

Genetics of primary open angle glaucoma

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Glaucoma is an heterogeneous group of disorders resulting from optic nerve damage and is the 2nd leading cause of irreversible blindness worldwide. It was originally described in Greece around 500 BC. It is distinguished in primary which is nonsyndromic and in secondary which can be syndromic. Primary open angle glaucoma (POAG), the most common form of this group of heterogeneous diseases, refers to the open, normal-appearing anterior chamber angle with normal trabecular meshwork in patients and it affects 1-2% of population older than 40 years. At the genetic level a distinction between juvenile onset with age at diagnosis of 10-35 years and adult onset with an age at diagnosis above 35 years has been made. Although the etiology of POAG is unknown, at least 8 genetic loci are involved. MYOC is an important gene for juvenile onset of POAG and a proportion of adult onset POAG. Many more major POAG genes are likely to exist and POAG is believed to have a digenic or multigenic inheritance. It is important in order to study the genetics of POAG, to keep in mind that, first, when mapping "monogenic" traits in large pedigrees, more than 1 gene might be segregating; Some affected individuals might represent phenocopies; Some affected individuals might be homozygous for a dominant mutation, and precise clinical delineation of the phenotype is mandatory in linkage mapping. This is the way to design a rational future therapy depending on pathophysiology, genes and pathways and based on pharmacogenetics and gene therapy.

Biography

Marilita M. Moschos graduated the Pharmacy School of the University and the Medical School of the University of Athens. She is actually working as Ass. Professor of Department of Ophthalmology of Athens University where she has the clinical and scientific co-responsibility of the Laboratory of Electrophysiology of Vision and the department of Glaucoma. She authored over 50 scientific papers in PubMed reviewed journals and presented over 60 at international conferences, in some of them as invited speaker. She is a member of many international ophthalmological societies and reviewer in several ophthalmological journals.