

## HAEMOGLOBIN S-D DISEASE IN A BAHRAINI CHILD

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Four month old Bahraini infant presented with severe haemolytic anaemia and was found to be double heterozygous for haemoglobins S and D. This condition has resulted in a severe clinical expression at a very early age unlike sickle cell disease in Bahraini children. Bahrain Med Bull 1995;17(4):

A number of red cell genetic traits such as sickle haemoglobin, thalassemias, red cell G6PD deficiency and hereditary elliptocytosis are prevalent in the Bahraini population<sup>1,2</sup>. Thus double heterozygosity for these conditions is a common occurrence. Another abnormal haemoglobin, HbD is not prevalent in this population. We report here a rare instance of an infant with both HbS and HbD coinheritance.

### THE CASE

Four months old male child was admitted to Salmaniya Medical Centre through the Accident and Emergency section, with fever, cough and pallor. Child was a product of full term normal delivery and fifth child in the family of Bahraini parents. There was no significant family history. On clinical examination the child was pale, dyspnoeic with 38° body temperature but active. There was no jaundice or lymphadenopathy. His spleen was enlarged 8 cm and liver 4 cm below the left and right costal margins respectively.

Haematological investigations showed Hb 4.4 g/dl, haematocrit 14.4 l/l, total red cell count 1.68 X10<sup>12</sup>/l, total WBC count 17.2 X10<sup>9</sup>/l, platelets 57 X10<sup>9</sup>/l, reticulocyte 15.0% and circulating nucleated red cells 170/100 leukocytes. Peripheral blood smear showed severe anisopoikilocytosis, hypochromia, microcytes, target cells, and ligamented cells. Foetal haemoglobin level and G6PD screening test were normal. Sickling test was positive and electrophoresis at pH 8.4 showed two major bands at positions HbS and HbF. There was no HbA band (Fig 1). The provisional diagnosis of either HbSS disease or HbSB thalassemia was made. When parents were investigated, the father had a pattern of AS in the electrophoresis but sickling test was negative. He was confirmed to have HbD trait on acid agar gel electrophoresis. The mother showed

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AS pattern in electrophoresis and was positive for sickling thus confirming her to be HbS trait. This led us to retest the infant's haemoglobin on acid agar gel which showed the presence of both HbD and HbS, thus confirming him to be a double heterozygous for HbS and HbD (Table 1).

Table 1  
 Relevant haematological data of the patient and his parents

	Sickling Test	HbElect at pH 8.6	HbElect at pH 6.0	HbS %	HbD %	HbF %
Patient	Positive	F S	F S D	11.3	17.3	71.4
Father	Negative	A D	A D	Nil	46.3	<1
Mother	Positive	A S	A S	46.5	Nil	<1

## DISCUSSION

HbS is highly prevalent among the Bahrainis. The incidence is around 10%. HbD however is a very rare among the Bahrainis. This communication reports simultaneous occurrence of HbS and HbD in a Bahraini child for the first time. On routine alkaline haemoglobin electrophoresis the condition is similar to HbSS disease manifesting in a single major haemoglobin band in the position of HbS, but in haemoglobin electrophoresis at acid pH the two abnormal haemoglobins were demonstrated.

Both Hbs S and D are asymptomatic when present as traits<sup>3-4</sup>. In homozygous form Hb D is almost always asymptomatic<sup>5</sup> and homozygous S has a mild presentation at this age in the Bahrainis. In contrast the double heterozygosity of HbD and HbS is associated with moderately severe haemolytic anaemia with crisis<sup>6,7</sup>. There are several HbD of which only HbD Punjab (B121 Glu-Glu) interacts with HbS to form a clinically significant condition<sup>5,6</sup>. This is ascribed to a specific interaction in the fiber due to a possible role of residue 121 in beta chain in stabilising the polymer and thus increasing intracellular polymerization of HbS<sup>8</sup>. Thus abnormal haemoglobins resulting from substitution at position 121 of the beta globin chain when co inherited with HbS enhance the sickling phenomenon<sup>9</sup>.

Thus our case presented with clinically significant disease at a very early age of 4 months.

## CONCLUSION

This is the first case of Hb S-D disease in Bahrain and additional cases may be found in other parts of the Arabian Gulf.

## REFERENCES

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