



Alliance For Eye  
And Vision Research

**AEVR**

*Educating about the Value of Vision Research*



**ALLIANCE FOR EYE AND VISION RESEARCH (AEVR)  
in conjunction with  
THE FOUNDATION FIGHTING BLINDNESS (FFB)**

*continues its series of educational briefings on exciting new  
developments in eye and vision research*

**Please join our presenters:**

**Paul Sieving, M.D., Ph.D. (NEI)**

**Jean Bennett, M.D., Ph.D., and Al Maguire, M.D.  
(University of Pennsylvania)**

**For a Luncheon Briefing**

***“Human Gene Therapy for Treating Blinding Eye  
Diseases: Off and Running”***

**Tuesday, June 24, 2008**

**12:00 Noon – 1:15 pm**

**House Rayburn B-339**

**Please R.S.V.P. to**

**Dina Beaumont @ 202-530-4672 or [dbeaumont@FightBlindness.org](mailto:dbeaumont@FightBlindness.org)**

**Note: AEVR and FFB are 501(c)3 Non-Profit Educational Foundations hosting this  
widely attended event with meal value less than \$50**

**AEVR/FFB Luncheon Briefing**  
**“Human Gene Therapy for Treating Blinding Eye Diseases: Off and Running”**  
**Tuesday, June 24, 2008 12 Noon - 1:00 pm, House Rayburn B - 339**  
**R.S.V.P. to: 202-530-4672 or [dbeaumont@FightBlindness.org](mailto:dbeaumont@FightBlindness.org)**

**What is the focus of this briefing, and why is it important?**

The briefing reports on very promising results of Phase I clinical trials designed to examine the safety of gene transfer to treat a debilitating form of childhood blindness called Leber congenital amaurosis (LCA), which is a degenerative disease of the retina, the light sensitive neural tissue in the back of the eye. Two papers describing these exciting results appeared in the April 27<sup>th</sup> online edition of the *New England Journal of Medicine*. A third important clinical trial of this treatment is well underway. The treated individuals have a specific form of LCA caused by mutations in the RPE65 gene. The normal RPE65 gene codes for the RPE65 protein, an enzyme that generates the type of vitamin A that the retina requires to respond to light. To circumvent this problem, the researchers injected millions of copies of the normal RPE65 gene into the tiny space beneath the retina using an inactivated virus. This would allow the retina to produce the normal RPE65 gene, thereby restoring not only light sensing capability to these individuals, but also potentially enabling them to actually “see” more.

Initial observations with six participants indicate that this gene transfer therapy appears to be safe and well-tolerated. While four of the six participants also reported an improvement in vision, three participants saw enough improvement that they could read 3 ½ more lines on an eye chart. Investigators will soon have the opportunity to also examine the published data from the third Phase I clinical trial. The results from all three Phase I trials will enable researchers to refine the gene transfer techniques and will inform the design of the next set of clinical trials to evaluate the efficacy of this treatment in a rigorous, objective way. If LCA can be treated effectively with gene transfer, it will point the way toward treatment of other eye diseases caused by single, malfunctioning genes, facilitating treatment of many previously untreatable blinding conditions, such as childhood macular degeneration (Stargardt’s disease), blindness in deaf-blindness (Usher syndrome), and retinitis pigmentosa.

**How is this breakthrough research being funded?**

Before the gene transfer procedure could be tested on people, four critical milestones had to be met: discovery of the RPE65 gene; creation of animal models that illustrate the gene’s functions and what happens when it is missing; development of a safe way to carry healthy replacement genes to the target within the eye; and studies of the procedure in a large animal model. Most of this work was funded by the National Eye Institute (NEI) within the National Institutes of Health (NIH) and co-funded by the Foundation Fighting Blindness (FFB), a private funding foundation. The seminal proof-of-concept for gene transfer as a treatment for LCA was demonstrated by investigators supported by the NEI and FFB in a dog breed that exhibits LCA, most notably “Lancelot” who visited Congress in 2001. Restoration of visual function in these affected dogs has been remarkable and long-lasting, with Lancelot maintaining vision for more than eight years. The two U.S.-based, independent investigative teams are funded by the NEI, FFB, Research to Prevent Blindness, and other organizations. A third team based in the United Kingdom is funded in part by FFB and Fight for Sight.

**About the speakers...**

- **Paul Sieving, M.D., Ph.D.**, the NEI’s Director, is known internationally for his research into the genetic basis of retinal neurodegenerative diseases
  
- **Jean Bennett, M.D., Ph.D.**, the F.M. Kirby Professor of Molecular Ophthalmology and the Vice Chair for Research in Ophthalmology at the University of Pennsylvania, is the Scientific Director of the LCA study, and **Al Maguire, M.D.**, an Associate Professor at the Scheie Eye Institute/University of Pennsylvania, is Principal Investigator.

**About AEVR...**

The Alliance for Eye and Vision Research (AEVR) is a 501(c)3 non-profit foundation dedicated to education about the importance of federal funding for eye and vision research.

**About FFB...**

The Foundation Fighting Blindness, Inc., is a 501(c)3 non-profit foundation dedicated to research that will provide preventions, treatments, and cures for people affected by retinitis pigmentosa (RP), macular degeneration, Usher Syndrome, and the entire spectrum of retinal degenerative diseases.

**AEVR and FFB are pleased to host this widely attended event, with a meal value of less than \$50.**