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Genetic Variants Associated with Primary Open-Angle Glaucoma in a Multiethnic Population

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Primary open-angle glaucoma (POAG) is a leading cause of irreversible blindness and is influenced by complex genetic and environmental factors. Understanding genetic predisposition aids early diagnosis and targeted therapies. This genome-wide association study (GWAS) evaluated DNA samples from 2,000 participants representing diverse ethnic backgrounds to identify genetic variants associated with POAG susceptibility. Analysis revealed multiple loci with significant association, including known risk genes MYOC and OPTN, as well as novel variants unique to specific ethnic groups. These findings highlight the genetic heterogeneity underlying POAG and suggest ethnicity-specific risk profiles. Further functional studies of these loci demonstrated

involvement in intraocular pressure regulation, optic nerve resilience, and trabecular meshwork function. Integration of genetic risk scores with clinical parameters improved prediction accuracy for disease onset.

This study underscores the need for personalized glaucoma screening programs considering genetic background. Additionally, gene therapy targeting implicated pathways offers future therapeutic potential. Limitations include sample size constraints for rare variants and the need for replication in independent cohorts. Overall, elucidating genetic determinants enhances understanding of POAG pathogenesis and moves the field toward precision medicine.

Biography

Dr. Ling Wei is a geneticist and ocular disease researcher at the National Eye Institute in Bethesda. She leads pioneering genomic studies focused on inherited eye disorders and the development of gene-based therapies. Dr. Wei's research aims to uncover the genetic foundations of vision loss and advance precision medicine in ophthalmology.