Molecular genetic study of Factor V deficiency in two Iranian families

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Donya Salehtash[1], Shermin Daneshpour[1], Sara Amini[1], Faezeh Sadat Mollazadeh[1], Hamideh Bagherian[1], Maryam Abiri[1,3] *,Sirous Zeinali[1,2]*

1. Dr. Zeinali's Medical Genetics Laboratory, Kawsar Human Genetics Research Center, Tehran, Iran

2.Department of Molecular Medicine, Biotechnology Research Center, Pasteur Institute of Iran, Tehran, Iran.

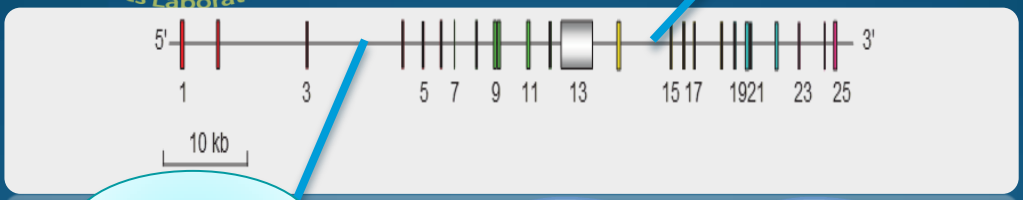
3.Department of Medical Genetics, School of Medicine, Tehran University of Medical Sciences, Tehran, Iran

*Corresponding authors:
Sirouszeinali@yahoo.com
Mary_abiri86@yahoo.co

Introduction

The F5 gene

80kb in Length



Contains 25 exons

F5 gene is located on chromosome 1q23-24

❖Factor V deficiency is a rare autosomal recessive disorder

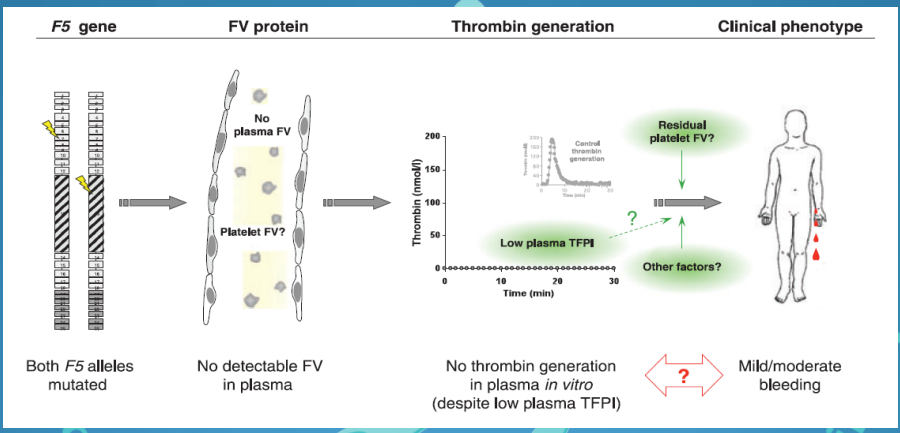
➨ Caused by mutations in F5 gene

(Homozygous or compound heterozygous state)

- **Factor V antigen levels**
  - FV levels below 10 to15% → Severe FV deficiency
  - FV plasma levels >20 to 30% → Mild to moderate FV deficiency

(Mutations in the heterozygous state)

Factor V deficiency; Genotype to phenotype



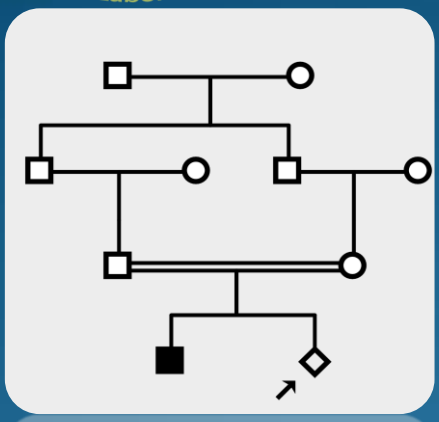
Factor V deficiency phenotypes

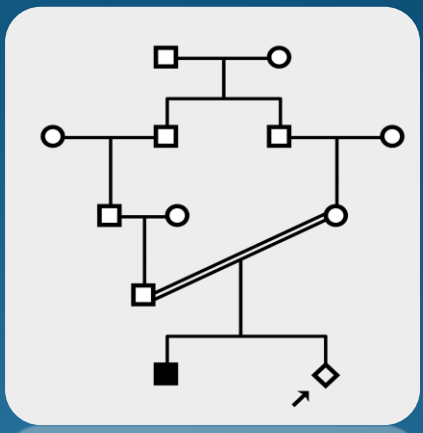
The Most Common Symptoms

- **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

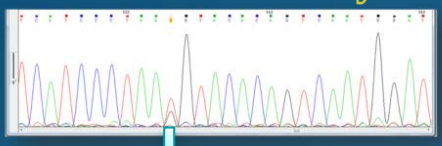
4 Extraction of DNA Using Salting Out Method

5 Direct sequencing
6 Pathogenicity prediction of identified variants

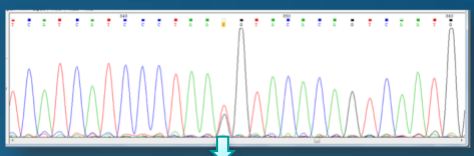
Results & Discussion

- ✓ Two different mutations inexons 5 and 10 of the F5 gene
- ✓ These mutations were Missense mutations

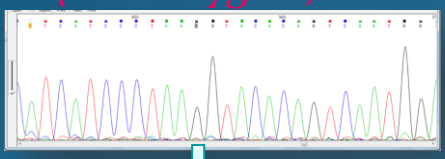
The First Family



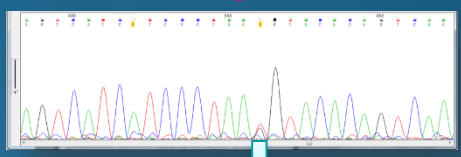
Father
(heterozygote)



Mother
(heterozygote)

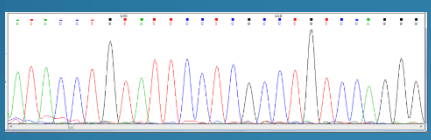


Affected individual
(homozygote)

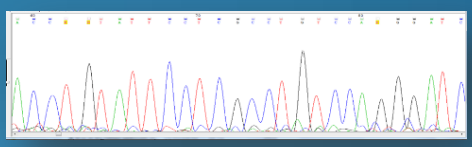


Proband
(heterozygote)

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.