Last results of the Spanish Multicenter Genetic Glaucoma Group

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Purpose: To present the last results of the Spanish Multicenter Genetic Glaucoma Group.

Methods: Complete ophthalmic examination of a Spanish population with familial glaucoma. Genetic analysis of MYOC and CYP1B1 genes by DNA extraction, PCR amplification and direct sequencing.

Results: We recruited 302 individuals with a positive family history of glaucoma or OHT coming from 18 different Spanish eye centers. We identified two novel MYOC mutation in two primary open angle glaucoma (POAG) families, and five previously reported MYOC mutations in seven families affected with POAG (5), juvenile glaucoma (1) and normotensive glaucoma (1). CYP1B1 mutations were found in sixteen index patients affected with primary congenital glaucoma (9), POAG (3), juvenile glaucoma (2) and Axenfeld Rieger syndrome (2).

Conclusions: The mutational ratio coincided with other occidental populations with low rates of consanguinity; genotype-phenotype correlation and genetic counseling were provided to our patients; molecular diagnosis contributed in our therapeutic decisions in order to better control our glaucoma patients.