

## ABSTRACT

Atonal homolog 7 (*ATOH7*) belongs to basic helix-loop-helix (bHLH) protein family which controls photoreceptor development. *ATOH7* is a transcription factor largely concerned with differentiation of retinal ganglion cells (RGCs) and optic nerve formation. It has also been reported that abnormal *ATOH7* expression resulted in an increased number of differentiated RGCs in an animal model of glaucoma. The present study aims to identify genetic variants in *ATOH7* gene and haplotypes association of *ATOH7* gene polymorphism with the onset of Glaucoma in Pakistani population. Three SNP's (rs7916697, rs61854782, rs3858145) of *ATOH7* gene was evaluated for their genetic association with glaucoma. For this purpose DNA was isolated from blood samples of 100 clinically diagnosed glaucoma patients and 100 control objects. Genotyping was done by PCR and Direct sequencing followed by RFLP. Genotypic frequencies of two SNP's rs7916697 "CT" (0.52), "TT" (0.48) and rs61854782 "AC" (0.54), "CC" (0.46) was higher in patients as compared to controls and were significantly linked ( $p < 0.05$ ) with glaucoma in Pakistani population. It was estimated that rs3858145 polymorphism was not significantly associated with onset of glaucoma. Haplotype analysis indicated that the frequency of CAA, TAA, CAC and TAC was higher in patients as compared to controls and significantly associated ( $p < 0.05$ ) with the disease onset. Moreover, no linkage disequilibrium was observed between rs7916697 and rs61854782 except rs3858145 ( $D=0.95$ ). It was concluded that *ATOH7* polymorphism was significantly associated with glaucoma in Pakistani population. Furthermore, large scale genetic screening is required to built a set of genetic markers that can be used for the genetic testing for early diagnosis and prevention of disease.