

CASE REPORT

CASE REPORT: A RARE PRESENTATION OF HEMOGLOBIN D TRAIT

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INTRODUCTION: Hemoglobin D Punjab was first discovered in the early fifties in a mixed British and American family of Indian origin from the Los Angeles area; hence it is also sometimes called as "D Los Angeles". Hemoglobin D is the 4th common hemoglobin variance. It developed as a response to the selective pressures of malaria in these regions. The substitution in Hb D is a glutamic acid to glutamine at the 121st amino acid of the beta -globin chain (121Glu Gln). Hb D has an S-like mobility on alkaline electrophoresis but co-migrates with HbA on acid pH. Osmotic fragility may be decreased. Blood films may show target cells. [1]

Two to 3 percent of the population of Punjab carries the Hb D gene. Subsequently, it has also been found in a number of other populations including Europeans, Mediterranean region, and Americans of African descent.[2]

Hb D heterozygotes (Hb D trait) are usually asymptomatic, not anemic, and have normal red cell indices.[3] Hb D homozygotes (Hb D disease) manifests as mild hemolytic anemia and splenomegaly.[4] But here we present a case of symptomatic Hb D trait, who presented with hemolytic anemia and splenomegaly from Hubli, Karnataka which is a rare phenomenon.

CASE: 45 years old male born of non-consanguineous marriage, hailing from Hubli presented with complaints of easy fatigability and exertional dyspnea of 30 days duration and jaundice of 20 days duration. Patient had a history of fever one month back prior to onset of above mentioned symptoms which lasted for one week duration and subsided on its own. Patient also had episodes of jaundice 7 months and 5 months back which subsided on its own without any medication. There was no past history of blood transfusion. Patient also had family history of similar complaints with grandfather and his younger brother (age 30 years) having recurrent episodes of jaundice. (Figure no. 1: pedigree chart) Patient was non-alcoholic and non-smoker. On examination he had severe pallor, icterus and massive splenomegaly

INVESTIGATIONS: Investigation reports revealed Hemoglobin of 4.5 gm/dl, Platelet count of 89,000/mm³, Total WBC count of 4,500 cells/mm³, LFT showed Total bilirubin of 8.2 mg/dl, Indirect bilirubin of 7.2 mg/dl, SGOT of 116 IU/L, SGPT of 45 IU/L, Alkaline phosphatase of 408 IU/L. Peripheral smear showed microcytic hypochromic anemia with few target cells. MCV was 68 fl.

Serum Iron profile showed mildly increased ferritin level of 300 ng/ml. Corrected reticulocyte count was 5%. LDH was 1500 IU. Renal function tests, Urine routine, Prothrombin time, Chest X ray, 2D Echo were normal. Peripheral smear for malarial parasite was negative. Upper gastrointestinal endoscopy was normal and did not show any esophageal varices. Bone marrow showed hypercellularity. Ultrasound of abdomen showed massive splenomegaly. CT abdomen revealed

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multifocal non enhancing hypodense areas seen in the spleen suggestive of splenic infarcts. (Figure no. 2: CT of splenic infarcts) Portal vein diameter was normal.

He was further investigated for hemolytic anemia with G6PD and pyruvate enzyme levels being normal. Coombs test and ANA profile were negative. Osmotic fragility test was normal

His hemoglobin electrophoresis showed abnormal band in HbS/HbD region. (Figure no:3 hemoglobin electrophoresis). Sickling test was negative, suggestive of HbD trait.

Patient was treated with blood transfusion and discharged

DISCUSSION: Though Hb D is not very uncommon in northern parts of India, very few cases have been reported from the south. HbD trait usually does not produce any clinical symptoms or hematological manifestations. [5] HbD disease is usually associated with mild hemolytic anemia and splenomegaly. Coinheritance with HbS mimics sickle cell disease [6] Our patient had HbD of only 16% with tests for sickling and hemoglobin electrophoresis not suggestive of associated sickle cell disease. Our patient with Hb D trait manifested with hemolytic anemia and massive splenomegaly. The presence of splenic infarcts in the absence of associated sickle disease is also a rare phenomenon.

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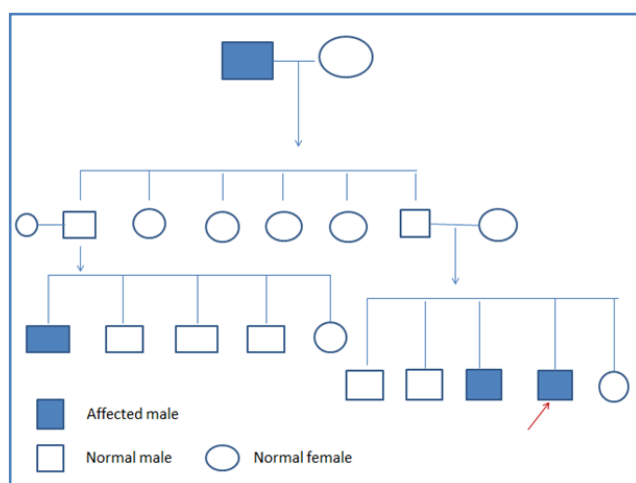


Fig. 1: Pedigree chart

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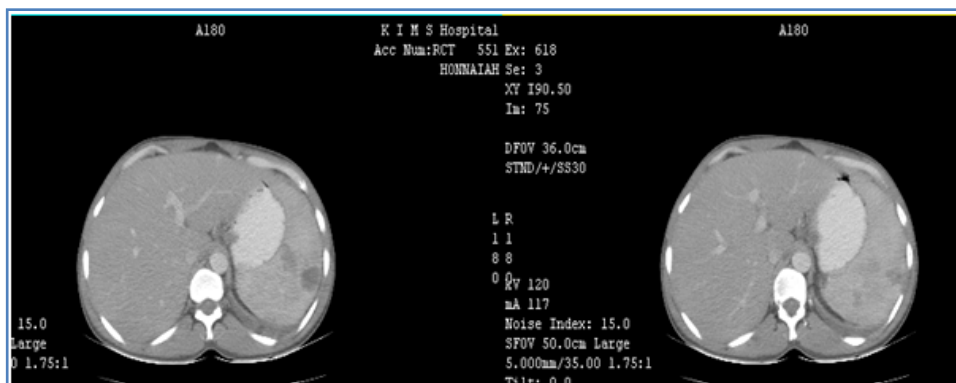


Fig. 2: CT abdomen showing multiple hypo echoic lesions in spleen suggestive of infarcts

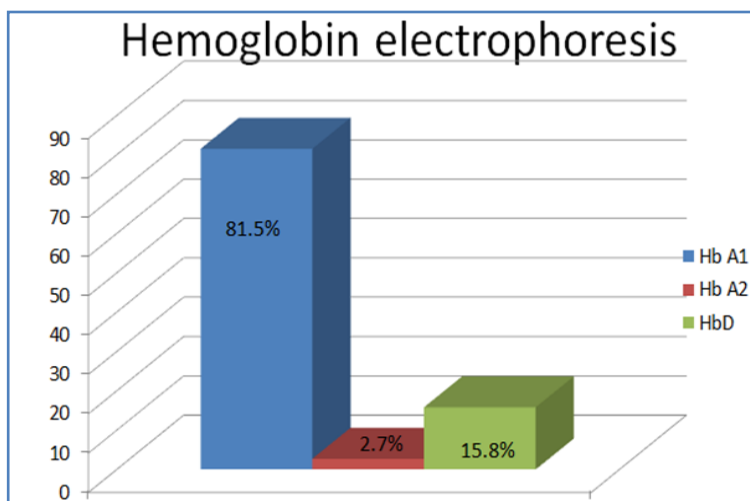


Fig. 3: Hemoglobin electrophoresis

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