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Pregnancy with Haemoglobin SD Disease- A Rare Case Report.

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ABSTRACT

Haemoglobin SD – Punjab is a rare compound heterozygous haemoglobinopathy characterised by presence of two beta globin gene variants : beta 6(GAG to GTG) and beta 121(GAA to CAA). These patients' clinical and haematological features mimic haemoglobin S disease. This case highlights the propensity for occurrence of rare phenotypes and emphasises the importance of accurate diagnosis especially during ANC checkup and to plan an effective patient management strategy before complications evolve.

Keywords: Haemoglobin S D –Punjab, compound heterozygous , haemoglobinopathy.

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 haemoglobin 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] The average frequency of sickle cell disorders in India is 4.3%.The highest prevalence has been recorded in state of Orissa (1-44.4%), followed by Madhya Pradesh (1-40.0%), Andra Pradesh (1-35.7%), Assam (1-35.5%) , Maharashtra (0.8-35.0%), Gujarat (1-31.4%), Kerala (1-30.0%), Uttar Pradesh(1.5-18.5), Karnataka (1-8.0%).[4] Hb D-Punjab is one of the most common hemoglobin variants worldwide, after Hb S and Hb C. It is prevalent in Punjab region, Northwest Indian, with an estimated frequency of 2.0%. In western India, more specifically in the Gujarat region, its frequency drops by one half.[5] Hb D-Punjab is also common in countries such as Italy,[6] Belgium,[7] Austria and Turkey.[8,9] A person with HbSD disease may suffer from anemia and bouts of pain called crisis.

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There also may be problems like frequent infections and unexplained fever. Pregnant women with Hb D disease may have complications varying from mild anemia to frequent pain and infection. Herein, we describe a case of pregnancy with sickle cell disease - D variant and emphasis the importance and need of Hb electrophoresis in cases with moderate degree of anaemias.

CASE REPORT

A 24 year Sikh Punjabi female G2P1L1 presented to antenatal clinic with 8month pregnancy with complaint of dyspnoea while climbing the stairs, palpitation, numbness in extremities and dark yellow coloured urine since 1 month. Past history revealed atleast 2 blood transfusions during previous pregnancy and 1 during current pregnancy. Family history revealed jaundice ++, anaemia++ and history of blood transfusion to her sister. On examination severe pallor and icterus was present .No splenomegaly. Abdominal examination suggested single live intrauterine gestation of about 32 weeks and confirmed by ultrasound. Her investigations showed Hb- 6.0gm%, PCV-17.8%, MCH-27.5pg, MCHC-33.7g/dl, MCV-81.7fl, RDW- 16.3%. Peripheral blood smear showed marked anisopoikilocytosis, microcytes, sickle cells, normocytes. Polychromasia seen, 160 nucleated RBCs/100 WBCs. Reticulocyte count was 22%. Sickling test with 2% metabisulphite was positive. HbS solubility test revealed crystals of HbS with turbid solution. Serum bilirubin (Total) 9.7mg/dl, direct bilirubin-1 mg/dl, indirect bilirubin-8.7 mg/dl. SGOT 110 IU/L, SGPT 78g/L. Routine Urine

examination was NAD but due to raised indirect bilirubin urine for urobilinogen was done which comes out to be positive upto 1:1280 dilutions. Her haemoglobin HPLC showed HbS -35.9%, HbD – 47.4%, HbF-10.9%, HbA-2.1%, and HbA2-2.4 %. HPLC of her husband revealed Beta Thalassemia trait with HbF – 1%, HbA – 80.7%, HbA2 – 4.5%. Though there was history of blood transfusion during previous pregnancy but it was home conducted so she remains undiagnosed.

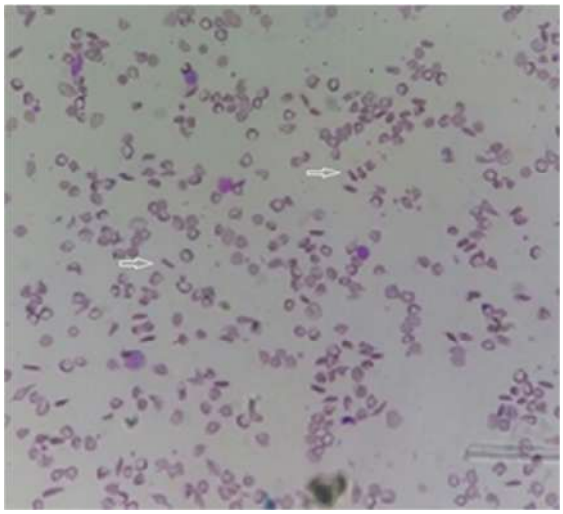


Figure 1: PBF showing sickled cells.

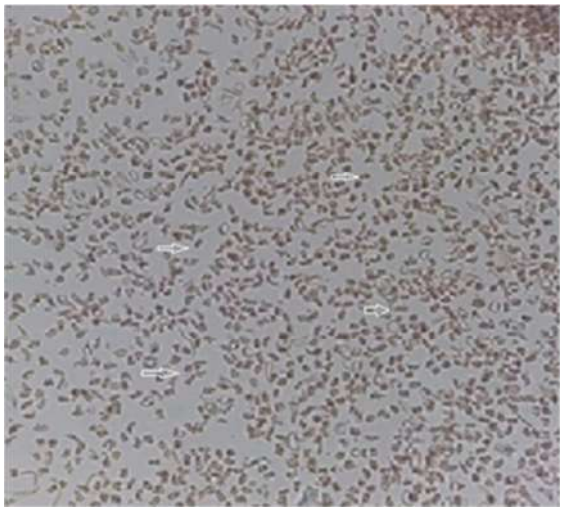
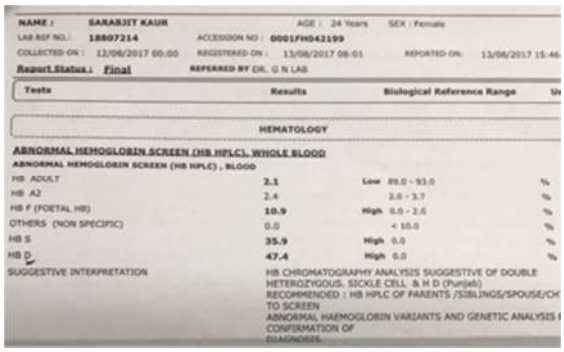


Figure 2: photomicrograph showing sickled cells (arrows) in sickling test by 2% sodium metabisulphite



NAME : SARABJIT KAUR AGE : 24 Years SEX : Female
LAB REF NO.: 18807214 ACCESSION NO : 0001FH042199
COLLECTED ON : 13/06/2017 00:00 REGISTERED ON : 13/06/2017 08:01 REPORTED ON: 13/06/2017 15:46
Report Status : Final REFERRED BY DR. G N LAB

Tests	Results	Biological Reference Range	Units
HEMATOLOGY			
ABNORMAL HEMOGLOBIN SCREEN (HB HPLC), WHOLE BLOOD			
ABNORMAL HEMOGLOBIN SCREEN (HB HPLC) , BLOOD			
HB ADULT	2.1	Low 89.0 - 93.0	%
HB A2	2.4	2.0 - 3.7	%
HB F (FOETAL HB)	10.9	High 0.0 - 2.0	%
OTHERS (NON SPECIFIC)	0.0	< 10.0	%
HB S	35.9	High 0.0	%
HB D	47.4	High 0.0	%
SUGGESTIVE INTERPRETATION	HB CHROMATOGRAPHY ANALYSIS SUGGESTIVE OF DOUBLE HETEROZYGOUS. SICKLE CELL & H D (Punjab) RECOMMENDED : HB HPLC OF PARENTS /SIBLINGS/SPOUSE/CH TO SCREEN ABNORMAL HAEMOGLOBIN VARIANTS AND GENETIC ANALYSIS CONFIRMATION OF		

Figure 3: photograph showing HPLC report

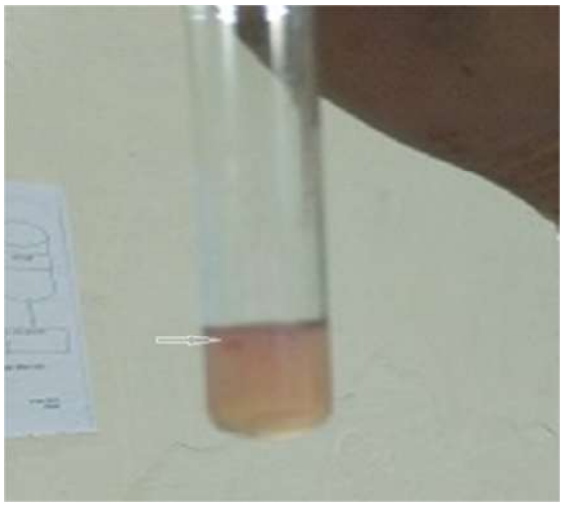


Figure 4: photograph showing HbS solubility test showing crystals of HbS (arrows) with turbid solution

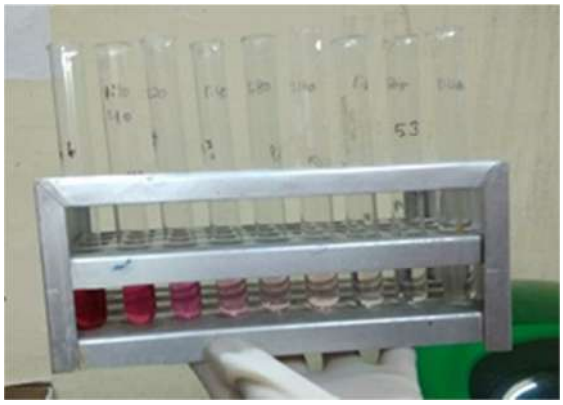


Figure 5: photograph showing positive Ehrlich's aldehyde test for urobilinogen in urine. Test Positive till 1:1280 dilutions

DISCUSSION

Hb SD Punjab disease is a double heterozygous S and D Punjab disorder and is associated with significant sickling of the several variants of haemoglobin D, only Hb D Punjab (Los Angeles) interacts with HbS, however the nature of this interaction is not known.[10] Earlier studies from Pakistan, Iran UAE and Mexico have shown that the clinical presentation of HbSD disease cases is similar to that of patients with severe form of sickle cell anemia. However reports from India have shown variable manifestations of HbSD disease. In HbSD disease HbD does not take part in the sickling process, as patients homozygous for HbD do not sickle.[11] However it is indicated that although HbD itself does not polymerize, it facilitates the polymerization of HbS, thus enhancing the severity of the disease. If one parent has Hb D trait and one parent has sickle cell trait then there is 25%chance in each pregnancy that the child will inherit one haemoglobin D gene and one sickle cell gene leading to this disorder. A person with Hb SD disease may suffer from anemia and bouts of pain

called crisis. There also may be problems like frequent infections and unexplained fever.[12] Pregnant women with Hb SD disease may have complications varying from mild anaemia to frequent bouts of pain, as observed in the present case. Therefore it is advisable that if HbSD disease is confirmed in pregnancy, then the possible complications and interventions have to be explained. Counselling through a genetic counsellor should be done. It is also important to screen the newborn for Hb -SD disease and observe the baby for any splenomegaly or vaso occlusive crisis or repeated respiratory tract infections.

CONCLUSION

HbD Punjabis the only variant with HbD mobility which leads to a severe disease when associated with HbS. Premarital screening and counselling paves the way for a healthy reproductive life for couples. Measures such as health education, carrier screening and premarital counselling should be explored by various NGO.s, in order to reduce the frequency of affected births.

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