ABSTRACT

T1D is the polygenic as well as the multifactorial disease caused by the autoimmune destruction of pancreatic β-cells. Present study was designed to investigate the association of polymorphism in *INS* gene with T1D of Pakistani population. Three SNPs of *INS* gene rs689 (*HphI*), rs3842748 (*DraIII*) and rs3842752 (*PstI*) were selected for the association with T1D and genetic analysis. For this purpose blood sample of 50 patients and 50 controls were collected along their anthropometric data. According to WHO criteria all these patients were clinically diagnosed with T1D and they were interviewed for assessment of clinical, physical and social aspects. Genomic DNA was isolated from each blood sample and PCR was done to target the required sequence. Genotyping was carried out by sequencing of DNA and PCR-RFLP. The current study demonstrated that males were at high risk of disease development as compare to females and age of diagnosis was higher during puberty. Low BMI (<18) as well as depression and positive family history were significantly associated with T1D. Overall two SNPs rs689 and rs3842752 were significantly associated ($P < 0.05$) with T1D in Pakistani population. Haplotype analysis indicated that CGA and TGT haplotypes were significantly associated with T1D ($P < 0.01$) while the frequency of CGT haplotype was significant in control and plays a protective role against disease development. *INS* gene polymorphisms were significantly associated with T1D and can be used as genetic marker for the diagnosis of T1D.