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**RESEARCH ARTICLE**

# HAEMOGLOBIN E/BETA-THALASSEMIA: A CASE REPORT FROM WEST MAHARASHTRA, INDIA

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**ABSTRACT**

Haemoglobin E- Beta thalassaemia is the commonest form of severe thalassaemia in many Asian countries In India; it is prevalent in North-Eastern region, but relatively rare in the rest of the country. Identification of this Hb variant thalassemia is important, because doubly heterozygous state for HbE and beta-thalassemia is characterized clinically by thalassemia major. Manifestations of E-beta thalassemia include refractory anemia, splenomegaly and sometimes, unexplained Jaundice. In addition these patients have additional complications like iron overload, hypercoagulable states (post-splenectomy), pulmonary hypertension and cardiopulmonary disease. Thus the affected individual may be symptomatic and transfusion dependent at an early age. This paper reports a case with Hb E- Beta thalassemia from West Maharashtra



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## INTRODUCTION

Hb E/beta-thalassemia (E/β thalassemia) is the commonest form of severe thalassemia in many Asian countries. It has become an increasingly severe public health problem in west Maharashtra region of India. Hb E/ β-thalassemia is inherited in an autosomal recessive manner; both parents of an affected individual are obligate carriers for one of these haemoglobin variants. Both variants are benign when they occur alone.

## MATERIALS AND METHODS

A 13 year old boy with severe anaemia and hepatosplenomegaly. About 2ml intravenous blood sample was collected from the patient in EDTA (Ethylene Diamine Tetra Acetic acid) coated vaccutainers and another 1ml blood was collected for serum.

The hematological analysis was performed on cell counter to obtain the Red cell indices (Hb, MCV, MCH, MCHC and RDW). Hb typing was performed by Hb electrophoresis and high performance liquid chromatography (HPLC). The serum ferritin level was determined. Finally the patient was treated with chelation therapy.

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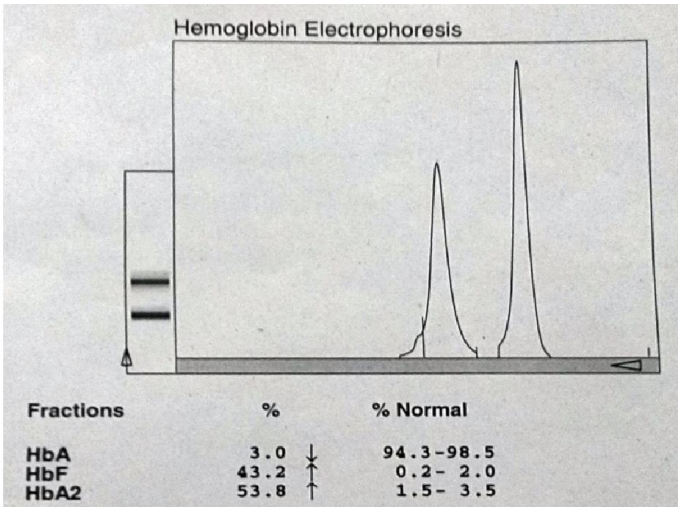
**Case report**

A 13 year old boy belonging to labourer family presented in the outpatient department of paediatrics with complaints of weakness, pallor and abdominal swelling for 2 months with history of 4 units of blood transfusion 4 months back. He had history of repeated cold, cough and diarrhoea with loss of appetite. On examination, patient was found severely anaemic and weak. On systemic examination liver and spleen was found to be enlarged.

Patient was admitted; red blood indices showed Hb of 6.3 gm/dL, MCV of 57 fL, MCH of 15.7 pg, MCHC of 27.6g/dl and RDW of 25.7 fL. RBC morphology was severely hypochromic and predominantly microcytic, Marked anisopoikilocytosis, basophilic stippling with presence of target cells,tear drop cells and schistocytes. Hb typing report show 53.8% Hb A2/E and 43.2% Hb F.Serum unconjugated bilirubin was 5 mg/dl. When correlated with red cell indices, the findings along with his clinical presentation were determined to be consistent with diagnosis of Hb E/β thalassemia. For confirmation of diagnosis, parental and sibling screening has been done and mother was detected as β-thalassemia trait, Father was detected as Hb E trait and his sister had compound heterozygous for Hb E-Beta Thalessemia, his brother was Beta Thalessemia trait. Reticulocyte count was 5 %. His ferritin level was 3011ng/ml. Following chelation therapy by using desferrioxamine, folic acid course and 4 units of blood transfusion the condition of the patient improved.

After showing improvement in the haemoglobin percentage the patient was discharged. At follow up after one month the ferritin level of the patient decreased.




## DISCUSSION

The clinical course of individuals with Hb E/β-thalassemia varies widely, but typically involves anaemia (often requiring blood transfusions) and hepatosplenomegaly, and may involve skeletal disease. The severity of Hb E/β-thalassemia varies from mild to severe. About half of individuals who have Hb E/β-thalassemia have severe manifestations that resemble thalassemia major, requiring regular blood transfusions to treat severe anaemia. Without treatment, this condition can result in lethargy, pallor, growth delay, developmental delay and hepatosplenomegaly.

β-thalassaemia is a major monogenic single gene disorder resulting from a reduced or absent synthesis of β-globin chain. The frequency of beta-thalassemia trait has variously been reported from <1% to 17% and an average of 3.3% in India. (Madan *et al*., 2000) The pathophysiology of Hb E/β-thalassemia is related to many factors including reduced β chain synthesis resulting in globin chain imbalance, ineffective erythropoiesis, apoptosis, oxidative damage and shortened red cell survival. (Datta *et al*., 2006; Pootrakul *et al*., 2000)

In most cases of Hb E/β-thalassaemia regular blood transfusion is required to maintain an adequate supply of haemoglobin. Chronic blood transfusions inevitably lead to iron overload and serious clinical sequelae and patients' receiving such transfusions requires lifelong chelation therapy. (Poggiali *et al*., 2012) Elevated serum ferritin predicts end-organ involvement in non-hereditary iron overload conditions, such as transfusion-associated iron overload in myelodysplastic syndromes, thalassemias and haemoglobinopathies. Levels less than 1500 ng/ml indicated mostly acceptable iron overload; levels greater than or equal to 3000 ng/ml were specific for significant iron-overload and were associated with liver injury. (Wang *et al*., 2010) It is now generally appreciated that no patient with Hb E/β-thalassaemia should be placed on a regimen of regular transfusions without an extended period (of at least 3-6 months without intercurrent illness) in which growth, pubertal development if applicable, quality of life, symptoms and signs of anaemia including changes in spleen size, are monitored. (Olivieri *et al*., 2011)

Prenatal Diagnosis can be done by Chorionic Villous Sampling at 10-12 weeks gestation in couples with identified _Beta-thalassemia and HbE mutations. Special DNA testing can also be done to identify mutation/disease. (Phadke and Agarwal, 2003) During childhood regular follow of growth and facial deformities, hemoglobon level, prophylaxis of infections causing worsening of anemia with vaccines, treatment of potential infectious sites are essential. Daily oral penicillin is recommended. (Kishore *et al*., 2007; Doctor Dora Bachir, 2009)

**Conclusion**

The only way to prevent the disease is carrier detection and awareness among the people about this emerging epidemic. An effective strategy of preventing the progression of the disease in the West Maharashtra might be a screening program employing more sophisticated techniques like polymerase chain reaction (PCR) followed by direct sequencing, genetic counseling.

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