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Last Results of the Spanish Multicenter Genetic Glaucoma Group

We present the last results of our multicenter, prospective, observational, case-series study of a Spanish population affected by family glaucoma (G) or ocular hypertension (OHT). Our purposes were to establish the clinical characterization and perform a mutational analysis of MYOC and CYP1B1 in patients and their relatives and to describe their genetic result.

Patients & Methods

Complete ophthalmic examination in order to describe their type of glaucoma and stage of the disease. Ancillary tests included computerized perimetry, ultrasound pachymetry, gonioscopy as well as fast-RNFL OCT in the OHT cases and Heidelberg tomographic treatment according to the literature.

Results I

Clinical forms: 14 PCG, 10 OHT, 6 NTG, 3 PEX, 3 PIGM, 1 RIEG.

Clinical forms: 84 POAG, 18 JOAG, 14 PCG, 10 OHT, 6 NTG, 3 PEX, 3 PIGM, 1 RIEG.

Ophthalmologic evaluation: -BCVA: 0.71 RE / 0.69 LE

Discussion & Conclusions

• We established the phenotypic and genotypic profile of a population of patients affected by family glaucoma or OHT coming from different parts of Spain.

• The majority of clinical forms were moderate-severe cases of POAG.

• Mutational screening results were similar to other genetic diagnosis series published in the literature.

Results II

CYP1B1 Mutations

Mutation families 18 Spanish hospitals belonging to the Spanish Multicenter Genetic Glaucoma Group (EMEIGG, estudio multicéntrico español de investigaciones genéticas en glaucoma) from different cities of Spain participated in the study.

139 index patients from 139 unrelated Spanish families were included in the study with a mean age of 60.09 y (5-98 y); 55% women – 45% men

Non consanguineous cases were reported but 10% of cases were originary from small villages with less than 500 inhabitants.

All were familial G/OHT cases and 42% were true hereditary cases (propositus case + two affected first degree relatives from consecutive generations).

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Discussion & Conclusions

• We found two novel MYOC mutations and one novel CYP1B1 mutation.

• We regarded the genotypic diagnosis as a risk factor to modulate the therapeutic approach of the patients.

• Considering the high percentage of CYP1B1 mutations present in our PCG population we regard this test as mandatory for the management of these cases.