SEPTEMBER 2022 ISSUE 24

RETINA AUSTRALIA QUARTERLY

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Welcome to the spring edition of Retina Australia Quarterly 2022. As we gradually emerge from the restrictions imposed as a consequence of the coronavirus pandemic, and the winter viruses, it is exciting to be able to report on our achievements during recent months.

As you would be aware, this year Retina Australia held its 24-hour Matched Giving Day on Tuesday 21 June 2022 to raise money for research. Thanks to the very generous support of our members, their family and friends, as well as donations from the broader community, I am pleased to announce that over $75,000 was raised this year. These funds will be utilised for investigations into inherited retinal disease being undertaken by successful recipients of Retina Australia research grants in future years.

Thank you to all members who have renewed their membership for this current financial year. Many of you have been able to access the new online system through Vega and, although there is more work to be undertaken behind the scenes, already this process has streamlined the collection of payments for staff and established a more straightforward method of contact for our members. If you still wish to pay your subscription, please do so at your earliest convenience. For those members who don’t have access to the online environment please contact the office on Tuesdays or Thursdays between the hours of 9:30am and 3:00pm, as all payments can be completed over the phone.

Work has continued on the establishment of the new website which will be fully accessible and provide a new modern and professional online presence for the organisation. The design for the front page of the website is now finalised, a new colour palette has been chosen, and a number of other page designs are well advanced. The next stage includes building the actual site as per the structure developed earlier in the process. I am hopeful that all work will be completed soon, and that the website can be launched by the end of October.

Congratulations to Jeremy D'Souza, former Director of Retina Australia, who has been elected to the Board of Retina International. Jeremy was nominated by Retina Australia for this position, and we wish him well as he works with colleagues from across the globe to support and enhance the activities of the Retina International membership which includes many patient-led organisations like us.

In June, the Retina International World Congress took place in Reykjavik, Iceland. Members present reported that it was an incredible experience to have so many people in the retina community come together from around the world, after almost two years of virtual meetings.

The Congress started with two days of engagement with the Retina International Youth Council. Twenty-three young people, including Jane Cherry, one of our directors, met in person to discuss how the Retina International strategy could address their specific priorities. Next was the Continuous Education Programme which brought delegates together to present and exchange experiences, share learnings and discuss the future path of Retina International. Following these activities were two days of presentations on research developments at both the scientific and lay sessions of the Congress. For the first time, members of the Retina International executive team presented the results of their own in-house research.

If you are interested in reading more about this Congress, a full publication is available on the Retina International website:Final-RIWC-2022-Publication.pdf (retina-international.org) The next Retina International World Congress will be held in Dublin, Ireland in 2024.

The 38th Annual General Meeting (AGM) of Retina Australia will be held on Saturday 15 October 2022 by Zoom video conference commencing at 1:30pm (AEDT).

If members do not have computer access, it is possible to participate by phone. The agenda will include the presentation of the 2021-2022 Retina Australia Annual Report and the audited financial statements. An election of Directors will take place if required. Further details will be circulated by late September to members entitled to vote at the AGM. If you have questions about this process, please do not hesitate to contact the office.

The next webinar will be held in conjunction with the AGM and commence at 2:00pm (EDST). Information about this webinar and details of how you can join, will be circulated by email, social media or in print to all members soon.
Thank you for your continued membership and support,

Leighton Boyd
Chairman

Research Reports 2021

Final reports from our research grant recipients for 2021 are now available to view on the Retina Australia website. These brief reports provide an insight into the projects that you have funded through your generosity. You can view them here: https://www.retinaaustralia.com.au/research-reports/

Applications for the 2023 grants round are currently being considered by the Grants Advisory Committee – the successful projects will be announced later this year.

Giving Day Success

Our Second Giving Day was held on Monday 21st June, and we were delighted to raise over $75,000 in 24 hours! Thank you to all our supporters for your generous gifts, which will all go towards supporting the very best Australian researchers in their work searching for a cure to inherited retinal disease.

Membership Reminder

It’s not too late to renew your membership! While you can receive this newsletter and attend our webinars for no cost, becoming a member of Retina Australia is a great way to support our operations.

There are two options:

- Member ($50), which entitles you to vote at the AGM and also to be eligible for election to the Retina Australia board,
- Associate ($30), which provides valuable support to the organisation and our operations.

You can renew now by clicking on the link below, or you can call the Retina Australia office on 03-9650 5088 between 9.30am and 3.00pm on Tuesday or Thursday.
https://www.retinaaustralia.com.au/membership-renewal/

Free Webinar Series 2022

The latest in research from Janssen

Date: Saturday 15th October
Time: 2.00pm AEDT (1.30pm in SA, 12.30pm in NT, 1.00pm in QLD and 11.00am in WA), following the Annual General Meeting.

Retina Australia is delighted to have the support of Janssen, who have recently provided generous funding for the redevelopment of the Retina Australia website, which will go live in October, as well as the introduction of Vega, an exciting new database system that has transformed the way we are able to interact with our members. This webinar provides an opportunity for our members to hear about their work in research, testing, and treatment efforts for those living with inherited retinal disease.

janssen | PHARMACEUTICAL COMPANIES OF Johnson&Johnson

At Janssen, the Pharmaceutical Companies of Johnson & Johnson, we work tirelessly to make a future without disease a reality for patients by fighting sickness with science, improving access with ingenuity, and healing hopelessness with heart.

We focus on areas of medicine where we can make the biggest difference: Cardiovascular & Metabolism, Retina, Immunology, Infectious Diseases & Vaccines, Neuroscience, Oncology, and Pulmonary Hypertension.

For Janssen, it is not just about leading innovation and driving medical breakthroughs. We go beyond the medicine by working with patients through the entire process to ensure the best possible experience and health outcomes. We are driven by our belief that “patients are waiting” and there is no time to waste.

Register here

If you need any further information, please contact us on 03-9650 5088 or email us at info@retinaaustralia.com.au

Queen's Birthday Honours

We are delighted to announce that our Chairman Leighton Boyd, and Company Secretary Rosemary Boyd have both been recognised in the 2022 Queen's Birthday Honours List.

Leighton has been appointed a Member (AM) of the Order of Australia (General Division) in recognition of his “significant service to people who are blind or have low vision”, and Rosemary has been appointed the Medal of the Order of Australia (OAM) (General Division) in recognition of her “service to people who are blind or have low vision”.

Leighton has been involved in Retina Australia and its predecessor organisations, Retina Australia (Victoria) and The Retinitis Pigmentosa Society of Victoria, since joining the committee in 1982. He served as the Victorian President since 2008, and as Chairman of the national organisation Retina Australia since 2015. Leighton was also a Vice President of Retina International from 2015 - 2021 and a member of the organising committee for four World Congresses.

Leighton has also contributed across the sector through his participation as a board member of the former Royal Victorian Institute for the Blind; a volunteer and committee member with Blind Citizens Australia from 2000 - 2015; a committee member focussing on the needs of the blind and low vision communities with Vision 2020 Australia since 2011, and by being a supporter of Bionic Vision since 2010.

In his local community in Nillumbik, Leighton has served as a member of the Disability Advisory Group from 2001 – 2009, the Community Inclusion Advisory Committee 2010 -2014 and was recognised as Nillumbik's Volunteer of the Year in 2009.

"It is a great honour to receive this award. I would like to thank everyone who has supported me in my endeavour to make a difference to the lives of Australians who are affected by an inherited retinal disease." Leighton Boyd, June 2022.

Rosemary has been involved in Retina Australia and its predecessor organisations, Retina Australia (Victoria) and The Retinitis Pigmentosa Society of Victoria since joining the committee in 1986. She served as the Victorian Honorary Secretary from 2008 – 2019 and has been a Board member of Retina Australia since 2008, and Company Secretary since 2013. Rosemary has served as a delegate to Retina International for over 10 years and was the organiser of the Australian Retinitis Pigmentosa Conferences in 2002 and 2015.

Rosemary has also worked as Executive Officer for Blind Citizens Australia from 2013 – 2015, and was Chairman of Ross House Association, the current home of Retina Australia, from 2009 – 2013.

"I am extremely proud to receive this award. It has been a privilege to work with, and for, the inherited retinal disease community throughout Australia." Rosemary Boyd, June 2022.

We would like to congratulate both Leighton and Rosemary for this well-earned recognition and thank them for their huge contribution to Retina Australia and the blind and low vision community.

Topical ocular administration of progesterone could be viable treatment for retinitis pigmentosa

Date: 30th May, 2022
Source: news-medical.net
By: Emily Henderson

Retinitis pigmentosa is a degenerative disease affecting the photoreceptor cells in the retina, known as cones and rods. Of genetic origin, this disease first affects vision in low light conditions, and progressively peripheral vision and the central field of vision until total sight loss occurs, as the photoreceptor cells gradually die. Noting recent research into the role of hormones, particularly progesterone, in preventing cell death due to oxidative stress, the CEU UCH Drug Delivery Systems (DDS) research group has successfully developed and tested a range of methods of delivery of this hormone into the eye to slow the degenerative process characteristic of retinitis pigmentosa.

The research undertaken by the team is detailed in Dr Adrián Alambiaga's doctoral thesis, supervised by Professor Alicia López Castellano and Dr Aracely Calatayud, and which he successfully defended at CEU in April. His findings, already published in several journals, show that progesterone can be delivered in various forms, such as eyedrops, micelles and inserts, in sufficient quantity to penetrate the surface of the eye and reach the neuroretina without causing significant toxicity or irritation.

The use of ocular inserts enables progesterone to be delivered in greater quantities than when aqueous solutions are used. Although we saw that aqueous solutions can also permeate the sclera and the cornea, inserts performed best in our tests, as they liberate progesterone for absorption by the neuroretina over a sustained period and in greater quantity", says Dr Adrián Alambiaga.

Topical administration, the best option

Progesterone is virtually insoluble in water and it only dissolves in gastrointestinal fluids slowly and incompletely. This means that when it is administered orally, it quickly becomes ineffective.

In addition, high levels of progesterone need to be administered orally so that just a small amount can reach the eye. "That's why it was important to study the different ways of administering progesterone topically or locally in the eye, thereby limiting the amount that has to be administered."

Over the course of his doctoral research, Dr Alambiaga, under the supervision of Prof López Castellano and Dr Calatayud, developed a range of pharmaceutical formulations of progesterone for topical delivery to the eye. These included aqueous solutions, which increase the durability and diffusion of the molecules on the ocular surface, and ocular inserts, which increase the contact time of the drug on the ocular surface, increase drug availability to the body, and enable a controlled release, more precise dosages and less frequent administration.

Aqueous solutions provide a successful pathway for topical delivery of progesterone

New treatment strategies

For the group's lead researcher, Professor Alicia López Castellano, who specializes in pharmaceutical technology at CEU UCH, Dr Alambiaga's thesis shows that "we have demonstrated for the first time that topical administration of progesterone in the eye is viable. This opens up possible new therapeutic strategies for retinitis pigmentosa patients, and by extension for patients with other eye conditions in which oxidative stress is a risk factor, such as glaucoma, age-related macular degeneration, macular edema due to retinal vein occlusion, cytomegalovirus retinitis, posterior uveitis and diabetic retinopathy."

Parts of Dr Adrián Alambiaga's thesis have been published as research articles in journals such as Pharmaceutics, the Journal of Pharmaceuticals and Biomedical Analysis, and the International Journal of Pharmaceutics.

A new hope for a therapy against retinitis pigmentosa

Date: 17th June, 2022
Source: Université de Genève, reviewed on sciencedaily.com

Retinitis pigmentosa, a degenerative genetic disease of the eye, is characterized by progressive vision loss, usually leading to blindness. In some patients, structural defects in the photoreceptor cells have been observed, but the molecular mechanisms involved are not understood. A team from the University of Geneva (UNIGE), in collaboration with the University of Lausanne (UNIL), has identified the essential role played by a molecular zipper formed by four proteins. The absence of this zipper leads to cell death in retinal cells. This discovery could lead to the development of therapeutic approaches for retinitis pigmentosa. This work can be read in the journal PLOS Biology.

Retinitis pigmentosa is the most common hereditary retinal disease in humans, with a prevalence of one in every 4,000 people worldwide. The first symptoms usually appear between the ages of 10 and 20 with a loss of night vision. Thereafter, the visual field narrows into a "tunnel vision" to finally lead to blindness around the age of 40. This disease is characterized by a degeneration of the light sensitive cells, the photoreceptors.

These specialized neuronal cells of the retina are responsible for the conversion of light into a nerve signal. The outer segment of the cell is made up of stacks of discs on which the light-sensitive pigments are located. The inner segment contains all the metabolic machinery essential to the functioning of the cell and is linked to the outer segment by the connecting cilium.

A molecular zipper

Mutations in the genes of four proteins located in this connecting cilium are all associated with retinal pathologies presenting degeneration of photoreceptors. These four proteins had been identified by the laboratory of Paul Guichard and Virginie Hamel of the Department of Molecular and Cellular Biology of the Faculty of Science. They are located in centrioles, cylindrical structures made of microtubules and present in all animal cells.

"In the centriole, these proteins ensure the cohesion of the different microtubules by acting like a zipper.

We wondered if they did not play the same role in the tubular structures of the connecting cilium," explains Virginie Hamel, last author of the study.

Observations with unprecedented precision

Thanks to an expansion microscopy technique optimized by the group of Virginie Hamel and Paul Guichard, which allow cells to be inflated without deforming them, the scientists were able to observe retinal tissue with a resolution never achieved. The biologists focused on the structure of connecting cilia from mice that had -- or did not have -- a mutation in the gene for one of the four mentioned proteins. These observations were conducted at different life stages. "In the absence of the mutation, we found that these proteins ensure, just as we had previously seen in centrioles, the cohesion between microtubules by forming a zipper that closes as development proceeds," explains Olivier Mercey, researcher in the Department of Molecular and Cellular Biology and first author of the study.

On the other hand, when the gene for this protein is mutated, although the structure of the microtubules appears normal in the first days, the microtubules gradually become less and less attached to each other. In adulthood, the affected mice have microtubules that are no longer "zipped" together at all and eventually collapse, leading to cell death of the photoreceptors.

Restoring the "molecular zipper" to prevent cell death

This work, supported by the European Research Council (ERC) and the Pro Visu Foundation, has led to a better understanding at the molecular and structural level of retinitis pigmentosa, which allows to consider therapeutic treatments that act upstream of cell degeneration.

"By injecting the protein into patients suffering from certain types of retinitis pigmentosa, we can imagine that the molecular zipper could be restored to ensure the structural integrity of the microtubules of the connecting cilia, thus preventing the death of photoreceptor cells. We are evaluating this approach in collaboration with our colleagues from UNIL and the Jules-Gonin Ophthalmic Hospital, Yvan Arsenijevic and Corinne Kostic," concludes Paul Guichard, coauthor of the study.

Roundworms offer new insights into Bardet-Biedl syndrome

Date: 31st May, 2022
Source: University of Michigan, reviewed on sciencedaily.com

Scientists have identified a new role for a protein complex at the center of a human genetic disorder called Bardet-Biedl syndrome, or BBS, for which there is currently no cure.

Bardet-Biedl syndrome arises when the BBSome protein complex malfunctions. Because the BBSome regulates the form and function of cilia, the hair-like structures on the surface of cells, BBS has been classified as a disease of the cilia.

But the wide spectrum of symptoms associated with BBS -- the most common of which is vision loss, as well as obesity, extra fingers or toes and kidney malfunction -- have led to hypotheses that the cause of the syndrome may not lie solely within the cilia.

In a new study published in Developmental Cell, a team from the University of Michigan Life Sciences Institute now offers the first known direct evidence for these hypotheses. Their findings demonstrate that the BBSome operates outside of cilia to support sight, at least in one common model species.
The discovery began when scientists in the lab of LSI faculty member Shawn Xu were investigating how tiny roundworms called Caenorhabditis elegans can sense light despite having no eye-like organs. Because C. elegans have a simple and well-mapped nervous system, the Xu lab uses them as a model to understand the fundamental biology behind various forms of sensation.

The team performed a genetic screen, a process of introducing random mutations to identify which genes are required for a given biological process, to find the genes involved in the worms' ability to respond to light. Most of the mutations that caused worms to stop sensing light turned out to be in the BBSome. And, like the progressive vision loss that BBS patients experience, the worms with BBSome mutations progressively lost the ability to sense light as they aged.

Through a series of several more experiments, the team discovered that the BBSome plays a role in light sensation independent of its role in the cilia. In one scenario, they mutated C. elegans to remove all cilia; in a second experiment, they left the cilia on the worms but prevented the BBSome from getting to the cilia. In both cases, the worms were still able to sense light, so long as the BBSome functioned in the rest of the cell.

"It's a great demonstration of the power of model organisms," said Xinxing Zhang, a postdoctoral researcher in the Xu lab and the study's lead author. "Cilia are essential for most organisms. But we can remove cilia from the C. elegans and they still survive, allowing us to uncover this unexpected role for the BBSome completely independent of the cilia."

Xu's lab previously discovered that C. elegans sense light through a receptor protein called LITE-1 that sits at the surface of neurons and sends signals to the central nervous system to respond to the light (in the worms' case, by moving away from it).

Because the roundworm, C. elegans, has a simple structure, it is a useful model organism for many aspects of biological research

In this latest study, the team found that when BBSome malfunctions within the cell, LITE-1 receptors are pulled back into the cell from the surface and then broken down, preventing the worms from sensing light.

In a second genetic screen, the scientists discovered that the process of degrading LITE-1 is controlled through another protein called DLK. The BBSome prevents DLK from starting a chain reaction that inappropriately breaks down LITE-1.

Both BBSome and DLK are conserved in humans, and the researchers were able to show that BBSome similarly blocks DLK expression in human cells.

They believe that this BBSome-DLK-photosensor pathway could be involved in the vision loss that is so prominent in patients with Bardet-Biedl Syndrome.

"Because BBS is known to be caused by defects in the BBSome, there has been a longstanding assumption that the disorder must be tied to the cilia," said Xu, who is also a professor of molecular and integrative physiology in the U-M Medical School. "We are not disputing that BBS is tied to defects in the cilia. We are just offering direct evidence that the BBSome can also function outside of cilia, and it has a role there related to light sensation. Perhaps this can broaden the view of how to develop treatments for BBS."

This research was supported by the National Institutes of Health.

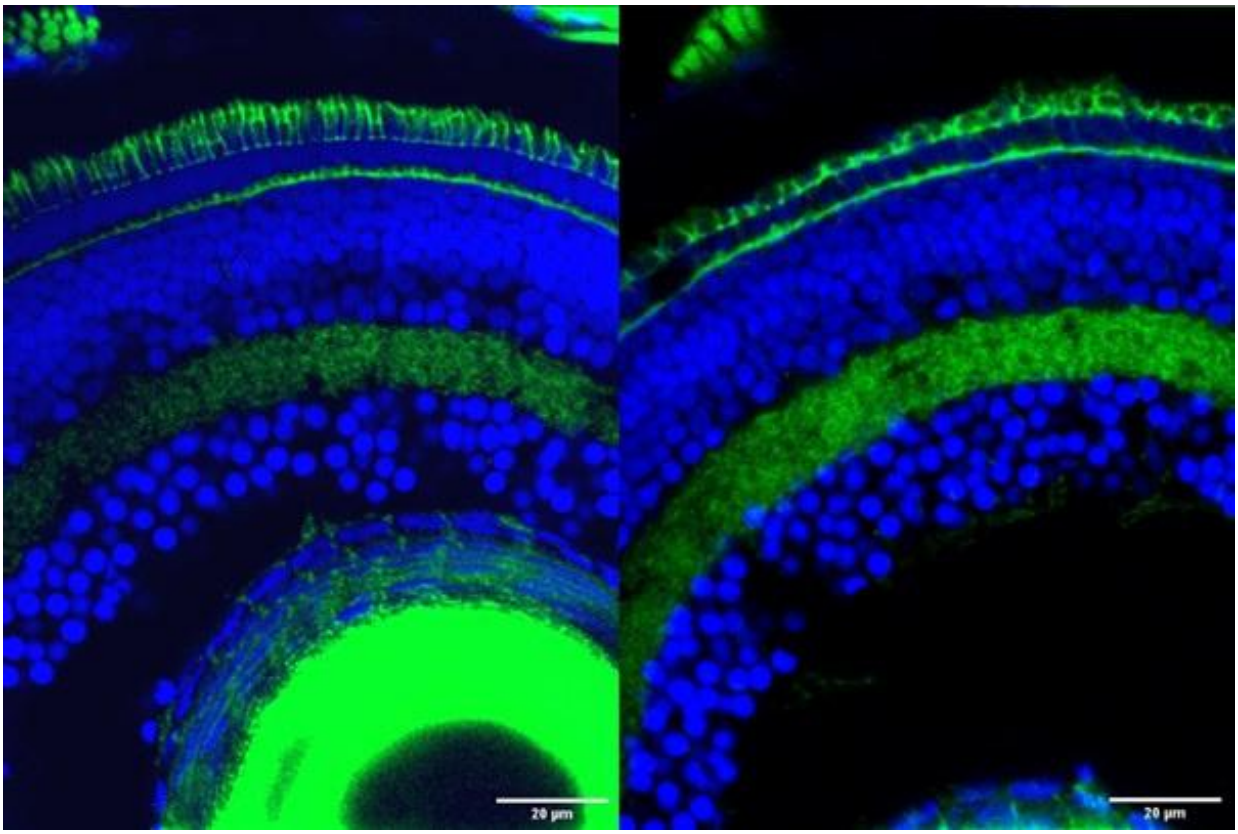
Researchers discover the gene essential for the growth of photoreceptors

Date: 14th July, 2022
Source: Centre for Gene Regulation

Srrm3 is a master regulator gene essential for the growth of photoreceptors, cells in the rear of the retina that catch and process light and transmit signals to the brain that allow vision, according to research, from the Centre for Genomic Regulation (CRG) in Barcelona. Zebrafish with the gene knocked off had severely impaired vision.

According to the study, Srrm3 controls alternative splicing in vertebrates, a process that enables cells to produce multiple types of proteins from a single gene and is particularly prevalent in neural cells. Misregulation of alternative splicing can have a terrible effect on a person’s health, such as in cases of cancer or neurological conditions.

Researchers discovered that Srrm3 primarily controls the splicing of microexons, little DNA segments that are only 3-27 letters long. Despite their small size, it has been demonstrated that microexon control is crucial for protein and cellular function.



Retinal cells in zebrafish with the outer segment, the part of photoreceptor cells responsible for transforming light into nerve signals that enable vision, stained in green at the top of the image. The outer segment is significantly degraded in retinal cells with the Srrm3 gene knocked out (right) compared to normal retinal cells (left). Image Credit: Ludovica Ciampi/CRG/Proceedings of the National Academy of Science

Numerous distinct microexons that are primarily found in photoreceptors but absent from other neurons were discovered by the researchers.

A significant number of these microexons have an impact on the activity of about 70 genes crucial for the growth of a photoreceptor's outer segment, which is the area of the cell that absorbs light. The results are published in the journal Proceedings of the National Academy of Sciences.

The study indicates a new level of cellular specialization needed for retinal cells' distinctive cellular form and function, one of the body's most sophisticated and specialized cells. Due to their intricacy, retinal cells are dependent on numerous distinct genes for their development—any one of these genes could be mutated to produce disease and cause blindness.

Retinitis pigmentosa, a hereditary condition for which the underlying molecular pathways are poorly understood, is one of the most frequent causes of inherited eyesight loss.

About 40–50% of retinitis pigmentosa cases are unexplained, meaning they involve mutations in unidentified genes. The study's authors set out to determine whether Srrm3 or the microexons concerned can explain some of these situations, and will conduct future research.

"The Srrm3 gene has neither been associated with the development of photoreceptor cells nor with the pathogenesis of retinal diseases before. We are already exploring the gene's role in patients without a genetic diagnosis. If we find cases with mutations in this specific gene, or on any retinal microexons, it could lead to potential new therapeutic strategies to manage the condition", states Ludovica Ciampi, Study First Author and PhD Student, Centre for Genomic Regulation.

For finding new therapeutic targets, microexon regulation in certain cell types must be understood, claims ICREA Research Professor Manuel Irimia. "Photoreceptors have unique properties thanks to the regulation of alternative splicing and microexons. This helps make the cell more specialized but also perhaps more susceptible to genetic diseases. Modulating splicing activity is now possible, so the more intricate biology we uncover, the more likely we are to find therapeutic targets to treat retinal diseases," concludes Dr Irimia.

New Stargardt's Support Group in Perth

A new group has been set up to support those living with Stargardt's Disease in Perth. The Order of the Stargardtians aims to provide information, support and social opportunities for those living with Stargardt's Disease. Supported by the Lion's Eye Institute (LEI) and Visibility, the group has already hosted two presentations with Associate Professor Fred Chen, from LEI. You can access a recording of the first of these, an introduction to Stargardt's Disease, on YouTube at: https://youtu.be/zVlUKBvv9kM

They have also set up a Facebook page – just search for Order of the Stargardtians and request to join them.

If you are based in Perth and would like more information, you can contact Madga Blaszkowska on 08-9381 0707 or Lynne Smithies on 08-9381 0790.

Karan Nagrani is using social media to raise awareness about the 'spectrum of blindness'

Date: 8th August, 2022
Source: ABC Great Southern, story by Asha Couch.

What comes to mind when you think of blindness? Is it a person donning dark sunglasses, possibly with a cane, or a guide dog? There are certainly people with vision loss who fit this bill, but for many others, their experience of blindness is not quite so black and white.

Karan Nagrani is legally blind, but if you passed him in the street, it's likely you wouldn't know. Diagnosed at the age of 11 with a degenerative genetic condition called retinitis pigmentosa, the now 36-year-old only has a fraction of his vision remaining."It starts off as night blindness and loss of side vision, and then the central [vision] starts to get affected," Mr Nagrani said.

"When people look ahead, they see 180 degrees … I see less than three degrees, and at night, it's completely black."

From his home in the southern coastal city of Albany, Western Australia, Mr Nagrani has made it his mission to educate people on what he calls the "spectrum of blindness". "I think people have this misconception that if you're blind, your eyes don't look normal," he said. "I can still make eye contact because I can still see a little bit, so people get a little confused."

When meeting new people, Mr Nagrani said he often felt he had to "convince" them of his disability. "I feel a sense of fear until I've convinced them that I have a disability because I don't want to be called a fraud. That is the fear that people are going to say, 'His eyes look normal, he's making eye contact, I think he's faking it'."

Knowing there would come a day when he would lose his sight, he didn't let his diagnosis deter him from pursuing his dream career.

"Growing up, I knew I was going to go blind, but I didn't want to pick a career based on that - I wanted to live my life and do something that I enjoy," he said.

Karan Nagrani is sharing his personal experience to foster a greater understanding of all forms of blindness

"Being creative, I got into graphic design and filmmaking, and I did that for 14 years.I'm proud to say I had a really successful career in marketing that I had to give up because I can't use laptops or computers anymore."

He's still got it

With the knowledge and skills gained from his career, Mr Nagrani is putting them to use by creating infographics and videos for social media using his smartphone.

"Growing up, I never saw any content that prepared me for what it is that I will or won't see," he said. "Now, I'm using my graphic design skills while I still can, to create resources that other people are using."

His Instagram account showcases a sense of humour that hasn't happened totally by chance.

"Social media is all about entertainment … you can present serious information, within reason, in a fun manner. Going by the responses that I get, it's actually the entertaining, informative posts that are most engaging because people actually stop and read and comment," he said.

But not everyone on the internet has his positive energy.

"There's always that one person who has something nasty to say," he said.

"I remember putting up a post once where I showed people what it's like to wake up with retinitis pigmentosa … one of the shots was on the balcony, showcasing the beautiful Albany landscape.

"Someone commented, 'What a waste of such a beautiful view on someone like you'. I get those comments, but I actually think that's a reflection on them, and I brush it off."

Social stigma an obstacle

Blind people experience an extra layer of difficulty navigating day-to-day life because of social stigma, according to eye expert Professor William Morgan. Often patients put in a lot of effort to appear "normal".

"Many people will think they're just normal people and get irritated and annoyed if they bump into them, for example, or take longer to sit down on a bus because they're having to feel their way around the seat," Professor Morgan, from the University of Western Australia and managing director of Lions Eye Institute in Perth, said.

"I do get those comments from patients actually; that they put an enormous amount of effort into nullifying the disability as much as possible."

Professor Morgan said services had improved dramatically for vision-impaired people in recent years, but there was still a way to go in regard to awareness.

"These people are putting a huge effort into mixing in society, and so increasing the tolerance [would help, as well as] an awareness of the different sorts of vision that you lose with these broad categories of diseases."

For Mr Nagrani, sharing his personal experience online is about fostering acceptance for all forms of blindness.

"It makes me so happy to see people from across the globe message me, asking me if they can share my posts to raise awareness," he said.

"I feel like even though I've had to give up my marketing career, I'm actually finding this more fruitful, in the sense that I feel like I'm really making a difference now."

Hot off the Press

World Research Summary by Dr Catherine Civil

Here are a few more snippets for you from the promising world of IRD research:

- In Italy, the miniscule telescope I mentioned previously has now been implanted into 3 people with late-stage AMD (age related macular degeneration) during routine cataract surgery. The telescope enlarges what it sees by 2.7 x and projects the images on to healthy areas of the retina. This amazing telescope is so tiny that it is nearly invisible in the eye...watch this space for the trial outcomes....
- An update on the phase 1/2 trial of botaretigene sparoparvovec gene therapy for the treatment of x-linked retinitis pigmentosa (XLRP) has showed some improvement in photoreceptor function under the bleb of injected genes and around it over the following 12 months. Phase 3 trials are starting.....
- This is an example of a gadget used to inject a gene mixture underneath the retina. Interesting, eh?

Advent R400e Subretinal Gene Therapy Delivery System

- A reminder of what optogenetics means ...most of the IRDs (inherited retinal diseases) destroy rods and cones but the inner retinal neurons remain intact and functional but without image-forming light sensitivity. The goal of optogenetics is to add a new gene to the surviving inner retinal neurons to add a new light sensitive function to these cells. This method is applicable to patients with late-stage disease where there are no more photoreceptors left to preserve or repair.
- Don’t forget your follow up appointments after your anti-VEGF eye injection treatment is finished: Recurrence of the nAMD is common. Even if you have finished the full course of treatment, there is a 50% risk of recurrence of the disease within a year. Beware!

- Two bionic eyes were compared for safety. a) the 256-channel intelligent micro implant eye (IMIE256) and b) Allogeneic RPE Cell Bioengineered Implant for Advanced Dry Age-Related Macular Degeneration. The IMIE256 implant was found to be safer than the Argus II implant. Out of the 5 study participants, 2 participants had severe adverse eye events but were managed by surgery. Visual function was improved in all patients. In the Allogeneic RPE Cell Bioengineered Implant AMD trial, 4 out of 16 subjects had serious ocular adverse events such as retinal haemorrhage, oedema, and retinal detachment. In some of the subjects there was an improvement of >5 letters on the standard eye chart. Overall, the implant was safe and well tolerated at one year. ..... Slow but steady progress.....
- Very excitingly 72 million US dollars has recently been donated by the FFB (foundation Fighting Blindness) to 9 US portfolio companies that are developing new therapies for retinal degenerations. The FFB has managed to leverage a further 5.6x this amount from other investors. That is a lot of research money, and hopefully it will translate to great outcomes for us.
- At the RI congress, Professor Rob Koenekoop from Montreal presented 4 exciting new therapies that are currently in clinical trials. Two therapies are targeted for Stargardt's (STGD) diseases, one for LCA and RP and one for Retinitis pigmentosa. Prof Koenekoop stated that revision therapeutics is testing a Cyclodextrin to remove Bisretinoids from the RPE in STGD patients (awaiting phase 1 trial), Alkeus therapeutics is testing Deuterium Vit A to prevent binding of Vit A derivatives and prevent Bisretinoid build up in STGD patients (awaiting phase 3 trial), Zuretinol, a 9 cis retinyl ester that recovered visual function in LCA patients with RPE65 or LRAT mutations in earlier clinical trials and is awaiting phase 3 trial and the N-acetylcysteine (NAC) Attack study, a phase-3 clinical trial study funded by National Eye Institute to test the effect of NAC in Retinitis Pigmentosa is starting soon.
- Again, at the congress, Dr Slaven Erceg from Spain presented their work on Genetically corrected iPS (induced pluripotent stem cells)-derived RPE cells for cell therapy of hereditary retinal dystrophy. Dr Erceg stated that they were able to generate and genetically correct cells that had the same property as retinal RPE cells. He further stated that future studies will determine if these cells have the potential to restore vision in patients with inherited retinal disorders.

That's all for this edition folks. Stay well and don't forget to keep your details up to date on the Australian IRD register - giving you the best chance of being first in line for any up-coming treatment trials in Australia and around the world.

Get in Touch

Volunteers fighting blindness

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Retina Australia Staff:

- Sally Turnbull, Administrative Officer
- Junxia Xu, Finance Officer

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- DEPUTY CHAIRMAN: Peter Maas
- TREASURER: Joshua Ginpil
- COMPANY SECRETARY: Rosemary Boyd
- DIRECTORS: Mary-Anne Carmody, Jane Cherry, Jess Coleman, Lindsay Da Costa, Julie Demarte, Heather Mack