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Prevalence and Spectrum of β-Thalassemia Mutations in Baghdad, Iraq: Data from the Premarital Screening Program

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Abstract

The knowledge of the prevalence and molecular basis of β-hemoglobinopathies constitutes an important prerequisite for an effective prevention program. To address this issue in Iraq's capital, Baghdad, a total of 12526 individuals (6263 couples) attending three main Premarital Screening centers were enrolled. Individuals were labeled as β-hemoglobin disorders based on full blood counts and high-performance liquid chromatography. For those identified as β-thalassemia trait, molecular characterization was achieved by multiplex PCR and reverse hybridization, followed by next-generation sequencing where appropriate. The prevalence of β-thalassemia and δβ-thalassemia traits were 3.5% and 0.01% respectively. For structural variants: sickle cell, hemoglobin D, C, and E traits were documented in 0.37%, 0.07%, 0.05%, and 0.04% respectively. Twenty-two couples were identified as couples at risk of having affected babies with hemoglobinopathies (3.5/1000). A total of 23 different β-thalassemia mutations were identified in studied samples, the eight most frequent of which were IVS-II-I (G > A), IVS-I-110 (G > A), IVS-I-6 (T > C), Codon 44 (-C), IVS-I-5 (G > C), IVS-I-1 (G > A), IVS-I-130 (G > C), and IVS-II-745 (C > G), accounting for 74.7% of the total mutations. In conclusion, the study illustrates the heterogeneity of β-thalassemia mutations in Iraq's capital, and identified several service indicators for prevention.

Accordingly, it constitutes an important step in the setup for an effective prevention program of hemoglobinopathies.

Keywords: Baghdad; Iraq; premarital program; prevalence; β-thalassemia.