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Newsletter for Retina Australia (ACT) Inc

EDITION 1/JUNE 2015

Message from the President

I must first of all apologise to all members for the lack of a newsletter since September last year. This is due largely to the time constraints put on executive committee members in organising not one but two fundraising functions in relatively quick succession. First came the regular Annual Spring Racing Luncheon at Thoroughbred Park on 16 November, after which we were given the opportunity to organise another fundraiser, an extremely successful concert at the Southern Cross Club. Reports on both events can be found below. In the meantime please note opposite the interim details for this year's major fundraiser. It would be good to have every member of Retina Australia (ACT) there!

Also in this edition are details of Retina Australia's triennial conference, this time being held in Melbourne in October. Members are encouraged to attend if they possibly can, to learn about all the latest research into a cure for retinal diseases.

ROBIN POKE AM
President
Retina Australia (ACT)

"ANNUAL SPRING RACING LUNCHEON"

SUNDAY 15TH NOVEMBER 2015
THOROUGHBRED PARK
CANBERRA

Invite all your friends to this fun afternoon with raffles and racing, food and fun and, importantly, fundraising that will support the work being carried out into the prevention and cure of Retinitis Pigmentosa.

We are fortunate to once again have the support of Women in Racing (WiR). Last year we had some 200 to our lunch, easily a record. Let's make a big effort to beat that number this year. With WiR's support we have been able to contribute not only to the ACT's share of funds for the Australian Inherited Retinal Diseases Register but also to funding for other research.

Retina Australia (ACT) is proudly
sponsored by

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ANNUAL SPRING RACING LUNCHEON A SUCCESS – YET AGAIN

Our 2014 Spring Racing Fundraiser, held at Thoroughbred Park on 16 November was probably our best ever. A fun-packed afternoon of racing, tipping competitions, raffles and auctions raised approximately \$6000 for research into a cure for retinal diseases.

The highlight was a hugely interesting presentation by Professor Jan Provis of the Australian National University, who, invoking her talents as an educator *par excellence*, took us through all facets of the scientific research taking place on our behalf and made what is quite an esoteric subject stunningly simple. This and other facets resulted in the event being widely applauded, witness this 'thank you' note from Lizzy Wagner:

Allow me the opportunity to thank you most sincerely for the pleasure of being able to assist and support Retina Australia with The Lizzie Wagner Group contribution. The day was most enjoyable and the event was run with style and the utmost professionalism. It was an honour to be associated and I look forward to joining you for many more celebrations and fundraisers in the future.

Special plaudits go to our Secretary, Jan James, for her tireless preparation and administration, and the ever-professional David Kilby, our regular MC.

NEW YORK-BASED 'LOCAL BOY' PLAYS BENEFIT CONCERT FOR RETINA AUSTRALIA

An Australian musician rapidly making an international reputation for himself in New York played a Retina Australia (ACT) benefit concert in Canberra on 16 March – especially for local family members.

Singer/songwriter Yanni Burton is the grandson Barbara Burton, our very popular committee member, whose other grandson Chris, Yanni's cousin, suffers retinitis pigmentosa. Barbara was delighted to inform her fellow members last year that Yanni and his band would soon be coming to Australia – and that all proceeds from the Canberra concert would go to our charity. "It was Yanni's one window of opportunity on the tour," Barbara says. "Naturally, we jumped at the prospect".

The concert was held at the 'Top of the Cross' at the Southern Cross Club, Woden, on 16 March – and received 'rave reviews'. Yanni brought with him his internationally acclaimed New York City Band, for whom their debut tour of Australia was a celebration of the Australian release of Yanni's debut EP, *Same Old Love*. Yanni and the band took their audience on "a journey through all the ups and downs we encounter in love, no matter who we are and who

we end up falling for". The pre-event promotion of the concert referred to it as "a rare chance to catch home grown talent that now performs on the international stage", and the audience were delighted they had taken the opportunity.

REQUEST FROM AIRDR REGISTER MANAGEMENT

The Australian Inherited Retinal Disease Register and DNA Bank (AIRDR) is funded by Retina Australia to collect DNA from people affected with an inherited retinal disease and their family members, and to analyse this DNA to establish the genetic cause of the disease within each family. We realise that, sometimes, a patient will have the genetic cause of their disease established as a result of investigations carried out by their ophthalmologist or genetic counselling service. The AIRDR would appreciate a copy of any results established in this manner, for recording in its register, to help complete the overall picture of the causes of IRD in Australia. These results will remain completely confidential to the research team, as are the results established by the AIRDR. If you have such results clinically established and you are happy to have them recorded in the AIRDR database, please contact Ms Ling Hoffmann on (08) 9346 2449, or ling.hoffmann@health.wa.gov.au.

Thank you.

Dr John De Roach
Principal Investigator
Australian Inherited Retinal Disease Register and DNA Bank

RETINA AUSTRALIA NATIONAL CONGRESS – GLOBAL EYES 2015

Melbourne October 23rd – 25th 2015 ibis Hotel Melbourne.
Bionics, Gene Therapy, Stem Cells and More.....
For more details 'phone 03 9650 5088
email : support@retinavic.org.au

Planning for the next Retina Australia Congress is almost complete. The event promises to be very interesting and an opportunity to learn about the latest developments in research as well as network with fellow Australians who are affected by inherited retinal degenerative diseases. Conference details containing a comprehensive and eclectic range of topics are appended to newsletter, with registration details available on request should you decide to attend. One of the 'star performers' will be Dr Gerald Chader, last seen when the conference was held in Canberra in 2006.

A number of Retina Australia ACT members hope to attend the National Congress in Melbourne this year. If you would like to attend and need assistance to make the arrangements of to discus transport options etc I would be pleased to assist you in making these arrangements. Please call me on 6258 4823 or email me on mf_jm@optusnet.com.au

GENERAL NEWS

AMANDA VANSTONE AT THE PRESS CLUB

Addressing the National Press Club in Canberra on 8 October, the Chair of Vision 2020 Australia Amanda Vanstone said Australia is facing staggering blindness and vision loss projections as the population ages.

The former parliamentarian also used the World Sight Day Address to call for a smarter, more coordinated, approach to eye health.

Presenting a plan to tackle avoidable blindness and vision impairment, Ms Vanstone told the audience that investing in eye health makes both social and economic sense in Australia and our region.

Ms Vanstone said the cost of vision loss in Australia in 2009 was \$16.6 billion. "With an ageing population it will keep rising," she said.

"People who are blind or vision impaired are twice as likely to have a fall, four to eight times likely to suffer from a hip fracture, enter nursing homes five years early, have a reduced life expectancy and are at three times the risk of depression.

"Doing something effective about vision loss and eye care offers tremendous savings."

Ms Vanstone said government, non-government, private and community sectors need to work together more effectively and spend eye health dollars more wisely so as to avoid duplication, overlap, and waste.

"Australia cannot afford to keep ballooning its health care costs. We have to invest in well founded, evidence-based prevention, early detection and early intervention, and support measures that keep people engaged in the social and economic life of our communities."

Ms Vanstone said failure to invest in eye health in Australia and the Indo-Pacific region would be short-sighted.

"There are oceans of people out there with preventable blindness and vision loss. It has a dramatic effect on their capacity as individuals to contribute to the economy.

"Doing something effective about preventable vision loss is good for economies and especially good for women and girls."

Ms Vanstone said the scale of the problem was staggering and the impacts of blindness and vision loss far reaching.

“One in five of the world’s poorest people live with a disability,” she said.

“Disability can leave you shut out in so many ways and reinforces the problem, which is why there is a strong correlation between blindness and vision impairment, poverty, education levels, and housing access.”

VISION AUSTRALIA CELEBRATES TACTILE FEATURES COMING TO AUSTRALIAN BANKNOTES

Vision Australia applauds the Reserve Bank of Australia, who today announced that the next generation of Australian banknotes will include a tactile feature to assist people who are blind or have low vision differentiate between different denominations.

A recent Vision Australia survey found that almost half of totally blind respondents feel they are being short changed on occasions. The same survey found 61 per cent of respondents with severe to total blindness had trouble differentiating between banknotes.

Existing features of banknotes such as bright colours and large bold numbers can help people who have low vision tell the difference between denominations of Australian banknotes, but this is not enough.

General Manager of Advocacy and Engagement, Maryanne Diamond AO, said the announcement from the RBA is a significant step that will have a measurable impact on the independence and confidence of the 350,000 Australians who are blind or have low vision.

“All members of the community should be able to simply differentiate notes by touch, without using additional devices or asking others for assistance,” she said.

“We have led the campaign for tactile features on banknotes for many years. Countless submissions have been made to the RBA and the Federal Government by generations of advocates, so it’s pleasing to hear the commitment to tactile features made today.”

RESEARCH NEWS

VISION LOSS GENE THERAPY HOPE RISES

Scientists in the United States have taken a step toward personalised gene therapies for a leading cause of inherited vision loss.

The researchers used "induced" stem cells derived from ordinary skin cells to investigate patient-specific causes of retinitis pigmentosa. By testing retinal cells created from the stem cells in the laboratory, they linked RP in two patients to mutations in a gene called MFRP.

The scientists then used a virus to deliver normal copies of the gene into the retinal cells and restore their function. In further experiments, the gene therapy was used to rescue the vision of genetically modified mice with RP caused by the same problem.

"The use of patient-specific cell lines for testing the efficacy of gene therapy to precisely correct a patient's genetic deficiency provides yet another tool for advancing the field of personalised medicine," said lead scientist Professor Stephen Tsang, from Columbia University in New York.

With more than 60 different genes having been linked to RP, it is a difficult disease to study. Prof Tsang's team used induced pluripotent stem (iPS) cells in their research. These are ordinary adult cells that have had their genetic clock re-set, causing them to revert to a basic embryonic stem cell-like state. They are "pluripotent" meaning that they can be transformed into a wide array of different cell types.

"Through genome-sequencing studies, hundreds of novel genetic spelling mistakes have been associated with RP," said the professor. "But until now, we've had very few ways to find out whether these actually cause the disease. In principle, iPS cells can help us determine whether these genes do in fact cause RP. It also helps us understand their function, and, ultimately, to develop personalised treatments."

The research appears in the online edition of the journal *Molecular Therapy*.

STEM CELL INJECTION MAY SOON REVERSE VISION LOSS CAUSED BY AGE-RELATED MACULAR DEGENERATION

Date: April 14, 2015

Source: Cedars-Sinai Medical Center

An injection of stem cells into the eye may soon slow or reverse the effects of early-stage age-related macular degeneration, according to new research from scientists at Cedars-Sinai. Currently, there is no treatment that slows the progression of the disease, which is the leading cause of vision loss in people over 65.

"This is the first study to show preservation of vision after a single injection of adult-derived human cells into a rat model with age-related macular degeneration," said Shaomei Wang, MD, PhD, lead author of the study published in the journal STEM CELLS and a research scientist in the Eye Program at the Cedars-Sinai Board of Governors Regenerative Medicine Institute.

The stem cell injection resulted in 130 days of preserved vision in laboratory rats, which roughly equates to 16 years in humans.

Age-related macular degeneration affects upward of 15 million Americans. It occurs when the small central portion of the retina, known as the macula, deteriorates. The retina is the light-sensing nerve tissue at the back of the eye. Macular degeneration may also be caused by environmental factors, aging and a genetic predisposition.

When animal models with macular degeneration were injected with induced neural progenitor stem cells, which derive from the more commonly known induced pluripotent stem cells, healthy cells began to migrate around the retina and formed a protective layer. This protective layer prevented ongoing degeneration of the vital retinal cells responsible for vision.

Cedars-Sinai researchers in the Induced Pluripotent Stem Cell (iPSC) Core, directed by Dhruv Sareen, PhD, with support from the David and Janet Polak Foundation Stem Cell Core Laboratory, first converted adult human skin cells into powerful induced pluripotent stem cells (iPSC), which can be expanded indefinitely and then made into any cell of the human body. In this study, these induced pluripotent stem cells were then directed toward a neural progenitor cell fate, known as induced neural progenitor stem cells, or iNPCs.

"These induced neural progenitor stem cells are a novel source of adult-derived cells which should have powerful effects on slowing down vision loss associated with macular degeneration," said Clive Svendsen, PhD, director of the Board of Governors Regenerative Medicine Institute and contributing author to the study. "Though additional pre-clinical data is needed, our institute is close to a time when we can offer adult stem cells as a promising source for personalized therapies for this and other human diseases."

Next steps include testing the efficacy and safety of the stem cell injection in preclinical animal studies to provide information for applying for an investigational new drug. From there, clinical trials will be designed to test potential benefit in patients with later-stage age-related macular degeneration.

GENE THERAPY OFFERS BLINDNESS HOPE

Date: December 9, 2014

Source: *Fighting Blindness UK*

A radical form of gene therapy that remodels eye cells into light receptors holds out the promise of restoring sight to people who are completely blind. The new approach is able to create replacement photoreceptors from cells that do not normally react to light.

In early tests on blind rescue dogs with an inherited disease similar to the human condition retinitis pigmentosa, scientists were able to restore sufficient light sensitivity for the animals to distinguish between flashing and non-flashing lights.

Blind mice given the same treatment became as good at navigating a water maze as normal mice. The research, reported in the journal *Proceedings of the National Academy of Sciences*, sets the stage for future clinical trials in humans with degenerative eye diseases. Two components of the "hybrid" treatment involve a gene that alters non-light sensitive cells and an injected chemical "photo switch".

US lead scientist Professor Ehud Isacoff, from the University of California at Berkeley, said: "The dog has a retina very similar to ours, much more so than mice, so when you want to bring a visual therapy to the clinic, you want to first show that it works in a large animal model of the disease.

"We've now showed that we can deliver the photo switch and restore light response to the blind retina in the dog as well as in the mouse, and that the treatment has the same sensitivity and speed of response. We can reanimate the dog retina."

The therapy is one of a number of potential treatments for blindness at early stages of development, two of which yielded exciting trial results this year. In October scientists from the US company Ocata Therapeutics, formerly known as Advanced Cell Technology, showed that stem cell-derived retinal cells could safely be implanted into patients and improve vision in some cases.

Earlier this year scientists at Oxford University hailed trial results from a genetic therapy for choroideremia, a rare inherited cause of blindness that affects one in 50,000 people. Inserting a missing gene called REP1 prevented progression towards blindness and led to dramatic improvements in sight for two men at an advanced stage of vision loss.

The new treatment employs a virus to insert a gene into normally non-light sensitive cells in the retina that gives them the potential to "see". The gene makes a protein that acts like a lock. When the right molecular key from the photoreceptor switch is slotted into the lock, light sensitivity is turned on. At present a new injection of the photo switch has to be made every week to maintain its effect, since the molecule is naturally removed after a period of time.

Several of the dogs have been treated and are currently undergoing tests to determine what level of light sensitivity they now have. The dogs already had the genetic disease when they were rescued from breeders and recruited for the study.

Co-author Dr William Beltran, from the University of Pennsylvania School of Veterinary Medicine in the US, said: "Use of such a clinically relevant large animal model allows us to begin tackling the next challenges on the road to translating this novel therapeutic strategy to human patients."

The therapy is said to show promise because although diseases such as RP destroy the eye's photosensitive cells, other cells in the retina are often left intact and unharmed. They include bipolar and ganglion cells, which both transmit visual nerve signals but do not contain photoreceptors.

In tests on mice, the gene was successfully inserted into almost every one of the rodent's million or so retinal ganglion cells. According to the researchers, this should be enough to restore useful vision.

"So far we can say that the treated mice can distinguish between steady light and flashing light," said Prof Isacoff. "Our next step is to figure out how good they are at telling images apart."

The scientists are also looking to see if the photo switch can be used to activate other receptor types, including some that might allow perception of fainter light.

One of the leading scientists involved in the quest to find a treatment for RP in the UK said the research was "exciting". Professor Paul Bishop, a consultant ophthalmologist at the Manchester Royal Eye Hospital, said: "Converting non-light sensitive cells in the retina into light-sensitive cells is an exciting new approach that has potential to restore sight in people who are blind from hereditary retinal degenerations".

Prof Bishop chairs the medical advisory board of RP Fighting Blindness, a charity dedicated to funding research into a cure or treatment for the condition and providing support to those with it.

The charity's chief executive David Head said: "This is highly-credible work which has been emerging in very recent years." The principles behind so-called 'optogenetics' are fascinating and as ever we are amazed by the research that is being carried out around the world. "We will watch developments very closely and we will certainly be making our members and patients aware of this work. It is important to recognize that it is still at the stage of animal testing, but exciting nevertheless."

RESEARCHERS GRANTED FUNDING BY RETINA AUSTRALIA

Following reviews of submissions to the Retina Australia board the researchers listed below have been granted research funding during 2015:

Dr Sandy Hung with Dr Raymond Wong; Dr Kathryn Davidson; Dr Alex Hewitt; and Dr Alice Pebay of Centre for Eye Research Australia received \$39,551 for their project "Correcting inherited retinal disease through gene editing".

Breakthroughs in cellular technology have led to the ability to generate stem cells from adult tissue. This offers the unique ability to interrogate pathological processes in tissue, which cannot be easily obtained pre-mortem (e.g. retina). In this study we will use recently developed molecular techniques for genome editing to correct and assess three specific mutations, which cause three distinct blinding retinal diseases (Best Disease, Doyne Honeycomb Retinal Dystrophy and Sorby Fundus Dystrophy). Combining these technologies for ocular disease is novel and will lead to the next generation of gene therapy.

Professor Nigel Lovell with Dr Amr Al Abed of University of New South Wales received \$39,405 for their project "Evaluation of electrical stimulation strategies for selective activation of neurons by a retinal prosthesis".

A retinal prosthesis, or the bionic eye, is a promising treatment for the restoration of vision in patients with retinitis pigmentosa and age-related macular degeneration. The device utilises an array of fine electrodes implanted in patient's eyes to deliver electrical pulses to activate the surviving cells in the retina, in particular the retinal ganglion cells which transmit impulses encoding visual information to the brain. In the mammalian retina distinct neural pathways are specialised to detect and transmit certain features of the visual image to the brain. In order for the artificial electrical stimulation delivered by the bionic eye to reproduce physiological vision, the stimulation strategies of these devices have to be programmed to mimic the natural retina in their ability to selectively target neurons. This can be achieved by arrangement of stimulating electrodes or configuring the pattern of electrical pulses delivered by each electrode, areas which our group have been actively researching and developing over the last 15 years. This proposal aims to use a combination of experimental, imaging and computational modelling to elucidate the mechanisms of selective neuronal recruitment of distinct retinal pathways by high frequency stimulation and thence optimise the stimulation strategies delivered by retinal prosthesis. Retinal tissue preparations will be stained for ganglion cells and high resolution 3D geometrical representations of these cells will be reconstructed. The responses of retinal ganglion cells to light stimuli will be imaged in mass by advanced microscopy and populations of these cells would be functionally classified. The neuronal cells shapes and functional responses will be captured into a computational model of the retina that would simulate the effects of electrode size and configuration in a multi-electrode array of a retinal prosthesis, on retinal cell population responses and advance our understanding of the mechanisms underlying selective neural recruitment. Our objective is to introduce and test a new approach to retinal neurostimulation that will allow targeted neural

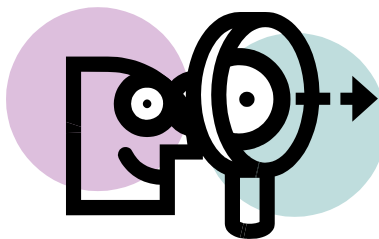
recruitment. While contributing directly to our knowledge of neural activation in general and retinal neurophysiology specifically, the work will also be a critical enabler of improved vision processing and stimulation strategies in future generations of bionic eye.

Dr Marc Sarossy with Behzad Aliahmad and Prof Dinesh Kumar of RMIT University, Melbourne received \$40,000 for their project "Hyperspectral funduscopy for non-invasive detection of retinal ischemia".

Retinal ischemia is the cause of a large number of blindness cases. Successful completion of this project will lead to a novel way to diagnose retinal ischemia and the extent of retinal blood flow without fluorescent dye injections, which is invasive and requires extensive infrastructure, and has the risk of allergic reactions.

To test this hypothesis, a hyperspectral camera, a camera with many more colour channels than commonly used RGB cameras, will be integrated with fundus imaging equipment, and retinal images will be taken of volunteers (both healthy and diseased) prior to their fluorescence angiogram. Spectral analysis of the images will be performed to automatically detect abnormalities (features) corresponding to retinal ischemia.

Such a method will provide non-invasive detection of ocular ischemia without the extensive infrastructure that is required for fluorescein angiography, which is currently used for this purpose.



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).

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.

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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