Genotype Haemophilia Screening Program Identified Two Novel Variants Including a Novel Variant (c.5816-2A>G) Causing Pathogenic Variant of Factor 8 Gene

Abstract:

Establishing a national screening program for hemophilia patients is highly encouraged by the World Health Organization and the World Federation of Hemophilia. Hence, this study aimed to analyze the variant spectrum of *F8* and *F9* genes in Arab hemophilia patients. Molecular genetics and sequencing studies were performed on a cohort of 135 Saudi hemophilia patients. Out of all screened hemophilia patients (97 hemophilia A and 39 hemophilia B), 15 (11.1%) were positive for inversion 22 and 4 (3%) for inversion 1. Out of a total of 32 (23.7%) substitution/deletion mutations, 2 novel variants were identified: a novel splice acceptor site missense mutation (c.5816-2A > G) causing a pathogenic variant of the *F8* gene and another splicing site point mutation in intron/exon 23 (g.164496G > A). The frequent *F8* variants were (c.409A > C, p.T137P) in exon 4, (c.760A > G) in exon 6, and (c.1835G > C, p.R612P) in exon 12, while the frequent *F9* variants were (c.580A > G) in exon 6 and (c.880C > T) in exon 8. These study data will enrich the spectrum of the genetic databases in the Arab population that could be applied in the future for national genetic counseling.

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