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Case Report

An interesting case of anemia with jaundice

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ABSTRACT

Hemoglobinopathies are haematalogical disorders that afflict millions of individuals worldwide. HbE is a hemoglobin variation caused by a mutation in the β globin gene that results in the substitution of glutamic acid for lysine at position 26 of the β globin gene. Hemoglobin (Hb) synthesis abnormalities are among the most prevalent inherited disorders. They can be quantitative (thalassemia syndrome) or qualitative (variant HbS). Hemoglobin E (HbE) is the second most common hemoglobin variation after hemoglobin S (HbS).

Keywords: Hemoglobin E disease, Hemoglobinopathies, HbE Thalassemia

INTRODUCTION

HbE is a haemoglobin variant that has a mutation in the β-globin gene. Here the defect lies in the substitution of glutamate or lysine at position 26 of the β-globin gene. Hb-E disease is defined as the coexistence of 2β-E alleles. Patients with genotype EE are either completely asymptomatic or mildly anaemic.[1] In this case, the person has been suffering from anaemia and jaundice for 15 years.

CASE REPORT

A 23 year old male presented for the evaluation of anemia and jaundice which was present for 15 years. There was no history of blood transfusion, bleeding diathesis, cholelithiasis and any stigmata of chronic liver disease. He was a non-smoker and non-alcoholic. Family history showed no other similar illness.

On examination, patient had pallor and icterus. Vital signs were normal. Examination of the abdomen revealed mild hepatomegaly and moderate splenomegaly and remainder of the examination was normal

Hemogram showed hemoglobin of 8.7gm% with normal cell counts. Peripheral smear showed microcytic hypochromic cells with anisopoikilocytosis, elliptocytes, target and teardrop cells. Iron studies showed no evidence of iron deficiency. LFT showed total bilirubin - 9.2mg/dl and indirect bilirubin- 8.4 with normal transaminases levels.

Serum LDH was normal ruling out intravascular hemolysis. Hemoglobin electrophoresis showed HbF-48.3%. HbA2/E-39.0% (Figure 1). Thus, confirming HbE thalassemia disease with thalassemia intermedia phenotype.

DISCUSSION

Hemoglobin E (Hb E) is known to be one of the world's most prevalent and vital mutations. This results in a heterogeneous group of disorders whose phenotype can vary from asymptomatic to severe transfusion dependent thalassemia major. Out of these, Hb E trait and Hb EE are considered to cause mild disorder.[2] Thalassemias' and hemoglobinopathies are autosomal recessive inherited

disorders, that primarily affect the globin chain of the hemoglobin.

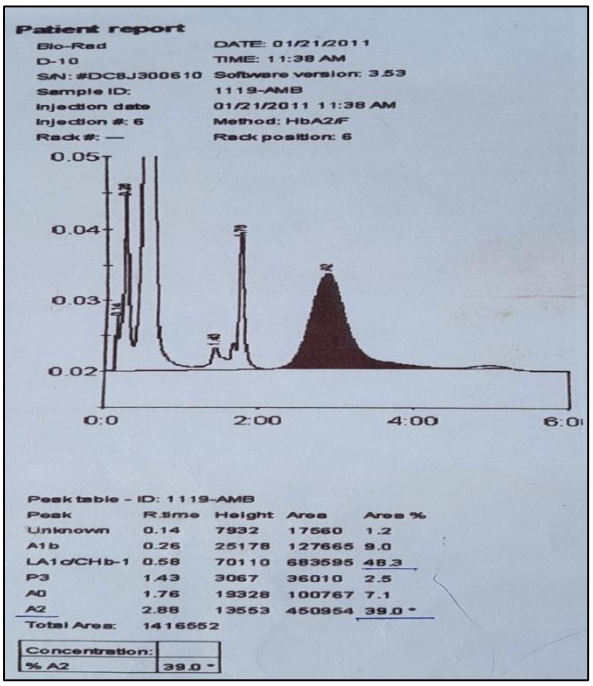

Figure 1: Hemoglobinopathy screening by HPLC.

These illnesses which were once exclusive to specific regions, tribes particularly those with endogamous marital practices, are now widespread throughout the world. This could be because of marriages between different races and communities across the world. The presence of Hb E used to be generally restricted to the Northeastern states of India with few occasional reports of it from an Indian state named Uttar Pradesh.[3] Clinical phenotypes of Hb E disorders particularly Hb E/ β-thalassemia have a wide range of clinical characteristics. These individuals' hematological studies indicate mild microcytosis, hypochromia and erythrocytosis as found in the β-thalassemia trait. However, identifying these individuals is critical because they may be carriers of the defective gene, resulting in a variety of hemoglobinopathies and thalassemias in their offspring. Hb E disease has a mild to moderate manifestation. By the age of ten, the majority of children would have had developed clinical symptoms. The most prevalent symptoms and presentation of Hb E disease include no or mild anemia, jaundice, fever, abdominal pain and gastrointestinal disturbances with or without splenomegaly. Microcytic hypochromic red cells with or without hemolysis are visible in the blood image. Recurrent episodes of jaundice are seen in some patients with Hb E illness. The clinical picture of Hb E/β-thalassemia varies, ranging from β-thalassemia minor through β-thalassemia intermedia to β-thalassemia major, however the majority of patients have moderately serious illness.[3] Hb E is mutation caused when the β globin gene undergoes substitution of glutamic acid with lysine at codon 26. Following this, cryptic mRNA splice site gets activated which reduces the production of β-E chain leading to thalassemia phenotype.[4] Hb E has a weaker α/β interface, that causes some instability when exposed to oxidant stress. There is no clinical significance to the Hb E trait in most cases. Although microcytosis without anemia is possible in certain patients. Unless laboratory tests are obtained, the image could be mistaken for iron deficiency.[2]

CONCLUSION

The given patient was treated symptomatically with H2 blockers, folic acid tablets and injections. This case signifies the necessity of lateral thinking in microcytic hypochromic anemia and appropriate workup revealed thalassemia, which was caused due to ineffective erythropoiesis leading to intramedullary hemolysis presenting clinically as hemolytic anemia, i.e., anemia with jaundice which is a rare presentation.

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REFERENCES

1. Jain D, Gupta SK, Singh T. Hemoglobin E disease with concomitant tuberculosis. Indian Journal of Pathology and Microbiology. 2008;51:570.
2. Vichinsky E. Hemoglobin E Syndromes. American Society of Hematology. 2007;(1):79-83.
3. Patne SCU, Shukla J. Hemoglobin E disorders in Eastern Uttar Pradesh. Indian Journal of Pathology and Microbiology. 2000;52(1):110-2.
4. Mias DD, Gulberson RD, Keren DF. The Range of Hemoglobin A2 in Hemoglobin E Heterozygotes as Determined by Capillary Electrophoresis. American Journal of Clinical Pathology. 2009;132:34-8.

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