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Prevalence of Complement Factor H (Y402H) Gene Polymorphism and Its Impact on the Predisposition of Syrians to Age-Related Macular Degeneration (AMD) and Response to Bevacizumab Intravitreal Injection

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Abstract : Age-related macular degeneration (AMD) is one of the leading causes of blindness worldwide. Complement factor H polymorphism (Y402H) is thought to play a potential role in the predisposition to AMD and response of patients with exudative AMD to treatment with anti-Vascular Endothelial Growth Factor (anti-VEGF). This study aimed to investigate the frequency of Y402H among Syrians, its impact on their susceptibility to AMD, and the hypothesized role of Y402H in patients' response to intravitreal anti-VEGF (i.e.,, bevacizumab). Our case-control study encompassed unrelated 54 AMD cases and 44 controls. Genotyping was determined by standard sequencing of PCR products. Frequency was compared between patients and controls, and correlation between genotype and response to treatment was assessed in 20 patients with wet AMD who received a therapeutic course of three intravitreal bevacizumab injections (once monthly). Our results revealed a significantly higher prevalence of the risk allele C among AMD cases (51.9%) in comparison with controls (37.5%) (P= 0.04, OR= 1.386, CI= 0.999-1.923). Patients with the TT genotype (no risk allele) exhibited a significantly better primary response rate, reached 87.5% compared to only 41.7% in patients carrying the risk allele C (TC + CC), (P= 0.04, OR= 9.8, CI=0.899- 106.84). The findings of this study prove the importance of investigating Y402H polymorphism as a prognostic marker for predicting response to bevacizumab in AMD patients.

Keywords : age-related macular degeneration, bevacizumab, complement factor H gene, polymorphism, Y402H
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