Author's response to reviews

Title: Evaluation of variants in the selectin genes in age-related macular degeneration.

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Dear Editors,

Please find attached our article “Evaluation of variants in the selectin genes in age-related macular degeneration”. In this report we describe a focused association study in which variants in the genes for E-selectin, L-selectin and P-selectin were assessed in a cohort of AMD patients and controls. We hypothesized that, in light of current evidence for inflammatory processes in AMD, genes involved in regulating leukocyte-endothelial interactions could play a role in AMD.

We confirmed a possible role of the selectins in AMD pathogenesis by showing expression of these genes in retinal microglia, drusen cores, and the choroidal vasculature of human eyes. Further, we tested a large cohort of AMD patients and controls (n=741) with densely packed SNPs that are distributed across the selectin genes. We show that there is no evidence that ancestral mutations in the selectin genes are a common cause of AMD. Given the prior successes of association studies identifying risk alleles for AMD (i.e. CFH) and the high interest of the field in genes involved in inflammatory processes, we believe the readership of BMC Medical Genetics would be interested in the findings of our report.

The manuscript includes 1775 words, two figures, and 1 table. We look forward to your review of our manuscript.

Best regards,

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Associate Professor