Genotype/ phenotype correlations in primary congenital glaucoma patients in the Lebanese population

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Purpose: To investigate the roles of Cytochrome P1B1 (CYP1B1) gene and Myocillin (MYOC) gene mutations in primary congenital glaucoma (PCG) in the Lebanese population and perform genotype/phenotype correlations.

Methods: This was a prospective study conducted at the Ophthalmology Department at the American University of Beirut. Lebanese patients with unilateral or bilateral PCG and their first degree relatives (parents and siblings) were screened for CYP1B1 and MYOC mutations. Demographic and phenotypic characteristics were recorded. Phenotypic characteristics pertaining to disease severity and outcomes were compared.

Results: Eighteen Lebanese families (66 subjects) with at least one member affected with PCG were included in this study: thirteen from the Muslim community, four from the Druze community, and one was a Christian family. Mutations in the CYP1B1 gene were detected in 6 families (33%). Five previously described mutations (R444Q; E229K; R469W; G61E; M1T) and one new single nucleotide deletion were identified (1793delC). The mutation group tended to have more severe phenotype with earlier age at diagnosis, higher rate of bilateral disease, and higher number of glaucoma surgeries. MYOC gene mutations were not detected in any of the families.

Conclusions: The rate of mutation in our population is lower than that reported in the rest of the Arab and Middle Eastern populations and suggests other culprit genes may need to be uncovered through additional analytical efforts.