عنوان مقاله:

Prevalence of intron 22 inversion mutation of FVIII gene in Iranian severe hemophilia A patients

محل انتشار:

کنگره بین المللی علوم و مهندسی (سال: 1396)

تعداد صفحات اصل مقاله: 9

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خلاصه مقاله:

Background & Aims: Hemophilia A is a common disorder of blood coagulation caused by deficiency of factor VIII with X-linked recessive inheritance affecting approximately 1 in 5000-10,000 male births worldwide. Hemophilia A caused by a heterogeneous spectrum of molecular defects in factor VIII gene .The most common mutation is the intron 22 inversion which can be analyzed by several methods, such as; Southern blotting , LD PCR or IS-PCR. This study aimed to identify the prevalence of intron 22 inversion mutation in Iranian patients with hemophilia A using IS-PCR method.Materials & Methods: This study performed on 30 hemophilia A patients. After extracting DNA from peripheral blood leukocytes by salting out method , Inverse shifting PCR was performed to detect intron 22 inversion and then PCR products were analyzed on 1/5% agarose gel electrophoresis.Results: According to results , in 20 patients with severe hemophilia A ,7/20(35%) were found to have the Intron 22 inversion . 28% (7/2) of patients had a inversion type I and 72% (7/5) inversion type II. Conclusion: The results of this study showed that the frequency of the intron 22 inversion mutation in Iranian sever hemophilia A patients were almost similar with those found in other population but the frequency of inversion type II were significantly higher than frequencies of other reporting population. Also this study demonstrate that using Inverse shifting PCR is a precise and rapid method for assessment of intron 22 .inversion mutations in hemophilia A patients

کلمات کلیدی:

Hemophilia A , Factor VIII , Intron 22 inversion , IS-PCR