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Case Report

Hemoglobin D Disease: An Atypical Presentation

Rai S[1], Sethi G[2], Sethi A[1], Kaur K[3]
[1]Assistant Professor, [2]Professor, [3]Resident, Department of Pediatrics, GGS Medical College and Hospital Faridkot, Punjab.

ABSTRACT
Haemoglobinopathies are characterised by structurally abnormal haemoglobin variants of the normal adult haemoglobin (HbA) which led to increase hemolysis and dependency on blood transfusion for maintaining normal range hemoglobin. HbD is characterized by point mutation of Glu→Gln substitution at codon 121 with a GAA→CAA change at the DNA level on chromosome number 11.
Keywords: Hemoglobinopathy, anemia, electrophoresis, haematological.

Corresponding Author: Dr. Seema Rai, Assistant Professor, Department of Pediatrics, , GGS Medical College and Hospital Faridkot, Punjab.

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INTRODUCTION

In 1951, Itano1 observed new haemoglobin variant, which co-migrated with haemoglobin S (Hb S) at alkaline pH, but failed to sickle, it was labelled as Hb D. Haemoglobinopathies are characterised by structurally abnormal haemoglobin variants of the normal adult haemoglobin (HbA) which led to increase hemolysis and dependency on blood transfusion for maintaining normal range hemoglobin. Haemoglobin C, D, E and S are classical examples of β chain variants. HbD Punjab also known as HbD Los Angeles is a β-chain variant and is characterized by point mutation of Glu→Gln substitution at codon 121 with a GAA→CAA change at the DNA level on chromosome number 11 and the electrophotetic mobility at alkaline pH is similar to HbS (β6, Glu→Val)[1] . Each hemoglobin chain has a molecular weight of 16000 daltons, four of such chains bind together loosely to form one haemoglobin molecule[2]. Hemoglobin D-Punjab can be inherited in heterozygosis as well as homozygosis which affects clinical presentation. Hb D can coexist with other haematological disorder like thalassemias[3]. Hemoglobin D (Hb D) is relatively common in India; its homozygous form is very rare. Hemoglobin D, hemoglobin variant, occurs mainly in north-west India Pakistan and Iran. Hemoglobin D-Punjab occurs in 2 - 3 % of the population of Punjab carries the Hb D gene[4]. Most of the patients with Hb D have mild anemia and mildly enlarged spleen. We are presenting a case of 2 year male with atypical presentation (requiring repeated blood transfusion).

CASE REPORT

A 2 year old male presented with marked pallor started at age of 6 month progressive in nature associated with weight loss, required multiple blood transfusions without any history of jaundice, bruisability, born to parents with non consanguineous marriage at term gestation, birth weight 2250 gms, cried immediately after birth, mental –motor milestone were normal for age and immunized per age. On physical examination:
Pulse rate, respiratory rate within normal limits. Weight 8/11.5 kg, height 75/85 cm, head circumference 50/ 50.5 cm respectively. Pallor present, oedma absent, cyanosis absent, lymphadepathy anterior cervical 1 cm enlarged, nontender, not attached to underlying tissue or overlying skin, clubbing absent. Abdomen distended soft ,nontender hepatomegaly present span 7 cm smooth surface, firm consistency, rounded margins moves with respiration, splenomegaly present 3 cm below left subcoastal margins.respiratory system ,cardiovascular system and central nervous system showed no abnormal finding.
. Laboratory investigations revealed haemoglobin 6.5 g/dl, white count blood cell count 10.6×10^3/uL with 65% lymphocytes, 32% neutrophils and platelets 450000/cumm, peripheral blood film showed microcytic hypochromic, elliptical cells, macrocytes. Ultrasound examination showed hepatomegaly with normal echo texture, splenomegaly with normal echo texture. High performance liquid chromatography Hb adult 62.5%, Hb A2 2.1%, Hb F 1.6% Hb D 27.1%, suggestive of haemoglobin D trait as shown in figure 1.

DISCUSSION

Hemoglobin electrophoresis mobility detects various variant like HbD such as Hb D Punjab, Hb D Iran, Hb D Ibadan and Hb D Bushman. Except for Hb D Punjab, compound heterozygous states of HbS with HbD variants were clinically innocuous. Hb-D Punjab arises from the substitution of glutamine for glutamic acid in the 121st position of Beta heterozygote state for HbD. The electrophoretic mobility of Hb-D on cellulose presented with acetate is identical to that of Hb-S on agar gel[5]. Like the other structural mutations of hemoglobin, haemoglobin D trait is the heterozygous state for hemoglobin D and haemoglobin A, whereas the homozygous state for hemoglobin D is designated hemoglobin D disease. The heterozygous state for hemoglobin D is entirely asymptomatic. The abnormal hemoglobin constitutes between 35 and 50 percent of the total haemoglobin. Homozygous haemoglobin D disease is very rare, and some patients originally believed to be homozygous for hemoglobin D subsequently were found to be heterozygous for
haemoglobin D and β thalassemia[6]. Extravascular hemolysis takes place whenever red cells are rendered less deformable, hemolysis, hemoglobinemia and hemoglobinurea are not observed and the principal clinical features are anemia and jaundice. There is often splenomegaly. Biochemically, 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 associated with a clinical disorder similar to less severe than, sickle cell anemia.Hb D has been reported in association with hematological malignancy such as leukemia and Hodgkin's lymphoma[7]

CONCLUSION

Regular clinical examination of clinically silent hemoglobinopathy should also be recommended as chronic extramedullary erythropoeisis can lead to organomegaly and can make patient blood transfusion dependent.

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