Case Report
Haemoglobin Defect, HbSD, with Recurrent Episodes of Jaundice

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Introduction
HbD Punjab, also known as HbD Los Angeles is a β-chain variant of HbD. It is characterized by Glu-Gln substitution at codon 121 with a GAA→CAA change at the DNA level and electrophoretic activity at alkaline pH, which is similar to HbS. Out of the 16 variants, HbD Punjab is the most common HbD variant. HbD has been described in homozygous and heterozygous forms. Heterozygous forms of HbS or HbD are usually silent. HbD Punjab interacts with HbS to produce mild haemolytic anaemia but cases have been reported with severe haemolytic anaemia and splenomegaly. It occurs in 1-3% of Western population. In the Indo-Pak subcontinent, HbD has been observed in 0.86% of the population, with a frequency of 3.6% in Punjab.1,2 We describe a case report from Rawalpindi, Punjab-Pakistan, of HbSD disease with recurrent episodes of jaundice.

Case Report
A 50 year old married woman presented in February 2010 with increasing yellowish discoloration of sclera, with no history of itching or clay colored stools. She gave history of repeated episodes of jaundice in the past, more so in summers, with complete recovery each time. She is a known Type 2 Diabetes Mellitus patient on insulin 70/30. There was no history of intravenous drug abuse or alcohol intake. There was no family history of hepatitis or hereditary blood disorders. On examination, she was found to be pale and icteric. Pulse was 68/min, blood pressure 140/90 mmHg and she was afebrile. Abdominal examination revealed a lax, non-tender abdomen with no visceromegaly or free fluid. Her initial workup showed microcytic hypochromic anaemia with marked red cell anisocytosis, target cells and poikilocytosis. Reticulocyte count was 3% and MCV 52. LFTs showed bilirubin of 9.7mg/dl, ALT of 952, while ALP was 41 and Gamma GT 142.

Patient was investigated for infective and autoimmune hepatitis, but relevant tests were negative. Abdominal Ultrasound showed multiple small stones in the gall bladder with normal liver parenchyma and biliary tract, with no signs of obstruction. Hb Electrophoresis showed a strong band in HbSD; 98%. HbA2 was 2.8% and there were no HbA or HbF bands. Diagnosis of HbSD disease was made and patient advised regular follow up with conservative treatment. During follow up, she was found to have bilateral pedal oedema. Investigations showed a bilirubin of 6.0 mg/dl, ALT of 147, and an alkaline phosphatase of 340. Synthetic functions of the liver were disturbed with a prolonged Prothrombin Time and decreased serum albumin levels. There was no history of diarrhoea. Prolonged jaundice, causing chronic cholestasis, leading to malabsorption was the only plausible explanation. On follow up, her symptoms had completely resolved. There was no clinical evidence of jaundice and malabsorption. Her bilirubin had returned back to normal along with the rest of investigations.

Discussion
This particular case of HbSD disease is an adult, while HbSD is usually seen predominantly in children. Prolonged repeated episodes of jaundice, elevated liver enzymes and malabsorption are the salient features of this case. Patient’s presentation is her complete spontaneous recovery after every episode of jaundice.

HbD Punjab is the most common variant of HbD. It is the only variant which interacts with HbS. There are case reports of HbSD disease with moderate to severe haemolytic anaemia and hepatosplenomegaly. In HbSD disease, HbD does not sickle but facilitates sickling of HbS.3,4 Moreover, enhanced levels of HbF have inhibitory effect on clinical expression of the disease. In our patient, absence of HbA and HbF can account for the severity of her anaemia.

References