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Quarterly National Newsletter of
Retina Australia

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CHAIRMAN'S REPORT

Congratulations to Fraser Alexander and his Congress committee for hosting the highly successful 2018 Retina International Congress at the University of Auckland between 8 & 12 February 2018. More than 400 people attended the public sessions, including many Retina Australia members. It was exciting that 21 Retina International member countries were represented at the business meetings. The entire event ran like clockwork because of the significant time and effort that went into its planning.

The Congress was officially opened by Ms Christina Fasser, President, Retina International, who gave a brief synopsis of the developments in research throughout the world. She mentioned that the first inherited retinal disease causing gene was found in 2004 and in the intervening years researchers have discovered an additional 289 genes and believe there are more to come. Ms Fasser also stated that there are currently 17 clinical trials underway which are full of promise for treatments for some strains of the disease, with many more on the horizon. She indicated that Retina International is concerned that when treatments are approved for general use, they will need to be available in every country, easily accessible for those who may be advantaged by such treatments, and most of all will need to be affordable for everyone. Ms Fasser also said that the diagnosis of specific strands of inherited retinal disease and counselling of the affected person will be extremely important to ensure that the appropriate treatment regime is applied.

The speakers taking part in the Congress were selected for their expertise from a considerable array of well-known researchers into inherited retinal diseases from across the world. Dr Elise Heon from Canada, Professor Jean Bennett from Pennsylvania and Professor Eric Pierce from Massachusetts were keynote speakers. Other major presenters included Australian researchers Associate Professor Alex Hewitt, Associate Professor Alice Pebay, Dr Tom Edwards, Associate Professor Penny Allen and Professor Mark Gillies. Associate Professor Andrea Vincent headed the New Zealand scientific team and Dr Gerald Chader, from California, presented the closing remarks.



Christina Fasser



Elise Heon



Jean Bennett



Eric Pearce

Researcher's presentations focussed on the four major areas outlined below.

- **Genetics and Gene Therapy**

There have been significant advances in genetic testing for inherited retinal diseases, however all researchers mentioned that the genetics of inherited retinal disease is complex and that all the clues must come together to give an exact classification. For testing, the options are: a single gene screen, a retinal panel, whole exome sequencing and whole genome sequencing. Scientists reiterated that the technology of testing is constantly changing and so this field is an exciting one to be involved in.

Attendees also heard about the novel, or non-coding causes of inherited retinal disease, how scientists identify patients for clinical trials, and the progress and lessons learned from clinical trials which are underway. Other factors included the cost of such treatments, the processes involved in the various trials being carried out, genetic discrimination, inconclusive and surprise results from the research and why certain gene types were more suited for treatments at this stage of the investigations.

- **Stem Cells and Research Challenges**

In this section of the program an explanation of the use of stem cells to model eye disease was given. It was fascinating to learn about how stem cells can be made into any kind of cells, including eye cells, and that researchers can get a patient biopsy from stem cells without having to take a biopsy from the eye. Information was also provided about gene editing, retinal ganglion cells, the use of a robot to feed the experimental cells, and why it takes a long time for treatments to be developed.

This area of research is moving rapidly, however there is still a long way to go before the knowledge gained is transformed into a practical application, or treatment, for those affected by inherited retinal disease.

- **Artificial Vision**

The history of prosthetic devices was outlined and an explanation of the varying approaches to electrical stimulation of the visual cortex was given. Information was provided about the epiretinal, subretinal, and superchoroidal prostheses and details of the current stages of research were provided.

There are currently two devices available for patient use, the Argus II and the Alpha. Both are extremely expensive and currently only available in the USA and Europe respectively, however the companies involved in producing these devices are investigating distribution in Australia.

• Scientific Breaking News

This section provided a snapshot of the current and emerging gene therapies for inherited retinal disease, the ReNeuron clinical trial, AMD therapeutic trials and pathogenesis, as well as robotic surgery for inherited retinal disease. Details were also given about Ranizumab and Brolucizumab, which are the new and emerging treatments for AMD, and which appear to be superior to the currently used Lucentis, Avastin and Eylea treatments.

All research presentations provided great hope for the future and were provided by enthusiastic and dedicated researchers who know how important it is to find suitable treatments for all inherited retinal diseases as quickly as possible.



In addition to the research presentations, sessions were held to discuss:

- the latest vision aids and technology which could be used to support independence,
- international perspectives on vision rehabilitation,
- patient registries,
- paediatric low vision support services,
- advanced and future technologies,
- travelling with sight loss, and
- the perspectives of parents of children with inherited retinal disease.

Overall the 2018 Retina International Congress program was packed with information. Participants also had the opportunity to interact and socialise with presenters, and others affected by inherited retinal disease, from all corners of the globe. It was clear from all presentations that there are many people whose paramount importance in their work is to find ways to improve the life experiences for all those affected by vision loss as a result of inherited retinal disease. I found this to be extremely exciting and look forward to reporting on any developments during the next year or two leading up to the next Retina International Congress which will be held in June 2020 in Reykjavik, Iceland.

Through attending this Congress and the associated business meetings, I am convinced that Retina Australia needs to become more involved with Retina International, to work closely with its CEO Ms Avril Daly, to utilise the information provided on its website, to keep abreast of the research around the world, and to monitor the clinical trials which may eventually lead to treatments for the various forms of inherited retinal disease experienced by Australians. It is also obvious from comments made by the various researchers that retinal disease registers, such as the Australian Inherited Retinal Disease Register & DNA Bank, are increasingly important as a way to recruit participants for clinical trials, and we need to be on the lookout for members who can assist with this important resource.

It is clear that the future of Retina Australia and Retina International is through its youth. To this end, a youth program, convened by New Zealander, Zane Bartlett, was held prior to the 2018 Retina International Congress in Auckland. Melanie Chatfield, one of our Retina Australia Board members, provided a thought provoking and inspiring presentation to the assembled youth. The program was extremely successful and will hopefully encourage those in attendance to become more proactive with their lives and potentially contribute to local and international groups. A more detailed account of the youth program appears later in this newsletter. I would encourage all Australians with inherited retinal disease under the age of 35 to become involved in the Retina International youth movement. If you would like more information please contact Melanie on melaniecchatfield@hotmail.com or Zane on youth@retina.org.nz.

I am fortunate to have attended many Retina International Congresses. This one was quite significant in that a number of the presenters, or members of their research teams, have been recipients of grants from Retina Australia in past years. It clearly indicates, thanks to the generous donations of our members and friends, how much of an influence we have had in shaping research into inherited retinal disease in Australia and internationally.

Retina Australia

Chairman

Leighton Boyd



NOTE: *If you are interested in attending the 21st Retina International World Congress in Reykjavik Iceland in 2020 please log on to the Congress website: www.riwc2020.is for information. This website will be updated on a regular basis as plans for the Congress begin to take shape.*

GRANTS FUND INNOVATIVE RESEARCH

From an article by Helen Carter, Optometry Australia – 22 January 2018

Professor Erica Fletcher, deputy head of the University of Melbourne's Department of Anatomy and Neuroscience, recently received two new federal government grants totalling nearly \$1.6 million to help fund her research.

A \$933,952 grant to investigate novel mechanisms of early age-related macular degeneration (AMD) will enable her to examine a mechanism by which retinal pigment epithelial cells contribute to vision loss early in the disease.

'This four year project focuses on how the retinal pigment epithelium changes during the early stages of AMD and whether treatment prevents cellular changes,' Professor Fletcher told Optometry Australia.

'Cells and their subcellular components need to renew themselves to stay healthy. We think that the way the RPE cells renew themselves decreases with age, and is a critical factor in the way the RPE changes during the early stages of AMD.

'We will use animal models as well as RPE cells derived from patients with AMD to determine how the signalling pathways important for cell renewal are affected during age and AMD.' We will then trial a novel nanosecond laser and/or two currently used drugs to see if we can slow disease progression.

The second grant, of \$665,582, will enable investigation of the role of microglia in early diabetic retinopathy.

'This project will examine the role that retinal immune cells called microglia have in causing early changes in the vasculature in those with diabetes' she said. 'We will examine whether diabetes changes the way neurons communicate with blood vessels, opening up a possible treatment target that could prevent the progression to more advanced disease.'

In addition to the two NHMRC project grants, Professor Fletcher also received an Australian Research Council grant which focuses on the biology of resident immune cells in the retina and the mechanisms by which they regulate the development of cone photoreceptors.

'Our work has shown that cone function doesn't develop in a normal manner when microglia signalling is disrupted. This project will examine the molecular biology - the genes expressed by individual microglia - and explore how they communicate and change photoreceptors,' she said.

'Although this project is highly focused on biology of the normal retina, it has long term benefits for our understanding of AMD because patients with AMD often have mutations affecting microglial function.'

In the same funding round, Dr Lauren Ayton also received \$179,118 to improve patient management pathways in AMD, a project which will continue a collaboration with Professor Robyn Guymer from the Centre for Eye Research Australia (CERA), and develop new training methods to up-skill optometrists on the latest advances in AMD from international research.

Dr Ayton, an honorary senior research fellow at the Department of Surgery (Ophthalmology) at the University of Melbourne and CERA, is currently working as Director of Clinical and Regulatory Affairs at the Boston Bionic Eye program in New York, affiliated with Harvard and Cornell universities. She will postpone her 2018 NHMRC Medical Research Future Fund Next Generation Clinical Researchers program Translating Research into Practice (TRIP) Fellowship until her return to Australia. The project focuses on improving patient management pathways in AMD.

‘There are new advances in the field of AMD occurring at a rapid rate, and it is vital that optometrists are educated on these changes in diagnostic and management protocols,’ Dr Ayton told Optometry Australia.

‘The TRIP fellowship will provide support for new research translation modalities. I am extremely honoured to have been awarded this prestigious fellowship, and am excited about working closely with Optometry Australia and the optometry profession on this project.’

Congratulations to Erica and Lauren who have both been recipients of research grants from Retina Australia and who are great advocates and supporters of our organisation. These grants are recognition of their significant contribution to IRD research and thoroughly deserved.

GOOD DIETARY SOURCES OF NUTRIENTS IMPORTANT TO EYE HEALTH

Vitamin E : Whole grains, nuts and legumes;
polyunsaturated oils (vegetable oils); monounsaturated oils
(olive oil)



Vitamin C : Fruits and vegetables
(especially citrus fruits, melons, and broccoli)

Zinc : Meat, poultry, fish, and dairy products



Carotenoids (Lutein and Zeaxanthin) :Fruits and vegetables
(particularly dark leafy greens and those that are yellow or green)

THE BENEFITS OF PSYCHOTHERAPY

John Delany, Senior Counselling Manager, Insight Counselling Service,
Fighting Blindness, Ireland.

Sight is probably our most treasured sense, and the thought of losing it naturally gives rise to feelings of fear and uncertainty about the future. The issues that a person with sight loss faces are both practical and psychological in nature as one makes the transition from a sighted to a partially or even non-sighted lifestyle. The journey of adapting and coping with new and uninvited circumstance is unique for each individual. That needs to be understood and respected by all who offer encouragement and assistance in order to ensure that the process of adjustment is not impeded.

At such a challenging time, engaging in psychotherapy can help. Working with a psychotherapist in a safe, trusting and non-judgemental environment allows the individual with sight loss the time and space to make sense of what is actually happening to them, away from the pressure of well-meaning but often ill-informed and anxious family and friends. With the appropriate support and guidance it is possible to work through and surmount difficulties to find new and life-enhancing meaning and purpose in living.

The psychotherapist offers a relationship where the person's innate inner wisdom and potential for positive change can be realised and harnessed. By freeing the natural healing process in the client, the integrity of personhood is honoured and championed. It takes time, commitment and courage to undertake such a journey but with the help and facilitation of a trained psychotherapist it is possible to reach the desired destination.

Sometimes life can be unfair. We study hard but don't achieve the hoped for grade; we train assiduously but never get picked for the first team; we work enthusiastically but are overlooked for promotion; we invest cautiously but the stock market crashes; we experience bereavement and the grief that ensues; and we risk loving another only to be rejected. But we still survive. The human being is designed to heal given the right circumstances and opportunities. We can't stop bad things happening; we can only choose how to respond. We take our life experiences and mine them for the gold of wisdom that lights our path through dark and alien territory. We grow and evolve so that sight loss informs but does not have to determine how we live our lives.

It is not just the individual with sight loss who is affected but also their partner, children, extended family and friends. At Insight, we work with all those affected directly and indirectly by sight loss. We offer a range of services including psychotherapy, support groups and technological assistance in conjunction with other relevant agencies.

Sight loss is a life changing experience, but not a life ending one.

INTRODUCING THE GOOGLE DISABILITY SUPPORT TEAM

Date: Thursday, January 25, 2018

The Google accessibility team has launched a dedicated disability support team. The support team is available to answer questions about using assistive technology with Google products and accessibility features and functionalities within Google products. Read more to learn about the team and plans for the future.

Additional support was a popular request from the Google accessibility community. We're pleased to announce the launch of our first dedicated Google disability support team. Agents are available Monday through Friday, 8am-5pm PST (Australian Eastern Standard Time is 18 hours ahead of Pacific Standard Time hence availability is 2pm-11pm Eastern Standard Time).

The support team is available through email and only in English language. Contact the team at disability-support-external@google.com and you will receive an answer by a support representative within 72 hours.

The support team is exploring adding additional languages and support channels in the future. Be sure to check back on this [Google accessibility blog](#) for more updates and announcements from the team. Read below to learn about additional resources offered and ways to keep in touch with the Google accessibility team.



Get additional support through communities

- [Join the eyes-free community](#) for Android eyes-free products
- [Join the accessible community](#) for general Google accessibility inquiries

Stay updated

- Stay up to date by following us on Twitter ([@googleaccess](#))
- Visit the [Google accessibility blog](#) monthly for new posts

Learn more

- Explore the [Google Accessibility website](#)
- Help us test! [Sign up to participate in user studies](#)

RETINA INTERNATIONAL CELEBRATES

RARE DISEASE DAY 2018

The promotion and development of research into rare and inherited forms of retinal disease (IRD) is a fundamental objective of the Retina International community. On February 28th 2018 we joined the multi-stakeholder global community to celebrate Rare Disease Day and called for structured collaboration among stakeholders that will lead to sustained funding, promotion and protection of rare disease research.

Rare disease research represents a broad range of scientific investigations to establish knowledge on rare diseases. Basic rare disease research involves the biochemical study of underlying pathophysiological mechanisms and their genetic and molecular characterisation. Clinical research focuses on the development of diagnostic tools and therapeutic solutions while translational research accelerates the transfer of knowledge from basic “bench-side” research into clinical “bedside” applications.

Equally important, in this multidisciplinary field of research, are studies on the social consequences of disease, health economics, communication and culture as well as epidemiological studies and research into the natural history of a disease. These studies help develop improved standards of care, treatment and a better quality of life for people living with rare disease. Specific and careful allocation of budgets and coordination of activities at national, regional and international levels are imperative for accelerating the journey towards the ultimate goal - effective treatments for unmet need.

Why is Research so Important for those affected by Rare Diseases?

Members of Retina International agree with their peers in other areas of rare disease that the greatest barrier to prevention, diagnosis and treatment is insufficient knowledge. Lack of investment therefore leads to delays in the development of fundamental scientific progress resulting in under-diagnosis, misdiagnosis and delays in diagnosis.

The key to developing this knowledge in the retina space is in supporting and promoting all elements of research into Inherited Retinal Disease (IRD). Investment in IRD research will improve the identification, understanding and development of therapeutic options for these conditions.

Due to their low individual prevalence, complexity and requirement for a multidisciplinary approach, the field of rare disease research is one in which the benefits of specific and targeted coordination and collaboration are most

obvious and pressing. Traditional funding mechanisms including “normal” market conditions, patient organisation fundraising efforts, and public funds are not adapted to the reality of rare disease research requirements, which include concerted efforts at both the national and international levels through the development of national plans and strategies.

Patients as partners

Retina International believes that patients should be partners in research not only as subjects, but as advocates for fundraising and key stakeholders in the drafting of guidelines and policies, and should always be consulted in the drafting and evaluation of national research policy in the context of strategic plans.

For more information refer: <http://www.retina-international.org>

RARE DISEASE DAY AWARD 2018

Congratulations to:

Professor John De Roach, Principal Medical Physicist at the Australian Inherited Retinal Disease Register who was recognised on Rare Disease Day for his important work in the field of genetic and rare diseases.



APP FOR THE VISION IMPAIRED COMMUNITY

ViaOpta Hello is a multi-tool application that can assist blind or vision impaired people by using cognitive services and Microsoft latest image analysis technology to identify people, items, and scenes.

This application is an example of how people can use their knowledge to partner with tech companies to assist the vision impaired to make their daily lives more accessible. The application can be downloaded on any Smartphone from the app store. Coming in twelve different languages, this application takes away the stress of potential language barriers, making it useful for anybody on-the-go. By using the Smartphone's camera to take pictures of objects or people, the back-end cognitive services provide a description of the content.

ViaOpta Hello has just been launched in Switzerland, Hong Kong, Malaysia, Thailand, East Africa, Italy, Nordics, Spain, Australia, Turkey, South Africa, China, Ireland and Singapore. A launch of the app is planned for Germany, USA, France, and the Middle East in the not too distant future.

To date there have been over 3500 downloads of the app and it has been well received by the community and seems to be filling an unmet need. For more information log onto <https://www.viaopta-apps.com/Viaopta-hello.html>

SHARE YOUR STORY

Sharing your story could help others affected by Inherited Retinal Disease

Retina Australia is looking for volunteers to share their story of living with an IRD. The new look Retina Australia website will feature a selection of short stories from people directly affected by an IRD or who provide support to someone close to them such as a child, sibling or partner.

Being diagnosed and living with an IRD can be difficult, and it helps to know you are not alone. Personal stories can provide hope, practical advice and challenge some of the assumptions of what it means to be vision impaired. They might encourage someone to reach out for help, give them greater confidence or inspire them to get involved in a new activity.

If you would like to tell your story or would like further information please email admin1@retinaaustralia.com.au.



Stay up to date with the very latest information, including research, events and notice of the Retina Australia website launch, by 'Liking' the **Retina Australia** Facebook page.



5 THINGS I LEARNED FROM MY TRIBE AT THE 2018 RETINA INTERNATIONAL CONGRESS

By Melanie Chatfield

We live in a largely sighted world. You just have to take a peek at social media to see the value placed on images and what things look like. But we don't all see stuff the same.

As someone living with a degenerative retinal condition I am used to being the only legally blind person kicking around. So it is a real treat when I get the chance to hang out with people who relate to what it is like to have low vision.

From 7-9th February, around 20 young people gathered in Auckland New Zealand to take part in the 2018 Retina Youth Event. The program comprised a mix of networking, cultural and leisure activities, as well as guest speakers – of which I was one.

Many youth participants stayed on for the weekend, joining the delegation of researchers, patients, service providers and families at the Retina International Congress scientific program that followed.



It was such a privilege to hang out with this crew for a few days, forging lifelong friendships with fun loving people from across the planet. Here is what I learned:

1. Diversity is the norm

Each of us was there because we had a problem with our eyes. Yet everyone had a different story to tell. Our history, disease symptoms, life experience, preferences and personalities combined to create a unique mix of individuals.

Amongst the group were PhD candidates, community service professionals, students of law and life, athletes and artists. A vibrant collection of inquiring minds, advocates, comedians and social change makers.

Whilst diverse, we quickly found something in common with one another. Be it a love of dad jokes, metal music, fantasy novels, philosophy or food, there was plenty to talk about.

Our magnetic personalities were strong enough to attract a friend from Argentina. Whilst not an official member of the youth group, his infectious positivity and perseverance in communicating through Google translate was immediately endearing. He soon cemented himself as an honorary member of the gang and had an uncanny knack for finding us wherever we were.

2. Make no assumptions

Many of the youth group agreed that others often made judgements about them without knowing their situation. People underestimated what they could do, and overestimated what they could see, defaulting to pre-conceived ideas and assumptions.

The resounding advice was simply – Just Ask. If you don't know or are unsure be curious. "What do you like, need or want?" "Would you like help?"

It is also important to remember that such an approach means that each of us needs to have the confidence to speak up and assert what it is that we prefer.

3. Be authentic

Be frank and talk about the things that matter. More often than not we are afraid to ask or verbalise our personal fears and aspirations. If you feel safe and supported give it try. Talk about love, heartbreak and family. Sex, drugs and mental health. Failure, desire and pride. Your openness and vulnerability will likely encourage others to share their thoughts.

If you are listening, do so without judgement. If you don't agree with a point of view, gently delve deeper and try to understand where the other person is coming from. You will be rewarded with an opportunity for connection far deeper than small talk offers.

During an intimate visit to the Te Tahawai Marae, participants learned of the value of disclosing a little bit about who they are and where they come from.

Listening, singing, sharing and learning about Maori culture provided a wonderful opportunity to connect and relate to others. The experience so profound for some that they likened it to a powerful connection with family.

4. Have courage



Physical activities such as hiking to Kite Kite waterfalls and surfing at Piha beach offered the chance to be brave and stretch beyond comfortable experience. Whether it was jumping under a freezing stream of water, stepping into the ocean, standing up on a surfboard, clambering over rocks or dodging low hanging branches, it took courage and resilience to give it a go irrespective of the outcome.

Discovering that you are more capable than you think builds confidence and helps you to grow. The experience adds to your story and can give you courage to try things that may be more difficult.

5. Get creative

Have you ever tried to grab a handful of sand? A few grains always drop out and get lost. The same could be said for gathering a group of low vision people in a strange city for a night out on the town.

While we misplaced a few people at times, various coping techniques and tricks emerged to make life easier. I learned that articulating the mating call of a rare exotic native bird is an effective way to echolocate your mates, while looking through your phone's camera can help you navigate a dark crowded venue.

Stopping in at every bar to ask if anyone has seen a rowdy bunch of blind people is a tenacious approach and means you won't be separated from the pack for too long.

We all have things we are good at. Be generous with your life hacks and open to learning from others. If something is proving challenging or difficult take some time to think about it from another angle. Perhaps there is a way around it after all.

6. Look forward

One reason to look forward is the chance to reduce the number of things you run into. Another is to stay focused on opportunities rather than dwell in what could have been.

While most of us were adept at seeing with our shins, toes and knees, the scientific conference offered hope for an alternative. Developments in stem cell therapy, gene editing and technological advances were encouraging for many of us with rare eye diseases. While the international network of researchers were quick to keep patient expectations in check, it was difficult not to be excited by the innovations already underway.

Experienced members of the Retina International executive encouraged younger delegates to look towards the future. There is considerable opportunity for younger members to consider the role they might play in not only continuing the existing legacy, but forging their own paths both locally and globally.

Paralympic swimmer Mary Fisher provided a great example of looking forward. As a guest speaker she gave an incredible account of her journey as an elite sports person, inspiring the group with her tale of focus, commitment, ambition and achievement.

Participating in a Retina Youth event offers an incredible opportunity to be a part of something special. If you are aged between 16-33, I encourage you to sign up for Iceland 2020 and join others from around the world. You won't regret it.

USING GENES FROM ALGAE TO RESTORE LIGHT SENSITIVITY

By Donna Dovey - *Newsweek*

Date: 16 January 2018

A French biopharma company has announced their plans to carry out human trials of a new treatment that would insert genes from light-seeking algae into the eyes of patients with inherited blindness in order to help them regain sight. The treatment involves optogenetics, a technique that converts nerve cells into light sensitive cells. Although optogenetics is commonly used in animal experiments, its effects on humans are far less known.

Recently GenSight Biologics announced that the UK Medicines and Healthcare Regulatory Agency accepted the company's clinical trial application to start Pioneer Phase I/II. The trial, expected to begin on UK patients in the first quarter of 2018, plans to study gene therapy as a treatment for patients with the genetic condition of retinitis pigmentosa.

Retinitis pigmentosa is an inherited condition that damages photoreceptor cells, or cells that convert light into electrical signals. As a result, this slowly changes the way the retina reacts to light and makes it progressively more difficult for patients to see. Eventually patients become completely blind. In order to be eligible for the trial, candidates must not yet be completely blind and at least have the ability to see five fingers held up from about a foot and a half away, *New Scientist* reported.

A combination of both optics and genetics, optogenetics involves inserting a light sensitive gene into neuronal cells, cells in the brain, spinal cord, or in this case, the eye. The gene is actually derived from single-celled algae and helps the algae detect light, *New Scientist* reported.

Normally optogenetics is used in animals to help researchers better understand how the brain works as it allows them to illuminate certain areas of the brain and document brain behaviour. However, in this trial, the doctors hope that the gene will convert ganglion cells, which normally communicate information from the eye to the brain, into photoreceptor cells. By recruiting cells to replace those damaged by retinitis pigmentosa, scientists believe they can restore vision, *New Scientist* reported.

At the moment it's unclear just how successful the treatment will be, but experts are hopeful.

"This therapy is novel in its approach," Dr. Anne Negrin, an ophthalmologist in Purchase, New York who is affiliated with Greenwich Hospital and not involved in the trial or research told *Newsweek*. "This is the first time we are trying to give vision to people with degenerative eye disease by recruiting ganglion cells to receive light signals to the brain, rather than trying to repair already damaged photoreceptor cells."

The gene will be injected into one eye and the hope is that it will only boost the detection of red light. Patients will then use special goggles to help them improve their overall vision. According to the organization, Colour Blind Awareness, photoreceptors in our eyes normally perceive three types of light, red, green and blue. However, the new photoreceptors created in this project can only perceive red light. That means the eye will not be able to interpret colour and the patients will only be able to see in black and white.

The primary analysis is meant to test the safety of the treatment over the course of a year.

Retina Australia looks forward to hearing the results of this very interesting research.



IN YOUR STATE - UPCOMING EVENTS

Victoria

Morning Tea

Retina Australia (Vic) would like to invite all members and friends to share in their regular Morning Teas which will be held on 13 March, 8 May, 14 August and 13 November 2018. These social events have proven to be very popular as they provide a means to network with others. Hope to see you there.



FIRST DATE: Tuesday 13 March 2018 between 10:30 am & 12:00 noon

PLACE: Hayden Raysmith Meeting Room, 4th Floor Ross House
247-251 Flinders Lane, Melbourne VIC

RSVP: to 03 9650 5088 by Thursday 8 March 2018 to confirm your attendance.

Queensland

Coffee Morning



Held on the 4th Tuesday of the Month, this is an opportunity for members and friends to get together and share a morning tea in a friendly, congenial atmosphere.

As well as the Community Meeting Room of the Brisbane City Library, there will be some other interesting venues this year, so make sure you keep in touch by phone or email. On 27th March we will be meeting at the BCC library at the top of Queen Street, opposite the casino, from 9:30 until 11:30ish. There is a \$2 charge to cover morning tea. Planning is under way for a trip out to the beautiful Sandgate foreshore in April.

If you would like to be on the email list to be notified of upcoming events, or to make your own suggestion, email retinafriends@gmail.com, comment on the Facebook page or phone Graeme Ferguson on 03849 7752 by the Sunday evening prior.

All welcome, but for catering, please let us know you will be coming through any of the above contacts.

The next dates are: 27th March, 24th April and 22nd May.

South Australia

Luncheon

On the **3rd Tuesday of every month** at the Strathmore Hotel in North Terrace, members of RASA get together for lunch arriving between 12 noon and 12.30.

This event has been going on for many years and not only members attend but sometimes ex-staff members also. The group welcomes new members and encourages them to come regularly.

Further information can be had from Chris Hicks, telephone 0497 491 115.



Western Australia

GWCF - Guildford Heritage Festival - March 2018

The organiser of this event has advised RAWA that due to unforeseen circumstances the Festival scheduled for March 2018 has had to be cancelled. We are thoroughly disappointed as last year's event was a huge success - fingers crossed that the Festival will appear on the calendar for 2019!

Friends of RAWA Social Gathering

We will be meeting again on **Sunday 18 March 2018** for a Friends of RAWA Social Gathering Coffee Catch Up - we would love to see you there!

Venue: The First Edition Café, State Library of WA,
25 Francis Street, Perth

Time: 10:30 to 12:30

Members, family and friends, guide dogs all welcome!

HBF Run For A Reason - The "Visionaries" Team

Once again RAWA is chasing a cure for blindness in the HBF Run For A Reason. Applications are now open to join the "Visionaries" Team.

The run takes place on **Sunday 27 May 2018**

<https://www.registernow.com.au/secure/PersonalDetails.aspx>

Just follow the above link to register to run now! Please spread the word to your family, friends and neighbours to be part of this fantastic fundraising event in Perth. Any questions please contact Tracie Troth, Admin Assistant, RAWA on (08) 9388 1488 or email fundraising@rawa.com.au

ORDER YOUR ENTERTAINMENT BOOK 2018/19



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HOT OFF THE PRESS

WORLD RESEARCH UPDATE FEBRUARY 2018

Summary by Dr Cathy Civil

Real treatments are so close we can almost touch them..



The first patient with the LCA10 form of Leber's Amaurosis has received a drug called QR-110; This is for a phase 1/2 trial Leber's is a common form of inherited retinal blindness in children worldwide. Some early results should be available this year, and the rest in 2019.



With gene therapy trials becoming more common, finding out which genes are causing the IRDs becomes more important. It gives more of those affected by IRDs the option to be included in clinical trials. The use of high-throughput next-generation gene sequencing is speeding up this process enormously. (And our own AIRDR team in Perth are right at the forefront of this work, meaning that those on their register have the best chance possible of being included in trials)



"Luxturna" is a gene therapy for the RPE65 mutation of Leber's Congenital Amaurosis. So far, at least 2/3 of those treated with it have had improved vision, without significant side effects. This is a one-off treatment, and has been approved in the USA. Spark, the company who make it, have negotiated with US insurance companies, so that there are no out of pocket costs to the patient for treatment or follow up. This could be the first of a whole cascade of new accessible gene therapy treatments for different IRDs. the light at the end of the tunnel is getting brighter all the time.....



The transplantation of healthy RPE (Retinal Pigment Epithelial) cells to support diseased photoreceptor cells, is being trialled.

In the past, the transplanted RPE cells have not lasted long, however new trials have found that by boosting the OTX2 gene in the transplanted cells, the cells seem to be more durable.

Subjects with end stage IRD, have had improved vision for up to 13 months with this treatment



Human foetal-derived stem cells (RPCs) were transplanted into the retina of rats affected by RP. There was a significant improvement in vision in five of the eight rats, and increased retinal sensitivity in the other three which lasted for 6 months. However, the improvements had gone by 12 months. Keep trying!



It is thought that one of the reasons photoreceptors die in RP is because glucose doesn't get broken down properly in the photoreceptor cells, so it accumulates in the RPE cells, which causes cell death. The abnormal signalling associated with this process might be a new direction for research.



In X-linked RetinoSchisis, the blisters on the retina (bullous schisis) may be present at birth, or develop soon afterwards. It usually presents with strabismus (flicking eyes). The blisters can progress to retinal detachment.



Biotech company Sirion, a world leader in virus based gene delivery, has signed a two year development agreement with Acucela Inc. for the development of new and better, commercially viable, viral vectors for the delivery of gene therapies for RP.



IRD Patients who have cataract surgery usually do well, but their risk of complications is higher than in those without IRD.



IRDs have replaced diabetes as the leading cause of blindness among working age adults in England and Wales, as diabetic treatment has improved, and IRD treatment has not.



Curcumin (present in turmeric) might help delay the progress of RP.



Gene therapy for Stargardt disease has been difficult so far because the offending gene is too big to fit into the usual viral vector. However, a team in Italy have designed dual viral vectors to overcome this problem. Results are looking promising.



“EURORDIS-Rare Diseases Europe” is a company which seeks to put pressure on European governments and corporations to speed up the development of treatments for rare diseases. The ambition of EURORDIS is to have 3 to 5 times more new rare disease therapies approved per year, 3 to 5 times cheaper than today by 2025



Gene therapy in RP43, a type of RP, has resulted in sustained vision improvement in mice



In California, a protein switch that responds to light, has been developed and attached to healthy retinal ganglion cells in mice with RP. This has given them some basic vision.



Specific retinal patterns have been found in human female Choroideremia carriers which may help an accurate diagnosis, appropriate investigations, and genetic counselling for future pregnancies.



A 5 yr review of the Argus 2 bionic eye shows that it has improved vision, function, and quality of life in patients with advanced RP without significant side effects. It enables perception of light/dark and shapes/objects. The benefit has been sustained for up to 5 yrs.



Genetic analysis of IRDs has shown involvement of at least 260 genes so far, with 70 of them associated just with RP. It is becoming more and more important to identify the remaining dysfunctional IRD genes, as gene therapies become more viable.



BEWARE! With progress in stem cell research, there are “for-profit” stem cell clinics opening up around the world. There have been a number of people with macular degeneration and with IRDs who have paid for treatment with autologous adipose stem cells (stem cells made from their own fat cells). After the treatment, their vision has become worse, and some have developed retinal detachments. **BEWARE!** If a clinic is not doing clinical trials approved by the ethics committee, then you are risking a worse deterioration in your remaining vision.

We really are at the cutting edge of a whole new era in sight. Every month brings new and exciting developments. Watch this space....



Did you know.....?

GC2018 – the Commonwealth Games at the Gold Coast is going for gold in its quest to deliver an inclusive Games and will proudly host the largest integrated Para-Sport program in Commonwealth Games history.

Tickets are still available for competitions such as the lawn bowls and you may even get a chance to see one of Australia’s top vision impaired lawn bowlers, Lynne Seymour with husband Bob as director. When she was first diagnosed Bob convinced her to go from her home town of Amberley, Western Australia to compete in the Queensland championships where she won everything she competed in as well as Bowler of the Tournament.

Since then Lynne has competed internationally as well as nationally but says the main reason she loves it so much is “You can go anywhere in Australia to a bowls club and you’re always welcome. It’s such a friendly sport.”



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Website - <http://www.retinaaustralia.com.au>

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Our Vision

We have a vision we want people to see.

Our Mission

We will assist those affected by vision loss through inherited retinal diseases while working towards the global eradication of such diseases.

Our Goals

To offer support to individuals, families and friends affected by inherited retinal disease.

To be the central source of information related to inherited retinal diseases.

To raise and distribute funds, in association with states and territories, for research into the prevention, diagnosis, treatment and cure of inherited retinal diseases.



Retina Australia needs your support in fighting blindness. You can help by subscribing to our quarterly Retina Australia National News Newsletter, fundraising and or making a donation to Retina Australia or your local state RA Group. Your donation enables Retina Australia to support the great work by Australian researchers; it also provides information and peer support within Australia for those 1 in 3000 affected by inherited retinal disease.

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