Abstract: Introduction; Hemophilia A is an X-linked recessive coagulation disorder because of deficiency of blood coagulation factor VIII. In view of the size and the complexity of the factor VIII gene and large numbers of mutations reported associated with hemophilia A, molecular diagnosis of the disease using polymorphic markers, located in the F8 gene and closed it, is favorable. Thus, analysis of informative markers in F8 gene region is an important step in linkage analysis and molecular diagnosis of the disease in Iranian population.

Method: In this study, by use of different bioinformatic tools, a single nucleotide polymorphic marker (rs 28370188) at 5' region of the F8 gene were investigated. This marker is located 1850 bp upstream of exon 1 in promoter of the F8 gene. Enzymatic DNA was extracted from blood sample of 160 unrelated individuals and 10 non-hemophilic family. Tetra-primer ARMS-PCR was used for genotype detection of C/T SNP. Primers were designed using oligo software. Genome amplified by ARMS PCR and 2 allele specific amplicons separated by gel electrophoresis. Results: we have detected genotype of F8 gene C/T SNP in healthy cases by gel electrophoresis. After genotyping of marker, allele frequency and heterozygosity of the marker is under investigation. Conclusion: given the importance of the 5' region of F8 gene and different mutations involved in the hemophilia A, introduction of informative molecular markers in this region and genotyping them could be useful to determine phase and heterozygosity and following, molecular diagnosis of the disease in Iranian population.