CURE BLINDNESS AUSTRALIA

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Volunteers Fighting Blindness

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Cure Blindness Australia Inc.

FDA APPROVAL OF RETINAL DYSTROPHY DRUG LAUNCHES ERA OF OCULAR GENE THERAPY

Date: 12 February 2018

Source: Modern Medicine Network

In December 2017, the US Food and Drug Administration (FDA) approved Luxturna (brand name for “Vortigene”) by Spark Therapeutics, a gene therapy for the treatment of biallelic RPE65 mutation-associated retinal dystrophy (commonly known as Leber's Congenital Amaurosis (LCA))

With the regulatory agency’s decision, Luxturna became the first pharmacologic treatment for this inherited disease that often leads to nearly complete blindness and the first gene therapy in the United States indicated for treatment of a genetic disease.

Its approval was granted based on clinical trial results showing that the gene therapy had an acceptable safety profile and resulted in rapid improvements in functional vision and visual function that were sustained with follow-up to 2 years. Data reported at the 2017 American Academy of Ophthalmology meeting show the treatment benefit persists to at least 3 years.
“The approval heralds a new horizon for treatment of inherited retinal dystrophies (RDs),” said Alex V. Levin, MD, MHSc, chief, paediatric ophthalmology and ocular genetics, Wills Eye Hospital, Philadelphia. “It encourages affected patients to get accurate molecular diagnosis of their underlying mutations so that they can bring themselves closer to intervention.” (continued on page 8)

IN THIS ISSUE

PRESIDENT'S REPORT

This is my first opportunity to pass on my Very Best Wishes to all our members for a prosperous and healthy New Year.

In the past few months a lot has happened.

The new name approved at the AGM in November has now been registered; we are now in the process of transitioning all of our banking to the new name and attending to the other legal and administrative requirements. The new Constitution has been registered as part of that process.

Patron – I am very pleased to announce that Judy Horton has agreed to become our new patron. Judy has been involved with RANSW for over 20 years; she has attended most of our fund raising events and was responsible for Yates supporting us through the sale of Cosmos Bright Eyes seeds which brought over $117,000 of funds to the organisation. With the new name now in place it was great that Judy accepted our invitation to be the Patron of the new organisation. A profile of Judy is in this edition of RODS.

Logo – this edition of RODS has our new logo and this logo was approved by the Board at the meeting on 10 March 2018.

A very extensive process of reviewing options was carried out including independent market research before we were in a position to make a final decision. Of the options available, over 75% of respondents chose this version, which made the decision of the Board relatively easy. The art work for all new stationery will be available in the next few weeks and we will then be able to have the stationery printed.

Web Site – The completion of the new web site is progressing well and whilst we would have liked to have it up and running by now, some glitches have meant we needed to slow down, and get it right rather than rush forward and not have it functioning in the manner that we wanted.

Hunter Chapter – I am delighted to advise you that we are opening a Hunter Chapter which will raise money and awareness in the Hunter

Region. We have six members of the management team, all very reputable business people in the Hunter Region.

Newcastle is the 7th most populous city in Australia and so we are obviously very excited about this new initiative. We are very grateful to our Board member, Deb Hescott, for putting this together. Deb travels to Sydney from Newcastle every month for our Board meetings and has been instrumental in a number of very successful fund raising events in that city. This latest initiative has grown out of the profile that Cure Blindness Australia has in the area, and all because of the tremendous work that Deb does and the commitment she shows to growing our profile in the Newcastle area.

Business and Funding Manager – We have been in discussions with a person to join us as a Business and Funding Manager and have reached an 'in principle' agreement with this person to join us. Commencement is not likely to take place before the end of April due to other commitments however we are confident that final agreement is not far away. Until the final agreement is in place I cannot tell you more at this time but should be able to give you more information in the next edition.

Bruce Richards

President

FDA APPROVAL OF RETINAL DYSTROPHY DRUG LAUNCHES ERA OF OCULAR GENE THERAPY

(continued from page 1)

"FDA approval of Luxturna is a landmark event that represents the culmination of nearly 20 years of hard work and a breakthrough for patients affected with LCA, their families, and the scientific community," added Paulo Falabella, MD, medical affairs ophthalmic lead, Spark Therapeutics, Philadelphia. "Needless to say, Spark Therapeutics is very pleased and excited to bring it to market."

Not a magic bullet

Dr. Levin pointed out that gene therapy with voretigene is not a magic bullet, even for patients who are appropriate candidates.

"We still have a lot to learn, and gene therapy needs to be more affordable and accessible," Dr. Levin explained. "But the availability of this approved therapy that can deliver a replacement or repair mechanism for this one abnormal gene in the eye, halt progression of a retinal degeneration in some patients, and perhaps even restore sight brings us through a doorway to an exciting future."

The treatment delivers a normal copy of the gene encoding the RPE65 protein to RPE cells. It thereby has the potential to restore the visual cycle and vision in individuals with RPE65 gene mutations, who have reduced or absent levels of RPE65.

The first of its kind treatment for blindness will cost US$425,000 per injection, or US$850,000 for both eyes, one of the most expensive medicines in the world. It is not clear at this time what plans there might be for its introduction in Australia.

WELCOME TO OUR NEW PATRON: JUDY HORTON

Cure Blindness Australia is thrilled to welcome its new patron, Judy Horton. Judy is a long-time friend of CBA and her name will be well known to many of our members who have a love for the garden. The bio that follows is courtesy of the ABC.

Judy is a horticulturist who is responsible for the regular updating of Yates Garden Guide, the best-selling Australian gardening book.

Judy began her career as a primary schoolteacher before falling in love with her five acre bush garden and studying horticulture in the early 1980s. After graduating from Ryde School of Horticulture she worked as a retail nursery garden adviser and a bush regenerator for the NSW National Trust for a number of years. Her radio experience began more than twenty years ago and over the years she has also filled in for Graham Ross and Don Burke on their respective shows. For the last fourteen years, Judy has been one of the weekend regulars who answer gardening questions on ABC 702 “Weekends with Simon Marnie” program.

Judy has been the recipient of the Horticultural Media Association Hall of Fame Gold Laurel and the NSW Nursery Industry Association’s Allan Seale Award for her contribution to gardening media. In 2012 she was presented with the Golden Wattle Award, the highest award from the Australian Institute of Horticulture, for her contribution to horticulture. A few weeks later she received an Award of Merit from the Nursery & Garden Industry of NSW & ACT for her outstanding contribution to the industry.

NIGHTSTAR THERAPEUTICS ANNOUNCENES INITIATION OF PHASE 3 TRIAL FOR CHOROIDEREMIA

Date: 5 March 2018

Source: Nightstar Therapeutics

Nightstar Therapeutics announced the initiation of the company's STAR Phase 3 registrational trial to study the safety and efficacy of NSR-REP1 in patients with choroideremia. In data from 32 patients treated with NSR-REP1 across four open-label Phase 1/2 clinical trials, over 90% of treated patients maintained or improved their visual acuity over a one-year follow-up period.

The STAR trial is expected to enroll approximately 140 patients across 18 clinical sites in the United States, Europe, Canada and South America, of which six sites will be surgical centers. Patients in the STAR trial are expected to be recruited primarily from the existing Nightstar-sponsored natural history observational study (NIGHT study) in order to accelerate Phase 3 enrollment from this well-characterized patient population.

"The Choroideremia Research Foundation is encouraged by the advancement of this gene therapy and congratulates the Nightstar team for their unrelenting commitment to serving patients," said Randy Wheelock, chief advisor for research and therapy development for the Choroideremia Research Foundation (CRF, http://curechm.org/).

Dr. Christopher Moen, president of the CRF commented, "Not only is this important for choroideremia patients and their families, but it is another important step toward developing therapies for the many people affected by blinding inherited retinal diseases, of which over 200 have been identified. The CRF is proud of its contributions in helping Nightstar achieve this milestone, including grants for initial research and preclinical studies. We look forward to realizing the full potential NSR-REP1 could have for patients with this challenging condition."

$7.5 MILLION FUNDING FOR PROQR'S PRE CLINICAL TRIALS FOR USHERS SYNDROME TYPE 2A

Date: 12 February 2018

Source: Foundation Fighting Blindness and ProQR

Foundation Fighting Blindness and ProQR Therapeutics N.V. announced that they have entered into a partnership to develop QR-421a for Usher syndrome 2A caused by an exon 13 mutation of the causative USH2A gene. Under the agreement, Foundation Fighting Blindness will provide up to $7.5 million in funding to ProQR for the preclinical and clinical development of QR-421a, which is expected to advance towards the clinic in 2018, and safety and efficacy results from the Phase 1/2 trial in Usher syndrome patients are expected in 2019.

Usher syndrome is a devastating genetic disease in which patients first develop hearing loss and then progressive vision loss, thereby threatening their independence and quality of life. Currently there is no treatment for the ophthalmic manifestation of Usher syndrome type 2A. QR-421a is a first-in-class RNA oligonucleotide that is being developed for the treatment of vision loss associated with the disease. QR-421a is designed to modify the RNA such that functional usherin protein is produced in the retina with the goal of stopping the progression of the disease and potentially gaining peripheral vision. ProQR in-licensed the technology underlying QR-421a from Radboud University Medical Center in the Netherlands, where it was invented by lead investigator Dr. Erwin van Wyck.

Foundation Fighting Blindness' Clinical Research Institute (FFB-CRI) has also launched a natural history study in 120 people with USH2A mutations. The study — known as RUSH2A ("R" stands for "rate of progression") — was launched in 2017 and is being conducted at about 20 clinical sites

around the world. RUSH2A investigators will use a variety of technologies to monitor changes in vision and retinal structure to document and analyse disease progression. Knowledge and data obtained from this trial are intended to provide a better understanding of how USH2A mutations affect the severity and progression of vision loss and help to inform the development of QR-421a.

“Teaming with corporate partners to help promising therapies move through preclinical and clinical development is central to FFB’s strategy so we are very pleased to enter into this partnership with ProQR,” said Benjamin R. Yerxa, PhD, CEO at Foundation Fighting Blindness. “The fact that there are currently no available treatments for Usher syndrome type 2A makes this work that much more exciting and critical.”

QR-421a for Usher syndrome is the second program in ProQR’s growing ophthalmology pipeline scheduled to enter clinical trials. The lead program in the ophthalmology pipeline, QR-110, is currently in a Phase 1/2 safety and efficacy trial in adult and paediatric patients with Leber’s congenital amaurosis 10, due to the p.Cys998X mutation in the CEP290 gene. This pipeline also contains several other molecules for genetic eye diseases, including QR-411 for Usher syndrome type 2A due to the PE-40 mutation, QRX-1011 for Stargardt’s disease and QRX-504 for Fuchs endothelial corneal dystrophy.

“We are excited to team up with the Foundation Fighting Blindness to develop QR-421a for patients that suffer from Usher syndrome due to exon 13 mutations“, said Daniel A. de Boer, CEO of ProQR. “They are the leading private funder of retinal disease research with a very patient centric approach which is a core pillar of our strategy. Through this partnership with the Foundation we plan to gain access to important know-how to develop programs in retinal diseases. We expect that the additional funding will allow us to rapidly advance this novel therapy for this orphan disease with a severe unmet need.”

FRENCH BIOTECH STARTS OPTOGENETICS TRIALS TO RESTORE SIGHT IN ALL TYPES OF RP

Date: 10 January 2018

Source: GenSight Biologics, Labiotech.eu and Newsweek

GenSight Biologics will start a clinical trial in the UK testing a combination of gene therapy and a wearable device, called GS030, to restore sight in patients with all forms of retinitis pigmentosa.

GS030 is a combination of a gene therapy and a wearable “optronic visual stimulation device.” The gene therapy step introduces a gene that encodes for a protein sensitive to light into specific neurons. The wearable device is a pair of goggles that capture images and transform them into light patterns that stimulate the light-sensitive proteins in the neurons, which send the visual signal to the brain. The need to transform the visual signals stems from the fact that the light-sensitive protein delivered to the neurons has less sensitivity than that of healthy human photoreceptors. This information is processed in a pocket computer the size of a mobile phone.

Normally optogenetics is used in animals to help researchers better understand how the brain works as it allows them to illuminate certain areas of the brain and document brain behaviour. However, in this trial, the doctors hope that the gene will convert ganglion cells, which normally communicate information from the eye to the brain, into photoreceptor cells. By recruiting cells to replace those damaged by retinitis pigmentosa, scientists believe they can restore vision, New Scientist reported.

At the moment it's unclear just how successful the treatment will be, but experts are hopeful.

“This therapy is novel in its approach,” Dr. Anne Negrin, an ophthalmologist in Purchase, New York who is affiliated with Greenwich Hospital and not involved in the trial or research told Newsweek. “This is the first time we are trying to give vision to people with degenerative eye

disease by recruiting ganglion cells to receive light signals to the brain, rather than trying to repair already damaged photoreceptor cells."

The gene will be injected into one eye and the hope is that it will only boost the detection of red light. Patients will then use a special goggles to help them improve their overall vision. According to the organization, Color Blind Awareness, photoreceptors in our eyes normally perceive three types of light, red, green and blue. However, the new photoreceptors created in this project can only perceive red light. That means the eye will not be able to interpret color and the patients will only be able to see in black and white.

The primary analysis is meant to test the safety of the treatment over the course of a year.

Although there are other gene therapies being developed for retinitis pigmentosa that do not require the use of wearable devices, they are limited to targeting specific mutations causing the disease. For example, Paris-based Horama is developing a gene therapy to cure the disease in patients with mutations in the PDE6β gene, while British NightstaRx is testing another gene therapy for mutations in the RPGR gene.

GenSight estimates that there are over 100 genetic defects that can cause retinitis pigmentosa, which would make it a very hard, long and expensive process to develop individual gene therapies for all of them. Its solution has the potential to address all types of the disease, for which there is no treatment available despite it being the most common genetic cause of blindness. CEO and co-founder Bernard Gilly stated that the therapy could also be extended to other retinal diseases such as dry age-related macular degeneration (dry-AMD).

Date: 19 February 2018

Source: Optometry Today (OT)

The possibility of ending sight loss is no longer a fantasy, it really could become a reality. This is one of the first things that Fight for Sight's interim director of research, policy and innovation, George McNamara, tells OT when speaking about the current picture of eye research in the UK.

However, for this to be achieved there needs to be a marked change of narrative when it comes to the way sight loss is discussed, Mr McNamara states. "There seems to be a very worrying perception in society that once you have lost your sight, what you require is care and support," he explains.

"But actually, what people with sight loss, or at risk of sight loss, want is for it to be halted in its tracks and disease to be stopped before it starts," he added.

While this conversational change may seem like a big one, the director points out: "We are working from this perspective when we talk about cancer and Alzheimer's, for example, so why wouldn't we take this perspective for sight loss?"

Research ready

According to the statistics, there are over two million people in the UK with sight loss, but many are living with a condition for which there is currently no treatment.

Since its establishment in 1965, Fight for Sight's mission has been to stop sight loss, which it still strives for today.

The charity awards over £3 million each year to researchers across the UK, and is currently funding 159 research projects at 44 different universities and hospitals.

Over the years, a number of Fight for Sight-funded projects have experienced success. An example of this is the establishment of the Tommy Salisbury Choroideremia Fund, Mr McNamara shares. “There are a range of success stories, but one that really springs to mind is around the inherited disease, choroideremia, for which there have been massive developments over the last decade,” he said. As a rare inherited disease that causes severe sight loss in men, there is no treatment for it.

However, in 2008, Fight for Sight awarded £300,000 to a research project into the condition led by Professor Miguel Seabra at Imperial College London. The professor’s work was fast-tracked by the grant and played a key role in identifying the function of the protein REP-1, which causes choroideremia. Professor Seabra went on to collaborate with Professor Robert MacLaren at the University of Oxford, who subsequently received a £1.1million grant to conduct the world’s first clinical trial of a treatment for choroideremia.

Six months after treatment with this therapy, the first six patients reported that their vision had improved in dim light. Two of the six were also able to read more lines on the eye chart. The success was significant and resulted in the establishment of NightstaRx, a company that has raised over $157m, which aims to bring gene therapies for inherited eye diseases into the clinic.

While this is just one story from many examples that Fight for Sight could share, what it demonstrates is the importance of funding very early stage research.

Fight for Sight was the catalyst for the successes that have unfolded in terms of finding a treatment for choroideremia, Mr McNamara highlights. “If we were not there in the beginning, venture capitalists, private investors, nor the Government would have seen this as an area that they felt comfortable funding research it,” he said.

Importance of partnerships

Fight for Sight realised the importance of collaborating with others in order to bring about change. In 2017, it agreed a record number of partnerships with 15 fellow charities and organisations.

“Many people living with sight loss are also living with other long-term conditions, such as diabetes. These partnerships are raising the profile and the benefits of eye research into neglected and forgotten diseases, demonstrating that now is the time for eye research to step out of the shadows as it has huge potential to make a difference.”

Looking forward

Mr McNamara is confident in his belief that sight loss can and should be halted because of the innovation that he has observed in research over the last decade.

“When you look at the developments in stem cell research and gene therapy over the last decade, as well as the role that technology now plays, you can see the potential for how we can realistically set out a clear ambition to end sight loss. Take gene therapy, for example, it is the here and now in eye research and we must be ambitious and grasp this opportunity to progress the breakthroughs in research,” he said.

The changes that Fight for Sight is implementing are being done in order to make sight loss a thing of the past and for Mr McNamara this is the perfect outcome.

“People may question this belief and say that it is not possible,” he said, “but I truly believe it is. The reality is that we are working towards ending cancer, and we are trying to stop dementia, and eradicate HIV, so why should we not want to end sight loss?”

“It won’t happen overnight,” he admits, “nor probably in the next 10–15 years,” he adds. “But we can make significant progress towards achieving it, and that can only be to the benefit of patients,” he concluded.

RODS FEEDBACK

RODS is the key publication of Cure Blindness Australia produced for our members, by our members. We are keen to ensure that it continues to be relevant, interesting, and easy to read for all our members, and would love to hear your feedback.

In recent editions, we have added more images in our stories, and ran printed copies of RODS in colour. Tell us what you think:

- Do you like the use of additional images?
- What assistive technologies do you use for reading RODS, and what issues arise by the use of images (if any)?
- To what extent does colour printing enhance your experience of reading the printed copy of RODS?

We'd love to hear any other feedback as well.

ELECT TO RECEIVE RODS BY EMAIL

As a not for profit organisation, every dollar we save on operating overheads is a dollar we can allocate to research to find a cure for RP and related blinding conditions. We are always striving to keep operating overheads as low as possible.

YOU CAN HELP US SAVE COSTS

By electing to receive RODS by email instead of a mailed print copy, saves on printing costs and postage. Contact us today and let us know if you would like to help us save costs by stopping your printed copy of RODS, and to receive instead by email.

See the back page of this newsletter for our contact details.

DONATIONS AND BEQUESTS TO CURE BLINDNESS AUSTRALIA

Help make a difference by donating to Cure Blindness Australia

Help us to find treatments and cures for retinitis pigmentosa and related conditions by donating to Cure Blindness Australia. Donations of $2 or more are tax deductible and go directly towards funding relevant medical research.

Donations can be easily made at everydayhero by clicking here (https://nfp.everydayhero.com/au/retina-australia-nsw-inc).

What is a bequest?

A Bequest is a gift through a will. Your gift, be it large or small, to Cure Blindness Australia Inc. can make a significant and lasting difference to our mission to provide research funds which allow us to continue to fight blindness.

Remembering Cure Blindness Australia Inc in your will is a way to ensure that your legacy carries on long after you have gone. The next time you update your will, your solicitor can help you to make a bequest.

OPPORTUNITIES TO JOIN THE CURE BLINDNESS AUSTRALIA BOARD

The Cure Blindness Australia Board is very proud of it comprising a good mix of diversity and a broad skill base.

We are currently in need of someone who can take over the role of Treasurer, and we also have room for up to five other members. If you have the skills or know someone who might like to join the Board, please contact Bruce Richards on 0418 963 806 or write to president@retinaaustraliansw.com.au

CONGRATULATIONS LINDY HOU: 2 GOLD AT THE 2018 ROAD NATS PARA CYLCLING

The 2018 Cycling Australia Road National Championships (RoadNats) cycling event was held in January in Ballarat where for the first time in its history, para-cycling road races and time trials were introduced. Our long time member and previous board member, Lindy Hou participated in the two womens races, and brought home gold medals in both. Well done Lindy. We're so proudy of you!

RECIPE: PUMPKIN PIE

Ingredients:

- 1 sheet puff or shortcut pastry.
- Mashed pumpkin. Enough to cover pie dish to 1 inch depth.
- 6 sliced roma or round tomatoes, or can tomatoes drained.
- 1 onion finely sliced.
- 200g grated tasty cheese.

Method:

1. Line pie dish with pastry.
2. Blind bake for 20 minutes.
3. Top with mashed pumpkin.
4. Layer thickly sliced tomatoes above pumpkin.
5. Add layer of sliced onions.
6. Top with grated cheese.
7. Bake at 180-200C for 45 minutes.

TOOL SHED

Tools for assisting people with vision impairments:

TAP TAP SEE

This App describes the world around you. Simply double-tap on the screen and it will take a photo that gets uploaded to its servers for processing. A few seconds later, assuming everything worked, the voiceover assistant provides an impressively detailed and accurate description, such as “Keyboard on white desk with glass mug”.

Available on Apple and Android mobile devices. Download the app from the iTunes App Store and Google Play Store.

IDENTIFI

iDentifi is an app that uses artificial intelligence to enable a visually impaired user to click a photo and get an instant description. It’s able to recognise objects, brands, colours, facial expressions, handwriting and text, and delivers an audible description of the image’s contents to the user. The interface is very accessible and gives the option to choose from three different modes of object and text recognition, as well as how fast you want the app to speak

Available on Apple mobile devices. Download the app from the iTunes App Store.

BUMP DOTS

Bump Dots are ideal for low vision. They come in a wide range of different sizes and colours to provide tactual identification of everyday items. Use clear bump dots to allow surfaces to show through such as on computer keyboards, musical keyboards, telephone keypads, television keypads, remote control keypads, and more. Use black dots on a white background (and vice versa) or fluorescent orange dots on patterned or darker backgrounds.

Available from the Vision Australia online shop and many other retailers.

UPCOMING EVENTS

Vision Australia maintains an excellent listing of events including audio described theatre shows, blind cricket competitions, Vision Australia client days and more at: http://www.visionaustralia.org/about-us/events

IN THE NEXT ISSUE

- Latest research information
- Your CBA Committee news

CURE BLINDNESS AUSTRALIA

Volunteers Fighting Blindness

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Please contact Cure Blindness Australia Inc. if you wish to speak to one of our volunteers. Office hours are Thursday 9.30 – 3.30. However, our telephone is monitored each day and you may leave a message which will be attended to as promptly as possible.

Cure Blindness Australia Inc. needs your support in fighting blindness. You can help by subscribing to our quarterly RODS magazine, fundraising and or making a donation to Cure Blindness Australia Inc.
This support will enable our volunteers in providing funds for much needed research, information and peer support within Australia for those 1 in 3000 affected by inherited retinal disease such as RP and related conditions.

COUNCIL MEMBERS - 2017

PRESIDENT:	**Bruce Richards**	**SECRETARY:**	**Robyn Richards**
VICE-PRESIDENT:	**Debra Hescott**	**TREASURER:**	**Vida Hung**
Councillors:	**Andrew Dunn, Michael Astrinos**		