AB099. Cognitive impairment and age-related macular degeneration: a possible genetic link

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Background: The number of older adults affected by age-related macular degeneration (AMD) and early cognitive changes is on the rise. Recent studies have shown a high co-occurrence of these conditions. This, along with shared risk factors and similar histopathology suggests they may share genetic risk factors as well. The goal of this study was to explore the possibility of known AMD SNPs contributing to the co-morbidity.

Methods: Participants (AMD and controls) aged 70 years or older with no known neurological or cognitive impairments were recruited for this study. Visual function was evaluated using ETDRS visual acuity, Mars Contrast sensitivity and the scanning laser ophthalmoscope. Cognitive status was measured using the Mini-Mental State Exam (MMSE) and the Montreal Cognitive Assessment (MoCA). Genotyping was conducted using a panel of AMD single nucleotide polymorphisms (SNPs). Analysis was focused on the CFH Y402H and ARMS2 A69S SNPs due their association with drusen and evidence of their association with cognitive impairment.

Results: According to the MMSE, two participants from the AMD group (N=21) and none from the control group (N=18) scored positive for cognitive impairment. The MoCA indicated 33.3% of the AMD group and 27.7% of the control group had MCI. There were no significant differences between MoCA scores based on the carrier versus non-carrier status of either the CFH or ARMS SNPs. The SNP in FADS1 (rs174547) that was part of the original panel, but not in the analysis, was found in a large number of participants. All those who scored positive for MCI were homozygous carriers of the FADS1 SNP.

Conclusions: Although more people from the AMD group scored positive for MCI, scores between groups were significantly different. The AMD and control groups did differ on which cognitive domains they had difficulty with, indicating those with AMD and MCI may be at a higher risk of converting to AD. There were no significant differences on cognitive scores between CFH and ARMS2 SNP carriers and non-carriers. The FADS1 SNP, not originally intended to be part of this study, will be included in future analyses to explore the possibility of a founder effect and a potential link to mild cognitive impairment (MCI).

Keywords: Age-related macular degeneration (AMD); mild cognitive impairment (MCI); Alzheimer's disease (AD); genetics

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