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Spectrum of β-Thalassaemia Mutations in Bahrain♣

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Objectives : To study the molecular characterization of β-thalassemia defects among Bahrainis.

Methods : We used a variety of polymerase chain reaction (PCR)-based procedures including reverse dot blot (RDB), denaturing gradient gel electrophoresis (DGGE) and DNA sequencing, to study the β-thal mutation in 87 Bahraini individuals from 51 unrelated Bahraini families.

Results : Thirteen different β-thal mutations were identified. Four mutations (Intervening Sequence I (IVSI)-3' end (-25 base pairs (bp)) deletion; Codon (Cd) 39 (C→T) and IVSI-5 (G→C), account for 80% of all β-thal alleles.

Conclusion : We conclude that IVSI-3' end (-25bp) deletion is the major β-thalassemic allele in Bahrain.

Recommendations : Based upon our findings, a preventive approach of β-thalassemia needs to be embloyed for the Bahraini people. This study can be used in implementing a cost effective strategy for screening and diagnosis of Beta thal among Bahrainis.

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