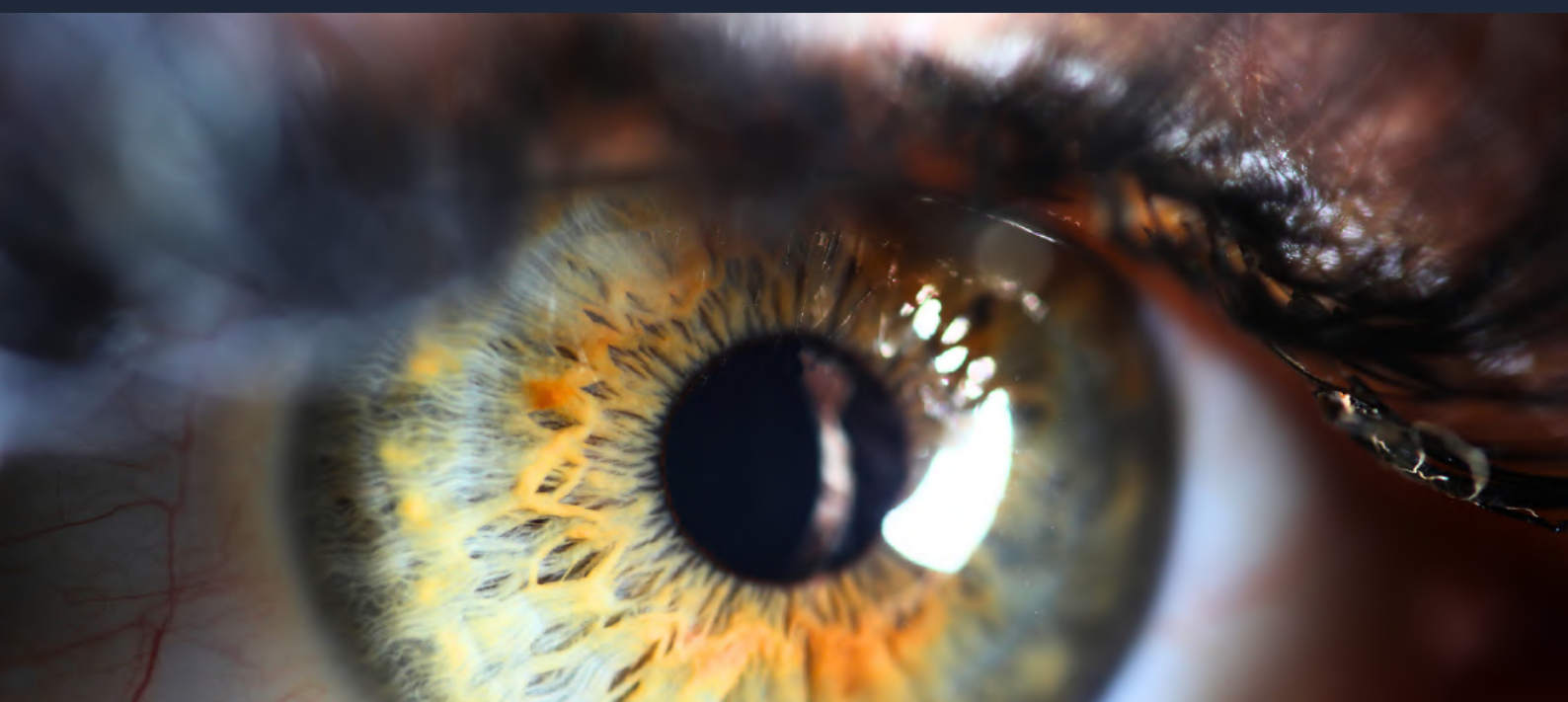


RETINA REPORTER

Retina Australia's Bi-Annual Newsletter

SUMMER 2024/2025



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World Geographic Atrophy Day 5 December 2024

Retina Australia is joining the global campaign to raise awareness for World Geographic Atrophy Day on 5th December 2024. As one of the leading causes of vision loss and blindness, particularly in Australians over 65 years of age, we encourage you to embrace the movement.

1. Educate yourself
2. Get your eyes checked
3. Spread the word

Read on for more information.

Message from the CEO



The launch of the “Top 10 Research Priorities for Inherited Retinal Disease” and the “Geographic Atrophy: Can’t Wait and See” report, Medicine Australia’s PharmAus24 event, World Retina Day, World Sight Day, the Vision 2020 Australia Parliamentary Friends Group for Eye Health and Vision Care Cocktail Function and Exhibition, and the (The Royal Australian and New Zealand College of Ophthalmologists) RANZCO Congress 2024, were all wonderful opportunities to spread the word about inherited retinal diseases and other retinal dystrophies, and to highlight the critical need to fund and facilitate medical research.



As we continue to raise awareness, we are also proud to support two new research projects for 2025, both focused on finding new treatments. These will be funded by Retina Australia’s Research Grants Program. You can help support these projects by making a gift of hope to our 2024 Christmas Appeal, thank you so much.

Retina Australia welcomed the Federal Government's recent decision to end discrimination based on genetic test results in life insurance underwriting. Removing barriers to genetic testing will allow Australians to make health decisions without worrying about being denied life insurance. Genetic testing is an essential step to determine eligibility for the only treatment currently available in Australia for inherited retinal disease, and for participating in clinical trials and future therapies.

We warmly welcome our new Scientific and Medical Advisory Committee (SMAC). Our expert members will advise on medical developments and we look forward to keeping you updated on these advancements. The SMAC includes the Chair of our Grants Advisory Committee, Professor John Grigg, and our two delegates to Retina International’s Scientific and Medical Advisory Board, Professor Erica Fletcher and Associate Professor Lauren Ayton. As the only Australian member of Retina International, Retina Australia benefits from global insights, enhancing our knowledge and awareness on an international scale.

Retina Australia’s Board and staff wish you all a happy and safe festive season. Thank you for your incredible support and we look forward to another 12 months of progress for better health outcomes for people affected by inherited retinal diseases.

Warmest regards,

A handwritten signature in blue ink that reads 'Julia Hall'.

Julia Hall, Chief Executive Officer

Message from the CEO



Pictured clockwise from the top left: Advisory committee representatives of the "Geographic Atrophy: Can't Wait and See" report; Jerome Laxale MP golf putting with Do It in the Dark (DITD) glasses; Professor John Skerritt AM golf putting with DITD glasses; Julia Hall with A/Prof Lauren Ayton presenting the "Top 10 Research Priorities for Inherited Retinal Diseases" poster at the RANZCO Congress; Rachie Williams MP and Geoff Provest MP wearing the DITD glasses and holding the June Retina Reporter newsletter; Carly Iles CEO Vision 2020 Australia completing a DITD puzzle, Retina Australia Directors Dino Farronato and Josh Ginpil with Julia Hall and Senator Tammy Tyrrell; Retina Australia Company Secretary Rosemary Boyd OAM and Chair, Leighton Boyd AM at the RANZCO Congress; Steering Committee representatives of the "Top 10 Research Priorities for Inherited Retinal Diseases" at NSW Parliament House with Tim James MP.



Scientific and Medical Advisory Committee



Associate Professor
Lauren Ayton

Departments of Optometry and Vision Sciences and Surgery at the University of Melbourne and Co-Lead of the Retinal Gene Therapy Unit at the Centre for Eye Research Australia



Associate Professor
Tom Campbell

Consultant ophthalmologist, Coastal Eye Centre, Sunshine Coast University Hospital, the Centre for Eye Research Australia at the University of Melbourne, and the NQ Eye Foundation



Associate Professor
Fred Chen

Principal Research Fellow at the University of Western Australia and a Clinical Associate Professor at University of Melbourne



Dr Glyn Chidlow

Research Scientist, South Australian Institute of Ophthalmology and University of Adelaide



Professor
Erica Fletcher

Head of Visual Neuroscience Laboratory, Department of Anatomy & Cell Biology, The University of Melbourne



Professor
John Grigg

Head of the Speciality of Clinical Ophthalmology and Eye Health, Save Sight Institute, The University of Sydney



Professor Alex
Hewitt

Professor of Ophthalmology at the Menzies Institute for Medical Research at the University of Tasmania



Dr Michael
Hogden

Retinal specialist and Consultant vitreoretinal surgeon at the Princess Alexandra Hospital, Brisbane



Professor Michael
Kalloniatis

Professor at the College of Optometry, University of Houston Texas USA



Professor David Mackey

Head of Research, Clinical Genetics & Epidemiology, Lions Eye Institute, Centre for Ophthalmology and Visual Science, University of Western Australia



Dr Jon Ruddle

Consultant ophthalmologist at the Royal Children's Hospital and Royal Victorian Eye and Ear Hospital.

2025 Research Grant



Retina Australia is delighted to support two new Research Grants in 2025 with the following researchers and their projects awarded for the coming year.



Therapies for currently untreatable autosomal recessive inherited retinal diseases

Chief Investigator

Professor Robyn Jamieson
Children's Medical Research Institute (CMRI), Sydney
Sydney Children's Hospitals Network
Save Sight Institute, University of Sydney

Grant awarded - \$60,000 (2025)

Project Aim

This project aims to develop gene replacement therapies for autosomal recessive (AR) inherited retinal diseases (IRDs), where no treatments currently exist. It focuses on targeting genes small enough for adeno-associated virus (AAV)-mediated gene therapy, which has shown promise in other cases and will target AR IRDs caused by variants in photoreceptor specific genes.

Project Summary

The project will generate patient-derived stem cells from three individuals with small gene AR IRDs and develop retinal organoids—3D models mimicking the retina—to study biomarkers and test AAV-mediated gene replacement therapy. There are approximately 120 IRD disease genes which are small and where the diseases they cause would also be likely to be amenable to gene replacement therapy. This project will refine the process of creating retinal organoids and identify disease biomarkers while optimising the gene therapy.

Expected Outcomes

The project expects to identify biomarkers that help characterise the disease, which could also assist in diagnosing patients with uncertain genetic variants. It expects to demonstrate a successful gene replacement therapy in retinal organoids, with the hope to then progress to clinical trials. This would offer potential new treatment options for small-gene AR IRDs, paving the way for gene therapies and diagnostics for other similar IRD conditions.

2025 Research Grant



Advancing Usher syndrome type 1B gene therapy with split intein

Chief Investigator

Dr Jiang-Hui (Sloan) Wang - Centre for Eye Research Australia (CERA)

Co-investigators

Associate Professor Guei-Sheung (Rick) Liu, CERA
Dr Thomas Edwards, CERA

Grant awarded - \$60,000 (2025)



Project Aim

Usher syndrome type 1B (USH1B) is caused by mutations in the MYO7A gene, leading to combined hearing and vision loss. However, the MYO7A gene is too large for delivery using standard adeno-associated virus (AAV) gene therapies. This study is exploring a new approach using a method that joins split proteins efficiently. The aim is to deliver the full MYO7A protein in a mouse model of USH1B through a less invasive injection into the eye, which could lead to better treatment outcomes.

Project Summary

Early experiments have successfully reassembled a split green fluorescent protein (GFP) in cells, laying the foundation for this MYO7A study. The goal is to find the best position to split the MYO7A gene for effective reassembly. Different split sites will be designed, focusing on specific amino acids that help the process work smoothly, and testing of which split produces the full-length MYO7A protein will be conducted. The therapy will be delivered into the eye of a mouse model of Usher syndrome type 1B using this new method to target photoreceptor cells. By splitting the MYO7A gene into two parts and delivering them with this new technique, the aim is to produce the full MYO7A protein, advancing treatment for inherited retinal diseases that involve large genes.

Expected Outcomes

The project expects to identify the ideal split site for effective reassembly of the MYO7A protein and demonstrate successful delivery of the full MYO7A protein to retinal cells via a two-part AAV vector. If successful, this method could help reverse retinal disease in the USH1B model and offer a pathway for treating other inherited retinal diseases caused by large genes like ABCA4 and CEP290.

2024 Christmas Appeal



Help us fund two new research grant projects that will focus on the discovery of potential new gene therapies.



2024 CHRISTMAS APPEAL

Give the gift of hope by funding life-changing research into inherited retinal disease

[DONATE NOW](#)

Give an everlasting and impactful gift of hope with a donation this festive season.

Your donation will contribute to accelerating research into inherited retinal disease with the hope of finding new treatments to stop vision loss.

Every donation helps. Together we CAN make a difference!

You can donate via the QR code or at www.retinaaustralia.com.au/donate.



We sincerely thank you for your support.

[DONATE NOW](#)



All donations \$2 and above are tax deductible

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

World Geographic Atrophy Day



Join us to raise awareness on 5 December 2024 for World Geographic Atrophy Day

As we age, our vision can change in ways we might not expect. One such change is the development of geographic atrophy (GA), a condition that affects many older adults but is often misunderstood. On December 5, in honor of World Geographic Atrophy Day, we want to shed light on this important degenerative eye condition, raising awareness and understanding in our community.

What is Geographic atrophy (GA)

Geographic atrophy is a form of advanced age-related macular degeneration (AMD), which primarily affects the macula, which is the part of the retina responsible for sharp, central vision. GA occurs when cells in the macula begin to deteriorate, leading to the gradual loss of vision. This condition can significantly impact daily activities like reading, driving, and recognising faces.

In Australia, there is an urgent need to address the impact of GA. The financial burden of GA-related vision loss exceeds \$1.8 billion annually, including \$377.23M in direct healthcare costs, \$312.74M in indirect healthcare costs and \$1,112.49M in wellbeing costs.

World Geographic Atrophy
Day Exclusive Sponsor



World Geographic Atrophy Day



Recognising the Symptoms

Knowing the symptoms of GA is crucial for early detection. Here are some signs to watch for:

- Gradual central vision loss - Over time, you might develop blind spots in your central vision, making it hard to focus on details.
- Blurred or distorted central vision - You might notice that straight lines appear wavy or that objects seem blurry.
- Difficulty with low-light vision - You may struggle to see in dimly lit environments or at night due to reduced night vision.
- Decreased contrast sensitivity - It may be harder to distinguish objects of similar shades or colours, affecting activities like reading and navigating unfamiliar areas.

If you experience any of these symptoms, it is advised that you consult your eye care professional as soon as possible.

Risk Factors

Several risk factors can increase your likelihood of developing GA:

- Family History - Genetics can significantly increase the risk of geographic atrophy (GA) and age-related macular degeneration (AMD).
- Lifestyle Choices - Smoking is the most significant modifiable risk factor for GA, along with diet and UV light exposure. Protecting your overall health can help preserve your vision.
- Ageing - This is the main non-modifiable risk factor, with risk increasing significantly after the age of 50.
- Inflammation - Retinal inflammation can cause dysfunction in the retinal pigment epithelium (RPE) and photoreceptor damage, resulting in GA lesions.



World Geographic Atrophy Day



The Importance of Early Detection

Early detection of GA is key to managing the condition. Regular eye exams can help identify changes in your vision and allow for timely intervention. Early diagnosis and recording of progression can help make informed decisions about your eye health and lifestyle adjustments that may assist in slowing deterioration.

Emerging Treatments

Two treatments for GA have been approved by the Food and Drug Administration (FDA) and are available in the US: Avacincaptad pegol (Izervay, Astellas Pharmaceutical) and Pegcetacoplan (Syfovre, Apellis Pharmaceuticals), with additional clinical trials ongoing. These medications are not currently approved for use in Australia and are undergoing review by the Therapeutic Goods Administration (TGA). It is hoped that the first treatments for GA will become available in Australia within the next 12 months to prevent vision loss.

Join the Movement

On December 5th, we invite you to participate in World Geographic Atrophy Day. Here's how you can get involved:

1. Educate Yourself - Take the time to learn more about GA and its implications. Stay aware of any changes in your vision. You can find more information about GA at www.retinaaustralia.com.au.
2. Have your eyes checked - Regular check-ups can help ensure that any potential issues are caught early, allowing for potential timely intervention.
3. Spread the Word - Share this article, talk to friends and family about GA, and help us raise awareness in our community.

Understanding geographic atrophy is especially important for those in at-risk groups. By raising awareness, we can foster a supportive community and help those affected feel empowered in managing their vision health.

Together, let's make a difference this World Geographic Atrophy Day!

In Focus: Inherited retinal disease



What is an inherited retinal disease?

An inherited retinal disease (IRD) is a condition caused by a genetic anomaly that leads to loss of vision and in some cases, legal blindness.

They are also known as inherited retinal dystrophies or inherited retinal degenerations.

There are over 20 types of inherited retinal diseases. Depending on the disease, you may have vision loss at birth, or it may occur later in life.



What causes an inherited retinal disease?

Genetic error

An IRD is caused by a variation in your genetic code or DNA.

You can think of DNA as a recipe book, which contains all the instructions needed for your body to make the proteins to survive and thrive. Proteins are important as they provide cell structure, function and regulation of the body. In genetic diseases, there is an error in one of the recipes, which can lead to disease.

In IRDs, the genetic error usually results in an abnormality of the photoreceptor cells in the retina. The retina is the light-sensitive tissue at the back of the eye that senses and transmits light signals to the brain through the optic nerve to form images, enabling people to see.

Over 300 IRD associated genes

Researchers have found over 300 genes that cause IRDs. (1)

Some IRDs, such as choroideremia or X-linked retinoschisis, are caused by one or only a small number of genes (known as a “monogenic” disease). Other diseases can be caused by many different mutations in many different genes. For example, over 70 different genes have been identified that can cause retinitis pigmentosa. Some gene anomalies cause more severe inherited retinal diseases than others.

In Focus: Inherited retinal disease



60% successfully diagnosed

Currently, we are only able to find the causative gene mutation for about 6 in 10 people. This is because we still do not know all the genes that cause IRDs and, in some cases, our testing is not sensitive enough to detect certain changes in the genetic information. However, researchers are working on more sensitive ways to examine a person's genetic code in more detail, which should improve this number over time.

Inheritance patterns

The way that each IRD is passed from generation to generation (inheritance pattern) can be different in each family.

- You may have a strong family history and know a family member with the condition.
- Or you may be the first one in your family to be diagnosed.

The different inheritance patterns of IRDs mean that there are different chances of children inheriting a condition, depending on the pattern. For example, recessive conditions can skip generations, whereas dominant inheritance is more likely to be present in most generations. In addition, it is possible for a person to develop an entirely new genetic mutation – this is known as a “de novo” gene variant. For more details, see the “Inheritance Patterns” section on the Retina Australia website at www.retinaaustralia.com.au.

Syndromic IRDs

In most people, IRDs only affect the eyes. However, some types are linked with other health issues, such as Usher syndrome (which also affects hearing). (2)

How many people live with an inherited retinal disease?

In Australia, it is estimated that ~ 1 in 1380 people are living with an IRD, which is around 19,000 people.(3) IRDs are currently:

- the leading cause of blindness in working age adults and
- the second leading cause of blindness in children in Australia (following cerebral visual impairment).(4)

Disease progression

IRDs can affect individuals of all ages and genders and can progress at different rates. Some may be born with or experience vision loss in infancy or early childhood. Some may experience a gradual loss of vision later in life.

In Focus: Inherited retinal disease

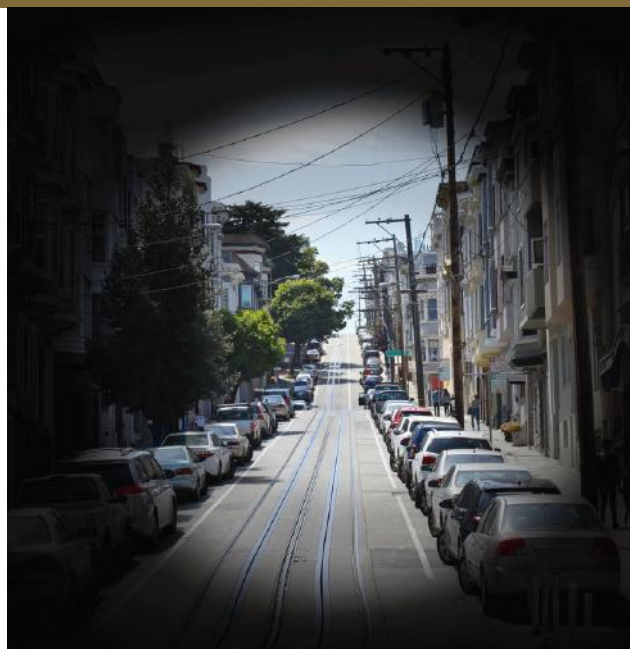


The prognosis of an IRD also varies – some people will lose all vision, whilst others will have more mild disease.

Common symptoms of IRDs

Each IRD is different, but common symptoms include:

- Low vision at night, or in dim or dark settings
- Loss of central and/or peripheral vision
- Sensitivity to light, glare and difficulty in bright light
- Farsightedness or shortsightedness
- Blind spots
- Colour differentiation difficulties or not seeing all colours
- Uncontrolled eye movements (nystagmus)



Vision loss can vary between different IRDs

In some conditions, only the central vision is lost, while others lose peripheral vision first, with a slow progression into the central vision areas. Some people will only ever have mild vision loss, whilst others can progress to lose all vision, also known as “no light perception” or “black-blindness”.

Your ophthalmologist may be able to give you some indication of the expected future for you based on your clinical presentation and genetic testing, but this is not an exact science. Sometimes, it can be unpredictable as to how the disease will proceed.

Obtaining a confirmed diagnosis through genetic testing is the only way to verify the exact gene anomaly(s) that is the underlying cause of an IRD.⁽⁵⁾

References

- (1) University of Texas–Houston Health Science Center Laboratory for the Molecular Diagnosis of Inherited Eye Diseases. <https://retnet.org/summaries>.
- (2) Nisenbaum E, Thielhelm TP, Nourbakhsh A, et al. Review of Genotype–Phenotype Correlations in Usher Syndrome. *Ear Hear*.
- (3) Hanany M, Rivolta C, Sharon D. Worldwide carrier frequency and genetic prevalence of autosomal recessive inherited retinal diseases. *Proc Natl Acad Sci U S A*. Feb 4 2020;117(5):2710–2716. doi:10.1073/pnas.1913179117
- (4) Heath Jeffery RC, Mukhtar SA, McAllister IL, Morgan WH, Mackey DA, Chen FK. Inherited retinal diseases are the most common cause of blindness in the working-age population in Australia. *Ophthalmic Genet*. May 3 2021:1–9. doi:10.1080/13816810.2021.1913610
- (5) Grigg J, Jamieson R, Chen F, et al. Guidelines for the Assessment and Management of Patients with Inherited Retinal Degenerations (IRD). 2020.

This article has been supported by Novartis.

Diagnosis and genetic testing



How do I know what type of IRD I have?

Difficulty in diagnosis

When first diagnosed with an IRD, the disease name can vary between doctors due to the complexity of diagnosing these conditions, involving over 300 known genes with overlapping effects. Genetic testing, known as a "molecular diagnosis," identifies the specific gene causing the IRD. For example, the RPE65 gene can lead to different clinical presentations such as retinitis pigmentosa (RP) or Leber congenital amaurosis (LCA). To minimise confusion, some professionals refer to the condition by the gene name, like RPE65-associated retinal degeneration.

Genetic testing

What is genetic testing?

Genetic testing, also known as DNA testing, is a medical process undertaken to examine a person's DNA, to identify changes in genes which can be used to confirm or rule out a genetic condition. It is the only way to be certain about what genetic mutation is causing a person's IRD. Testing is also sometimes referred to as obtaining a "molecular diagnosis". To have a genetic test, you need to give a DNA sample. This can be collected from a blood test, saliva sample, or a buccal swab (using a cotton bud to collect some of the cells from inside your cheek).

How do I access genetic testing and what is involved?

If you are interested in obtaining a genetic test, the first person to speak to is your ophthalmologist. You will however, usually need a referral from a general practitioner or optometrist before you can visit an ophthalmologist. Your ophthalmologist will be able to advise on whether a genetic test is suitable for you, and explain the potential costs, risks and benefits. If you would like to see another healthcare professional (i.e. a clinical geneticist or a specialised ocular genetics clinic), your doctor can arrange a referral.

Before providing a sample for a genetic test, you will undertake counselling with either your doctor or a specialised genetic counsellor. They will inform you of the details of the process, and ask you to sign a form to acknowledge you consent to the procedure.

You will then be asked to supply a sample of either blood, saliva, or a cheek swab. The sample is then sent to a testing laboratory, where the genes of most likely involvement are tested.

Diagnosis and genetic testing



The other information which is vital to genetic testing is knowledge of your family history. Before you start the process, it is important to find out if anyone else in your family has an IRD. You will be asked to provide this information to your doctor, as it helps them identify the most likely gene to test for.

How much does genetic testing cost?

The cost of a genetic test varies significantly depending on the test type and testing facility. Australians have a choice to fund this testing privately (i.e. pay out of your own pocket), or through the public health system (which covers the costs for some patients through government funding programs but may have long wait times). For some people with a clinical diagnosis of retinitis pigmentosa (RP) or Leber congenital amaurosis (LCA), there is also currently a sponsored genetic testing program called the Vision Genome Program. In addition, there may be avenues to have genetic testing via participation in natural history studies for IRDs. We advise that you discuss your options for genetic testing with your ophthalmologist and that your genetic testing process be accompanied by consultation with a genetic counsellor. Find out more information about genetic testing including contact details of public clinical IRD genetic testing providers and natural history studies, and what to expect when results are ready, in the “Genetic Testing” section on the Retina Australia website at www.retinaaustralia.com.au.

The importance of genetic testing for treatment access

In order to access the only available treatment in Australia (refer details on the following page) and to participate in certain clinical trials with early access to potential new therapies, you will be required to have a genetic test to determine eligibility with an accurate diagnosis for which the treatment is targeted.

You can find a current listing of research projects and clinical trials open for participation on the “IRD Research Project and Clinical Trial Register” section on the Retina Australia website at www.retinaaustralia.com.au. Projects on the Register have also been summarised later in this newsletter. Retina Australia recommends that you talk to your ophthalmologist regarding any treatment or interventional clinical trial participation to discuss the best options for you.

This article has been supported by Novartis.

Retina Australia Christmas Office Hours

The office will be closed from Friday 13th December 2024
and reopen on Tuesday 7th January 2025

Are there treatments available?



One treatment in Australia

In Australia, there is currently one regulatory-approved therapy for an IRD, a gene therapy called Luxturna® (voretigene neparvovec-rzyl, Novartis), for those related to mutations in the RPE65 gene. Associated IRD conditions for the RPE65 gene may include retinitis pigmentosa (RP) or Leber congenital amaurosis (LCA).

Luxturna® was approved by the Australian Therapeutic Goods Administration (TGA) in 2020 and is currently available at two treatment sites – the Sydney Eye Hospital and the Royal Victorian Eye and Ear Hospital. We recommend that you speak to your ophthalmologist to find out more.

Emerging treatments

There are also a number of emerging treatments currently in the pipeline. These include gene therapies, stem cell therapies, optogenetics, pharmaceutical compounds and vision prostheses. There is no guarantee that they will become available however, as stringent clinical trials must be performed first to show safety and efficacy, and they are all in various stages of development.

Phase 3 interventional clinical trials can serve as beacons of hope for the discovery of potential new treatments. This phase is typically the last stage of testing to provide evidence of effectiveness, which is necessary for seeking regulatory approval from bodies such as the Therapeutic Goods Administration (TGA) in Australia, the Food and Drug Administration (FDA) in the USA, and the European Medicines Agency (EMA) in Europe. However, exceptions exist. Pathways like accelerated approval or conditional approval can allow for earlier submissions for treatments addressing serious conditions with unmet medical needs.

Are there treatments available?



Currently, there are two trials showing great promise for inherited retinal diseases. They are awaiting final Phase 3 results or are in discussion with regulatory authorities.

Bota-vec for X-linked Retinitis Pigmentosa (XLRP)

In a collaborative investigation with MeiraGTx Holdings plc, Janssen Pharmaceuticals has been conducting a [Phase 3 trial](#) called LUMEOS for Botaretigene Sparoparvovec (Bota-vec), formerly AAV-RPGR, for the treatment of X-linked retinitis pigmentosa (XLRP) associated with the RPGR gene. X-linked conditions are caused by gene mutations on the X-chromosome and are typically more severe in males.

Bota-vec is administered via a single injection into the sub-retinal space, located between the light-sensitive retina lining the back of the eye and the underlying posterior blood supply of the eye. This one-time gene therapy is designed to deliver functional copies of the RPGR gene aiming to preserve and prevent retinal cell loss in people living with XLRP. Preliminary results from earlier Phase 1/2 trials demonstrated significant improvements in both retinal sensitivity and vision-guided mobility, highlighting the drug's potential benefits. The Phase 3 trial is expected to have been recently completed, with results anticipated to be reported within the next 6–9 months.

MCO-010 for Retinitis pigmentosa and Stargardt disease

MCO-010 (sonporetigene isteparvovec), developed by Nanoscope Therapeutics, is a gene therapy designed to restore light sensitivity in patients with inherited retinal diseases, regardless of specific mutations. Delivered via a single eye injection, it enables bipolar cells to produce the MCO protein, improving light sensitivity in cases of severe vision loss. This technique is known as optogenetics.

In a previous article, "[Optogenetics win for retinitis pigmentosa after changing primary endpoint](#)", we highlighted [the Phase 2 clinical trial success](#) of MCO-010 for retinitis pigmentosa. In October 2024, Nanoscope Therapeutics announced that after a productive meeting with the FDA, it will initiate a Biologics License Application (BLA) in Q1 2025, with reference to its fast track designation received from the FDA.

Also resulting from this trial success, Nanoscope Therapeutics is set to launch a Phase 3 clinical trial for MCO-010 targeting individuals with advanced Stargardt disease, with site selection in Q4 2024 and patient dosing in Q1 2025.

We eagerly await further announcements of these studies and potential new therapies and are hopeful for the delivery of new treatments for IRDs.

Research participation



An interview with Dino Farronato – A participant's perspective



Dino Farronato is a Director of Retina Australia. In our recent research update webinar (available for viewing on our website), Dino kindly shared his story about living with an inherited retinal disease, his genetic testing experience and why he recommends participation in research studies.

Thank you for sharing your story with us Dino.

I am a lawyer, company director, husband, father, bush walker, rugby tragic and a person with low vision as a result of retinitis pigmentosa (RP).

I was diagnosed with RP in my 30s. With neither parent or extended family having vision problems, but my only sibling and I having RP, it is likely due to a recessive gene. Since my initial diagnosis, I have consulted experts in Australia and elsewhere searching for answers, all to no avail.

How and why did you seek genetic testing?

In early 2023, I became involved in the VENTURE study at the Centre for Eye Research Australia. I was introduced to it via a Retina Australia webinar. My objective in participating was to identify the genetic cause of my RP.

It was a painless process involving a number of clinical tests and a blood sample, with a delightful, caring team – Janice and Justin were wonderful. My initial reservations about possible uses of my genetic information were well understood and made clear to me that it was not an issue. Unfortunately, my results were not determinative. I do not have any of the genetic spelling mistakes which are currently known to lead to RP. Further research is being undertaken to expand the knowledge of RP and its causes. I am very happy to add my data to the study and hopefully help researchers find additional genetic causes of RP.

Why do you recommend people participate in research?

Studies are crucial to research which lead to clinical trials and hopefully, the discovery of new treatments. You will also be contributing to science, helping others, and learning more about your disease. Recent breakthroughs reflect a real impetus in RP research. If you can, get involved in research such as the VENTURE study. If not for yourself, contribute for others around you and for future generations.

Research Project and Clinical Trial Register



Summary of Projects Currently Recruiting Participants

SUNDEW: ADOA Interventional Clinical Trial

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. Recruiting participants Australia wide. Sponsor: PYC Therapeutics
Contact: Sundew@pyctx.com

An Observational Clinical Trial of PRPF31 (RP11)

Disease: Retinitis pigmentosa **Participants:** Carriers, Patients

This study aims to observe the progression in patients with the inherited retinal disease (IRD) retinitis pigmentosa II (PRPF31 or RP11) over the period of four years.

Recruiting: Centre for Eye Research Australia, East Melbourne, Victoria and Lion's Eye Institute, Western Australia. Sponsor: PYC Therapeutics Contact: quokka@lexitas.com

Investigating the genetic basis of undiagnosed inherited retinal diseases

Disease: All inherited retinal diseases **Participants:** Patients

This study aims to investigate both known and new genetic changes that may be associated with inherited retinal diseases (IRDs). The researchers will search for genetic changes that could be linked to IRDs, but may have not been identified before.

Recruiting participants in Victoria. Sponsor: Centre for Eye Research Australia and the University of Melbourne Contact: IRD@groups.unimelb.edu.au

Perspectives on social wellbeing programs: adults with dual sensory impairment (DSI) and their communication partners

Disease: Dual sensory impairment - vision and hearing loss **Participants:** Family Members, Parents and Guardians, Patients

The purpose of the study is to understand:

- the impacts of dual sensory impairment (DSI) on social participation from your perspective as a person with DSI or a communication partner and
- your perspective towards participating in a health and wellbeing program.

Recruiting participants Australia wide. Sponsor: Macquarie University
Contact: sensoryloss.research@mq.edu.au

Assessing the caregiver experience for patients with inherited retinal disease (IRD) diagnosed in childhood

Disease: All inherited retinal diseases **Participants:** Parents and Guardians

The study aims to explore how caring for a person diagnosed with an IRD as a child impacts the carers' life and if the impact changes as the child gets older.

Recruiting participants Australia wide. Sponsor: University of Technology, Sydney
Contact: Maria.H.Kokoszka@alumni.uts.edu.au

Research Project and Clinical Trial Register



Summary of Projects Currently Recruiting Participants (continued)

Enabling Accessible and Inclusive Playgrounds for Children and Carers with Vision Impairment

Disease: All inherited retinal diseases **Participants:** Parents and Guardians, Health Professionals, Patients

This study will begin our work by investigating the experience of playgrounds for children with vision impairment and their parents/carers with the aim to determine a set of best practice guidelines for designing accessible and inclusive playgrounds that allow social inclusion and enhance community integration, belonging, health and wellbeing.

Recruiting participants Australia wide. Sponsor: University of Sydney

Contact: sue.silveira@nextsense.org.au

The Victorian Evolution of Inherited Retinal Diseases Natural History Registry (VENTURE) Study

Disease: All inherited retinal diseases **Participants:** Carriers, Patients, Family

The VENTURE registry collects retrospective and prospective clinical and genetic information from people living with an inherited retinal disease. Recruiting participants in Victoria.

Sponsor: Centre for Eye Research Australia, University of Melbourne

Contact: IRD@groups.unimelb.edu.au

The Australian Inherited Retinal Disease Register (AIRDR) and DNA Bank

Disease: All inherited retinal diseases **Participants:** Carriers, Patients, Family

The primary aim of the AIRDR is to characterise the genetic spectrum of IRDs in the Australian population in order to guide research into treatments and cures for IRDs.

Recruiting participants Australia wide. Sponsor: Sir Charles Gairdner Hospital, Perth, Western Australia Contact: SCGHMTP@health.wa.gov.au

Save Sight Institute IRD Registry

Disease: All inherited retinal diseases **Participants:** Carriers, Patients, Family

IRD management involves detailed ophthalmic structural and functional assessment.

Recruiting participants in New South Wales. Sponsor: The University of Sydney, NSW

Contact: ssi.operations@sydney.edu.au

For more project details, refer to our website at www.retinaaustralia.com.au.

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