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Molecular diagnosis of Factor VII gene in 5 Iranian families

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Introduction

Congenital factor VII (FVII) deficiency is a rare autosomal recessive coagulopathy. Pathologic mutation in **FVII gene** causes coagulation defect via decreasing biosynthesis or synthesis of dysfunctional molecule of FVII and consequently reduced ability of FVIIa to activate other coagulation factors.

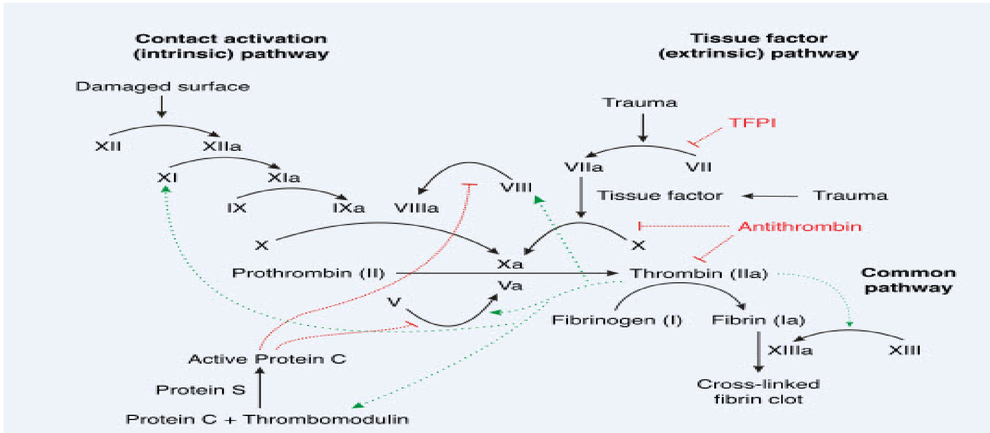
Material &methods

Molecular investigation on patients were done by checking the whole FVII gene including exons and exon-intron flanking sites using **direct DNA sequencing.** Also gained results were confirmed via **linkage analysis**.

Result

Five unrelated families with at least one affected patient who were result of **consanguineous marriage** were investigated. All cases had normal platelet count, prolonged Prothrombin Time(PT), International Normalized Ratio INR (>5) and normal Activated Partial Thromboplastic Time APTT. FVII activity in all cases was less than 1%. Different missense mutation named **c.288G>T, c.635G>A, c.790Del C, c.910G>A** in **exons 2, 6, 7 and 8** respectively were found in patients in homozygote state. The **c.790Del C** was **the most common** mutation and **c.288G>T hadn't been reported previously**.

To confirm the findings ,the **parents hetrozigosity** were checked.



Mutation	Exon	Frequency
c.790Del C	E7	50%
c.635G>A	E6	25%
c.288G>T	E2	16.7%
c.910G>A	E8	8.3%

Conclusion

According to the high rate of consanguineous marriage in Iran, molecular investigation of FVII gene in the families with FVII deficiency history can be effective in decreasing frequency of FVII deficiency in newborn baby and increase healthy baby.