CYP1B1 GENE ANALYSIS AND PHENOTYPIC CORRELATION IN CHILDREN WITH PRIMARY CONGENITAL GLAUCOMA

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Purpose: To investigate the prevalence of CYP1B1 mutations in Portuguese children with primary congenital glaucoma (PCG) and attempt to correlate the mutation status with clinical features of the disease. Material and Methods: DNA sequencing analysis of the CYP1B1 gene was used to screen 21 children with PCG followed on Pediatric Ophthalmology and Medical Genetics consultations at Hospital D. Estefânia (Centro Hospitalar de Lisboa Central). We also examined the effect of mutations on the phenotype of the patients. Results: We detected mutations in the CYP1B1 gene in six patients (28.57%), all compound heterozygotes. Seven types of mutations were identified: c.182GA, c.317CA, c.535delG, c.1064_1076del, c.1159GA, c.1310CT and c.1390dupT. All patients with these mutations developed bilateral PCG, whereas in the group without mutations only seven (46.67%) showed bilateral disease. Age at diagnosis was lower in the group of patients with these mutations (0.0±0.00 months vs. 4.5±2.63 months, p 0.01). In the remaining variables (age at first surgery, postoperative intraocular pressure and corneal diameter, final VA, number of surgical reinterventions and the need for anti-glaucoma medications postoperatively) there was no significant difference between the two groups (p 0.05 for all comparisons). Conclusion: This study demonstrates the diversity of CYP1B1 mutations and its impact on clinical PCG. In the future, genetic analysis can be a useful tool for the diagnosis, prognosis and treatment of this pathology.