

عنوان مقاله:

Relationship between factor VIII genetic pattern and presence of mild hemophilia, severity of bleeding

محل انتشار:

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خلاصه مقاله:

We aimed to assess mild hemophilia and relationship between blood parameters and genetic pattern and bleeding in patients with mild hemophilia. This cross-sectional descriptive-analytic study was performed on 50-member family was screened for people with mild hemophilia. To detect gene mutations in patients, peripheral blood was taken DNA was extracted to assess DNA variant of factor VIII by the PCR method and finally DNA sequences was determined using the ALFexpress auto read sequencing. The rate and severity of bleeding was evaluated using a special questionnaire. Out of 50 subjects studied, 32 (64.0%) including 16 men and 16 women were carriers for hemophilia, while 18 (36.0%, 13 men and 5 women) were normal for the disease. The mean level of factor VIII was significantly lower in those suffering mild hemophilia as compared to normal people (23.69 ± 4.30 versus 73.39 ± 3.42 , $p < 0.001$). Similarly, the mean PTT was also higher in mild hemophilia group than in healthy group (34.00 ± 1.69 versus 26.95 ± 0.60 , $p < 0.001$). Most cases with mild hemophilia had no bleeding or suffering mild bleeding. There was no significant correlation between severity of bleeding and the level of factor VIII in mild hemophilia. The gene analysis could confirm the genotype of hemophilia in those patients who were homozygous for factor VIII gene. Lowering factor VIII with overexpression of its related gene is dominant in patients with mild hemophilia; however, it may not be associated with the severity of bleeding in such patients.

کلمات کلیدی:

Hemophilia, Factor VIII, Genetic Pattern

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