

## GENETIC ANALYSIS OF *CYP1B1*, *MYOC*, *OPTN* AND *OPTC* GENES IN GLAUCOMA PATIENTS FROM SOUTH INDIA

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Glaucoma represents a group of optic neuropathies with different genetic basis. In India, ~1.5 million people are blind due to glaucoma. Mutations in the *MYOC* gene at the *GLC1A* locus on chromosome 1q21-q31, *CYP1B1* gene at the *GLC3A* locus on chromosome 2p21 and the *OPTN* gene at the *GLC1E* locus on chromosome 10p14 have been found in patients with glaucoma. The purpose of the present study was to screen sequence variations in these genes as well as in *OPTC* in a total of 146 glaucoma patients ascertained mostly from the state of Karnataka and its border areas with states of Andhra Pradesh and Tamilnadu, south India. Of these, 116 were affected with adult-onset primary open-angle glaucoma (A-POAG), 18 were with juvenile-onset primary open-angle glaucoma (J-POAG) and 12 were with primary congenital glaucoma (PCG). The coding regions of *CYP1B1* and *MYOC* were screened for mutations and variations in all of the 146 patients using a combination of PCR-SSCP and DNA sequence analyses. Our analysis revealed a total of five mutations (two novel and three known) and seven variants/polymorphisms in *CYP1B1*. No mutation was found in *MYOC*. However, we have detected one known and one novel polymorphisms in this gene. Screening of 116 A-POAG patients showed one novel and one known risk-factor associated mutations in *OPTN*. In addition, two novel and one known polymorphisms were also detected in *OPTN*. Screening of *OPTC* in 116 A-POAG patients showed one novel and two previously known polymorphisms. (This work was supported by a grant from the CSIR, New Delhi).