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Optineurin (OPTN) screening in Primary Open Angle-Glaucoma (POAG)

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Purpose: To determine the role of *OPTN* in POAG with high intraocular pressure (IOP). **Methods:** 271 primary open-angle glaucoma probands with IOP > 22 mmHg were screened for *OPTN* mutations.

Results: We found causative mutations in three probands: one with an E50K and two with R545Q mutations. In addition, we found 34 individuals with the risk-associated alteration, M98K.

Conclusions: In screening a set of subjects with POAG with elevated IOP, causative OPTN mutations were found in 3/261 (1.15%). In contrast, a risk-associated alteration was found in 13 %.

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Topic (Complete): 5.6 Genetics Additional (Complete): Financial interest : No Approved by IRB : Yes Permission to Record : Yes Study Design : Cohort Study Precis : Although OPTN is reportedly important in normal tension glaucoma, it may play only a minor role in primary open-angle glaucoma, being the cause of 1.15% of POAG cases with IOP > 22 mmHg.