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Sickle-Cell Disease-A Mini Review

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ABSTRACT

Sickle-cell disease is a group of blood disorders inherited from parents; sickle-cell anemia is the most common type. In this mini-review, we focused on the symptoms, reasons for causing the disease and some other factors like treatment, types. This review gives the knowledge over sickle-cell anemia disease.

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INTRODUCTION:

The erythrocytes of certain individuals possess the capacity to undergo reversible changes in shape in response to changes in the partial pressure of oxygen. When the oxygen pressure is lowered, these cells change their forms from the normal biconcave disk to crescent, holly wreath, and other forms. This process is known as sickling[1]**.** It is now recognized that both the sickling phenomenon and the disease are inherited; that both types of manifestation have been found in communities in which sickling has been detected; that the conditions are found in Africans or people who must have had blood connections with Africans; and that the sickling phenomenon is connected with the chemical and physical properties of the hemoglobin in the red cells[2]**.**

This is a hereditary disease which results in the retarded growth. Usually, the red blood cells contain normal hemoglobin and are disc shaped, but people suffering from sickle cell anemia have the sickle-shaped RBC due to the formation of stiff rods by the hemoglobin. These sickle-shaped cells are not flexible and cause the blockage of blood vessels which stops the blood flow resulting in the insufficient delivery of oxygen to tissues.

The blockage of blood vessels also causes pain and damage of organs. The people with sickle cell anemia produces abnormal sizes of RBC which do not last as long as the normal RBC. Many consider that sickle cell anemia is mostly afflicted to blacks like Africans, but it doesn't depend on the skin color. The gene variant of the sickle cell disease is related to malaria and the regions like Africa are most prone to malaria, so the people over that region are mostly affected by sickle cell anemia.

This disease affects million people worldwide mainly in the regions like Africa, East Mediterranean, also in India Sickle cell disease is considered as the most common inherited blood disorder in America. Though doctors provide many preventive or cure methods it is possible for some people to die due to illness caused by this disease.

Reasons for the cause of sickle cell anemia:

Normal adult hemoglobin, designated hemoglobin A, consists of two α-globin chains and two β- globin chains. The cause of sickle cell anemia is a point mutation in β-globin gene. This genetic abnormality leads to the production of sickle hemoglobin, a protein that has unique property of polymerizing into long fibers when deoxygenated, thereby decreasing red cell deformability and damaging the cell membrane[3].

This is generally caused by the genetic abnormality in gene for hemoglobin which possesses sickle hemoglobin. This is not contagious. When a child inherits two sickle hemoglobin genes it results in this disease. In many cases, the child inherits two sickle hemoglobin genes, but in some cases, the child receives only one sickle hemoglobin gene. This condition is known as 'sickle trait'. This is mostly not considered as disease because the complication or problems caused in this case is very rare.

The mutations in the gene generally involve replacement, insertion or deletion of one to four nucleotide bases from the DNA sequence of normal (β^A) gene. Sickle cell anemia which is found primarily in African populations is caused by homozygosity for a unique DNA base pair substitution (β^S) in the sixth codon of the gene[4].The hereditary nature of sickling of erythrocytes of certain persons has been recognized and accepted for many years. The exact pathogenesis of genetic pattern; however, been the source of controversy.

Earlier investigators recognized no genetic difference between sickle cell disease and asymptomatic trait (sicklemia). They regarded both conditions as being due to a single dominant gene which in some persons produce active disease and in others only an asymptomatic trait. More recently Neel and Beet have taken issue with the older hypothesis and have presented an alternate concept. According to these workers, the asymptomatic sickle trait is due to a gene which is represented but once in the patient's genetic system (heterozygous state), whereas active sickle cell disease appears when the gene occurs twice (homozygous state)[5].

The abnormal hemoglobin causes sickle cells which stick to the walls of blood vessels and finally leads to the insufficient supply of oxygen. These sickle cells are fragile and prone to rupture. This condition is known as hemolysis. When the number of red blood cells decreases it results in the anemia. This is known as sickle cell anemia.

SYMPTOMS:

Sickle-cell anemia is a hereditary disease which causes persistent anemia, often resulting in retarded growth; by recurrent crises followed by fever, headache, and bone and joint pains; and by signs of hemolysis (jaundice-leucocytosis) and of blood regeneration.

The main symptoms are a severe anemia with or without jaundice, pyrexia of unknown origin with leucocytosis, cirrhosis of the liver, bossing of the skull, retarded growth, signs suggestive of Perthes's disease, the acute abdomen, or hemiplegia. Despite this individual

with sickle cell disease exhibit significant morbidity and mortality. Symptoms include chronic anemia, acute chest syndrome, stroke, splenic and renal dysfunction, pain crises, and susceptibility to bacterial infections.

Pediatric mortality is primarily due to bacterial infection and stroke. In adults, specific causes of mortality are more varied. Disease expression is variable and is modified by several factors, the most influential being genotype and other factors include β-globin cluster haplotypes, α-globin gene number, and fetal hemoglobin expression[6].

TYPES:

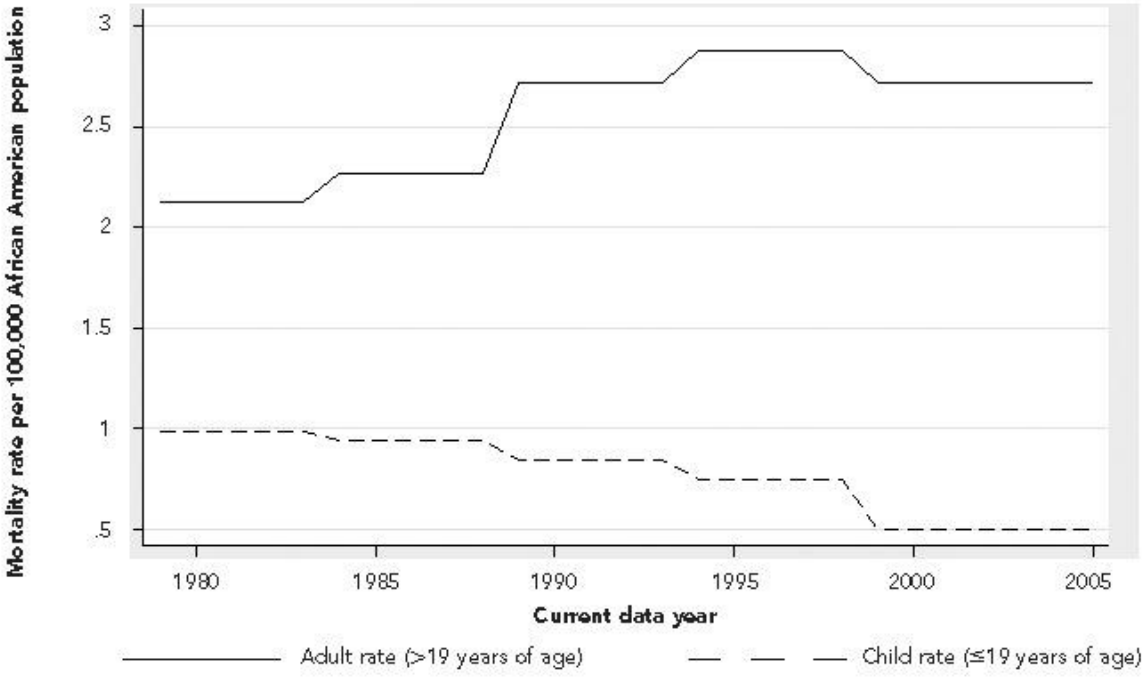
Hemoglobin is a protein which carries oxygen to different tissues and it consists of two α globin chains and two β globin chains. Due to the different mutation in these genes causes different types of sickle cell anemia such as Hemoglobin SS Disease, Hemoglobin SC Disease, Hemoglobin SB+ (Beta) Thalassemia and Beta-Zero Thalassemia[7].

Table: State-specific average annual mortality rates for sickle cell disease per 1,00,000 individuals during 1979-1995[8]

State	Overall
Alabama	5.5
Alaska	0.5
Arizona	0.8
Arkansas	2.8
California	1.5
Colorado	0.7
Connecticut	1.4
Delaware	2.8
District of Columbia	14.6
Florida	3.5
Georgia	5.3
Hawaii	0.3
Illinois	2.6
Indiana	1.7

Lowa	0.3
Kansas	1.3
Kentucky	1.3
Louisiana	5.3
Maine	0.1
Maryland	2.7
Massachusetts	0.7
Michigan	2.5
Minnesota	0.3
Mississippi	6.6
Missouri	2.1
Montana	—
Nebraska	0.4
Nevada	1.3
New Hampshire	<0.1
New jersey	2.4
New Mexico	0.4
New york	2.9
North Carolina	4.2
North Dakota	0.1
Ohio	1.9
Oklahoma	1.5
Oregon	0.3
Pennsylvania	1.8
Rhode island	0.8
South Carolina	6.3
South Dakota	0.2
Tennessee	3.3
Texas	2.1
Utah	0.2

Virginia	2.2
Washington	0.5
West Virginia	0.7
Wisconsin	1.0
Wyoming	0.6



GRAPH: Mortality rates for adults and children with sickle cell disease: U.S., 1979–2005[9]

TREATMENT:

There have been numerous references to the effect of acidosis on sickling; the most relevant study is that by Greenberg and Kass. Physicians have used an alkali regime in the treatment of sickle-cell anemia for some time. Alkali treatment has its place in the prevention of sickling rather than in the treatment of a crisis, where its role is that of preventing further sickling rather than of affecting the wedging of the already sickled cells[10].

Bone marrow transplantation (BMT) is one of the treatment done to sickle cell disease patients, but the main disadvantage in this treatment is that we can't easily predict the suitable persons that should be offered BMT. Risk factor is more in this treatment and it should be

done carefully. Blood transfusion is also one of the major treatment done to the sickle cell disease patients.

CONCLUSION:

Hereby we conclude that sickle cell anemia is one of the main diseases which causes the high mortality rate all over the world, mainly in the regions of Africa, Eastern Mediterranean and some tropical regions.

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