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

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 %.

Commercial Relationship: R.M. Ayala-Lugo, None; P.R. Lichter, None; H.S. Pawar, None; A. Aledavood, None; S.E. Moroi, None; C.A. Downs, None; T.M. Guckian, None; W.C. Bromley, None; C.T. N'tim-Amponash, None; A. Mendoza, None; V. Azocar, None; E.J. Maul, None; B.F. Boyd, None; S. Boyd Lewis, None; J.E. Richards, None.

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.