Capitol Hill Briefing Highlights Promising Human Gene Therapy Trial

On June 24, two top retinal researchers educated 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 AEVR and the Foundation Fighting Blindness (FFB), a nonprofit organization that funds research to cure retinal degenerative diseases and a NAEVR member organization.

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 NEI and FFB made their advancement possible.

NEI Director Dr. Paul Sieving also presented 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.

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.

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.

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.