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DOUBLE HETEROZYGOUS HbS/HbD INCIDENTALLY DIAGNOSED DURING PREGNANCY -A CASE REPORT

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ABSTRACT
Compound heterozygous for HbS/HbD results in severe hemolytic anemia and a clinical syndrome similar to that of sickle cell disease. Here we report a case of pregnancy with HbSD in 23 yrs old G2P1L1 residing at Nagour district in Rajasthan in rural community presented in antenatal OPD at 35 weeks of pregnancy with complains of bodyache, chest pain and fever for 2 days. On examination she had icterus, pallor and bilateral pitting edema. Her height was 160 cm. and weight were 52 kg. abdominal examination suggested live intrauterine gestation of 34 weeks. Spleen and liver was not palpable.

KEYWORDS: HbS-hemoglobin S, HbD-hemoglobin D, HbSD variant, sickle cell disease.

INTODUCTION
Hemoglobin(Hb) abnormality is the most frequent Genetic disease, affecting approximately 7% of the world population.[1] HbS worldwide is the most frequent clinically severe variant.[2] HbSD is an inherited autosomal recessive variation of HbA that occurs in beta globin protein chain of HbA by substitution of glutamic acid for glutamine at codon 121 of beta chain. In HbSD, 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 hemoglobinD. If one parent has Hb D trait and one parent has sickle cell trait then there is 25% chance with each pregnancy that the child will inherit one hemoglobinD gene and one sickle cell gene.[3] HbD, unlike the normal adult HbA, can participate in the polymerization process with HbS, thus leading to atypical hemolytic anemias with enhanced sickling.[4]

CASE REPORT
A 23-year-old pregnant female (G2P1L1) Nagour district in Rajasthan in rural community presented in antenatal OPD at 35 weeks of pregnancy with complains of bodyache, chest pain and fever for 2 days. Patient has history of jaundice at 13 yrs. of age. 1 units of blood transfusion was given at that time. At previous pregnancy patent developed preeclampsia and patient was delivered by LSCS at 33 weeks and 2 unit blood transfusion was given. Post-delivery period was uneventful.

On examination she had icterus, pallor and bilateral pitting edema. Her height was 160 cm. and weight were 52 kg. abdominal examination suggested live intrauterine gestation of 34 weeks. Spleen and liver was not palpable. Her investigations showed Hb-9.8 gm/dl, MCV-94.7 mm3, RDW-CV-22.9%, corrected WBC count - 15240/mm3. Peripheral blood examination reveals microcytic hypochromic RBCs, moderate anisopoikilocytosis, few macrocytes, many sickle cells, target cells, polychromatic RBCs and 35 NRBCs/ 100 WBCs are seen. Mild neutrophilic leukocytosis. parasite not seen. Reticulocyte count was 22%. Serum total bilirubin 6.26 mg/ dl, direct bilirubin 4.18 mg/dl. Serum LDH was 2258. Coombs test was negative. Hemoglobin electrophoresis by showed – sickle cell -WINDOW—28%, D-WINDOW-34%, C-WINDOW-0%, HbA2 level -2.2%, HbF level –9.7%. These features were suggestive of sickle cell trait and D trait. Patient was delivered at 36 weeks by LSCS. Post LSCS period uneventful and child weight was 2.6gm.

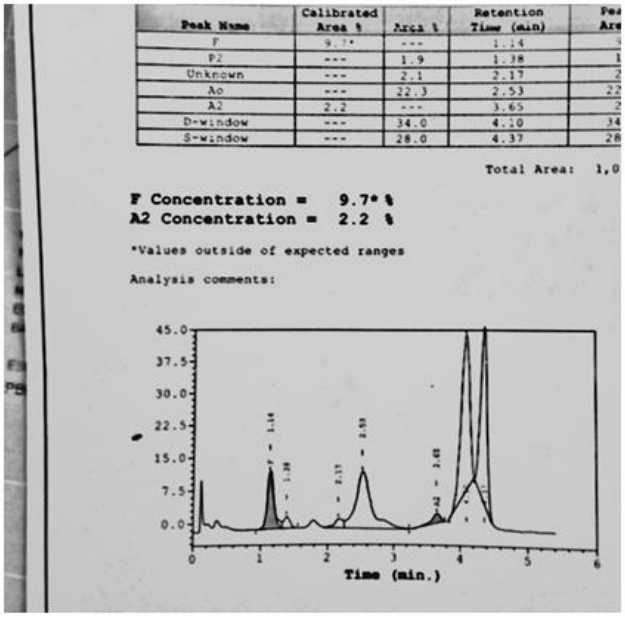

Figure 1: Graphical representation of HPLC results.

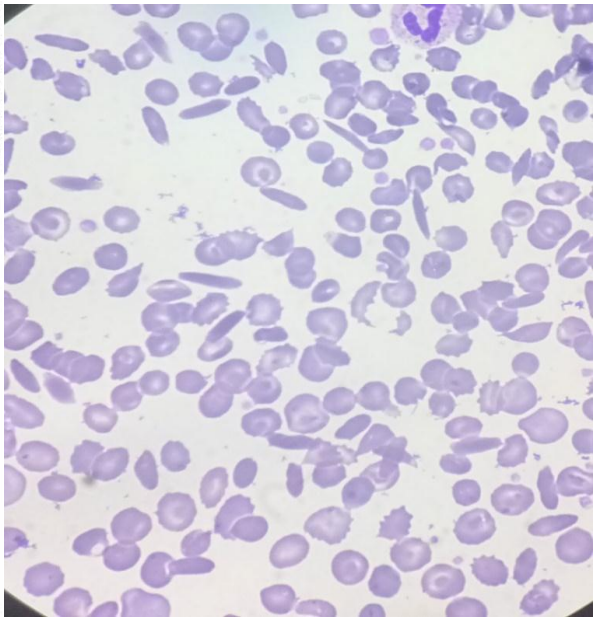

Figure 2: Peripheral blood smear of patient.

DISCUSSION

Hemoglobinopathies are characterized by structurally abnormal hemoglobin variants of the normal adult hemoglobin (HbA). Hemoglobin S, C, D and E are classical examples of β chain variants. Hb S (β6(A3), Glu→Val) variant has a worldwide distribution with highest gene frequencies seen in equatorial Africa, Quatif osases of eastern Saudi Arabia and parts of India. In Malaysia, Hb S variant is seen amongst the Malaysian Indians at a low frequency.[5] HbS heterozygotes (βS/βA) are usually asymptomatic but rarely may be associated with clinical and hematologic manifestation of significance. Homozygotes (βS/βS), however, has enormous clinical importance. Co-inheritance of both hemoglobin S and hemoglobin D, termed HbSD disease, may manifest with mild to moderate hemolytic anemia resembling those of sickle cell anemia. The distinction between sickle cell anemia and SD disease is important because of the different prognosis in the two diseases.[6] Sickle cell - Hb D double heterozygous state- Can mimics sickle cell anemia clinically and hematologically. The electrophoretic mobility of HbD is identical to that of HbS at alkaline pH and absence of sickling of erythrocytes in PBF[7], whereas PBF of HbSD shows the presence of sickle cells. On Hb electrophoresis AS pattern seen and on HPLC D window is seen with retention time of 4.09-4.14 min.[8,9] 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.[10] Pregnant women with Hb D disease may have complications varying from mild anemia to frequent pain and infection. Therefore, it is advisable that if HbSD disease is confirmed in pregnancy, then the possible complications and interventions have to be explained. Counseling through a genetic counselor 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. A pediatric hematologist should be consulted regarding patient evaluation and possible disease management. It is suggested that Good obstetric management is likely to play part in reducing maternal mortality. Exhausting and difficult labors are undesirable, but, as operative procedures are not without risk, caesarean section should not be routinely undertaken.[11]

In considering transfusion, it is important to realize that sickle-cell hemoglobin releases oxygen more effectively than normal hemoglobin[12] and one should not be over alarmed by low hemoglobin levels. Indeed, sudden over transfusion may precipitate crisis by increasing blood viscosity. Further, Labor and immediate postpartum period, when complications are most likely to occur, especially when the patients are not transfused, vaginal delivery instead of caesarean section, be aimed as there is increased risk of preterm labor, fetal distress and still birth in such patients.[13] Prolonged labor, Gestation over 40 wks and routine Caesarean section or induction, if not indicated, should be avoided. Low birth weight of babies of mothers with sickle-cell anemia is said to be due to high fetal wastage is high attributed to intrapartum anoxia.[14,15]

CONCLUSION

It appears that pathological processes leading to complications in pregnant patients with hemoglobinopathies may be multi-factorial and need to be diagnosed accurately in order to prevent possible complications outlined above when the different disease processes are individually considered.

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