

Sickle Cell Anemia: An Inherited Blood Disease

Background:

- Sickle cell anemia has several alternative names, such as Hemoglobin SS disease (Hb SS) and Sickle cell disease (Medline Plus, 2005).
- Sickle cell anemia is an inherited disease in which the red blood cells, normally disc-shaped, become crescent shaped. As a result, they function abnormally and cause small blood clots. These clots give rise to recurrent painful episodes called "sickle cell pain crises". There are several types of crises (Medline Plus, 2005).
- Hemolytic crisis occurs when damaged red blood cells break down (Medline Plus, 2005).
- Splenic sequestration crisis is when the spleen enlarges and traps the blood cells (Medline Plus, 2005).
- Aplastic crisis results when an infection causes the bone marrow to stop producing red blood cells (Medline Plus, 2005).
- Repeated crises can cause damage to the kidneys, lungs, bones, eyes, and central nervous system (Medline Plus, 2005).
- Sickle cell anemia is caused by an abnormal type of hemoglobin called hemoglobin S. Hemoglobin is a protein inside red blood cells that carries oxygen (Medline Plus, 2005).
- Hemoglobin S, however, reduces the amount of oxygen inside the cells, distorting their shape. The fragile, sickle-shaped cells deliver less oxygen to the body's tissues, and can break into pieces that disrupt blood flow (Medline Plus, 2005).
- Sickle cell anemia is inherited as an autosomal recessive trait , which means it occurs in someone who has inherited hemoglobin S from both parents (Medline Plus, 2005).
- Someone who inherits hemoglobin S from one parent and normal hemoglobin A from the other parent will have sickle cell trait. Someone who inherits hemoglobin S from one parent and another type of abnormal hemoglobin from the other parent will have another form of sickle cell disease, such as thalassemia (Medline Plus, 2005).
- Common symptoms include: paleness, yellow eyes/skin, fatigue, breathlessness, rapid heart rate, delayed growth, puberty susceptibility to infections, ulcers on the lower legs (in adolescents and adults), jaundice bone pain, attacks of abdominal pain, and fever (Medline Plus, 2005).
- Patients with sickle cell disease need continuous treatment, even when they are not having a painful crisis. Supplementation with folic acid, an essential element in producing cells, is required because of the rapid red blood cell turnover (Medline Plus, 2005).
- The purpose of therapy is to manage and control symptoms and to try to limit the frequency of crises (Medline Plus, 2005).
- Bone marrow transplants can be curative, this therapy is indicated in only a minority of patients, predominantly because of the high risk of the

procedure (the drugs needed to make the transplant possible are highly toxic) and difficulty in finding suitable donors. Also, bone marrow transplants are much more expensive than other treatments (Medline Plus, 2005).

- Antibiotics and vaccines are given to prevent bacteria infections, which are common in children with sickle cell disease (Medline Plus, 2005).

Statement of the Problem:

- Scientists have learned a great deal about sickle cell anemia during the past 30 years - what causes it, how it affects the patient, and how to treat some of the complications. They also have begun to have success in developing drugs that will prevent the symptoms of sickle cell anemia and procedures that should ultimately provide a cure (ASCAA, 2000).
- Hydroxyurea appears to work primarily by stimulating production of fetal hemoglobin. There is some evidence that administering hydroxyurea with erythropoietin, a genetically engineered hormone that stimulates red cell production, may make hydroxyurea work better. This combination approach offers the possibility that lower doses of hydroxyurea can be used to achieve the needed level of fetal hemoglobin. However, both of these drugs may produce serious side effects, so researchers continue to search for safer agents that are just as effective (ASCAA, 2000).
- Clotrimazole, an over-the-counter medication commonly used to treat fungal infections, is under investigation as a treatment to prevent the loss of water from the red blood cells that contributes to sickling. It is hoped that this medication, used alone or in conjunction with other anti-sickling agents, may eventually offer an effective long-term therapy for sickle cell anemia patients (ASCAA, 2000).
- The ultimate cure for sickle cell anemia may be gene therapy. In sickle cell anemia, the gene that switches on production of adult hemoglobin shortly before birth is defective. Two approaches to gene therapy are being explored. Some scientists are looking into whether correcting this gene and inserting it into the bone marrow of people with sickle cell anemia will result in the production of normal adult hemoglobin. Others are looking at the possibility of turning off the defective gene and simultaneously reactivating another gene that turns on production of fetal hemoglobin. In both cases, the research is at a very early stage. Progress is being made, however, and there is a real possibility of an eventual clinical cure for sickle cell anemia (ASCAA, 2000).
- Although the genetic defect that causes sickling was identified more than 40 years ago, until very recently, research into the development of treatments for the disease was hampered by the lack of an animal model that could be used to test experimental drugs and gene therapy. Recently, however, scientists were able to genetically engineer a line of mice that exhibit some of the characteristics of sickle cell disease in much the same way humans do. This is an important advance in the search for an effective treatment and eventual cure for sickle cell disease (ASCAA, 2000).

Epidemiologic Picture of the Problem:

- The sickle cell disorders are found in people of African, Mediterranean, Indian, and Middle Eastern heritage. In the United States, these disorders are most commonly observed in African Americans and Hispanics from the Caribbean, Central America, and parts of South America (Black Health Care, 1999).
- As populations migrated, the sickle cell-mutation spread to other Mediterranean areas, further into the Middle East and eventually into the Western Hemisphere. In the United States and other countries where malaria is not a problem, the sickle hemoglobin gene no longer provides a survival advantage. Instead, it may be a serious threat to the carrier's children, who may inherit two abnormal sickle hemoglobin genes and have sickle cell anemia (ASCAA, 2000).
- Approximately 1 out of every 500 African Americans is affected by sickle cell disease (Medline Plus, 2005).
- Approximately 1 out of every 1200 Latinos is affected by sickle cell disease (Insel and Roth, 2006).
- In this country, it affects approximately 72,000 people, most of whose ancestors, come from Africa (ASCAA, 2000).
- Although sickle cell disease is present at birth, symptoms usually don't occur until after 4 months of age (Medline Plus, 2005).
- In the past, death from organ failure often occurred between the ages of 20 and 40 in most sickle-cell patients. More recently, because of better understanding and management of the disease, patients live into their forties and fifties (Medline Plus, 2005).
- Sickle cell trait might have developed in tropical regions as an adaptation to the widespread presence of malaria (Insel and Roth, 2006).
- Sickle cell anemia can only result when two carriers with sickle cell trait have a child together. Therefore, genetic counseling is recommended for all carriers of sickle cell trait (Medline Plus, 2005).
- Approximately 2 million Americans, or 1 in 12 African Americans, and 1 in 16 Hispanic Americans carry the sickle cell trait (ASCAA, 2000).

Solutions to the Problem:

American Sickle Cell Anemia Association-

www.ascaa.org

The American Sickle Cell Anemia Association (ASCAA) is a private nonprofit organization in Cleveland, Ohio. The ASCAA was founded in 1971 and is the oldest sickle cell research, education, and social services organization in the United States. Within the Ohio Department of Health, the ASCAA is designated as Region V and comprises Cuyahoga, Geauga, Lake, Medina and Lorain counties. Region V has the highest incidence of sickle cell disease and

variants of the disease in the state. The organization currently provides a wide range of services to those individuals and families with either sickle cell trait or variants of the disease itself. Key services include: ongoing follow-up diagnostic testing, counseling, and tracking services for parents with infants who screen positive by the State Laboratory; Family Counseling and Support Services; coordination of medical, social services, education and support for the program's clientele; teacher education and screening services delivery at upwards of 75 local health fairs. In addition, ASCAA makes outreach to the region's African American, Hispanic, Mediterranean, and Arab communities for family education and the identification of the incidence of sickle cell disease. Since its inception, the organization has tested approximately 100,000 at-risk individuals and has been a repository of statistical, case information, and data beneficial to patients, affected family members and the medical established locally and globally.

Sickle Cell Society-

www.sicklecellsociety.org

The Society provides a wide range of services from information, advice and counseling, to financial help, holidays, briefings, seminars and training. The Society is committed to ensuring that sufferers benefit directly from voluntary donations that make up the welfare fund. Therefore, the Society has a special welfare fund to assist individuals and their families in need of extra help. Thousands have benefited since the fund was first set up in 1982, getting extra help with heating bills during the cold weather, money for warm clothing, and much more. Many people who suffer from a sickle cell disorder find their education constantly disrupted by illness. As a result, they often leave school without qualifications, and need extra help to obtain the qualifications they need. The Bryan Jones Fund now provides that help, offering financial support for sickle cell sufferers who wish to further their education. Each year, through its Children's Holiday Fund, the Society provides a much-needed holiday for 30 children with sickle cell disorders. Many of the children come from low-income families, and would otherwise not have a holiday.

Sickle Cell Center-

<http://www.cincinnatichildrens.org/>

The experts at the Cincinnati Children's Comprehensive Sickle Cell Center provide care for all the routine problems and complications of sickle cell disease. In partnership with families and the community, we try to maximize the growth and development of each patient. Through medical treatment, psychological evaluation, social services, education, counseling, research and working collaboratively with families, they are dedicated to prolonging and improving the lives of our patients. Dr. Marilyn Gaston, the Sickle Cell Awareness Group, concerned parents and hospital administration established the Comprehensive Sickle Cell Center (CSCC) at Cincinnati Children's in 1971. The center created one of the first newborn screening programs for sickle cell in the United States.

Doctors at Cincinnati Children's performed the first bone marrow transplant to cure sickle cell disease. To prevent the complication of acute chest syndrome, researchers at Cincinnati Children's developed incentive spirometry, which is the standard of care across the country. The CSCC provides important leadership in the National Sickle Cell Disease Program and plays a significant role in advancing sickle cell disease research and patient care.

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(Class Hand-out)

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