World Academy of Science, Engineering and Technology
International Journal of Biomedical and Biological Engineering
Vol:11, No:12, 2017

Clinical and Molecular Characterization of 120 Families with Sporadic Juvenile Onset Open Angle Glaucoma

Authors : Bindu I. Somarajan, Viney Gupta, Gagandeep Kaur Walia, Jasbir Kaur, Sunil Kumar, Shikha Gupta, Abadh K. Chaurasia, Dinesh Gupa, Abhinav Kaushik, Aditi Mehta, Vipin Gupta, Arundhati Sharma

Abstract : Background: Juvenile onset primary open angle glaucoma (JOAG), affects individuals under the age of 40 years. Studies on a few families of JOAG, that led to the discovery of the Myocilin gene, reported the disease to have an autosomal dominant pattern of inheritance. However, sporadic forms of JOAG been seen to be more common in some populations. Most pathological homozygous mutations in the CYP1B1 gene associated with JOAG have been seen among sporadic cases. Given the higher prevalence of sporadic JOAG cases in our population, we aimed to look for common mutations E229K and R368H, the two most common variants in the CYP1B1 gene associated with glaucoma. Objective: To determine the frequency and evaluate genotype phenotype correlation of CYP1B1 E229K and R368H mutations in a cohort of 120 sporadic Juvenile open angle glaucoma patients.Methods: Unrelated JOAG patients whose first degree relatives had been examined and found to be unaffected were included in the study. The patients and their parents were screened for E229K and R368H mutations. The phenotypic characteristics were compared between probands with and with out these mutations by SPSS v16. Results: Out of 120 JOAG patients included in the study, the E229K mutation was seen in 9 probands (7.5%) and R368H in 7 (5.8%). The average age of onset of the disease (p=0.3) and the highest untreated IOP (p=0.4) among those carrying mutations was not significantly different from those who did not have these mutations. The proportion of probands with angle dysgenesis among those with E229K and R368H mutations was 70% (11 out of 16) in comparison to 65% (67 out of 104) of those who did not harbour these mutations (p=0.56). Similarly the probands with moderate to high myopia among those with E229K and R368H mutations was 20% (3 out of 16) in comparison to 18% (18 out of 104) of those who did not harbour these mutations(p=0.59). Conclusion: The frequency of E229K and R368H mutations of the CYP1B1 gene is low even among sporadic JOAG patients. Moreover there is no clinical correlation between the presence of these mutations and disease severity

Keywords : CYP1B1, gene, IOP, JOAG, mutation
Conference Title : ICHGG 2017 : International Conference on Human Genetics and Genomics
Conference Location : Rome, Italy
Conference Dates : December 11-12, 2017

Open Science Index, Biomedical and Biological Engineering Vol:11, No:12, 2017 publications.waset.org/abstracts/77908.pdf