

ABSTRACT

Glaucoma is an irreversible blindness caused by the elevation of Intra ocular pressure (IOP), optic nerve damage, followed by visual field defects and ultimately vision loss if left untreated. Apolipoprotein E (*APOE*) gene was considered to be susceptible to cause Primary Open Angle Glaucoma (POAG) in Pakistani population. The current study was aimed to identify the association of genetic variations in *APOE* gene and risk of glaucoma development. For this purpose, blood samples were collected from 100 patients and 100 controls along with their clinical characteristics. All patients were clinically diagnosed with glaucoma according to WHO criteria. Genomic DNA was isolated from the blood. DNA was amplified with Polymerase Chain Reaction (PCR) followed by DNA sequencing and Restriction Fragment Length Polymorphism (RFLP) analysis. Moreover, genotypic frequencies of three SNP's (rs769445T/C (0.0015), rs405509G/T (0.0016) and rs449647A/T (0.0016) were higher in patients as compared to controls and were significantly associated ($p < 0.05$) with POAG in the population of Pakistan. The haplotype analysis indicated that C T T, T T G, T T T and C T G frequencies were higher in patients as compared to controls ($p < 0.05$). While no linkage disequilibrium was seen between (rs769445, rs405509 and rs449647). In conclusion, the polymorphism between *APOE* was significantly associated with glaucoma. A large scale genetic screening is required to build a set of genetic markers that can be used for genetic study for early diagnosis and prevention.