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Aqueous humor complement factor I concentration is associated with SNP of *CFI* gene

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Complement activation is a central innate immune mechanism. Aberrant alternative complement activation has been linked to the pathogenesis of age-related macular degeneration (AMD). Complement factor I (FI, encoded by the *CFI* gene) is a negative regulator of complement and plays an important role in limiting complement activation. There is increasing evidence to suggest that local alterations in FI levels may be a priming event in AMD.

Here we evaluated the association of systemic and locally produced FI levels with AMD and a regulatory *CFI* SNP (rs2285714). FI levels were measured by ELISA in plasma (54 AMD cases and 25 age-matched controls) and in aqueous humor (24 AMD cases and 22 age-matched controls). Aqueous humor (AH) samples were obtained from AMD patients before intravitreal injection of anti-VEGF and elderly controls during cataract surgery. rs2285714 was genotyped by real-time PCR. There was no difference in the FI plasma levels in AMD (27.8 ± 7.1 μg/mL) and control (25.3 ± 5.3 μg/mL) patients. The median FI level in AH was lower in AMD patients (33.1 ng/mL) compared to controls (58.4 ng/mL), although the difference was not significant ($p = 0.12$). Huge concentration gradient between the plasma and AH was found. rs2285714 was significantly correlated with FI concentration in AH, but not in plasma. The median FI level in AH was significantly higher in individuals with the CC genotype (82.5 ng/mL) compared to individuals with the heterozygote (35.9 ng/mL, $p = 0.002$) and TT genotypes (25.9 ng/mL, $p = 0.0002$). These findings support that the importance of local rather than systemic FI production in AMD.

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