

Sickle Cell Anemia: A Brief Synopsis

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ABSTRACT

Sickle cell disorder (SCD), one of the most common genetic disorders worldwide, is now considered as of global importance and medical significance. SCD is a group of red blood cell disorders inherited from a person's parents where both the parents are carrier for the gene. Sickle cell anemia is an autosomal linked recessive trait, causing severe associated health problems leading to reduced life span. With the help of improved novel strategies and therapies, it is utmost important that the treatment is availed by the less-resourceful, impoverished countries.

Keywords: Anemia; Sickle crisis; Hydroxyurea; Blood transfusion

INTRODUCTION

The studies have shown high mortality cases in African continent while in Indian sub-continent; it is mostly prevalent in certain tribes in South-India, Assam, Bihar and Odisha. SCD is a group of red blood cell disorders inherited from a person's parents where both the parents are carrier for the gene. The most common type is Sickle cell anemia (SCA) results due to the abnormality in the oxygen-carrying, red pigmented protein called haemoglobin present in the red blood cells.

COMMENTARY

In 1958, the genetic basis of the disease was found out which suggested that the disease is caused due to the substitution of amino acid valine for glutamic acid at the sixth position of the beta-haemoglobin chain [1]. This substitution, now termed as single point mutation (GAG to GUG) within β -globin gene on Chromosome-11, results in change in the shape of haemoglobin under low oxygen tension [2]. Upon deoxygenation, the mutant hemoglobin called Sickle haemoglobin (HbS) undergoes polymerization tending to change the shape of the red blood cells from biconcave, disc-shaped into crescent, sickle-like [3]. However, the sickling becomes permanent, and the red cells are irreversibly sickled with reoxygenation. HbS confers some protection against Plasmodium falciparum malaria so that these patients are relatively resistant to malaria [4].

Certain factors like low pH (acidosis), low oxygen concentration, fever, sluggish blood flow, reduction in red cell-water content promote the event of sickling. In this condition, the rigid and sticky sickle cells get stuck to the blood vessel walls and block the

free flow of blood and prevent the oxygen from reaching to the tissues [5]. There are clinical consequences associated with the disease such as pain attacks ("sickle cell crisis"), stroke, anemia, infections, swelling and organ damage. The severity of the symptoms may differ from person to person and may lead to acute and chronic complications.

In children suffering from SCD, the clinical signs are absent in the first 6 months of birth due to the high levels of fetal hemoglobin, Hb-F that prevents the red cells from sickling. Early symptoms may include painful swelling of the hands and feet, acute chest syndrome, splenomegaly (due to the abnormal rupture of red blood cells in spleen), damage to kidney and liver, hence making them susceptible to pneumococcal and bacterial infections. Dactylitis may occur in children with this trait. In adults, the "sickle cell crisis" includes anemia (due to secondary folate deficiency), haemolytic crisis (due to rapid enlargement of liver and spleen, destruction of red cells and abrupt fall in Hb), vaso-occlusive crisis (involves organs like penis or lungs; often results in ischaemia, pain, necrosis) and aplastic crisis (results from temporary marrow suppression following infections, particularly with human parvovirus B19) [6].

However, blood tests can show if a person has SCD or the sickle trait. In the complete blood count test, the significant reduction in HbA levels with high reticulocyte (immature red cells that develop in bone marrow) count or high Hb levels can be obtained [7]. The disease can also be diagnosed in babies before birth by taking the sample of tissue from the placenta (the organ that develops in uterus to provide nutrients and oxygen to growing baby) or from amniotic fluid (the fluid surrounding the

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baby in the sac). Screening tests are performed to substantiate the sickle phenotype which includes sodium metabisulphite sickling test, sickle solubility test (based on the principle that HbS becomes insoluble upon deoxygenation) and electrophoresis and chromatography as confirmatory tests. Haemoglobin electrophoresis, isoelectric focusing (IEF), and high performance liquid chromatography (HPLC) are three extensively used tests.

With the new scientific approaches and developments, the longevity of the patients has shown to be increased. The preventive drug treatments such as antibiotics, pain management, blood transfusions benefit people suffering with SCD. Treatments of infections with antibiotics, pneumococcal immunization in younger children, folic acid supplementation, pain relievers for acute and chronic pain and management using iron chelators (like deferoxamine) are some management measures. For patients with severe symptoms, hydroxyurea, an anti-tumor drug, increase the production of Hb-F and reduces vaso-occlusive crisis. Blood transfusions are required for severe anemia due to aplastic crisis, acute chest syndrome attacks and stroke. Transfusions are done to increase oxygen carrying capacity, replace the mutant sickle cells, thus reducing haemolysis and allowing unrestricted blood flow.

CONCLUSION

As of currently, bone marrow transplantation is the only cure to sickle cell disease. In this medical treatment, bone marrow of a

sick patient is replaced with the bone marrow of a healthy, genetic compatible donor, thus improving the quality of life. Since SCD involves a defective gene, its replacement with a normal gene by gene therapy is a promising cure for researchers.

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