

## Genetic Association of SIX6 Gene with Pathogenesis of Glaucoma

**Authors :** Riffat Iqbal, Sidra Ihsan, Andleeb Batool, Maryam Mukhtar

**Abstract :** Glaucoma is a gathering of optic neuropathies described by dynamic degeneration of retinal ganglionic cells. It is clinically and innately heterogenous illness containing a couple of particular forms each with various causes and severities. Primary open-angle glaucoma (POAG) is the most generally perceived kind of glaucoma. This study investigated the genetic association of single nucleotide polymorphisms (SNPs; rs10483727 and rs33912345) at the SIX1/SIX6 locus with primary open-angle glaucoma (POAG) in the Pakistani population. The SIX6 gene plays an important role in ocular development and has been associated with morphology of the optic nerve. A total of 100 patients clinically diagnosed with glaucoma and 100 control individuals of age over 40 were enrolled in the study. Genomic DNA was extracted by organic extraction method. The SNP genotyping was done by (i) PCR based restriction fragment length polymorphism (RFLP) and sequencing method. Significant genetic associations were observed for rs10483727 (risk allele T) and rs33912345 (risk allele C) with POAG. Hence, it was concluded that Six6 gene is genetically associated with pathogenesis of Glaucoma in Pakistan.

**Keywords :** genotyping, Pakistani population, primary open-angle glaucoma, SIX6 gene

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