عنوان مقاله:

Complete Genotype and Clinical Phenotype of Hemophilia B: A Study on Iranian Patients

محل انتشار:

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خلاصه مقاله:

Background: Hemophilia B which refers to the deficiency or functional defect of factor IX (FIX) is typically an X-linked bleeding condition that arises from heterogeneous mutations of the FIX gene (F۹). The number of hemophilia cases in Iran is considerable and currently, about ۱۱۱۸ Iranian patients are suffering from hemophilia B, although a small number of them underwent genetic investigations. Here we assessed molecular defects and also laboratory and clinical findings of ۱۰ Iranian cases with hemophilia B.Materials and Methods: A total of ۱۰ cases with hemophilia B were enrolled in the study. Patients were clinically examined by a hematologist and their previous medical documents were surveyed carefully. Routine coagulation tests and FIX activity and antigen assays were performed for the studied patients. Genotyping of F۹ for identifying genetic mutations was conducted by the Sanger sequencing method following PCR amplification of the promoter region and all the eight exons of the F۹ gene.Results: The mean age of patients was ۴ years (۹ months to ۱۶ years) and consanguinity was reported in ۸۰% of cases. Patients were commonly manifested by hematoma (۹۰%), epistaxis (۸۰%), and hemarthrosis (۷۰%) and the severity of the disorder was severe (۷۰%) or moderate (۳۰%). In nine out of ۱۰ patients a genetic defect in F۹ gene we detected including three missense (c.۳۰۴T>C, c.۱۰۰۷T>A, c.۱۹۱G>A) and three nonsense mutations (c.۸۹۲C>T, c.۸۸۰C>T, c.۱۱۱۳C>A). Based on the FIX variant database (http://www.factorix.org), five mutations have been reported previously, but mutation c.۱۰۰۷T>A (p.Ile۳۳۶Asn) seems to be a novel mutation.Conclusion: Our results indicated the heterogeneous molecular defects of hemophilia B in Iran, as recorded in the FIX mutation database. Moreover, no specific genotype-phenotype association was observed in studied subjects.

کلمات کلیدی:

Hemophilia B, Factor IX deficiency, Genotyping, Mutation, Clinical phenotype