



**RETINA SOUTH AFRICA**  
Fighting Blindness

# RETINA E-News

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

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An amazing retinal research partnership!

## RETINA E-NEWS to YOU

### SPECIAL CONGRESS EDITION

### HELSINKI HIGHLIGHTS

The 15<sup>th</sup> Biennial Retina International World Congress was held in Helsinki, Finland from 2<sup>nd</sup> July to 5<sup>th</sup> July 2008. Representing Retina South Africa were James Cape and Claudette Medefindt. Sandra Cape accompanied us to provide much needed visual support.

Helsinki was a fitting venue for the presentation of the success of the first ever human clinical trial for the replacement of a defective gene causing a retinal degeneration. Fitting, because it was here in 1984 that Professor Shomi Battacharya announced the localization of the first X-linked RP gene. This was groundbreaking news then, just as Professor Robin Ali's work is today. Professor Ali's presentation on the replacement of the defective RPE65 gene that causes Lebers Congenital Amaurosis was a highlight of the conference that was already brimming with good results in many scientific areas.



From left: James Cape, David Waldron, President Fighting Blindness Ireland, Claudette Medefindt, Prof Robin Ali and David Head, CEO British RP Society at the congress Gala Dinner

Professor Ali demonstrated with an exciting video the remarkable improvement in night vision that Stephen Howarth had achieved within 6 months of receiving the gene therapy. He reminded us however that this success was the result of 15 years of research. Proof of principle

A lifetime of dedication  
honoured

Promising treatments  
of both RP and dry  
AMD

Artificial Retina Update

that the replacement therapy worked in animal models was already established in 1999. The planning of the trials started in 2004 and involved dozens of researchers from many different research modalities. The 1<sup>st</sup> phase was conducted on 3 patients and was mainly to determine safety factors. Further trials would investigate higher doses and younger patients who would probably have better results. The success of this trial would also speed up the process for other trials. Prof. Ali said that the trial had demonstrated that we can deliver genes safely to the retina but more investment was needed to move the research forward.

### **PROFESSOR ALAN BIRD**

Professor Alan Bird of Moorfield's Eye Hospital, London was honoured at the congress for a lifetime of dedication to the understanding of Retinal Degeneration. Professor Bird's key note lecture was an elegant presentation of the past, present and future of research into Retinal Dystrophies. He highlighted, as many others did during the congress, the importance of having well documented patients available to the clinicians for future treatment. He also highlighted very effectively the difference between treatment for patients that had photoreceptor cell dysfunction and for those where loss of vision was caused by cell death. The other speakers at the congress presented papers on various types of research that targeted these 2 differing scenarios.

### **DR WENG TAO**

Dr Tao presented the results of the phase 2 and 3 studies of the effect of growth factors on patients with both RP and Dry AMD. The growth factor CNTF is delivered via an encapsulated cell which is implanted in the vitreous part of the eye. The therapy is not gene dependant and is being tested on many types of RD including Choroideremia. Early results are very encouraging and some patients had experienced a significant improvement in vision. Growth factors would be effective where some photoreceptor cells are still viable.

### **DR JOHN FLANNERY**

Dr Flannery was one of the guests on our speaker's tour of 2006 and is one of the leading neuro-scientists in the world. He presented a remarkable new and innovative way of possibly restoring vision in advanced RD where photoreceptor cells have been completely lost. His project involves manipulating retinal ganglion cells to convert them into light sensitive cells. These ganglion cells transport message from the photoreceptors to the optic nerve and survive even in advanced stages of RD. These cells are being effectively used in the artificial retina research projects. Although still in the early stages, Dr Flannery's research is a very promising avenue of treatment in advanced stages of photoreceptor cell death.

### **ARTIFICIAL RETINA**

There were excellent presentations on 2 different projects to develop an

artificial retinal implant. These are being developed to restore vision when photoreceptors have been completely destroyed.

Professor Eberhart Zrenner presented results of his sub retinal chip. The chip is 3 x 3 mm in size and consists of 1500 micro photodiodes arranged in a 4 x 4 array. The array is stimulated in various patterns and patients could distinguish horizontal lines from vertical lines and their positions. They could also detect dot alignment and direction of movement. With the correct stimulation patients could also find a white plate on a black background.

Professor Mark Humayun presented the findings of his project with an epiretinal prosthesis. The project is called Second Sight and the implants were tested on 4 patients blind from end stage RD. They were assessed in 4 different tasks and scored significantly better than chance in 83% of the tests. They had to locate and count objects, differentiate between 3 objects, determine the orientation of a capital L, and judge the direction of a moving object. His conclusion was, that using the device blind subjects can differentiate and localise objects in their environment. He felt that the development of prosthesis with more electrodes provides higher spatial resolution vision to blind patients.

#### **PROFESSOR THEO VAN VEEN**

Professor van Veen whose work on anti-oxidants in animal models of RD was first presented in Brazil gave an update on his findings and other similar work into the effect of anti-oxidants. Studies in 9 different mouse models of RD have shown oxidative damage in the photo receptors that is probably due to the hyper-oxic environment created by the progression of the disease. Oxidative stress can also be caused by genetic defects or lack of neurotrophic factors. The 2 studies in animal models showed that the use of a combination of high levels of anti-oxidants delayed photoreceptor cell death. Ophthalmologists have generally been reluctant to advise RD patients to use these anti oxidants due to the lack of a patient trial. The results of a patient trial in Spain into the effect of these anti-oxidants are expected by the end of 2008. At the Annual Retina International Scientific and Medical Advisory Board [SMAB] meeting held at the ARVO congress in April the board issued the following statement on the use of anti oxidants for RP:

*“Independent evidence from well respected laboratories agrees that combinations of antioxidant supplements are successful in slowing retinal degeneration in RD animal models. Positive effects in all these animal models may indicate that such treatment could be effective in most or all forms of RP and allied diseases irrespective of molecular diagnosis.*

*Safety seems assured from the animal testing done to date and from the fact that the supplements used are known to be well tolerated in humans and are not controlled substances.*

Retinal anti-oxidants  
safe to use – RI SMAB

*Retina International looks forward to the results of human clinical studies for this promising treatment to slow the progression of Retinal Degeneration. These studies are ongoing at the Mediterranean Ophthalmology Foundation, Valencia, Spain (Prof. F.J. Romero)”.*

For a copy of this report please contact Retina South Africa.

Other interesting papers were given on:

- **Stem Cells** – by Professor M Perez who highlighted the success but also the major challenges still faced by this type of research.
- **Usher Syndrome**- Professor John Flannery gave cautious hope that at least for Usher Type 3a genetic replacement was possible.
- **Early onset RP, LCA**- Professor Elise Heon gave an excellent overview of the complexity of this rare but topical condition. The gene therapy trials of Ali, Bennet and Jacobson are all focused on the RPE 65 subtype but Professor Heon reminded us that to date 11 different gene mutations have been identified and a further 2 loci on Chromosome 1 and 14 have yet to be finally understood. Gene therapy for each different gene mutation in each different condition seems to be an almost insurmountable goal.
- Excellent papers were also given on all the forms of Macular Degeneration – Age Related, Stargardt and Best. Many treatments are in or progressing toward clinical trials.
- The control of risk factors for AMD such as Blood pressure, obesity, smoking, exposure to Sunlight and nutrition were also highlighted.
- Genetics, Genetic Counseling and the importance of Patient registries were the focus of numerous papers. This was also discussed at the business meetings of Retina International and South Africa can be very proud of the progress we have made in this area.

### **PROFESSOR JOE HOLLYFIELD**

Professor Hollyfield is co-chair of the Retina International SMAB and also a valued member of Retina South Africa’s SMAB. He gave an excellent overview of the advances in the understanding of the complex disease processes in AMD. Professor Hollyfield’s recent success in the development of a mouse model will significantly affect the development of treatments for this important and widespread condition.

### **PROFESSOR JERRY CHADER**

Professor Chader is the secretary of the Retina International SMAB and also a valued member of Retina South Africa’s SMAB. Professor Chader gave 2 papers at the congress - one on therapy trials and the second an overview of present and future therapy for Retinal Degenerative

Diseases. He reminded us that it is estimated that only half of the genes for RP have been identified and probably most of the genes for AMD. On therapy trials he stated that clinical trials were an important link between the laboratory and the patient. Phase 1 trials were primarily to test for safety factors and therefore only a small number of patients were selected. Phase 2 and 3 trials were again to test for safety but also for effectiveness. Clinical trials had already resulted in treatments for Wet AMD. There were 5 important areas of research where clinical trials are in progress or being planned:

- Gene therapy- 3 clinical trials for RPE65 in LCA by Ali, Bennet and Jacobson were underway and reports by Ali and Bennet showed great early results. Gene therapy trials to deliver growth factors for wet AMD were also underway. Gene therapy trials for Stargardts and Usher Syndrome are in the planning stage.
- Pharmaceutical intervention- Encapsulated cell technology to deliver growth factors for Dry AMD and RP was in clinical trials.
- Nutritional Studies- the AREDS study had shown the importance of Anti oxidants in AMD and the AREDS2 trial is testing the effectiveness of Lutein and Zeaxanthin in AMD. The patient trial to test Anti-Oxidants in RP was underway.
- Artificial Retina- Many different centres were investigating this and results of 2 trials were both showing great promise.
- Photoreceptor Cell transplantation trials have not been that successful. Stem cell research offers great hope for the future but some challenges remain. Both of these areas and the artificial retina would hopefully one day replace dead or non – functioning photoreceptor cells.

He ended with a true message of hope: “We are beginning to see that research is turning into practice – giving hope to all RD patients”

## **RI BUSINESS MEETINGS**

The Continuing Education and General Assembly meetings were held at the Iris Centre, the headquarters of the Finnish Society for the blind. The range of services and the adaptive technology available made us painfully aware how few of our partially sighted South Africans have access to even basic rehabilitation services.

Christina Fasser was once again elected as the President of Retina International and Claudette Medefindt was re-elected onto the management committee for another 2 year term. The next RI World congress will be held in Stresa, Italy from 25<sup>th</sup> to 27<sup>th</sup> June 2010 . The CE program featured short presentations from various countries and the excellent presentation on Ethics by Avril Daly from Ireland was the highlight of this part of the conference. A new mission statement for Retina International was agreed upon-

“..... hope to all RD patients”

*“Retina International promises to facilitate urgently the development, and ensure the world-wide availability and accessibility, of proven treatments and cures for Retinitis Pigmentosa, Macular Degeneration, Usher Syndrome, and allied retinal dystrophies”*

This highlights the advocacy role the patient organizations need to play to encourage National Governments to invest in present and future therapy for RD emanating from research successes.

### **VISIONS 2008 - BIENNIAL OPTOMETRY CONGRESS**

The South African Optometric Association kindly sponsored an awareness table for Retina South Africa at their National Congress held at Emperors Palace, Gauteng from 12<sup>th</sup> to 14<sup>th</sup> July. The congress was attended by over 500 Optometrists and support staff and afforded us an excellent opportunity to create awareness of the necessity for referrals from the optometrists. Their patron Zinzie Mandela made a brief stop at our table and was so moved when she tried on a pair of RP simulators that she took a pair of home.



Claudette Medefindt  
with Faith Chabedi,  
President of the South  
African Optometrists  
Association at the  
Visions 2008 Congress

## **Interesting Links**

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International

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