



I-C

Newsletter for Retina Australia (ACT) Inc

EDITION 2/OCTOBER 2015

Message from the President

Welcome to the second newsletter of the year, which we, your committee, naturally hope you find interesting.

Appended to this issue of the newsletter is a small survey which we encourage all members to participate in. We are very keen to hear from you all your views on how your committee operates, and how we can do better on your behalf.

In the meantime please note opposite further details for this year's major fundraiser. It would be good to have every member of Retina Australia (ACT) there!

ROBIN POKE AM
President
Retina Australia (ACT)

Retina Australia (ACT) is proudly
sponsored by
Elders Real Estate Belconnen

Suite 4, Bank Building, Jamison Centre, Macquarie, ACT 2614
Phone: 02 6251 2088

"ANNUAL SPRING RACING LUNCHEON"

**SUNDAY 15TH NOVEMBER 2015
THOROUGHbred PARK
CANBERRA**

Invite all your friends to this fun afternoon with raffles and racing, food and fun - and, importantly, fundraising that will support the work being carried out by your local Retina Australia branch into the prevention and cure of Retinitis Pigmentosa.

We are fortunate to once again have the support of Women in Racing (WiR). Last year we had some 200 to our lunch, easily a record. Let's make a big effort to beat that number this year. With WiR's support we have been able to contribute not only to the ACT's share of funds for the Australian Inherited Retinal Diseases Register but also to funding for other research.

We also have a special guest. Chris Burton, grandson of committee member Barbara Burton and an RP sufferer since he was eight, recently flew with his dad, Noel Burton, to Canada, where he acquired a new pair of revolutionary eSight glasses. The difference in his sight was instant, and the glasses have made a difference to all facets of his life. Come and hear Chris's amazing story.

Jan James, Secretary

GENERAL NEWS

RETINA INTERNATIONAL

During July, the president of Retina International, Christina Fasser from Switzerland, announced the appointment of Ms Avril Daly as the first CEO of Retina International. Avril, who is currently the CEO of Fighting Blindness Ireland, will join Retina International later this year. Retina International, has been run by a volunteer President and Management Committee since its inception in 1978. At the 2012 Retina International General Assembly held in Hamburg, a motion was passed to establish a dedicated Secretariat and appoint a permanent administrative office. At the 2014 General Assembly, held in Paris, it was unanimously agreed to locate the office in Dublin. Since then, the Management Committee has been following a process to establish this office and appoint a CEO.

Christina said the Retina International Management Committee was looking forward to a fruitful and beneficial collaboration for all involved, especially for those living with a retinal degenerative disease and their families. "Retina International is very pleased to appoint Ms Avril Daly as its first CEO," she said. "Avril has worked as an advocate in the international patient community for fifteen years and represented Retina International on the Board of the European Patients' Forum and the European Organisation for Rare Diseases. She has also presented on our behalf at many international conferences in the United States and in Europe."

Leighton Boyd, the president of RA Victoria, says he has had the pleasure of meeting and working with Avril since 2009. "I believe her appointment is well justified," he says. "Avril has an excellent reputation across international retina organisations as a patient advocate and through her work in policy development. She has also focussed on the areas of medical research, clinical trial development, orphan drug designation and health technology assessment.

"So well done to Retina International," said Leighton, "for taking this big leap forward in a bid to more adequately support a global retina community determined to do all that is possible to find treatments and cures for all types of inherited retinal diseases".

WANT TO WALK OR RUN AROUND THE LAKE? – THEN THIS IS FOR YOU

"Achilles Canberra is a walking and running group for people with disabilities. Most of our members with disabilities have vision impairment. Achilles Canberra promotes involvement in active lifestyles and achieving personal fitness goals in a socially supportive environment. Regular training takes place at Lake Ginninderra on every 1st, 3rd and 5th Sunday of each month. Participants who are vision impaired are paired up with volunteer guides who run or walk alongside them during training sessions. Our president, Peter Ralston, is also an accredited athletics coach. We welcome any new members whether they are walkers, walkers wanting to be runners or established runners.

Our most recent achievements include the Canberra Times Fun Run where we entered 7 walkers and runners into the 10km and 5km events. We also have 2-5 teams participating in weekly 5km parkrun events at Ginninderra and Gungahlin.

If you would like to know more about Achilles Canberra or would like to participate, please contact us at achillescanberra@gmail.com "

RESEARCH NEWS

CARRIER FEMALES AFFECTED BY X-LINKED RP

by Ben Shaberman, 4 August 2015

X-linked retinitis pigmentosa (XLRP) is an inherited retinal disease that causes significant vision loss, sometimes complete blindness, in males. Females are often considered to be unaffected carriers of the condition, with a 50 percent chance of passing XLRP to their sons.

However, women can have vision loss from XLRP as well, and a Foundation Fighting Blindness-funded study of 242 XLRP carriers, led by Jason Comander, M.D., Ph.D. at the Massachusetts Eye and Ear Infirmary, indicates that it may happen more than previously thought. Results of the study were published in the journal *Ophthalmology*.

"I thought that most XLRP carriers would be normal", says Dr. Comander. "That is the traditional clinical teaching. However, when you put carriers 'under the microscope,' you find retinal abnormalities in most. Usually the abnormalities are mild and don't change quality of life. Only 2.5 percent of XLRP carriers are legally blind."

Dr. Comander and his team found that 40 percent of genetically tested XLRP carriers showed a definitive abnormality in at least one of three vision tests: visual acuity, visual field or dark adaptation.

Why Women May be Affected

In XLRP and other X-linked diseases, the mutated gene is on an X chromosome. Women aren't expected to have vision loss from XLRP because they have two X chromosomes — a healthy X in addition to the X with the mutation. Because men have one X and one Y, they experience severe vision loss if they inherit the X with the mutation from their mothers.

However, some copies of a woman's healthy X chromosome are inactivated as a result of a process aptly called "random X inactivation." That means women can experience vision loss from XLRP because some copies of healthy X chromosomes in the retina are turned off. Because X inactivation is random, there's variability from woman to woman in how much of the retina is affected.

See more at: <http://www.blindness.org/blog/index.php/a-surprising-number-of-carrier-females-are-affected-by-x-linked-retinitis-pigmentosa>

IN BLINDING EYE DISEASE, TRASH-COLLECTING CELLS GO AWRY, ACCELERATE DAMAGE

Source: www.nih.gov, 2 July 2015.

Spider-like cells inside the brain, spinal cord and eye hunt for invaders, capturing and then devouring them. These cells, called microglia, often play a beneficial role by helping to clear trash and protect the central nervous system against infection. But a new study by researchers at the US National Eye Institute (NEI) shows that they also accelerate damage wrought by blinding eye disorders, such as retinitis pigmentosa. NEI is part of the National Institutes of Health.

"These findings are important because they suggest that microglia may provide a target for entirely new therapeutic strategies aimed at halting blinding eye diseases of the retina," said NEI Director, Paul A. Sieving, M.D. "New targets create untapped opportunities for preventing disease-related damage to the eye, and preserving vision for as long as possible."

The findings were published in the journal *EMBO Molecular Medicine*. Lead investigator, Wai T. Wong M.D., Ph.D., chief of the Unit on Neuron-Glia Interactions in Retinal Disease at NEI, studied mice with a mutation in a gene that can also cause retinitis pigmentosa. The researchers observed in these mice that very early in the disease process, the microglia infiltrate a layer of the retina near the photoreceptors, called the outer nuclear layer, where they don't usually venture. The microglia then create a cup-like structure over a single photoreceptor, surrounding it to ingest it in a process called phagocytosis. Dr Wong and his team caught this dynamic process on video. The whole feast, including digestion, takes about an hour.

Phagocytosis is a normal process in healthy tissues and is a key way of clearing away dead cells and cellular debris. However, in retinitis pigmentosa the researchers found that the microglia target damaged but living photoreceptors, in addition to dead ones. To confirm that microglia contribute to the degeneration process, the researchers genetically eliminated the microglia, which slowed the rate of rod photoreceptor death and the loss of visual function in the mice. Inhibiting phagocytosis with a compound had a similar effect. The microglia seem to ignore cone photoreceptors, which fits with the known early course of retinitis pigmentosa.

"These findings suggest that therapeutic strategies that inhibit microglial activation may help decelerate the rate of rod photoreceptor degeneration and preserve vision," Dr Wong said.

So what triggers microglia to go on this destructive feeding frenzy? Dr Wong and his colleagues found evidence that photoreceptors carrying mutations undergo physiological stress. The stress then triggers them to secrete chemicals dubbed "find me" signals, which is like ringing a dinner bell that attracts microglia into the retinal layer. Once there, the microglia probe the photoreceptors repeatedly, exposing themselves to "eat me" signals, which then trigger phagocytosis. In response to all the feasting, the microglia become

activated. That is, they send out their own signals to call other microglia to the scene and release substances that promote inflammation.

Other potential treatments for retinitis pigmentosa, such as gene therapy, are progressing, but are not without challenges. Gene therapy requires replacing defective genes with functional genes, yet more than fifty distinct genes have been linked to the disease in different families, so there's no one-size-fits-all gene therapy. "A therapy targeting microglia might complement gene therapy because it's an approach that's independent of the specific genetic cause of retinitis pigmentosa", said Wong.

A clinical trial is under way at NEI to see if the anti-inflammatory drug minocycline can block the activation of microglia and help slow the progression of retinitis pigmentosa. The trial is currently recruiting participants.

Wong's lab colleagues Lian Zhao, Ph.D., Matthew Zabel, Ph.D., and Xu Wang, M.D., Ph.D., played key roles in conceiving and conducting this research. The NIH is a component of the U.S. Department of Health and Human Services, and is the nation's medical research agency, which includes 27 Institutes and Centres.

GENE TIED TO PROFOUND VISION LOSS DISCOVERED BY SCIENTISTS

Date: January 15, 2015

Source: University of Texas Health Science Center at Houston

An exhaustive hereditary analysis of a large Louisiana family with vision problems has uncovered a new gene tied to an incurable eye disorder called retinitis pigmentosa, according to an examination led by scientists at the University of Texas Health Science Center at Houston (UTHealth). It is a family of eye diseases that affects more than 200,000 in the United States and millions worldwide.

The retina converts images into electrical signals that can be processed by the brain. It acts much like the film in a camera. Retinitis pigmentosa damages this film (the retina) and its early symptoms include decreased night vision and peripheral vision. Once it starts, the loss of vision is relentlessly progressive, often ending in blindness.

In the journal *Investigative Ophthalmology & Visual Science*, UTHealth's Stephen P. Daiger, Ph.D. and his colleagues report their discovery of a new gene tied to retinitis pigmentosa, which brings the total of genes associated with this sight-threatening disease to more than 60. The gene is called hexokinase 1 (HK1).

This information is important because it helps affected families cope with the disorder, helps explain the biologic basis of these diseases and suggests targets for drug treatments and gene therapy, said Dr Daiger, the report's senior.

"The challenge now is to block the activity of these mutations and clinical trials are underway to do just that," he said.

"Dr Daiger is trying to make a breakthrough in potentially blinding diseases with no known treatments," said Richard S. Ruiz, M.D., professor of ophthalmology and holder of the John S. Dunn Distinguished University Chair in Ophthalmology at UTHealth. "Right now, we address the symptoms of the disease and help patients make the most of their existing vision."

For approximately three decades, Daiger, a member of the Human Genetics Center at the UTHealth School of Public Health, has been following the progress of hundreds of families across the country with retinitis pigmentosa. "We've found the cause of disease in 80 percent of the families we have studied," Daiger said. "Our goal is to find the cause in the remaining 20 percent."

Equipped with the genetic profiles of family members, Daiger's team has identified differences in the genetic makeup of those with the disease. The researchers also use family histories and DNA tests to glean information about the condition's hereditary nature.

There are different types of retinitis pigmentosa and Daiger's laboratory is focused on the autosomal dominant type. This means that only one parent needs the mutation in order to pass the disease to a child. This type accounts for about a third of all cases and many of its disease-causing genes have been discovered, several by Daiger's research group.

"The story of the HK1 mutation is itself interesting," Dr Daiger said. "What we found is a mutation present in families from Louisiana, Canada and Sicily. Our evidence suggests the mutation arose in a common ancestor who lived centuries ago. The mutation spread in Europe and North America, and may be common among Acadians in Louisiana. This is called a founder mutation."

STEM CELL INJECTION MAY SOON REVERSE VISION LOSS CAUSED BY AGE-RELATED MACULAR DEGENERATION

Date: April 14, 2015

Source: Cedars-Sinai Medical Center

An injection of stem cells into the eye may soon slow or reverse the effects of early-stage age-related macular degeneration, according to new research from scientists at Cedars-Sinai published in the journal *Stem Cells*. Currently, there is no treatment that slows the progression of the disease, which is the leading cause of vision loss in people over 65.

"This is the first study to show preservation of vision after a single injection of adult-derived human cells into a rat model with age-related macular degeneration," said Shaomei Wang, MD, PhD, lead author of the study and a research scientist in the Eye Program at the Cedars-Sinai Board of Governors Regenerative Medicine Institute.

The stem cell injection resulted in 130 days of preserved vision in laboratory rats, which roughly equates to 16 years in humans.

Age-related macular degeneration affects upward of 15 million Americans. It occurs when the small central portion of the retina, known as the macula, deteriorates. The retina is the light-sensing nerve tissue at the back of the eye. Macular degeneration may also be caused by environmental factors, ageing and genetic predisposition.

When animal models with macular degeneration were injected with induced neural progenitor stem cells, which derive from the more commonly known induced pluripotent stem cells, healthy cells began to migrate around the retina and formed a protective layer. This protective layer prevented ongoing degeneration of the vital retinal cells responsible for vision.

Cedars-Sinai researchers in the Induced Pluripotent Stem Cell (iPSC) Core, directed by Dhruv Sareen, PhD, with support from the David and Janet Polak Foundation Stem Cell Core Laboratory, first converted adult human skin cells into powerful induced pluripotent stem cells (iPSC), which can be expanded indefinitely and then made into any cell of the human body. In this study, these induced pluripotent stem cells were then directed toward a neural progenitor cell fate, known as induced neural progenitor stem cells, or iNPCs.

"These induced neural progenitor stem cells are a novel source of adult-derived cells which should have powerful effects on slowing down vision loss associated with macular degeneration," said Clive Svendsen, PhD, director of the Board of Governors Regenerative Medicine Institute and contributing author to the study. "Though additional pre-clinical data is needed, our institute is close to a time when we can offer adult stem cells as a promising source for personalized therapies for this and other human diseases."

Next steps include testing the efficacy and safety of the stem cell injection in preclinical animal studies to provide information for applying for an investigational new drug. From there, clinical trials will be designed to test potential benefit in patients with later-stage age-related macular degeneration.

GENE THERAPY OFFERS BLINDNESS CURE HOPE

Date: December 9, 2014

Source: Fighting Blindness UK

A radical form of gene therapy that remodels eye cells into light receptors holds out the promise of restoring sight to people who are completely blind. The new approach is able to create replacement photoreceptors from cells that do not normally react to light.

In early tests on blind rescue dogs with an inherited disease similar to the human condition

retinitis pigmentosa, scientists were able to restore sufficient light sensitivity for the animals to distinguish between flashing and non-flashing lights.

Blind mice given the same treatment became as good at navigating a water maze as normal mice. The research, reported in the journal *Proceedings of the National Academy of Sciences*, sets the stage for future clinical trials in humans with degenerative eye diseases. Two components of the 'hybrid' treatment involve a gene that alters non-light sensitive cells and an injected chemical 'photo switch'.

US lead scientist Professor Ehud Isacoff, from the University of California at Berkeley, said: "The dog has a retina very similar to ours, much more so than mice, so when you want to bring a visual therapy to the clinic, you want to first show that it works in a large animal model of the disease.

"We've now shown that we can deliver the photo switch and restore light response to the blind retina in the dog as well as in the mouse, and that the treatment has the same sensitivity and speed of response. We can reanimate the dog retina."

The therapy is one of a number of potential treatments for blindness at early stages of development, two of which yielded exciting trial results this year. In October scientists from the US company Ocata Therapeutics, formerly known as Advanced Cell Technology, showed that stem cell-derived retinal cells could safely be implanted into patients and improve vision in some cases.

Earlier this year scientists at Oxford University hailed trial results from a genetic therapy for choroideremia, a rare inherited cause of blindness that affects one in 50,000 people. Inserting a missing gene called REP1 prevented progression towards blindness and led to dramatic improvements in sight for two men at an advanced stage of vision loss.

The new treatment employs a virus to insert a gene into normally non-light sensitive cells in the retina that gives them the potential to "see". The gene makes a protein that acts like a lock. When the right molecular key from the photoreceptor switch is slotted into the lock, light sensitivity is turned on. At present a new injection of the photo switch has to be made every week to maintain its effect, since the molecule is naturally removed after a period of time.

Several of the dogs have been treated and are currently undergoing tests to determine what level of light sensitivity they now have. The dogs already had the genetic disease when they were rescued from breeders and recruited for the study.

Co-author Dr William Beltran, from the University of Pennsylvania School of Veterinary Medicine in the US, said: "Use of such a clinically relevant large animal model allows us to begin tackling the next challenges on the road to translating this novel therapeutic strategy to human patients."

The therapy is said to show promise because although diseases such as RP destroy the eye's photosensitive cells, other cells in the retina are often left intact and unharmed. They include bipolar and ganglion cells, which both transmit visual nerve signals but do not contain photoreceptors.

In tests on mice, the gene was successfully inserted into almost every one of the rodent's million or so retinal ganglion cells. According to the researchers, this should be enough to restore useful vision.

"So far we can say that the treated mice can distinguish between steady light and flashing light," said Prof Isacoff. "Our next step is to figure out how good they are at telling images apart."

The scientists are also looking to see if the photo switch can be used to activate other receptor types, including some that might allow perception of fainter light.

One of the leading scientists involved in the quest to find a treatment for RP in the UK said the research was "exciting". Professor Paul Bishop, a consultant ophthalmologist at the Manchester Royal Eye Hospital, said: "Converting non-light sensitive cells in the retina into light-sensitive cells is an exciting new approach that has potential to restore sight in people who are blind from hereditary retinal degenerations".

Prof Bishop chairs the medical advisory board of RP Fighting Blindness, a charity dedicated to funding research into a cure or treatment for the condition and providing support to those with it.

The charity's chief executive David Head said: "This is highly-credible work which has been emerging in very recent years. The principles behind so-called 'optogenetics' are fascinating and as ever we are amazed by the research that is being carried out around the world. We will watch developments very closely and we will certainly be making our members and patients aware of this work. It is important to recognise that it is still at the stage of animal testing, but exciting nevertheless."

RETROSENSE THERAPEUTICS' LEAD GENE THERAPY CANDIDATE GETS FDA CLEARANCE TO PROCEED TO FIRST-IN-HUMAN CLINICAL TRIALS

Gene therapy application of optogenetics hopes to restore sight to patients affected with retinitis pigmentosa

ANN ARBOR, Mich. RetroSense Therapeutics LLC, a privately-held biopharmaceutical company, today announced the Company's Investigational New Drug (IND) application for RST-001 received clearance from the United States Food and Drug Administration (FDA). RetroSense is developing RST-001 for the treatment of retinitis pigmentosa (RP), a genetic

condition that leads to the progressive degeneration of rod and cone photoreceptors (cells found in the retina that sense light), resulting in severe vision loss and blindness. With its IND now in effect, RetroSense expects to initiate a Phase I/II clinical trial by year-end in order to evaluate the safety and, potentially, efficacy of RST-001.

"There is great promise for the clinical application of optogenetics and this first human clinical trial should provide key insights into the potential for this therapy to treat diseases affecting the eye or brain," a company spokesperson said.

RetroSense Therapeutics is developing RST-001 as a first-in-class gene therapy application of optogenetics. Optogenetics refers broadly to a means of conferring light sensitivity to cells that were not previously, or natively, light sensitive. By applying optogenetics to retinas in which rod and cone photoreceptors have degenerated, RetroSense is working to confer new light sensitivity to the retina, with the expectation of some degree of improved or restored vision for affected patients.

In 2014, the FDA granted Orphan Drug designation for RST-001 based on its development as a treatment of RP, a rare disease that affects an estimated 100,000 people in the United States. As a designated Orphan Drug, RST-001 is eligible for various development incentives under the Orphan Drug Act, including a potential waiver from FDA's application user fees, certain tax incentives and Orphan Drug exclusivity.

"The IND for RST-001 is an important milestone for the company. This brings us one step closer to realizing our ambition of improving vision in those individuals with currently incurable blindness," said Sean Ainsworth, CEO of RetroSense Therapeutics. "There is great promise for the clinical application of optogenetics and this first human clinical trial should provide key insights into the potential for this therapy to treat diseases affecting the eye or brain."

Dr. Zhuo-Hua Pan, the inventor of RetroSense Therapeutics' optogenetic approach added, "My hope from early on was to see our work improve the lives of people with vision defects. It is great to see the approach moving imminently into human clinical studies."

About RetroSense Therapeutics

RetroSense Therapeutics is a privately-held biotechnology company developing life-enhancing gene therapies designed to restore vision in patients suffering from blindness due to retinitis pigmentosa and advanced dry age-related macular degeneration. There are currently no FDA-approved drugs to improve or restore vision in patients with these retinal degenerative conditions. The company's approach to using optogenetics in vision restoration is based on pioneering, proprietary research conducted at Wayne State University and Massachusetts General Hospital. RetroSense has worldwide exclusive rights to the relevant intellectual property from both institutions. RetroSense is led by a team of seasoned veterans with deep experience in taking products from the discovery stage through to the clinic.

CANADIAN GENE THERAPY TRIAL TO TREAT CHOROIDEREMIA

Biopharmaceutical company Nightstar has announced that a new gene therapy clinical trial for Choroideremia will take place at the University of Alberta in Canada. Choroideremia (CHM) is a rare inherited cause of severe sight loss that only affects the male population. Symptoms start in childhood with trouble seeing at night and eventually lead to complete blindness by around the age of 40. There is currently no treatment.

The condition is caused by faults in the CHM gene, which codes for a protein called REP-1. Initial work supported by the Tommy Salisbury Choroideremia Fund at Fight for Sight meant that researchers at Imperial College London were able to find out what REP-1 does.

Promising results

A team led by Professor Robert MacLaren at Oxford's Nuffield Laboratory of Ophthalmology, then developed ground-breaking therapy to repair the fault. The world's first clinical trial of the therapy reported promising results in *The Lancet* in 2014.

Six male patients will take part in the Canadian Phase II study to find out more about how safe and effective the treatment is. They will each receive an eye injection of a virus that's been engineered to safely carry a healthy copy of the choroideremia gene into cells in the light-sensitive part of the eye.

Ian MacDonald, Professor of Ophthalmology and Visual Sciences, University of Alberta, commented: "We are very excited to be working with Nightstar and to have treated our first choroideremia patient. Choroideremia is a devastating condition for individuals and families, but we believe our new gene therapy will arrest any further deterioration of vision and will provide long lasting benefit. The next challenge will be making this therapy available to all individuals with the condition as soon as we possibly can."

ARVO 2015 HIGHLIGHT: NEW RESEARCH BOOSTS PROSPECTS FOR SAVING VISION WITH RDCVF

An eye doctor could preserve meaningful vision in people with advanced retinitis pigmentosa (RP) by saving just five percent of their cones, the cells concentrated in the central retina enabling us to read, recognise colours and see in lighted conditions.

That was the impetus for Foundation Fighting Blindness (FFB)-funded researchers Jose Sahel, MD, and Thierry Leveillard, PhD, to search for, and ultimately discover, a cone-saving protein they call rod-derived cone-viability factor, or RdCVF. After years of refinement and testing in animal models, the emerging therapy is about a year and a half from moving into a clinical trial at the FFB-funded Paris Research Centre for the Study of Retinal Degenerative Diseases.

While the two scientists from the Institut de la Vision have been optimistic about the ability of RdCVF to save vision in people, results from a new study, reported at ARVO (The Association for Research in Vision and Ophthalmology) and published in the journal *Cell*, have taken their confidence to a new level. By studying RdCVF in mice and chickens, the latter of which have cone-rich retinas, they identified its mechanism of action. In other words, now they know precisely why it works so well.

The researchers found the RdCVF boosts the uptake of energy to the cones. Also, the protein may help regenerate cone outer segments, the protrusions which make vision possible by capturing and processing light, but are often lost on retinal degenerative diseases.

"The mode of action of RdCVF is appealing because it opens a new avenue of research aimed at promoting the regrowth of cone outer segments and restoring lost visual acuity in RP," says Dr Leviellard. "It is also an entirely novel neuroprotective mechanism for preserving cones."

RdCVF will be administered to patients as a gene therapy. A human engineered virus known as an adeno-associated virus (AAV) will deliver copies of a gene that expresses RdCVF to cones. A single injection of the treatment, made underneath the retina, will likely work for several years. A key benefit of RdCVF is that it works independent of the mutated gene causing vision loss.

While it has been evaluated for several different forms of RP, RdCVF may also be effective for other retinal conditions.

"This new research represents a massive amount of work, led by Thierry, and provides a valuable elucidation of RdCVF's mechanism of action," says Dr Sahel. "It follows decades of FFB support for Thierry and me, and the advancement of RdCVF toward a human study."

Source: Ben Sharberman, 13 May 2015, www.blindness.org

HELPING BLIND DOGS SEE COULD HELP FIGHT HUMAN BLINDNESS

Dogs have lent their eyesight to people who need it, perhaps since the friendship between human and beast began. And now, mapping the genes of blind dogs could lead to treatments for the visually impaired. By uncovering canine eye mutations, veterinary researchers are coming closer to understanding two of the most common diseases that cause blindness: glaucoma and retinitis pigmentosa.

It turns out that dogs' eyes are similar to humans', the veterinary researchers say, and what goes for theirs often goes for ours, too. So much so, that a U.S. foundation for research into blindness has funded some of their work. Andras Komaromy's research journey began 10

years ago with a phone call from a breeder, who was watching dogs slowly go blind from a strange retinal disease.

"I drove more than 500 miles from Philadelphia to Michigan to examine the affected dogs," the research veterinarian said in a statement. He eventually moved there and researched the disease at Michigan State University. The dogs were all the same breed, Swedish Vallhunds. The hereditary disease appeared to be relatively new. Scandinavian veterinary eye examiners began seeing it in the late 1990s.

The Swedish Vallhund is small, stocky and pointy-eared, with thick grey to red fur in a pattern resembling that of a German shepherd. Despite its compact size, it's tough and fearless, the American Kennel Club says – a "big dog in a small body." It's an athletic herder and a friendly family dog. It impresses in show competitions - dashing through "flyball" obstacle courses or walking through obedience drills.

It's also called a Viking dog because its ancestors go back at least to those times, and the breed almost died out during World War II in Sweden until a nobleman intervened. Swedish Vallhunds are still not found in many places.

So, Komaromy hit the road to dog shows in Scandinavia and North America to examine the Viking dogs. And he found colleagues Hannes Lohi and Paivi Vanhapelto doing similar research in Finland. They covered seven countries on three continents to examine 324 dogs, and with the dog owners' permission and supervision by an animal ethics group, they tested blood samples from the dogs to study their genomes. The researchers nailed the degenerative retinal disease down to a mutational defect on a gene designated as MERTK (c-mer proto-oncogene tyrosine kinase).

They published the results last December. Problems with the gene are already associated with incurable retinal blindness in humans. When things go wrong with the gene, tissue in the retina can slowly atrophy. By marking the gene, they can help breeders avoid spreading the hereditary disease, but they will also work to develop a treatment that inhibits the mutated gene to put the brakes on the disease.

The researchers hope it will lead to similar possibilities in humans stricken with retinal disease, since not only the anatomy of a dog's eye is similar to a human's, but how genes shape them is as well. "Canine retinal disease models contribute significantly to our understanding of retinal disease mechanisms and the development of new therapies for human patients," they said.

Studies on blindness in another dog, also believed to be of Viking origin, have led the same research team to make advances in the fight against glaucoma, a much more widespread cause of blindness in humans. It affects roughly 66 million people worldwide, the researchers

said. The Norwegian elkhound is sometimes called the "national dog of Norway." It and the Vallhund look like they could be cousins.

It's also stocky but not as slight as the Swede and more of a hunter and guard dog as a herder. It has similar markings but is silver-grey, the American Kennel Club says. It has a low-key temperament but gets very attached to its family and has a hard time being away from it.

Human glaucoma is caused by multiple factors, the scientists said in a study. But they found that a major form of glaucoma in the elkhound is associated with a mutation on the so-called ADAMTS10 gene.

Here, too, their work makes it possible to develop a genetic test to help breeders stop passing on the hereditary disease. And Komaromy's laboratory is working on a treatment for the blindness it causes.

The San Francisco-based Glaucoma Research Foundation has supported his work with a grant. *Source: Ben Brumfield, CNN, 3 January 2015.*

OCULAR NUTRITIONAL SUPPLEMENTS: ARE THEIR INGREDIENTS AND MANUFACTURERS' CLAIMS EVIDENCE-BASED?

Purpose

To compare ingredients contained in top-selling brands of ocular nutritional supplements with the Age-Related Eye Disease Study (AREDS) and AREDS2 formulae and investigate the validity of claims made by manufacturers of leading brands of ocular nutritional supplements.

Methods

We examined the five top-selling brands of ocular nutritional supplements in the United States according to dollar sales tracked by SymphonyIRI (Waltham, MA) from June 2011 to June 2012. We reviewed the ingredients and manufacturer claims of 11 ocular nutritional supplements on the companies' consumer information websites; the ingredients were compared with those contained in the AREDS and AREDS2 formulae.

Main Outcome Measures

Proportion of ocular nutritional supplements that contained the same ingredients, in the same doses, as the AREDS or AREDS2 formula; proportion of nutritional supplements with unsubstantiated claims made by the manufacturer.

Results

All of the ocular nutritional supplements contained the ingredients from the AREDS or AREDS2 formula; 36% (4/11) of the supplements contained equivalent doses of AREDS or

AREDS2 ingredients; 55% (6/11) included some information about the AREDS on their consumer information websites. Product descriptions from 4 of the 11 supplements (36%) stated that the supplements were important to maintain general eye health; none of these supplements duplicated the AREDS or AREDS2 formula. All the individual supplements claimed to "support," "protect," "help," or "promote" vision and eye health, but none specified that there is no proven benefit in using nutritional supplements for primary prevention of eye disease.

Conclusions

The majority of top-selling ocular nutritional supplements did not contain the identical ingredient dosages of the AREDS or AREDS2 formula and had product description claims that lacked level 1 evidence, underscoring the importance of ophthalmologists educating their patients on the evidence-based role of nutritional supplements in the management of eye health.

Source: Ophthalmology, March 2015. Yong JJ, Scott IU and Greenberg PB, from Department of Internal Medicine, Yale-New Haven Hospital-Waterbury Hospital, Departments of Ophthalmology and Public Health Sciences, Penn State College of Medicine, Pennsylvania and Section of Ophthalmology, Providence VA Medical Centre, Rhode Island, US.

ABOUT US

Retina Australia (ACT) Inc. is a member of the national body, Retina Australia (RA). Other members of RA are Retina Australia (NSW), Retina Australia (VIC) – which incorporates activities in Tasmania – Retina Australia (QLD), Retina Australia (SA) and Retina Australia (WA).

Our role, and that of our fellow organisations, is to provide information and support to people and families affected by Retinitis Pigmentosa and other retinal dystrophies. We also raise funds for scientific research into the causes and prevention of these dystrophies.

Retina Australia is a member of Retina International, which has members and affiliates in more than 50 countries. It is estimated that more than 20 million people worldwide are affected by some form of retinal dystrophy.

Mission Statement: To provide information and support to people and families affected by inherited retinal dystrophies, and, with the support of the Australian community, to raise funds to finance scientific research into the causes, prevention and cure of retinitis pigmentosa.

Ways You Can Help Retina Australia (ACT)

There are many ways you can help and support us. Listed below are some suggestions.

1. Suggest to family and friends that they hold a special event (such as a sponsored walk or fundraising dinner) to support our work.
2. Include a reference to us when updating your Will.
3. Continue with your much-appreciated cash donations.
4. Tell your local eye professionals (and your family and friends) if you have found the information provided by Retina Australia (ACT) and its support services useful.
5. Offer your services to a member of the executive as there are numerous small jobs that need to be done. Many of these do not require you to be on a committee.
6. If approached, consider allowing your medical information and DNA to be stored on the inherited retinal diseases database so it is readily available to eye researchers both here and overseas.
7. When you hear that someone in your community has been diagnosed with sight loss tell them about Retina Australia (ACT) and the support services offered.

Council Members 2013-2014

PATRON AND EXECUTIVE MEMBER:

Mr David Kilby
198 Brooklands Road
HALL ACT 2618
laststopambledown@westnet.com.au
(02) 6230 2280

VICE PRESIDENT:

John Barlow
C/- Elders Belconnen Real Estate
Jamison Centre
JAMISON ACT 2614
Barlowjohn@bigpond.com
0411097900

MEETINGS/MINUTES SECRETARY:

Mrs Lyn Barlow
Meriden
Wallaroo Rd
HALL ACT 2618
lynbarlow@optusnet.com.au
(02) 6230 2382

Mrs Barbara Burton
Mahbrook
100 Wyoming Rd
BUNGENDORE NSW 2621
Mahbrook@bigpond.net.au
(02) 6236 9210

PRESIDENT:

Mr Robin Poke AM
53 Glasgow Street
HUGHES ACT 2605
robin.poke@grapevine.com.au
(02) 6281 4519
0431 970 850

SECRETARY:

Mrs Jan James
12 Wynn Place
FRASER ACT 2615
mf_jm@optusnet.com.au
0421277880

TREASURER:

Mrs Doris Wallace
133 Mount Vernon Drive
KAMBAH ACT 2902
idwallaces@hotmail.com.au
(02) 6231 3822
0412305175

Mrs Elizabeth McLarnen
7 Stewart Street
MELBA ACT 2615
liz.mclarnen@gmail.com