

Molecular Basis of α -Thalassemia in Bahrain

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Objectives: This study was designed to delineate the molecular lesions, on DNA level, that lead to α -thalassemia in the population of Bahrain.

Methods: Various polymerase chain reaction (PCR)-based methodologies were involved, namely, differential PCR amplification, PCR-restriction fragment length polymorphism (PCR-RFLP), and direct PCR-amplified genomic DNA sequencing.

Results: Five α -thalassemia determinants were identified. These include three deletional type, the rightward 3.7 kilobase (kb) deletion, the leftward 4.2 kb deletion, and the pentanucleotide deletion in 5' splice donor side of intron I in $\alpha 2$ -globin gene (GGTGAGG \rightarrow GG-----), and two nondeletional α -thal determinants, the Saudi type polyadenylation (polyA) signal mutation in the $\alpha 2$ -globin gene (AATAAA \rightarrow AATAAG), and the Turkish type polyA signal mutation (AATAAA \rightarrow AATGAA), also in $\alpha 2$ -globin gene.

Conclusion: Three α -thalassemia mutations, the Saudi type polyA signal mutation, the pentanucleotide deletion and the rightward 3.7 kb deletion, account for 97% of all α -thalassemia determinants in Bahrain.

Recommendations: A well-tailored genetic counseling approach, supported by molecular studies, is advised for family members affected with α -thalassemia, and in particular for carriers of the polyA signal mutations.