

Thao tran



Thao Tran Prof. Thomas Hematology 09/08/2012 Sickle Cell Disease Blood has been considered the essence of life for centuries, and our blood is a living tissue made up of liquid and solids. There are many diseases related to blood such as anemia which is the decrease in oxygen supply to tissue and causes hypoxia, thalassemia is the mutation in one or more globin gene of hemoglobin, etc. Therefore, one of blood disorders is the most common symptomatic hemoglobinopathy with the greatest prevalence in worldwide which is called sickle cell disease. Sickle cell disease is popular in tropical Africa, Mediterranean, the Middle East, India, Nepal, and in geographic regions where there has been migrations from endemic area such as North, Central and South America. According to Center for Disease Control, sickle cell disease affects from 90, 000 to 100, 000 people in United States and causes death among Black or Africa-American children from 4 years of age by 42% from 1999 through 2002. ([www.cdc.gov](http://www.cdc.gov)). It is interesting to note that the areas with the highest frequency of sickle cell disease also have infection with *Plasmodium falciparum* that causes malaria in human. In the United States, there is an estimated population of 270 millions about 1, 000 babies are born with sickle cell disease each year. It means there are 1-2% of all babies having this disease in the total of 25% of people with sickle cell disease. Moreover, sickle cell disease had already spread from Africa to American by the time of slave trade. Sickle cell anemia is caused by an abnormal type of hemoglobin called hemoglobin S due to the replacement of a non-polar valine amino acid for a polar glutamic acid at the seventh position of beta chain. This substitution produces a net of decrease in negative charge; thus, it changes the molecule's electrophoretic mobility. Hemoglobin S distorts the shape of red blood cells, especially when exposed

to low oxygen levels. The distorted red blood cells are shaped like crescents or sickles. These firm, sickle-shaped cells deliver less oxygen to the body's tissues. They can also clog more easily in small blood vessels, and break into pieces that block the blood flow. Our bodies make healthy hemoglobin known as hemoglobin A, and people with sickle cell anemia produce hemoglobin S instead. (Clinical Lab Hematology, 216-217). Sickle cell disease is a serious condition in which erythrocytes become sickle shaped like C. They are stiff and tend to form clumps, so they cannot be easy to move in the blood vessels. This disease is considered an inheritance because the sickle cell gene is passed from one generation to other generation which is called autosomal recessive inheritance. In the other term, it means that both parents must pass on the defective form of gene to their children, and this disease occurs on the family tree by parents who have sickle cell trait. The gene involved in this disease control the production of hemoglobin, which binds to oxygen in the lungs and delivers to the peripheral tissues, such as liver and muscles. Sickle celled people carry one normal and one gene for sickle cell disease, and it is developed as people get older. People cannot lose their sickle cell genes at any period of time, but the severity of sickle cell disease can change over time. The first clinical signs of this disease appear at about 6 months of age when the concentration of hemoglobin S predominates over hemoglobin F. The sickle cell disease is also associated with hemolytic anemia, vaso-occlusions of the microvasculature, overwhelming infection, and acute splenic sequestration. (Clinial Lab Hematology, 218) As we can see in two pictures above, the picture on the left is a normal red blood cell, and the picture on the right is sickle cell. The erythrocytes change their shapes because of lacking oxygen affinity. They

become longer and curved. Sickle cells get stuck in blood vessels and prevent blood from reaching the internal organs because of their poor deformability. Nevertheless, they block all of the blood vessel in the arms, legs, chest or abdomen and cause a strong pain crisis. Especially in children who get sickle cell disease, they might get bacterial infection and inflammation by Streptococcus pneumonia and Hemophilus influenza since their spleen is damaged. One of spleen's main jobs is to protect against infection. It keeps neutrophils from relocating to areas of inflammation. (Clinical Lab Hematology, 219). Besides that, if there is the presence of sickle cell in brain, it can cause stroke due to the brain dies. Stroke affects about one in every ten children with sickle cell disease. In agreement with the Cooperative Study of Sickle Cell Disease, the individuals homozygous for hemoglobin S have higher leukocyte and platelet counts than normal people, especially for children less than 10 years of age. ([www. biolincc. nih. gov](http://www.biolincc.nih.gov)). Moreover, The blood smear shows variable anisocytosis with polychromatophilic macrocytes and poikilocytosis with the presence of sickle cells and target cells. (Clinical Lab Hematology, 220) There are many signs and symptoms associated with sickle cell disease such as swollen hands or feet, sudden paleness of the skin or nail beds, yellow color of the skin or eyes, fever or signs of infection, swelling in the abdomen, sudden tiredness with no interest in what is going on, erection of the penis for a long time, trouble hearing or seeing, weakness on one side of the body, sudden change in speech, headache, trouble breathing, and/or joint, stomach, chest, muscle pain or limping. ([http://www. nih. gov](http://www.nih.gov)). Associating with those signs and symptoms, sickle cell disease also causes many complicated disease, and they can lead to death. For example, stroke, acute chest syndrome,

organ damage, blindness, gallstone, and ulcers. Stroke is one of the most serious complications of disease since sickle cells block the blood flow to the brain. Early diagnosis of sickle cell anemia is very important so that children who have the condition can get proper treatment. Screening tests for sickle cell anemia and sickle cell trait are done on newborn infants in most States. One blood test used to diagnose sickle cell anemia looks at how hemoglobin moves in an electric field. This is called electrophoresis, and it's usually used to diagnose older children and adults. | ï· | Effective treatments are available to help relieve the symptoms and complications of sickle cell anemia, but in most cases there's no cure. (Some researchers believe that bone marrow transplants may offer a cure in a small number of cases.) | ï· | Sickle cell anemia is treated with medicines, blood transfusions, and other treatments specific to certain complications. |