Spring 2025

visionary

Hope in sight®

Next-generation gene therapy in Australia

Be a sport

Help advance breakthroughs in eye research

CENTRE FOR
Eye Research
Australia

HOPE IN SIGHT
GIVING DAY
9 October 2025

Kicking goals together

There are so many remarkable people who have joined CERA's cause, working together to end blindness from inherited retinal diseases (IRDs).

Twenty-eight-year-old Olivia Depares and 15-year-old Louis Shepard both live with Usher syndrome – a genetic form of deaf blindness – and are outstanding advocates for CERA's research.

While conditions like theirs were recently considered untreatable, we're now close to a host of new ways to protect the vision of people diagnosed with these diseases.

This includes the work of Dr Jiang-Hui (Sloan) Wang, who is coming back to CERA after a stint at a prestigious US lab to continue his research in Australia, bringing back new skills to accelerate gene therapy research.

This year, CERA's Annual Hope in Sight Giving Day is directly supporting his work, and I'm delighted that your donations, up to our $200,000 goal, will be matched dollar for dollar.

In this edition of Visionary you'll also read about Associate Professor Raymond Wong's work that has made exciting progress since CERA's community gave it a Giving Day boost in 2021.

I'm also proud to share that alongside my collaborator Dr Ceecee Britten-Jones, we've now helped 100 people with an unknown IRD undergo specialised genetic testing to find an answer to their vision loss.

We've come so far thanks to your support, but we still have work to do. Please consider making a gift on our annual Hope in Sight Giving Day on Thursday 9 October to drive research towards real-world treatments for vision loss.

Together, with your support, we can put hope in sight.

Professor Lauren Ayton AM
Deputy Director
Centre for Eye Research Australia

Centre for Eye Research Australia

+61 3 9959 0000
cera@cera.org.au
cera.org.au

CERA.eye
Centre for Eye Research Australia (CERA)

CENTRE FOR Eye Research Australia | THE UNIVERSITY OF MELBOURNE | the royal victorian eye and ear hospital

CERA gratefully acknowledges Australian Vision Research's support of our work and of Professor Keith Martin as Director of Research (Victoria).

Photo: Ben Jones

Championing eye research

Olivia Depares hopes a scientific breakthrough can transform sight as much as it has hearing.

Olivia Depares is a whip smart and capable young woman determined to live her life on her own terms and break down barriers for others with Usher syndrome.

A lawyer specialising in immigration law and a career in government, she's also a keen boxer.

"I think sports is so fundamental for us all – to be physically active and social builds confidence and friendships," she says.

Olivia was born with Usher syndrome 1B – a rare inherited genetic condition that causes profound hearing loss, a progressive decline in vision from teen to adult years and balance problems.

"It's only through living with the condition that you can truly understand how it affects all aspects of your life," she says.

Scientific breakthroughs have helped her keep her hearing and achieve her goals, and she is doing everything she can to advocate for research into vision loss.

Journey to diagnosis

When Olivia was only a few weeks old, her mother Jenny was suspicious that her daughter could not hear.

"Despite several visits to our then GP, these concerns were initially dismissed, but with the insistence of my mum, a referral to an

(Continued Page 4)

Photo: Ben Jones

ear, nose and throat specialist confirmed my profound hearing loss," says Olivia.

Olivia was fitted with powerful hearing aids at 11 months, but they were not enough for her to hear speech clearly. The only way she could hear speech was through a multi-channel cochlear implant.

Independently developed and commercialised in Australia, the multi-channel cochlear implant has transformed the lives of people with Usher syndrome as well as the lives of millions of people worldwide, enabling them to hear.

After months of auditory therapy, Olivia received a cochlear implant first in her right ear when she was 11 months of age, and in her left ear at nine years old.

"My cochlear implants have been crucial for me to learn to hear and then speak," Olivia says.

"They are certainly a part of me."

"They have given me the ability to pursue my goals and passions which would have no doubt been a lot harder to achieve had my parents not decided to proceed with cochlear implantation.

"Now I feel that it is my duty to do what I can to raise awareness, raise understanding, raise funds and raise hope, to give those living with eye disease the chance to improve their future."

But without research, people like Olivia with Usher syndrome will continue to lose their sight.

This loss of sight associated with Usher syndrome is caused by an inherited retinal disease (IRD) called retinitis pigmentosa (RP) – which is the degeneration of photoreceptor cells in the retina.

Children typically experience a gradual loss of their peripheral vision between 10 and 15 years. Then, as a young adult – from their mid-20s to early 30s – they progress towards becoming legally blind.

Fighting fit: Olivia and her mum Jenny are proud advocates for CERA research.

Olivia's field of vision is currently 10 degrees instead of the usual 180 degrees.

"I have no peripheral vision, and my sight is deteriorating slowly," says Olivia.

As a toddler Olivia's balance was also an issue, and at the age of four she showed the initial signs of night blindness.

"I was late to walk and had lots of therapy to encourage my gross motor skills," says Olivia.

"Mum also describes me freezing up when I found myself in a dark room or hallway.

"It was at this time that I was diagnosed with RP, the missing link completing the Usher syndrome diagnosis."

On her own terms

Olivia aspires to build on her already impressive achievements.

"I completed my secondary education in 2015 and then went on to complete a Bachelor of Laws and Arts (Communications) at Macquarie University.

"In 2021, I was admitted as a lawyer and spent some time working in immigration law before moving to my current role in government which has a focus on legislative amendments and policy reform.

"Away from work, I love to exercise with a regular combination of boxing and strength training as well as walking. I have also recently become involved in my partner's food truck business, which has been lots of fun, and hard work – venturing out on the weekends to regional NSW and Sydney surrounds."

Olivia's passion for advancing the health and rights of people with disabilities could one day also lead to her taking on an advocacy role: "I would love to move into the social justice field with a focus on disability policy and reform in the future," she says.

"I feel that it is my duty to do what I can to raise awareness, raise understanding, raise funds and raise hope, to give those living with eye disease the chance to improve their future."

– Olivia Depares

Future impact

There is currently no cure for Usher syndrome, but CERA's work into emerging gene therapies is giving Olivia hope.

"Comprehending and acknowledging the struggles people have with deaf-blind conditions is a step towards recognising the enormous importance of CERA's research into Usher syndrome.

"My hope is that by sharing my story, other people with Usher syndrome can have the confidence to be able to live a life without further limitations and like me can continue living independently and contribute to society to their full ability.

"I hope people consider this and donate what they can towards this worthwhile research. It is research that can have a life changing impact."

Photo: Anna Carlile

Genetic testing milestone

Five hundred and fifty people with an inherited retinal disease are now involved in the VENTURE project.

Cutting-edge research is making it possible to prevent vision loss caused by inherited retinal diseases (IRDs).

However, with over 300 genes linked to different inherited conditions, people often don't know the exact gene that is causing their blindness, and it leaves them unable to access emerging treatments.

The VENTURE Study, a partnership between CERA and the University of Melbourne, involves more than 550 people living with inherited retinal disease.

One hundred of them are now receiving an advanced test as the result of a partnership with The Advanced Genomics Collaboration (TAGC), which aims to find the exact genetic cause of their blindness.

The TAGC is a partnership between the University of Melbourne and biotech company Illumina.

"We are thrilled to be working with Illumina and TAGC to help develop other avenues for advanced testing for these people, in the hope that an answer may be achievable," says Professor Lauren Ayton AM, who co-leads the project at CERA alongside Dr Tom Edwards.

Expanding view

"The VENTURE registry is driving inherited retinal disease research, giving access to research opportunities for individuals with inherited retinal diseases."

Team work: Professor Lauren Ayton and Dr Alexis Ceecee Britten-Jones.

"With emerging treatments like gene therapy in the pipeline for people with an IRD, it is now more important than ever to solve the mystery for those people for whom standard testing has not revealed the cause of their vision loss."

VENTURE works alongside people with IRDs and their families to track the progress of their vision loss, as well as discovering if they are suitable for new treatments and clinical trials as they become available.

Under the leadership of ocular genetics expert Dr Alexis Ceecee Britten-Jones from the University of Melbourne, some VENTURE participants now have access to advanced forms of genetic testing to try and solve more complicated cases.

This testing is showing researchers and clinicians how to best use new genetic testing technologies for IRDs.

Professor Ayton says a specific diagnosis can also be very reassuring for a person with an inherited retinal disease.

"The value of genetic testing cannot be overstated."

Of great importance to the team is a collaborative approach to research with consumers, clinicians, researchers and industry that is driven towards better outcomes for people living with vision loss and blindness.

"We work incredibly closely with community groups like Retina Australia, UsherKids Australia and Blind Citizens Australia to make sure our work is relevant and needed for the community," says Professor Ayton.

"We also work closely with scientific and clinical colleagues across the globe to ensure the research has the highest impact."

Professor Ayton is a member of the Retina International Scientific and Medical Advisory Board, and CERA is also a site for the international Foundation Fighting Blindness clinical consortium.

"The value of genetic testing cannot be overstated."

– Professor Lauren Ayton

Getting involved with VENTURE

"We are keen to find more participants for clinical trials quickly, and to also know more about IRDs in this era of possible treatment," says Professor Ayton.

VENTURE has already been a valuable program for participants – from people who have answered surveys to those participating in more involved research.

"We have been able to offer genetic testing to many people, working in partnership with services such as the Ocular Genetics Clinic at the Royal Victorian Eye and Ear Hospital," says Professor Ayton.

"I would love to see a world where these conditions are considered curable, but we need more support to do that," explains Professor Ayton.

"I'm thrilled to see the impact of our research to date."

Photo: Anna Carlile

Next steps in restoring vision

Associate Professor Raymond Wong's research has taken its next step.

The retina at the back of the eye plays a crucial role in vision.

It's home to millions of photoreceptors – light-detecting cells that send signals to the brain which are turned into images.

But for people who live with retinal degeneration – a group of diseases that includes age-related macular degeneration, retinitis pigmentosa and Stargardt's disease – the death of these cells means a progressive loss of vision.

Once photoreceptors die off, they can't be restored, but cell reprogramming has the potential to change that.

Head of Cellular Reprogramming at CERA, Associate Professor Raymond Wong is taking major steps towards restoring vision by using cell reprogramming to help the photoreceptors grow back.

"We are working on a treatment to stimulate the stem cells in the eye to develop into new photoreceptors by injecting safe engineered viruses into the eye to deliver reprogramming genes," says Associate Professor Wong.

Crucial support

Associate Professor Wong's work would not have progressed to where it is today

← *Restoring vision: Associate Professor Raymond Wong in the laboratory.*

without the support of CERA's donors and the National Stem Cell Foundation of Australia.

"We received a major boost after donors gave generously to our Hope in Sight Giving Day back in 2021, raising $191,000 for our research," says Associate Professor Wong.

"It was a game changer for us, and we are so thankful."

This support gave his team the opportunity to undertake creative and 'big sky' planning as they scaled up their work towards the development of real world and accessible treatments for retinal degeneration.

"We would not be where we are today without our generous CERA donors who gave to us and continue to do so.

"With research at this stage, it's difficult to find financial support as it doesn't yet have a clear path to the clinic, but it is absolutely necessary for the discovery of treatments.

"Philanthropic support, whether it's large or small, makes all the difference," says Associate Professor Wong.

Major growth

In the discovery phase of the research, Associate Professor Wong received support from government and philanthropic organisations, including the National Health and Medical Research Council (NHMRC), Medical Research Future Fund (MRFF), National Stem Cell Foundation of Australia, the CERA Foundation, and Retina Australia.

After this phase was complete, CERA established the startup company Mirugen to secure commercial partners to support the research even further.

Last year, Mirugen received a $1.92m grant from biotech incubator CUREator+ through the MRFF.

CUREator+ is a national program funded by the Federal Government's Medical Research Future Fund and delivered by Brandon BioCatalyst and ANDHealth.

"Philanthropic support, whether it's large or small, makes all the difference."

– Associate Professor Ray Wong

This year, Mirugen secured $4.5m in seed funding from a consortium of investors through the University of Melbourne Genesis Pre-Seed Fund, Tin Alley Ventures and Brandon Capital.

"Mirugen is on the cusp of translating our research into a possible therapy that could transform lives by restoring vision, so this funding comes at a crucial time," says Associate Professor Wong.

Securing these partnerships is a huge moment for moving the research closer to the clinic.

"With this support, it shows that there is hope from many people to make this treatment available to patients as soon as possible."

Photo: Anna Carlile

Gene editing for the next generation

Gene editing that could help halt or even reverse vision loss in people like Louis Shepard with Usher syndrome 1B.

Louis Shephard, like all people born with Usher syndrome 1B, has experienced profound hearing loss, balance problems, and a progressive decline in vision throughout his life.

"But my vision is not the only thing about me – it does not define me," he says.

"What does define me are my personality traits, such as being honest, having integrity, and my hobbies such as playing sports like cricket, or gaming, reading a book."

Louis received two cochlear implants – one at 11 months old and then one at 12 months old – which today lets him hear the ring of the ball used in blind cricket as it spins down the pitch to him, ready to hit.

These cochlear implants are crucial for Louis to play as he currently has 15 degrees of peripheral vision compared to the standard 180 degrees.

Medical breakthroughs have helped Louis keep his hearing, and new technologies in gene therapies are pointing towards making similar achievements in protecting vision possible.

RNA editing

To create and develop treatments for inherited retinal diseases (IRDs), one of the exciting technologies being used by CERA's Head of Genetic Engineering Research Associate Professor Rick Liu and his team is the Clustered Regularly Interspaced Short Palindromic Repeat (CRISPR)-Cas System.

Future focus: (L-R) CERA advocates Emily and Louis Shepard, and Associate Professor Guei-Sheung (Rick) Liu.

CRISPR is a technology that makes it possible to correct genetic errors in RNA molecules – the carriers of genetic information for all living cells in the body.

Now researchers are using this technology in the lab to develop treatments which could one day edit RNA directly in the patient's retina, correcting faulty genetic messaging and helping retinal cells function properly.

"Finding treatments and cures for inherited retinal diseases, like Usher syndrome, caused by large-sized gene defects, is urgent and at the forefront of our advancement in the field of research," says Associate Professor Liu.

"We don't have time to waste – early intervention for inherited retinal diseases before anything happens to the retina of a patient is an important focus.

"CRISPR technology is ready, and now we must continue to advocate for support, raise awareness, and secure further funding to turn this breakthrough into a real solution for those in need."

Finding treatments

Because it does not permanently alter DNA, RNA-targeted approaches provide a safe way to correct genetic errors. It can also target diseases caused by both large and small genes.

"Finding treatments and cures for inherited retinal diseases like Usher syndrome is urgent."

– Associate Professor Rick Liu

"This means that the treatment won't be limited by the size of the mutated gene that causes the disease," Associate Professor Liu says.

"It will significantly advance CERA's research efforts to delay or cure eye diseases caused by mutations in large genes, such as Usher syndrome.

"With over 300 genes known to be associated with inherited retinal diseases, affecting over 2 million people worldwide, customised gene editing that achieves safe and effective treatment to prevent vision loss is our priority," he says.

"We are excited by the possibilities CRISPR offers and the real-world impact it could have for individuals like Louis."

As Louis Shepard says: "It's amazing that there is now research that could stop people from losing their sight from Usher syndrome – and one day help someone like me have their full 180 degrees of vision."

Gene therapies' new home

A new CERA lab led by Dr Jiang-Hui (Sloan) Wang is bringing a cutting-edge technique for developing gene therapies to Australia.

Dr Jiang-Hui (Sloan) Wang is a rising star in eye research and is bringing his cutting-edge gene therapy capabilities back to Australia from the United States.

His new lab at CERA will be one of only a handful in the world capable of developing new viral vectors to treat inherited retinal diseases (IRDs) that involve large genes, like Usher syndrome.

As well as his own projects, Dr Wang's work is anticipated to help accelerate research across CERA that is aiming to develop gene therapies for several sight-threatening conditions.

Mutations in the MYO7A gene – which direct the making, development and function of photoreceptor cells in the retina – is the major cause of Usher syndrome type 1B.

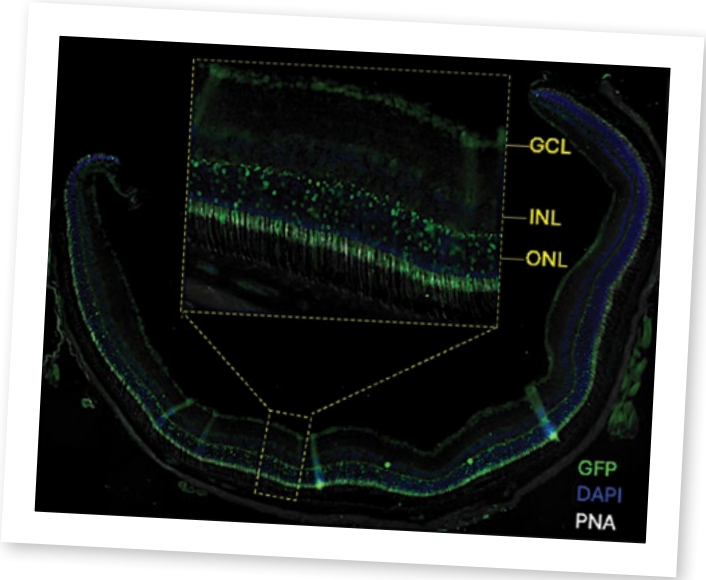
It leads to profound hearing loss, a progressive decline in vision from teen to adult years and balance problems.

Safer procedures

IRDs that involve large genes like Usher syndrome were once considered untreatable, but gene therapies that can correct faulty genes and prevent vision loss are beginning to emerge.

← *Dr Jiang-Hui (Sloan) Wang returns to Australia.*

→ *The split green fluorescent protein is reassembled in the deeper layers of the mouse retina.*



These therapies often require delicate, invasive injections of harmless viral vectors – known as adeno-associated viruses (AAVs) – to be delivered beneath the retina carrying the corrected gene.

However, there is a limit to how much AAVs can carry.

"The MYO7A gene that causes Usher syndrome is too large for a single AAV to deliver to fix the mutations, and previous attempts to deliver the genes in two AAVs has had mixed results," says Dr Wang.

Whilst the AAVs themselves are safe, the deep injections that deliver them to the eye have the risk of damaging the very cells that they're meant to save.

By contrast, the simple eye injections used for common conditions like macular degeneration, are much safer but can't penetrate deep enough to reach the light sensing cells.

It's like having a lifesaving medicine that stops at the gate and never gets delivered.

That's why researchers consider it the 'holy grail' to engineer AAVs capable of traversing the eye's natural barriers via minimally invasive injections.

Greater view

Dr Wang's revolutionary approach uses a less invasive injection to deliver the full MYO7A gene into the eye.

By splitting the large MYO7A gene and reassembling it inside the retina using a specially engineered AAV vector he developed, his aim is to reverse the effects of retinal disease in a mouse model of Usher syndrome.

Having developed techniques and processes under the mentorship of Professor Guangping Gao at UMass Chan in Massachusetts, United States – one of the world's leading experts in AAV and gene therapy – Dr Wang is now bringing cutting edge technology back to Australia for further development and eventual clinical use.

Dr Wang is deeply appreciative that his work is being supported by CERA's community of donors.

"These generous gifts will make a meaningful difference in accelerating our efforts to develop gene therapy for Usher syndrome.

"If successful, this method could offer a safe and efficient way to treat other inherited retinal diseases with large genes, using the same injection approach, and will benefit teams working to develop gene therapies across CERA, including Associate Professor Rick Liu's Usher RNA editing research. This support comes at a pivotal moment, as I prepare to return to Australia from the US and establish my new lab. I'll be able to hit the ground running and continue pushing forward research that we hope will change lives."

Celebrating our donors

We're deeply grateful to the late Marjorie Roberts, who has left a gift in her will to support CERA's research.

Marjorie Roberts' hope was that eye problems could one day be cured.

Born into a pioneering family and living on a wheat farm in Kunat (just south of Swan Hill), conditions were often harsh for young Marjorie and her family.

She attended boarding school until she was nine years old, and when her father died, her mother, sister and Marjorie continued working the family farm. In a statement, her family wrote to CERA: "She was an inspirational example of strength and resilience."

Marjorie and her husband Ken both grew up during the Depression.

"They struggled," said the statement. "But they both felt fortunate to be able to get good jobs, work hard, and raise us in the family home. They also both had a keen sense of duty to help those less fortunate than themselves."

Driven by their strong Christian faith, both Marjorie and Ken were, "strongly and passionately involved in the activities of their local church, helping in the community, volunteering within many organisations and regularly donating to many and varied charities throughout their lives.

"They were generous in their donations to overseas, poverty-stricken areas and inspired by the impact of treatments on improving quality of life across the world."

"Mum started wearing glasses for short sightedness as a 12-year-old and had cataract surgery in her later years. One of her children also struggles with poor eyesight. Mum was sad when she could no longer clearly see the beauty of the bush, and it was her hope, that eye problems could one day be cured."

We thank you Marjorie, for choosing to leave a gift in your will to CERA and bring hope to people affected by vision loss and blindness.

"We are so extremely proud of the life she has led and the example she has left us. We were not at all surprised by the generous donations in her will to CERA."

A gift in your will leaves a legacy of sight for future generations

For a free copy of our ***Gifts in Wills*** information pack, or if you would like to chat to us about leaving a gift in your will to CERA, please contact us on **1300 737 757** or **giftsinwills@cera.org.au**

cera.org.au/shine

CENTRE FOR
Eye Research
Australia

Help us hit blindness for a six on Hope in Sight Giving Day

HOPE IN SIGHT
GIVING DAY

Donate on Thursday 9 October and see your impact doubled!

Help us raise $200,000 for research

Every dollar you give up to $200,000 will be doubled by a generous anonymous donor

$20 → $40
$100 → $200
$200 → $400

CENTRE FOR
Eye Research Australia

Sign up at charidy.com/HopeinSight