Case Report

A Rare Presentation of Haemoglobin D-thalassemia

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Abstract:- Hemoglobin D is the fourth most common haemoglobin variant occuring in a group of Asian population particularly from India,Pakistan, Iran and Iraq. In India, it is mainly reported in north western states of Haryana, Punjab and Gujarat. Homozygous Hb D disease is a rare disease and usually presents with mild haemolytic anemia with mild to moderate splenomegaly. Heterozygous form of Hb D is mild, clinically silent, but coinheritance of Hb D with Hb S or thalassemia produces clinically significant conditions like sickle cell anemia and chronic haemolytic anemia of moderate severity. We present a similar case of compound heterozygous Hb D/ beta thalassemia minor with hemolytic jaundice and hepatosplenomegaly.

Key Words:- Haemoglobin D disease, Homozygous HbD, Haemoglobin S, Thalassemia

Introduction

Hemoglobin D-Punjab occurs with greatest prevalence (2%) in Sikhs of Punjab, India, whereas Gujarat, the province in the west from where the case was reported, has a prevalence rate of 1%.[1] Encountered by Itano,[2] in 1951, Hb D differs structurally from normal Hemoglobin A at 121 position on beta chain, where glutamine replaces glutamic acid.[3] The electrophoretic mobility of Hb D is identical to that of Hb S at alkaline pH in cellulose acetate, but HbD can be distinguished from Hb S by its normal solubility as well as different mobility from HbS on citrate agar electrophoresis at pH 6.2.[4] Hb D gene can be detected by DNA amplification and globin chain analysis.[4,5,6] There are several hemoglobin D variants, amongst them Hb D-Punjab (also known as Hb D- Los Angeles) is by far the commonest.[1,7] Individuals with compound heterozygosity for Hb D-Punjab / β-thalassemia have mild to moderate disease [8]. A subset of patients may manifest moderately severe anemia with splenomegaly and chronic hemolysis, while others may show very mild disease with no apparent clinical signs. Hb levels range between 8 and 12 g/dl. Patients primarily have Hb D and a small percentage of Hb A depending on the type of β-thalassemia mutation (β+or β0). Hb A_2 levels may be normal or increased while Hb F levels are usually normal, although slightly increased in a few cases. Hb D occurs in four forms: heterozygous Hb D trait, Hb D-thalassemia, Hb S-D disease and the rare homozygous Hb D disease, which is usually associated with mild haemolytic anemia and mild to moderate splenomegaly.[1]

Case Report

We report the case of a 30-year-old female who presented with complaints of jaundice since one week, pain abdomen since 3 days and generalised weakness since one week. Physical

examination revealed a palpable spleen and hepatomegaly.The laboratory results were Hb: 8.3 g/dl, TLC-7400/cmm, RBC count- 6.62 million/cmm, MCV: 58.80fl, MCH: 18.00 pg, and RDW: 14.5%, total bilirubin-4.23(indirect-1.09, direct-3.14). Peripheral blood film showed microcytes, tear drop cells, pear shaped cells, target cells, polychromasia +, 2nRBC/100 WBC, shift to right. An impression of microcytic hypochromic anemia was given. Ferritin level was 90.30 ng/ml. The HPLC showed hemoglobin in D window of 77.90%, Hb A2 of 4.5%, Hb A of 3.00%, and Hb F of 1.10%. Electrophoresis showed properties of an Hb D Punjab with beta thalassemia minor. The results showed that the individual was a compound heterozygote for Hb D / β-thalassemia.

Discussion

Hb D/β-thalassemia patients have a disease that has clinical manifestations ranging from mild to moderate disease, resembling either thalassemia minor or thalassemia intermedia. This case report confirms the benign nature of coinheritance of Hb D-Punjab and β+-thalassemia. In our case, the patient presented with jaundice and hepatosplenomegaly and the patient had a microscopic picture of microcytic hypochromic anemia with normal ferritin levels suggestive of the non-iron deficient status. In another study conducted, a case of Hb D had presented with hemolytic jaundice after a physiological stress of twin delivery, diagnosed as Hb D heterzygote on electrophoresis.[9] In another study, it was recommended that cases of Hb D-Punjab, or any other hemoglobin (Hb) variant appearing as homozygous, are carefully evaluated if microcytic hypochromic parameters not associated with α-thal are present.[10] In our case, electrophoretically hemoglobins showed mobility at the position of Hb D, Hb A_2 and Hb A.RBC indices were in the range of thalassemia as well, confirming that the individual was a compound heterozygote for Hb D / β-thalassemia.

In today's ever changing population demographics with racial inter mixing, hemoglobin D disease should no longer be considered as an entity confined to the south Asian region. It is important for a hematologist to keep this important differential in mind when dealing with any suspected hemoglobinopathy.

Conflict of Interest None

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