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AEVR in Action

Hill Briefing Highlights Research Effort that is Helping the Blind to See

Two of the world's top retinal researchers came to Capitol Hill on June 24 to educate congressional staff about their landmark clinical trial in which they used gene therapy to restore some vision in three young adults who were virtually blind from a severe form of retinitis pigmentosa known as Leber congenital amaurosis (LCA). Seventy people attended the briefing, which was hosted by the Alliance for Eye and Vision Research (AEVR) and the Foundation Fighting Blindness (FFB).



University of Pennsylvania researchers Jean Bennett, M.D., Ph.D., and Al Maguire, M.D., respond to attendee questions

The researchers' historic advancement opens the door for using gene therapy to treat a variety of blinding retinal diseases, including age-related macular degeneration (AMD), the leading cause of blindness in people over 50, as well as a number of other health conditions that impact the quality of life of millions and cost taxpayers many billions of dollars. Vision loss and blindness alone cost Americans nearly \$70 billion annually.

Jean Bennett, M.D., Ph.D., the study's Scientific Director, and Al Maguire, M.D., the study's Principal Investigator, both of whom are at the University of Pennsylvania, provided details of their clinical study, which is taking place at the Children's Hospital of Philadelphia (CHOP). The researchers noted that critical funding from the National Eye Institute (NEI) within the National Institutes of Health (NIH) and the Foundation Fighting Blindness, a nonprofit organization that funds research to cure retinal degenerative diseases, made their advancement possible.



NEI Director Paul Sieving, M.D., Ph.D., who is known internationally for his research into the genetic basis of retinal neurodegenerative diseases, provided an historical context to the current human gene therapy trials

Paul Sieving, M.D., Director of the National Eye Institute, was also on hand to deliver remarks, in which he called the researchers' work a "stunning outcome" and a great testament to the value of research funding. He added, "This effort is the tip of the iceberg. This validates the process of putting genes in the body for the purpose of restoring vision, liver function, heart function, and treating many other conditions." Initial results of the study were published in the *New England Journal of Medicine* on April 27, 2008. News of the advancement was carried by dozens of major media outlets around the world including: ABC (Good Morning America), NBC, CBS, *The Wall Street Journal*, and *The Washington Post*.

Seven years ago, Dr. Bennett came to Capitol Hill to share the results from a preclinical study of the same gene therapy, which was at the time successfully giving vision to dogs born blind from LCA. During her June 24 Hill visit, she said, "We predicted seven years ago at a similar venue that this approach could cure blindness in humans. We are thrilled to be here today to tell you that this prediction appears to be coming true." Dr. Maguire, a vitreoretinal surgeon, provided technical details of the study, including how the corrective gene is delivered to the retina using a therapeutic man-made virus or adeno-associated virus (AAV) — an approach that is also being used in studies of diseases such as muscular dystrophy and hemophilia.

Though the primary goal of the Phase I study at CHOP is to ensure safety of the treatment, Dr. Maguire noted that the patients' vision was also tested objectively and subjectively. After treatment, they were able to read several lines on an eye chart. They also had improved peripheral vision and better eyesight in dimly lit settings. Dr. Maguire said that he knew the treatment was working well when the patients asked to receive the therapy in their untreated eyes. The investigators will be treating LCA patients as young as eight years old. They believe the most

dramatic results will be seen in young children.

Dr. Bennett is also leading an effort that will use gene therapy to treat an inherited form of macular degeneration known as Stargardt disease. She hopes to begin a clinical study of that treatment in about 18 months. Commercial companies are also investigating gene therapy for treating age-related macular degeneration — a condition that affects more than 10 million Americans and for which few treatments exist.

In concluding the briefing, Dr. Sieving put historical perspective on the breakthrough. "The roots of this trial go back many years. A gene that causes this disease, RPE65, was found 15 years ago," he said. Dr. Sieving also noted that the discovery that vitamin A is essential for vision — a Nobel-Prize-winning finding made by George Wald, M.D. (hon), Ph.D., in the 1930s — was also a crucial stepping stone to the recent gene therapy advancement. "This is a very exciting time for all of medicine," he added. "Because this is a time when charitable and taxpayer dollars are being converted into treatments for people."

The research teams working on these projects also received funding from private sources Research to Prevent Blindness and Fight for Sight.



Dr. Bennett and Dr. Maguire with Edward Gollob, a member of the Boards of Directors of both AEVR and FFB



FFB Chief Research Officer Stephen Rose, Ph.D. (second left) is joined by representatives of the Blinded Veterans Association, including Tom Zampieri, Ph.D., Steve Matthews, and Claudia Perry



Dr. Bennett and Dr. Maguire met with Elizabeth King (right), Senior Legislative Assistant to Cong. Chaka Fattah (D-PA), in whose district the University of Pennsylvania is located. Cong. Fattah is a member of the House Committee on Appropriations.



Left to right: AEVR Executive Director James Jorkasky, NEI's Tom Hoglund, FFB's Ben Shaberman, and NEI's Shefa Gordon, Ph.D.



Left to right: Yewlin Chee and Lauren Stein, two students conducting research at University of Pennsylvania, speak with Norm Lanphear of the American Academy of Ophthalmology



Left to right: Steve Grossman of the Alliance for a Stronger FDA discusses with FFB's Dr. Rose the potential FDA requirements of a treatment emerging from the trials