AUSTRALIAN & NEW ZEALAND
REGISTRY OF ADVANCED GLAUCOMA

Issue 1 / October 2010

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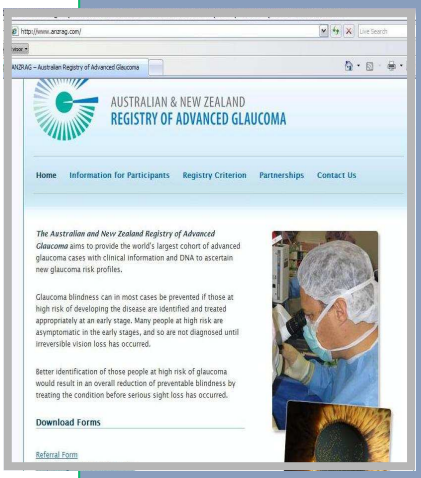
Welcome to the first newsletter for the Australian & New Zealand Registry of Advanced Glaucoma (ANZRAG). The project commenced in April 2007 and was initiated as a 5-year project, funded by the Eye Foundation. The aim of the Registry is to recruit the world's largest collection of Advanced Glaucoma cases, and other glaucoma subtypes, including clinical details and DNA samples. "Advanced Glaucoma" refers to cases where a substantial amount of vision has already been lost. The Registry offers genetic screening, for participants identified with Advanced Glaucoma, of a known glaucoma-causing gene (Myocilin) and will also use DNA samples to search for new genes linked to causing glaucoma. Every DNA sample brings us closer to our goals. If you would like to assist further, please complete the enclosed questionnaire and return it in the reply-paid envelope provided

Your interest and participation in this project is invaluable - Thank you.



OFFICIAL WEBSITE

The official Australian & New Zealand Registry of Advanced Glaucoma website can be viewed at www.anzrag.com.

There is information about the project for participants and eye specialists, links to our project partners and downloadable referral forms.

We will also be publishing this newsletter and other updates to the website to keep everyone informed of the research status.

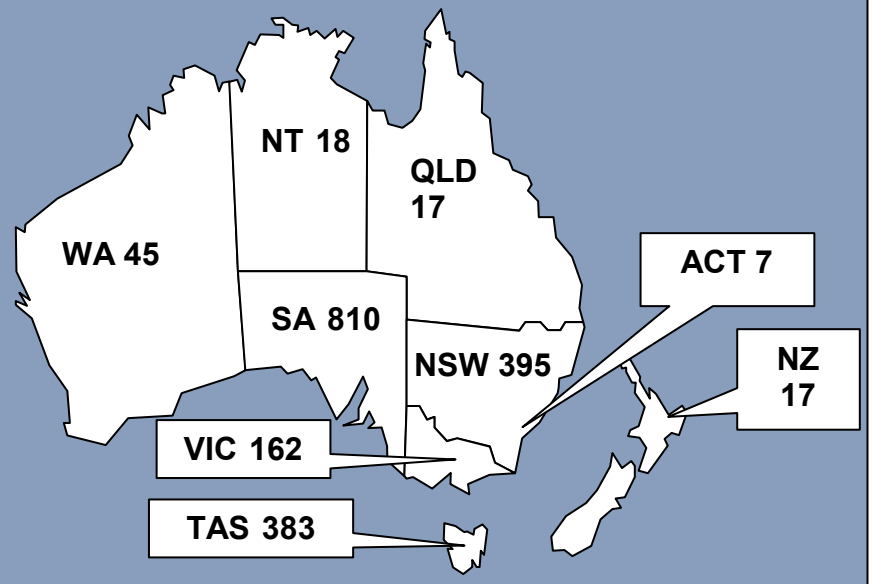
Please feel free to have a look around the website and pass the link to anyone you think may benefit or be interested participating.

FINDINGS PUBLISHED:

Thanks to the fantastic response we have had from referring specialists and willing participants, the Registry is very pleased to announce that world-first results have recently been published in the prestigious journal *Nature Genetics.*

The study entitled; *Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma*, was performed in collaboration with the deCODE genetics in Iceland and was published online on September 12th. It will be available in print in the coming months. This work involved over 3,500 glaucoma patients and goes some way to understanding which common gene variants contribute to the risk of getting glaucoma. We are also generating exciting results about which specific genes contribute to the risk of Advanced Glaucoma, so watch this space as we will report soon.

Glaucoma blood samples and referrals have been received from every state and territory of Australia and from New Zealand.



SO FAR……..

- 1088 Advanced Glaucoma samples collected
- Of these, 50 Advanced Glaucoma participants have a mutation in the *Myocilin* gene
- Many family members of those showing *Myocilin* gene mutations have now also been screened.
- 418 DNA samples from participants with Pseudoexfoliation being utilised in searching for new genes linked to this condition
- Presentation of information to the Royal Australian and New Zealand College of Ophthalmology conference in Brisbane 2009 and the International Society for Eye Research conference in Montreal and the World Ophthalmology Conference in Berlin during 2010

The prevalence of visual impairment from Primary Open Angle Glaucoma (POAG) in Australia is expected to grow from 24,000 people in 2005 to 52,000 people in 2025.

Diagnosis rates for POAG could be improved by: 1) at the current diagnosis threshold, enhancing the quality of care by ensuring eye practitioners include the Visual Field tests when appropriate when examining patients' eyes; 2) careful examination of the optic disc in all patients regardless of (pressure); and **3) improving the focus on family history, since people with a family history of glaucoma have a much higher chance of developing POAG.**

Excerpt from "Tunnel Vision—The Economic Impact of Primary Open Angle Glaucoma" (Centre for Eye Research Australia, University of Melbourne, Australia, February 2008).

ANZRAG; Saving Sight

Mr Fellows (*name changed*) was informed, through the work of ANZRAG, that he carried a gene mutation in the *Myocilin* (MYOC) gene. It did not have an affect on his condition or treatment, but it did have an impact on the members of his family.

Mr Fellows has a sister who was also tested and was found not to carry this mutation. She was very relieved to learn that her risk of developing glaucoma, although not nil, was lowered and is probably more around the same as the general population.

However, Mr Fellows' son found out that he does carry the same genetic mutation as his father.

Although he had not yet shown major elevation of his eye pressures, this new information, coupled with other slight changes in his eyes, was enough reason for the Chief Investigator to recommend he commence glaucoma treatment.

This action has most likely prevented Mr Fellows' son experiencing the same devastating vision loss, due to glaucomatous changes, as his father.

Frequently Asked Questions:

What will I be asked to do as a participant?

You will be asked to provide a blood or saliva sample, for DNA extraction, and to sign consent forms to allow us to perform genetic screening on your sample and to obtain clinical information related to your eye disease from your treating specialist.

Will it cost me anything?

It does not cost to be involved in the Registry. You will be provided with a kit through Australia Post to collect the blood sample. We cover the expenses for return packaging and postage of the kit, and for any invoices related to having the blood taken.

What is genetic testing?

Genetic testing is the analysis of DNA in order to detect changes related to a genetic disorder. Genetic testing is different from other kinds of laboratory tests. These tests are done in specialised, accredited laboratories and they can provide beneficial information not only for the patient but also for other family members.

Did You Know???

More than 50% of people in developed countries who have glaucoma have not been diagnosed.

Glaucoma is the leading cause of preventable blindness in the world.

What if a gene mutation is found in my sample?

You will receive a phone call from our Genetic Counsellor. The findings will be explained to you in detail and you will be able to ask any questions. If a gene mutation is found, there may be implications for you and for other members of your family. The Genetic Counsellor will discuss the option of providing a screening service to other relatives. Your result will be completely confidential and we will not initiate contact with any family members.

What is genetic counselling?

Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of how genetics contribute to a condition.

What should I expect from a genetic counselling consultation?

You should expect the Genetic Counsellor to document your medical and family history, to provide you explanations regarding the course of the condition as well as its inheritance, to evaluate the risk of occurrence in other family members, to discuss genetic testing options and/or results as well as their implications for you and your family and to promote informed choices and adaptation to the risk or condition.

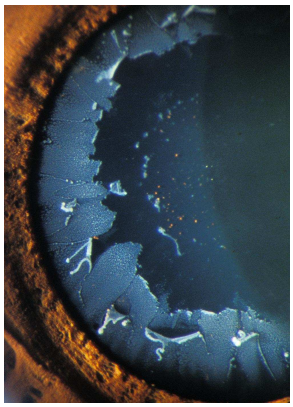
Can I refer other members of my family who have glaucoma?

If other members of your family have advanced glaucoma with severe visual field loss, or one of the secondary types of glaucoma we are studying (pseudoexfoliation, angle closure, pigment dispersion, steroid response, primary congenital glaucoma or anterior segment dysgenesis) you can refer them **if you have their permission** to do so. We can take referrals over the telephone, by fax or by email. We will need to verify their diagnosis with their treating specialist.

Terminology:

- **Congenital glaucoma:** A form of glaucoma that occurs in babies or young children.
- **DNA:** The molecule that makes up genes (deoxyribonucleic acid)
- **Gene:** The specific sequence of DNA that contains the instructions for a specific characteristic or function.
- **Glaucoma:** A group of eye diseases in which the optic nerve at the back of the eye is damaged. Glaucoma is a leading cause of vision damage and blindness in people over 40 years of age but it can affect people of any age.
- **Mutation:** A change in a gene that can be responsible for a genetic condition.
- **Pigment dispersion syndrome/glaucoma:** A condition where pigment cells are shed from the back of the iris and float in the liquid within the eye. If the cells get caught in the drainage area of the eye, pressure may increase which may lead to glaucoma.
- **Pseudoexfoliation syndrome/glaucoma:**

A condition where flaky deposits appear on the surface of the lens (appearance like dandruff - see picture). If the particles affect drainage within the eye, increased pressure may lead to glaucoma.



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
◊SA Pathology
◊Flinders Medical Centre
◊Glaucoma Australia
◊The Ophthalmic Research Institute of Australia
◊ NHMRC Centre for Clinical Eye Research
◊Royal Australian and New Zealand College of Ophthalmologists

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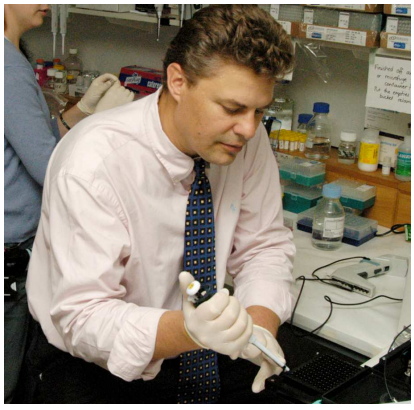

Associate Professor Jamie Craig at the Australian and New Zealand Registry of Advanced Glaucoma laboratory