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## AEVR Congressional Briefing Addresses the Importance for Vision Health of the Interplay Between Genes and Lifestyle

On February 20, AEVR sponsored a Capitol Hill briefing featuring Debra Schaumberg, Sc.D., O.D., M.P.H. (Harvard Medical School), who described her National Eye Institute (NEI)-funded research into the interplay of genes and lifestyle in age-related macular degeneration (AMD), the leading cause of vision loss that affects approximately nine million Americans and will impact many more as baby boomers age. AEVR's briefing was held in conjunction with the Women's Eye Health Task Force and the Society for Women's Health Research.



Dr. Schaumberg spoke about the importance of population-based NIH-sponsored studies with Dr. David Noll of the office of Sen. Ted Kennedy (D-MA), Chair of the Senate Health, Education, Labor, and Pensions (HELP) Committee, with jurisdiction over the NIH.



[www.eyeresearch.org](http://www.eyeresearch.org)

12300 Twinbrook Parkway, Suite 250  
Rockville, Maryland 20852-1606

Phone: 240-221-2905

Fax: 240-221-0370

Email: [jamesj@eyeresearch.org](mailto:jamesj@eyeresearch.org)



Dr. Schaumberg's cutting-edge research uses statistical and epidemiological methods to study AMD, a complex disease where multiple genetic and lifestyle variables play a role. Through uncovering the genetic secrets of AMD, as well as studying the potential impact of lifestyle factors such as obesity and cigarette smoking that appear to influence the expression of these genes, Dr. Schaumberg can more fully quantify the risk for incidence of the disease, thereby enabling disease prevention or more rapid diagnosis and treatment.

Dr. Schaumberg's work arises from findings from the National Institutes of Health's (NIH) Human Genome Project, announced in 2005, in which two genes (Complement Factor H, or CFH, and LOC387715) were found that contain variants that account for the majority of cases of AMD. However, after studying large populations of individuals with these gene variants—representing studies conducted by various Institutes within NIH—Dr. Schaumberg observed that many people with these variants do not develop AMD. “The high prevalence of the CFH and LOC gene variants in the population suggested to us that lifestyle factors could strongly influence the effect of the genetic variants on risk of AMD,” noted Schaumberg.

For example, in people with the CFH gene variant, cigarette smoking increases the risk of AMD by 8-fold, and obesity increases the risk by 12-fold. In people with the LOC gene variant, cigarette smoking increases the risk of AMD by 20-fold, and obesity increases the risk by 9-fold. “Obviously, knowing what a single gene variant is doesn't reveal the whole story, which likely includes multiple genetic variants, possibly some with a protective influence, and their interplay with other factors



Dr. Schaumberg speaks, then meets with Martha Nolan of the Society for Women's Health Research, which along with the Women's Eye Health Task Force co-sponsored this AEVR event.

about which we are still learning, including some modifiable risk factors,” said Schaumberg, who added that this research into AMD will also help to establish a paradigm for the study of other serious eye disorders—such as glaucoma, cataract, diabetic retinopathy, and dry eye syndrome—where genetics and lifestyle also play a role. She concluded by stating that, “Right now, it's important to pay attention to one mystery we have solved, which is that development of AMD can be influenced by smoking and obesity, particularly among those with certain common genetic variants.”

AEVR Executive Director James Jorkasky concluded the briefing by emphasizing that the research being conducted by Dr. Schaumberg's team is a prime example not only of trans-Institute NIH research, but of the new paradigm for research and clinical practice for the 21st century as described by NIH Director Dr. Elias Zerhouni—that which is predictive, preemptive, preventive, and personalized to each individual.