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Molecular Basis of Benign form of Sickle Cell – B thalassemia Syndrome in two Bahraini Patients

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Objective: The aim was to study the different molecular determinants that might cause an extremely mild form of sickle cell beta thalassemia syndrome among our population.

Method: Two Bahraini students belonging to two unrelated families with normal clinical picture were noticed to have sickle cell beta thalassemia syndrome through hemoglobin electrophoresis. Different molecular genetic techniques were employed to study blood samples from these girls, namely, the polymerase chain reaction-restriction fragment polymorphism (PCR-RFLP), denaturing gradient gel electrophoresis (DGGE), and differential PCR amplification.

Result: Three different molecular determinants were found in these students for the beta – globin gene: Compound heterozygosity for the sickle cell mutation and nt 88 (C→ A) mutation. Haplotype were shown to be the Saudi-Indian haplotype for the sickle cell mutation and haplotype No. IX for nt – 88 (C→ A) mutation. Alpha- globin gene mapping revealed homozygosity for the rightward deletion (--$\alpha^{3.7}$/ -$\alpha^{3.7}$/) for both students.

Conclusion: different molecular determinants were found in association with this mild form of sickle cell β-thalassemia disease: namely inheritance of mild β+ thalassemic mutation, HbS haplotype- associated high HbF expression, and coinheritance of α-thalassemia. All of these modulators were found to give a mild state of sickle cell disease in our patients. This indicates that, molecular diagnostics techniques are of invaluable importance in giving a precise and definitive diagnosis, and to predict the clinical manifestation.

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