



# I-C

## Newsletter for Retina Australia (ACT) Inc

EDITION 2-2011

June 2011

### ***Message from the President***

It has been exciting to note in the past few months the significant amount of reporting of retinal research, not only in the media but also online. A selection of these reports is contained in this issue of I-C.

Also contained within is news of the constant efforts by your executive to raise funds for research into retinal dystrophies, and information about new products with the potential to make life just that little bit easier for those whom we serve by the work we do.

With that last thought in mind, could I please urge all members to help us with an annual donation, however small, and so support a dedicated group of people who not only work tirelessly on your behalf in an entirely honorary or *pro bono* capacity but also reach frequently into their own pockets in order to do so?

Thank you very much.

**ROBIN POKE**

### ***MEMBERS' "MONTHLY LUNCH DATE"***

Where: West Belconnen Leagues Club

Hardwick Crescent  
Holt ACT 2615  
0419 201 815

Date: 1<sup>st</sup> Monday of the Month

Location: West Belconnen Leagues Club, part of the Canberra Raiders Sports Club Group is located in the heart of West Belconnen, close to bus stops and Kippax Shopping Centre, and has ample on-site parking.

Lunch time specials are very reasonable priced and the quality of the food is high.

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**

## GENERAL NEWS

### THE IRDR AND DNA BANK – THE GOOD NEWS...

Courtesy of our members and supporters, Retina Australia (ACT) recently contributed \$2373.13 to the Australian Inherited Retinal DiseaseS Register and DNA Bank. This money has been raised through donations by our members and friends, as well as through fundraising events such as the Thoroughbred Park function in December. Retina Australia (ACT), together with our partner interstate Retina organisations, undertook to help fund the IRDR & DNA Bank project for three years, commencing 1st April 2009.

### ...AND THE NOT SO GOOD NEWS

With the assistance of Retina Australia and Retina Australia (ACT), Dr John De Roach from the Sir Charles Gairdner Hospital in Perth, which administers the IRDR & DNA Bank, recently prepared a submission to the National Health and Medical Research Council for funding amounting to almost \$1million to ensure that the second three-year phase, from 2012 to 2015, would see not only the continued collection of DNA material but most importantly the sequencing of the DNA collected during the first three year phase. Unfortunately Dr De Roach has learned in the last few days that the submission has been unsuccessful. It was emphasised by the NH&MRC [that this had nothing to do with the quality of the submission: rather, other areas of medical research had been perceived as in greater need of the funds. Dr De Roach and his staff, with the support of Retina Australia, are working on alternative proposals and strategies, to ensure that this important resource continues to develop.](#)

### MYER GENEROSITY

We have been heartened, however, by the news that the Myer Group in the ACT will assist our fundraising efforts by contributing monies from its Community Fund. This results from a suggestion by Sue Morcombe, an executive at Myer Tuggeranong, who very generously supplied a variety of Myer products for our use when she attended our Thoroughbred Park fundraiser in December 2010.

### DIARY DATE

Speaking of which, make a note in your diaries: our major fundraiser in 2011, another fun-filled afternoon at Thoroughbred Park, is booked for Sunday 27 November. We hope to see as many members there as possible.

### CONSORTIUM BEING CONSIDERED

In a bid to explore *all* fundraising options Retina Australia (ACT) recently approached four other blind and vision impaired advocacy groups in Canberra with a view to establishing a fundraising consortium. Aware as we are that a number of large corporations here regularly support large charity organisations, we suggested to Canberra Blind Society, Blind Citizens Australia, Radio 1 RPH and VisACT that a joint approach might reap dividends. A meeting was held on 20 June, and each organisation has agreed to consider our proposal.

### TAX REBATES FOR ADAPTIVE TECHNOLOGY

As we approach the end of the financial year the Australian Tax Office has advised that the cost of some equipment designed for people who are vision impaired is now eligible for the medical expenses rebate. The rebate means that 20 per cent of all eligible expenses above \$1,250 can be claimed. Approved products include a talking computer, reading system and CCTVs, provided they have been prescribed by a medical doctor, optician or optometrist.

Consult your tax pack for details.

### **WEBSITE GAINS PRAISE**

The Retina Australia (ACT) website, created by Chris Martin and now fully functional, has gained high praise from our state counterparts. The URL is:

<http://www.raact.org.au/wordpress>

Take a look - and please let us have your ideas for additional information.

### **'ON YER BIKE' WITH VISACT**

For the past seven years VISACT has been the major promoter of blind and low vision sports in the ACT. VISACT works closely with the Canberra Blind Society in supplementing the recreation services offered by the Society. An example of this is VISACT and Females in Training, or FIT, joining forces to continue the very popular tandem cycling program. There are weekly rides organised, from short rides to long-distance road trips, with a large number of pilots and a range of bikes available, including two new bikes purchased last year with the assistance of sponsors ACT Health and the Lords Taverners. For more information contact Lauren Brand 0403 602 577, Lindy Hou 6156 2964 or Rosemary Robinson 0416164922.

VISACT also has an accessible website with more information at [www.visact.org.au](http://www.visact.org.au). To keep up to date with what's happening you can become a member of VISACT sport and receive regular updates on that sport as well as what's happening in the world of blind sports.

### **TECHNOLOGY TOOLS AND TIPS FOR THE VISION IMPAIRED**

Modern technology can make life so much easier for everyone, including those with vision impairment, but with the trend towards making everything smaller, sometimes it can be difficult to utilise all the new tools and functions available. Here we look at just a few ways to make things easier for those with poorer eyesight.

#### **Large Button Mobile Phone**

The Seniorphone has been designed specifically to be easy to use with its big buttons and easy to use menu style. The phone features:

- Large buttons
- Easy to use menu
- Loud ring tones
- Programmable SOS button
- Easy calling with speed dials
- Completely unlocked—will work with any Sim Card
- No contracts or monitoring fees

For more details go to [www.seniorphone.com.au](http://www.seniorphone.com.au) or phone 1300 821 660.

#### **Microsoft Software Help**

With every new version of Windows or the Office suite, Microsoft enhance the features for those who find it harder to see to navigate around the screen. When using Office, type "Accessibility Features" into the Help section and it will bring up the helpful features for your version of Office. With Windows you can use the *Accessibility Wizard* (XP or earlier) or *Ease of Access Centre* (Vista & 7). These offer a number of features that allow you to change some of the settings on your computer to make them easier to see.

You can change settings to:

- Increase the font size for Windows title bars, menus and other features.
- Increase the size of items on screen, including the text inside windows.
- Use Microsoft Magnifier, which opens a floating window that displays an enlarged view of part of the screen.
- Select the size of scroll bar and window borders.
- Choose the size of icons on your desktop.
- Choose high contrast display colours that make text easier to read.
- Choose the size and colour of your mouse cursor .
- Change the blink rate and width of the cursor.

Additionally, many other programs and hardware are compatible with Windows and available to help individuals who are blind. These include screen readers, Braille output devices, and other useful products. For more information, go to

<http://www.microsoft.com/enable/>

## **YOUTH AMBASSADORS**

*Recently Karen Junge (RA Qld), Melanie Chatfield (RA WA) and Liz Wheeler (RA NSW), were asked to be Retina International Youth Ambassadors in Australia. Ambassadors need to be suffering an inherited retinal disease and be willing to put some time and effort into discussing and raising awareness of youth issues relating to our conditions. This is their story, related by Liz...*

Our first task arrived in March when we were asked to contribute some suggestions for the Retina Youth Program, a part of the 2012 Retina Youth Congress. It was very exciting to receive emails from all across the world from others suggesting topics of interest. For Australia we put forward the following suggestions.

### **Social and confidence**

- Understanding body posture/body language – how to hold yourself with confidence and interact with other people.
- Learning how to ask for help – building confidence, strong networks and friends.
- Options for being physically active/interesting – recreational activities you can do with a vision impairment, examples being Brazilian jui jitsu, tango dancing, skiing.

### **Family Planning, including:**

- Understanding genetics: what is known and available to determine feasibility of passing on the disease?
- Other options for those wishing to become parents, and overcoming obstacles with a disease.
- Shared stories and experience. Current parents can share benefits, fears, difficulties.

### **Financial Planning**

- What to consider when faced with a degenerative illness that may limit your career span or earning capacity in the future.

Both Melanie and I are hoping to get across to the 2012 Retina International Congress so we can network, learn as much as we can and then share all this at the Retina Australia Conference in Sydney in October 2012.

We have also set up a group called Retina Australia – Youth on Facebook. It is a closed group for people between the ages of 18 and 35 who suffer an inherited retinal disease. The group will enable us to share information on youth issues fed through the international group and build a support network. If you meet the criteria we encourage you to join.

Cheers,  
Liz



**LIZ**



**KAREN**



**MELANIE**

## **PRODUCT INFORMATION**

### **RETINAL PROSTHESIS APPROVED**

After more than 20 years of research and development involving a team of international specialists, Second Sight Medical Products, Inc., has announced that its Argus II Retinal Prosthesis System ("Argus II") is now approved for sale in the European Economic Area (EEA). After a successful clinical trial involving more than 30 blind patients around the world, and a very thorough review of the product's safety and performance by an independent expert body, this device becomes the first approved treatment available for sightless people.

"After years of research, we are very happy to be able to offer a viable long-term solution for people suffering from advanced retinal degenerative diseases such as retinitis pigmentosa (RP)," said Robert Greenberg, MD, President and CEO of Second Sight. "The CE Mark approval, which comes after intense regulatory review of our trial and our device, represents a huge step forward for the field and for these patients who have, until now, had no proven treatment alternatives."

Argus II is Second Sight's second generation implantable device intended to treat profoundly blind people suffering from degenerative diseases such as RP. The system works by converting video images captured from a miniature camera, housed in the

patient's glasses, into a series of small electrical pulses that are transmitted wirelessly to an array of electrodes on the retina. These pulses then stimulate the retina's remaining cells resulting in the corresponding perception of patterns of light in the brain. Patients learn to interpret these visual patterns, thereby gaining some functional vision. Thirty patients participated in the clinical trial, using the device at home and in their daily lives since the trial started.

Although the resulting vision is far from normal, investigators in the clinical trial of the Argus II are excited by the results. "After more than three years of clinical trials, we were happy to demonstrate the performance, safety and long-term reliability of Argus II," explained Professor José-Alain Sahel, Chairman, Department of Ophthalmology: Quinze-Vingts National Ophthalmology Hospital, Paris, France.

Adds Dr. Lyndon da Cruz, MD PhD Consultant Retinal Surgeon at Moorfields Eye Hospital in London, UK: "The fact that nearly all patients had a stable, safe and functioning system and that a majority of patients could recognise large letters, locate the position of objects and the best could read short words impressed us beyond our most optimistic expectations."

"This 'artificial retina' brings hope to thousands of people with advanced retinal diseases" added David Head, Chief Executive of the British Retinitis Pigmentosa Society. "The restoration of an element of vision may bring with it the restoration of independence and mobility that would greatly improve a patient's quality of life."

*Source: Adapted from Second Sight Media Release, Lausanne, 2 March 2011.*

### **ISRAELI COMPANY ADVANCES RETINAL IMPLANTS**

An Israeli company has announced plans for a retinal implant device that uses nano technology and may help millions of blind people. The start up company founded in 2009 aims to make artificial vision a reality by building a simpler, smaller and less invasive device. The heart of the system is a wi-fi powered chip the size of a child's fingernail that is attached over the damaged retina within the eyeball. The company claim in their press release that the 30 minute operation would be a relatively simple surgical procedure in which a five millimetre incision is made in the sclera and the device slid inside and glued to the retina.

This device, which the firm calls Bio-Retina, is powered by a small battery mounted in a normal looking pair of eyeglasses that broadcasts an under one-milliwatt WiFi signal. The electronic radiation level is equivalent to sunlight and is said to pose no risk to the user. Bio-Retina then uses the power to operate like a digital camera's image recording chip, light naturally entering through the pupil falls on the chip which then sends a 24x24 black and white pixel image via a series of nano-scale electrodes. Later versions are aiming for a 72x72 resolution, according to the firm, essentially giving the blind and near blind back their vision. Gefen, the managing director, said the uniqueness of their idea is the number of pixels crowded on the sensor, and the way it is glued in place. The firm has demonstrated proof of concept, and they are working on a prototype to show the image end-to-end, and hope to start clinical trials on humans by 2013. They are hoping the blind will be able to identify people in front of them, and distinguish images instead of dark and light.



## RESEARCH NEWS

*The Research Update in this edition focuses in part on developments in orphan drug designations. But what exactly are orphan drugs?*

An orphan drug is a pharmaceutical agent that has been developed specifically to treat a rare medical condition, the condition itself being referred to as an orphan disease.

The assignment of orphan status to a disease and to any drugs developed to treat it is a matter of public policy in many countries, and has resulted in medical breakthroughs that may not have otherwise been achieved due to the economics of drug research and development.

The Orphan Drug Act (ODA) of January 1983, passed in the US, with lobbying from the National Organisation for Rare Disorders, is meant to encourage pharmaceutical companies to develop drugs for diseases that have a small market. Under the law, companies that develop such a drug may sell it without competition for seven years, and may get clinical trial tax incentives. The European Union (EU) has enacted similar legislation, through which pharmaceuticals developed to treat rare diseases are referred to as "orphan medicinal products."

Orphan drug legislation also exists in Australia. The Australian orphan drugs program was set up in 1997. The program aims to ensure the availability of a greater range of treatments for rare diseases and allows the Therapeutic Goods Administration (TGA) to use information from the US Food and Drug Administration (FDA) Orphan Drugs Program as part of the Australian evaluation process.

So what is an Orphan Drug Designation and why is it so important to all in the rare disease community?

Pharmaceutical and biotechnology companies research and develop new medications to treat medical conditions, but people who have rare diseases have not always had as much attention from researchers. This is because numbers are small and therefore the potential market for new drugs to treat them is not economically attractive.

A rare disease is considered as one which occurs in less than 200,000 individuals in the United States, less than 5 per 10,000 individuals in the European Union, or less than 2000 individuals in Australia.

Government regulatory agencies in the United States and the European Union have taken steps to change this reality. The Food and Drug Administration (FDA), which is responsible for ensuring the safety and efficacy of medications on the market in the United States, established the Office of Orphan Product Development (OOPD) to facilitate the development of orphan drugs including offering research grants. Then in 1983 the Orphan Drug Act was passed. This for the first time offered incentives to induce companies to develop medical products for individuals with rare disorders.

In 1995 the European Medicines Agency (EMA), which is responsible for ensuring the safety and efficacy of medications on the market in the European Union (EU), established the Committee on Orphan Medicinal Products (COMP), which oversees the development of orphan

drugs in the EU. Like the US Congress, the EU government recognised the need to increase research and development of orphan drugs.

An Orphan Drug Designation is not easy to achieve so it is very exciting for patients who have supported research into rare retinal degenerative disease to see so many orphan drug designations being applied to potential treatments.

Every year brings us a step forward to a cure or treatment for retinal degenerations and we hope that the successes of the past year can be built upon and treatments will become a reality in the shortest possible time frame.

### **RETINA INTERNATIONAL CELEBRATES RARE DISEASE DAY 2011**

For over thirty years patient-led research groups have formed partnerships with leading researchers across the globe and together they have blazed a trail in the development of treatments and cures for rare retinal diseases such as Retinitis Pigmentosa, Usher Syndrome, Stargardts Disease, Macular Degeneration and Lebers Congenital Amaurosis (LCA).

Over the years these groups have raised significant funds to support world-leading research, and advocated at the highest level for national and international support of their efforts.

In this time patients and researchers have become true partners and in so doing expedited the search for cures and treatments. Patient groups are now working closely with industry to ensure that the work they have supported can be facilitated through clinical trial development and the regulatory process to ensure treatments are available to those losing their sight as soon as possible. And the results of this intense work are now coming to fruition.

With new orphan drug designations granted for treatments with the potential to cure and to treat rare forms of rare retinal degenerations over the last few months, on Rare Disease Day 2011 Retina International is delighted to highlight their progress.

### **PATIENT-LED RESEARCH**

From RP Fighting Blindness (the British Retinitis Pigmentosa Society)

Our sister charity in Dublin, Fighting Blindness Ireland, has reported success in securing Orphan Drug Designation for adeno-associated viral (AAV)-based therapy containing DNA encoding an RNAi targeting rhodopsin and a replacement rhodopsin gene for the treatment of rhodopsin-linked Retinitis Pigmentosa, currently being developed by Genable Technologies.

Genable Technologies Ltd. is a company formed by Fighting Blindness (Ireland) researchers at Trinity College Dublin (TCD) to progress the development of gene therapies for Retinitis Pigmentosa (RP) and other retinal diseases. Fighting Blindness Ireland was a founding investor in the company.

Avril Daly, CEO at FB Ireland, said "This is a very important step towards a gene therapy product for RP, which is a rare disease, and it is believed to be the first time orphan drug designation has been granted for a gene therapy in Ireland."

At least 150 different alterations in rhodopsin-linked RP have been identified worldwide, which makes developing a gene-based therapy very complex. Researchers at the Smurfit Institute



of Genetics in TCD have been working for over 20 years to identify the genes and potential treatments for RP. This rare, genetically inherited condition is caused by gene mutations which lead to the degeneration of photoreceptors at the back of the eye and eventual sight loss.

This therapy switches off both copies of the gene, the normal and the altered copies. Simultaneously, a replacement rhodopsin gene is introduced which has been subtly altered so it cannot be suppressed. It encodes normal protein, which allows the photoreceptors to work normally.

Ms Daly welcomed this development, saying: 'Research into a gene therapy for rhodopsin-linked RP was one of the first projects funded by Fighting Blindness Ireland in the mid 1980s. It is hugely exciting to learn that this project has led to a potential gene therapy that in turn has received an orphan drug designation; it is a great day for patient-led research. The relationship between researchers and patients is vital, particularly in the area of rare diseases, where some projects would not be funded without patient groups taking the initiative. Today we are witnessing the results of these relationships. We know this is only one step towards a therapy for a dominant form of RP, but it is a major step and gives real hope to those living with this degenerative disease not only in Ireland but internationally'.

There is still a journey ahead. This potential therapy must go through the human clinical trials process before it can be administered as a treatment.

Professor Jane Farrar of Trinity College Dublin and Genable Technologies Limited commented: "We are extremely pleased to receive orphan drug status in Europe. This decision by the EMA will help raise awareness of Retinitis Pigmentosa as a serious disease and ultimately help more patients receive therapy for their disease".

Ms. Daly added "This development would not have been possible without the support and perseverance of the members and friends of Fighting Blindness and the determination and skill of the researchers at TCD over the past three decades. We will follow the progress of this therapy closely and look forward to bringing more good news to the patients affected by RP in the not too distant future".

Prof. Tony Moore of the RP Fighting Blindness Medical Advisory Board said "Rhodopsin mutations are a common cause of ADRP. However we should caution that this is but a first step and that there is still a lot of work to be done before clinical trials in man will start."

### **Research Round-Up from RP Fighting Blindness (UK)**

There have recently been several news stories in the mainstream and scientific press about the progress of various threads of research. Here we briefly round them up.

#### **Gene Therapy**

Always an interesting area, as it is one of the most promising and advanced lines of research. In April Prof. Robin Ali from the Institute of Ophthalmology presented to our Retinal Awareness Group (RAG) weekend in Blackpool and brought the audience up to date with progress. Prof. Ali explained how gene replacement therapy for Lebers' Congenital Amaurosis (LCA) is now being trialled at much higher doses than previously, and in children as young as five years old. He also highlighted some of the genes that are likely to be the subject of the

next rounds of clinical trials and appeared to be optimistic about the potential benefits as the technology involved is being refined and enhanced as trials progress.

At about the same time as Prof. Ali's presentation, separately we learnt that in the USA the FDA have given approval for clinical trials of a gene therapy for Stargardts Disease, a form of RP. Similar work using a different viral vector to deliver the gene is also progressing at Oxford Eye Hospital (see our later item about patient information day in July).

Also in the USA REGENX BioSciences, a privately held biopharma company and developer of NAV, a unique gene delivery technology, has announced a \$2.8 million grant from the US National Institute of Health which will support the advancement of NAV technology to treat X-Linked RP for which there is currently no treatment. Kenneth Mills, CEO at REGENX said "We ... recognise the critical importance of accelerating early stage development into meaningful treatments ... NAV technology is attractive because of its unique ability to safely and effectively deliver genetic material to target cells in the eye".

### **Stem Cells**

Prof. Ali also discussed recent progress on the use of stem cells to generate photoreceptors suitable for transplantation into the retinae of people with RP. Though anxious to point out that clinical trials of such treatment are some way off, he was optimistic about recent progress, including news from Japan where a respected research group have succeeded in generating mouse photoreceptor cells using stem cells.

Created by coaxing mouse stem cells into a precise three-dimensional assembly, the 'retina in a dish' is by far and away the most complex biological tissue engineered yet, scientists say. "There's nothing like it," said Prof. Ali (who was not involved in the study). "When I received the manuscript, I was stunned, I really was. I never thought I'd see the day where you have recapitulation of development in a dish."

If the technique can be adapted to human cells and proved safe for transplantation it could offer a supply of tissue to replace damaged retinas. However this could still take many years, though more immediately synthetic retinal tissue could help researchers in the study of RP and in identifying therapies.

David Head, Chief Executive at RP Fighting Blindness welcomed the news, saying: "The prospect of replacing failed retinal cells is really exciting, and this is a crucial and significant step in that direction.

"The last three or four years have brought some fantastic developments in retinal research. What these research groups achieve is amazing and they deserve accolades."

### **Optogenetics**

Optogenetics can bypass damaged photoreceptors by genetically engineering other types of cells to respond to light. This is done by adding genes that code for proteins called channelrhodopsins, which normally sit in the photoreceptor cell membrane.

Several research groups have been exploring the possibilities of optogenetics. Prof. Alan Horsager's (USC Institute for Genetic Medicine) recent work has been focused on making retinal bipolar cells sensitive to light and in a new study in mice the team have induced

enough sensitivity in the retina of blind mice and restored enough vision that the mice can navigate a maze.

"It's a very targeted approach that maintains the natural processing of the retina," said Horsager. "There is a lot more to understand and preclinical studies are the next step to determining the potential therapeutic benefit for humans."

Prof. Tony Moore at UCL Institute of Ophthalmology and a member of the RP Fighting Blindness Medical Advisory board welcomed the news, though added: "We must stress that this work is in its infancy and is at the stage of work in animal models, but may be applicable to human disease in future."

## **RESULTS OF ELECTRICAL STIMULATION THERAPY STUDY FOR RP PATIENTS**

Okuvision GmbH, which specialises in the field of electrical stimulation therapy (EST) for early and intermediate stage retinitis pigmentosa patients, presented data from the company's first pilot study at the annual meeting of the ARVO (Association for Research in Vision and Ophthalmology, Ft. Lauderdale). The study included 24 patients suffering from early and intermediate stage retinitis pigmentosa who were separated into three groups and received EST at varying strengths for 30 minutes once a week for six weeks. The presentation was delivered by lead author, Dr. Florian Gekeler of the Centre for Ophthalmology, Tuebingen University Eye Hospital.

Dr. Gekeler's poster presentation titled, "Transcorneal Electrical Stimulation in Patients with Retinitis Pigmentosa," highlights findings from Okuvision's first clinical trial which began in 2007 using a thread electrode to stimulate the retina with small amounts of current. Twenty-four patients with early and intermediate stage retinitis pigmentosa were randomised and separated into three different groups. Each group received different levels of stimulation. The first was treated without electrical current, the second with 67 percent of the individual threshold, and the third with 150 percent of the patient's threshold – all for 30 minutes a week for six weeks. Final evaluation showed a +20 percent statistically significant improvement in the field of vision by patients who received the 150 percent stimulation. The findings from this study emphasise that electrical stimulation of the retina liberates growth factors which may be able to delay retinal degeneration.

"Our team began this study with the goal of determining whether electrical stimulation therapy could safely and effectively preserve vision for early stage retinitis pigmentosa patients," said Dr. Florian Gekeler, consultant at the Centre for Ophthalmology of the University of Tuebingen, Germany. "The visual results achieved surpassed our initial expectations and it is our hope that these results will be the first step in ensuring that EST is considered a viable treatment option to slow the degenerative progress for retinitis pigmentosa patients."

"The results of our study show promise that a treatment option, while not a cure for retinitis pigmentosa, could be available in the future," said Dr. Walter-G. Wrobel, chairman and founder, Okuvision GmbH. "While much is still unknown about EST technology, the results of the study are a great step toward determining efficacy of this treatment and we look forward to conducting further studies to potentially reach out to those who are hopeful in delaying the effects of retinitis pigmentosa."

## **PROMISING CONE PRESERVING TREATMENT**

A promising treatment aimed at preserving cones, the retinal cells that provide central and daytime vision, is poised to move into a Phase I clinical trial within the next year. Known as rod-derived cone viability factor (RdCVF), the therapeutic protein has consistently preserved vision in several preclinical studies.

RdCVF has received an orphan medicinal product designation from the European Commission, a regulatory agency similar to the FDA in the U.S. The orphan designation provides marketing, financial, and clinical research benefits to Fovea Pharmaceuticals, the French company developing the treatment.

In 2005, Drs. José-Alain Sahel and Thierry Léveillard received the Foundation Fighting Blindness Annual Trustee Award for their discovery of RdCVF as a potential vision-saving treatment. The Foundation-funded French research team screened 210,000 genes to find a rod-derived protein that would protect cones.

A majority of retinal degenerative diseases, including retinitis pigmentosa (RP), are caused by mutations in genes that affect rod cells. As a result, rods are the first photoreceptors to degenerate. Rods provide peripheral eyesight and vision in dark settings.

However, once rods are gone, cones subsequently degenerate. This phenomenon led researchers to suspect that rods were secreting a factor (or multiple factors) that helped to preserve cones.

Initially, the France-based clinical trial will involve monthly ocular injections of RdCVF into people with RP. The investigators are also evaluating gene therapy as a delivery mechanism to provide long-term, sustained release of RdCVF. One gene therapy treatment will likely be effective for several years.

While the researchers will initially evaluate RdCVF in people with RP, they believe the treatment may preserve vision in people with a wide range of retinal degenerative diseases.

Dr. Sahel notes that by keeping as few as 5 percent of cones alive, a person can continue to function independently.

Drs. Sahel and Léveillard are co-founders of Fovea Pharmaceuticals. Dr. Sahel is centre director for the Foundation's Paris Research Centre for the Study of Retinal Degenerative Diseases.

*Source: [www.blindness.org](http://www.blindness.org)*

## **CONSORTIUM MAKES MAJOR STRIDES IN THE DEVELOPMENT OF TREATMENTS FOR X-LINKED RP**

After three years of intensive research on multiple fronts, a consortium funded by the Foundation Fighting Blindness is reporting significant progress in the development of vision-saving treatments for X-linked retinitis pigmentosa (XLRP).

"X-linked RP is a particularly aggressive retinal degenerative disease, and at the same time, is relatively prevalent. So the need to take action is strong," says Dr. Stephen Rose, chief research officer, Foundation Fighting Blindness. "By establishing the consortium, we are able to collaborate as a team to identify treatment opportunities and then apply a full court press in developing them. The strategy has paid off well."

The XLRP consortium is focusing its efforts on multiple gene therapy approaches, as well as a protein-based treatment that inhibits retinal cell death. The team is also performing natural history studies to better understand the progression of XLRP and how well potential treatments are working. Gene therapy for XLRP is furthest along in development and could move into clinical trials within the next two to three years.

Dr. Rose says that consortium investigators have been able to leverage the research successes for other retinal conditions, including gene therapy clinical trials that are restoring vision in children and young adults with Leber congenital amaurosis (LCA).

Dr. Jean Bennett, the lead investigator for the LCA gene therapy clinical trial at The Children's Hospital of Philadelphia, is achieving excellent results in preclinical studies of XLRP gene therapy, which uses the same gene delivery technology — adeno-associated viruses (AAVs) — used in the LCA trial.

Gene therapy is an attractive approach to treating XLRP, because 70-80 percent of XLRP cases are caused by a single gene called RPGR. In other words, an RPGR gene therapy could potentially help 70-80 percent of all people with XLRP.

One of the challenges in delivering the RPGR gene to the retina is its size. As genes go, it is rather large. To overcome that limitation, Dr. William Hauswirth of the University of Florida is developing an XLRP gene therapy that delivers a region of RPGR known as ORF-15 to the retina. ORF-15 is where disease-causing variations in RPGR commonly occur. He, too, has obtained excellent results in preclinical studies.

On another research front, Drs. David Zack and Debra Thompson at the University of Michigan are conducting preclinical experiments of a protein called X-linked Inhibitor of Apoptosis Protein (XIAP) to slow or halt vision loss from XLRP. The investigators have shown that XIAP slows retinal cell death and loss of vision in preclinical testing.

Functional and structural evaluation of the retina is critical to the development of these various XLRP treatments. Dr. John Heckenlively at the University of Michigan, a leading retinal physician, is an expert in conducting both retinal examinations and tests that measure retinal activity. He will help investigators understand how well emerging treatments are working in both lab and clinical studies.

Dr. Rose notes that while the consortium is aiming at XLRP, the knowledge coming into and out of their studies is relevant to a wide range of retinal conditions. "The consortium has benefited greatly from the knowledge gained in the LCA clinical trials and other cross-cutting studies, and I know other research groups are watching the progress being made in XLRP very closely," he says.

## **VITAMIN D CONSUMPTION AND REDUCED RISK OF MD: 2011 STUDY**

Consuming vitamin D from foods or supplements might reduce the risk of developing age-

related macular degeneration (AMD) in women younger than 75, according to a study published by the Archives of Ophthalmology. A team led by researchers at the University at Buffalo, N.Y. examined data on blood levels of vitamin D among 1,313 women, 50 to 79, enrolled in the ongoing Women's Health Initiative study. In women younger than 75, vitamin D from foods and supplements (but not from time spent in direct sunlight) was linked with decreased risk of developing early age-related macular degeneration. Those who consumed the most vitamin D had a 59 percent decreased risk of developing it compared with women who consumed the least.

## **NUTRITION AND EYE HEALTH**

### **Vitamin A**

At the recent Retina International congress in Italy, it was disclosed that contrary to advice given as a result of a US study, the use of Vitamin A is in doubt for some types of retinal dystrophy. It was originally claimed, following extensive research, that Vitamin A in prescribed amounts could slow the progress of Retinitis Pigmentosa. It has now been found that in some cases of retinal dystrophy including Stargardts disease, that taking Vitamin A in the prescribed form had the opposite effect, in that it can cause more damage to the eye. It is now advised that people with retinal dystrophy should not use Vitamin A without first discussing the matter with their ophthalmologist.

### **Eating For Eye Health: The Macular Degeneration Cookbook**

#### **Chapter 9, Buttrose & Jones, 2009**

Diet can play a role in good eye health. Studies have shown that Macular Degeneration responds to antioxidants, vitamins, minerals and other nutrients.

### **Antioxidants**

Antioxidants are important to our health and are found in the foods we eat. When cells turn food and oxygen into energy they also produce 'free radicals'. The free radicals are thought to be a contributing factor in the cause of MD and other diseases. Free radicals can be neutralised by antioxidants.

Antioxidants for eye health include:

- Lutein and Zeaxanthin – These are particularly important for eye health, lutein is found in high concentrations in the peripheral retina, while zeaxanthin is found predominately in the central macular (source Wikipedia). It is found in high levels in dark green leafy vegetables such as spinach, kale and chard.
- Vitamin C – This is necessary for normal structure and function of connective tissue. Found in citrus fruits, papaya and rockmelon, and vegetables such as broccoli, brussel sprouts and capsicum.
- Vitamin E – Necessary for cell production from the damage caused by free radicals. Vitamin E is found in nuts especially almonds, whole grains and green leafy vegetables.
- Zinc – Contributes to the normal structure of the skin, normal wound healing and contributes to a healthy immune system. It is found in meat and seafood (particularly oysters), seeds, nuts and whole grains.

Other nutrients for eye health:

- Selenium – This is necessary for cell protection from some types of damage caused by free radicals. Brazil nuts are rich in selenium.



- Omega-3 – Omega-3 supports normal development of the brain, eyes and nerves. Cold water, oily fish such as salmon, trout and sardines are rich in omega-3.

The 'Eating for Eye Health' Cookbook can be ordered from the MD Foundation, Phone 1800 111 709 or on the website [www.mdfoundation.com.au](http://www.mdfoundation.com.au)

### **OMEGA-3**

Studies continue to show the benefit of incorporating fish regularly into your diet. "We found that it (Omega-3) had as big an effect as the current drugs that are used in these diseases, so this is huge in terms of increasing medical costs that are occurring in all countries," Dr Lois Smith, a professor of ophthalmology at the Harvard Medical School said. So with that in mind, here's a recipe from "Cooking for VIPs" by Maxine Turkington ([www.tips4vips.co.uk](http://www.tips4vips.co.uk))

### **PEPPERED TUNA STEAKS**

Like good steak, fresh tuna should be eaten as rare as possible – so make sure it's very fresh. Rated E – Easy and requires minimum effort in preparation.

Serves 4

#### **INGREDIENTS**

250 ml (1Cup) orange juice  
110g unsalted butter, softened  
1 tea/s lemon juice  
1 tab/s whole grain mustard  
Salt & pepper to taste  
4 fresh tuna steaks of similar thickness  
2 tab/s olive oil  
4 tea/s cracked black pepper

#### **PREPARATION**

Gently boil the orange juice in a small saucepan until reduced and thickened. Slowly whisk in the butter on medium heat: remove from the heat and add the lemon juice, mustard, salt and pepper. Brush both sides of the tuna steaks with olive oil and a sprinkle of cracked pepper. Grill on high or sear in a hot pan until medium rare, about 2-3 minutes each side. Serve immediately with the sauce (reheated if necessary). Goes well with mashed potatoes and grilled sweet peppers, or salad.

## **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.

## MEMBERSHIP DONATIONS

Dear fellow members of Retina Australia (ACT)

As you all know, once you became a member of our organisation you are a member for life and are not required to pay annual renewal fees or subscriptions. We do, however, ask that you consider making an annual tax-deductible donation.

Retina Australia (ACT) receives neither government nor any other form of funding. The organisation therefore relies on your donations so that we can maintain our services to you, our members. Among these services are the provision of telephone-based peer support, the printing and distribution of the "I-C" newsletter and information kits, the organising of social events, and the raising of funds for research into the causes of retinal diseases. A percentage of the monies raised through your donations are also channeled into Australian research. Sundry other administrative tasks, such as printing, photocopying and postage must also be paid for.

It should also be noted that executive members of Retina Australia (ACT) are all volunteers, giving their time in a bid to ensure a brighter future for family, friends and fellow citizens who are facing the onset of blindness.

Please therefore send your donation, to Doris Wallace, Treasurer, Retina Australia (ACT), 40 McKillop Circuit, Kambah, ACT 2902 as soon as possible.

Thank you for your continued membership and support.

Yours sincerely

ROBIN POKE  
President

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**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 an

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