

AEVR Briefing Addresses the Interplay between Genes and Lifestyle in Vision Health

On February 20, AEVR sponsored a Congressional briefing featuring Dr. Debra Schaumberg (Harvard Medical School), who described her NEI-funded research into the interplay of genes and lifestyle in AMD. The briefing was held in conjunction with the Women's Eye Health Task Force and the Society for Women's Health Research.

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 NIH's *Human Genome Project*, announced in 2005, in which two genes (Complement Factor H, or CHF, and LOC387715) were found to 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 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.”



Dr. Schaumberg speaks about the importance of NIH-sponsored trans-Institute large population-based 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.



Changing the Face of Medicine

Vision Research Community Discusses Pivotal Role of Ocular Epidemiology in the Translation of Basic Research

The *Fourth U.S. Symposium on Ocular Epidemiology*, held January 29-31 by Johns Hopkins University through an NEI grant, drew vision researchers throughout the country. In keynote comments, Dr. Harvey Fineberg, President of the Institute of Medicine of the National Academy of Sciences, described the challenges that ocular epidemiology faces, which parallel the new paradigm for healthcare research and delivery envisioned by NIH Director Dr. Zerhouni.



Dr. Fineberg and NEI Director Dr. Paul Sieving

Joyce Schiffman (Bascom Palmer Eye Institute), Dr. Rohit Varma (Doheny Eye Institute), and NEI's Dr. Emily Chew