Sickle cell anemia essay

Literature, Russian Literature

Blood is essential to human survival. This fluid is the transportation system of the body; it delivers all necessary nutrients that cells need in order to function properly. The largest component of blood is comprised of red blood cells. These cells are the bodys principle means of transporting oxygen from the lungs to all body tissues. They are perfectly suited for this function because red blood cells contain hemoglobin, an iron-based protein molecule that binds to oxygen. This bond results in oxygen being circulated as blood travels (Medical and Health Encyclopedia, 1995, p. 411). Blood is essential to human survival.

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Normally, in an average man, there are 5 million red blood cells per cubic millimetre. If this number decreases to 4 million or less, it results in a condition known as anemia. Any abnormalities in size, shape, or hemoglobin content can also account for an anemic state. Many types of this disorder exist, including sickle cell anemia, a disease that is closely related to hemoglobin (Medical and Health Encyclopedia, 1995, p. 35).

What is it? Sickle cell anemia is a disorder that directly affects the circulatory system, or red blood cells in particular. Normally, the body manufactures red blood cells that are disc-shaped. All are smooth and flexible, enabling them to pass easily through the smallest blood vessels. However, the red blood cells of individuals affected by sickle cell anemia are distorted. They take on a sickle shape, also becoming sticky, rigid, and fragile.

The Cause Unlike other cells, red blood cells lack a nucleus and organelles. About 97% of the red blood cells dry content is composed of the macromolecule, hemoglobin. This molecule is key for explaining the sickle cell phenomenon. The DNA required to make hemoglobin involves six genes. Four of these genes code for one half of the molecule, called the alpha chain while the remaining two code for the other half, the beta chain. Each chain is divided into two subunits (or globins) with its own non-protein heme group that is able to bond with one oxygen molecule. Therefore, each hemoglobin molecule can bind to four oxygen molecules in total. These are the characteristics of the normal hemoglobin A.

However, if one of the six genes is altered, forms of hemoglobin other than hemoglobin A will result . The beta-globin gene that is found on the short arm of chromosome eleven is the determinant of sickle cell anemia. A slight alteration in just one nitrogenous base in the DNA sequence coding of this gene is simply all that is necessary to cause a lifetime of medical complications. This change causes a point mutation in the beta-globin chain.

In other words, glutamic acid, an amino acid that normally fills the sixth position in the beta chain, is replaced by another amino acid called valine

(Biology 11, 2002, p. 202). When two mutated beta-globins unionize with the two wild-type (normal) alpha globins, hemoglobin S is created. This variant has different properties than hemoglobin A. Under both oxygenated and deoxygenated conditions, hemoglobin A molecules remain separate from each other, allowing the red blood cells to maintain the same shape. However, even though hemoglobin S allows the red blood cells to retain their normal shape in an oxygenated environment, they become drastically different once they are oxygen-deprived. Subsequently, the abnormal amino acid valine causes a weak attraction force between the hemoglobin S molecules, causing them to polymerize. They form a twisted bundle made up of fourteen strands.

Problems arise because these polymers cause the cell membrane to distort out of their round shape, becoming rigid and sickle-shaped instead. Once the erythrocytes pick up oxygen again, the hazardous attraction force disappears, returning the membrane to its original shape. This cycle of assembling and dissembling quickly deteriorates both the cell membrane and the hemoglobin itself. Although erythrocyte production is increased significantly, it still cannot keep up with the destruction rate of those sickle cells. A normal red blood cell has an average lifespan of about 120 days, while a sickle cell can only live for about 20 days. This reduces blood count drastically.

Inheriting the disease Sickle cell anemia is an autosomal recessive genetic disease, inherited from the parents. This means that the child must receive two sickle cell genes, one from each parent, in order to be fully affected. In

other cases where one sickle cell gene is inherited along with a normal gene, the child has a condition known as sickle cell trait. Sickle cell trait is different from sickle cell anemia. People who have sickle cell anemia are affected by the abnormal hemoglobin SS, while people with sickle cell trait do not.

Most people who have sickle cell trait do not even know it because they never experience the side effects. These people simply have the gene that can cause sickle cell anemia. Therefore, they run the risk of passing the disease down to their own children.

Parents who both have sickle cell trait may produce a child who has a 25% chance of having sickle cell anemia and 50% chance of having the sickle cell trait. The risk increases if either parent has sickle cell anemia. Although anyone can inherit this disease, it is known that the victims most affected have ancestors from the African heritage. Others include those from the Mediterranean, Middle East and India. History and Evolution Scientists and researchers believe that sickle cell anemia evolved many years ago from a spontaneous single mutation in the Hemoglobin gene.

This mutation was found in people originating from Asia and three parts of Africa, where malaria was common. Malaria is a disease caused by protozoan parasites within mosquitoes. Female mosquitoes transfer the parasite into humans with a single bite. This small amount of parasites will multiply into an entire population within the red blood cells. As a result, the red blood cells die and parasites continue to take over, causing pain and symptoms of anemia such as light-headedness and fever.

However, the resistance to this disease would soon be found in the mutated genes. Now, a person with a mutation in one of their hemoglobin genes would be protected from malaria. There are theories developed as to why this occurs: 1.

Parasites in the red blood cell produce an acid and with this acid, Hemoglobin S has a tendency to polymerize causing cells to sickle. Then the sickled cells are destroyed as they pass through the spleen, therefore destroying the parasites as well. 2.

The parasites do not live long in low oxygen levels, and since the oxygen level in the sickled red blood cell and the spleen are low, the parasites die easily. 3. Also, under low oxygen levels, potassium leaks out of the red blood cells containing hemoglobin S. Since parasites need the high level of potassium to survive, they have trouble living in that environment In that case, as people begin to survive malaria, they live on to pass the mutated gene to their children. That is where the sickle cell gene causes the sickle cell disease throughout the world. Today, more than one million people are affected by sickle cell anemia. Symptoms/ Long Term Effects This chronic blood disease indicates one huge, general problem: blockage of blood vessels. As a result, numerous symptoms and complications can result.

Most people experience painful episodes called crises. The following are some common forms of crises: Hemolytic crisis: Occurs when the body cannot produce red blood cells fast enough to replace those destroyed. Since sickle cells have a much shorter lifespan then normal erythrocytes, people with sickle cell anemia are especially susceptible to this crisis. Splenic

sequestration crisis: When sickle cells block the blood vessels leading out of the spleen, blood remains in the organ, unable to exit. The spleen enlarges, sometimes causing pain.

This is a dangerous condition because as more blood becomes trapped, a drop in blood count will take place, leading to other serious implications. Usually after a patient recovers, their spleen would have been damaged beyond repair. Consequently, they will be more prone to infections such as pneumonia. Aplastic crisis: A virus called Parvovirus B19 enters the body, directly preventing the bone marrow from producing blood for two to three days. Though this is a small matter in normal people, it is life-threatening for sickle cell anemia patients. They must immediately be admitted to the hospital and may even need blood transfusions. Vaso-occlusive crisis: Any obstruction in blood vessels leading to an organ that results in an insufficient oxygen supply and damage of tissues. This condition leads to a wide variety of other complications due to the lack of oxygen in organs such as a stroke .

Other symptoms include painful swelling of the hands and feet; pain occurring randomly at any organ or joint; and acute chest syndrome. These all occur due to blockage of blood vessels. Exhaustion, paleness, and shortness of breath, yellowing of skin and eyes, delayed growth and puberty, formation of gallstones and eye damage can also happen because of a deficiency of red blood cells. In addition, victims can also suffer leg ulcers, though the cause is not clear.