

☒ | Pseudoexfoliation glaucoma has a strong genetic component

The Science behind the Tip

Pseudoexfoliation or exfoliation syndrome (PEX) is the most common identifiable condition associated with glaucoma. Individuals can have PEX and not develop glaucoma, but they are at high risk. In a recent population-based study with long-term follow-up, 16% of PEX patients required treatment upon presentation and of the remaining patients, 44% received therapy over the next 15 years¹. Further, the prognosis of pseudoexfoliation glaucoma is worse than that of primary open-angle glaucoma. In EMGT, progression risk was doubled among patients with PEX².

Screening of relatives of PEX patients is useful because several clinical studies have indicated that PEX runs in families³. Research into the genetic basis of PEX has recently led to the discovery that variants in the LOXL1 gene on chromosome 15 are strongly associated with pseudoexfoliation glaucoma^{4,5}. The LOXL1 protein encoded by the gene is involved in the formation of elastin fibers and appears to play a role in the accumulation of the deposits that causes PEX.

References

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