Sickle Cell Disease Essay, Research Paper

Sickle cell disease is not contagious. It is a genetically inherited disease that affects the red blood cells. Normal red blood cells are round, but sickled red blood cells are crescent shaped. There are many people living with sickle cell, but it can be fatal. Usually people with the disease die between the ages of 20 and 40 due to organ complications.

Red blood cells contain a special protein called hemoglobin (Hb) that carries oxygen from the lungs to all parts of the body. Hemoglobin is produced in the bone marrow. This chemical substance is what gives red blood cells their color. The main hemoglobin in normal red blood cells is hemoglobin A (HbA). People with Sickle Cell Disease have Sickle hemoglobin (HbS)(General, 1). Normally, red blood cells live for about 120 days before new ones replace them, but sickled red blood cells only live for about 16 days.

Normal red blood cells can bend and flex easily to pass through veins. When sickle hemoglobin gives up its oxygen to the tissues, it sticks together to form long rods inside the red blood cells making these cells distorted and rigid. The red cells take on the appearance of the C-shaped farm tool called a sickle. Because of their shape, sickled red blood cells can t squeeze through small blood vessels and may get stuck. This will cause the cells to pile up and block the blood vessel, which stops the oxygen from getting to where it is needed(Sickle, 2-3). Without oxygen, the area becomes inflamed and may cause severe pain to the patient.

Sudden outbursts of pain in sickle cell patients are called crises. These episodes can range from a mild attach lasting for only a few minutes to a severe pain lasting days or weeks requiring hospitalization. Pain-killing drugs are sometimes used to treat severe crises. Most pain occurs in the arms, legs, back, and stomach. Thirst, overexertion, cold weather, cold drinks, and swimming may cause the pain in children, while stress triggers the crises in adults. Swelling of the hands and feet can also occur during blood vessel blockage.

If the blockage is long lasting, it can destroy areas of tissue. Damage to vital organs may also result. Sickle cells tend to become trapped and destroyed in the liver and in the spleen. This results in a shortage of red blood cells called anemia which, when extreme, can cause the patient to be pale, short of breath and easily tired. Certain conditions, including infections, may worsen a patient s anemia by speeding up the destruction of red blood cells or reducing red blood cell production(What, 2).

Bacterial infection is one of the main causes of mortality for patients with sickle cell disease. Young children are especially at risk, but patients of any age can suffer a rapid death from sepsis. The spleen’s function is markedly decreased in sickle cell patients, leaving the person at high risk for salmonella, meningococcus, and streptococcus pneumonia. More serious infections such as meningitis, pneumonia, and osteomyelitis must be treated empirically early to prevent death(Sickle, 2).

Another common symptom of Sickle Cell is Jaundice. Jaundice occurs when there is excessive disintegration of red blood cells as in anemia. It is an abnormal condition in which body fluids and tissues, particularly the skin and the eyes, take on a yellowish color(Frequently, 4). This symptom is not a concern unless it noticeably worsens. However, this condition may indicate upcoming crises.

All sickle cell patients should be under the care of a medical team that understands sickle cell disease. Newborn babies detected with sickle cell disease should be placed on daily penicillin to prevent serious infections. Getting plenty of rest, drinking lots of water, avoiding extreme temperatures, and taking the vitamin folic acid daily are some ways to keep a sickle cell patient healthy. Fevers, chest pain, shortness of breath, abdominal swelling, unusual headache, and sudden vision changes are some of the conditions that should be watched for in the patients. These symptoms need urgent medical evaluations.

Sickle cell occurs mainly among Africans and people of African descent. About 1 in every 12 African Americans carries the gene for the sickle cell trait and about 1 in every 400 600 American blacks is born with the disease. Africans are not the only ethnic group that can develop sickle cell disease. Arabs, Greeks, Italians, Latin Americans, and Native Americans are also effected by sickle cell. An estimated 70,000 Americans of different ethnicity have sickle cell disease(Information, 1). It is recommended that all races should be screened at birth.

Most states now perform a sickle cell test when babies are born. It is a simple blood test called the hemoglobin electrophoresis. This test can be done by a doctor or a local sickle foundation. Because the HbS and the HbC amino aid substitutions change the electrical charge of the protein, the migration pattern of the hemoglobin with electrophoresis results in diagnostic patterns with each of the different hemoglobin variants(Sickle, 1-2).

Because sickle cell disease is an inherited blood disease, the only way for a person to acquire it is from their parents. The conditions are inherited from parents much the same way as blood type, hair color, and other physical traits. Every person has 2 copies of the hemoglobin gene, one from their mother and one from their father. To have sickle cell you must receive the sickle cell gene from both parents. If only one gene carries sickle cell, then that person has a sickle cell trait. People with a sickle cell trait are not said to have the disease. They do not experience painful episodes and are generally not affected by the sickle hemoglobin. They cannot later develop sickle cell disease. However, when two carriers have a child, their child may inherit two sickle cell genes and have the disease.

Sickle cell anemia is one type of disease that can result from sickle cell. There are other types of sickling disorders as well. Sickle C disease is also a common form of sickle cell. It is caused by a substitution of lysine in the 6th amino acid position of the beta globin chain of hemoglobin from both parents. Sickle beta thalassemia is a less common form of sickle cell disease. This variant is caused by genetic mutations that abolish or reduce production of the beta globin subunit of hemoglobin(What, 2). The effects of these diseases vary greatly from one person to the next. Some affected may rarely see their doctors for sickle cell-related complaints, while others may be frequently hospitalized.

For now, no medicines exist to effectively treat sickle cell anemia. Treatments usually aim to prevent complications. Crises are treated with

painkillers, fluids, and oxygen. Folic acid and a B vitamin are also used to help patients. They should also be vaccinated against pneumonia because sickle cell patients are prone to getting it(Frequently, 1). The only way to prevent sickle cell disease is to find out whether or not you carry the genes for the disease before getting pregnant.

After conception, sickle cell anemia can be diagnosed by amniocentesis in the second trimester of pregnancy. This test involves placing a needle in to the womb through the women s abdomen and obtaining a small sample of the amniotic fluid. The amniocentesis test carries a small risk of causing a miscarriage, about 1 in 100, but other complications are very unusual(Amniocentesis, 1). If the fetus has sickle cell, the parents may choose to terminate the pregnancy.

Chorionic Villus Sampling (CVS) can be carried out earlier then amniocentesis. This test is usually taken during the 9th or 10th week of pregnancy. It takes a small amount of material from the developing placenta. This test can be done by placing a thin tube through the cervix for a sample or by placing a needle in the womb and using an ultrasound scanner to guide it. Though CVS is usually the technique used to determine sickle cell, it has more risks than amniocentesis(Chorionic, 1). This is a relatively new test and there is not as much information on reliability either.

If a couple does decide to terminate their pregnancy after either an amniocentesis or a CVS test, this should not affect their fertility in future pregnancies. The couple may wish to be referred to a genetic counselor to discuss the risk of an affected baby next time. Together they can comprehend the medical facts and understand the available alternatives for dealing with the risk of recurrence. Then they can choose the course of action that seems to fit best to their family goals, ethics, and religion. Finally the counselor can help the couple seek appropriate medical and educational care.

Although there is no known universal cure for Sickle Cell Disease, there have been new advancements that are known to cure the disease. A few children with sickle cell disease have been cured through a bone marrow transplant. The transplant used bone marrow from a sibling with similar genetic makeup. However, this cure carries a high risk. About 5 to 8 percent of children who undergo bone marrow transplants for severe hemoglobin disorders die. This new form of treatment may not be suited for all persons with sickle cell.

Chemotherapy drugs have also been known to cure sickle cell. A number of other new therapies, including hydroxyurea, for reducing the severity and frequency of complications of the disease are also being tried(Sickle, 2). Limiting the damage to the organs through medical care also improves the survival and the quality of life for many affected people. Scientists say that gene therapy may someday have a cure at less risk as well. Many organizations, such as the March of Dimes, have been major supporters of sickle cell disease research.