



SCREENING OF OPTINEURIN GENE MUTATIONS IN JAPANESE NORMAL TENSION GLAUCOMA PATIENTS

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Background: In patients with autosomal dominantly inherited adult-onset POAG, T. Rezaie, et al identified a causative gene on chromosome 10p14 and designated it OPTN (for "optineurin") (Rezaie, T et al. Science, 2002). We investigated whether the OPTN contained the missense mutations reported by Rezaie et al. in Japanese patients with normal tension glaucoma.

Design: Genetic screening study in Japanese NTG patients

Participants: Sixty-seven Japanese normal tension glaucoma patients (19 males and 47 females)

Testing: All patients were screened for mutations in OPTN by DNA direct sequencing.

Main Outcome Measure: Mutations in OPTN

Results: According to the Rezaie's report, we checked missense mutations that resulted in "disease causing" Glu50?Lys (E50K) amino acid change, 2-base pair "AG" insertion and Arg545?Gln (R545Q) residue change, and a "risk-associated" Met98?Lys (M98K) amino acid change, in OPTN. Glu50?Lys (E50K) amino acid change was not present in patients with normal tension glaucoma, however, Arg545?Gln (R545Q) residue change was present in one patient (1.5 %) and "risk-associated" Met98?Lys (M98K) amino acid change was detected in 7 patients (10.7 %). In addition, part of exon 5 in OPTN was defective in two patients (3.0 %).

Conclusion: Our results indicated that the mutations in OPTN might be a contributing factor for developing normal tension glaucoma in Japanese patients.