

☒ | Chase the family of primary-open angle glaucoma patients

The Science behind the Tip

Various modes of screening for primary open-angle glaucoma (POAG), i.e. different combinations of tonometry and structural or functional examinations, are to date not cost-effective¹. Case-finding is considered economically sounder. A positive family history is a strong, if not the strongest, risk factor for POAG. Many patients have first degree or more distant relatives with POAG². Thus, one of the most efficient ways to screen for undiagnosed disease is at present to encourage POAG patients to have their family members examined.

In order to maximize the efficacy of these efforts, 3 recommendations can be made. (1) If the POAG patient is aged under 50 and especially 40, all siblings (brothers and sisters) should be examined. (2) If there are more than 2 first degree relatives with confirmed POAG, all relatives should be screened. (3) Relatives over age 50 who show no sign of POAG should be examined at a 4 to 5 years interval.

Various POAG genes have been identified, the myocilin (MYOC or TIGR)³ and optineurin (OPTN)⁴ genes being among the most extensively studied. Mutations of these genes could be linked to specific subsets of POAG, namely to juvenile-onset and normal-tension glaucoma cases, respectively. They only account however for a small percentage of POAG cases overall. The heredity of this multifactorial disease appears to be mostly more complex, precluding at this time routine genetic testing for identifying individuals genetically predisposed to it.

References

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