Particularly important is the constant medical check-up young children with sickle cell disease to prevent early life-threatening infectious complications. Anemia may worsen as a result of an infection or a sudden increase in the spleen, may require hospitalization to treat the infection or pain. Pregnancy and surgery, as well as diseases of the eye, require the attention of specialists who have experience working with patients suffering from sickle cell anemia.

A vaccine against pneumococcal disease and early treatment with antibiotics has salvaged the lives of scores of young children with sickle cell disease. Weak aching in the bones can be managed at home with rest, plenty of fluids intake and pain medications, over-the-counter. Sometimes these bouts of pain can be severe, and hospitalization of the child may be required to get rid of the pain. Most children with sickle cell disease are normally developed mentally and can succeed in school (Gordon, 2000).

In case of serious respiratory diseases hospitalization is required to provide

adequate levels of oxygen to the patient. New methods of treatment using a laser beams can be applied to patient with clogged retina capillaries. Sickle cell disease cannot be prevented. However, couples with the characteristic of sickle-cell anemia are advisable to seek genetic counseling.

Implications for the Nursing Practice

The prevalence of sickle cell disease necessitates various interventions by nurses to help manage the disease better. One such intervention is managing genetic information in families of children with sickle cell disease. This is achieved by providing tailor made education on the genetic features of sickle cell disease. A recent study by Gallo, Knafl, Angst and Feetham seeks to identify information management models by unfolding parents' knowledge, attitudes, and approaches used in accessing interpreting and relaying information about sickle cell disease to others (2007).

One common practice in the care of sickle cell patients usually involves the red cell exchange programme. This process entails the removal of sickled cells (HbS) while concomitantly transfusing red blood cells from a donor with the aid of an apheresis machine. This strategy sometimes encounters problems in accessing the veins. According to Freires, frequent venous cannulations over several years cause relentless " scarring, thrombosis, infection, stenosis and loss of function of the veins" (2011). This implies that there is a need for better alternatives that are less intrusive than the currently used ones. The accessibility of dual lumen portacaths gives an opening for the nurse-led central venous access devices (CVAD). This leads to a less painful and convenient way of accessing the veins during this crucial procedure. Therefore, this procedure ought to be implemented by all

health facilities to reduce the side effects encountered from the other traditional methods of the red cell exchange programme.

Conclusion

Sickle cell anemia is considered to be a dangerous disease as it has a high mortality rate especially in the early childhood years. This disease is mostly present in Black people especially from the African region. Patients suffering from Sickle cell anemia should be educated regarding their situation, which will enable them to recognize the early symptoms so that they can seek medical aid. Having proper information regarding the disease will decrease the mortality rate as people will be aware of the symptom of the disease and its treatment.

References

Explore Sickle Cell Anemia. (2012). Retrieved October. 5, 2012. from, http://www. nhlbi. nih. gov/health/dci/Diseases/Sca/SCA_All. html.

Freires, M. (2011). Breaking boundaries: A nurse-led portacath insertion service for sickle cell patients. Mary Seacole Award Report. Retrieved from http://www. rcn. org. uk/__.../Mary_Seacole_Award_report_by_2010_winner_Mylene_Freires. pdf.

Gallo, A., Knafl, K., Angst, D., & Feetham, S. (2007). Managing genetic information in families of children with sickle cell disease. Nursing and Health Practices. 9. 3. 249. DOI: 10. 1111/j. 1442-2018. 2007. 00357. x

Gordon, M. A. (2000). Lets talk about sickle cell anemia. New York: PowerKids Press.

Maakaron, E. Joseph (2012). Sickle Cell Anemia. Retrieved October 5, 2012 from http://emedicine. medscape. com/article/205926-

overview#aw2aab6b2b2aa

Passarge, E. (2007). Color atlas of genetics (3rd ed.). New York, NY: Thieme.

Sickle cell anemia. (2011). Retrieved October 5, 2012, from http://www. mayoclinic. com/health/sickle-cell-anemia/DS00324