

# [Sickle cell disease essay sample](https://assignbuster.com/sickle-cell-disease-essay-sample/)

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Introduction

Sickle cell is one of the most prevalence genetic diseases in the United States. The disease affects more than 70, 000 people and it is estimated than more than 2 million people carry the trait for the disease.  Like many disease like Diabetes, high blood pressure and others, sickle cell has shown ethnic variability in epidemiological studies. Although the disease affects millions of people in the world, there are some races and ethnic groups that appear to be at an increased risk. In American, epidemiological studies have shown that African Americans are at a higher risk than others. Statistics in the world also confirm that people with sub-Saharan origin are at a higher risk of contracting the disease as compared to the rest of the population. Sickle cell is a disease that affects people with ethic backgrounds in the United States making the disease is most common in African Americans.

Prevalence of Sickle cell anemia in African Americans

There are more than 70, 000 Americans with sickle cells condition while another 2 million Americans have carrier traits for the condition. However epidemiological studies all over the world have revealed that the disease is prevalent among African Americans, and in Indians or Saudi Arabians. In world wide epidemiological studies, the disease is prevalent in West and Central Africa where the rate of prevalence is sometimes as high as 25%. In this region 1-2% of the all children are born in a form of sickle cells condition.

Epidemiological studies in the United States show that in a population of more than 280 million people, 1, 000 babies are usually born with sickle cell condition. The prevalence is however higher in African in a population of around 90 million people in Nigeria, around 45, 000 -90, 000 babies are born with sickle cell condition every year. In the United States, 1 in every 500 African Americans birth has a sickle cell condition. In total 2 million Americans or approximately 1 in every 12 African Americans is a carrier of the sickle cell trait. (SCDAA, 2009)

Following these statistics, research has indicated that transatlantic slave trade can be held responsible for introduction of the gene to America and the Caribbean region.   However, this is usually controversial as the condition had already spread to other continent even before the slave trade (SCDAA, 2009).

Sickle cell anemia

Sickle cell is an inherited blood disorder which mainly affects the red blood cells.  Patients exhibiting symptoms of the disease have blood cells with hemoglobin S, which can be described as abnormal hemoglobin.  In most cases, the red blood cells assume a sickle shape or a crescent shape and exhibit difficulty in traveling around the blood systems through blood vessels. The abnormal hemoglobin is C-shaped, appearing like a sickle and it is also very stiff (Sickle Cell Disease Association of America, Inc 2008).

Hemoglobin is the substance that makes up the red blood cells. The red blood cell is vital in the body as it assists in the transport of oxygen in the body organs.  Hemoglobin assists the red blood cell to transport oxygen from the lungs to other vital organs in the body like brain which cannot survive without oxygen (March of Dimes Foundation, 2008).  There are three types of hemoglobin including Hemoglobin A, C, and S.  A normal red blood cell has hemoglobin A.  The other two, Hemoglobin C and S are abnormal hemoglobin. Hemoglobin A or the normal hemoglobin is  usually very soft, round in shape, and can contour around the body through  blood squeezing past  tiny blood vessels.   Normal hemoglobin is round shaped and very flexible which make it easy to flow through the blood vessels (Sickle Cell Disease Association of America, Inc 2008).

The abnormal hemoglobin S is prevalent among individuals with sickle cell condition.    Hemoglobin S (where S is used to stand for sickle) will initially appear like normal hemoglobin-A, but in a different form.  Unlike the red blood cells with normal hemoglobin-A which can live for up to 120 days, red blood cells with hemoglobin S can live for only 16 days.  They are also distorted in shape and also become very stiff. When traveling in blood vessels, there is a high likelihood that the sickle shaped blood vessels will block small blood vessels due to their shape.  When they block the blood vessel, they usually act like a clot which blocks blood from reaching the vital organs.  The abnormal sickle cell is very stiff and when it is lodged in the blood vessels, it usually cut off blood supply which causes pain and sometimes this can cause adverse damage on tissues to inefficient supply of blood leading complication referred to as sick cell disease. Currently, there is not cure that has been found for sickle cell disease (March of Dimes Foundation, 2008).

Types of sickle cell anemia

There are many types of sickle cell condition. These include Sickle Cell Anemia (SS), Sickle-Hemoglobin C disease (SC), Sickle Beta-zero Thalassemia, and Sickle Beta-Plus Thalassemia.

* Hemoglobin SS or sickle cell anemia

For this condition, individual usually inherit a sickle cell gene from every parent. This is the most prevalent and common type of sickle cell condition   where the genes fully express themselves and individual exhibit sickle cell symptoms.

* Hemoglobin SC or Sickle-Hemoglobin C disease

Under this condition, individuals usually inherit one sickle cell gene and another gene for abnormal hemoglobin C.  This condition may not be fully expressed like the sickle cell disease we have reviewed above.

* Hemoglobin S-beta thalassemia

An individual will inherit one sickle cell gene and another gene for beta thelassemia which is another inherited form of anemia.

Pathophysiology of the disease

The condition is caused by mutation where glutamic acid is usually replaced with valine. This leads to loss of elasticity of the red blood cells.  This loss of elasticity in the red blood cells has been described as the central factor in pathophysiology of the condition.  There is low oxygen tension which promotes the repeated episodes of red blood cell sikcling and damage of the cell membrane. Even when the oxygen tension is returned, the affected cells will not resume their original shape.  As they pass through the blood vessel, they lead to vessel occlusion and later ischemia. (March of Dimes Foundation, 2008)

The disease is mainly inherited from the parents. Those who have expressive sickle cell disease have inherited one sickle cell gene from every   parent.  Those who have only a copy of the gene are carried and therefore they will not express symptoms of the diseases except when they are at a higher attitudes like during a flight (Centers for Disease Control and Prevention, 2008).  At a higher attitude, oxygen supply is less and therefore the red blood cells may become fragile and highly deformed which increases hemolysis process (breaking up of red blood cells)

Therefore the main way through which an individual can develop the sickle cell disease is through inheritance of the sickle cell genes from the parent. The condition is inherited in the same was as blood type, the color of the hair or any other trait.  The kind of hemoglobin that an individual will make in their blood will depend on the hemoglobin genes that have been inherited from their parents.  This may explain the reasons why the disease has continued to be confined in some races only.

Symptoms of the disease

There are a number of medical conditions that occur together with the condition. However, the effects of the condition are highly variable from one individual to another. While some individual will be generally healthy, others will be frequently hospitalized.   The following are the common health problems which are likely to occur in individuals with a sickle cell disease; (Centers for Disease Control and Prevention, 2008)

The first most prevalent condition is anemia. Sickle cells condition leads to destruction of red blood cells resulting to reduced number of hemoglobin. Individuals therefore appear pale, tired and will experience shortness of breath from time to time.

The other prevalent medical condition is infections.  Infants and young children who have sickle cell diseases are likely to have an increased risk of bacterial and viral infections. There are increased incidence of infections like pneumonia, meningitis, and many others in sickle cell individuals.  Viral and bacterial infections have been identified as leading cause of death in children with sickle cell conditions. However, there have been reduced incidents of death since the medical practitioners started treating children with Influenza shot, pneumococcal vaccine, meningococcal vaccines, and many others.

The third medical complication is frequent pain episodes which is prevalent among sickle cells individuals especially children. This also varies from individuals to individuals.  Some individuals will have one or very few pain episodes in a year while others will have more than 15 episodes. The pain episodes usually lasts for some few hours or   few days but some may prolong up to one week.  The pain is usually experienced around body organs or in joints where the sickle cells pile blocking the blood vessels.  Other patients have exhibited hand-foot syndrome where the hands may swell when the blood vessels are cut short due to blockage.  This is usually the first symptoms in children if later followed by fever (Sickle Cell Disease Association of America, Inc 2008).

There is also a high rate of stroke which has been reported for individuals with sickle cell disease. More than 10 percent of children who have sickle cell anemia usually develop stroke which can lead to lasting disabilities and to some extent learning problems.

Cure of the disease

Although there is not cure for the diseases, a number of children with sickle cell disease have been successfully cured through blood stem cell transplant.  The stems cells are immature blood cells which develop to mature red blood cells. They are obtained from bone marrow or in umbilical cord blood. More than 85% of the stem cell transplants have been cured while 5% had died from the procedure (Sickle Cell Disease Association of America, Inc 2008).   Future research shows that gene therapy may eventually offer a cure for the condition.

Conclusion

Sickle cell disease is an inheritable condition which is passed on from parent to children. Epidemiological studies show that the disease is highly prevalence to African Americans and individual with sub-Saharan origin. The sickle cell condition leads to deformation of Hemoglobin which assumes a sickles shape increasing their likelihood of blocking the blood vessels. This cuts off oxygen supply to vital body organs which may be damaged.  Individuals with the condition exhibit various medical complications. Although there is no cure for the disease, stem cell transplant has been used to treat children with the condition.

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