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Newsletter of Retina Australia (ACT) Inc

EDITION 4-2012 NOVEMBER 2012

MESSAGE FROM THE PRESIDENT

Welcome to the fourth and final newsletter for 2012, in which the major item is a report on the Retina Australia Congress, held at Parramatta in October. There is also a report on the national organisation's Annual General Meeting.

You can read, too, about updated plans for Retina Australia (ACT)'s major annual fundraiser, a 'must' for the diary, while members are also encouraged to attend the organisation's Annual General Meeting on 27 November. Both events will provide the opportunity for members to mix and mingle before the Christmas and New Year festivities, and are opportunities your local executive welcomes very much. We hope to see as many members as possible at both functions.

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 8 January 2013

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

The **Fundraiser at Thoroughbred Park** is our major activity of the year to raise money for the DNA Database and also monies for our researchers who are working so hard on our behalf. If you have contacts that would be happy to donate goods for our raffle I would be so grateful for this assistance. Lyn and I will pick them up if needed. Please contact us if you would like to discuss further.

Jan James
Secretary

DATES FOR THE DIARY!

As indicated in the October newsletter 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. We have also extended an invitation to Dr Terry Diamond, Retina Australia's 'marathon man', who in March ran a marathon every day for seven days in seven states and in so doing raised approximately $150,000 in research funding for the national organisation. Lyn Barlow and Jan James are working on ensuring that this year's event will be better than ever, so put the date in your diary and ask as many of your family and friends attend as possible. **A flyer is included with this newsletter.**

Please also note that the **Annual General Meeting of Retina Australia (ACT)** is to be held on **Tuesday 27th November at 6.30 pm at Elders Real Estate Office, Jamison Centre.** We hope very much to see as many members as possible at the meeting. Refreshments will be served.

RETINA AUSTRALIA NATIONAL CONGRESS 2012

The theme of the 2012 national Congress, which was held on October 20 and 21 at the Novotel Hotel in Parramatta, NSW, was **"Hope in Sight".** The Congress, which was very well organised, provided a variety of excellent presentations and a choice of workshops for the 140 people who attended.

The **Hon. Barry Jones AO** officially opened the Congress. Dr Jones spoke about his work with Vision 2020 and about eye care in general. In particular he mentioned that the message of Vision 2020 International is that early detection of eye disease is imperative and so people should not put off going to their eye health professional.

The major thrust of the overall program was to inform delegates of the latest research involving inherited retinal diseases being undertaken in Australia and throughout the world. Details of presenters and their presentations are as follows:

Professor Robin Ali, the Professor of Human Molecular Genetics at UCL Institute of Ophthalmology, London, was the keynote speaker. Professor Ali participated at various times throughout the Congress within workshops and led the scientific panel at question times. He provided an overview of gene and stem cell therapy for inherited retinal diseases and explained some of the research he has been leading during the past fifteen years. In particular he spoke about:

- the clinical (or human gene therapy trials for patients with Lebers Congenital Amaurosis (LCA) and the fact that early results indicate that the participants have improved night vision.
- the gene therapy treatment for at least 11 forms of early onset severe retinal disease, which his team have developed using mouse models. They are now at the point that they can slow degeneration, but it will be some time before these therapies will be tested in humans.
- his investigative work on X-Linked RP, with particular consideration of the RPGR defective gene. He reported that some progress has been made but the work is quite difficult because of the "bottleneck" of genes in the photoreceptor.

- regenerative medicine, an interdisciplinary approach to the repair, replacement and/or regeneration of damaged tissues and organs involving tissue engineering, stem cell biology and gene therapy. The timescale for delivery of such treatment can vary between five and 30 years as there are many challenges to overcome before a product can be developed.
- stem cell therapy for retinal repair, which is potentially a regenerative medicine strategy for many forms of retinal disease. Currently investigations are under way for the transplantation of Retinal Pigment Epithelium(RPE) cells and photoreceptors. These strategies are regarded as important because the loss of photoreceptor cells is the main reason that people with inherited retinal diseases go blind, except for those with Stargardts disease, where it is the RPE that is diminished.
- his laboratory, which has demonstrated that effective photoreceptor cells can be transplanted using mice models. They have proven that the vision of the mice has improved, but are still investigating how long these integrated photoreceptors will survive.

In conclusion, Professor Ali said that his overall aim is to find therapies independent of the type of genetic inherited retinal disease.

Dr John De Roach, the Director of the Australian Inherited Retinal Disease Register & DNA Bank (IRDR), and **Ms Tina Lamey** who has responsibility for the day to day operations of the IRDR, spoke about their work. John gave an historical account and reported that overall there are now over 5000 registered participants. He also spoke about the limits of the work and their goals for 2013. John also explained that a clinical reporting service is beginning to emerge and he is seeking government funding for this. He believes this is an unmet need and an important development for the future. For people interested in looking at the data collected to date, or to find out more about the work of the team, information is provided on the two websites:
www.IRDregister.org.au and www.scgh.wa.gov.au/Research/InheritedRetinal.html.

Tina then spoke about the genetic analysis component of the work. She explained that it was important to understand the clinical manifestation of each disease to understand what may happen in specific cases and to develop a genetic picture of inherited retinal diseases across Australia. She also said that it was important to confirm clinical diagnosis so that genetic counselling can occur, and that analysis was also an important step for development and application of personalised gene therapy. Tina also spoke about some of the results their team had produced and in particular those for which clinical trials are underway or in the pipeline. She said the emergence of new technology will assist with future work.

Dr Lauren Ayton, the Clinical Research Co-ordinator for the bionic eye project, explained that the project team were currently working towards giving some level of sight back to people who have no vision or very limited light perception. She spoke about the different types of prosthetic devices currently available throughout the world and explained that most bionic eye researchers are working with patients with RP and hoping to follow up with AMD patients in the longer term.

Lauren then spoke specifically about the recent developments in Victoria, where three people have recently received a sub-retinal, 17mm long silicon implant with tiny

platinum electrode spots. These "bionic eye" recipients regularly attend the laboratory to undertake various tests, which are conducted by connecting the device directly through a connector plug worn just behind the ear like a cochlear implant. They are not hooked up to any camera. This research with human patients is in its infancy but has already been of great benefit to the work of the research team. Lauren mentioned that progress in the last two years, and particularly the last two months, has been amazing. This has been assisted by the number of people, particularly members of Retina Australia Victoria, who have volunteered to assist the project by offering to have their eyes photographed, answer questions or participate in orientation and mobility trials.

Lauren said the Bionic Vision project outcomes were concerned with improved visual acuity, orientation and mobility, daily living activities and improvements in the quality of life. The next device to go to clinical trial will be the wide view suprachoroidal type of device, which has 98 electrodes, and this study will be concerned with orientation and mobility in particular. In the future the team will test a high acuity epiretinal device with up to 1000 electrodes. It is believed that such a device could allow reading of large text or even facial recognition.

Associate Professor Erica Fletcher heads the Visual Neuroscience Laboratory at the Department of Anatomy and Cell Biology at the University of Melbourne. She has a strong interest in the mechanisms of photoreceptor degeneration and spoke about her research investigating the role of purines in photoreceptor death. Dr Fletcher's results indicate clearly that purines, which are themselves released from dying photoreceptors, exacerbate the death of neighbouring photoreceptors. Currently she is investigating chemical compounds which will reduce photoreceptor death.

Dr Una Greferath works within the Department of Anatomy and Cell Biology, University of Melbourne, where much of her research involves aspects of the development of the retina. She reported on her work, which was funded by Retina Australia. The main aspects of her talk was that Mueller cells are crucial support cells in the retina and important to photoreceptor integrity. Mutations in genes affecting Mueller cell function lead to Lebers Congenital Amaurosis.

Professor Robyn Guymer is Head of the Macular Research Unit, Centre for Eye Research Australia, University of Melbourne, where her research is mainly in the area of macular degeneration. Robyn is also part of the Bionic Vision Australia team. She explained that AMD starts with subtle changes in the eye and for some patients it involves bleeding at the back of the eye, which is known as Wet AMD. The treatment for Dry AMD involves injections into the eye, and although many people regain vision and retain this vision for the rest of their life, there are still 15 per cent of patients whose vision does not improve. Currently Professor Guymer is looking at the genetic structure of each patient's AMD to determine why some people gain much more vision than others with the treatments currently available, namely Lucentis and Avastin. It is believed that results of these studies may lead to individualised treatment regimes and a much higher success rate.

Associate Professor Robyn Jamieson is involved with Paediatrics, Genetic Medicine and Ophthalmology at the Medical School, University of Sydney, and is Head of Developmental Genetics Research at the Children's Hospital, Westmead NSW. She spoke about her work with cone-rod and rod-cone dystrophies. Robyn believes it is

crucial to identify the gene and the inheritance patterns so that factual advice can be given to families in the first instance so that when potential treatments become available families can decide whether to access them or not. She explained clearly the different types of inheritance patterns by using diagrams of chromosomes. Some data she provided included the fact that everyone has 23,000 genes and 3,000,000 base pairs so there are lots of things to consider when providing advice. If people are interested in finding out about local clinical testing, or the human genome project they should look at either http://www.genetics.edu.au/ or http://genome.ucsc.edu/.

Professor David Hunt is Winthrop Professor at the School of Animal Biology, University of Western Australia. A considerable amount of his time is involved with research into dominant macular dystrophies. He explained that he had chosen to study dominant inherited disorders because these run in families and so over years can be mapped into the future. His studies have found that high levels of calcium have been responsible for the death of photoreceptors and so these patients may be suitable for gene therapy. He is working with Dr Robyn Ali in the development of such therapies.

As well as these scientific workshops, **Mr Michael Simpson**, from Vision Australia, spoke about the government and non-government entitlements of a vision impaired person within Australia.

There were also a number of workshops provided for delegates. The topics included: What is it like to be blind?; Retina Youth: "Me Too"; Vision impaired people and the kitchen; Sighted Guide Skills; Ophthalmic issues and gene therapy or stem cell therapy; Youth Issues (Genetic and financial advice); The impact of vision impairment on the family; and the latest technology for vision impaired people.

Overall it was a very successful Congress with "information overload" for delegates. Some of the "Take home messages" were:

- Over time a number and variety of treatments for inherited retinal disease will become available and it should be noted that some treatments will suit some people and their eye disease better than others.
- With the potential for treatments and possibly even cures being developed for widespread usage within the next decade or so, it is imperative that people with inherited retinal disease have an exact diagnosis of their genotype and also that they have access to genetic counselling.
- Those suffering retinal diseases should be aware that advertised treatments in some magazines and on the Internet could well be shams or potentially fraudulent. All legitimate treatments or cures are published in reputable scientific journals and through mainstream media.

RETINA AUSTRALIA NATIONAL AGM 2012

The Retina Australia Annual General Meeting was held following the Congress on Monday 22 October 2012. The directors elected for the next twelve months were as follows:
RA (NSW) Graeme Banks (President), Betty Ghent (Secretary)
RA (ACT) Robin Poke, John Barlow
RA (WA) Pina Faliti-Smith, Sean Smith

RA (Queensland) Anne Housego, Geoffrey Munck
RA (Victoria) Leighton Boyd (Vice President), Rosemary Boyd
RA (SA) Philippa Cooper (Treasurer), Orm Cooper

At the meeting the President, Graeme Banks, and each of the State Presidents presented their reports, which outlined what activities each organisation had been undertaking during the previous financial year. The audited financial reports were also presented and accepted. The general business discussed included:

- continued funding of the IRDR and DNA Bank as a priority for the next three years,
- a proposal by the Retina Australia Youth Ambassadors that a trek with Inspired Adventures to become a potential national fundraising activity,
- the success to date of the Facebook page, "Retina Australia – Youth", which has a number of participants aged between 18-35 sharing their stories,
- a proposal to include links to social media, including Twitter and Facebook, on the national and state websites,
- a decision that the 2013 AGM would be held in Canberra, and
- a decision that the 2015 national Congress would be held in Melbourne.

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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