Beta thalassemia

Red blood cells in our bloodstream contain a crucial component known as hemoglobin, responsible for giving blood its characteristic red color. Hemoglobin plays a vital role in absorbing oxygen from the lungs and transporting it to various parts of the body. Structurally, hemoglobin consists of two essential parts: heme and globin. The heme portion is predominantly composed of iron, while the globin part comprises of two alpha and two beta protein chains.

The production of beta chains relies on both the beta genes inherited from the father and mother. Each individual possesses two beta genes, one from each parent. Interestingly, if one beta gene malfunctions due to a genetic mutation, normal beta gene produces adequate number of beta chains. Hence, it does not lead to any significant issues for the person. When one beta gene functions correctly and the other is faulty, this condition is known as beta thalassemia trait.

People with the beta thalassemia trait can pass either a completely normal beta gene or a defective one to their children. The specific gene transmitted to the child cannot be predicted in advance. If both the father and mother carry the beta thalassemia trait, there is a 25 percent chance of passing on both non-working (defective) beta gene to the child. In the event that the child inherits two non-working genes, it will result in the absence of beta globin chain production. Without this essential chain, hemoglobin cannot be formed, leading to a severe condition known as beta thalassemia major.

In cases of beta thalassemia major, affected children require regular blood transfusions to manage severe anemia since their blood lacks sufficient hemoglobin. Despite receiving red blood cells through transfusions, these cells have a limited lifespan and are eventually destroyed by the spleen after approximately 2-4 weeks. Consequently, the anemia recurs, necessitating further transfusions for the child's well-being.

Individuals with the thalassemia trait have an inherent advantage against malaria as malaria parasitess cannot thrive within their red blood cells. In ancient times, when malaria ravaged the world, the human population faced extinction. However, those carrying the thalassemia trait survived the epidemic, while others succumbed. Their descendants spread across the globe, leading to a prevalence of the beta gene problem. In India, thalassemia is particularly common among communities such as the Lamanis, Gauda Saraswat Brahmins, Sindhis, Rajputs, Jains, Shias, and Pathans.

As mentioned earlier, thalassemia major occurs when both genes responsible for the beta chain fail to function correctly. This condition results in approximately ten thousand children with thalassemia major being born in India each year.

Children with thalassemia major experience a lack of oxygen in their blood, leading to bone marrow enlargement in various bones, causing facial deformities. Anemia increases iron absorption from the intestines, and each blood transfusion introduces a significant amount of iron into

the body. This excess iron accumulates in organs like the heart and liver, potentially causing damage and organ failure. Additionally, hormonal imbalances due to the lack of certain hormones may result in stunted growth, thyroid issues, and impotence in these patients.

As blood production decreases in the bone marrow, the spleen attempts to compensate by producing blood, despite its inability to do so. This causes a considerable enlargement of the spleen and shortens the lifespan of transfused red blood cells, leading to an increased need for blood in the patient. Furthermore, individuals with thalassemia major are more susceptible to infections and blood clotting within veins.

Children with thalassemia major may begin showing symptoms like body paleness and irritability around the age of four to six months. A blood test at this stage would reveal a hemoglobin level of less than 5%. Microscopic examination of the blood under microscope would indicate various changes characteristic of thalassemia major. Confirmatory tests, such as hemoglobin electrophoresis or HPLC, would show elevated levels of Hb-A2 and HbF, further confirming the presence of thalassemia major. In some cases, genetic tests are also conducted to detect abnormalities in the beta genes.

Without regular blood transfusions, children with this condition typically survive for less than four years. However, with proper and frequent blood replenishment and iron removal therapy, their life expectancy can be similar to that of normal individuals. It's crucial to maintain the hemoglobin level above 9 and consider iron removal therapy when blood ferritin levels exceed 1000 units. Deferasirox and Deferiprone tablets are used for this purpose. When using these medications, caution must be exercised to avoid contact with iron utensils. Deferasirox may cause loose motions, while Deferiprone can lead to joint pain and reduced white blood cell count.

Surgical removal of an enlarged spleen is sometimes necessary. However, a bone marrow transplant is the only potential cure available in India. For this treatment, a sibling with a matching HLA (Human Leukocyte Antigen) is required, and it's best to conduct the transplant when the child is 4-5 years old, as success rates decrease after seven years due to high iron stores.

Dietary adjustments are essential for patients with thalassemia major. It is advisable for them to avoid meats other than chicken. Consuming black tea can help reduce iron absorption. Foods rich in vitamin C, such as lemon and orange, should be limited during meals, while increased consumption of milk and fish can be beneficial. Incorporating wheatgrass juice into their diet can also be advantageous.

Medicines like Hydroxyurea, Thalidomide, Decitabine, and Luspatercept, can be used in a patient with thalassemia, under the guidance of a doctor.

Additionally, gene therapy treatment is now available in western countries. This innovative approach involves replacing the malfunctioning beta gene with a healthy one. The modified cells are then introduced into the patient's bone marrow. Compared

to traditional bone marrow transplants, gene therapy treatment has fewer side effects.

Preventing beta thalassemia major is entirely possible. Detecting the presence of the beta thalassemia trait can be achieved through a Hemoglobin HPLC blood test. Couples who both have the beta thalassemia trait have a 25 percent chance of having a child with thalassemia major. However, advancements in prenatal testing allow the detection of this condition while the baby is still in the womb. Procedures like Amniocentesis or Chorionic villi biopsy are used to collect cell samples from the baby for genetic testing. If both beta genes of the child are found to be abnormal, it confirms the presence of beta thalassemia major. In such cases, pregnancy termination can be considered to prevent the birth of a child with thalassemia major.

Therefore, it is advisable for all adults planning to have children to undergo a hemoglobin HPLC test. In some states, like Gujarat, this test has already become mandatory. Opting for this affordable test (usually around 1000 rupees) is a wise decision instead of dealing with the challenges of raising a child with thalassemia major. Governments should take action in making this blood test free and compulsory to promote preventive measures against beta thalassemia major.

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