“Anyone Can Be An Advocate”

Advocacy for people affected by blindness and low vision is an important and much-needed cause to which we can all relate. Blindness and low vision advocacy means taking an active part in improving treatment, health care, and daily living for those among us who cannot, or do not, always speak for themselves.

Formal training is not required to be an advocate, but the title is usually justified by knowledge, experience, and passion. The cause could be as far-reaching as human rights, education, and poverty, or it could be as simple as speaking out about litter in the neighborhood.

Advocacy is fueled by natural born empathy. The capacity for empathy can be innate, it can be acquired by way of relationships with others, or it can be activated by memories of one’s own personal experiences. But it cannot be taught. Having walked in someone else’s shoes is the best way to learn it.

And all of us are walking in on those shoes, which means that not only do we have the right to speak out, we have an obligation to do so. I hope this month’s presentation will encourage us to speak or act out when opportunities arise that call for it.

Dan Roberts
News & Information

First Gene Therapy for AMD Performed

Researchers at the University of Oxford have carried out the world’s first gene therapy operation to tackle the root cause of age-related macular degeneration (AMD).

An 80-year-old woman with AMD is the first of ten people to receive the treatment. The procedure was carried out at the John Radcliffe Hospital by Professor Robert MacLaren, Professor of Ophthalmology, in a clinical trial (FOCUS) sponsored by Gyroscope Therapeutics. It is too early to know if the patient’s sight loss has been halted, but her vision and all that of all other subjects will be closely monitored. This first stage of the trial is primarily designed to check the safety of the procedure.

The operation involves detaching the retina and injecting a solution containing a benign virus into the retinal pigment epithelium (RPE) layer underneath. The virus contains a modified DNA sequence, which infects cells and corrects a genetic defect that causes AMD. Ideally if successful, gene therapy would only need to be performed once, as the effects are thought to be long-lasting.

A key factor in AMD is the complement system, a system of proteins in our immune system that fights bacteria. In macular degeneration, these proteins are over-active and start to attack the retinal cells, similar to how they would attack bacteria.

"The idea of this gene therapy is to ‘deactivate’ the complement system, but at a very specific point at the back of the eye, so the patient would otherwise be unaffected by it." If successful, it is hoped that gene therapy can be used in the future on patients with early AMD to halt the disease before their vision has started to deteriorate.

It is important to emphasize that this study is a very early-stage intervention that will not restore sight, but it is expected to prevent further deterioration. Additionally, the defective CF-1 gene occurs in only about five percent of AMD patients, so the number of candidates for the treatment is relatively small. Still, this is a very important proof-of-concept study that holds a great deal of promise for the future.
What is Artificial Intelligence and How Can It Help Me?
by Dan Roberts

Artificial intelligence (AI) makes it possible for machines to learn from experience, adjust to new inputs, and perform human-like tasks. It provides us with human-like interactions with computers and helps us with difficult decision making.

AI works by super-speed processing of large amounts of information (data), allowing the software to learn automatically from patterns or features in the data, and then to predict likely outcomes. In other words, it imitates the learning and decision-making functions of the human brain. But it is much faster, often more reliable, and it requires minimal human intervention. AI still requires human interaction, however, so it is not ready to completely replace us. Instead, it will allow us to be more efficient in our work and daily life, especially those of us who are blind and visually impaired.

AI relies heavily on deep learning and natural language processing. Deep learning is a type of machine learning that trains a computer to perform human-like tasks, such as recognizing speech, identifying images or making predictions. Natural language processing helps computers understand, interpret, and manipulate human language. This is what helps computers communicate with humans by reading text, hearing and interpreting speech, recognizing emotion, and determining which of those parts are important.

AI is in the process of incorporation into many devices for blind and visually impaired people. Such devices include self-driving vehicles, reading machines, face recognition software, and navigation software. In addition, the capabilities of AI in tandem with eye care, diagnosis, and treatment are being dramatically realized, with new uses seemingly being discovered daily.

The full potential of AI is yet to be seen. In the areas of eye care and low vision rehabilitation, we can look forward to it playing a major role in helping to:

- Ensure drug safety by enabling pharmaceutical manufacturers to quickly determine the quality, efficacy, and safety of new products
- Get new therapies to market faster
• Speed up clinical trials
• Ensure more accurate clinical diagnoses
• Ensure optimum treatment choices by doctors
• Make more precise decisions about optimal daily living choices

Thinkers like Steve Wozniak, Bill Gates, and the late Stephen Hawking have warned about AI getting out of hand. Control, after all, is usually enjoyed by the smartest among us, so who wouldn’t worry about machines that are more intelligent than we are? The technology discussed here, however, has been of the “narrow AI” variety, wherein humans set the limits of the machines, keeping them subordinate to us and our whims. There is room for concern about “general AI” machines that might actually approach consciousness, but that is a topic for another time and place. For now, we can enjoy and benefit from the evolution of yet another direction science is taking to make the world a safer and more accessible place.

New Findings in Genetic Pathologies of AMD

With recent successes in gene replacement therapy, scientists are enthused about studying how genetic variants play a part in age-related macular degeneration (AMD).

Previous research has identified 34 small genomic regions ("loci") on the DNA molecule and 52 genetic variants (mutations) within those loci that are associated with AMD. Variants can regulate certain genes to either turn on or off, so the question is “which genes are being regulated by the variants in AMD?”

Led by Anand Swaroop, Ph.D. (chief of the Neurobiology-Neurodegeneration and Repair Laboratory at the National Eye Institute), a study published Feb. 11 in Nature Genetics identified target disease genes at 6 of the 34 AMD loci, the most likely of which are genes labeled B3GLCT and BLOC1S1. Both of these could affect several of the AMD-related cell functions. In addition, three additional target genes were discovered, which had never before been associated with the disease, and 20 more genes were also found that could be candidates for involvement in development of AMD.

This work is exemplative of the exponentially expanding research in the genetics of AMD. Future studies will aim to explain the function
of the target AMD genes to determine how they relate to AMD development and to look for targets for new treatment strategies.

Study Finds That Anti-VEGF Drug Treatments For Wet AMD Do Not Cause Strokes

Three anti-VEGF drugs are in clinical use for treatment of wet age-related macular degeneration (wAMD). These are Lucentis (ranibizumab), off-label Avastin (bevacizumab), and Eylea (ranibizumab).

• The rate of stroke from intravitreal injections of Lucentis was found in the early clinical trials to be 0.2% (1 of 525) in the combined group of patients compared to 0.4% (1 of 260) in the control arms. No followup studies, however, were done to confirm the risk.

• In 2015, The Centre for Adverse Reactions Monitoring (CARM) received two reports of stroke after intravitreal injection of Avastin. Again, the association was not confirmed by further research.

• In addition, some evidence has been reported (3,4) showing a possible association between Eylea and incidence of stroke, with followup research still to come.

This evidence has caused concern among patients who must undergo anti-VEGF injections to save their eyesight. Recent good news, however, is that a 5-year cohort study of 504 patients from Olmsted County, Minnesota has found no consistent associations in the risk of stroke, myocardial infarction, or death among wAMD patients receiving anti-VEGF injections compared with control groups with and without wAMD.

This appears to be the first effort to scientifically pursue the evidence, and the findings may help to ease the concern of both patients and doctors. More such studies, however, should be initiated before any change in protocol is made.
Yet Another Sustained-Delivery Anti-VEGF Drug Ready For Phase 2

A report at the Hawaiian Eye & Retina 2019 conference revealed another sustained-delivery drug, GB-102, for treatment of wet age-related macular degeneration (AMD). Graybug Vision’s ADAGIO study has now provided evidence that GB-102 can continuously inhibit activity of VEGF for several months. The treatment was well-tolerated in the study and found to be free of dose limiting toxicities, drug-related serious adverse events, or inflammation.

88% and 68% percent of evaluable study subjects were maintained on only a single dose of GB-102 at 3 and 6 months respectively. Currently, patients with wet AMD require intravitreal injections every 6 to 8 weeks. Graybug Vision’s Phase 2b study of GB-102, is expected to begin enrollment in 2019.

This is the second anti-VEGF drug in as many months designed to increase the time between injections. Read about all ongoing research for wet AMD treatment.

New Genes Linked to AMD

New research published this week in Clinical Epigenetics, has identified two new genes linked to the development of age-related macular degeneration (AMD).

Led by Dr. Louise Porter at the University of Liverpool, the team identified genes that may become new targets for treatments. 44 human donor eyes were profiled for levels of DNA methylation (a chemical change that may be influenced by sex, age, smoking and diet) and and underlying gene changes in AMD. The researchers were able to identify changes in the newly-discovered genes, SKI and GTF2H4, which have not been previously associated with AMD but are now implicated in the regulation of the disease pathways.

Next Month: “Learning to Live with Low Vision”

An encore presentation from June 2006 featuring a man’s first-person account of his journey from discovery to independence through low vision rehabilitation.