Molecular genetic study of Factor V deficiency in two Iranian families

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Factor V deficiency is a rare autosomal recessive disorder

- Caused by mutations in F5 gene

(Factor V antigen levels)

- FV levels below 10 to 15% → Severe FV deficiency
- FV plasma levels > 20 to 30% → Mild to moderate FV deficiency

(Factor V deficiency phenotypes; Genotype to phenotype)

F5 gene is located on chromosome 1q23-24

- Contains 25 exons

The Most Common Symptoms

- Nose bleeds
- Easy bruising
- Bleeding following surgery
Material & methods

Our Cases

 ✓ The levels of Fv antigen was Low (FV=1 & FV<1) in affected individuals

 ✓ These families were referred to Dr.Zeinali medical genetic laboratory for confirmation of the clinical diagnosis & carrier detection for other family members

1. Sample collection
2. Genetic Counseling
3. Questionnaire and consent form
4. Extraction of DNA Using Salting Out Method
5. Direct sequencing
6. Pathogenicity prediction of identified variants
Results & Discussion

✓ Two different mutations in exons 5 and 10 of the F5 gene
✓ These mutations were Missense mutations

The First Family

- Father (heterozygote)
- Mother (heterozygote)
- Affected individual (homozygote)
- Proband (heterozygote)

The second family

- Affected individual (homozygote)
- Proband (homozygote)

- Heterozygosity of the identified mutations was confirmed in the parents and was not present in healthy members of the family.
- To date, the present study is the first report of genetic study of Factor v deficiency in Iran.
- Patients and their families face lots of difficulties because of the symptoms of the disease and using factor replacement as treatment strategy.
- More studies are recommended to update the mutation spectrum and their clinical significance.