On September 20, AEVR and its co-sponsors hosted a Congressional briefing in recognition of International Age-related Macular Degeneration (AMD) Awareness Week 2012. Since AMD is the leading cause of blindness and low vision in the developed world due to the loss of central vision, events held globally this week recognize its impact in terms of decreased productivity, loss of independence, and reduced quality of life. However, with each year, there is encouraging new research into diagnostics and treatments for this disease as the first generation of FDA-approved drugs to inhibit neovascularization.

He noted that a refined understanding of genotype and phenotype associations—clinical, biological, pathological—in disease development is a critical step toward the development of effective diagnostics and therapies. In that regard, his research suggests that AMD is likely multiple diseases with substantial overlap and that it co-segregates with other major systemic diseases. He reported that late-stage AMD can develop independently from either the chromosome 1 or the chromosome 10 genes and that some individuals can develop late-stage AMD in the absence of drusen. This has led him to propose that macular drusen are not significantly associated with pure Chromosome 10-directed AMD. As evidence, he notes that AMD in African Americans is associated with drusen, but neovascularization is rare, likely due to a scarcity of chromosome 10 gene variants, while AMD in Asians is primarily neovascular and drusen are uncommon, likely due to a scarcity of chromosome 1 gene variants.

Additionally, he discussed the association between reticular pseudodrusen and late-stage AMD.

Research suggests that AMD is likely multiple diseases with substantial overlap and that it co-segregates with other major systemic diseases. – Dr. Hageman

Reticular pseudodrusen are macular lesions that are similar in size and distribution to unique choroidal (a vascular layer next to the retina) capillaries called the choriocapillaris. Although the lesions had previously been thought to be AMD-related drusen, advances in imaging techniques have demonstrated it as a separate disease phenotype. In a Utah cohort, the reticular pseudodrusen phenotype is significantly associated with nearly 70 percent of all severe late-stage AMD, is female predominant, and can occur in the absence of both chromosome 1 and chromosome 10 gene variants, suggesting that it may be a new AMD disease entity.

Armed with these data, Dr. Hageman is directing a team of clinicians and researchers to identify and validate therapeutic targets for early-stage AMD and its co-segregating diseases, thereby shortening the drug-development process. "There is a compelling national need to develop diagnostic tests and therapeutics to treat and prevent this devastating condition," he concluded.

On September 13, AEVR joined Lighthouse International and other co-sponsors in hosting a Congressional Briefing on low vision and vision rehabilitation, featuring Lighthouse Chief of Low Vision Bruce Rosenthal, O.D.

Lighthouse Educates About Low Vision

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