Veterinary & Biomedical Sciences Seminar

Speaker

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Thursday 29 October
12.30pm – 1.00pm
VB3.23

The Genetic Epidemiology of Age-Related Macular Degeneration: Investigating the Role of the C2 and CFB Complement Genes

Age-Related Macular Degeneration (AMD) is a debilitating eye disease that is the leading cause of blindness in the elderly throughout the developed world. It is characterised by a variety of pathological changes (such as neovascularisation, atrophy and deposition of waste matter) in the macula, a region of the retina responsive for detailed central vision.

We have investigated the association of Complement Component 2 (C2) and Complement Factor B (CFB) variants with AMD progression and severity phenotypes in a comprehensively phenotyped cross-sectional case series of 1013 AMD cases collected as part of the Western Australian Macular Degeneration Study. A tagging set of 20 Single Nucleotide Polymorphisms (SNPs) were genotyped in the adjacent C2/CFB genes.

We have found some significant associations between C2/CB SNPs and different AMD phenotypes. Our results suggest that C2/CFB genetic variants lead to altered/compromised functioning of the alternative complement pathway, and that this is concomitant to increased risk of developing neovascular or atrophic AMD.

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