

RETINA REPORTER

Retina Australia's Bi-Annual Newsletter

JUNE 2023

We have a vision. A world without inherited blindness.

There are no cures. There are very few treatments.
Help us change that.

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New website launched

Retina Australia launched its new [website](#) in April. It delivers you a rebranded information and communications platform on inherited retinal disease (IRD), upgraded for functionality and designed to comply with global accessibility standards.

You can now review summaries of our research grants for the first time, find out about who we are including our Team, Board, Strategic Plan and principles of operation, easily access our information resources and past Retina Reporter newsletters, register directly for upcoming webinars and watch previous webinars online, automatically renew or apply for membership, and also find out about the many ways you can help us. Details of our revised Research Grants Program and how to apply are also available.

A message from the CEO



Over the past few months, I have had the pleasure of meeting some of our wonderful long time members, including Warren Colledge, Keith Duhigg and their lovely wives, Rhonda and Teresa respectively. It was very special to hear about their involvement with Retina Australia over the years and about their continued interest in research progress and the organisation's developments. Thank you so much for your continued support to Retina Australia.

I have also had the opportunity to meet many of our researchers, including both our 2023 Research Grant awardees, Dr Anai Gonzalez-Cordero from the Children's Medical Research Institute, Sydney, and Dr Adrian Cioanca from the John Curtin School of Medical Research. This has provided a small glimpse into the innovative worlds of stem cell and gene therapy research. It's a truly exciting time with new technologies and research developments progressing rapidly.

Medical research however, can often be complicated and difficult to comprehend for those of us who are not researchers. Our new website now offers summaries of our research grant projects and Retina Australia will continue to work hard to present research, information and associated communications in clear and simple dialogue for easier understanding in an accessible way.

I hope you find this edition of the Retina Reporter full of interesting new research developments, particularly those resulting from our completed 2022 research grants, providing hope and precursors to possible new therapies and treatments in the future.

Julia Hall

Chief Executive Officer
Retina Australia

Research Grants Program Applications close 30 June

Retina Australia's Research Grants Program is currently open for applications to fund medical research projects focused on inherited retinal disease in 2024. Grants are open to all Australian researchers. Applications close on 30 June 2023. Details of Funding Guidelines and Conditions along with the Grant Application Form can be found on our website, www.retinaaustralia.com.au.



2023 Annual Appeal



2023 ANNUAL APPEAL

Support life-changing research into inherited retinal diseases

[DONATE NOW](#)

Retina Australia's 2023 Annual Appeal is now underway for the month of June and we need your support.

This is our one major fundraising event of the year and is critically important to us in providing funding for our research grants program and our information and support services. We can only do this with your generosity.

Your donation will support :

- Future funding of the Retina Australia Research Grants Program, which drives life-changing research into inherited retinal disease, with a focus on promoting early detection, discovering preventions, advancing treatments, with the ultimate goal to progress towards unlocking cures into inherited retinal disease
- The provision of information and support to those affected by inherited retinal disease, including access to essential resources, hosting research webinars on the latest developments, and providing access to support groups
- The raising of awareness and understanding about inherited retinal disease to the general community

Join us in our Mission to support Australians affected by inherited retinal disease and together we can strive for a World Without Inherited Blindness.

With many thanks,

Leighton Boyd

Chair
Retina Australia



Retina Australia Research Grant Impact Reports

We are delighted to provide updates on previous grant projects that were awarded by Retina Australia in 2022 and are now completed and reported on.

RNA base editing strategies as potential therapeutic of inherited retinal dystrophies

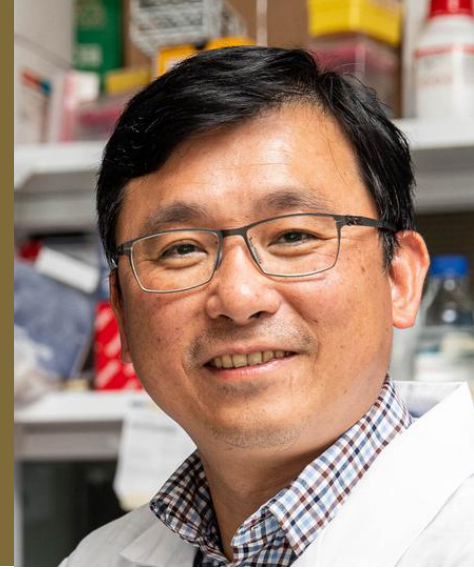
Chief Investigator

Associate Professor Guei-Sheung (Rick) Liu, Centre for Eye Research Australia, Melbourne (CERA)

Co-investigators

Professor Bang Bui, University of Melbourne
Dr Thomas Edward, CERA

Grant awarded - \$40,000 (2022)



Project Aim

This study aimed to use a new genetic technology called "RNA gene editing" to correct genetic mutations that cause inherited retinal diseases, which are a leading cause of blindness in working-age adults in Australia. The goal was to also develop a more effective and generalisable treatment approach, which could have a significant impact on the management of inherited retinal diseases and other genetic diseases.

Project Results and Impact

In this project, the research team designed and compared two newly developed RNA base editors (CRISPR-dCas13 and CRISPR-inspired RNA targeting system) on the editing of the RPE65 gene, the common single-base mutation found in Leber Congenital Amaurosis. By using retinal pigment epithelium cells which are the cells that support and nourish the light sensitive cells in the retina, their results showed that the CRISPR-dCas13 RNA base editor could effectively correct the single-base mutation on the RPE65 gene and restore the protein expression terminated by the mutation.

The team are now expanding this study to investigate the feasibility of using this RNA base editor to correct mutations in other inherited retinal diseases, such as Usher Syndrome type 2A, Usher Syndrome type 1F, and X-linked Retinoschisis. Moreover, they are also ongoing to validate the efficacy of RNA base editing in clinically relevant animal models to demonstrate the efficacy or preservation of the retinal function from mutation-caused retinal degeneration. The outcome of their study would be a significant step toward a viable treatment for inherited retinal degeneration.

Published peer-reviewed journal article

Kumar S and Liu GS. Recent advances in RNA-targeting therapy for neurological diseases (2023). Neural Regeneration Research, accepted on 21/12/2022



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

Chief Investigator
Associate Professor Penelope Allen, CERA

Co-investigators
Dr Janine Walker, CSIRO; Dr Matthew Petoe, The Bionics Institute;
Professor Nick Barnes, Australian National University, Canberra;
Dr Carla Abbott, CERA

Grant awarded – \$39,862 (2022)

Project Aim

This project aimed to measure visual outcomes of people implanted with a bionic eye using a new method called Local Background Enclosure (LBE) compared to an older method called Lanczos2. Testing would be compared both in a lab and in the real world (on a street). The study also aimed to test the safety and effectiveness of the bionic eye 4 years after it was implanted on the project participants.

Project Results and Impact

The project investigated the effectiveness of a new visual processing method, called Local Background Enclosure (LBE), in helping people who had been implanted with bionic eyes to navigate obstacles in both a controlled laboratory environment and a real-world setting. The results showed that LBE performed better than the traditional intensity-based method, called Lanczos2, for detecting low-contrast obstacles, and was also able to detect high-contrast obstacles as well. LBE also outperformed Lanczos2 for detecting objects such as mannequins, overhanging boxes, and large bins, while

Lanczos2 was better for detecting ground-based boxes.

In the real-world environment, the bionic eye was also able to detect objects more effectively than a cane or relying on the user’s own body senses. Participants reported that the device helped them navigate their environment with greater confidence and awareness of their surroundings.

The study also monitored the safety and functionality of the bionic eye over a 4-year period, and found that it remained stable and reliable for long-term use, with no adverse impact on participants.

These findings are important for optimising the visual processing strategy and establishing the real-world effectiveness of the Australian bionic eye. The depth-based vision processing method could potentially be incorporated into the bionic eye system to improve navigation. Overall, the study demonstrated the potential of the bionic eye to help people with visual impairments navigate their environment safely and effectively.

Published peer-reviewed journal article

Lauren Moussallem; Lisa Lombardi; Matthew A. Petoe; Rui Jin; Maria Kolic; Elizabeth K. Baglin; Carla J. Abbott; Janine G. Walker; Nick Barnes; Penelope J. Allen, Navigational outcomes with a depth-based vision processing method in a second generation suprachoroidal retinal prosthesis, ARVO, April 23-27 2023, New Orleans, USA.

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

Chief Investigator

Dr Rabab Rashwan, Lions Eye Institute, Perth

Co-investigators

Miss Annie Miller, Lions Eye Institute, Perth

Dr Livia Carvalho, Lions Eye Institute, Perth

Grant awarded – \$40,000 (2022)



Project Aim

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. This project focused on the development of a broad treatment approach that can be used on RP patients by investigating the effect of an FDA-approved HDAC inhibitor (SAHA) to protect photoreceptor cells, especially cones, from degeneration in two mouse models of RP.

The intraocular administration (via injection to the eye) of SAHA to the mice, was tested for the first time at the early and late stages of the disease to achieve the best results on photoreceptor survival and function. The ultimate aim 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.

Project Results and Impact

This study was important in assessing whether the drug SAHA could protect the cells in the eye that help us see (called photoreceptor cells) in mice with RP. They found that the drug did rescue some of the central retina cells and may have a net protective effect, but it did not improve the vision of the mice significantly.

Due to the time constraints of the study, the team was not able to study how the drug works at the genetic level in this project but they are planning to do so in the future. Samples were collected and have been sent for next generation sequencing. We are currently awaiting results.

In future studies, priority should focus on understanding exactly how SAHA works to protect the photoreceptor cells and whether this protection is due to the drug's molecular or pharmacological effects. This is important because RP is a complex disease that affects people in different ways and can have a big impact on their lives, so finding effective treatments, like SAHA, is very important. Additionally, it would be helpful to find treatments that work regardless of the specific genes involved in the disease.

Webinar 1 Summary (May 2023) – "Bionic Eye"

Associate Professor Penelope Allen from CERA, discussed her involvement in the development of the suprachoroidal retinal prosthesis, also known as a "Bionic Eye". This started with the first clinical trial in 2012 with three patients, then a second trial of a fully implantable device with four patients in 2018. The aim has since been to improve device functionality with the most recent study in 2022, funded by Retina Australia, confirming the benefits of the new depth algorithm and also the safety of the surgical procedure and implant after 4 years with no loss of functionality, (results summarised on page 5).

Versions of the Bionic Eye in the United States, France and Germany have ceased production as they were more complicated with more risks, being implanted on the surface of the retina or sub-retinal. Associate Professor Allen believes the simplicity of their surgical approach and the stability of the device within the suprachoroidal space (the potential space between the sclera and choroid that covers the circumference toward the back area of the eye) makes it a really good long-term option for patients.

Updates on Clinical Studies for IRD

Highlights from Associate Professor Lauren Ayton, CERA/University of Melbourne:

- Associate Professor Ayton introduced us to the VENTURE Inherited Disease Registry. With almost 300 people now registered, VENTURE aims to learn more about IRD and genetic testing so people can be identified for upcoming treatments and recruitment for clinical trials, of which there are currently seven underway. VENTURE welcomes all registrants, from those with early low vision loss to ultra low vision, as there are studies for different stages of vision loss.
- The VENTURE team recently published on its first 150 registrants, finding the most common type of IRD to be Retinitis Pigmentosa followed by Usher syndrome. The results show similar breakdowns of IRDs subtypes seen in the US and in Europe.
- Luxturna is the first and only regulatory approved gene therapy available in Australia. It is a gene replacement treatment for a form of Leber Congenital Amarois that's caused by a gene called RP65 and involves a once-off injection underneath the retina that gives the correct version of the gene. Other types of IRDs strongly researched and with clinical trials currently underway include Leber Hereditary Optic Neuropathy, Choroideremia, X-linked Retinitis Pigmentosa and Stargardt Disease. This is where genetic testing is important to find those who can benefit from potential future treatments.
- Interestingly, a research survey (funded by Retina Australia) of an estimated 5% of Australians with an IRD, found that 92% of respondents would have gene therapy if available to them, however, only 28% thought they had a good idea of what it entailed. Most people had not sought any treatments at all. Some had tried vitamin A, herbal remedies, and acupuncture. Key barriers to gene therapy were cost, potential side effects and early stages of treatment.

Webinar 1 – watch the recording

You can watch a full recording of this webinar, including more detailed study presentations and results on the Retina Australia website or click [HERE](#) to watch now.





Invitation to Webinar 2 8 August 2023

This webinar will feature:

- Associate Professor Guei-Sheung (Rick) Liu, reporting on "RNA base editing strategies as potential therapeutic of inherited retinal dystrophies" (summary page 4)
- Miss Annie Miller, co-investigator presenting on "Neuroprotective effect of SAHA in Retinitis Pigmentosa. Do time & frequency matter?" (page 6)

You can register your interest via our [website](#) or contact us on (03) 9650 5088 or info@retinaaustralia.com.au.

Our 40 year Anniversary

Retina Australia will be celebrating its 40 year anniversary in October 2023. We would love to hear any stories, receive any messages, photos or even videos to reflect upon your experience with the organisation over this time. These will be compiled as part of our celebration and recognition of Retina Australia's commitment and contribution to the inherited retinal disease community. Please send your messages and any enquiries to Jane at jane.cherry@retinaaustralia.com.au. Thank you so much for helping us celebrate this wonderful milestone!

Retina Australia Membership

2023/2024 financial year memberships are coming up for renewal. Current members will either receive a personalised email or letter shortly to complete your renewal. Your engagement is very important to us and we look forward to improving our support to you over the coming year.

If you aren't already Retina Australia member, please consider joining our community for information and support. It only costs \$30 for an Associate Membership or \$50 for a Full Membership. Only members will now have the option to receive newsletters in print or audio form. You can find out more and apply for new membership via our [website](#) or contact us on (03) 9650 5088 or info@retinaaustralia.com.au.

