

# Medical Case Reports

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ISSN: 0976-8726 & E-ISSN: 0976-8734, Vol. 2, Issue 1, 2011, PP-21-22

Available online at <http://www.bioinfo.in/contents.php?id=93>

## PREGNANCY WITH SICKLE CELL DISEASE –D VARIANT: A CASE REPORT

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Received: October 06, 2011; Accepted: November 02, 2011

**Abstract-** Compound heterozygosity 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 sickle cell disease - D variant. A 25yrs old G2P1L1 residing at Raigad, from tribal community presented for the first time in antenatal OPD at seven months pregnancy with complaints of fever, cough and cold. She had severe anemia, jaundice, splenomegaly and frontal bossing. Initially, she was thought to be a case of severe sickle cell anemia or sickle cell trait. However with the help of alkaline electrophoresis it was confirmed that she is a case of HbS /D disease.

**Keywords** – HbSD variant, sickle cell disease,

**[Key message:** 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 should be kept in mind that patient can have sickle cell crisis and the severity of which is enhanced by HbSD. 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 paediatric hematologist should be consulted regarding patient evaluation and possible disease management.]

### Introduction

Haemoglobins C, D and E are three other common abnormal haemo-globins. The carriers of haemoglobins C, D and E are quite symptomless and have normal haemoglobin. The homozygote states, haemoglobin C, D disease and E disease are chronic hemolytic anaemias of variable severity. They usually manifest with splenomegaly[1]. 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. HbD has been described in both heterozygous and homozygous states as well as in combination with HbS and beta thalassaemia[2]. Simple heterozygous and homozygous individuals are asymptomatic, whereas association with HbS is characterized by a mild to moderate hemolytic anemia. HbS and HbD are the most commonly encountered variants worldwide[3]. In India prevalence of Hb S gene varies from 0-34% in different tribal and scheduled caste groups and the average frequency of Hb D is observed to be 0.86%. Hb D disease (HbDD) occurs when an infant inherits two copies of HbD variant gene, one from each parent, and the chance of which is 25%. 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 haemoglobin D gene and one sickle cell gene[4]. Homozygous HbD is not associated with health problems. A person with HbSD disease may suffer from anemia and bouts of pain called crisis. 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[5].

### Case report

A 25 yrs old G2P1L1 residing at Raigad district from tribal community presented for the first time in antenatal OPD at seven months of pregnancy. She complained of fever, cough, cold and body ache since 8 days. On examination she had frontal bossing of forehead, icterus, severe pallor, bilateral pitting edema feet (grade 2). Her height was 158 cm and weight was 38.8 kg. Abdomen examination suggested single live intrauterine gestation of 28 weeks. Spleen was palpable, but its exact size could not be ascertained. Liver was not enlarged. Her investigations showed Hb-6.2gm%, PCV-19%, MCH-24.3pg, MCHC-32.6g/dl, MCV-74.5um<sup>3</sup>. Peripheral smear showed anisopoikilocytosis, macrocytes, microcytes, hypochromasia, polychromasia, nucleated RBC, pencil cells, tear drop cells schistocytes along with irreversibly sickled cells. Reticulocyte count was 24%. Sickling test was positive and malarial parasite were not seen. Serum bilirubin (Total) 6.19mg/dl, direct bilirubin - 2.09 mg/dl, indirect bilirubin -4.1mg/dl. Her Alkaline electrophoresis of Hemoglobin showed HbS/D - 63.8%, HbF-30.2%, HbA-4.1%, HbA2-1.9%. Her USG showed splenomegaly of 13.4 cm., single live intrauterine pregnancy of 29 wks of gestation, expected baby weight 1.425kg, placenta of grade 3

maturity . Patient had a preterm vaginal delivery after 4 weeks. Baby was kept in neonatal intensive care unit in view of preterm status .Baby's blood was sent for Hb electrophoresis which showed HbS D disease. Sibling's Hb electrophoresis also showed the presence of Hb SD disease.

### Discussion

Hb-D is found sporadically in many parts of the world. It occurs somewhat more commonly in certain regions of India but even there the maximum incidence of the trait is 2% in Sikhs of the Punjab[6]. There are several variants of haemoglobin D such as Hb D Punjab (Los Angeles) Hb D Ibadan, Hb D Iran. Of these variants only Hb D Punjab only interacts with HbS, however the nature of this interaction is not known. HbD has also been reported with other haemoglobinopathies like  $\beta$ -thalassaemia without any additional clinical or hematological abnormalities[7].

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[8]. However it is indicated that although HbD itself does not polymerize, it facilitates the polymerization of HbS, thus enhancing the severity of the disease. Hb D disease (HbDD) occurs when an infant inherits two copies of HbD variant gene one from each parent, and the chance of which is 25%.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 haemoglobin D gene and one sickle cell gene. Homozygous HbD is not associated with health problems. 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[9]. Pregnant women with Hb D disease may have complications varying from mild anemia to frequent pain and infection, as observed in case reported in this article.

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 paediatric 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[10].

In considering transfusion, it is important to realize that sickle-cell haemoglobin releases oxygen more effectively

than normal haemoglobin[11] and one should not be over alarmed by low haemoglobin 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 [12]. Prolonged labor, Gestation over 40 wks and routine Caesarean section or induction, if not indicated, should be avoided<sup>14</sup>. 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 [13,14]

### Conclusion

It appears that pathological processes leading to complications in pregnant patients with haemoglobinopathies 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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