AUSTRALIAN & NEW ZEALAND
REGISTRY OF ADVANCED GLAUCOMA
Newsletter

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Department of Ophthalmology
Flinders University
Adelaide, South Australia

High Profile Visit

The Royal Australian and New Zealand College of Ophthalmologists (RANZCO) Eye Foundation's Patron, Her Excellency Ms Quentin Bryce AC CVO, visited the Australian and New Zealand Registry of Advanced Glaucoma on the 3rd July 2012 at the Flinders Medical Centre, Eye Clinic. She praised the work of the research project and emphasised how important it is to encourage regular eye health checks.

Her Excellency also visited the Australian and New Zealand Ophthalmic Surveillance Unit (ANZOSU), which is headed by Associate Professor Richard Mills, assisted by the Scientific Coordinator Lynda Saunders. The aim of this project is to establish resources that facilitate national active surveillance of rare eye conditions that are either important for public health or whose natural history or management is of clinical or scientific interest.

(Photo: Government House) L to R: A/Prof Jamie Craig, A/Prof Richard Mills, Dr Sharon Morton, Professor David Day, Her Excellency Ms Quentin Bryce AC CVO.

Her Excellency was taken on a tour of the Flinders Medical Centre Eye Clinic, including an inspection of the Day Ophthalmic Surgery Unit. She met with clinical staff, research staff and patients and witnessed use of clinical diagnostic and imaging equipment.

Ms Bryce addressed the group and discussed the importance of research into eye diseases and the implications of strong competition for funding research.

The tour ended with an afternoon tea in the Flinders Eye Clinic.

HAPPY NEW YEAR!!

2013

We hope you are all experiencing a wonderful start to the year.

2013 marks the fifth year of the Australian and New Zealand Registry of Advanced Glaucoma. Some of our participants originally gave a DNA sample when the project was in the trial stages back in 2005. We have passed many milestones since then including the wonderful success rate of recruitment across Australia and New Zealand. As mentioned in our last newsletter, there are now a growing number of samples being provided from international sources.

Clinical information and data collected through the registry has been used and reported in an increasing number of scientific papers published in ophthalmology and research journals. The findings of this research has also led to new projects being funded to further explore the findings. The Australian and New Zealand Registry of Advanced Glaucoma looks forward to continued long-term recruitment and aiding further research into the causes of blinding glaucoma as further numbers of cases enhance the power to make new discoveries.

Family Matters:

The Australian and New Zealand Registry of Advanced Glaucoma is looking to identify how glaucoma is passed through families. We believe multiple genes may cause glaucoma, and many are yet to be discovered. To assist in discovering these genes we are now looking closely at families that have a reported history of glaucoma. We believe these families can significantly assist in our search for new genes that can cause glaucoma. The more samples we collect from members of the same family who are affected by glaucoma, the more likelihood we will find a gene common to these.

One current focus is on recruiting people from the families of our existing participants who also have glaucoma. Family members interested in taking part will need to provide a blood sample, details regarding their treating specialist, and sign a consent form so we can use their sample in our research.

You may already have been contacted regarding the possible involvement of your family. So far, 253 additional family members with glaucoma, from 140 families, have agreed to assist us in our research.

We hope that by discovering new disease-causing genes, new screening and treatment techniques may be developed in the future to prevent vision loss, and even blindness, for people at risk of developing glaucoma. There may be benefits in the future for family members who have not yet been affected by this eye disease.

If you have family members affected with glaucoma interested in taking part in the research, please contact us on (08) 8404 2035 or via email at info@anzrag.com. Please note that if your family members are living overseas, we would still be pleased to hear from them as we have options to also have them included.

The Eye Bank of South Australia:

For many people the faces of their loved ones are just a blur. Imagine for a moment what that must be like. The sight of many people whose vision is affected by corneal disease can be restored by an operation known as a corneal graft or corneal transplant.

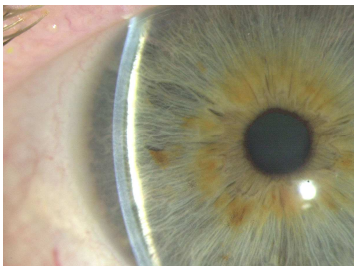
The cornea is the clear surface at the front of the eye. Should the cornea become cloudy from disease, injury, infection or any other cause, vision will be dramatically reduced. A corneal transplant is the surgical procedure which replaces the defective cornea with a similarly shaped piece of a healthy donor cornea.

The two most common reasons for requiring a transplant are bullous keratopathy and keratoconus. Other reasons include eye injury, Fuch's Dystrophy, herpes virus infection of the eye, corneal scarring due to trauma, hereditary or congenital corneal clouding, or severe bacterial infection.

Almost anyone can donate eyes regardless of age, sex, race, or previous physical condition. Wearing glasses or having poor vision does not prevent eye donation. The eyes must be removed within 12 hours of death.

Patients with certain diseases, or who are at risk of these diseases, are not suitable eye donors. People who wear glasses, or who suffer from cataracts or glaucoma **can** donate their eyes. Other than ocular cancer, most other types of cancer do not prevent eye donation. The research team is always grateful to receive eyes that are likely to be considered unsuitable for transplant, to be used in research into prolonging graft survival and all kinds of eye disease.

Please contact the Eye Bank for more information. Eye Bank of South Australia: Ophthalmology Department, Flinders Medical Centre, Bedford Park, SA, 5042 (Telephone: 08 8204 4928 Email: fmceyebanksa@health.sa.gov.au).



The Big Picture:

One aim of the Australian and New Zealand Registry of Advanced Glaucoma is to accumulate the world's largest repository of genetic and clinical information from advanced glaucoma cases. The registry now has over 1,500 advanced glaucoma cases comprising of DNA samples and clinical information collected by treating specialists.

Although the number of cases is important and reflects the commitment of ophthalmologists to supporting the registry, it is important to not lose sight of the overall aim of the research the registry is performing.

Glaucoma can sneak up on patients and steal their sight without them even knowing. We have heard from many of you how on the first diagnosis of glaucoma a significant amount of vision had already been lost. One main goal of the registry is to discover new genes to assist in the development of new screening techniques that will identify those most at risk of developing glaucoma, and therefore allow early intervention to prevent any vision loss. DNA testing may give ophthalmologists the head start they need to better prescribe therapy and prevent patients from losing their vision. We thank all our participants for assisting us to work towards these goals.

Predicting the Future:

As part of our registry, we screen the DNA of individuals with severe glaucoma for a gene known to cause glaucoma, the *Myocilin* gene. When an individual is identified as having a change in that particular gene, we offer to test their relatives, as other family members can have the same gene change, and as such be at a very high risk of developing glaucoma at a younger age than in most glaucoma patients. This familial predictive testing has been successful at identifying at-risk individuals who can now hope to avoid glaucoma blindness through regular glaucoma check-ups and appropriate interventions.

Recently, we aimed to understand individuals' motivations as well as the personal and familial impacts of predictive genetic testing for glaucoma. We sent a questionnaire to family members of individuals with a *Myocilin* gene change who chose to be tested for the familial gene change to obtain their feedback.

The main benefit brought up by 80% of the participants was the availability of monitoring for early detection and prevention of glaucoma, and the most often cited motivations for being tested were to take appropriate interventions (73%) and to remove uncertainty (62%).

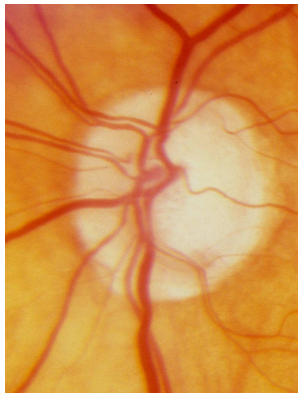
The third commonest cited motivation for being tested was to provide information to children (59%). All participants who had children above 18 years old discussed their test results with them. Additionally, the majority of the participants have discussed their genetic result with their family (95%). It seems that familial testing globally brings better awareness among relatives, including children of adult age for being tested.

The main concerns expressed by carriers were related to the fear of developing glaucoma, the efficacy and cost of treatments, the possibility of transmission to children, and the travelling distance to the clinic when living in rural areas.

Such findings are very valuable in helping health professionals provide better support and addressing the relevant personal and familial issues with patients undergoing predictive genetic testing for glaucoma.

Terminology:

- **RNA:** Some participants may have been asked to provide another blood sample taken in a blue tube for RNA analysis. Ribonucleic acid (RNA) differs from DNA in that DNA's function is solely to store genetic information whereas RNA presents in 3 different types—each with a specific function. rRNA exists not in the nucleus but the cytoplasm of the cell in ribosomes, where protein synthesis takes place. mRNA records information from DNA and takes it to the ribosomes where the rRNA exists. tRNA deliver amino acids one by one to the protein chains being manufactured at the ribosomes.
- **Angle Closure Glaucoma:** If the drainage angle between the cornea and iris is very narrow the iris itself can block the drainage of fluid from the front part of the eye. In some cases the pressure in the eye may raise very quickly and this can cause intense pain for the patient. Immediate medical intervention is required to prevent permanent loss of vision.



- **Selective Laser Trabeculoplasty (SLT):** Is a low level laser treatment used to open up the drainage angles in the eye. It is usually used after medication has been trialled and before surgery. SLT is used to treat open angle glaucoma and may be performed in an outpatient clinic. SLT leaves less scarring than the older laser treatments (ALT) and therefore can be repeated if necessary.

Would you like to know more?

If you have any questions regarding this research and how you can contribute, please feel free to contact the office at Flinders Medical Centre/Flinders University in Adelaide as follows:

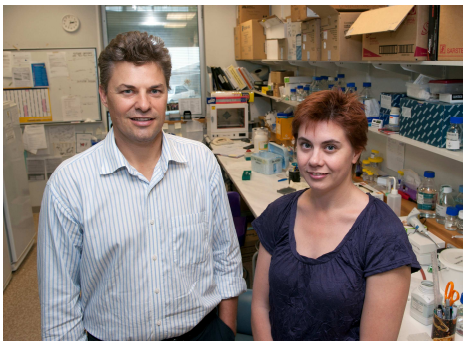
Phone: +61 08 8404 2035 Fax: +61 08 8204 6722 Email: info@anzrag.com

*The **Australian and New Zealand Registry of Advanced Glaucoma** wishes to acknowledge and thank the organisations that support our work and research. Without their assistance this project would not be possible:*

◊Eye Foundation
◊Flinders University
◊Royal Society for the Blind
◊IMVS
◊Flinders Medical Centre
◊Glaucoma Australia
◊The Ophthalmic Research Institute of Australia (ORIA)
◊ National Health and Medical Research Council
◊Royal Australian and New Zealand College of Ophthalmologists (RANZCO)

Members of the Research Team:

Chief Investigator	Prof Jamie Craig
Post Doc Research Fellow	Dr Kathryn Burdon
Genetic Counsellor	Emmanuelle Souzeau
Coordinator	Bronwyn Ridge
Research Assistants	Dr Jude Fitzgerald
Technical Assistant	Win Kee Beh



Assoc/Prof Jamie Craig and Dr Kathryn Burdon
Photo by: Ashton Claridge, Flinders University