RETINA REPORTER

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Retina Australia's Bi-Annual Newsletter

WINTER 2026

CONTENT

Latest research on inherited retinal diseases

This edition of the Retina Reporter brings you a comprehensive update of the latest research developments in inherited retinal diseases (IRDs), including reports from our two 2025 research grant recipients, an extensive summary of the progress that has been made on IRDs over the past decade, and a list of participation opportunities open on our IRD Research Project and Clinical Trial Register.

Message from the CEO

As we move into the middle of 2026, it is a timely opportunity to reflect on both the progress we are making and the growing momentum across inherited retinal disease (IRD) research, advocacy and support.

Research remains at the heart of Retina Australia's mission. In this edition, we highlight the impact of our Research Grants Program, with new Impact Reports demonstrating how your support is advancing understanding, improving diagnosis, and contributing to the development of new treatments for IRDs. These are tangible steps toward preserving sight and changing lives.

We are also pleased to share that our 2027 Research Grants Program is now open, and we look forward to receiving innovative applications that will advance the detection, prevention and treatment of inherited retinal diseases. Alongside this, our 2026 Annual Appeal is underway - an important opportunity for our community to continue driving this critical research forward.

This edition's In Focus section looks back on the past decade of IRD research, highlighting how far the field has progressed, from gene discovery through to emerging therapies and clinical trials. We also provide a current research update and a summary of clinical trials open for participation, recognising the importance of connecting people with opportunities to engage in research where appropriate.

Importantly, while research brings hope, it is also critical to understand the scale of the challenge we are working to address.

Inherited retinal diseases are the leading cause of blindness in people of working age in Australia, and one of the leading causes of blindness in children. However, their impact extends far beyond vision loss alone.

At a high level:

- IRDs have a lifelong impact, often beginning early and progressing over time
- The economic and social burden is significant, affecting individuals, families and the broader community
- Most of this impact sits outside the health system, including reduced workforce participation and reliance on informal care
- Employment is significantly affected, particularly as vision declines
- Families and carers play a critical role, often at personal and financial cost
- Impact increases substantially as vision loss progresses, reinforcing the importance of early intervention

Message from the CEO

These insights reinforce a clear message: investment in research is not only about future treatments, it is about reducing lifelong impact, preserving independence, and improving participation across society.

On Saturday, May 2, the Retina Australia Board came together for our annual Strategy & Planning Day, a highly constructive session that sharpened our focus, strengthened alignment, and set clear priorities for the year ahead.

We were pleased to be joined by Catherine Brooks from Equitable Philanthropy, who provided practical and actionable insights into strategic fundraising. This has helped us identify priority funding streams and shape a clear roadmap to support sustainable growth and long-term impact.

A key outcome of the day was a reaffirmed commitment to our role within the broader ecosystem.

Retina Australia will continue to play a proactive and collaborative role alongside clinicians, researchers, industry and government, as well as organisations such as Vision 2020 and its members. We will also strengthen our engagement across adjacent sectors including disability and rare disease.

While challenges remain, there is real reason for optimism. The pace of scientific discovery, combined with growing collaboration and community support, is bringing us closer to a future where inherited blindness can be prevented or treated.

Thank you for being part of this journey.

Warmest regards,

Jono Brookes
Chief Executive Officer

Pictured on the next page: Clockwise from the top: Retina Australia Board Directors with Catherine Brooks, CEO of Equitable Philanthropy, who was a guest presenter at the recent Strategic Planning day; Leighton Boyd AM and Rosemary Boyd OAM representing Retina Australia at a Guide Dogs Victoria community day in April; Jono Brookes (CEO) and Sara McKenzie (Operations Assistant) who both joined Retina Australia since the last edition of Retina Reporter.

Message from the CEO

Research into inherited retinal disease matters

This year, we’re inviting you to be part of something powerful.

For decades, inherited retinal diseases (IRDs) were considered untreatable, with no therapies able to slow or prevent progressive vision loss. That picture has changed dramatically over the past ten years, as advances in genetics and molecular medicine have driven an unprecedented wave of discovery and clinical progress.

Today, the underlying genetic cause of disease can be identified in around 60% of people with IRDs, and efforts are ongoing to close the remaining gap.

Since 1989, **Retina Australia has invested over $6.8 million into more than 150 research projects** — helping drive discoveries that are changing what’s possible for people living with inherited retinal diseases.

It’s a breakthrough that opens the door to possible new therapies for many other IRDs linked to over 400 different genes.

For over 19,000 Australians living with IRDs, the hope this innovation has provided matters.

2026 Annual Appeal

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Today, there are more than 60 clinical trials underway globally. Behind each of these is years of research, driven by dedicated scientists working to better understand, treat, and ultimately cure these conditions.

Dr Livia Carvalho is working on gene therapy approaches and trying to understand how retinal cells degenerate—research that is critical to developing future treatments for IRDs.

Retina Australia is proud to support the work of Dr Carvalho and other Australian researchers investigating treatments for IRDs through our Research Grants Program.

“The main thing for us is that Retina Australia is exclusively about IRDs — it becomes a crucial lifeline for IRD researchers,” Dr Carvalho says. “For early career researchers, they can get a Retina Australia grant that encourages them to stay in IRD research, and keep their passions in the field.”

Our goal this year is to raise $100,000 to help IRD researchers like Dr Carvalho continue this vital work.

Together, we can back the researchers bringing hope into sight.

Help us show our community they matter.

This End of Financial Year, we invite you to support crucial research by donating to the Retina Australa 2026 Annual Appeal.

As more is learned about inherited retinal diseases, progress in research and clinical trials continues to provide hope for individuals and families affected by inherited blindness. Your support can help accelerate progress and back the researchers turning hope into sight.

Donate to the Retina Australia 2026 Annual Appeal: retinaaustralia.com.au/help-us/annual-appeal

Thank you for standing with us.

DONATE NOW

All donations $2 and above are tax deductible

www.retinaaustralia.com.au (03)9650 5088

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In Focus: The Past 10 Years of IRD Research

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Overview: An exciting time for IRD research, as new discoveries are being made every year

Inherited retinal diseases (IRDs) are a group of genetic conditions that lead to progressive vision loss and are a leading cause of blindness in working-age adults.

For decades, IRDs were considered untreatable, with no therapies able to slow or prevent progressive vision loss. That picture has changed dramatically over the past ten years, as advances in genetics and molecular medicine have driven an unprecedented wave of discovery and clinical progress.

Since the first IRD-associated gene was identified in 1984, researchers have uncovered more than 400 genes linked to these conditions, with new discoveries continuing at a steady pace each year. Today, the underlying genetic cause of disease can be identified in around 60% of people with IRDs, and efforts are ongoing to close the remaining gap.

Alongside improved understanding of the genetic causes of IRD, there is also growing research to better understand these conditions - how they progress over time, why they can present differently even within the same gene, and how we can better monitor vision changes - to improve diagnosis, monitoring, and support the development of treatments.

In Focus: The Past 10 Years of IRD Research

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Sustained investment has played a critical role in this progress. Since 1989, Retina Australia has contributed over $6.8 million to support pioneering research, playing a critical role in an international pipeline that is now bringing effective treatments within reach for a growing number of IRDs.

With the field advancing so rapidly, keeping up with the latest developments can be challenging for individuals living with IRDs as well as for the researchers working to better understand and treat them. Behind each project is years of research, driven by dedicated scientists working to better understand, treat, and ultimately cure these conditions.

Here are some of the ways that treatment has progressed in recent years.

Treatments

Gene therapy is transforming how we treat retinal diseases by fixing genetic errors or replacing faulty genes. At the heart of this progress is the adeno-associated virus (AAV), a harmless virus modified to act as a "delivery truck" for therapeutic genes.

For certain single-gene disorders with a known causal mutation, surviving retinal cells, and an available approved therapy, gene replacement therapy are among the best disease-targeted treatments currently available.

In Focus: The Past 10 Years of IRD Research

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A major milestone in this area came in 2017 with the approval of Luxturna, the world's first ocular gene therapy treatment. Luxturna was approved in December 2017 by the US Food and Drug Administration to treat IRDs associated with the RPE65 gene.

Subsequent approvals for Luxturna in Europe and Australia marked the beginning of a new era in ophthalmology, demonstrating that gene-based treatments could move from concept to clinic and paving the way for a growing pipeline of gene therapy treatments we see today.

One such example is GS010 (ND4), an experimental gene therapy developed for Leber hereditary optic neuropathy (LHON), a condition typically caused by mutations in mitochondrial DNA, most commonly in the ND4 gene. These mutations impair the function of retinal ganglion cells, leading to rapid central vision loss.

Like Luxturna, the therapy uses a modified virus. GS010 uses a clever workaround for mitochondrial genetics: a healthy copy of the ND4 gene is packaged into a viral vector and is then injected into the eye. The gene is delivered to retinal ganglion cells, which produce functional ND4 protein, helping to restore mitochondrial function. This approach is called allotopic expression.

GS010 has completed Phase III clinical trials and demonstrated promising results, representing an important step forward in the development of gene therapies for mitochondrial eye diseases. While not yet approved for use in Australia, this work highlights the growing momentum in IRD research.

In Focus: The Past 10 Years of IRD Research

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Building on these advances, several gene therapy candidates have progressed to Phase III clinical trials in humans, reflecting both the maturity of the field and the increasing momentum toward effective treatments:

- **Laruparetigene Zosaparvovec/AGTC-501 (Beacon Therapeutics)**
  - Targeted at X-linked retinitis pigmentosa (XLRP) caused by mutations in the RPGR gene, being tested in the VISTA Phase 2/3 study.

- **Timrepigene Emparvovec (Biogen)**
  - A gene therapy (AAV2-REP1) for choroideremia, designed to deliver a functional CHM gene, with data from Phase 3 studies showing improvements in some participants.

- **OCU400 (Ocugen)**
  - A pioneering "gene-modifier" therapy delivering the NR2E3 nuclear hormone receptor gene to restore retinal homeostasis. Unlike gene-specific replacement, it is being targeted at multiple IRD types (e.g., RPGR, RPE65, ABCA4) where other therapies may not apply.

- **Cotoretigene Toliparvovec (BIIB112, Biogen) and Botaretigene Sparoparvovec (MeiraGTx, Johnson & Johnson)**
  - Other treatments for X-linked retinitis pigmentosa that have been studied in Phase II/III.

Outside of gene therapy, many gene agnostic treatments (such as oral medications, neuroprotection, and optogenetics) have also progressed to Phase II trials in humans.

In Focus: The Past 10 Years of IRD Research

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How patients can help advance treatment discovery by joining IRD registries in their states

IRD knowledge/registries

Developing effective treatments for IRDs depends on a detailed understanding of their genetic causes, clinical features, and natural history. This knowledge is required to target the right cause, treat the appropriate individuals, and accurately assess whether a treatment is actually working. To support future IRD research in Australia, having access to robust genetic and clinical data is essential, not only to clarify genotype–phenotype relationships, but also to identify patient groups who may benefit from emerging therapies.

In recent years there has been significant growth in this work in Australia. In 2020, researchers at the Centre for Eye Research Australia and University of Melbourne collaboratively established the VENTURE registry, which now includes more than 650 participants from across Australia and New Zealand.

This resource has become a cornerstone for IRD research and clinical trials across Australia and overseas. Complementary efforts include the Australian Inherited Retinal Disease Registry and DNA Bank (AIRDR) and the Western Australian Retinal Disease (WARD) study in Perth, and at the Save Sight Institute in Sydney. These groups collaborate closely to support research and advance the development of treatments for IRDs throughout Australia.

In Focus: The Past 10 Years of IRD Research

Together, these registries advance IRD research and support the development of improved diagnosis and treatments in Australia. They also support trial feasibility by defining the local IRD landscape, supporting advocacy to industry partners, and enabling patient identification.

Importantly, these research groups have supported IRD families in Australia in obtaining a diagnosis through various genetic testing programs. Securing a precise genetic diagnosis is increasingly important, as it is often a prerequisite for participation in targeted gene therapy trials and treatments.

Genetic testing

In Australia, genetic testing for childhood hearing loss was approved for Medicare funding in 2023, which is a significant progress not only for conditions like Usher syndrome (IRDs with hearing loss) but also sets an example for other rare diseases.

Although IRD genetic testing is not yet Medicare funded, Australian IRD research groups have played a major role in enabling access to genetic diagnosis, helping many individuals obtain answers that support clinical management and family planning decisions.

Testing approaches have also evolved from single-gene analysis to broader multi-gene panels and increasingly comprehensive genomic methods. As genomic technologies improve, involvement in ongoing research programs is also helping to identify diagnoses for IRDs that were not previously possible.

In Focus: The Past 10 Years of IRD Research

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Future directions

Over the past 10 years, there have been more advances in IRD research than ever before, including the approval of the first gene therapy and many other therapies now being tested in clinical trials. This progress is expected to continue and accelerate.

Future treatments may pair gene therapy with anti-inflammatory drugs or neuroprotectants. Machine learning analysis of data from primate studies may help researchers to create capsids with better targeting and lower immune triggers.

As genetic technologies improve and understanding of IRD disease-causing genes expands, diagnosis will become more accurate and new treatments will continue to emerge.

At the same time, registries and research studies learning more about different IRDs are helping to improve how treatments are developed and how clinical trials are designed.

Together, this progress is expected to improve how IRD clinical trials are designed and allow more patients to access emerging therapies sooner.

With more discoveries arriving every year, it is important to stay hopeful, and stay informed. Over 60 trials have been conducted globally. While not all will succeed, each teaches us more about curing inherited blindness.

Retina Australia Research Grant Impact Report

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Retina Australia is delighted to provide the final report summary on a grant project awarded in 2025 that is now complete.

Advancing Usher syndrome type 1B gene therapy with split intein

Chief Investigator
Dr Jiang-Hui Wang
Centre for Eye Research Australia
Melbourne

Co-Investigators
Professor Guei-Sheung Liu
Dr Thomas Edwards
Centre for Eye Research Australia, Melbourne

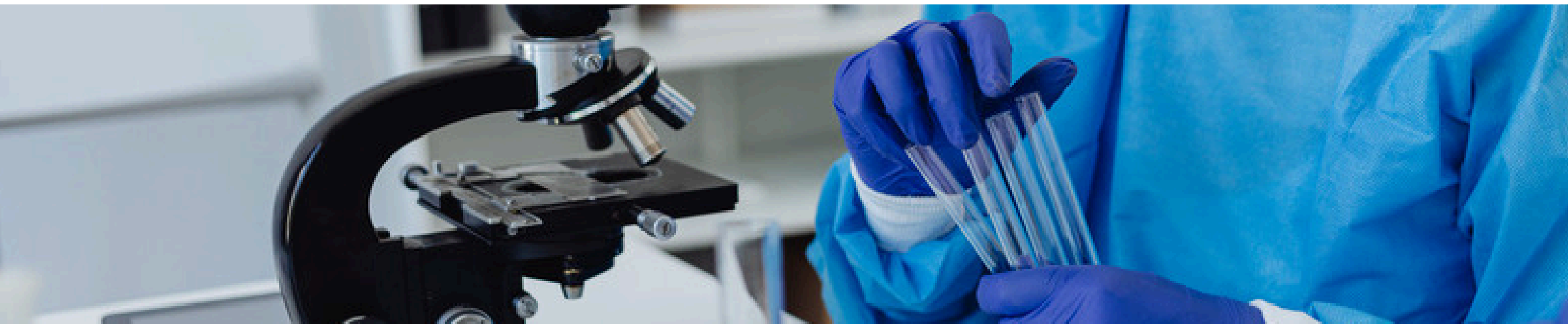
Grant awarded - $60,000 (2025)

Project Aim

Usher syndrome is the leading cause of combined deafness and blindness in the world. Usher syndrome type 1B (USH1B) is associated with mutations in the MYO7A gene.

Although the FDA has approved adeno-associated virus (AAV) for other genetic diseases like voretigene neparvovec (Luxturna™) for retinitis pigmentosa, a single AAV cannot deliver the large MYO7A gene needed for treating USH1B due to AAV's packaging limitation. Using two AAV vectors to deliver the MYO7A gene in halves at RNA level has shown limited success.

Protein trans-splicing, which efficiently joins split proteins, is a promising solution. Our study aims to show effective MYO7A protein reconstitution by finding the best split site in the gene. We aim to deliver full-length MYO7A protein in a USH1B mouse model using intravitreal injection. This less invasive administration method ensures wider vector spread in the retina compared to traditional subretinal injection, utilizing our new AAV vectors.



Retina Australia Research Grant Impact Report

Research Impact and Significance

This project made important progress toward developing better gene therapies for inherited retinal diseases that cause vision loss.

First, we identified the best way to split the large MYO7A gene so it can be packaged more effectively into viral gene therapy tools. This allowed us to achieve the highest level of MYO7A protein production in cells so far, which is a key step toward treating diseases such as Usher syndrome type 1B.

Second, we developed an improved AAV delivery vector that can reach light-sensing retinal cells, called photoreceptors, much more effectively through a simple injection into the eye. This is important because current delivery methods often struggle to reach enough of these cells safely and efficiently.

Together, these advances address two major challenges in retinal gene therapy: how to deliver a very large disease gene, and how to get that gene into the right cells in the retina. The long-term impact is the potential to create safer, less invasive, and more effective treatments for patients with inherited blinding diseases.

This work also provides a strong foundation for future studies and, ultimately, translation toward clinical use.

Retina Australia Research Grant Impact Report

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Retina Australia is delighted to provide the final report summary on a grant project awarded in 2025 that is now complete.

Therapies for currently untreatable autosomal recessive IRDs

Chief Investigator
Professor Robyn Jamieson
Children's Medical Research Institute,
University of Sydney

Grant awarded - $60,000 (2025)

Project Aim
This project aims to develop new treatments for certain inherited retinal diseases (IRDs) that currently have no cure. These diseases are caused by faults in small genes that are small enough that they can be replaced using gene therapy. About one-quarter of all IRDs fall into this category. Gene replacement therapy using AAV (adeno-associated virus) has already worked for one condition caused by variants in the RPE65 gene, giving us confidence that this approach can help others.

In this project, we use stem cells from patients and grow them into "retinal organoids" which are miniature versions of the retina created in the lab. These allow us to study the disease closely and test new gene therapy treatments. By focusing on small genes linked to autosomal recessive IRD, we aim to speed up the development of future therapies for these currently untreatable conditions.

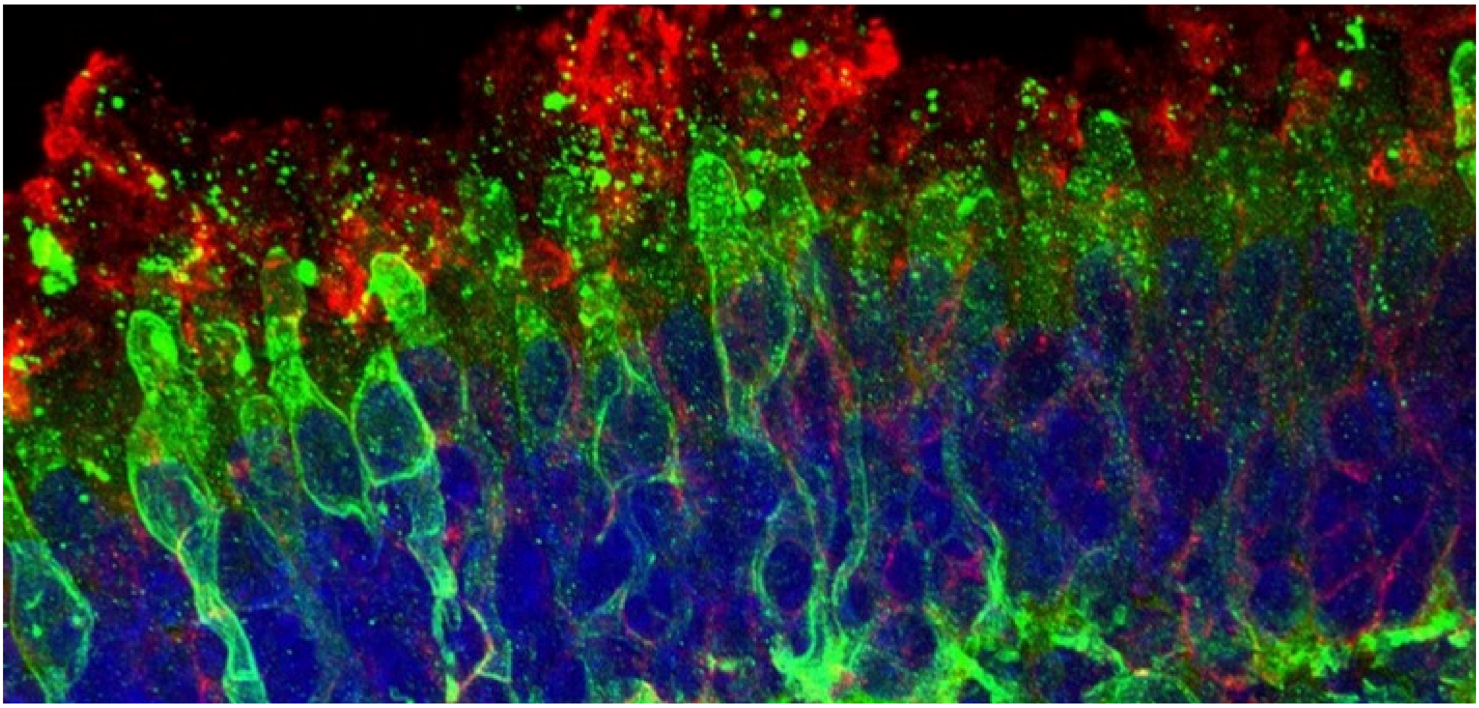
Project summary
In this project, we created stem cells from patients who have changes in a small gene that causes inherited retinal disease (IRD). There is currently no treatment for this condition. Because the gene is small and the disease happens when the gene stops working properly, they are good candidates for a form of gene therapy where we add back a healthy copy of the gene.

Research Grant Impact Highlight

Project Summary (cont'd)
We turned these stem cells into "retinal organoids," which are tiny 3D models of the retina grown in the lab. These organoids allow us to study the disease in a realistic way. We also created healthy control organoids for comparison.

Using these organoids, we looked for signs of damage to the photoreceptor cells which are responsible for vision. We examined their structure, specific markers in the cells, and patterns of gene activity. After identifying reliable signs of disease, we developed gene therapy constructs and delivered them to the organoids. Early results suggest that the replacement therapy approach is promising.

Impact
This project has helped us discover important markers of disease and investigate new gene therapy approaches. Since there are around 120 other similar small genes that can cause related IRDs, many of these conditions may also benefit from the same type of gene-replacement therapy. This work strengthens our pipeline for producing retinal organoids, studying disease changes, and developing new gene therapies. This paves the way toward treatments for many currently untreatable IRDs.



A retinal organoid used for functional genomic investigations for variant classification.
Image: Prof Robyn Jamieson

Research Project and Clinical Trial Register

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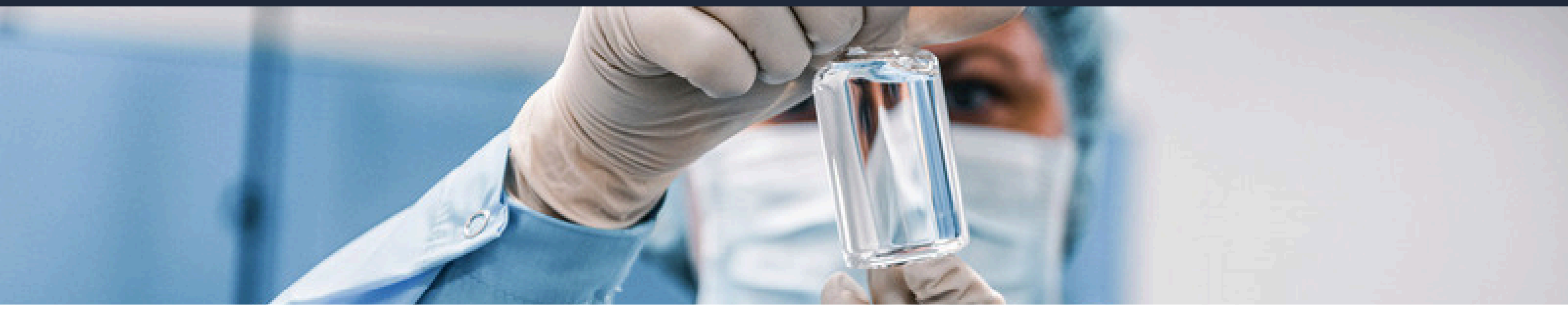

Summary of Projects and Trials Currently Recruiting Participants

The IRD Research Project and Clinical Trial Register provides information about research projects and clinical trials in IRDs. This Register is for informational purposes only, and further details can be sought via the email contacts. For more detail on each project, refer to our website at: retinaaustralia.com.au/inherited-retinal-disease/ird-research-project-and-clinical-trial-register/

Sydney Eye Hospital PRIMAlia clinical trial
Disease: Stargardt disease, Best disease, some patients with retinitis pigmentosa
Participants: Patients
The Save Sight Institute have recently commenced the PRIMAlia clinical trial at Sydney Eye Hospital / University of Sydney, which is relevant to those with inherited retinal disease affecting the macula. The trial is currently recruiting patients with inherited photoreceptor degeneration involving central vision loss. This study evaluates the PRIMA subretinal photovoltaic implant, a novel retinal prosthetic approach aimed at restoring central vision in patients with severe macular photoreceptor loss.
Recruiting: https://clinicaltrials.gov/study/NCT07266584

Sponsored post
A Phase 2 Study of the Safety and Efficacy of a Potential New Treatment for Patients with Late-stage Retinitis Pigmentosa - Sponsor: Kiora Pharmaceuticals Pty Ltd
Disease: Retinitis Pigmentosa **Participants:** Patients
If you have been diagnosed with retinitis pigmentosa (RP), you may be eligible to participate in a clinical study. Kiora Pharmaceuticals is sponsoring a study to evaluate the safety and efficacy of a potential new treatment to improve vision in individuals with profound vision loss due to RP. Conducted at 5 locations in Australia, this clinical study will require you to attend the study site for study visits every 3 weeks for up to 30 weeks. During the study, you will receive 3 injections of the potential new treatment into each eye, and you will be asked to undergo assessments and tasks to assess safety and effects on your vision.
Recruiting: Save Sight Institute, Sydney, contact: john.grigg@sydney.edu.au,
Queensland Eye Institute, Woolloongabba, contact: brett.caldwell@qei.org.au
Royal Adelaide Hospital, Adelaide, contact: melanie.willoughby@sa.gov.au
Cerulea Clinical Trials, East Melbourne, contact: llombardi@ceruleaclinicaltrials.org.au
Lions Eye Institute, Nedlands, Western Australia, contact: tammy.corica@lei.org.au

Research Project and Clinical Trial Register

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Sponsored post
A phase I/II dose-escalating study of the safety, tolerability and efficacy of KIO-301 administered intravitreally to patients with retinitis pigmentosa and choroideremia (ABACUS) – Substudy - Sponsor: Kiora Pharmaceuticals Pty Ltd
Disease: Retinitis Pigmentosa (RP) or Choroideremia (CHM)
Participants: Patients
This study aims to evaluate the use of specialised tasks to assess vision in individuals with profound vision loss due to RP or CHM. Led by Professor Robert Casson and the Royal Adelaide Hospital, this clinical study will require you to participate in non-invasive tasks (e.g., navigation and object identification) to help validate these functional vision tests so they can be used to assess potential new treatments.
Recruiting: South Australia Contact: Melanie.Willoughby@sa.gov.au

Sponsored post
SUNDEW: ADOA Interventional Clinical Trial - Sponsor: PYC Therapeutics
Disease: Autosomal dominant optic atrophy (ADOA)
Participants: Patients
This study is a first step to test a new treatment for individuals with a confirmed mutation in the OPA1 gene, which causes a condition called autosomal dominant optic atrophy (ADOA) that affects vision. It targets the root cause of the genetic mutation and involves giving a single, increasing dose of the treatment directly into the eye to see how safe it is and how well people can tolerate it to find the safest and most effective dose of PYC-001 for treating ADOA.
Recruiting: Centre for Eye Research Australia, East Melbourne, Victoria and Lion's Eye Institute, Western Australia. Contact: Sundew@pyctx.com

Sponsored post
An Observational Clinical Trial of PRPF31 (RP11) - Sponsor: PYC Therapeutics
Disease: Retinitis Pigmentosa
Participants: Patients, Carriers
This study aims to observe the progression in patients with the inherited retinal disease (IRD) retinitis pigmentosa 11 (PRPF31 or RP11) over the period of four years.
Recruiting: Australia-wide Contact: quokka@lexitas.com

Research Project and Clinical Trial Register

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Understanding rod function changes in choroideremia and retinitis pigmentosa

- Sponsor: University of Melbourne and Centre for Eye Research Australia
Disease: choroideremia, and retinitis pigmentosa related to the RPGR and USH2A genes
Participants: Patients, Carriers
This study is looking to validate new clinical trial outcomes, and involves standard imaging and vision tests as well as a light detection task. You would be required to come to Melbourne University in Carlton for one 3-hr session, with a $50 travel reimbursement available.
Recruiting: Melbourne Contact: IRD@groups.unimelb.edu.au

Exploring Barriers and Facilitators to Social Participation in Blind or Vision Impaired Australian Young Adults - Sponsor: Griffith University
Disease: All Inherited Retinal Diseases **Participants:** Patients
This research aims to understand the barriers and facilitators influencing social participation, with the goal of identifying strategies to foster inclusion and accessibility in educational, social, and community settings.
Recruiting: Australia-wide Contact: chrisy.mowbray@griffithuni.edu.au

Perspectives of stem cell therapies for retinal conditions - Sponsor: University of New South Wales and Children's Medical Research Institute
Disease: All Inherited Retinal Diseases and Age-Related Macular Degeneration
Participants: Patients
By completing a survey, we are seeking your perspectives of stem cell therapy, a novel therapy being researched in the laboratory as a potential option for these eye conditions.
Recruiting: Australia-wide Contact: visionlossPSP@unsw.edu.au

Exploring therapies for those who have none - Sponsor: Perron Institute for Neurological and Translational Science
Disease: All rare genetic retinal diseases with no current treatments
Participants: Family Members, Patients
The study will provide a genetic diagnosis by identifying specific changes in the patient's DNA, confirm that is causing the disease, and explore new treatments using our three decades of experience with a type of drug called Antisense oligomers. These drugs could help to restore normal gene function.
Recruiting: Tasmania, WA, Victoria, NSW Contact: MolecularTherapy@murdoch.edu.au

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