Sickle Cell Anemia

Sickle cell anemia is a genetic disorder characterized by abnormal red blood cells that take on a sickle shape, primarily due to variations in the structure of hemoglobin. Normally, red blood cells are disc shaped, but in this condition, the altered hemoglobin causes hemoglobin molecules to clump together, leading to distorted red blood cell shapes. Consequently, these sickle-shaped cells are prone to premature destruction, resulting in anemia. The disease is hereditary and has a higher prevalence in the African continent. Lamanis and other tribal communities in India are notably affected.

Interestingly, individuals with sickle cell anemia have some level of protection against malaria. In ancient times, during widespread malaria outbreaks, many succumbed to the disease, while those with sickle cell anemia showed greater survival rates. Over generations, this disease has become more common in various populations.

The misshapen red blood cells can become trapped in small blood vessels, causing blockages that restrict blood flow. This blocking most commonly affects organs such as bones, kidneys, eyes, and the spleen. The lack of blood supply to these organs can lead to excruciating pain, particularly in areas like the chest, spine, and limbs. The spleen's blood flow disruption can cause it to shrink, increasing the risk of various infectious diseases. Inadequate blood circulation in the eyes can result in blurred vision, while similar issues in the brain may lead to paralysis. In the lungs, improper blood flow can lead to high blood pressure in the pulmonary blood vessels. Sickle cell anemia patients may suffer from a potentially causing a life-threatening condition known as acute chest syndrome. Symptoms of this syndrome include fever, cough, chest pain, difficulty breathing, and low oxygen levels.

In sickle cell patients, certain conditions can exacerbate the sickling of red blood cells. Infections, fever, exercise, stress, low oxygen levels in the blood, dehydration, high acid content in blood, and elevated hemoglobin content can lead to higher risk of sickling, causing disruptions in blood flow and various complications. Consequently, it is crucial to take preventive measures to minimize these factors.

To diagnose sickle cell anemia, various blood tests are conducted. Typically, the hemoglobin level in these patients is lower, ranging from 6 to 10 grams. Microscopic examination of blood reveals sickle-shaped red blood cells. Confirmation of the disease is achieved through hemoglobin electrophoresis (HPLC), which measures the amount of sickle hemoglobin (HbS) present. Additionally, PCR testing can detect variations in the beta globin gene.

Hydroxyurea is a medication used in the treatment of sickle cell anemia. It works

by reducing HbS levels and increasing HbF levels, which decreases the sickling of red blood cells and minimizes the blood vessel occlusion events. This results in reduced bone pain and improved life expectancy. Folic acid tablets are often prescribed alongside hydroxyurea to support red blood cell production in the bone marrow.

A new drug called Crizanlizumab is now available, which prevents red blood cells from becoming trapped in blood vessels. It requires monthly injections. In patients with sickle cell anemia, the spleen may be absent or not function properly, making them more susceptible to infections from microorganisms such as Pneumococcus, Meningococcus, and Hemophilus, which can be life-threatening. As a preventive measure, penicillin V tablets or vaccines are administered. Additionally, getting hepatitis B and influenza vaccines annually is recommended.

In case of severe anemia in sickle cell patients, red blood cell transfusions may be necessary. Exchange transfusion is a procedure used in critical situations or when the patient requires general anesthesia. During this procedure, the patient's red blood cells are replaced with healthy donor red blood cells.

In the event of an infection and fever, prompt administration of antibiotic injections is crucial for these patients. Pain relievers should also be given to manage severe pain. Additionally, maintaining adequate oxygen levels, providing red blood cell transfusions, and administering saline solutions are important measures.

Boys with sickle cell anemia may experience persistent erection of the penis, accompanied by unbearable pain, and in some cases, it can lead to impotence. It is essential for boys to be aware of this issue, and if such problem arises, seeking immediate medical attention can prevent it from escalating into a major problem.

Patients should promptly consult a doctor if they experience breathing difficulties or fever. Oxygen therapy and, in some cases, ventilator support may be required. Sickle cell patients often face significant mental distress, and seeking the help of a psychological counselor can be beneficial.

Bone marrow transplant offers the possibility of a complete cure for this disease. However, it is a risky treatment for sickle cell patients, as around ten to twenty percent of patients may die due to treatment related side effects. Therefore, this treatment is only recommended in select special cases.

Gene therapy treatment is now available in western countries, wherein a normal gene is inserted to replace the defective gene in the patient. This leads to the production of normal hemoglobin. Efforts are being made to introduce this treatment in India as well, with some major medical institutions exploring its implementation.

Women with sickle cell anemia face a significantly higher mortality rate due to various complications if they become pregnant, making it advisable for those with this condition to avoid pregnancy. However, for those who desire children, several measures can be taken to ensure a safer pregnancy. It is crucial to manage surgeries and anesthesia in coordination with a hematologist to minimize complications.

While it is possible to detect the presence of sickle cell disease in the fetus, it is not necessary to do so, as the severity of the disease in the child cannot be accurately determined while still in the womb.

Individuals with sickle cell anemia should have regular check-ups with their doctors every three months, with CBC (Complete Blood Count) tests conducted during each visit. Additionally, RFT (Renal Function Test), LFT (Liver Function Test), hemoglobin electrophoresis, eye tests, TCD (Transcranial Doppler) tests, lung and dental examinations should be performed annually.

While people with sickle cell anemia do not require specific dietary regimens, staying well-hydrated is important. They should avoid extreme hot or cold weather and refrain from excessive exercise. Reducing the consumption of painkillers and avoiding smoking and excessive alcohol intake is also advised.

As the disease is hereditary, it is beneficial to do hemoglobin electrophoresis test in patient's siblings, blood relatives, and children.

If one of the two beta globin genes is normal, the individual will not experience any problems; this condition is known as sickle cell trait. However, when two people with sickle cell trait marry and have children, there is about a 25 percent chance that their children may develop sickle cell anemia.

It is essential to know that the disease is not transmitted through contact with the patient.

This article provides a brief overview of Sickle Cell Anemia. For further information, one can contact their treating medical team.

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