

NEI's Dr. Sieving Educates Capitol Hill about Rare Eye Diseases



At an October 31 Congressional briefing sponsored by the Foundation Fighting Blindness (FFB) and AEVR, top leaders from the vision research community spoke about the critical need for research funding to eliminate retinal diseases that affect more than nine million Americans.

In his welcome, Cong. Pete Sessions (R-TX) stated that, "Today the message is very clear: There is much that can and must be done to overcome these blinding conditions. It is our responsibility through good public policy to recognize that we have the ability to find cures for these diseases." Cong. Sessions, who has a teenage son affected by retinitis pigmentosa (RP), added, "When you have a 16-year old son who wants to be a physician and has a great life ahead of him, you're crushed when you learn he is losing his eyesight to RP." He also stated that it is in the public's best interest to ensure that the best, most-promising research technologies are investigated and employed, including the potential of stem cell therapies for treating



Cong. Pete Sessions (R-TX) offers a welcome to the 70-plus attendees

vision loss. "We need to find out what stem cells can do for us in order to make wise public policy," he said.

Stephen Rose, Ph.D., Chief Research Officer for FFB, which is the largest source of non-governmental funding for retinal degenerative research in the world, and NEI Director Paul Sieving discussed why partnerships between government and the private sector are beneficial to advancing research. For example, both Dr. Rose and Dr. Sieving noted that an FFB-NEI partnership helped to advance a treatment called Encapsulated Cell Technology (ECT), which is now in Phase II/III studies across the country for treatment of a variety of retinal degenerative conditions, including RP, the "dry" form of AMD, and Usher syndrome (combined blindness and deafness). "In an FFB-funded lab study, ECT showed promise for saving vision, so the NEI subsequently conducted Phase I safety studies of it," said Dr. Rose. During the Phase I trial, ECT saved and in some cases restored vision. More than 150 people are now enrolled in the Phase II/III studies, which are funded in part by FFB.

Dr. Sieving, who showed the tiny ECT to attendees added, "What is learned in treating the eye may translate into benefits for treating other neurodegenerative diseases such as ALS, Parkinson's disease, and Alzheimer's disease. One of the advantages of working in the eye is that it is easier to



NEI Director Dr. Paul Sieving, FFB CEO Bill Schmidt, and Bobby Hillert from the office of Cong. Sessions

get the device into the eye than into the skull." In addressing the genetic basis of eye disease, Dr. Sieving noted that of the 25,000 genes in the body, about 10 percent or 2,000 of those have been cloned, and about one-quarter of these have been found to cause eye disease.

Though many retinal diseases are considered to be rare or "orphan"—meaning that they affect less than 200,000 people—research for orphan diseases is a critical public health issue. Collectively, 25 million Americans are affected by some orphan disease. Dr. Sieving said that, over time, many common conditions will be subdivided into smaller, genetically related groups. "Heart disease, diabetes, and cancer are all genetically driven diseases. We are finding that all common diseases in fact splinter into groups of rare genetic diseases, and we are beginning to treat these conditions based on their genetic profile."

Dr. Rose remarked that it is an extraordinarily promising time in vision research because of the numerous human studies emerging and underway to save and restore vision. He said, "Ten years ago, a person with a retinal degenerative disease was told 'There's nothing we can do for you, go home and learn Braille, you will go blind.'" Today, thanks to funding from the NEI and FFB, there are clinical trials underway to eradicate these diseases. There is a lot of hope."



FFB's Dr. Stephen Rose details research into rare eye diseases