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Hemoglobin SD disease – A case report

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ABSTRACT

Itano first described hemoglobin D in 1951 in a Caucasian family who had hemoglobin S interacting with D. Hemoglobin D was the fifth hemoglobin to be described. Compound heterzygosity for bs/bd results in a severe hemolytic anaemia and clinical syndrome similar to that of sickle cell disease. Here, we report a case of Hb SD Punjab disease. A 13 year old female presented with hemolytic anaemia, hepatosplenomegaly and occasional pain in abdomen. Initially, she was thought to be a case of sickle cell anaemia, however, with the help of HPLC it was confirmed as Hb SD disease. © 2012 Trade Science Inc. - INDIA

KEYWORDS

Hb–hemoglobin;
HbS–hemoglobin S;
HbD–hemoglobin D;
HPLC–high performance liquid chromatography

INTRODUCTION

Hemoglobin (Hb) abnormality is the most frequent genetic disease, affecting approximately 7% of the world population[1]. Hemoglobin S worldwide is the most frequent clinically severe Hb variant[2].

There are many different kinds of hemoglobin genes. The normal hemoglobin gene is called A, however there are over 400 abnormal hemoglobin genes. The most common are S, C, D, O, B-thalassemia because you inherit one gene from each parent it is possible to get many different combinations of genes[3].

In Hb SD, one β-globin gene encodes a chain leading to formation of hemoglobin S (HbS) and the other β-globin gene encodes a β -chain leading to formation of hemoglobin D. HbD, unlike the normal adult HbA, can participate in the polymerization process with HbS, thus leading to atypical hemolytic anaemias with enhanced sickling[4].

CASE HISTORY

A 13 year old female child was admitted to the institute with fever for 1 week and pain abdomen for 1 month. Physical examination revealed short stature, pale tongue, pale spoon shaped nails, enlarged tender spleen 8cm in size and liver 3cm below right costal margin.

INVESTIGATIONS

Haematological investigations of the patient showed Hb 4.10g/dl, RBC count 1.18 mill/mm^3, PCV 12%, MCV 101.70fL, MCH 33.50pg and RDW 37.90% (TABLE 2). Peripheral blood smear showed moderate anisopoikilocytosis, normocytes, teardrop cells, target cells, sickle like cells, ↑ed polychromasia (Figure 1). Total T4 levels was 3.29 μg/dl, total T3 levels was 0.725 ng/ml and TSH levels was 56.7mIU/L.

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TABLE 1 : Hemoglobin HPLC/ electrophoresis of patient, parents and siblings.

Test Name	Karanjit Kaur (Patient)	Partap Singh (patient's father)	Sukhwinder Kaur (patient's mother)	Sukhchain Singh (patient's brother)	Sharanjit Kaur (patient's sister)	Reference Range (% age)
Age	13 years			12 years	18 years	
Hb F	19.70	< 1.00	1.30	< 1.00	<1.00	< 1.50 %
Peak 2	1.30	2.80	3.10	2.80	3.90	< 9.60 %
Hb-Adult	12.30	51.80	55.80	55.50	87.30	83.24-90.79 %
Hb A_2	2.00	2.20	3.30	2.70	2.50	1.50-3.50 %
Hb D	38.70	35.60	---	31.80	------	%age
Hb Sickle	23.30	----	32.80	------	------	%age
Others	2.60	7.20	3.30	6.40	5.50	< 10.00 %

Hb-HPLC showed HbF 19.70%, Hb adult 12.03%, HbD 38.70%, Hb sickle 23.30%; suggestive of haemoglobin SD disease (TABLE 1). When parents were investigated, Hb-HPLC of father showed Hb adult 51.80 % and Hb D 35.60%, hematological index showed Hb 14.10g/dl with MCH 29.90 pg findings suggestive of Hb "D" Trait (heterozygous state) and Hb-HPLC of mother showed Hb adult 55.80%, Hb sickle 32.80%, hematological index showed Hb 11.30g/dl, PCV 35.70% with RDW 15.70%, suggestive of HbS (heterozygous state). Sickling test of both the patient and the mother showed positive results.

TABLE 2 : Hematological Indices of patient, parents and siblings.

Test Name	Karanjit Kaur (Patient)	Partap Singh (patient's father)	Sukhwinder Kaur (patient's mother)	Sukhchain Singh (patient's brother)	Sharanjit Kaur (patient's sister)	Reference Range
Hb	4.10	14.10	11.30	12.50	11.30	11.50-15.00 g/dl
RBC Count	1.18	5.25	4.10	4.65	3.91	3.80-4.80 Mill/mm3
PCV	12.00	42.70	35.70	37.00	34.50	36.00-46.00 %
MCV	101.70	81.50	87.20	79.50	88.30	80.00-100.00 fL
MCH	33.50	26.90	27.50	26.90	28.90	27.00-32.00 pg
RDW	37.90	14.00	15.70	16.70	17.10	11.50-14.50 %
Suggestive impression	Hb SD Disease	Hb D Punjab Trait Heterzygous	Hb S Trait Heterzygous	Hb D Punjab Trait Heterzygous	Normal	

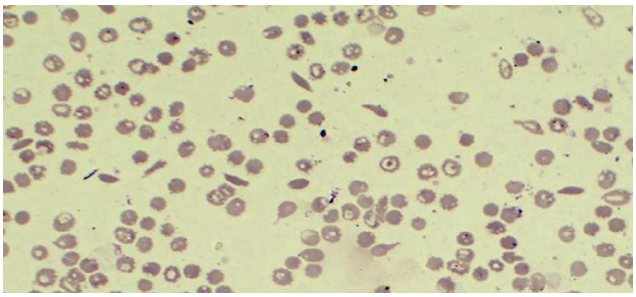

Figure 1 : Peripheral blood smear of patient.

DISCUSSION

Hb D is the fourth most common hemoglobin variant, which developed as a response to the selective pressure of malaria. It is most often found in people living in India, Pakistan, England, Ireland, Holland, Australia, China, Iran, Turkey and their descendants[5]. Hb D Punjab also known as Hb D Los Angeles is a β-chain variant and is characterized by substitution of glutamic acid for glutamine at codon 121 of β- chain. Hb D has been described in both the heterozygous and homozygous states[2].

Homozygous HbDD is rare and a relatively mild disease. (5). In heterozygous states it is seen in combination with HbS and β- thalassemia[6]. HbD interacts with HbS to form a clinically significant condition. This is ascribed to a specific interaction in the fiber due to a possible role of residue 121 in beta chain in stabilising the polymer and thus increasing intracellular polymerization of HbS[7].

The distinction between sickle cell anemia and SD disease is important because of the different prognosis in the two diseases[8].

The electrophoretic mobility of HbD is identical to that of HbS at alkaline pH and absence of sickling of erythrocytes in PBF[9] whereas PBF of HbSD shows the presence of sickle cells.

The patients also suffer from hypothyroidism the reason for this could be repeated transfusions leading to iron-overload-related complications which include insufficiency of the parathyroid, thyroid, and pituitary[10].

CONCLUSION

Due to increased prevalence of some genetic dis-

orders in some racial or ethnic groups like HbD disease, it being the fourth most common Hb variant. HbD Punjab is the only variant with HbD mobility which leads to a severe disease when associated with HbS. Premarital screening and counseling paves the way for a healthy reproductive life for couples. Measures such as health education, carrier screening and pre marital counseling should be explored by various NGO's, in order to reduce the frequency of affected births.

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