CASE REPORT

HAEMOGLOBIN E/BETA-THALASSEMIA: A CASE REPORT FROM UPPER ASSAM, INDIA
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ABSTRACT: We report here a patient from upper Assam region of India with Hb E/β thalassemia disease, whose father belongs to the tea garden community and mother is Assamese where Hb E is very common. The patient was suffering from severe anaemia and hepatosplenomegaly. After examination two units of blood transfusion and folic acid course were given to the patient. Patient was finally treated with chelation therapy.

KEYWORDS: Hb E/ β -thalassemia, Anaemia, Chelation therapy.

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 upper Assam 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.

We present a case of Hb E/ β-thalassemia from upper Assam region of India which was successfully treated with chelation therapy.

MATERIALS AND METHODS: A 5 year old boy with severe anaemia and hepatosplenomegaly presented in the outpatient department of pediatrics. 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 (SYMEX XS- 800i, Japan) to obtain the Red cell indices (Hb, MCV, MCH, MCHC and RDW). Hb typing was performed by high performance liquid chromatography (HPLC) using the BIO-RAD D10 Hemoglobin testing system. The serum ferritin level was determined in IMMULITE – 1000 (SIEMENS, UK). Finally the patient was treated with chelation therapy.

CASE REPORT: A 5 year old boy belonging to the tea garden community presented in the outpatient Department of Paediatrics with complaints of weakness, pallor and abdominal swelling for 6 months with history of 2 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. He was weighing 13kg (weight for age below third centile). On systemic examination liver and spleen was found to be enlarged.

Patient was admitted; red blood indices showed Hb of 4.0 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
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predominantly microcytic along with presence of target cells. Hb typing report show 59.5% Hb A2/E and 32.0% Hb F. 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 father was detected as β-thalassemia trait, mother was detected as Hb E/E homozygous and his sister had Hb A/E heterozygous. His ferritin level was 3011ng/ml. Following chelation therapy by using deferasirox, folic acid course and 2 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.[1] Ethnic groups in north-eastern India have among the highest known gene frequency for Hb E. However, there are few reports of Hb E/β-thalassemia from these ethnic groups. 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.[2-3]

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.[4] 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.[5] 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.[6]

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 upper Assam might be a screening program employing more sophisticated techniques like polymerase chain reaction (PCR) followed by direct sequencing, genetic counseling.
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REFERENCES:
1. Madan N, Sharma S, Sood SK, Colah R,(Late) Bhatia HM. Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India, Indian J Hum Genet. 2010; 16(1): 16–25.
2. Datta P, Basu S, Chakravarty SB, Chakravarty A, Banerjee D, Chandra S, et al. Enhanced oxidative cross-linking of hemoglobin E with spectrin and loss of erythrocyte membrane asymmetry in hemoglobin E beta-thalassemia. Blood Cells Mol Dis. 2006; 37: 77-81.
3. Pootrakul P, Sirankapracha P, Hemsorach S, Moungsub W, Kumbunlue R, Piangitjagum A, et al. A correlation of erythrokinetics, ineffective erythropoiesis, and erythroid precursor apoptosis in Thai patients with thalassemia. Blood 2000; 96: 2606-12.
4. Poggiali E, Cassinerio E, Zanaboni L, Cappellini MD. An update on iron chelation therapy. Blood Transfus 2012; 10: 411-22.
5. Wang W, Knovich MA, Coffman LG, Torti FM, Torti SV. Serum Ferritin: Past, Present and Future. Biochim Biophys Acta. 2010; 1800 (8): 760–769.
6. Olivieri NF, Pakbaz Z, Vichinsky E. Hb E/beta-thalassaemia: a common & clinically diverse disorder, Indian J Med Res. 2011; 522-531.

Photograph of the patient
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