



InVision

Canadian Trial of new Drug for Retinal Degenerations

A human trial testing a potential new treatment for two genetic forms of retinitis pigmentosa (RP) and Leber congenital amaurosis (LCA) has announced positive early results.

Canadian company, QLT Inc. is conducting a Phase I clinical trial of QLT91001, an oral drug for the treatment of some genetic types of retinitis pigmentosa (RP) and Leber congenital amaurosis (LCA). The trial is being led by Dr. Robert Koenekoop from the McGill University Health Centre Research Institute at the Montreal Children's Hospital.

"Only patients with mutations in the genes LRAT or RPE65 can participate," says Dr. Koenekoop, although he notes that trials of therapies for other genetic mutations may begin in time. Mutations in these genes cause some forms of LCA or RP.

This phase I proof-of-concept trial (see page 7) is designed to test the safety of the drug, and the principles behind it, in up to 28 people (half with LCA, half with RP).

Jörgen Gustafsson is part of the RP trial. He



Jorgen Gustafsson and Dr. Robert Koenekoop

developed night blindness in early childhood, and more advanced vision loss in his thirties. His intensive search for treatment led him to Moorfields Eye Hospital in England. After several years, genetic testing discovered Jörgen carried two mutations in his LRAT gene.

Jörgen's medical records were submitted for Dr. Koenekoop's review and Jörgen was deemed eligible for the QLT091001 trial.

For seven days this spring, he received

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Subscribe today to our monthly **Vision Research e-News** to receive emails about the latest sight saving research.

LETTER FROM THE EDITOR

Investing in the Future

Photo: Bryan William Jones, Ph.D.



Dr. Bill Stell

Research is our best hope for protecting and restoring vision – an investment in the future.

That is why I am proud to tell you about the return on your investment. In 2010, the Foundation Fighting Blindness invested 1.4 million

donor dollars in the ground-breaking work of 29 research groups from Atlantic Canada to British Columbia. The scientists you funded discovered new genes responsible for vision loss. They proposed potential new therapies. They showed that stem cells extracted from a human eye could restore sight to mice. They produced more than 50 scientific papers sharing their findings with a global network of collaborators.

Long-term support, from donors like you, is paying dividends. Your donations have trained some of our most influential scientists, such as Dr. Elise Héon at Toronto's Hospital

for Sick Children. Nearly three quarters of Foundation-supported research projects completed in the past 10 years went on to receive federal funding for related projects – making your donations reach even further.

Your investment has matured into an effective, internationally-connected community of Canadian scientists, but that doesn't mean our task is done. As our front page story illustrates, we now have the opportunity to move **scientific discoveries from Petri dishes to patients**. Your donation today brings us closer to treatments and cures tomorrow.

Please give what you can, to nourish your investment at this critical moment for scientific advancement. A donation card and return stamped envelope are enclosed.

Sincerely,



Dr. Bill Stell
Director of Research Programs

Are you receiving this newsletter and haven't made a donation to the Foundation Fighting Blindness this year?

It takes money to research, write, print and distribute our newsletters. Become a monthly donor today! Just \$5 a month will ensure we continue to provide you with the latest in sight saving research through our *InVision* newsletter and Vision Research e-News.

RESEARCH

Continued from page 1

daily doses of QLT091001 at the McGill University clinic. He will travel back to Montreal for follow-up appointments.

Preliminary results are now available from twelve people with LCA who have varying levels of vision. These results show improvements in visual acuity, visual field or both in 8 of 12 subjects. People with the most tunnel vision had the most striking improvements in visual field. Improvements in visual acuity were less common than visual field, but one person unable to see the eye chart at the start of the trial could read a few letters nine days after treatment. In two cases, improvements from this brief one-week treatment have lasted six months to a year.

Although short-term headaches and light-sensitivity were reported by some participants, there were no serious ill effects. Some patients had small increases in triglyceride levels, but these levels normalized during the two-week testing period.

“Clearly, additional studies are required to fully assess QLT091001 in these patients but we believe that these data are very

How Does QLT091001 Therapy Work? People with mutations in LRAT and RPE65 are not able to process vitamin A into a compound called 11-cis-retinal. This substance is necessary for the retina to capture light; if it is absent the photo-receptors in the retina become inactive, and eventually die causing blindness. QLT091001 is a synthetic, oral form of 11-cis-retinal, which people would take regularly to replace this “missing ingredient” and allow the retina to function.

promising,” said Bob Butchofsky, President and Chief Executive Officer of QLT Inc.

To benefit from treatments like QLT091001, people living with genetic retinal diseases will have to know which mutation(s) they carry. Fortunately, genetic testing is now more available in Canada, although testing may still be a long process.

Dr. Koenekoop is positive about the continued advancement of genetic testing. “A few years ago, we had no way to know the underlying mutations that caused retinal diseases, virtually all of these genes have been discovered in the past 25 years. Today, testing is available in centres across Canada, and the costs of gene sequencing and other techniques are dropping fast.”

Dr. Koenekoop and his Foundation Fighting Blindness-funded research have been an important part of genetic testing progress. He and his colleagues Dr. Anneke de Hollander and Dr. Frans Cremers have identified several genes responsible for RP and LCA. He also worked with Dr. Rando Allikmets to create a simplified genetic testing chip.

“Foundation Fighting Blindness donors have been and continue to be a critical part of my work,” says Dr. Koenekoop. “They have helped us identify genes and develop the chip technology that made this clinical trial possible and they continue to support my genetic testing work. Canadian donors should be proud of the advances their donations have made possible.”

Learn more in our **Genetic Testing Fact Sheet**. Find it at www.ffb.ca or call **1.800.461.3331**.

RESEARCH

Your FFB-funded Scientists

Today, with your donations, 18 research groups from Halifax to Vancouver are pursuing sight saving discoveries. More scientists will be funded this summer, as our Board of Directors acts on the expert advice of our Scientific Advisory Board.

Dr. Valerie Wallace, Chair of the Foundation Fighting Blindness (FFB) Scientific Advisory Board, knows our scientists well. For six years, she has volunteered her expertise to determine the projects that make the best use of donor funds.

“FFB-funded scientists work in a broad range of fields,” she says, “areas that bring us closer to effective treatments. In addition, the FFB matches donor dollars with federal funds from the Canadian Institutes of Health Research (CIHR) to bring together teams of experts for key treatment studies.”

How the Eye Develops

Understanding how the retina and optic nerve develop and how these delicate structures maintain their functions is vital. Your donations support Dr. David Eisenstat in Winnipeg and Dr. Judith West-Mays in Hamilton both studying genes that guide the developing eye. Other scientists, including Dr. Andrew Waskiewicz in Edmonton, Dr. David Picketts in Ottawa and Dr. Alicia Ebert in Calgary, look at how the cells of the eye communicate, and how cells prompt

others to grow and make connections.

“These projects give us critical information,” says Dr. Wallace. “For example, Dr. Picketts looks at how nerve cells build connections between the retina and the brain. To restore vision, we must understand how to stimulate and maintain these links.”

Specific Causes of Vision Loss

Research to understand and treat genetic diseases is making headlines, and Canadian scientists including Dr. Robert Koenekoop (see front page) and Toronto’s Dr. Jane McGlade, who also studies retinitis pigmentosa (RP) and Leber congenital amaurosis (LCA), are at the forefront. Gene therapies for Stargardt disease, cone-rod dystrophy and other conditions are being developed and tested under the direction of Vancouver’s Dr. Robert Molday, in an ambitious project with matching funds from CIHR.



Dr. Kevin Gregory-Evans

Photo: Martin Dee (UBC Photographer, Public Affairs)

“We’re seeing some positive results from gene therapy trials, so there is tremendous buzz in this area,” says Dr. Wallace. “However, other strategies are also needed – especially for conditions like age-related macular degeneration, which has a less clear-cut genetic cause, or for conditions like RP, where there

are many genes, making it challenging to develop specific gene therapies.”

That is why your donations also fund a major team project led by Dr. Kevin Gregory-Evans in Vancouver. This FFB-CIHR project brings together experts in the molecular biology of vision loss – exploring how drugs might be combined to slow disease. It will produce the most immediate benefits for people with age-related macular degeneration, but experts in several areas are involved. One of these is Dr. Orson Moritz, who is also funded for his work on the causes of RP vision loss (see page 7).

Preserving Vision

In addition to disease-specific treatments, your donations fund more general efforts to delay photoreceptor loss, thus preserving vision.



Dr. Catherine Tsilfidis

Ottawa’s Dr. Catherine Tsilfidis has developed XIAP gene therapy and has shown that it can prevent photoreceptor death in animals. With funds from FFB and CIHR, she and her colleagues are scaling up the therapy to prepare for human trials in the next few years. Other funded scientists, including Toronto’s Dr. Rod Bremner and Calgary student Robert Cantrup, study protein factors that might protect photoreceptors from destruction.

Restoring Sight

For many FFB supporters, the ultimate goal is to reverse blindness. Stem cell therapy is one promising approach. With your donations, Toronto scientist Dr. Vince Tropepe studies how stem cells rejuvenate the retina in zebrafish (see page 6). Montreal scientists,



Dr. Michel Cayouette

Dr. Michel Cayouette and Dr. Gilbert Bernier, explore ways to prompt stem cells to produce photoreceptors. Our largest stem cell project, a joint project with CIHR, is headed by Dr. Wallace herself and includes experts from Ottawa, Toronto and Calgary working to overcome the technical challenges of stem cell transplants.

“I believe that one day, stem cells will be used to restore sight,” says Dr. Wallace, “but stem cells are not the only therapy with this potential.” She points to Dr. Gautam Awatramani in Halifax, who is using light-sensitive molecules from plants to reprogram the eye’s nerve cells, making them capable of sensing light even after the rod and cone photoreceptors have been lost.

“People think of scientists as meticulous, and we are, but science is also a creative field. By supporting the innovation of Canadian scientists, FFB donors bring sight saving discoveries closer.”

RESEARCH

Research Findings from Fish

Next time you visit your local pet store, meet a vision research star – the zebrafish. This tiny creature has an important place in labs across Canada.

Over the last twenty years, zebrafish have helped scientists document the eye's development. A pair of fish produces 200 clear eggs weekly, so scientists can literally watch the eye grow, reaching full maturity in three weeks. Although the fish is small, the photoreceptors are larger than human ones, simplifying study. The fish are also easy to care for – hundreds live together happily in a single tank.

Although zebrafish and people don't seem to have much in common, we are all vertebrates. Zebrafish have a spine and the complex of nerves associated with it. There are also genetic similarities. For example, Dr. Andrew Waskiewicz, of the University of Alberta, discovered that a gene mutation which impairs the vision of zebrafish, was similar to a one found in some people with Leber congenital amaurosis. He is now using zebrafish to learn how this mutation causes vision loss and to screen potential therapies. Similarly, Dr. Alicia Ebert in Calgary is using zebrafish to understand how the eye builds connections to the brain through the optic nerve. Both scientists receive support from your donations.

Zebrafish eyes are different from human eyes in one critical way. The fish's eye can use stem cells in the retina to repair itself. Several years ago, a team of Foundation Fighting Blindness-



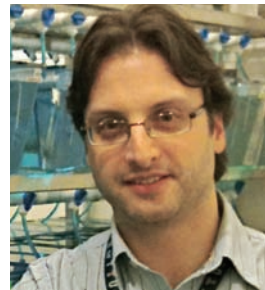
Zebrafish

Photo: Dries Knapen, Creative Commons

funded scientists, including Dr. Derek van der Kooy and Dr. Vince Tropepe, showed that adult human eyes also have stem cells, although the human version is relatively inactive.

Today, Dr. van der Kooy works to harvest stem cells from human eyes and harness them for transplants, but Dr. Tropepe has a different approach. He studies the genes which activate zebrafish stem cells.

"Imagine if we could learn from zebrafish how to stimulate stem cells in the human eye," says Dr. Tropepe. "A person could re-grow their lost vision."



Dr. Vince Tropepe
Associate Professor,
Cell & Systems Biology
and Ophthalmology
& Vision Sciences,
University of Toronto

"My dream is to learn from nature how to help the retina heal itself. I am grateful for the generous support of Foundation Fighting Blindness donors who are helping to make this dream possible."

Hear Dr. Tropepe talk about his FFB-funded work at www.youtube.com/user/FFBCanada.



FFB Science Class – Human Clinical Trials

Your donations continue to pave the way for new treatments. In addition to the QLT091001 trial (page 1), human trials for wet AMD are underway in Canada. The following phases of human trials are done, following successful laboratory studies, as the ultimate test of any treatment.

Phase 1 trials involve a few people, test the drug's safety, and help determine the dose. Phase 1 trials may give hints about the treatment's benefits, but there are too few participants to be conclusive.

Phase 2 trials involve more people. They aim to fine-tune dosages and to provide the first conclusive evidence that a treatment will be effective.

Phase 3 trials gather the evidence that a treatment needs for approval by government regulators, including details about treatment outcomes and side effects. Hundreds of people participate, although trials for rare genetic conditions will be smaller. In a phase 3 trial, the new treatment is often compared to a placebo (a fake treatment) or to existing treatments, to make its benefits and risks clearer.

Phase 4 trials are done after the drug has received government approval, to gather information about the treatment's long-term use.

The Science of Sunglasses

While many Canadian scientists study zebrafish, Dr. Orson Moritz uses tadpoles to understand how retinitis pigmentosa develops and to test new drugs to treat it.

“Tadpoles have many of the advantages of fish, such as quick reproduction and simple care, but it is easier to make genetic changes,” he says. This advantage has allowed Dr. Moritz to breed a family of frogs with autosomal dominant RP and to study influences on their vision.

Groups of these tadpoles were raised under different light conditions. This research has shown that brief exposure to bright light did not harm the tadpole's retinas, but prolonged exposure to short wavelengths of light (green and blue) caused blindness. Dr. Moritz's results have an immediate message: sunglasses that block short wave lengths of light (those with orange coloured lenses) may best protect the eyes of people with RP.

Light exposure has also been studied for those with age-related macular degeneration (AMD). Again prolonged exposure to bright light, particular short wavelength light, seems to accelerate vision loss. Studies like these have led ophthalmologists to recommend the regular use of sunglasses with yellow or orange lenses for all people with retinal degenerative diseases.

EVENTS

COMIC VISION

Proudly presented by **Bell**



Photo: Clive Tonge

Comic Vision took place in five cities across Canada: **London, ON** (April 26), **Oakville, ON** (April 27), **Toronto, ON** (April 28), **Vancouver, BC** (May 4) & **Calgary, AB** (May 5). Thanks to our supporters, volunteers and sponsors, Comic Vision 2011 reached a new high point raising over \$1 million for sight saving research.

Become a distinguished sponsor of our 2012 national tour; contact **Jamie**

King at jkking@ffb.ca. Get involved and volunteer; Visit www.comicvision.ca or call **1.800.461.3331** for more information.

Comic Vision: Last Call tickets now available. Comic Vision: Last Call will be held on Thursday, July 7th, 2011 at downtown Toronto's historic Steam Whistle Brewery. To purchase tickets, visit www.comicvision.ca.

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cycle for sight

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Participate in Cycle for Sight and help us reach our 2011 fundraising goal of \$300,000 for vision research!

Ride Solo – Ride Tandem – Ride Together
 Register at www.cycleforsight.ca
 Ottawa: Sat, June 18: 50km | 100km
 Toronto: Sat, June 25: 70km | 140km

For more information contact Cycle for Sight Event Coordinator, Stephanie Willet at info@cycleforsight.ca or 1.800.461.3331 ext 230.



Michael Ovens and Catherine Bancroft

Thank you to our 2011 event sponsors and supporters:





Ride for Sight supports vision research funded by:  The Foundation Fighting Blindness a cure is in sight

JUNE 11 2011
 Maritime Ride for Sight
 Steeltown Park, Trenton, Nova Scotia

JUNE 10-11-12 2011
 Alberta Ride for Sight
 Stoney Nakoda Resort & Casino, Morley
 (Hwy 40 & Hwy 1)

JUNE 17-18-19 2011
 Central Ontario Ride for Sight
 Tudhope Park, Orillia
 (Hwy 12 & Atherley Road)

JUNE 24-25-26 2011
 Newfoundland & Labrador Ride for Sight
 Gander Community Centre, Gander

Join the fight against blindness and start fundraising today.
 Call 1.800.461.3331 or visit www.rideforsight.com



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MAKING A DIFFERENCE

Monthly donor encourages others to do the same

Monthly donor Jan Ross first noticed her vision was blurry when applying eye makeup. Her ophthalmologist diagnosed her with wet AMD three years ago at age 69.

“When I first learned I had AMD, I was very concerned about losing my driver’s licence and my general independence,” says Jan who now finds it difficult to read product labels or the newspaper.

Jan’s made lifestyle changes to slow the disease’s progress. She wears tinted glasses, and a wide-brimmed hat. She takes AREDS vitamins (a combination of vitamins tested in the **Age-Related Eye Disease Study**) and tries to eat a healthy diet with “lots of spinach”.

Jan attended a Vision Quest conference in 2010 - her first contact with the Foundation Fighting Blindness. “It is very helpful to know that research is happening and that there are lots of other people out there with the same condition,” she shares.

For people like Jan, research has begun to deliver options. “I’m encouraged that I’m able to receive eye injections for AMD,” she says, referring to the wet AMD treatments approved in recent years. She believes her donations will lead to more progress.

“I donate to the FFB and encourage others to do the same to hopefully find a cure for AMD and other eye conditions,” she says.

Progress relies on consistent monthly donations like Jan’s – “A Cure is in Sight!”



Jan Ross

You can support sight saving research today. Call **1.800.461.3331** or visit **www.ffb.ca** to:

- Become a Monthly Donor
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- Start a Sight Saver fundraising page
- Shop our Eye Opening gift catalogue
- Celebrate a special occasion with a tribute gift

EVENTS



The Foundation Fighting Blindness

a cure is in sight

Vision 2011 Quest



Save the Date

Halifax, NS: October 22, 2011

World Trade Convention Centre
1800 Argyle Street
Chairs: Dr. Johane Robitaille; Dr. Alan Cruess

Toronto, ON: October 29, 2011

Toronto Reference Library
789 Yonge Street
Chairs: Dr. Elise Héon; Dr. Michael Brent

Vancouver, BC: November 5, 2011

Library Square Conference Centre
350 West Georgia
Chair: Dr. Kevin Gregory-Evans

Vision Quest 2011 – from coast to coast

Learn about your eye disease and the Foundation Fighting Blindness' sight saving research!

Sessions will include:

- New Developments, New Hope: Translating Research into Treatments
- Getting the Most of the Vision You Have
- The Promise of Gene Therapy
- Vision Research in Your Community

Register for:

Retinitis pigmentosa & related retinal diseases: 8:30 AM – 12:30 PM

Age-related macular degeneration:
1:00 PM – 4:00 PM

Register today for early bird rate: \$30
or after September 12: \$40

www.ffb.ca | 1.800.461.3331



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The Foundation Fighting Blindness

a cure is in sight

InVision



Your donation to the Foundation Fighting Blindness funds sight saving research.

InVision is our commitment to share the impact of your support. Be a part of the search for causes, treatments and cures of retinitis pigmentosa, macular degeneration, and related retinal diseases.

View **InVision** online and sign-up for Vision Research e-News at www.ffb.ca.

We want to hear from you!

Share your comments and stories, or update your contact information:

The Foundation Fighting Blindness

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Toronto, ON M4W 3P4
info@ffb.ca | 1.800.461.3331

We hope you have enjoyed the first edition of **InVision**. Our readers were an important part of selecting the new name.

We received plenty of great suggestions, but the winning title, **InVision**, came from Lynn Mercer-Boundridge of St. John's NL. In an online poll at www.ffb.ca, 50% of you chose this name over four other possibilities.

Thanks for sharing your ideas!

Thank You For Making a Difference!