

The Genetics of Glaucoma Study

Information for Participants

1. What is the purpose of the study?

A person's risk of glaucoma has been shown to have a hereditary component. Large-scale studies investigating genetic risk factors may increase our understanding of the disease process, identify people at high risk of developing glaucoma, and identify novel treatments. These studies need the participation of large numbers of people who have been diagnosed with the disease.

Therefore we are trying new ways to make contact with as many people as possible who have been diagnosed with glaucoma or have had treatment for glaucoma. One way of doing this is to approach people who have been prescribed medications that treat glaucoma. The other way is to inform people of our study aims through social media, and invite those who have been diagnosed with glaucoma to participate in our study. We have taken these approaches for other medical conditions and had a good response.

You have been invited to participate in this study either through the Department of Human Services or through our media campaign. If you have been contacted by the Department of Human Services, you have recently been prescribed one of (but not limited to) these glaucoma medications: **prostaglandin analogs** (e.g. Xalatan, Lumigan, and Travatan), **beta blockers** (e.g. Timoptol, Timoptol XE, and Nyogel), **alpha agonists** (e.g. Alphagan P and Iopidine), **carbonic anhydrase inhibitors** (e.g. Trusopt, Azopt, and Diamox), or **combined medications** (e.g. Cosopt/Cosopt PF, Combigan, Simbrinza, Xalacom, and Ganfort).

We hope that you will participate in the study. Your participation in this study is voluntary; there will be no cost to you whether you do or do not take part. Choosing not to take part in this study will not affect your future medical care in any way. Our aim is to find new genes for glaucoma and develop risk prediction tools that use genetic data to identify people at high risk of developing this disease.

2. Who can participate?

For this study we need to recruit both men and women who have been diagnosed with glaucoma by an ophthalmologist or optometrist, have been prescribed a glaucoma medication, or have family history of glaucoma.

3. Do I have to participate?

Your participation in this study is completely voluntary and there will be no cost to you. If you do not want to take part in this study you do not have to. You should feel under no obligation to participate in this study. Choosing not to take part in this study will not affect your current and future medical care in any way.

4. Can I withdraw from the study?

You are under no obligation to continue with the research study. You may change your mind at any time about participating in the research. People withdraw from studies for various reasons and you do not need to provide a reason.

You can withdraw from the study at any time by completing and signing the **‘Participant Withdrawal of Consent Form’**. You can contact the Project Coordinator Dr Puya Gharakhani by phone: 07-38453981 or email Glaucoma_Genetics@qimrberghofer.edu.au to request the ‘Participant Withdrawal of Consent Form’ which will be mailed to you to complete and sign.

If you withdraw from the study, you will be able to choose whether the study will destroy or retain the information it has collected about you. You should only choose **one** of these options. Where both boxes are ticked in error or neither box is ticked, the study will destroy all information it has collected about you. However, your data may not be able to be deleted if we have analysed and/or published the research results.

5. What does participation involve?

Participation involves providing your contact details and answering a questionnaire. To participate you are also required to donate a saliva sample. We will extract your DNA from your saliva sample to investigate genetic risk factors for glaucoma by comparing DNA from people with glaucoma against a control group who do not have this condition.

Participation also involves consenting to storage of your questionnaire and genetic information in a data repository for future use. This information may be stored indefinitely (if you have consent for us to access your Medicare Benefits Schedule and Pharmaceutical Benefits Scheme data, those data will not be stored indefinitely; see below and section 11) and pooled together with similar data from other participants. To see how your privacy is protected, please read Section 10 - ‘Is it confidential?’

Before you participate, though, we need your consent. When consent is given, we will begin the process of collecting your DNA. This will involve you providing a sample of saliva into a specialised collection container which we will send to you. You will be asked to return this sample to our laboratory via our delivery services, at no cost to you. Some details of your medical history that would be helpful to the project investigators (like how many prescriptions you may have had for various medications) would be hard for many people to remember. So we will ask for your permission to access your complete Medicare Benefits Schedule (MBS) and Pharmaceutical Benefits Scheme (PBS) claims information from the last five (5) years. Medicare collects information on your medical visits and procedures, and the associated costs, while the PBS collects information on the prescription medications you have filled at pharmacies. If you agree, you will be asked to fill out a consent form authorising the study access to your Medicare and PBS data as outlined on the consent form. The consent form will be sent securely to the Department of Human Services which holds your Medicare and PBS information confidentially. Consent to access your Medicare and/or PBS claims information is completely separate from consent for the rest of the study (questionnaire and biological sample). You can participate in the other parts of this study without consenting to the Medicare and PBS component.

6. Do I have to give a DNA sample?

To participate in this study you are required to provide a saliva sample, as explained in the paragraph above. Providing a sample can be done in your own home.

7. What is in it for me?

This study is unlikely to be of any immediate and specific benefit to you. Extensive research is required to find answers to the questions we are studying. However, future medical or scientific discoveries may come from the research in which you participate, and in turn, help develop risk prediction strategies and improve available treatments and outcomes for people suffering from glaucoma.

Due to the specific sampling design of the study, we will not be able to provide any individualised analytical feedback to participants about their health condition, biological sample, or DNA. However, researchers will be providing everyone who participates with a newsletter. In this newsletter we will give you information about the progress and outcomes of this study.

Our research team greatly value the time and effort that you give to research.

8. Are there any risks?

Researchers acknowledge that being invited into this research study may be a sensitive issue for you and may, therefore, cause you some discomfort. We would like to restate that we currently do not have any information about you.

You may feel that some of the questions we ask in the questionnaire are stressful or upsetting. If you do not wish to answer a question, you can skip it and go to the next question, or you may stop immediately.

If you have any questions or concerns about this research study, you may telephone the Project Investigator, Dr Puya Gharahkhani, Senior Research Officer, on 07-3845 3981. If you have any concerns or complaints regarding the conduct of this study, you may contact the Chairperson of the QIMR Berghofer Medical Research Institute Human Research Ethics Committee (QIMRB HREC) via the Secretary on Tel: 07-3362 0117 and quote reference number P3423.

9. Will I be contacted again about this study?

We plan to extend this study and may seek to re-contact some of the participants in the current study. Choosing to participate in the current study does not mean that you will necessarily be re-contacted. If we do contact you about a follow-up study, you can of course choose not to participate and it will not impact your participation in the current study in any way.

10. Is it confidential?

Yes. All information and data collected for the study remains confidential in accordance with The Australian National Health and Medical Research Council (NHMRC) Human Research guidelines and the Privacy Act 1988. Your personal details, questionnaire data, biological sample, and genetic data will all be stored separately. We have not been supplied your name, address, medical or other details. Your individual questionnaire, biological sample and genetic data files will have a number assigned to it, not your name. Your name and personal details will continue to be stored on file at QIMR Berghofer but will be stored separately from, and not linked with, your questionnaire information, biological sample and genetic data (de-identified data i.e. staff working on your data will not be able to identify you). The only link between your data and your personal details is your participant identification number (meaning your sample is potentially re-identifiable). Linking both your personal details and data file using this number

(re-identification) is not required for this study. However, if it becomes necessary for some reasons (for example to go back and check some details), it is strictly restricted to members of the QIMR Berghofer research team involved in this study.

Results of this research study may be presented in scientific papers in medical literature, or in public talks, but your identity will not be revealed. The data collected as part of this study will be combined at analysis with the data from many other people, and as such there will be no way of identifying you as a participant.

In accordance with the Privacy Act 1988 and other relevant legislation, you have the right to request access to the information about you that is collected and stored by the research team. You also have the right to request that any information with which you disagree be corrected. Please inform the research team member named at the end of this document if you would like to access your information.

By confirming your consent, you consent to the research team collecting and using personal, questionnaire and genetic information about you as described for the research study.

11. What will happen to information about me?

The researchers will store your personal, questionnaire and genetic information indefinitely (except for the Medicare and PBS data; see below) at QIMR Berghofer Medical Research Institute. The reason why we need to store this information indefinitely is because it will continue to be valuable to researchers many years into the future, and may be considered for use in future related projects. Before any future work proceeds, it will be subject to approval by the relevant ethics committees.

In addition to conducting academic research, we may share your de-identified data with others for the purpose of developing a genetic test for glaucoma risk (in healthy individuals) and glaucoma prognosis (in individuals affected by glaucoma).

Any Medicare and PBS data you consent to provide (including the consent form itself) will be used for the purposes of this study only, and will not be used for any other purposes. It cannot be shared with anyone outside the research team for this project without specific Commonwealth Government approval. The Medicare and PBS data will be held in secure, password protected servers at QIMR Berghofer Medical Research Institute to which access is restricted to personnel directly involved in the project. The original records supplied to the research team, and any copies, will be deleted from our computer systems 7 years after the publication of the final project report. However, any research findings associated with your Medicare or PBS data will not be able to be destroyed or recalled.

Your genetic information and some of your questionnaire information (but not your name, other personal details, Medicare or PBS data) may eventually be put into an international genetics data repository. Information in the database will be available only to researchers from around the world who are approved to study how genes cause a variety of health conditions. These scientists will not know your name or other personal information we learn about you.

12. What will happen to my biological and DNA samples?

This Study: We will use your biological (saliva) sample to extract one or more samples of DNA. The research team will then look for differences and similarities between participants' DNA samples. This information can help us understand why some people have a certain condition such as glaucoma and some people do not.

Your biological sample and samples of your DNA will be stored securely at QIMR Berghofer Medical Research Institute along with samples from many other people. They will be re-identifiable, which means that they will be stored with a barcode label, and can be identified as yours even though your personal details are stored separately. Linking your personal details with your biological sample or DNA using the barcode is restricted only to members of the QIMR Berghofer research team.

We may decide to send part of your biological sample and/or a sample of your DNA to another laboratory (which may be overseas) for processing or analysis. If this occurs, your part sample will only be labelled with a number, and transported along with samples from many other people. No information identifying you will be sent to or accessible by the other laboratory. Any sample remaining after processing or analysis by the other laboratory will be returned to QIMR Berghofer Medical Research Institute for indefinite storage. No samples will be retained overseas.

Future Studies: We would like to store your biological and/or DNA samples (except for your Medicare and PBS data) for use in future research studies that may or may not be related to this study. There is no direct benefit to you from the storage of your biological and/or DNA samples. In the future, other doctors and scientists at this and other medical and research centres may use your samples to learn about many different diseases and conditions. Their goal is to improve health outcomes and develop new treatments. The purpose of storing these types of samples is to answer questions in the future, so we expect to keep your samples indefinitely.

13. Who are the researchers?

This study is being conducted by the following researchers:

- Dr Puya Gharahkhani, QIMR Berghofer Medical Research Institute
- Associate Professor Stuart MacGregor, QIMR Berghofer Medical Research Institute
- Prof Alex W Hewitt, QIMR Berghofer Medical Research Institute

14. What if I have questions?

You can call or email the Project Coordinator by phone: 07-38453981 or email Glaucoma_Genetics@qimrberghofer.edu.au. We are happy to answer any questions you have before you agree to participate and also at any time throughout the study.