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BETA GLOBIN GENE HAPLOTYPES IN BAHRAINI PATIENTS WITH SICKLE CELL ANAEMIA

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Molecular genetic studies were undertaken to determine the haplotype of chromosomes carrying the sickle cell allele in Bahraini patients, and hence allow consideration of the possible source of these alleles. A total of 59 individuals from 19 families were studied. Of these, 35 were affected with sickle cell anaemia, and 24 were carriers.

Haplotypes were investigated by PCR amplification of globin target sequences followed by restriction digestion using HindIII, AvaII, HindII, and HinfI polymorphism.

In the 19 families the Bs gene was found to be linked to the haplotype +++++- (also known as the Asian haplotype) in 33 chromosomes (90%), to the haplotype +-+-++ known as the S2 haplotype in 2 chromosome (5%), to haplotype S1 (--++++) in one chromosome (2.5%), and to the haplotype --+--+ found in association with beta thalassaemia in one family (2.5%).

Our study shows that the Asian haplotype is predominant in Bahrain (90%). This haplotype has previously been found to be linked to a benign sickle cell anaemia. The African haplotype S1 was found in one family only.

Until the 1930s malaria was endemic in Bahrain, and complete eradication was not achieved until the 1970s. Selective pressure exerted by malaria may have contributed to the high prevalence of haemoglobinopathies in the country. It was found that one Bahraini in ten carries the Bs gene[1].

The clinical picture of Sickle Cell Disease (SCD) shows wide variability. Many patients have an illness characterised by severe anaemia, recurrent vaso occlusive episodes, end organ failure and increasing susceptibility to infection. In contrast in most patients from Bahrain, the Eastern province of Saudi Arabia, Iran, Kuwait and India, the disease is clinically mild with only moderate haemolytic anaemia[2-5,19]. Although the factors that modify the clinical picture of the disease are not fully understood, there is now good evidence that the co-existence of alpha thalassaemia gene and also a high level of HbF protect against many manifestations of SCD, probably because HbF interferes with Hbs polymerisation[2-5].

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