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visionary

SPRING 2019

Hope for the future
Sisters share their stories of living with vision loss

Testing times
Navigating the personal decision to take part in genetic research

CENTRE FOR
Eye Research
Australia

Welcome to the Spring edition of Visionary.

Centre for Eye Research Australia

T +61 3 9929 8360
E cera@cera.org.au
W cera.org.au

CERA.eye

EyeResearchAus

Centre for Eye Research Australia (CERA)

Nothing inspires the CERA team more than making a difference for people experiencing vision loss and blindness. In this edition, we meet two remarkable women, Diana and Sarah, who share their stories of living with Stargardt's macular dystrophy to raise awareness of inherited eye diseases and the importance of research.

We also feature Dr Tom Edwards and his gene therapy research; orthoptist Lisa Kearns explains what families need to consider before taking part in genetic research; and Dr Srujana Sahedbjada outlines her keratoconus research.

We are entering an exciting new era for vision research, with new gene and cell therapies and other technologies offering the potential of treatment for diseases previously considered 'untreatable'.

We could not do this without our many generous supporters including the many people who donated to our 2019 Tax Appeal. Your support brings us one step closer to developing these life-changing new treatments in Melbourne.

Finally, if you want to learn more about the future of eye research, you can attend our free community forum on World Sight Day on 10 October. I look forward to meeting you there!

Professor Keith Martin

Keith Martin

CENTRE FOR Eye Research Australia

the royal victorian eye and ear hospital

THE UNIVERSITY OF MELBOURNE

Gene therapies bring new hope for treatment

Inherited retinal diseases (IRDs) often strike people in the prime of their life, leading to irreversible vision loss or blindness.

The diseases are caused by genetic 'mistakes' which cause the cells of the retina to malfunction – and for many decades patients have been told there is no treatment.

But now the accelerating international pace of gene therapy research is bringing new hope that treatments to stall vision loss, or even partially reverse damage, may be on the horizon.

In Melbourne, CERA Principal Investigator Dr Tom Edwards' team is investigating a gene therapy for a particular inherited retinal disorder that is common in our region.

"We're looking at one particular gene and developing a strategy to introduce a correct copy back into the affected retinal cells," he says.

"We've had early success in delivering our gene of interest into cells grown in the lab, which has given us plenty of encouragement."

Dr Edwards hopes that, ultimately, he will be able to bring his research to a clinical trial in Melbourne.

"It is an exciting time, we have seen the first approved gene therapy in the US for a variety of retinitis pigmentosa and over the next few years we hope to conduct trials for IRDs here in Melbourne."

Dr Edwards says that Melbourne, with its unique combination of vision research and clinical expertise at CERA, the University of Melbourne and Royal Victorian Eye and Ear Hospital, is well positioned to become a Centre for Excellence in Ocular Gene and Cell Therapy.

"Our combined expertise at CERA, the University of Melbourne and the Royal Victorian Eye and Ear Hospital, can give Australians access to world-class research into inherited retinal diseases, and as research progresses, we may develop techniques that can be applied to more common eye diseases with a genetic basis such as glaucoma and age-related macular degeneration."

Testing times

After the initial shock of diagnosis, CERA's Lisa Kearns provides information and support to people with inherited eye disease deciding whether they will participate in research.

After being diagnosed with an inherited eye disease, the decision to participate in research is deeply personal.

"Often people contact us shortly after they have been diagnosed because they have been told they have an inherited eye condition with no effective treatment or cure," says research orthoptist and associate genetic counsellor Lisa Kearns.

For the past 16 years, Lisa has been the first point of contact for people who contact CERA's Clinical Genetics Unit because they want to learn more about their condition or participate in research.

The CERA team has long been involved in research to discover genes that cause inherited eye conditions. This information is used for research led by Principal Investigator Professor Alex Hewitt of CERA and Professor Alice Pébay of the University of Melbourne who are utilising cutting edge stem cell technology and gene-editing techniques in the hope of developing new treatments.

Lisa recruits participants for research into inherited retinal disease, inherited optic nerve disease, congenital cataract, strabismus, familial retinal detachment and glaucoma. She works closely with Professor Hewitt, and honorary fellows Professor David Mackey and Dr Jonathan Ruddle.

She says when patients decide to participate in research it's vital they have all the knowledge they need to make informed consent.

At the first meeting, Lisa works with the participants to put together a family tree, showing how family members are related, and who else could be affected with the same eye condition.

"Often having an eye condition can be overwhelming, and this meeting provides an opportunity to talk it through," says Lisa.

Patients who consent to research provide a blood, saliva or skin sample which is sent to the laboratory for testing.

"Finding the specific genetic cause of eye conditions can be useful for families but it is a complex process that requires working with experts in Australia and overseas, and sometimes the results can take a very long time," she says.

"If the cause is confirmed, this can provide a more definite diagnosis, and lead to a better understanding of whether the eye condition is likely to remain stable or deteriorate.

"The inheritance pattern can be difficult to predict based on family history alone. Knowing the genetic cause can help establish the correct inheritance pattern and likelihood of children or other family members developing the eye condition.

"Some people become anxious about going blind or knowing that loved ones can be susceptible.

"Some patients describe the emotional burden or the disappointment of learning a genetic cause cannot be found or that results are inconclusive. At CERA we have a highly experienced research team able to counsel participants."

Lisa says research has helped families to be referred to clinical genetics services to confirm results, have other family members tested or have informed family planning discussions. In Victoria, participants can be referred to the Ocular Genetics Clinic (OGC) at the Royal Victorian Eye and Ear Hospital.

Lisa says approval of the first gene therapy for an inherited retinal disease caused by a rare gene paves the way for other clinical trials which may come on line in the next few years.

"It is very exciting to see advances in genetics, upcoming gene and stem cell-based therapies entering clinical trial stage and beyond to potentially improve the quality of life for people with inherited eye disease," she says.

"But while researchers are working hard to develop treatments and make sure they are safe and effective before they are used in humans, we need to ensure that people are making the most of the vision they still have with aids, adaptive technology and by utilising support services.

"It's also very important for them to keep getting regular eye checks, so they can be checked for other eye conditions."

The Clinical Genetics Team receives funding from the Ophthalmic Research Institute of Australia (ORIA), Retina Australia, Glaucoma Australia, National Health and Medical Research Council (NHMRC) CRE Grant, Australian Stem Cell Therapies Mission through the Medical Research Future Fund and the Foundation for Children, Sydney.

Photo by Anna Carlile

Diana's story

Diana Cupido, 37, has lived with Stargardt's macular dystrophy – an inherited retinal disease which causes a serious loss of central vision – for almost 20 years. She is sharing her story to raise awareness of her disease and why research is important.

You can read more of Diana's story at **cera.org.au/dianas-story**

Photo by Anna Carlile

I had normal vision until I was 18. I was doing a Diploma of Advanced Tourism and I started to notice that I couldn't read things or recognise people, I'd wave to people thinking they were someone else.

I didn't know at that stage that it was anything serious, but I would read, and words would disappear before my eyes and I thought I just needed to see someone to get glasses and that would fix it.

I soon found out that glasses would not fix it and I was sent for a lot of tests. I was diagnosed with Stargardt's disease which typically happens in the teenage years. From there it spiralled and got worse very quickly and I lost most of my central vision.

None of the older people in my family – my parents or aunts and uncles – had any vision problems. It was just a combination of genes that came from my parents – and it has also affected two of my sisters.

Now my vision is all blurry in the centre, the detail is all confused and I use my peripheral vision to compensate. I have flashing sensations and motions that make it difficult to deal with glare and bright lights in places like shopping centres.

Overcoming fear

At the time I was dealing with what was happening with me, with so much fear about not having the life I thought I was going to have. You go from being a healthy 18 or 19-year-old to dealing with accepting what is happening to you and realising there is no cure.

I spent a lot of times in the early days thinking that I would be different, that my vision wouldn't get worse and for the next four or five years I made it my mission that there would be a miracle. It wasn't until I was in my late 20s that I really accepted it.

There are things that people normally do that I can't. I can't drive, I can't enjoy beautiful scenery, or a landscape or a sunny day like I used to.

For me, having a vision impairment has been a roadblock to my chosen career. I wanted to be a flight attendant. I was selected by an international airline and travelled overseas for the training and failed the medical and that was a huge blow. I speak several languages, I am well-educated, and I have a tourism degree but because of my vision I could not be a flight attendant. I also considered being a mid-wife but decided because of my vision, and finding it difficult to read charts and monitors it would be too dangerous.

Making the most of life

I have learned I have to make the most of life and look at how much vision I do have. I think about people who have no vision and how difficult and challenging life is for them.

I have two small children and it is the best job I have had! They are too young to understand my vision impairment, but it does not affect my ability to look after them. The only thing is I have to rely on others if we need to drive somewhere.

I have learned to live in hope not in fear, I am not waiting for a cure to be happy. I think of how a cure or treatment could help other people, people who have less sight than me and the next generation.

I am a glass half full person and I am grateful for the sight I have got, but I wouldn't want anyone else to have to live through this challenge.

Sarah's story

Sarah Ceravolo, 32, has forged a successful career as a designer. She is telling her story to give others hope.

I was diagnosed with Stargardt's macular dystrophy in 2012. I had suspected that I might have it because my sister had been diagnosed with the same condition years earlier.

While sitting in the State Library sketching inside the domed reading room, I attempted to read a plaque fixed to the desk. The words bent towards the middle and that was when I first became aware I was developing Stargardt's.

I often say this is one of the best times to be vision impaired as the adaptive technology available can help a lot – my iPhone and Siri work as an incredible aid, to the point, ironically, I almost go weeks without thinking about or noticing my condition.

I started out as a metal-smith and finished my academic career with a Master of Architecture and still had pretty good vision. At the time I was studying, I learned to use digital design tools and was a very capable 3D modeller – I haven't diverged from that path. Now, I use a digital magnifier to read and my 3D models rotate and enlarge so I am able look at them in detail.

Powered by creativity

In designing, my imaginative skills are very important – my results are not compromised by my impaired vision as the digital tools compensate for this. My creativity also applies itself to my adaptive nature in finding my way around my ailment, in a way, my eyes act really just as the middle man.

Some people don't know I am vision impaired - I have reached a point in my design career where I realise it is not a hindrance and often others don't even notice it. I haven't used it as a crutch. It has pushed me further.

I joined the board of Retina Australia (Victoria) soon after being diagnosed as I wanted to help others. I converted my personal experience into a tool as it would feel less like a burden if it became purposeful in facilitating me to help raise funds and create awareness.

One of my projects on behalf of R.A.V was the creation of a design studio module for the Product Design course aimed at vision impairment. I gave a lecture to students on the condition and where personal aids are lacking in this area. The students were super excited to participate and produced great designs as they felt it had such purpose.

Inspired by others

I know there are a lot of people who are worse off than me and I can rest assured that I will never be completely blind. I am inspired by so many people in the vision-impaired community and what they achieve.

I remember my sister Diana working hard to learn four languages. When I saw her tenacity, I knew I could do it too. People like Leighton Boyd (the Chair of Retina Australia) – who has worked all his life, now maintains the charity and looks after his

grandchildren, and so many other amazing vision impaired people I have met.

My vision appears as a mild version of what you see when you stare directly into the sun and look away – you try and wait for your eyes to recover from the bright light – it's not a black dot.

Some days I look at landscapes and remember how it felt when something was breathtaking on the horizon, now I don't have that same feeling – there is nothing to substitute for that exact moment, but you find other ways of seeing beauty.

Supporting eye research

My whole immediate family had a blood test to look at our genetics and help with research. Even if in this lifetime there is no cure, the only way we can find one is through assisting the research.

There is a lack of awareness about the disease and you are told all the time that there is nothing that can be done for you.

When you are first diagnosed and try to seek more information, you turn to Google and the first words you see are so disheartening and unhopeful.

I do think I will see a successful clinical trial that will help me in my lifetime but this could potentially take decades. I believe there will be genetic therapies which will help people who are younger and prevent them from losing their vision. I think it is something for people to look forward to and knowing this makes me happy and hopeful for them.

You can read more of Sarah's story at **cera.org.au/sarahs-story**

Photo by Anna Carlile

Be part of the future of macular research

CERA is devoted to discovering better treatments for age-related macular degeneration. To do this, clinical research is essential – and the patients who participate play an invaluable role.

As we age, our eyes go through some changes. For one in seven people over the age of 50, this includes signs of age-related macular degeneration (AMD).

The macula, the small central area of the retina at the back of the eye, is responsible for our sharp central vision. AMD occurs when the light-sensitive cells of the macula gradually break down, leading to blurred central vision. This can make it difficult to read, recognise faces, drive and perform other everyday activities.

As the disease advances, vision loss can become more serious. AMD is the leading cause of legal blindness and severe vision loss in Australia, responsible for 50% of all cases.

"Our research aims to improve our understanding of macular degeneration and discover treatment options," says Professor Robyn Guymer AM, Head of Macular Research and Deputy Director at CERA.

"We have a good treatment for wet AMD, the most devastating form of the disease, where vision loss can be sudden and dramatic – though there are still trials to find better treatments.

"But there is currently no treatment for dry AMD, which develops slowly and results in gradual vision loss.

"That's why clinical trials are so important – that's what will help us find an answer."

Macular research at CERA

The Macular Research team at CERA is investigating many aspects of the disease – such as genetics, environmental associations, biomarkers of AMD, and the risk factors for progression from early AMD to advanced.

A large recent study, the Laser Intervention in Early Age-Related Macular Degeneration (LEAD) trial, studied 300 people with AMD and found promising results using nanosecond laser technology to slow the progression of the disease. Future trials will continue the investigation of new laser treatments.

"We're also interested in following people with macular degeneration over time to learn more about who is at the greatest risk of losing vision," Professor Guymer says.

"For this type of research there is no treatment involved, but we monitor people with a range of novel instruments that aren't widely available elsewhere."

Why take part in a clinical trial?

By getting involved in a clinical trial, you can play an important part in the future of eye health.

"Clinical trials help us advance our knowledge of the disease, so that we can help more people with better treatments in the future," Professor Guymer says.

"The diseases we look at are often inherited, so a lot of people join our clinical trials with the idea that they can contribute to discoveries that may help their family members one day."

Clinical trials can also provide access to new interventions before they are widely available, in many cases participants may receive a treatment that could help their condition.

"We'd love to hear from anyone with AMD," Professor Guymer says. "In particular, we're looking for people with dry AMD, and people with high risk early disease, who haven't lost vision yet but have drusen, which tells us someone is at a greater risk of vision loss from AMD."

You can register your interest in clinical trials on the CERA website. The more information you can provide about your eye condition and history, the easier it is for CERA researchers to determine if there is a trial you might be suitable for.

To find out more about clinical trials at CERA and register your interest online, visit **cera.org.au**

Photo by Anna Carlile

A closer look at keratoconus

New research aims to uncover crucial insights into the genetic causes of keratoconus, a corneal condition that affects young adults and children.

The cornea, the front window of the eye, plays an important role in focusing our vision. In healthy eyes, the cornea is clear and curved in a dome shape, allowing light to enter the eye.

For people with keratoconus, the cornea gets progressively thinner, developing a cone-shaped bulge that distorts vision. As the disease advances, vision continues to deteriorate, and a corneal transplant may be required to restore vision.

"Keratoconus predominately affects teens to those in their early adulthood, when these young people are supposed to be living some of their best years," says CERA Research Fellow Dr Srujana Sahebjada.

"Although the prevalence of keratoconus is increasing, there is still a lot we don't know about the disease, its causes and the ways to prevent the condition. I am endeavouring to change that."

To uncover insights into the genetic causes of keratoconus, Dr Sahebjada is leading a new study that will examine the corneas of people with the condition.

Dr Sahebjada will collect the diseased tissue, which is usually discarded during a corneal transplant, and study the genetic material in each of the different corneal layers using advanced genetic techniques.

"This gives us a comprehensive look at the corneal tissue to help us try to identify which layers are being affected and where the disease is originating," says Dr Sahebjada.

In the long term, this work could provide insights that help identify high-risk patients and stop the progression of the disease to more advanced stages, hopefully reducing the need for corneal transplants.

"We are doing such important work at CERA and have achieved some significant advancements, but I'm determined to continue to strive towards improving the lives of those with keratoconus."

This research is supported by a significant grant from the Perpetual 2019 IMPACT Philanthropy Application Program.

Leading a global effort

CERA researchers have formed the Keratoconus International Consortium (KIC) – bringing together 40 keratoconus research groups around the world to create a shared database. This work at CERA is supported by the Lions Eye Donation Service and led by Dr Srujana Sahebjada, Professor Paul Baird and Associate Professor Mark Daniell.

"We hope that by collecting unified research data and sharing it with one another, we can develop a globally accepted classification system to monitor the disease progression and promote a more consistent approach to diagnosis and treatment," Dr Sahebjada says.

Backing a bold idea

The willingness of Australian philanthropists to back an idea 'out of left field' will see an innovative eye test to detect Alzheimer's disease trialled on patients this year.

The work of CERA researchers Associate Professor Peter van Wijngaarden and Dr Xavier Hadoux hit the headlines in June, when they received funding through the Alzheimer's Drug Discovery Foundation's Diagnostics Accelerator program.

The foundation – backed by a group of US philanthropists including Bill Gates, Leonard Lauder, Jeff and MacKenzie Bezos and the Dolby Foundation – funds innovative ways of diagnosing and treating Alzheimer's disease.

The funding will fast track CERA's research, enabling the technology to be tested on participants in the Healthy Brain Project, an innovative study conducted by our colleagues at the Florey Institute of Neuroscience and Mental Health.

Associate Professor van Wijngaarden says the eye test – which uses technology similar to that utilised by satellites – has the potential to revolutionise the diagnosis of Alzheimer's disease.

However, it's been a long research journey and recent advances could not have occurred without the initial generous support of Australian philanthropists Baillieu Myer, Samantha Baillieu and Jeanne Pratt via the Yulgilbar Alzheimer's Research Program. Other vital support has also been provided by the HL Hecht Trust, managed by Perpetual, the Viertel Foundation, managed by Equity Trustees, the Joan Margaret Ponting Trust, Coopers Brewery Trust, Cylite CEO Steve Frisken and National Foundation for Medical Research and Innovation.

Yulgilbar Foundation Chair Samantha Baillieu AM says the Yulgilbar Alzheimer's Research Program – an initiative of her parents Sarah and Baillieu Myer – is proud to have been involved with the project from the beginning.

"Associate Professor van Wijngaarden first came to the attention of Yulgilbar's scientific advisors and, with another philanthropist, we assisted with the purchase of the non-invasive retinal scan camera," she says.

"The appeal of funding this project was firstly, the camera was not available to patients in Australia and secondly, we knew that through Peter and his team's skills that the research could be taken all the way through.

"It was real, practical and offered an insight that otherwise had not been available, this resonated with our approach and ethos."

Sylvia's gift

CERA is grateful to the late Sylvia Gelman AM MBE (1919 – 2018) – teacher, activist and philanthropist – for her generous bequest for eye research.

A dedicated teacher, a women's rights advocate, a lover of the arts and sports – Sylvia Gelman lived a full and passionate life and made a lasting impact on all those around her.

For 20 years, Sylvia taught at Mount Scopus Memorial College, where she became a deeply respected Headmistress. She served as president of the National Council of Jewish Women of Australia (NCJWA) and was made Life Governor of NCJWA in 1988.

"She taught so many of the Jewish community," says Graham Solomon, Sylvia's nephew. "She, along with her husband, helped Jewish refugees who migrated to Australia after fleeing the Holocaust in Europe.

"Whenever I come up against a problem, I think to myself, how would Aunt have solved this? I will always remember all the successful people who came up to Sylvia and said, 'Without you, I would not be where I am today.'"

Sylvia's service to education, youth, the Jewish community and women's rights has been recognised with honours including an Order of the British Empire (MBE), a Member of the Order of Australia (AM), the Queen Elizabeth II Silver Jubilee Medal, induction into the Victorian Honour Roll of Women and the JCCV General Sir John Monash Award.

Sylvia began to take an interest in eye health when her sister, Peggy Solomon, developed age-related macular degeneration (AMD). Later on, her nephew Graham was diagnosed with early stage glaucoma, and Sylvia herself developed AMD.

All this led to Sylvia's decision to include a gift in her will to CERA among her many generous bequests.

Sylvia's generous gift will enable the purchase of a new OCT scanner, which is critical for CERA researchers, in particular those researchers investigating AMD, its early diagnosis, causes and new treatments for AMD.

On behalf of everyone at CERA, we are eternally grateful to Sylvia for leaving a gift in her will to CERA.

If you're considering leaving a gift in your will to CERA, please call our Donor Relations Advisor, Elaine Levine, on (free call) 1300 737 757 for a confidential discussion.

You can read more about Sylvia's generous gift at **cera.org.au/sylvias-gift**

What's On...

SUNDAY 6 OCTOBER | 10:30AM-4:00PM

2019 Victorian Seniors Festival – Celebration Day

Visit our stall at this year's Victorian Seniors Festival. Talk with CERA staff about the latest developments in eye research and take home a goodie bag!

Federation Square, Melbourne

No bookings required

WEDNESDAY 9 OCTOBER | 1:00PM – 1:30PM

2019 Victorian Seniors Festival Hub – Information Session

Set your sights on healthy vision presented by the Centre for Eye Research Australia.

Melbourne Town Hall

No bookings required

THURSDAY 10 OCTOBER | 10:30AM – 1:15PM

Looking to the Future Community Forum

Hear from Managing Director Professor Keith Martin and Deputy Director A/Prof Peter van Wijngaarden about gene and cell therapy and the latest in artificial intelligence and new technology in eye research.

Royal Australasian College of Surgeons, East Melbourne

RSVP essential

THURSDAY 21 NOVEMBER | 5:15PM – 7:30PM

Gerard Crock Lecture

A/ Prof Penny Allen will present on the Bionic Eye project.

Kenneth Myer Building, University of Melbourne, Parkville

RSVP essential

CONTACT US TO RESERVE YOUR PLACE NOW:
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