# Retina Australia Quarterly

# December 2021

**Contents**

[Chairman’s Report 3](#_Toc88569603)

[Christmas Greetings 5](#_Toc88569604)

[New Board Appointments 6](#_Toc88569605)

[Do It In The Dark 7](#_Toc88569607)

[Retina Australia Webinar 8](#_Toc88569609)

[Thru My Eyes – a New App for IRDs 9](#_Toc88569610)

[Retina Australia 2022 Research Grant Recipients 10](#_Toc88569611)

[Bionic Eye restores sight in world first trial 14](#_Toc88569612)

[The Australian Bionic Eye: The people behind the technology 16](#_Toc88569613)

[Brain Implant Gives Blind Woman Artificial Vision in Scientific First 17](#_Toc88569614)

[Genomic study revealing among diverse populations with inherited retinal disease 21](#_Toc88569615)

[Hot off the Press 24](#_Toc88569616)

[We’ve got Christmas covered 27](#_Toc88569618)

[Get in Touch 28](#_Toc88569619)

[Disclaimer 29](#_Toc88569620)

# Chairman’s Report

Welcome to the Summer edition of Retina Australia Quarterly 2021. I would like to thank all members and friends of Retina Australia, for their continued support and invaluable contributions, particularly during a second extremely challenging year where the coronavirus (COVID-19) pandemic has affected us all so significantly. I look forward to 2022 in the hope that our lives will return to a somewhat normal routine, and we can collectively continue to strive towards “a world without inherited blindness”.

As you may be aware, inherited retinal diseases are a group of conditions that disproportionally affect children and young adults and lead to blindness. In Australia, one in every 1,500 children is born with an inherited retinal disease. The patient burden is extremely high and the impact on family and friends can also be devastating.

Consequently, Retina Australia welcomes the news that Luxturna, a new targeted gene therapy that has the potential to improve vision, and prevent progression towards total blindness, for people with mutations in the RPE65 gene, is now approved by the Therapeutic Goods Administration for use in Australia. I understand that patients with this specific gene malfunction have been treated in Sydney recently and the results have been amazing. I look forward to learning how patients respond to Luxturna in the long term as the treatment is rolled out across the country for everyone who has been diagnosed with RPE65.

The approval of this life-changing therapy, and the knowledge that a significant amount of research is currently being undertaken world-wide for various forms of inherited retinal disease, brings hope to all of us that treatment may be possible for everyone affected at some future time.

Retina Australia held its 37th Annual General Meeting of members via Zoom videoconferencing on 9 October 2021. Members were provided with the Chairman’s Annual Report, which summarised the previous year’s activities of Retina Australia, as well as the 2020-2021 audited financial statements. The election of Board members was also endorsed.

I am pleased to be able to report that the composition of the Retina Australia Board for 2021-2022 is:

**Chairman:** Leighton Boyd

**Deputy Chairman:** Peter Maas

**Treasurer:** Joshua Ginpil

**Company Secretary:** Rosemary Boyd

**Board Members:** Mary-Anne Carmody, Jane Cherry, Jessica Coleman, Lindsay DaCosta, Julie Demarte, and Heather Mack.

Whilst welcoming new directors, Jane Cherry, Jessica Coleman and Lindsay DaCosta, to the Board, I would like to thank Jeremy D’Souza, Robert Craft and Melanie Chatfield for their contribution in recent years. Melanie has recently been appointed as an ambassador for Retina Australia and I look forward to her continued contribution for many years to come.

Since the AGM, the Board has met to consider the future strategic direction for Retina Australia and plans are well underway for 2022 and beyond. It is truly exciting to be involved in this new era for Retina Australia.

It is very satisfying to report that the Retina Australia Board have allocated $120,000 for the 2022 research grants pool. The ability to pledge this amount for research once again, has been made possible by the very generous donations, of members, friends, and supporters across the country, as well as from the much-appreciated funds gained from our first ever “Giving Day” which was held on 21 June 2021.

On your behalf, I would like to congratulate the successful grant recipients for 2022: Associate Professor Penelope Allen, Dr Rabab Rashwan, Associate Professor Guei-Sheung (Rick) Liu and their research teams. Specific details of their proposed research are included in this newsletter.

This year we again received many credible, and worthy, applications and I thank the researchers who put forward their unique proposals and wish them well for their future investigations. It is unfortunate that we do not have the capacity to fund all projects, however we are certainly interested in continuing to work closely with researchers investigating inherited retinal disease wherever possible. To this end, since the inception of the research grants program, Retina Australia has provided over $5.8million towards research.

Following an eventful year in which our activities have been curtailed by COVID-19, the Board and staff have continued to reach out to members and friends through the use of Zoom meetings, Webinars, and social media. We also implemented some new fundraising projects including launching the “Do it in the Dark” activities, holding our first “Giving Day” and hosting some excellent Trivia Nights online. We are looking forward to expanding our fundraising projects in 2022.

I am also hopeful that we will have new vigour in pursuing our mission of “assisting those affected by vision loss from inherited retinal diseases by providing support, information and funding for research”. Above all, the Board and staff will continue to raise awareness of inherited retinal disease in the community across Australia. I look forward to the next year with enthusiasm.

Thank you for your continued membership and support,

Leighton Boyd

Chairman, Retina Australia

# Christmas Greetings

The Board and staff of Retina Australia would like to wish all our members and supporters a very Happy Christmas and New Year and thank you for all your support during 2021. Our office will be closed from 3pm AEDT on Tuesday 21st December and will reopen at 9am AEDT on Tuesday 11th January 2022.

# New Board Appointments

We are delighted to welcome the following new appointees to the Retina Australia Board:

Jane Cherry  
Diagnosed with Retinitis Pigmentosa at 31, Jane has been actively involved in raising awareness of people living with Inherited Retinal Disease and raising funds for Retina Australia to support their medical research into early detection, treatment and cures. Jane is a member of the Retina International Youth Council and joined the planning committee to coordinate the virtual International Youth Conference held in August 2021. Jane is currently working to further develop her skills as a patient advocate through Retina International’s Education Programme.  
  
Jess Coleman

Jess began her involvement with Retina Australia through her non-profit platform – Running Against RP – which she launched in 2016 with all funds raised going directly to Retina Australia. Jess’ passion for raising awareness, support, education, & funding for inherited retinal diseases, particularly retinitis pigmentosa, was birthed from her personal experience of growing up with a father whose vision was gradually diminishing due to retinitis pigmentosa.  
  
Lindsay Da Costa

Lindsay is a senior human resources professional with extensive experience working in large, globally listed organisations across multiple industries. Lindsay works closely with senior commercial leaders and executive leadership teams, providing strategic HR advice and solutions on employment relations, developing leadership capability, and organisational design. Lindsay has a strong interest in community and culture and has contributed to the mission of various not-for-profit sector organisations and committees.

# Do It In The Dark

## Save the date for 2022!

After a very successful inaugural year in 2021, Do It In The Dark is coming back in 2022! During the months of March and April, you will have the opportunity of doing one of your favourite activities and raising money for Retina Australia at the same time!

More information will be available in early 2022, but now is the time to start thinking about what you can do to help raise awareness and improve understanding of inherited retinal disease, as well as raising much needed funds. For more information, contact our Fundraising and Marketing Manager Faik Demir at faik.demir@retinaaustralia.com.au or on 03-9650 5088.

# Retina Australia Webinar

**Clinical genetics services helping with genetic information, counselling and access to clinical therapy, trials and research in the IRDs.**

Following on directly after this year’s Annual General Meeting on Saturday 9th October, we were delighted to welcome Professor Robyn Jamieson, Dr Alan Ma and Ms Laura Wedd from the Department of Clinical Genetics, Western Sydney Genetics Program, Sydney Children’s Hospitals Network and Westmead Hospital, who spoke about referral pathways and possible outcomes of genetic testing for patients and families with IRDs. The patient journey was discussed, as it relates to the genetic information, research studies to clarify genetic variants, available gene therapy, clinical trials and novel pharmaceutical and genetic therapy approaches.

The session was recorded, and is available to watch for free on the Retina Australia YouTube channel, or at this link: <https://youtu.be/OyFnHBvYUMY>. If you subscribe to our YouTube channel, you will automatically be notified when a new video is uploaded, so you will never miss out!

We would like to thank Robyn, Alan and Laura for this excellent session, as well as all the presenters in our 2021 webinar program. We are planning further sessions in 2022, and as always, we are keen to hear from you with any suggestions for topics or presenters of interest. Just drop us a line at [info@retinaustralia.com.au](mailto:info@retinaustralia.com.au) or call us on 03-9650 5088.

# Thru My Eyes – a New App for IRDs

Have you ever wished you could show people what you experience visually as someone living with an IRD? Thru My Eyes is a free iOS app, developed by ProQR Therapeutics as an interactive tool to support inherited retinal disease (IRD) awareness. While the app provides an artist’s impression using different filters, the developers have calibrated visual acuity (sharpness of vision) to validated scales to be as realistic as possible.

The app is still in its early stages and its development is ongoing. For now it provides simulations of Leber’s congenital amaurosis (LCA) and retinitis pigmentosa (RP), with more conditions to be added in the future. For more information and to download it, visit: <https://www.thrumyeyes.app/>

# Retina Australia 2022 Research Grant Recipients

We are pleased to announce that the 2022 recipients of the Retina Australia medical research grants are Associate Professor Penny Allen (Centre for Eye Research Australia, Melbourne), Dr Rabab Rashwan (Lions Eye Institute, Perth), Associate Professor Guei-Sheung (Rick) Liu (Centre for Eye Research Australia, Melbourne). These grants have a combined total value of $120,000.

**Improving real-world mobility and assessing long-term safety outcomes with a retinal prosthesis (“Bionic Eye”)**

**Lead:** Associate Professor Penelope Allen -Centre for Eye Research Australia

**Research Team**: Dr Janine Walker, Dr Matthew Petoe, Professor Nick Barnes, Dr Carla Abbott

**Project Description:**

Participants with end-stage retinitis pigmentosa (inherited retinal disease) were implanted with a second-generation Australian bionic eye (retinal prosthesis) in 2018 as part of a clinical trial. The clinical trial finished in December 2020 and showed the device has an unsurpassed safety profile, with no serious adverse events and good stability over 3 years. Furthermore, laboratory-based measures showed that using an intensity-based method of visual processing (turning a stream of images from a camera into a series of flashing lights for the recipient to interpret) improves orientation and mobility, functional vision, and activities of daily living for recipients.

However, the team has now designed a promising cutting-edge visual processing strategy based on advanced depth processing algorithms with potential for further breakthrough visual improvements, especially in a real-world setting with varying levels of colour contrast. They have an extraordinary opportunity to measure visual outcomes with this advanced vision processing method in an extension study with three participants already implanted with a bionic eye in both a controlled laboratory environment (obstacle avoidance task) as well as in a real-world environment (novel outdoor streetscape protocol). The outcomes are critical to optimising the visual processing strategy and to establish the real-world efficacy of the unique Australian bionic eye. The team will also monitor the long-term safety of the device out to 4 years after implantation as a secondary outcome to check the device does not impact adversely on eye health and to check the long-term device functionality.

**Neuroprotective effect of SAHA in Retinitis Pigmentosa. Do time and frequency matter?**

**Lead:** Dr Rabab Rashwan - Lions Eye Institute Perth

**Research Team**: Miss Annie Miller, Dr Livia Carvalho

**Project Description**:

Retinitis pigmentosa (RP) is a genetic, blinding retinal disorder that affects approximately 2 million people worldwide. RP involves the death of the photoreceptor cells (rods and cones) that turn light signals into vision. RP begins with the death of rod photoreceptors, causing night blindness. As the disease progresses, the cone photoreceptors also degenerate, which causes the loss of central, fine and daylight vision leading to complete blindness in some people. RP has been proven to be caused by mutations in over 90 genes, targeting all causative genes being a challenge for treatment development.

For this reason, this project will focus on the development of a broad treatment approach that can be used on RP patients regardless of the underlying mutation causing the disease. Other studies showed that a molecule called histone deacetylase (HDAC) to be increased in the retina of many RP mouse models. In the present study, the team will investigate the effect of an FDA-approved HDAC inhibitor (SAHA) to protect photoreceptor cells, especially cones, from degeneration in two mouse models of RP with two different mutations. The intraocular administration of SAHA will be for the first time tested at the early and late stages of the disease to achieve the best results on photoreceptor survival and function. Additionally, multiple treatments and the long-term safety profile of SAHA will be examined, which is essential for clinical translation. The ultimate impact of this study is the development of gene-independent treatment strategies that preserve visual acuity and daylight vision in RP patients and several different types of vision loss, benefiting a more comprehensive range of patients.

**RNA base editing strategies as potential therapeutic of inherited retinal dystrophies**

**Lead:** Associate Professor Guei-Sheung (Rick) Liu - Centre for Eye Research Australia

**Research Team**: Associate Professor Bang Bui, Dr Thomas Edwards

**Project Description:**

Inherited retinal degenerations (IRDs) are significant contributors to global vision impairment and blindness, with no cure to date. Gene therapy by supplementation is restricted to certain forms of IRDs and therefore alternative therapeutic approaches are required. Specific mutations underlying these conditions have been identified, although efficient strategies to target these mutations have not been developed. Recently, CRISPR-based editing has allowed targeted modification of single bases using adenosine and cytosine deaminases without causing sequence breaks. Compact CRISPR/Cas enzymes that target RNA have also been demonstrated to perform efficient and specific base editing. Their compact size allows delivery using a single viral vector which is the delivery method of choice. These enzymes (Cas13X) do not require a flanking sequence which permits targeting of any locus within the transcriptome, while ensuring safety as the genome is not altered.

In addition, a novel CRISPR-inspired RNA targeting system (CIRTS) has been developed, which is smaller than all known CRISPR/Cas systems and able to perform efficient base editing. The team will compare these two established compact RNA base editors to investigate their potential in correcting point mutations present in the retina efficiently and specifically. Successful demonstration of ocular RNA base editing using these systems would provide a novel therapeutic approach against IRDs, one that is efficient, safe, cost-effective and provides long-term treatment.

# Bionic Eye restores sight in world first trial

**Source**: Georgia Linnell, 9now.nine.com.au

**Date:** September 2021

A whole new world has appeared for four [vision-impaired](https://www.9news.com.au/disability) Australians, with the creation of a bionic eye.

For the past decade, Associate Professor Penny Allen and her team at the Centre for Eye Research Australia have developed technology that can partially restore sight lost to a genetic condition of the retina.

Over the past two years, four Australians have been a part of a world-first trial of the [technology](https://www.9news.com.au/technology), including great-grandmother Colleen Knowles.

"Where we used to live, there were a lot of trees in the nature strip. I lived there for eight years and I never knew there were trees," Ms Knowles told A Current Affair reporter Sam Cucchiara.

"Once I put the device on, I was able to appreciate the landscape that was there and then I was getting the trees on the other side of the road."

Ms Knowles vividly remembers her "switch on" day.

"Probably the first seven or eight times, they were saying, 'Can you see anything?'" she said.

"'Nope, nope, nope'. Then all of a sudden, it's 'oh'.

"My family were in another room watching it on [CCTV](https://www.9news.com.au/cctv), so it was exciting for them, because I was sitting there thinking it wouldn't work and it did."

It was a similar experience for grandfather Mark Boyd.

"I'm 66 now. I first got diagnosed when I was about seven and it's gradually got worse over those 60 years," Mr Boyd said.

He's now able to enjoy trips to the theatre.

"I would stand in the foyer and hear people around me, but I wouldn't be able to know - except by sound - how close," he said.

"But with the device, you can scan around, and you know someone's there."

Professor Allen has dedicated her life to helping others see.

"It's very satisfying. The patients are able to do tasks, increase their social interaction by using the device and that's what drives us all," she said.

"That satisfaction leads to an appetite of more development, more work and making it better."

**How it works**

The cutting-edge surgical technique involves placing an implant on the skull, which is attached via a lead to electrodes within the back of the eye.

A pair of glasses containing two cameras captures footage which is then sent to a unit to convert the video into an electrical impulse.

The impulse is passed to the skull implant, and then back onto the device in the eye to stimulate existing tissue and help the wearer to see.

"It is history-making," Ms Knowles said.

"It's going to change the lives of vision-impaired people."

The device is being co-developed with Bionic Vision Technologies, which hopes to take it on the market by 2025.

"It's a revolutionary Australian-first that will give blind people the ability to regain their vision," Bionic Vision Technologies CEO Dr Ash Attia said.

"It's the next frontier when it comes to digital medicine and medical technology."

Dr Attia has pointed out the similarities between the bionic eye technology and another world-first technology also developed at Melbourne's Royal Victorian Eye and Ear Hospital.

"What (the) cochlear (implant) has done for the profoundly deaf, is exactly what this system is going to do for the blind," Dr Attia said.

"Eyesight is precious. To lose it is debilitating and to restore it is nothing short of miraculous and life-changing."

Before being made widely available, the bionic eye will undergo a third clinical study in early 2023.

This story was featured on Channel 9’s A Current Affair in September. You can view the story at [this link](https://9now.nine.com.au/a-current-affair/bionic-eye-device-restores-eyesight-for-vision-impaired-australians-during-world-first-trial/8d5a07d1-e725-4a3e-bed3-54f6046491d5)

# The Australian Bionic Eye: The people behind the technology

On 4th November, the Convergence Science Network hosted a webinar featuring some of the leading researchers in the bionic eye project, including Associate Professor Penny Allen from CERA (and one of our 2022 research grant recipients). A recording of this free event is available at this link: <https://youtu.be/la8m5T1kQoo>

# Brain Implant Gives Blind Woman Artificial Vision in Scientific First

**Source:** Carly Cassella, sciencealert.com

**Date:** 28 October 2021

A 'visual prosthesis' implanted directly into the brain has allowed a blind woman to perceive two-dimensional shapes and letters for the first time in 16 years.

The US researchers behind this phenomenal advance in optical prostheses have recently published the results of their experiments, presenting findings that could help revolutionize the way we help those without sight see again.

At age 42, Berna Gomez developed [toxic optic neuropathy](https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3116542/), a deleterious medical condition that rapidly destroyed the optic nerves connecting her eyes to her brain.

In just a few days, the faces of Gomez' two children and her husband had faded into darkness, and her career as a science teacher had come to an unexpected end.

Then, in 2018, at age 57, Gomez made a brave decision. She volunteered to be the very first person to have a tiny electrode with a hundred microneedles implanted into the visual region of her brain. The prototype would be no larger than a penny, roughly 4 mm by 4 mm, and it would be taken out again after six months.

Unlike retinal implants, which are being explored as means of [artificially using light to stimulate the nerves leaving the retina](https://www.sciencealert.com/scientists-have-created-an-artificial-retina-implant-that-could-restore-vision-to-millions), this particular device, known as the Moran|Cortivis Prosthesis, bypasses the eye and optic nerve completely and goes straight to the source of visual perception.

After undergoing neurosurgery to implant the device in Spain, Gomez spent the next six months going into the lab every day for four hours to undergo tests and training with the new prosthesis.

The first two months were largely spent getting Gomez to differentiate between the spontaneous pinpricks of light she still occasionally sees in her mind, and the spots of light that were induced by direct stimulation of her prosthesis.

Once she could do this, researchers could start presenting her with actual visual challenges.

When an electrode in her prosthesis was stimulated, Gomez reported 'seeing' a prick of light, known as a phosphene. Depending on the strength of the stimulation, the spot of light could be brighter or more faded, a white color or more of a sepia tone.

When more than two electrodes were simultaneously stimulated, Gomez found it easier to perceive the spots of light. Some stimulation patterns looked like closely spaced dots, while others were more like horizontal lines.

"I can see something!" Gomez [exclaimed](https://healthcare.utah.edu/moran/news/2021/10/artificial-vision.php) upon glimpsing a white line in her brain in 2018.

Vertical lines were the hardest for researchers to induce, but by the end of training Gomez was able to correctly discriminate between horizontal and vertical patterns with an accuracy of 100 percent.

"Furthermore, the subject reported that the percepts had more elongated shapes when we increased the distance between the stimulating electrodes," the authors [write in their paper](https://www.jci.org/articles/view/151331).

"This suggests that the phosphene's size and appearance is not only a function of the number of electrodes being stimulated, but also of their spatial distribution… "

Given these promising results, the very last month of the experiment was used to investigate whether Gomez could 'see' letters with her prosthesis.

When up to 16 electrodes were simultaneously stimulated in different patterns, Gomez could reliably identify some letters like I, L, C, V and O.  She could even differentiate between an uppercase O and a lowercase o.

The patterns of stimulation needed for the rest of the alphabet are still unknown, but the findings suggest the way we stimulate neurons with electrodes in the brain can create two-dimensional images.

The last part of the experiment involved Gomez wearing special glasses that were embedded with a miniature video camera. This camera scanned objects in front of her and then stimulated different combinations of electrodes in her brain via the prosthesis, thereby creating simple visual images.

The glasses ultimately allowed Gomez to discriminate between the contrasting borders of black and white bars on cardboard. She could even find the location of a large white square on either the left or right half of a computer screen. The more Gomez practiced, the faster she got.

The results are encouraging, but they only exist for a single subject over the course of six months. Before this prototype becomes available for clinical use it will need to be tested among many more patients for much longer periods of time.

Other studies have implanted the same microelectrode arrays, known as Utah Electrode Arrays, into other parts of the brain to help control artificial limbs, so we know they're safe in at least the short term. But it's still early days for the tech, which risks a steady [drop in functionality](https://iopscience.iop.org/article/10.1088/1741-2552/abc025) over just a few months of operation.

While engineers beef up the reliability of the devices, we still need to know exactly how to program the software that interprets the visual input.

Last year, researchers at Baylor College of Medicine in Houston [inserted a similar device](https://www.sciencealert.com/a-new-brain-implant-lets-blind-people-see-letters) into a deeper part of the visual cortex. Among five study participants, three of whom were sighted and two of whom were blind, the team found the device helped blind people trace the shapes of simple letters like W, S, and Z.

In Gomez's case, there was no evidence of the device triggering neural death, epileptic seizures, or other negative side effects, which is a good sign, and suggests microstimulation can be safely used to restore functional vision, even among those who have suffered irreversible damage to their retinas or optic nerves.

“One goal of this research is to give a blind person more mobility”, says bioengineer Richard Normann from the University of Utah.

"It could allow them to identify a person, doorways, or cars easily. It could increase independence and safety. That's what we're working toward."

Right now, it seems only a very rudimentary form of sight can be returned with visual prostheses, but the more we study the brain and these devices among blind and sighted people, the better we will get at figuring out how certain patterns of stimulation can reproduce more complex visual images.

Perhaps one day, other patients in the future will be able to trace the whole alphabet with this prosthesis because of what Gomez has done. Four more patients are already lined up to try out the device.

"I know I am blind, that I will always be blind," Gomez [said](https://healthcare.utah.edu/moran/news/2021/10/artificial-vision.php) in a statement a few years ago. But I felt like I could do something to help people in the future. I still feel that way."

Gomez's name is listed as co-author on the paper for all her insight and hard work.

The study was published in the [Journal of Clinical Investigation](https://www.jci.org/articles/view/151331).

# Genomic study revealing among diverse populations with inherited retinal disease

**Date:**19October, 2021

**Source:**University of California - San Diego

An international team of researchers, led by scientists at University of California San Diego and Shiley Eye Institute at UC San Diego Health, has broadened and deepened understanding of how inherited retinal dystrophies (IRDs) affect different populations of people and, in the process, have identified new gene variants that may cause the diseases.

The findings were published in the October 18, 2021 issue of PLOS Genetics.

IRDs are a group of diseases, from retinitis pigmentosa to choroideremia, that result in progressive vision loss, even blindness. Each IRD is caused by at least one gene mutation, though mutations in the same gene may lead to different IRD diagnoses.

IRDs are rare, but they affect individuals of all ages, progressing at different rates, even within families afflicted with the same disease. Specific diagnosis depends on finding the genetic causative mutations.

The U.S. Food and Drug Administration has approved gene therapy for treating one form of IRD involving the gene RPE65, but for other IRDs caused by mutations in more than 280 different genes, there are no cures or treatments proven to slow disease progression.

The researchers conducted whole-genome sequences (WGS) of 409 persons from 108 unrelated family lineages, each with a previously diagnosed IRD. WGS is a process of determining the entirety, or near-entirety, of the DNA sequence of an individual. It provides a comprehensive portrait of the person's entire genome, including mutations and variants, which can be used for broad comparative purposes.

Study participants were recruited from three different geographic regions: Mexico, Pakistan and European Americans living in the United States. Genomic analyses were conducted from blood samples taken from all participants, which revealed causative variants in 62 of the 108 lineages. A total of 94 gene variants were found in the 62 families: 52 variants had previously been identified as causative and 42 had not. Surprisingly, more than half of the new variants were not listed in the Genome Aggregation Database, an international compilation of genomic data.

Overall, causative variants were detected in 63 percent of Mexican participants, 60 percent of Pakistani, and 48 percent of European American.

The study also identified a large proportion of new IRD causative mutations specific to the populations studied and revealed the types of mutations contributing to inherited retinal dystrophies. Approximately 13 percent of the families displayed atypical or unexpected changes in the genome. Five of the family lineages had mutations in more than one gene in all affected individuals; one family carried mutations in different genes in different affected members and a de novo mutation was found in one patient that was not present in both parents.

An additional 8 percent of families had large changes in the structure of their genome causing the inherited retinal disease and the initial clinical diagnosis in four families was re-classified based on their genotype.

The authors said the new findings boost understanding of the distribution of IRD causative mutations in these three diverse populations, which will further assist our understanding of disease variation and presentation. That, in turn, will help design more efficient genetic testing strategies and therapies applicable to global populations.

The research team was led by Radha Ayyagari, PhD, professor of ophthalmology and pathology, and Kelly A. Frazer, PhD, professor of paediatrics and director of the Institute for Genomic Medicine, both at UC San Diego School of Medicine; and S. Amer Riazuddin, PhD, associate professor of ophthalmology at John Hopkins University, in collaboration with institutions in India, Mexico, Canada, Brazil, Pakistan and the United States.

# Hot off the Press

## World Research Summary by Dr Catherine Civil

Here are some more snippets for you which I have tried to make readable. With many thanks again to Christina Fasser who is a past president of Retina International and who compiles a list each month of the latest research from the leading journals and sends it on to us.

* The golden oldie drug methotrexate which is still often used for rheumatoid arthritis, has been given orphan drug status in the USA with a view to development as a potential treatment for RP.
* Did you know that there are at least 24 IRD (Inherited Retinal Disease) clinical trials ongoing in the USA alone, and many, many more around the world? There is a move to include children now in the trials as they can receive extra benefit by being treated early in their disease. There are several kids who have done well with Luxterna for LCA for example.
* Passing small electrical currents through the cornea sounds awful as a treatment for RP but seemingly it is helpful and safe! Further trials are continuing.
* Presbyopia drops are in phase 2 and 3 trials and being looked at by several companies. This is not IRD related, but I had to include it as presbyopia is the reason so many of us need glasses as we get older. Interesting!
* A Second-Generation (44-Channel) Suprachoroidal Bionic eye helped the 3 subjects who wore it for 2 years. They reckoned that completing visual tasks was easier, but that the vision related quality of life did not change. I expect that this will improve with the newer models now available.
* Retinal implants with intra-orbital parts (e.g. connecting cables) unfortunately reduce some eye movements which isn’t great cosmetically, but that doesn’t stop this bionic eye from working
* There is a variety of avenues being pursued for AMD (Age-related Macular Degeneration). Current clinical trials for dry AMD are investigating the complement cascade, vitamin A metabolism, metformin, and tetracycline; whereas clinical trials for wet AMD are aiming to decrease treatment burden through new port delivery systems, increasing drug half-life, and targeting new sites of the VEGF cascade. Stem cell and gene therapy are also being evaluated for treatment of both wet and dry AMD.
* Optogenetic therapy involves creating artificial photoreceptors in the retina: The Chrimson R gene is injected into the eye via a viral vector, where it turns retinal ganglion nerve cells into photoreceptors; Special goggles are worn which project light onto the retina of the user, and then eye training is given. Watch this space.
* A chest mounted camera which triggers collision warnings has been helpful in those with severe visual impairment and might save a few bruises and accidents.
* And even more exciting is a new generation white cane. This has a colour 3D camera, a sensor, and its own on-board computer. When paired with a building’s architectural drawing, it can accurately guide a user to a desired location with sensory and auditory cues (presumably vibrations and beeps), while also helping the user avoid obstacles like boxes, furniture, and overhangs.

There are still a few kinks to be worked out, but it is looking very promising. The cane can be used in both robotic and plain old white cane modes.

* An antioxidant gene has been found which could perhaps slow photoreceptor degeneration and so slow down visual loss.
* “Photobiomodulation”, or PBM has been trialled using 670nm LED light treatment to the eyes. It looked like this helped visual acuity in dry AMD…. without any adverse events…… further research will be interesting.
* Training helped those with scotomas (black spots in their vision) find their way around, with the help of a device on the finger which gave audio and visual feedback.
* A deep learning platform has been developed which can diagnose 39 different classes of retinal disease by comparing images with a massive database. It seems to be very accurate and should help earlier diagnosis and treatment for many people with IRDs in the future.
* Transplantation of Human Embryonic Stem Cell-Derived Retinal Tissue under the retina in animals and people has been successful but sadly (and understandably) can’t be used routinely due to ethical concerns and limited supply.

It is so good to see the pace of research escalating all the time, especially as treatments become more viable. The new developments astound me each month.

So that’s all for now folks!

All my best wishes to you all until next time.

Cathy

# We’ve got Christmas covered

With Christmas fast approaching and Australia Post busier than ever, here's a great idea for getting all your gifts sorted early.

Our friends at Goodwill Wine are offering a customised "Choose Your Own Six Gift Packs" service where you can save $45 on shipping by having six beautifully packaged Gift Packs delivered to your door in one big box.

As always they'll donate 50% of the profit from your order to us... it's a gift that keeps on giving.

Order yours here: <https://goodwillwine.com.au/pages/retina-australia>

# Get in Touch

Volunteers fighting blindness

Retina Australia Contact Details:

* Enquiry Line – 1800 999 870
* Office – (03) 9650 5088 between 9:30am & 3:00pm Tuesday or Thursday.
* Email – [info@retinaaustralia.com.au](mailto:info@retinaaustralia.com.au)
* Website - <http://www.retinaaustralia.com.au>
* Facebook – <https://www.facebook.com/RetinaAustralia/>
* LinkedIn – <https://www.linkedin.com/company/retinaaustralia>

Retina Australia Staff:

* Faik Demir, Fundraising and Marketing Manager
* Sally Turnbull, Administrative Officer
* Junxia Xu, Finance Officer

Retina Australia Board Members – 2020/2021

* CHAIRMAN: Leighton Boyd
* DEPUTY CHAIRMAN: Jeremy D’Souza
* COMPANY SECRETARY: Rosemary Boyd
* DIRECTORS: Mary-Anne Carmody, Melanie Chatfield, Robert Craft, Julie Demarte, Joshua Ginpil, Peter Maas, Heather Mack

# Disclaimer

Views expressed in this publication are not necessarily those of Retina Australia. Retina Australia accepts no responsibility and disclaims all liability for such views as well as for any information contained in articles and summaries of research reports, including but not restricted to, the use of pharmaceuticals or other products, items of equipment or practices. Retina Australia strongly suggests that persons seek advice from their medical practitioners before adopting any changed procedures, practices or products.