Case study

A case report on hepatosplenomegaly and cholelithiasis in a case of beta-thalassemia: Hb E type.

ABSTRACT

Beta thalassemia is an inherited blood disorder in which the body does not produce enough haemoglobin. It is a relatively uncommon condition. The association with beta-thalassemia is uncommon. An enlarged spleen in a child with beta-thalassemia can be caused by increased red blood cell destruction, the formation of blood cells outside of the bone marrow, repeated blood transfusions, or iron overload. We report a rare case of 17 years old patient admitted with multiple blood transfusions and pneumococcal and meningococcal vaccination done on 8th November 2021 and last blood transfusion on 13th November 2021. The patient was admitted to AVBR Hospital Sawangi (M) Wardha with the chief complaint of abdominal pain that has been constantly exacerbated by movement as well as in resting period over 7 days with the clinical findings of the backache and joint pain. He was having a continuous blood transfusion in the last 7 months. After the diagnostic investigation like ultrasonography and computed tomography, the client was diagnosed with hepatosplenomegaly and cholelithiasis. The physician is treating the child with continuous blood transfusion and with iron chelating therapy which would be beneficial to reduce the iron load. The patient has been planned for splenectomy, which was the long-lasting treating strategy for his clinical presentation.

Keywords: child, hepatosplenomegaly, Cholelithiasis, splenectomy, Beta-thalassemia

INTRODUCTION:

Thalassemia refers to a group of blood disorders that affect how the body produces hemoglobin. Hemoglobin is a protein that is found in red blood cells and transports oxygen throughout the body.[1] This disorder is classified into α and β types based on the abnormal globin chain, and each type is further subdivided into a major, intermedia, and minor subtypes based on the severity of the chain abnormality.[2] Beta thalassemia is an inherited blood disorder in which the body does not produce enough hemoglobin. Symptoms of beta-thalassemia can range from mild to severe, depending on the type.[3]The global annual

incidence of symptomatic individuals is estimated to be one in 100,000, with the European Union accounting for one in 10,000. Thalassemia is a monogenic hematologic disorder that is common in Iran, particularly in the country's south.[4]

PATIENT INFORMATION

A 17 years old patient with a known case of beta-thalassemia which was of Hb E type has been diagnosed seven months back. The patient came in the hospital now with 7 days complaints of severe abdominal pain continuously with joint pain and backache. The pain was rated by using a visual analog pain scale which has been rated as six out of ten. The patient was admitted to the hospital several times for continuous blood transfusion over 7 months. The last blood transfusion was done on 13th November 2021 and pneumococcal and meningococcal vaccination has been taken on 8th November 2021. The patient is Beta thalassemia major is caused by **a** homozygous mutation of the beta-globin gene, resulting in the total absence of beta chains. It manifests clinically as jaundice, growth retardation, hepatosplenomegaly, endocrine abnormalities, and severe anemia requiring life-long blood transfusions

On physical examination, a child looks pallor, poor intake habits due to abdominal distension. There is failure to thrive and physical underdevelopment. The patient has undergone a complete blood count in which Hb was 7.2g% whereas RBC was 3.97 million/cu.mm. The patient ferritin level was 1000 whereas the platelet count was 1.06 lakh per microliter of blood. All of the other findings, including electrolytes and urinalysis, were in a normal range Computed tomography Scan , which revealed that liver enlarged measures21cm(hepatomegaly), gallbladder shows radio-opaque calculi largest measuring 6×6mm and in spleen enlarged measures 24cm(splenomegaly). The ultrasonography revealed hepatosplenomegaly and cholelithiasis.

The physician advised the patient to continue blood transfusion which the patient is taking for the last 7months which is the mainstay of treatment. Iron chelating agents have been used to reduce iron load and consequent hemosiderosis. Tablet Deferoxamine 500mg once a day, tablet montair LS twice a day, has been administered with high protein diet and also planned for splenectomy.

DISCUSSION:

Beta-thalassemia is a group of hereditary blood disorders characterized by errors in the synthesis of the beta chains of hemoglobin, resulting in a wide range of phenotypes ranging from severe anemia to clinically asymptomatic individuals.[5] Individuals with thalassemia typically present with severe anemia within the first two years of life, necessitating regular red blood cell (RBC) transfusions.[6] If a regular transfusion program is started that maintains a minimum Hb concentration of 9.5 to 10.5 g/dL, growth and development tend to be normal for 10 to 12 years.[7] In the present case, the child was administered regular blood transfusion over 7 months. Clinical findings seen in developing countries due to untreated and poorly transfused individuals with thalassemia include growth retardation, pallor, jaundice, poor musculature, hepatosplenomegaly, leg ulcers, the development of masses from extramedullary haematopoiesis, and skeletal changes caused by bone marrow expansion. [8]

Thalassemia manifests clinically between the ages of 6 and 24 months. Infants who are affected do not thrive and become increasingly pale. Feeding difficulties, diarrhea, irritability, recurrent bouts of fever, and progressive abdominal enlargement due to spleen and liver enlargement may occur.[9] The child was pale which was inspecting and as time moved on the Ct- Scan and USG revealed hepatosplenomegaly and cholelithiasis.

Regular transfusion therapy causes iron overload-related complications such as growth retardation, sexual maturation failure, diabetes mellitus, and insufficiency of the parathyroid, thyroid, pituitary, and, less commonly, adrenal glands), dilated myocardiopathy, liver fibrosis, and cirrhosis.[10] In the present case to reduce the regular therapy and iron overload the chelating therapy has been started.

Regular RBC transfusions, iron chelation, and management of secondary complications of iron overload are all part of the treatment for thalassemia. Patients who have been transfused may experience complications due to iron overload. The complications are infinite due to iron overload which leads to death. Some studies show the specific iron overload complications in children include growth retardation and failure or delay in sexual maturation. Later iron overload-related complications include heart involvement (dilated myocardiopathy or, in rare cases, arrhythmias), liver involvement (fibrosis and cirrhosis), and endocrine gland involvement (diabetes mellitus, hypogonadism, and insufficiency of the parathyroid, thyroid, pituitary, and, less commonly, adrenal glands).[11] Other complications include hypersplenism, chronic hepatitis (caused by virus infection with hepatitis B and/or C), HIV infection, venous thrombosis, and osteoporosis. Patients with the liver viral infection and iron

overload are at a higher risk of developing hepatocellular carcinoma. Spleen removal may be necessary in some cases.[12] As same in the patient due to the spleen enlargement, the splenectomy is planned as a surgical intervention.

Compliance with iron chelation therapy (discussed later) has the greatest influence on the frequency and severity of iron overload-related complications. Individuals who have not been transfused regularly usually die before the second or third decade. Individuals who have been regularly transfused and treated with appropriate chelation live past the age of 40. [13]The most serious life-threatening complication of iron overload in beta-thalassemia is a cardiac disease caused by myocardial siderosis. In fact, due to cardiac complications, 71% of beta-thalassemia patients die. [14]

The only permanent cure currently available is bone marrow transplantation. Following recent medical advances in transfusion, iron chelation, and bone marrow transplantation therapy, the prognosis for people with beta-thalassemia has improved significantly in the last 20 years. However, cardiac disease is still the leading cause of death in iron overload patients. Regular medical care, including transfusions and chelation, is the best way for the patient to live the healthiest life possible.[15][16]

CONCLUSION:

The prognosis of thalassemia depends on the response and complication occurring due to the multiple blood transfusion and iron-chelating therapy as well as with the patient undergoing splenectomy surgical intervention. The case of 17 years old patient survive about 17 years and still surviving with multiple blood transfusion. The prognosis for this people is poor. There are several advances for treating major Beta thalassemia is Bone marrow transplantation which is one of the longest treatment modalities. As we present a case of 17 years old patient who is on continuous blood transfusion and iron-chelating therapy. The condition has an improvement but several complications occurred during this phase of time of 7 months and through CT- scan and USG it reveals that the patient developed hepatosplenomegaly with cholelithiasis. The surgeon planned for further surgical intervention of splenectomy.

INFORMED CONSENT

The authors got written informed consent from the patient for imaging and other clinical investigation or information for publication in the journal. The author keeps names and

initials that will not be published concealed. While every effort will be taken to disguise the identity, anonymity cannot be guaranteed.

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