

Sickle Cell Anemia

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The purpose of reviewing sickle cell disease is to identify the classification of this disease and what really are its causes and consequences. Sickle cell disease is one of the most common hereditary anemias that include disorders in the structure or synthesis of hemoglobin and it is mainly caused by congenital abnormalities in hemoglobin structure. As is known clinically, hemoglobin is the major part that is responsible to transport oxygen from the lungs to the rest of the body. Thus, due to the changing in the shape of erythrocytes, which become crescent in shape and fragile, this may lead to a hemolytic anemia, which causes a destruction of red blood cells by the liver and spleen. Accordingly, this microreview will briefly discuss how this occurs and what modern methods are utilized to distinguish this type of inherited disease and what the latest ways to treat and handle this disease are.

Introduction

Principally, let us start with the official definition of sickle cell disease; it is a blood disorder (hemoglobinopathy) that is characterized by a single amino acid substitution in the Beta globin chain; heterozygotes have one normal (B^A) and one affected B chain (B^S) gene (The beta-globin molecule contains a total of 146 amino acids), and produce about 60% (Hb A) and 40% (Hb S); homozygotes produce mainly (Hb S) with small amounts of HbF(fetus) HbA(Adult) and 40% (Hb S); homozygotes produce mainly Hb S with small amounts of Hb F. So individuals who inherit the sickle cell gene from one parent and B thalassemia from the other make predominantly sickle hemoglobin. Furthermore, the amino-acid substitution in the B globin chain results in insufficient oxygenation in blood circulation, and this may lead to an increase in the toughness and aggregation in the microcirculation. These changes appear as a hemolytic anemia, which may cause a tissue block due to red blood cells stacking together, which can lead to more cells rupturing and basically cause a painful crisis, a leg ulcer, bones necrosis, and heart enlargement. It is worth mentioning that there are people who just carry this disease in their genes and no symptoms or other complicated crisis are prominent in them, and clinically they are known as a sickle cell trait. Therefore, to limit the distribution of this disease,

premarital blood investigations for both couples are highly encouraged, especially when the carriers intend to getting married so they make sure that they are going to engage with someone does not have the disease

Recent Progress

Scientifically, the sickle cell anemia is resulted by a single nucleotide change of the (DNA) sequence and it considers as worse mutation that has severe consequences for an organism, because it causes a “frame shift”: all codons following this single nucleotide deletion most likely altered. So this one mutation in fact causes many changes in amino acid sequence. In addition, the frame shift mutation will likely introduce a top codon about 21 amino acids later, thus terminating a particular spot on the gene that is being affected by the frame shift mutation.

Sickle cell disease is clinically detected by many medical labs. Investigations such as Hemoglobin electrophoresis are considered the most widely used diagnostic test and also are a confirmatory examination of sickle cell disease. In addition, there is one test, which is the routine screening test that indicates the presence of sickle hemoglobin or not by obtaining inexpensive blood samples, and this is known as a qualitative test.

The principal of Hemoglobin electrophoresis procedure is to use an electrical current to separate different hemoglobin types, in which each Hg has special

electrical charges, and they move at different speeds in gel medium. Different types of Hg immigrate accordingly either into anode or cathode poles based on electrical charges they have on them. Thus, the test can distinguish the abnormal levels of Hemoglobin S as well as other abnormal hemoglobins. More than that, a peripheral blood smear is taken, which allows a medical lab technician to observe the morphology of red blood cells under a microscope by making a blood film collected on (EDTA) sample so cells can be seen as crescent in shape, target cells, and nucleated. Regarding the treatment and management of (S.C.D) in the meantime, all the medical researchers and scientists have found shows that supportive medications reduce and relieve the pain and the symptoms of periodic sickle cell attacks. For instance, supportive medications include hydration (e.g. Intravenous fluids), anti-inflammatory agents, and pain medication (e.g., non-steroidal anti-inflammatory drugs and narcotic analgesia). In the same vein, there are many preventive tips for sicklier patients to overcome this crisis or regular bouts such as drinking plenty amounts of water to be well-hydrated, and they also should avoid extreme physical activity and take 1mg dose of folic acid daily and for life

Regarding the recent medication that has been discovered lately and is known as chemotherapy, hydroxyurea is one of the most significant drugs that is associated with sickle cell anemia treatment which reduces the annual rate of crisis and disease attacks, and the goal of taking this medication is to reduce the symptoms and complications that may lead to strokes, chest syndrome or even splenomegaly (i.e. an enlargement in the spleen due to severe hemolytic anemia). However, there are many concerns in regards to long-term chemotherapy, and there might be many clinical implications of using it such as leukocytopenia, cancer, gout, fever, nausea, and other implications could result due to this sickle cell anemia disease. It should be noted that Hydroxyurea doses should be taken under the supervision of a hematologist.

Discussion

What is the future of sickle cell anemia treatment?

In the meantime, there is no radical solution for sickle cell disease at all. However, many kinds of therapy have been improved, but still they are at early stages and under trial. Genetic therapy is one of the genetic strategies to treat a sickle cell disease, and in fact it is fast moving nowadays. Actually, it is concerned in modification of human Beta globin that is being mutated or defected in the case of sickle cell anemia. Furthermore, there is another treatment which is bone marrow transplantation, and this procedure has many risks, and indeed it is still not entirely approved since a recipient must have appropriate matching donor for blood transfusion to take place accordingly. However,

researchers are still working to create new techniques that contribute in reducing the risks of the bone marrow transplantation procedure. Nevertheless, in both cases (gene therapy & bone marrow transplantation) the research is still at a very early stage, but the question still remains, will those kinds of treatment be highly improved, extensive and effective in the forthcoming future?

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