



RETINA E-News

No. 11 January 2010

Seeking a cure for Retinitis Pigmentosa, Macular Degeneration and allied Retinal Dystrophies

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RETINA E-NEWS to YOU

MESSAGE FROM THE CHAIRMAN - James Cape

There can be no doubt that the global financial meltdown has had an adverse effect on the Foundation. 2009 Was filled with challenges, yet it has been rewarding with the amazing progress made in the scientific field. It is indeed pleasing to see that our hard work and effort is starting to bear fruits and the light at the end of the tunnel is no longer a dream. The dedication and focus of branches, volunteers and staff in times when it is easy to lose sight of our goals is much appreciated. We should be proud of our collective achievements and performance in a year that can safely be described as the toughest in a decade.

Perhaps the best measure of our worth is not the success achieved in good times, but how we adapt and harness the opportunities when times are as difficult and challenging as they are now. As the saying goes: adversity has the effect of eliciting talents which in prosperous circumstances would have lain dormant. These testing conditions are set to continue in 2010, and will force us to remain vigilant and nimble. To a large extent we have been equal to the challenges of this difficult period and have measured up to the lean times that confront us. However, this journey is far from over and governments, businesses and individuals all face the same uncertain future. To my colleagues in the Executive and Branches, your unselfish dedication made the year possible by dutifully delivering the services that are the backbone of our Foundation and shared throughout the Retina family. Good luck for the year ahead.

A DECADE IN REVIEW By Claudette Medefindt, e-News editor

A new decade demands reflection on the past and encourages speculation on the future. What have we achieved during the last decade? Reviewing our past newsletters caused a certain amount of sadness coupled with tremendous pride and hope. Sadness, to remember all those who have passed away that fought so bravely against blindness: Edith Allpass from Pietermaritzburg, Tich Cousins, Kenneth Cape, Joyce Cousins and Adele Kearney from Gauteng, John Mansfield, John Foster Greenwood and Alec van Schoor from Cape Town and Fluris Strydom from Pretoria. We have also sadly

parted ways with many who gave many years to our cause: Our national chairman of 25 years Gordon Cousins, Pat Woodland, Willie and Elsabe Reichart, Poppie Strydom, Anne Heard [Metcalf] and many others.

But on the positive side so much has been achieved that we can all be very proud of the past decade. We changed our name from the Retinal Preservation Foundation, we became more professional, appointed a CEO- Ebrahim Patel, launched our Enews, continued to grow the Ride for Sight and achieved a high profile in the public media. Our remaining branches have matured into professional and dedicated teams and the Book of Retina – documenting procedures and practices is now a useful tool to them.

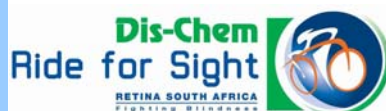
Our local researchers discovered 2 novel genes for Dominant RP. We supported the development of the Micro-array Chip for quick screening of RD and hundreds of families have had positive results. Most of these results have been delivered to them and only funding constraints prevent the balance from being delivered. This has made a huge impact on the lives and extended families of all these patients.

Internationally we have seen the advent of the first ever gene replacement trials to treat the RPE 65 form of Leber Congenital Amauroses [LCA]. This remarkable event is already paving the way for other genetic trials.

The first effective treatment for wet AMD – Lucentis is now saving the vision of thousands of patients around the world. The efficacy of Anti – oxidants to slow the progress of both AMD and RP is now widely accepted and we have managed to get a local supplier to produce the Anti – oxidant for RP at a reasonable cost.

And what of the next decade? It is never wise to speculate about science but we can reasonably expect treatment trials for more forms of Retinal Degeneration, and that some of these treatments will become commercially available. Stem cell therapy will probably come of age and this will offer real hope to those with advanced degeneration. The artificial retina will also provide some form of vision to those who have no photoreceptors to regenerate. But a quote from Time magazine that we used in a newsletter in 2002 is still relevant: *We know that with determination, money and energy you can hurry Science.*

DIS-CHEM RIDE FOR SIGHT 2010



Dis-Chem Pharmacies are once again the naming sponsors for the annual Ride for Sight. The event will be held on Sunday February 21st February 2010 at the Boksburg City Stadium. We are

appealing to all cyclists to help fight blindness by supporting the event and in particular the visually impaired tandem cyclists. Cash prizes will be given to the first 3 tandems in the Disabled Tandem category in the 116km event. For more details or to offer help on the day contact the national office.

WORLD RETINA WEEK: 21-27 SEPTEMBER 2009

Retina South Africa would like to thank Novartis South Africa who assisted us in creating awareness of Age Related Macular Degeneration during this global event. The campaign was a tremendous success and we received excellent exposure on TV, radio and in the print media. Hundreds of callers to our share call number resulted in many people receiving telephonic counseling about their retinal condition and the treatment options now available for wet AMD. The poster and pamphlets featuring the Amsler grid were distributed throughout South Africa and we are still able to supply on request.

GETPIX VISIONS MASKED BALL



Pictured at the ball- Norman and Claudette Medefindt with James Cape, National Chairman and his wife Sandra on the right. Paula Rutherford from GetPix presents the cheque for R24000 to Claudette

GetPix Photographic Studio nominated Retina South Africa as one of the beneficiaries of their Vision's ball held at Emperors Palace on 12th September 2009. Prizes were awarded to top photographic students in various categories and Retina South Africa was awarded R24 000 from the proceeds. We would like to thank GetPix for this generous donation and all the sponsors: Emperors Palace, Cést La Vie, Lakefield Photo Express, Colour Me Green, Kopanong Hotel, Space Light, Accor Hospitality, Mount Grace Hotel, Irene Country Lodge, Cullinan Diamond Inn, Crystal Restaurant, Add Web, Lenzz – Rezzt, Tao Designer Glass, Tiger Brands, Colgate Palmolive, Peppina. Phomola, Waterberg Transfers, Thandeka Lodge, Flora Farm, Monte Casino, Ocean Basket, International Travel and Tours, Canon, Singer, Authentic Events, Dunblane Estates and Sign Girl.

INTELLIPEN COMPETITION WINNER:

The Intellipen was won by Harry Schwarz, a friend of Retina South Africa.

The pen is a remarkable way to improve communication for partially sighted people. It digitally captures handwriting, drawings and sketches which can then be stored on the USB Flash drive and uploaded to a computer. The special offer of a 20% discount is still open to readers of Enews.



UPDATE ON RETINAL RESEARCH

The results on the LCA trial are continually improving. While all 3 centers doing trials are investigating the RPE 65 gene that causes Leber Congenital Amauroses [LCA] they are investigating various strengths of the gene therapy and areas of the retina to be treated. Over the past 2 years more than 20 patients have all had significant restoration of vision. The trials still need to go through 2 more stages before becoming freely available. The success has paved the way for other gene therapy trials and those starting soon include:

- Applied Genetic Technologies Corporation trials to treat LCA, Age Related Macular Degeneration, Retinoschises and Achromotopsia
- Oxford Biomedica trials for Stargardt dystrophy and possibly Usher Syndrome
- Genable [Ireland] therapy to treat Dominant forms of RP
- An international consortium including researchers from Saudi Arabia to treat the MERKT form of recessive RP

For more information see www.blindness.org.

IS A GENETIC TEST RIGHT FOR YOU?

Patients who wish to be eligible for gene specific therapies in the future will need to know precisely which gene mutation is causing their retinal degeneration. Most of the DNA samples that have been banked at UCT over the past years have been tested to some degree or another but not all the genetic mutations have been identified. Many families have had their genetic results but at least 250 results are waiting to be delivered and Retina South Africa does not have sufficient funding for this. If you donated a blood sample in the past and would like to know if a result is available for you; and if you or your Medical aid will fund the cost of the Genetic Counselor who can deliver the result [between R400 to R750] then please send an email with your full name, ID number, diagnosis, telephone numbers and email address to the national office. We will then forward these names to UCT. The delivery of results process will then start. Patients at State hospitals where there is a genetic counselor will only have to pay a nominal fee.

If you have not donated blood but would like to know which gene is causing your retinal degeneration you will need: A Confirmation of Diagnosis from an Eye Specialist; a pre- test session with a registered genetic counselor and be willing to contribute towards the costs. For LCA patients only, a limited amount of funding has been set aside to assist severely economically disadvantaged families for gene tracking. Comprehensive motivations for funding should be sent to the national office for consideration.

Knowing the genetic cause can also help patients understand their condition, determine risk factors, identify carriers and in some cases determine the rate of degeneration of the condition. For more details on gene tracking please contact the national office.

PROGRESS REPORT FROM UCT

Professor Ramesar gave an excellent update to the National Management committee meeting held in December.

Stargardt Project: Of the 180 SA families that have been screened for ABCA4 mutations, 105 families are now completed and can have their results delivered. UCT are also looking at the most common mutations in South Africa, ways of developing a quick test for these and the link between these and rates of degeneration.

Recessive RP: 57 DNA samples have been sent for Micro-array screening to Estonia and 4 families have been completely characterized* and can have results delivered.

Editors Note * Characterized means both DNA mutations found.

Due to the diversity of recessive RP, this chip has given us the poorest results. The Gene sequencing test now available in Germany would yield better results but the costs are astronomical.

LCA: In 2009, 3 families' results from 2008 were confirmed and have had their results delivered. An additional 3 DNA samples were tested on the LCA chip. None were completely characterized and their result delivery is pending.

Usher Syndrome: In 2009, 2 DNA samples were sent for testing. Neither was completely characterized and both results have been delivered.

Up to November, 2009 results from the UCT database are:

Condition	Families on Database	Families with gene result
Dominant RP	91	19
Recessive RP	91	5

Other RP	300	8
LCA	30	4
Usher Syndrome	72	7
RP multiplex*	325	7
Stargardt	226	121
Other RD	48	4
X- linked RP	24	3
Total	1207	181

*RP Multiplex is where there is more than one affected individual in a family but the exact inheritance mode is unclear.

From the 1207 families on our database, there are 2934 DNA samples stored at UCT. 181 Families [14.99%] are completely characterized and for 139 families, the DNA results have been verified. To date 220 individuals have received a result (either positive, negative or inconclusive) of which 28 had results delivered in 2009.

UPDATE FROM PROFESSOR JOHN FLANNERY

Drs Natalie Corporale and John Flannery are among the team of researchers at the University of California, Berkley working on increasing the ability of blind mice to respond to light by manipulating existing proteins that our brains normally use to transmit information between neurons. They have focused on an engineered protein, LiGluR (Light Activated Glutamate Receptor), which can turn neuronal activity on and off upon illumination with specific wavelengths of light. They are hoping that that this approach will lead to a viable therapy for people who have lost significant vision due to late stage retinal degeneration. They can consistently restore light responses in mice with no light perception using these photoswitch molecules. They now need to determine the quality and type of this vision. They are testing to see whether the animals can negotiate a maze and see edges and contrast. As yet they do not know if the mice can see shape and form or just the ability to sense the light. Their results will be presented at the ARVO conference in May, 2010.

NEWS ON THE ARTIFICIAL RETINA

If you are interested in the technical aspects of the retinal prosthesis see: http://news.cnet.com/8301-17938_105-10361725-1.html

This report features the MIT project but has links to other projects and interesting videos.

Interesting Links

www.retina-international.org

www.fightblindness.org

www.amdalliance.org

www.Novartis.com

www.intellipen.co.za

NEW ASSISTIVE DEVICES

Electronic Reader: A reading device has been developed by Intel who produce the electronic processors used in many computers. The device does not require a computer and will be wonderful for people who are not computer literate. For more information see:

<http://www.intel.com/healthcare/reader/index.htm>

Smartview Versa Plus Video Magnifier: This new device re-defines the handheld electronic magnifier. It also has multi media features with magnification up to 15X. For more information contact:

Anton@nctec.co.za.

RETINA INTERNATIONAL CONGRESS 2010

The 16th International congress will be held in Stresa, Italy on the 26th and 27th June 2010. James Cape and Claudette Medefindt will represent Retina South Africa. For more information see:

www.retinaitalia.org

ARTICLE ON VISION LOSS

Dr Shirley Murray has written an excellent article on Adult Vision loss which will help patients and their families understand the emotional turmoil of vision loss. For a copy contact national office.

ANNUAL MEMBERSHIP FEE

Annual Membership/Subscription fees of R100 are now payable to the National Office or your branch. Direct deposits/EFT payments to: Retina South Africa at Standard Bank, Branch Code 011-642 (Germiston), Account No. 020312164. Please add your name and branch in the reference line. For card payments please contact the National Office.

If possible PLEASE add a donation towards the shortfall of the research grant to UCT. We are truly grateful, thank you.



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