2nd International and 10th National Biotechnology Congress of Islamic Republic of Iran
August 29-31, 2017
Seed and plant Improvement Institute, Karaj, Iran

دومین همایش بین المللی و دهمین همایش ملی بیوتکنولوژی جمهوری اسلامی ایران
۷-۹ شهریور ماه ۱۳۹۶
کرج، سالن همایش های موسسه تحقیقات اصلاح و تهیه نهال و بذر

Different strategies of genetic investigation of Von Willebrand disease

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Introduction

Von willberand disease is a genetic bleeding disorder with autosomal recessive mode of inheritance. The disease is mainly categorized into 3 difference types. Initial diagnosis of the disease is based on the biochemical evaluation of hemostasis factors specific for VWD. Genetic testing can confirm the clinical diagnosis.

Method & Material:

A 3 years old boy suspected to Type 3 Von Willebrand disease was referred us . Fallowing genetic counseling, the patient was candidate to be tested with NGS method. Segregation analysis was done for further confirmation of the results. In addition linkage analysis with help of STR markers were done.

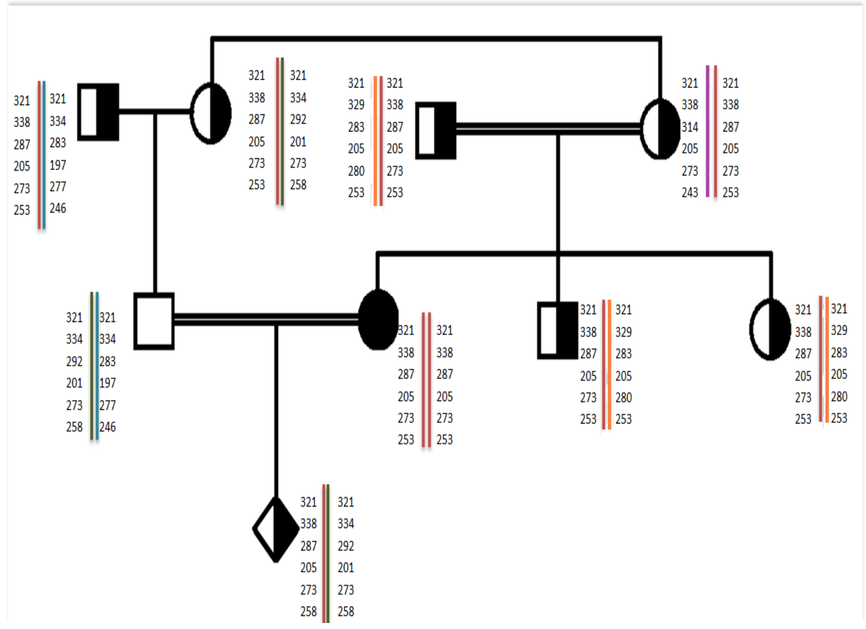
Result & Discussion:

The diagnosis of Von Willberand disease Type 3 was based on the measured activity of 2% for FVIII and VWF factors. NGS analysis of VWF gene showed a homozygote missense mutation. Segregation analysis showed heterozygous mutation in the parents. Haplotype analysis also confirmed the results. Analysis of the hemostasis factors specific for VWD can be very helpful in defining the type of the disease, but cannot help in finding the causative mutation. Finding the exact mutation is necessary for PND and PGD studies. To the best of our knowledge there are a few reports in genetic analysis of Von Willberand disease in Iran. This study showed different approaches for finding the mutation and its confirmation.

References :

Screening of Von Willebrand Disease in Iranian Women With Menorrhagia.

https://en.wikipedia.org/wiki/Von_Willebrand_disease



Key words: VWF, NGS, STR, Haplotype analysis