

## Abstract

Type 1 Diabetes is a polygenic and autoimmune disease caused by self-destruction of pancreatic *B*-cells. The present study designed to investigate the association of SNPs (rs5742909,rs231775, rs4553808 and rs733618) on *CTLA4* and rs1544410 on *VDR* gene with the onset of T1D in Pakistani population. For this purpose blood sample of 102 patients and 100 controls were collected along their anthropometric data. Genotyping was performed in the patients and the healthy control subjects by sequencing and polymerase chain reaction-fragment length polymorphism analysis using *MseI* and *BsmI* restriction endonucleases for the (rs5742909, rs4553808) and rs1544410 SNPs respectively. The current study demonstrated that males were at higher risk of disease development as compared to females and age of diagnosis was higher during puberty. Most of the patients were underweight with mean value of BMI 17.15 and 17.87 in males and females respectively. Positive family history was strongly associated with T1D. All the studied SNPs (rs231775, rs5742909, rs4553808, rs733618) of *CTLA-4* gene and rs1544410 of *VDR* gene were found to be involved in pathogenesis of type 1 diabetes (T1D) in Pakistani population. The frequency of ACAT and ACGC haplotypes was significantly higher in the patients as compared to controls while haplotype ACAT was significant in control and plays a protective role against disease development. So it is concluded that *CTLA-4* and *VDR* gene polymorphism is significantly associated with T1D and can be used as genetic marker for the identification of T1D.