



I-C

Newsletter of Retina Australia (ACT) Inc

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MESSAGE FROM THE PRESIDENT

Welcome to the third newsletter for 2012, within which comes further details of the Retina Australia Congress, being held in Sydney in October, and of the Retina International Congress, held in Hamburg recently. You can read, too, about plans for this year's major fundraiser, a 'must' for the diary, while there is again a considerable amount of heartening research to report.

I would like, on behalf of my colleagues on the committee, to thank all members most sincerely for the positive feedback they provided when approached recently about their involvement in the Inherited Retinal Diseases Register. The information provided has given us a far clearer idea of local involvement, thus enabling us to move forward in our dealings with the Register.

Finally, we forgot this year to make the usual end-of-financial year appeal to members for a small contribution towards our administration costs. Anything you can spare will help offset these expenses and ensure that just that little bit more can be contributed to those all-important research funds.

Best wishes to all...

ROBIN POKE

MEMBERS' "MONTHLY LUNCH DATE"

Where: Café Charisma
Shop 4
2 Colville Street
North Lyneham

Date: 2nd Tuesday of the Month

This is a light and bright café with a delightfully eclectic and very reasonably priced menu, a great ambience and extremely good service. For those who like a drink with lunch, it's BYO.

The next monthly lunch is on:

Tuesday 9 October 2012

Please let me know if you have a problem with transport and I will try to arrange for someone to pick you up.

Jan James
Secretary
Ph 6258 4823
0421 277 880

DATE FOR THE DIARY!

We can confirm that our major fundraiser this year is to be held on Sunday 16 December and will be based on the same very successful format as have been the previous four: an afternoon at Canberra racecourse, also known as Thoroughbred Park, enjoying tipping competitions, raffles and race watching, once more supported by the local Women in Racing group. Lyn Barlow and Jan James are working on ensuring that this year's event will be bigger and better than ever, so put the date in your diary and ask as many of your family and friends attend as possible. Further details will be available in the next newsletter, due to be released following the Retina Australia National Congress, being held at The Sebel at Parramatta, west of Sydney, on 20 and 21 October.

NATIONAL CONGRESS: PLACES STILL AVAILABLE

The Congress, being hosted this year by Retina Australia (NSW), is to be opened by the Hon Dr Barry Jones AO, a former Commonwealth government minister and still a leading advocate for those with vision impairment. Another special guest is Professor Robin Ali from the Moorfields Hospital in London, a globally recognised expert in stem cell research and gene therapy for those with RP. Professor Ali is the leader of the group of ophthalmic surgeons who performed the first of the human gene therapy trials at Moorfields.

But there will also be a host of other interesting speakers on developments in research, and a range of interesting workshops. The national Congress is organised so that people in Australia with retinal degenerative disease can meet with researchers and other experts not only to hear about advances in research but also to join in question time and get some answers to questions about the eye condition in their own family. The Congress will commence with a welcoming Cocktail Party on the evening of Friday 19 October, then on Saturday night there will be a Gala Dinner with professional entertainment and guest speaker Don Burke, Patron of Retina Australia (NSW).

Details of the Congress, including the program and registration details and a range of accommodation and booking information, accompanied the previous newsletter. If you have not yet booked, we encourage you to attend. For those who would prefer to attend just for one day, arrangements are being put in place by RA (ACT) for an excursion on the Saturday of the Congress. Please phone the office on 6281 4519 or 0431 970 850, or Jan James on 6258 4823 or 0421 277 880 if you would like to take up the offer.

THE INHERITED RETINAL DISEASES REGISTER

In the previous edition of *I-C* we asked that members and their families who had contributed their DNA to the Inherited Retinal Diseases Register being compiled by the Sir Charles Gairdner Hospital in Perth inform the executive of this fact. We pointed out that, within the bounds of personal privacy, it was vital that these statistics be known to us, because official figures appeared to conflict with our own.

We have been absolutely delighted by your response! Committee representatives assigned to elicit the information received remarkable cooperation from members, and we are very grateful to you all.

17TH RETINA INTERNATIONAL WORLD CONGRESS
12 – 15 JULY,
HAMBURG, GERMANY

Among the Australian delegates to this Congress were Retina Australia President Graeme Banks and his wife Lynette, and Leighton and Rosemary Boyd, President and Secretary respectively of Retina Australia (Victoria). It is Leighton and Rosemary who prepared this comprehensive summary of the Congress.

There were more than 700 people in attendance during each of the two days of the Congress, the theme of which was "Gateway to Vision". Although the majority of the participants were members of Pro Retina Deutschland, or German citizens, there were a significant number of people who had travelled a long way to attend the Congress. Overall, some 32 countries were represented and it is estimated that the combined membership of their national retina organisations, which make up Retina International, now exceeds 950,000.

The program for the Congress included keynote speakers and themed parallel sessions, wherein the presenters spoke about their research progress, the results so far and their aims for future investigations. There were fifty-two conference speakers, from Germany, the United Kingdom, the United States, Switzerland, Canada, Italy, Ireland, France and the Netherlands. Given many of the sessions were concurrent, Congress participants could choose which of the sessions listed in the program they were interested in and simply go to the allocated venue at the appointed time. The majority of the presentations were made in English and translated simultaneously into German, French or Spanish.

The first keynote lecture, entitled "Success of RPE65 gene- eplacement therapy: How did we get there?" was presented by Professor Andreas Gal, from Hamburg. Prof Gal spoke about his work with the RPE65 gene, more commonly known as Leber Congenital Amaurosis. Mutations in this gene cause the autosomal recessive childhood onset of Retinitis Pigmentosa. His work in gene therapy by injection had reversed the blindness from RPE65 gene in both a mouse and dog model. Also, RPE65 was a candidate for cell replacement therapy. Professor Gal stated that RPE is easy to target surgically and has a single layer of active cells. The gene vector can be turned on slowly and remain active for a long time. It is efficient and targets the gene directly. Human trials are now in progress. An article in the *New York Times* has referred to the trials as "giving sight by therapy with genes". Professor Gal also mentioned that there are a vast array of gene therapies now available and these are being trialled throughout the world.

The second keynote lecture was titled "Interdisciplinarity: The key to progress in therapy of retinal degeneration" and was presented by Professor Eberhart Zrenner from Tübingen, Germany. He said that if new strategies to prevent blindness are to be implemented there needs to have a great deal of interdisciplinary cooperation between doctors and patients, biologists, psychologists, clinics and laboratories, health departments, research institutions, engineers and pharmacologists. Professor Zrenner said that everyone has to work together to find a solution for every individual patient. This is difficult, he said, because there are different "languages" used in each field of medicine and research. He said everyone brings a specific skill but no one can achieve the result without each other and above all, there needs to be a mutual understanding of the outcome being sought. There follows an extract from his address:

"There is a long way from a good idea on a possible new therapeutic strategy to a product available for patients. Many steps have to be taken to transfer findings in basic research from the laboratory into a preclinical and finally into a clinical study, and such a project may take years if not decades. The process can be sped up considerably by interdisciplinary teams working closely together from the start. There are numerous examples of successful translational efforts:

1. Gene therapy

Specialist for virus vectors, biochemists, molecular biologists and geneticists have to work closely in a team to generate a construct, and be able to transport the proper genetic code into a cell that is not properly functioning. Animal models, created by biologists and investigated by physiologists for safety and efficacy are required for proof of concept. Retinal surgeons have to develop optimal procedures for application in man, and psychophysicists have to test the effect. Other specialists deal with the legal and regulatory issues involved in such therapies. Last but not least, patients as pioneers and partners of the team of scientists and physicians have to take some risk to create possible benefits. This particular type of partnership is an indispensable and extremely valuable prerequisite for success in research, showing safety and efficacy.

2. Retina implant studies

Similarly in electronic retinal implants, engineering specialists and material scientists as well as physicists and chemists have to closely work together with retina physiologists in order to create a device that can process images and transport them to retinal neurons. Biostability and biocompatibility of devices have to be established, and novel surgical techniques for applications have to be developed to allow for safe application. Studies in large animals require veterinary specialists and the development of particular testing routines. New tests for "machine vision" that assess the unusual ways of "seeing" have to be developed, including low vision specialists who are able to quantify the increase in mobility and quality of life provided by such devices.

3. Electrostimulation

It has been shown in several animals that electrostimulation releases various

endogenous growth factors. To translate this into clinical studies requires the close cooperation of biochemists and pharmacologists on the one side and electronic engineers and physiologists on the other. Again, pioneer patients have to join the team in order to allow for the development of this type of novel therapy to be first assessed in clinical studies.

Many more examples can be shown where partnership between disciplines as well as between researchers, patients and patient organisations are indispensable for achieving the many small steps of progress that are usually involved until a final goal is reached.

Awareness by the scientists in the various disciplines as well as by patients that novel therapeutic approaches of this kind require close cooperation has been considerably increased in recent years and quite often researchers and patients are presenting results of clinical studies together. This was previously quite unusual during the development of new drugs or devices”.

Final perspectives

As at previous Congresses, Professor Gerald Chader from Los Angeles presented in the final plenary session his perspectives on the progress being made in inherited retinal degeneration research. He commented that in the past no gene mutations for Retinitis Pigmentosa (RP) were known and there was very little idea about the mechanisms of photoreceptor cell death and there were no agents known that could slow photoreceptor degeneration and death. Nowadays, however, about half of the RP mutations are known, and much is known, too, about the basic mechanism of photoreceptor cell death and how to inhibit it. Electronic implants are available to patients and basic work in the fields of stem cell biology and in optogenetics show promise for future treatment.

Professor Chader also spoke about the clinical trials currently in progress for neuroprotection, gene therapy, antioxidant therapy and electronic implants that can improve vision. In summing up, Professor Chader stated: “We are finally passing out of the time of scientific darkness and into the enlightened era of clinical trials. These should soon lead to many new therapies that will save and restore vision in patients with retinal disease.”

Another of the last researchers to speak was Professor Joe Hollyfield, from Cleveland, USA, whose main interest is in Age-related Macular Degeneration (AMD). Professor Hollyfield began his summary by explaining in great detail what and where the macula is. He then spoke about the various treatments currently underway and of the trials for both wet and dry AMD. Although there are some therapies for wet AMD, there are none yet for dry AMD, despite the many studies, and there is still much work to be done. He completed his talk by discussing whether AMD could be prevented in the future.

Other presentations at the Congress fell into the following categories:

- Clinical Basis of Inherited Retinal Degenerations
- Cones in Focus
- Electronic Retinal Implants
- Genetic Diagnosis
- Current and Future Treatment Trials
- Clinical Trials Today and Tomorrow
- Coping with Retinitis Pigmentosa

There was also an AMD Symposium, sessions on Acquired Retinal Disease and Optical Aids, and parallel workshops entitled "Ask the Expert" about Retinitis Pigmentosa, Inherited Maculopathies, X-linked retinopathies, Usher Syndrome, Bardet Biedl Syndromes and Age Related Macular Degenerations. It was fantastic to note how much progress has been made since the previous Congress in Stresa, Italy, two years ago, and to hear about the many different types of clinical trials being conducted, or planned, across the globe. It was also great to see the enthusiasm of the researchers, who are sure that there will be therapies or prosthetic devices available, for all manner of retinal disease, in the not too distant future.

Another aspect that clearly came through the various presentations was that the researchers understood that people with a retinal disease:

- need and want precise diagnosis
- may want to know their visual prognosis
- will also want to know the effect on life determined by visual deficit, and ability to adapt, eg driving, work, pleasure and possible counselling
- are interested in the likelihood of recurrence in the family
- would like to know what research exists and if they have the ability to be treated.

In light of these factors it was stressed that everyone with a retinal disease should undergo annual checks because sometimes eye conditions change. They may also be affected by life situations such as stress, bereavement, loss of job, etc. At these annual checks ophthalmologists/optometrists can update their patients about any of the latest developments which may be relevant to them. In order to take advantage of these possible treatments or cures, it was also recommended that everyone with a retinal disease ensure that they have their details registered on a retinal disease registry in their home country as they may receive information directly from the administrators of the registry.

Note: Our Australian Registry is the Inherited Retinal Disease Register & DNA Bank in Western Australia in which many of our members have already registered. If you require information about this register, please contact the office.

Retina International General Assembly

This business meeting, held on Thursday 12 July, was attended by delegates from 32 countries. Apart from discussions about Governance, Budget, Work Plans and Business Rules, a new Retina International Mission Statement was agreed upon:

Retina International's Mission is to do all in its power to facilitate the earliest possible development of treatments and cures for Retinal Degenerative conditions, such as Retinitis Pigmentosa, Macular Degeneration, Usher Syndrome and allied retinal dystrophies and to ensure that all patients have access to accurate diagnosis, genotyping and registered treatments in the shortest time possible.

The major decision taken at this General Assembly was to establish a Retina International Secretariat to support the work of the President and Management Committee. All members voted in favour of this proposal and fundraising will commence in September this year with a view to employing a CEO in April 2013. The office will be established and hosted by Retina Ireland, who carried out the feasibility study and who will endeavour to source funds for the Secretariat as it was unanimously agreed that there will be no increase in fees for any member organisation in order to fund the Secretariat. If insufficient funds are raised by April, the establishment of the Secretariat will be delayed until such time as there is money to sustain the employment of a CEO.

The decision to establish a Retina International Secretariat was seen as a turning point for this organisation, which for the past thirty years has relied on volunteers to carry out all of the business and on member country contributions to support administrative costs. Apart from having full time paid staff to implement the work of the President and Management Committee, it is believed having our own Secretariat will facilitate international collaboration, be an access point for patient retinal organisations to converge, and give Retina International a much greater ability to lobby throughout the world.

The General Assembly was also an Annual General Meeting and therefore an election was held for positions within the Retina International Management Committee. Results of the election were as follows:

President - Christina Fasser (Switzerland)

Members of the Management Committee – Fraser Alexander (New Zealand), Claudette Medefindt (South Africa), Caiza Ramshage (Sweden), Maria-Antonietta Leopoldi (Brazil), Lorna Rosenstein (Canada) and K.P. Tsang (Hong Kong).

Those interested in finding out more about the work of Retina International and its member organisations should visit the website, www.retina-international.org

Retina International Continuous Education Program

This event is scheduled prior to the Congress so that members can share information about how their respective Retina organisations carry out their work. It also includes presentations of mutual interest. This year the program had a major focus on varying aspects of Clinical Trials. These included:

- presentations by two participants: one who was involved in the Tübingen

University retinal implant study and the other with an implant of ARGUS II from the company Second Sight

- points to consider before entering a clinical trial
- ethical issues involved in clinical trials
- an overview of current and planned clinical trials

Other sessions encompassed Research Support; Life with a Retinal Disease; Marketing and Fundraising, and Advocacy. It was very interesting to hear about the many facets associated with clinical trials, and to learn how other Retina organisations are addressing the challenges we all face in trying to support members, in spreading information about retinal diseases, and in trying to raise money for continued research.

RESEARCH UPDATE

**News Release, Weill Cornell Medical College
New York, August 13 2012**

AN ARTIFICIAL RETINA WITH CAPACITY TO RESTORE NORMAL VISION

For the First Time, Researchers Decipher the Retina's Neural Code for Brain Communication to Create Novel, More Effective Prosthetic Retinal Devices for Blindness

Two researchers at Weill Cornell Medical College have deciphered the neural code of a mouse's retina and coupled this information to a novel prosthetic device to restore sight to blind mice. The researchers say they have also cracked the code for a monkey retina — which is essentially identical to that of a human — and hope to quickly design and test a device that blind humans can use.

The breakthrough, reported in the Proceedings of the National Academy of Sciences (PNAS), signals a remarkable advance in longstanding efforts to restore vision. Current prosthetics provide blind users with spots and edges of light to help them navigate. The Weill device provides the code to restore normal vision. The code is so accurate facial features can be discerned and animals can track moving images.

The lead researcher, [Dr. Sheila Nirenberg](#), a computational neuroscientist at Weill Cornell, envisages a day when the blind can choose to wear a visor, similar to that used on the television show Star Trek. The visor's camera will take in light and use a computer chip to turn it into a code that the brain can translate into an image.

"It's an exciting time. We can make blind mouse retinas see, and we're moving as fast as we can to do the same in humans," says Dr. Nirenberg, a professor in the Department of Physiology and Biophysics and also in the Institute for Computational Biomedicine at Weill Cornell. The study's co-author is Dr. Chethan Pandarinath, who was a graduate student with Dr. Nirenberg and is currently a post-doctoral researcher at Stanford University.

This new approach provides hope for the 25 million people worldwide who suffer from blindness due to diseases of the retina. Because drug therapies help only a small fraction of this population, prosthetic devices are their best option for future sight. "This is the first prosthetic that has the potential to provide normal or near-normal vision because it incorporates the code," Dr. Nirenberg explains.

Discovering the Code

Normal vision occurs when light falls on photoreceptors in the surface of the retina. The retinal circuitry then processes the signals from the photoreceptors and converts them into a code of neural impulses. These impulses are then sent up to the brain by the retina's output cells, called ganglion cells. The brain understands this code of neural pulses and can translate it into meaningful images.

Blindness is often caused by diseases of the retina that kill the photoreceptors and destroy the associated circuitry, but typically, in these diseases, the retina's output cells are spared.

Current prosthetics generally work by driving these surviving cells. Electrodes are implanted into a blind patient's eye, and they stimulate the ganglion cells with current. But this only produces rough visual fields.

Many groups are working to improve performance by placing more stimulators into the patient's eye. The hope is that with more stimulators, more ganglion cells in the damaged tissue will be activated, and image quality will improve.

Other research teams are testing use of light-sensitive proteins as an alternate way to stimulate the cells. These proteins are introduced into the retina by gene therapy. Once in the eye, they can target many ganglion cells at once.

But Dr. Nirenberg points out that there's another critical factor. "Not only is it necessary to stimulate large numbers of cells, but they also have to be stimulated with the right code — the code the retina normally uses to communicate with the brain."

This is what the authors discovered — and what they incorporated into a novel prosthetic system.

Dr. Nirenberg reasoned that any pattern of light falling on to the retina had to be converted into a general code — a set of equations — that turns light patterns into patterns of electrical pulses. Researchers have been trying to find the code that does this for simple stimuli. "We knew it had to be generalisable, so that it could work for anything — faces, landscapes, anything that a person sees," Dr. Nirenberg says.

Vision = Chip Plus Gene Therapy

In a 'eureka moment', while working on the code for a different reason, Dr. Nirenberg realised that what she was doing could be directly applied to a prosthetic. She and her student, Dr. Pandarinath, immediately went to work on it. They implemented the mathematical equations on a "chip" and combined it with a mini-projector. The chip, which she calls the "encoder", converts images that come into the eye into streams of electrical impulses, and the mini-projector then converts the electrical impulses into light impulses. These light pulses then drive the light-sensitive proteins, which have been put in the ganglion cells, to send the code on up to the brain.

The entire approach was tested on the mouse. The researchers built two prosthetic systems — one with the code and one without. "Incorporating the code had a dramatic impact," Dr. Nirenberg says. "It jumped the system's performance up to near-normal levels — that is, there was enough information in the system's output to reconstruct images of faces, animals — basically anything we attempted."

In a rigorous series of experiments, the researchers found that the patterns produced by the blind retinas in mice closely matched those produced by normal mouse retinas.

"The reason this system works is two-fold," Dr. Nirenberg says. "The encoder — the set of equations — is able to mimic retinal transformations for a broad range of stimuli, including natural scenes, and thus produce normal patterns of electrical pulses, and the stimulator (the light sensitive protein) is able to send those pulses on up to the brain."

"What these findings show is that the critical ingredients for building a highly effective retinal prosthetic — the retina's code and a high resolution stimulating method — are now, to a large extent, in place."

Dr. Nirenberg says her retinal prosthetic will need to undergo human clinical trials, especially to test safety of the gene therapy component, which delivers the light-sensitive protein. But she anticipates it will be safe, since similar gene therapy vectors have been successfully tested for other retinal diseases.

"This has all been thrilling," Dr. Nirenberg says. "I can't wait to get started on bringing this approach to patients."

The study was funded by grants from the National Institutes of Health and Cornell University's Institute for Computational Biomedicine.

Both Drs. Nirenberg and Pandarinath have a patent application for the prosthetic system filed through Cornell University.

Look up <http://physiology.med.cornell.edu/faculty/nirenberg/lab/> to hear and watch a video from Dr Nirenberg. On the Nirenberg Lab site, third point down "A Prosthetic Eye to Treat Blindness". Don't miss it, it's excellent.

FIRST PATIENT IMPLANT OF A BIONIC EYE IN AUSTRALIA

Bionic Vision Australia (BVA), the Australian bionic eye consortium, has announced that after many years of preparation researchers there have successfully performed the first patient implant of an early bionic eye prototype, a major milestone for the project. This early prototype consists of a retinal implant with 24 electrodes. A small lead wire extends from the back of the eye to a connector behind the ear. An external system is connected to this unit in the laboratory, allowing researchers to stimulate the implant and elicit some vision. The prototype does not incorporate an external camera - yet. This is planned for the next stage of development and testing. While this is ahead of planned tests in 2013 with the full bionic eye system, such understanding early on in the development allows BVA to maximise the technology as it develops over the next 18 months.

Oxford BioMedica Announces Positive Review of Ongoing RetinoStat® and StarGen™ Clinical Studies

Oxford BioMedica, a gene-based biopharmaceutical company, and its partner, Sanofi, have announced a positive interim review of the RetinoStat® Phase I study in neovascular "wet" age-related macular degeneration (AMD) and the StarGen™ Phase I/IIa study in Stargardt disease by the Data Safety Monitoring Board (DSMB), an independent panel of specialists in the fields of ophthalmology, virology and vectorology. RetinoStat® and StarGen™ were designed and developed by Oxford BioMedica using the company's proprietary LentiVector® gene delivery technology.

Of the RetinoStat® Phase I study, the DSMB has reported successful retinal transduction, as shown by substantial increase in expression and secretion of endostatin and angiostatin proteins measured in the anterior chamber of the eye following a single administration of RetinoStat®. With regard to the ongoing StarGen™ Phase I/IIa study, the DSMB has reported that the long-term safety profile is now up to 12 months post-treatment (dose level 1).

John Dawson, Chief Executive Officer of Oxford BioMedica, said: "The continued progress of our ophthalmology portfolio, supported by another positive DSMB review, is encouraging – particularly given that early RetinoStat® data demonstrate sustained therapeutic protein expression in the eye following a single administration. The favourable safety profile of our novel ocular gene therapies further supports the wider LentiVector® platform safety package with over 33 patients treated to date across the ocular and Parkinson's disease programs."

The RetinoStat® open label, dose escalation Phase I study will enrol 18 patients with "wet" AMD and will evaluate three dose levels to assess safety and aspects of ocular physiology. The StarGen™ open label, dose escalation Phase I/IIa study will enrol up

to 28 patients and will evaluate three dose levels for safety, tolerability and aspects of biological activity.

Researchers Elucidate Cause of Death of Photoreceptor Cells in RP

Research conducted at the Angiogenesis Laboratory at Massachusetts Eye and Ear Infirmary has for the first time identified the mode of death of cone photoreceptor cells in an animal model of retinitis pigmentosa (RP). This groundbreaking study, led by Demetrios G. Vavvas, M.D., Ph.D., and including Joan W. Miller, M.D., Mass. Eye and Ear/Mass General Hospital Chief of Ophthalmology and Chair of Ophthalmology at Harvard Medical School, has further identified the receptor interacting protein (RIP) kinase pathway as a potential target for developing treatment for vision loss in patients with RP.

Previous studies have identified mutations in more than 50 genes that cause RP, but the mechanisms by which rods and cones die remain to be completely defined. Since many of the genes associated with RP produce proteins that are used specifically in rod cells, it is still a puzzle why and how cones, which in some cases do not use the mutant proteins, die after rods degenerate. Using an animal model of RP, the investigators studied whether RIP kinase mediated necrosis is involved in the death of photoreceptor cells, finding for the first time that it is involved in cone degeneration and that a deficiency of RIP kinase reduced cone loss. Moreover, the study found that treatment with a drug that inhibits RIP kinase significantly delayed cone cell death and preserved cone photoreceptors.

"Though the precise mechanisms involved in RIP kinase inducing necrosis remain unknown, our finding that necrosis results in cone cell death puts us one step closer to understanding this disease and, more importantly, moves us one step closer to being able to provide novel therapies to millions of patients with vision loss," said Dr. Vavvas.

Source: Massachusetts Eye and Ear Infirmary, Boston, 20 Aug 2012.

Is Turmeric the Spice of Life?

Dr. Ayyagari, Ph.D., associate professor of ophthalmology at the University of California San Diego, and her fellow researchers, fed curcumin to rats genetically engineered to have the P23H mutation in rhodopsin. Previous studies showed that these rats have eye problems reminiscent of RP. Specifically, the eyes of these rats have poor electrical responses to light and over time, the retina thins with the loss of photoreceptor cells. Feeding curcumin to the P23H rats alleviated these problems. Curcumin preserved the number of rods and cones in the retina and it increased the light-induced electrical response recorded from the rats' eyes. These results suggest that curcumin may support the alleviation of eye problems by preventing the loss of photoreceptors caused by the P23H mutation.

Source: Vasireddy et al. "Rescue of Photoreceptor Degeneration by Curcumin in Transgenic Rats with P23H Rhodopsin Mutation." PLoS ONE, June 2012, vol. 6(6), pp. e21193.

Health Services Utilisation and Cost of Retinitis Pigmentosa

Objective

To estimate annual per-patient health services utilisation and costs of retinitis pigmentosa (RP) in the United States. To our knowledge, this study is the first to estimate the health services utilisation and costs associated with RP, a rare disease with a long duration of visual disability relative to other common retinal disorders.

Methods

A retrospective claims analysis of patients with RP and a 1:1 exactly matched cohort of non-RP patients was conducted using the MarketScan Commercial and Medicare Supplemental Databases. Individuals were continuously enrolled in a commercial health plan or employer-sponsored health insurance for at least one year.

The annual outcomes were analysed using nonlinear multivariate models: inpatient hospital admissions, inpatient hospital days, emergency department visits, outpatient physician visits, and prescription drug refills and inpatient and outpatient medical, pharmacy and total health care costs.

Results

Patients with RP had 0.04 more hospital admissions, 0.19 more inpatient hospital days, 0.05 more emergency department visits, 2.74 more outpatient visits, and 2.18 additional prescription drug fills annually compared with their non-RP counterparts. Health care expenditures were significantly higher for patients with RP, who cost \$894, \$4855, and \$452 more for inpatient, outpatient, and pharmacy services respectively. Overall health care costs were \$7317 more per patient per year in the RP cohort. In the present analysis, the marginal effect of RP on total health care costs varied by age, with children younger than 12 years and adults aged 55 to 64 years having the highest incremental costs.

Conclusions

Patients with RP consume substantially greater amounts of health services with significantly higher health care costs.

Clinical Relevance

Treatments that slow, halt, or possibly restore RP-related vision loss may prove cost-effective for payers and society.

Source: Archives of Ophthalmology, 2012;130(5):629-634. Kevin D. Frick, PhD; M. Christopher Roebuck, MBA; Joshua I. Feldstein, BA; Catherine A. McCarty, PhD; Lori L. Grover, OD.

From the Foundation Fighting Blindness: Emerging Treatments for Retinal Diseases

In every cell in our body, DNA is sending messages, known as messenger RNA (mRNA), which tell our cells what proteins to make. Proteins are essential building blocks that provide our cells with structure and strength, regulate and protect our bodies' chemistry, and facilitate the transport of oxygen and other essential substances. Proteins are also essential to the health of our photoreceptors — the light-sensing cells in the retina — and the biochemical process that makes vision possible. It is therefore critical that the messages transmitted by our DNA are translated correctly, so that the right proteins are made.

In about 12 per cent of all degenerative retinal diseases, the translation of mRNA into necessary proteins stops prematurely, leading to the production of non-functional proteins and resulting in vision loss. In simple terms, it's as if someone stops reading a sentence halfway through, and the resulting message doesn't make sense. These traditional errors are due to what is known as premature termination codons or PTCs.

In a Foundation-funded study at the Johannes Gutenberg University in Mainz, Germany, Dr. Wolfrum and his team are evaluating a drug that can "read through" PTCs in retinal cell cultures and mouse models of Usher Syndrome type 1C (USH 1C). The drug enables the cell to read the complete message and make the right protein. Known as PC124, the drug has already been used in clinical trials for Duchenne muscular dystrophy and cystic fibrosis, both of which are devastating conditions caused by PTCs.

In a research paper published in the journal *Human Gene Therapy* in May 2011, Dr. Wolfrum reported that PTC124 was effective in initial USH 1C studies. He notes that in moving the treatment forward, his team needs to identify the optimal method for delivering the drug to the retinas in humans. The FFB-funded studies will evaluate a variety of administration options, including oral delivery, eye drops and sub retinal injections. As with any new drug, safety studies are essential, as well; it is important to verify that the drug doesn't cause problems in other parts of the body.

"We are excited about this potentially new method of treating genetic variants because it holds potential for treating a wide range of retinal conditions, including forms of retinitis pigmentosa and Usher Syndrome," says Stephen Rose PhD, chief research officer, Foundation Fighting Blindness. The fact that it has shown some success in clinical trials for other diseases is also a big plus. That gives us more confidence in the drug as we move it forward."

Dr. Wolfrum's team recently received a three-year grant from the Foundation to conduct safety, effectiveness and delivery studies for PTC124 and improved versions of the drug.

SHOPFRONT

Footwear for the Blind: Bluetooth Shoes

More than 285 million people across the globe suffer from visual impairment. Yet the tools to assist the blind in walking have changed little since the 1920s, when their canes started being painted white to make other pedestrians more aware of their presence. The gizmos that do exist have tended to be expensive and clunky, and have not caught on. This may change if Anirudh Sharma, a 24-year-old computer engineer from Hyderabad in India has his way.

His innovation, dubbed "Le Chal" ("take me along" in Hindi), pairs a smartphone app with a small actuator sewn inside the sole of one shoe via Bluetooth, the wireless technology used in exchanging data over short distances from fixed and mobile devices and creating personal area networks. The user tells the 'phone his desired destination, which is translated into electronic commands using voice-recognition software. The app, which can be programmed to run in the background, fetches the local map of the area. The phone's Global Positioning System (GPS) tracks the person's location in real-time, telling the actuator to vibrate when it is time to turn. The side of the shoe where the vibration is felt indicates which way to go.

Mr Sharma opted for a vibrating signal because for the blind, who rely on their sense of hearing to make sense of the environment, audio feedback is a distraction.

The system does not require constant internet access. Once downloaded, maps can be stored locally and combined with GPS data. The app uses Open Street Maps (OSM), an open-source rival to Google Maps. OSM allows editing, a helpful feature in updating rapidly changing urban landscapes. A speed-dial function can rapidly retrieve the most frequently visited routes.

The shoe pod is also equipped with an obstacle-detection mechanism. A sensor in the tip of the shoe, devised by Mr Sharma's business partner, Krispian Lawrence, scans the vicinity using sonar, which emits ultrasounds that bounce off obstacles, indicating their presence. The shoe sets off a distinct pattern of vibrations to alert the person of any obstruction and guides him around it.

For now, the footwear, being tested at the L.V. Prasad Eye Institute, one of India's biggest eye-health facilities, may be most useful in areas with little or no traffic, such as quiet residential streets or parks. The challenge, Mr Lawrence says, is to get the algorithm to tell an uncovered manhole from a flight of stairs, but he expects it to be able to do so in due course. Dealing with moving obstacles like cars may take longer, although the pair are working on ways to alert wearers not just about the presence of cars but also their speed.

To ensure that the final product resembles a regular shoe, fashion technologists are being consulted to help with ergonomics and design. Mr Sharma and Mr Lawrence say

their high-tech shoes should not cost more than an ordinary, stylish pair.

Source: A.A.K., The Economist, 14 July 2012, Mumbai

"To be blind is not miserable; not to be able to bear blindness, that is miserable"

John Milton 1608-1674

As Milton pondered over 400 years ago, the critical issue in learning to live without useful vision is how you personally adapt and learn to "bear" it.

Low vision aids are critical to learning to live successfully with vision loss, yet so many people are unaware of them.

Contact Quantum Technology **(02) 9479 3100**

E: info@quantumrlv.com.au

to get help on what is available for your needs.

LAST WORD

Twenty years from now you will be more disappointed by the things that you didn't do than by the ones you did do. So throw off the bowlines. Sail away from the safe harbor. Catch the trade winds in your sails. Explore. Dream. Discover.

MARK TWAIN, 1835-1910

ABOUT US

Retina Australia (ACT) Inc. is a member of the national body, Retina Australia (RA). Other members of RA are Retina Australia (NSW), Retina Australia (VIC) – which incorporates activities in Tasmania – Retina Australia (QLD), Retina Australia (SA) and Retina Australia (WA). There is also a newly-formed group in the Northern Territory.

Our role, and that of our fellow organisations, is to provide information and support to people and families affected by Retinitis Pigmentosa and other retinal dystrophies. We also raise funds for scientific research into the causes and prevention of these dystrophies.

Retina Australia is a member of Retina International, which has members and affiliates in more than 50 countries. It is estimated that more than 20 million people worldwide are affected by some form of retinal dystrophy.



FIGHTING BLINDNESS

Mission Statement: To provide information and support to people and families affected by inherited retinal dystrophies, and, with the support of the Australian community, to raise funds to finance scientific research into the causes, prevention and cure of retinitis pigmentosa.

Ways You Can Help Retina Australia (ACT)

There are many ways you can help and support us. Listed below are some suggestions.

1. Suggest to family and friends that they hold a special event (such as a sponsored walk or fundraising dinner) to support our work.
2. Include a reference to us when updating your Will.
3. Continue with your much-appreciated cash donations.
4. Tell your local eye professionals (and your family and friends) if you have found the information provided by Retina Australia (ACT) and its support services useful.
5. Offer your services to a member of the executive as there are numerous small jobs that need to be done. Many of these do not require you to be on a committee.
6. If approached, consider allowing your medical information and DNA to be stored on the inherited retinal diseases database so it is readily available to eye researchers both here and overseas.
7. When you hear that someone in your community has been diagnosed with sight loss tell them about Retina Australia (ACT) and the support services offered.

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