Association of copy number variations in complement factor H-related genes among age-related macular degenerative subjects

ABSTRACT

Age-related macular degeneration (AMD) is the most widely recognised cause of irreversible vision loss and previous studies have suggested that the advancement of wet AMD is influenced by both modifiable and non-modifiable elements. Single nucleotide polymorphism (SNPs) and copy number of variations (CNVs) have been associated with AMD in various populations, however the results are conflicting. Our aim is to determine the CNVs of Complement Factor H-Related genes among Malaysian subjects with wet AMD. 130 patients with wet AMD and 120 healthy controls were included in this research. DNA was extracted from all subjects and CNVs of CFH, CFHR1 and CFHR3 genes; determined using quantitative real-time PCR and were compared between the two groups. A consistent association was observed between CFH gene and wet AMD susceptibility ($P < 0.05$). The age-adjusted data suggests a possible increased risk of AMD disease ($P < 0.05$). No correlation was detected between CNVs and wet AMD for the remaining genes after we compared the frequencies of mean for that gene. An association was observed between CFH CNVs and wet AMD in the Malaysian population, however, strong evidence of a link with wet AMD was not found. Further investigative studies are needed using larger sample sizes to elucidate the role of CNVs in AMD pathogenesis.

Keyword: Age-related macular degeneration; CFH gene; Copy number variation; Malaysia