

Original Paper

## Beta globin gene haplotypes associated with hemoglobin D-Punjab in northern Iran

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### Abstract

**Background and Objective:** Hemoglobin D-Punjab is one of the variant of hemoglobin caused by a mutation on position 121 of beta globin gene which is frequent in India, Pakistan and Iran. Heterozygote form of this variant is mainly asymptomatic while in combination with hemoglobin S, severe form of anemia occurs. This study was carried out to determine the beta globin gene haplotypes associated with hemoglobin D-Punjab in Northern Iran.

**Methods:** This descriptive study was carried out on families of 18 individuals whom were carriers of hemoglobin D-Punjab in Sari in Northern Iran. Genomic DNA was extracted from peripheral blood samples using Phenol-chloroform standard protocol. In order to identify different haplotypes associated with hemoglobin D-Punjab, PCR-RFLP method and family linkage analysis were used.

**Results:** In 17 subjects hemoglobin D-Punjab was linked to [+ - - - + +] haplotype and in one case association with [- + + - + +] haplotype was observed.

**Conclusion:** The hemoglobin D-Punjab alleles have mainly unicentric origin and [- + + - + +] rare haplotype may have different genetic origin or is created as a result of gene recombination.

**Keywords:** Hemoglobin D-Punjab, Haplotype, PCR-RFLP, Iran

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