RECOGNIZING 5 NOVEL MUTATIONS IN IRANIAN FAMILIES AFFECTED BY FACTOR VII DEFICIENCY

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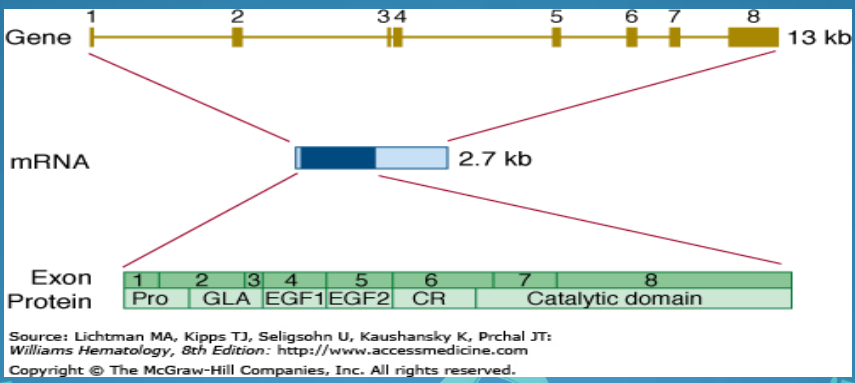
Introduction:

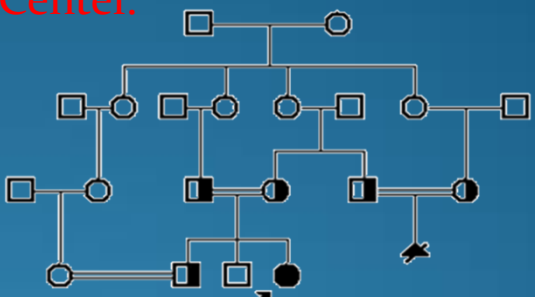
- Factor VII deficiency is a rare autosomal receive disorder involving in blood clotting in the coagulation cascade
- F7 encodes a vitamin k–dependent factor which is critical in hemostasis.
- The factor VII gene locus is on chromosome 13 (13q34).



Source: Lichtman MA, Kipps TJ, Seligsohn U, Kaushansky K, Prchal JT: *Williams Hematology, 8th Edition*: http://www.accessmedicine.com

Material and methods:

- In the present study, mutations in factor VII gene were analyzed in a total of 26 Iranian families referred to Kawsar Human Genetic Research Center.

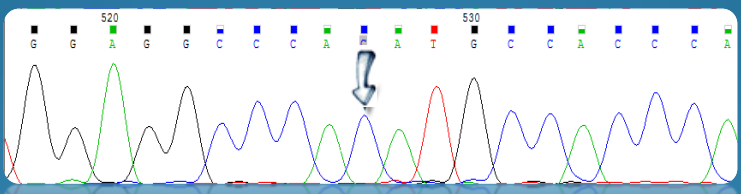


- DNA extraction was done using salting out procedure.
- Haplotype analysis was performed in all family members using short tandem repeat (STR) markers.
- All exons and intron boundaries of the factor VII gene were sequenced using Sanger sequencing.
- . Evaluating the pathogenicity of the novel mutations was done by online soft wares such as Sift, Polyphen-2, Mutation Taster, Hope .

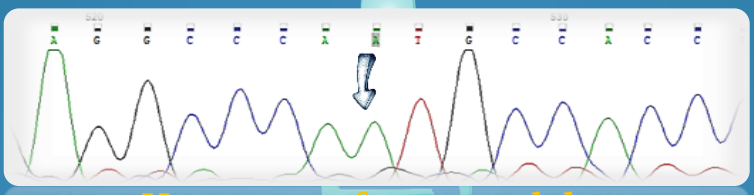
Results

after observing segregation of the disease with FVII gene in the families, the gene was sequenced. It was revealed novel mutations in 3 different kinds including three missense in exons 2-4, one nonsense in exon 7, and one deletion mutation in exon 8.

According to the above soft wares the mutations were all pathogenic ones.



Normal for c.1220delC



Homozygot for c.1220delC

Conclusion

the missense mutations might disrupt the protein structure and the nonsense and deletion caused releasing downstream part of the protein and abolished its function.