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Clinical study of the correlation between complement factor H polymorphism and age-related macular degeneration

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ABSTRACT. This study aimed to investigate the correlation between age-related macular degeneration (AMD) of the liver-kidney yin-deficiency type and complement factor H (*CFH*) polymorphism, and to determine whether the C allele of the T1277C (Y402H) variant is a risk factor for this condition. We performed a case-control investigation of 60 patients with liver-kidney yin-deficiency AMD and 60 normal control subjects. Peripheral blood was collected from each participant for DNA extraction. Following amplification by polymerase chain reaction, the DNA samples were sequenced, and polymorphism of the *CFH* gene was examined. Data were analyzed with the chi-square test, with $P < 0.05$ signifying statistical significance. The frequency of the C allele was significantly higher in the wet than in the dry AMD group ($P = 0.044$). In addition, the TC and CC genotypes were markedly more common in the former than in the control group ($P = 0.013$), and there

was a significant difference in the distribution of the T and C alleles between wet AMD patients and control subjects ($P < 0.05$). Based on this, we conclude that liver-kidney yin-deficiency AMD is associated with the C allele and TC and CC genotypes of the *CFH* Y402H polymorphism. Among patients with this condition, *CFH* genotypes were normally distributed. The principal *CFH* genotypes that induce liver-kidney yin-deficiency AMD are the mutant homozygote CC and heterozygote TC forms. Moreover, C allele carriers are at higher risk of developing this disease.

Key words: Liver-kidney yin deficiency type; Y402H polymorphism; Age-related macular degeneration; *CFH* gene