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The molecular analysis of β -thalassemia mutations in northeast Egypt

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Background: β -Thalassemia (β -thal), the most common genetic disorder in Egypt is widely distributed particularly in Mediterranean and Middle Eastern countries. β -Globin alleles were studied by different molecular methods which are known to be difficult, time consuming and more liable to contamination. The β -Globin StripAssay method is reported to be rapid, simple, reproducible and less expensive.

Aim: Our study aimed to evaluate the β -Globin StripAssay method based on reverse hybridization for detection of β -thal mutations in Egyptian children living in northeast Egypt and to detect possible genotype/phenotype correlation.

Subjects & Methods: Forty (40) children with β -thal major (20 males and 20 females) with mean age of 10.33 ± 4.75 years were recruited consecutively from outpatient Hematology Clinic of Mansoura University Children's Hospital, Egypt. In addition to full history, clinical and routine laboratory evaluation, mutation analysis was performed by the β -Globin StripAssay MED (Vienna Lab, Vienna, Austria).

Results: The most common genotypes encountered were; homozygous IVS 1.110 (6 patients, 15%) and IVS 1.1 (5 patients, 12.5%) while compound heterozygous genotypes were detected in the remainder mainly IVS 1.110/IVS 1.6 (8 patients, 20%), IVS 1.110/IVS 1.1 (5 patients, 12.5%). The most frequent mutant alleles detected were; IVS 1.110, IVS 1.1 and IVS 1.6, accounting for 33.75%, 27.5% and 18.75%, respectively; while the least frequent was Codon 39 that represented 2.5% of recovered alleles. The detection rate of the used method in our population was 90% where 8 alleles out of 80 (10%) remained uncharacterized. No genotype/phenotype correlation was demonstrated in studied patients.

Conclusion: β -Globin StripAssay is a fast, easy-to-perform and reliable method for genetic screening of β -thalassemia patients in Egypt. IVS 1.110, IVS 1.1 and IVS 1.6 are the most frequent mutant alleles with poor phenotype/genotype correlation.

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