NEI Collaborations Yield Exciting Results

NEI’s history of collaborations with other NIH Institutes and Centers, other federal agencies, and with private funding foundations is resulting in exciting results for patients.

- NEI’s research with the National Cancer Institute (NCI) and National Heart, Lung, and Blood Institute (NHLBI) into factors that inhibit new blood vessel growth (angiogenesis) has resulted in the first generation of ophthalmic drugs to treat the “wet” form of AMD, stabilizing and restoring vision. In “wet” AMD, abnormal blood vessels grow in the light-sensitive back of the eye called the retina, leaking serum and blood and damaging the macula, which is necessary for central vision used for reading and driving. One of these drugs, Lucentis, has also been approved by the Food and Drug Administration (FDA) for use in treating diabetic macular edema, while other drugs are currently undergoing regulatory review.

- NEI’s Diabetic Retinopathy Clinical Research (DRCR) Network—a multi-center network dedicated to facilitating clinical research—confirmed that laser treatment for diabetic macular edema, when combined with anti-angiogenic drug treatment, is more effective than laser treatment alone, the latter of which has been the standard of care for the past 25 years. The current DRCR Network is a successor to several previous networks in which NEI has collaborated with the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK).

- NEI and the National Aeronautics and Space Administration (NASA) reported on the use of a compact fiber-optic probe developed for the space program that is showing promise as the first non-invasive early detection device for cataracts, the leading cause of vision loss worldwide. Using a laser light technique called dynamic light scattering (DLS), which was developed to analyze growth of protein crystals in a zero-gravity environment, the probe measures the amount of light scattering by a protein in the eye’s lens called alpha-crystallin. The probe senses protein damage due to oxidative stress, a key process in many medical conditions including cataract and diabetes, as well as Alzheimer’s and Parkinson’s disease. The probe is not yet available to the general public, but is being used in research to investigate early stages of cataract formation and to develop potential therapies.

- The NEI-led Phase 1 human gene therapy clinical trial for Leber Congenital Amaurosis (LCA), a neurodegenerative eye disease that affects children and young adults, has resulted in patients experiencing visual improvement. NEI’s pioneering work conducted with the privately-funded Foundation Fighting Blindness, as well as subsequent refinement of gene therapy techniques, is enabling further privately-funded clinical trials for AMD, choroideremia, Stargardt disease, and Usher Syndrome (deafblindness). The latter three neurodegenerative diseases occur in early childhood and progressively destroy the retina, resulting in a lifetime of direct medical costs and indirect support costs.

NEI Leadership: Genetic Basis of Eye Disease

NEI-funded researchers have discovered more than 500 genes associated with both common and rare eye diseases, which is 7.5 percent of all disease-causing genes discovered to-date. NEI has especially made great strides in determining the genetic basis of AMD—the leading cause of vision loss in the U.S. and a disease for which little could be done just a few short years ago. NEI’s AMD Gene Consortium, a network of international investigators representing 18 research groups, has discovered seven new regions of the human genome—called loci—that are associated with increased risk of AMD. They also confirmed 12 loci already identified in previous studies. The loci implicate a variety of biological functions, including regulation of the immune system, maintenance of cellular structure, growth and permeability of blood vessels, lipid metabolism, and atherosclerosis. Understanding the genetic basis of the disease and underlying mechanisms will lead to better diagnostics and therapies.

Retinal Prosthesis System: A Patient Perspective

In February 2013, the FDA approved the implanted Argus II Retinal Prosthesis System to treat adult patients with advanced Retinitis Pigmentosa (RP), a rare genetic condition affecting 100,000 Americans and 1.5 million people worldwide. This inherited, progressive neurodegenerative disease causes rod and cone photoreceptors in the light-sensitive back of the eye to degenerate, leading to total or near-total blindness.

The device, manufactured by Second Sight Medical Products, Inc., is a result of collaboration between the Department of Energy’s National Laboratories and its Artificial Retina Program and the NEI, which funded research at four universities and at Second Sight. In the device, a small video camera mounted on a pair of glasses sends images to a video processing unit that converts them to electronic data that is wirelessly transmitted to an array of electrodes implanted onto the retina, referred to as a “chip.”

The device is enabling those who are otherwise blind to identify doors, crosswalks, and even utensils, as evidenced by Paul D’Addario, a former trade association database manager from Arlington, Virginia and RP patient who had not been able to see since 2004. In 2007, he had the second-generation 60-electrode chip implanted onto his retina at Johns Hopkins University. As soon as the system was activated, he saw “flashes of light,” which upon further training he was able to recognize as contrast between objects as he scanned with the device’s eyeglass-mounted video camera. “I had to train my brain as I scanned, interpreting the flashes to create a contrast, determining the outline and size of objects.”

He uses the device both at home and in the neighborhood, especially scanning objects at intersections, and is pleased that upgrades to the visual processing unit continue to improve his results. He is excited about the next generations of the product, where arrays of upwards of 1,000 electrodes on the chip may enable more users to read large print and recognize the faces of loved ones. “The future is looking better than I thought it would, much quicker than I thought it would.”

Eye Fact

In 2010, the World Health Organization (WHO) reported that 285 million people in the world are visually impaired, with 39 million blind and 246 million with moderate-to-severe visual impairment. Cataract, glaucoma, and AMD are the top causes of blindness, with 42 percent of visual impairment due to refractive errors.

Wearing the Argus II Retinal Prosthesis System, Paul D’Addario sorts white and black socks—an activity of daily living.