Abstract

AS3MT gene has been reported to be a candidate gene for inorganic arsenic methylation in which mutation causes inflammation in vascular tissues that ultimately causes CVD. Two SNPs of this gene rs10748835 and rs11191439 have been reported to be in strong association with CVD in different populations. This study aims to find association of these SNPs in Pakistani population. For this purpose blood samples were collected from 50 CVD patients and 50 healthy controls. Demographic data was also analyzed. Mean age, BMI, Cholesterol of patients was 49 years, 30.90kg/m^2 and 316mg/dl respectively. Using PCR and DNA sequencing methods two SNPs were genotyped. Genotypes were than analyzed using SHEsis software for allelic frequency, genotypic frequency and haplotype analysis. Analysis of the genotype and allelic frequencies distribution of both SNPs revealed a significant positive association. AA genotype of rs1191439 and CC genotype of rs10748835 was found significantly associated with CVD in Pakistani population as these genotypes having high frequency in cases and have p value less than that of 0.05. Haplotype analysis further supported the findings by showing that carriers of haplotype G-C had significantly higher frequency (0.614) in the patients. Further Linkage disequilibrium showed the association between both SNPs on chromosome 10 (r^2=0.78). In conclusion, the study proved that rs10748835 and rs11191439 polymorphisms of *AS3MT* gene are positively associated with CVD in Pakistani population as p-value is less than 0.05.