WHAT IS SICKLE CELL DISEASE?

Sickle Cell Disease (SCD), or sickle cell anemia, is an inherited blood disorder that affects the primary oxygen-carrying component of blood, the hemoglobin in red blood cells (erythrocytes). Red blood cells contain a protein-iron compound called hemoglobin (Hgb). When blood flows through the lungs, the hemoglobin picks up oxygen. Blood is then pumped by the heart to the body, where the hemoglobin releases the oxygen to the tissues. Except for the first few months of life, when fetal hemoglobin persists in all infants, normal red blood cells contain hemoglobin A. Persons with sickle cell disease have mostly Hemoglobin S, which is abnormal. Red blood cells containing Hemoglobin S do not live as long as those with Hgb A [about 16 (10-20) days vs. 90-120]. This leads to a shortage of red blood cells, a condition called anemia.

Normal red blood cells maintain a discoid (like a donut without a hole) shape even when they have released their oxygen. When "sickle" red blood cells give up their oxygen they become sickle, or crescent shaped. These cells do not move easily through the blood vessels. They tend to get stuck and block the flow of blood. When this happens, the blood supply to the tissues and organs is reduced. Eventually this causes the complications of sickle cell disease, such as fatigue, breathlessness, rapid heartbeat, delayed growth, susceptibility to infections, skin ulcers, and vision problems.

Sickle cells are destroyed rapidly in the body, causing anemia, jaundice and the formation of gallstones.

The sickle cells also block the flow of blood through vessels resulting in lung tissue damage (acute chest syndrome), pain episodes (arms, legs, chest and abdomen), stroke and priapism (painful prolonged erection). It also causes damage to most organs including the spleen, kidneys and liver. Damage to the spleen makes sickle cell disease patients, especially young children, easily overwhelmed by certain bacterial infections.

HOW DO YOU GET SICKLE CELL DISEASE?

The only way you can get sickle cell disease or sickle cell trait is by inheritance. You cannot get sickle cell disease by contact or by blood transfusions. If a person inherits one normal and one sickle hemoglobin gene, they have sickle cell trait; if one inherits two sickle hemoglobin genes one has the disease (sickle cell anemia).

HOMOZYGOUS AND HETEROZYGOUS

Genes are like blueprints or formulas which the body follows to make substances, including hemoglobin. People who have inherited two copies of the gene for Hgb S (one from each parent) are homozygous and have sickle cell disease.

They cannot make normal (A) hgb. People who have inherited only one copy of the gene for Hemoglobin S have Sickle cell trait. They also have one gene for Hgb A and can make normal Hgb. People with sickle cell trait usually do not manifest any of the problems associated with sickle cell anemia. Rarely, people with sickle cell trait can exhibit sickling of red cells under conditions of extreme physical stress or low atmospheric oxygen.

A person with sickle cell disease can also have other hemoglobin abnormalities, which increase(s) the severity of their disease.

Inheritance

- Unaffected persons have two copies of the gene for Hgb A (AA). Persons with the sickle cell trait have one copy of each gene (AS). Persons with the disease have two copies of the Hgb S gene (SS).
- If one parent has Sickle Cell Anemia (SS) and the other is unaffected (AA), all children will have sickle cell trait (AS). If one parent has Sickle Cell Anemia (SS) and the other has Sickle Cell Trait (AS), there is a 50% chance (or 1 out of 2) of having a baby with either sickle cell disease (SS) or sickle cell trait (AS) with each pregnancy. There is no possibility of unaffected offspring.
- When both parents have Sickle Cell Trait, they have a 25% chance (1 of 4) of having a baby with sickle cell disease with each pregnancy. Thus it is possible that ALL children will be unaffected, or ALL children will have the trait, or ALL will have the disease.
- If neither parent has the trait, no children will have the trait or the disease.
GEOGRAPHIC DISTRIBUTION

The disease originated in at least 4 places in Africa and in the Indian/Saudi Arabian subcontinent. It exists in all countries of Africa and in areas where Africans have migrated. It is also common in the Latino population.

The transatlantic slave trade was largely responsible for introducing the sickle cell gene into the Americas and the Caribbean. However, sickle cell disease had already spread from Africa to Southern Europe by the time of the slave trade, so it is present in Portuguese, Spaniards, French Corsicans, Sardinians, Sicilians, mainland Italians, Greeks, Turks and Cypriots.

Sickle cell disease appears in most of the Near and Middle East countries including Lebanon, Israel, Saudi Arabia, Kuwait and Yemen. The condition has also been reported in India and Sri Lanka. Sickle cell disease is an international health problem and truly a global challenge.

In the United States, about 1,000 babies are born with sickle cell disease each year.

IS THERE A CURE?

Only a closely matched sibling stem cell transplant offers a cure for sickle cell anemia, but these procedures are usually reserved for those under 16 years old. Hydroxyurea is used to prevent pain episodes (“crises”).

Health maintenance for patients with sickle cell disease starts with early diagnosis, preferably in the newborn period (all states and the District of Columbia include sickle cell testing in newborn screening) and includes penicillin prophylaxis, vaccination against pneumococcus bacteria and folic acid supplementation. Treatment of complications often includes antibiotics, pain management, intravenous fluids, blood transfusion and surgery as well as psychosocial support. Blood transfusions help benefit sickle cell disease patients by reducing recurrent pain crises, risk of stroke and other complications. Because red blood cells contain iron, and there is no natural way for the body to eliminate it, patients who receive repeated blood transfusions can accumulate iron in the body until it reaches toxic levels. It is important to remove excess iron from the body, because it can gather in the heart, liver, and other organs and may lead to organ damage. Treatments are available to eliminate iron overload.

HOW DO YOU KNOW IF YOU HAVE SICKLE CELL TRAIT?

The way to find out if you have the trait is a blood test, such as Hgb electrophoresis or chromatography. For University of Illinois students, Hgb electrophoresis is available at McKinley Health Center.

If you have sickle cell trait and wish to become pregnant, you and your partner should have genetic counseling to determine your risks for having a child with sickle cell trait or disease.

References

- American Sickle Cell Anemia Association at: http://www.ascaa.org/
- Sickle Cell Disease Association of America at: http://www.sicklecelldisease.org
- www.sicklecellinfo.net/info_links/sc_centers.htm lists sickle cell disease clinics by location.