Sickel Cell Anemia Essay, Research Paper

Sickle Cell anemia is a group of inherited red blood cell disorders, or a collection of recessive genetic disorders characterized by a hemoglobin variant called Hb S. Normal red blood cells are round like doughnuts, and they move through small blood tubes in the body to deliver oxygen. Sickle red blood cells become hard, sticky and shaped like sickles used to cut wheat. When these hard and pointed red cells go through the small blood tube, they clog the flow and break apart. This can cause pain, damage and a low blood count, or anemia. There is a substance in the red cell called hemoglobin that carries oxygen inside the cell. One little change in this substance causes the hemoglobin to form long hard rods in the red cell when it gives away oxygen. These rigid rods change the red cell into a sickle shape.

For such a miniscule mistake, the consequences are tragic. At the time of conception, a person receives one set of genes from the mother and a corresponding set of genes from the father. Sickle cell disease is a condition that is determined by a single pair of genes. The genes are those which control the production of hemoglobin in red cells. It is a member of the globin gene family, a group of genes involved in oxygen transport, and hemoglobin binds oxygen in the lungs and delivers it to the other tissues. Most people have two normal genes for hemoglobin. Some people carry one normal gene and one gene for sickle hemoglobin. This is called “sickle cell trait”.

These people are normal in almost all respects. Problems from having a single sickle cell gene develop only under very unusual conditions. People who inherit two genes for sickle hemoglobin (one from each parent) have sickle cell disease. It is believed that individuals with African and Mediterranean ancestry have unusually high frequency of sickle cell trait due to the reduced mortality from malaria infections when compared with individuals who do not carry the hemoglobin variant

Red blood cells carrying the abnormal gene (hemoglobin S) travel normally through circulation until they are deoxygenated. When this happens the hemoglobin S molecules form long, rigid rods, causing the normally donut-shaped cells to stiffen and distort into a sickle shape.

These sickle cells have a very hard time moving through the small capillaries. As blood flow slows, the cells lining the vessel walls become sticky, attracting the sickle cells like flypaper and causing massive circulatory gridlock. Although these changes are partly reversible through the normal process of reoxygenation, by the time this reversal takes place much damage has already been done. The spleen, acting as a policeman, traps and destroys many of the abnormal sickle cells, resulting in rapid turnover of red blood cells and chronic anemia.

This anemia results in fatigue and a number of the following problems;pain episodes, strokes, susceptibility to bacterial infections, particularly in children, leg ulcers, bone damage, yellow eyes or jaundice, early gallstones, lung blockage, increased infections, kidney damage and loss of body water in urine, painful erections in men, blood blockage in the spleen or liver, eye damage, low red blood cell counts (anemia), and delayed growth.

Each year in the US, an average of 75,000 hospitalizations are due to sickle cell disease, costing approximately $475 million. Sickle cell disease is also associated with significant mortality. Among children, the primary causes of mortality are bacterial infections and stroke. In adults, it is more difficult to attribute specific causes to mortality, but it appears that individuals with more indicative disease are at risk for early mortality.

Tremendous advances in detection and treatment mean that most patients now survive to adulthood, many into their 50s and 60s and beyond. Some patients lead fairly normal lives, attending school and work with only occasional pain episodes and only slowly progressive organ damage. Treatments, such as penicillin prophylaxis, have been developed that can significantly reduce the rate of disease and mortality of sickle cell disease patients. For this reason, several US organizations have supported screening all newborns for sickle cell disease. As a result, almost every US state and territory now screen their newborn infants for this blood disorder. Several test methods are available to detect sickle cell disease. Most tests examine an individual?s hemoglobin, although DNA testing is also now available. As a result of newborn screening, better medical care, parent education and penicillin prophylaxis, the mortality rate due to sickle cell is decreasing

There are several test methods available. These tests detect the beta globin gene product and are performed on blood samples, including cord blood and dried blood spots, which are collected at any time following birth. DNA testing can also be performed. DNA samples can be collected either prenatally, or postnatally, may be used for DNA testing.

The Agency for Health Care Policy and Research (AHCPR) has recommended hemoglobin electrophoresis, isoelectric focusing, and high-performance liquid chromatography as accurate methods for newborn screening. They state that DNA analysis may also be used, but that it is costlier than the other methods. AHCPR has also recommended that all diagnostic laboratories participate in quality assurance and proficiency testing programs, regardless of the type of test they perform. The Centers for Disease Control and Prevention currently conducts quality assurance evaluations of state newborn screening programs.

Tests used in the US however, may not be cost-effective for sickle cell diagnosis in other developing countries. In Kenya, another method, peripheral blood film (PBF) has been confirmed to be the most cost-effective diagnostic method. The sensitivity of PBF is 76%, and specificity is around 99.7%.

Here are a few key history notes. In 1972, the congress passed the National Sickle Cell Anemia Control Act, which called for screening programs. In 1975, the first US state began a newborn screening program for sickle cell disease. However, it was the late 1980?s before most states were performing sickle cell screening on newborns. This was most likely due to the publication of a study in 1986 that showed that oral penicillin could significantly reduce the rate and mortality of the disease in children.

In 1987, the National Institutes of Health held a conference that supported early diagnosis by newborn screening as being beneficial to infants with sickle cell disease. In 1993, another US agency, the Agency for Health Care Policy and Research (AHCPR), also concluded that newborn screening would significantly reduce mortality and problems in infants. AHCPR further recommended that ?all infants should be tested for sickle cell disease, regardless of race (universal screening) since targeting high risk racial or ethnic groups would not identify all affected infants due to the inability to reliably determine the infants? race by appearance, name or self-report.?

After screening there are several things a person can do to help slow and prevent complications. Take the vitamin folic acid daily helps make new red cells, along with iron. Daily penicillin until age six can prevent serious infections. Keep you body well hydrated, drinking plenty of water daily (8-10 glasses for adults). Avoid too hot or too cold temperatures, over exertion and stress. Get plenty of rest, and most important, get regular check-ups from knowledgeable health care providers.

Patients and families should watch for the following conditions that need urgent medical evaluation: fever, chest pain, shortness of breath, increasing tiredness, abdominal swelling, unusual headache, any sudden weakness or loss of feeling, pain that will not go away with home treatment, and sudden vision change. Following all the previous advice should allow anyone with the disease to lead a fairly normal life.