World Glaucoma Week 2014 Briefing Focuses on Genetic Basis of Glaucoma

On February 26, AEV R held its World Glaucoma Week 2014 Congressional Briefing two weeks earlier than other worldwide events held during March 9-15 since the American Glaucoma Society (AGS) was holding its Annual Meeting in Washington, D.C. Held in conjunction with the AGS Advocacy Day and co-hosted by all major glaucoma societies and research organizations (see boxes below), the briefing was the first time AEV R featured NEI-funded researchers engaged in studying the genetic basis of glaucoma—which will lead to new insights into the molecular pathogenesis, effective screening and prevention strategies, and more rational treatment approaches.

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—Dr. Wiggs

Entitled Determining the Genetic Basis of Glaucoma to Develop Novel Treatments, the briefing featured two researchers from Harvard Medical School’s Massachusetts Eye and Ear Infirmary—Janey Wiggs, M.D., Ph.D., the Associate Chief of Ophthalmology Clinical Research and Associate Director of the Ocular Genomics Institute and Louis Pasquale, M.D., Director of Glaucoma Service and Director of Telemedicine. Both participate in the NEI Glaucoma Human Genetics Collaboration (NEIGHBOR) Consortium and augmented Glaucoma Gene Environment Initiative (GLAUGEN), a joint project of the NEI with the National Human Genome Research Institute (NHGRI). These collaborative efforts are under the umbrella of a larger NEIGHBOR HERITABLE Overall Operational Database (NEIGHBORHOOD) that involves clinicians and geneticists at multiple institutions throughout the U.S. The consortium’s goal is to identify genetic variants associated with Primary Open Angle Glaucoma (POAG), the most common form of the disease. NEIGHBOR is unique because it is the largest Genome-Wide Association Study (GWAS) for POAG to-date and has identified the first genetic risk factors for normal pressure glaucoma.

Dr. Wiggs explained that the research’s overall goal is to identify glaucoma risk factors, which can be both genetic and environmental. This makes it possible to develop screening and diagnostic tests that can identify those at risk before irreversible damage to the optic nerve. Characterizing the genes and environmental exposures will help define the molecular abnormalities responsible for the disease, which is the first step toward developing novel therapies targeted to the disease-causing events. She spoke primarily about NEIGHBOR’s focus on adult-onset POAG, since it is strongly age-related and has “complex inheritance,” meaning that multiple genes, each with moderate effects, contribute to the disease susceptibility. NEIGHBOR created the NEIGHBORHOOD database to compare the distribution of genetic markers distributed through the human genome in a very large number of glaucoma cases and controls. She described the gene variants identified to-date, noting that there is still much to discover, especially with regard to rare gene variants and their impact on disease risk.

Dr. Pasquale described the use of The Nurses’ Health Study—which was begun at Harvard in 1976, initially included 120,000 participants, and tracks incidence of 40 different diseases, including glaucoma—to research a potential link between estrogen levels and POAG risk pre- and post-menopause. He cited a number of studies that have shown a relationship between attributes of female reproductive health and increased POAG risk. He also provided evidence that the relation between declining estrogen levels and POAG has a genetic basis. Specifically, he discussed how common genetic variants involved in estrogen metabolism and nitric oxide signaling are related to POAG. He concluded by noting that both environmental and genetic factors relating to estrogen levels are associated with POAG, and that gender biology research has identified several potential targets for POAG, including estrogen itself and components of the nitric oxide signaling pathway.

AGS Members Educate Capitol Hill about Glaucoma Research and Clinical Practice

AEVR coordinated its March 26 event with the AGS Advocacy Day, which was held just prior to the start of its Annual Meeting. A total of 52 AGS advocates from 23 states participated in 105 visits with Members of Congress, sharing their message about the impact of glaucoma, why it is vital to fund this research within the NEI, and the importance of the glaucoma sub-specialty clinical practice. NAEVR was pleased to assist AGS and to accompany its members on Hill office visits. Later that day, the AGS joined with the FDA in convening a workshop to advance the science of minimally invasive glaucoma surgery devices. At the AGS Annual Meeting, Dr. Wiggs provided the Clinician-Scientist Lecture, delineating two different approaches to using genetics in glaucoma research depending on whether the disease occurs early or late.

Lindsay Rhodes, M.D. (University of Alabama at Birmingham) meets with Sen. Richard Shelby (R-AL), a member of the Senate Labor, Health and Human Services, and Education (LHHS) Appropriations Subcommittee who previously served as its Ranking Member. Dr. Rhodes worked on Capitol Hill for Senate Majority Leader Harry Reid (D-NV) prior to attending medical school.

World Glaucoma Week History

The first World Glaucoma Day was held on March 6, 2008, and the U.S. House of Representatives passed H.R. 981, which recognized the event and supported NEI’s efforts to research the causes of and treatments for glaucoma. Since 2010, the day has expanded into a week of events held worldwide, with all major glaucoma professional societies and research organizations co-sponsoring AEV R’s 2014 event, including, AGS, ARVO, Glaucoma Research Foundation, Optometric Glaucoma Society, and The Glaucoma Foundation.

Prior to the briefing, Cong. Gene Green (D-TX) dropped by between hearings to meet the speakers and thank them for their dedication to glaucoma research.

Kelley Kurtz from the office of Cong. Rob Woodall (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma (R-GA) with Abdul Agermoune, a parent whose child was recently diagnosed with infantile-onset glaucoma.