QIMR Berghofer Medical Research Institute

THE UNIVERSITY OF MELBOURNE

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Genetics of Epilepsy

Information for Participants

Please read the following Information Sheet.

1. Introduction

You are invited to participate in this research project, which is called the 'Genetics of Epilepsy' (GenEp). We are seeking individuals 18 or over who have taken Anti-Epileptic medications to help us understand why epilepsy medications work for some people but not others, and why some medications cause side effects—such as fatigue, dizziness, or "brain fog".

This Participant Information Sheet and Consent Form tells you about the research project. It explains what is involved in the study to help you decide if you want to take part.

Please read this information carefully. Ask questions about anything that you don't understand or want to know more about. Before deciding whether or not to take part, you might want to talk about it with a relative, friend or your doctor.

If you decide you want to take part in the research project, you will be asked to provide your consent online. You will be able to save an electronic copy of this Participant Information Sheet and Consent Form to keep.

If you do not wish to take part in this study, you do not have to. The project investigators do not have your name or contact details unless you provide them, so if you do not wish to take part you do not have to do anything further.

2. What is the purpose of the study?

Our group at QIMR Berghofer Medical Research Institute has been researching complex health conditions and medications for over thirty years. In collaboration with other researchers, our Twin and Family Studies have shown a person's risk of epilepsy has a hereditary component – it is more common in those who have an affected family member.

Genes which contribute to the risk of developing this condition can be identified through Genome-Wide Association Studies (GWAS). GWAS is a DNA analysis method which compares the genetic profile of groups with a particular condition with a control group who have never had this condition, allowing researchers to reveal genes unique to each group.

GWAS need the participation of large numbers of people who have been diagnosed with epilepsy to increase our understanding of the disease process and identify novel treatments. So far, studies on epilepsy have lagged behind those on other common conditions (such as heart disease and cancers). Therefore we are trying new ways to make contact with as many people as possible who have epilepsy,

and one way of doing this is to approach people who have been prescribed medications that treat this condition. We have taken this approach for other medical conditions and had a positive response from the community.

Anti-Epileptic Drugs (AEDs) are medications used to manage symptoms of epilepsy. You have been contacted by the Services Australia because you may have recently been prescribed one of these medications.

We hope that you will participate in the study. This research will collect a world-first large and genetically informative cohort of 10,000 pharmacoresistant epilepsy patients with the ultimate aim of finding new treatments.

3. Who can participate?

For this study we need to recruit both men and women, aged 18 years and over, who have been prescribed anti-epileptic medications.

4. What does participation involve?

Participation involves providing your contact details and answering a short online questionnaire about your experiences with epilepsy. Then, if eligible, you will be asked to donate a saliva sample.

We will extract your DNA from your saliva sample to investigate genetic risk factors for epilepsy by comparing DNA from individuals with epilepsy against a control group who do not have this disorder.

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.

Medicare collects information on medical visits and procedures, and the associated costs, while the PBS collects information on the prescriptions that have been filled at pharmacies.

So we will ask for your permission to access your Medicare Benefits Schedule (MBS) and Pharmaceutical Benefits Scheme (PBS) claims information.

Consent to authorise the study to access your MBS/PBS information will be collected online, following on this study consent form. If you complete this consent form, it will be sent securely to Services Australia, which holds 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 (online questionnaire and biological sample). You can participate in the other parts of this study without consenting to the Medicare and PBS component.

Services Australia is not involved in this research other than to provide the information that you have consented to the release of, should you decide to participate in this study. Services Australia has

confirmed that this research and any associated documents have received approval from a Human Research Ethics Committee (HREC) that is registered with and operates within guidelines set out by the National Health and Medical Research Council (NHMRC).

We appreciate the time and effort you will give in this study. In return, we will provide you with an e-gift card to the value of $30 at the end of your participation in the study. You will still be reimbursed for your time in participating if you do not provide consent to access your Medicare and PBS information.

Participation also involves consenting to potential storage of your questionnaire and genetic information in a data repository for future use. This information may be stored indefinitely and pooled together with similar data from other participants. To see how your privacy is protected, please read Section 10 - 'Is it confidential?'

5. Do I have to give a DNA sample?

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

6. Do I have to take part in this research project?

Participation in this research project is voluntary. If you do not wish to take part, you do not have to. The project investigators do not have your name or contact details unless you provide them, so if you do not wish to take part you do not have to do anything. If you decide to take part and later change your mind, you are free to withdraw from the project at any stage.

You are under no obligation to continue with the research study. 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 contacting the research team and requesting a withdrawal of consent form for 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. If you withdraw from the study, your information that has already been analysed and/or included in a publication may not be able to be withdrawn or destroyed. In such circumstances, your personal information will continue to form part of the project/research study records and results. Your privacy will continue to be protected at all times.

7. What are the possible benefits of taking part?

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 improve the available treatments and outcomes for people with epilepsy and their future families. Many participants value the unique contribution that they can make to research.

Due to the specific sample design of the study, we will not be able to provide any individualised diagnostic 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, as well as that of several other studies.

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

8. Are there any risks and disadvantages in taking part?

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 Co-ordinator Richard Parker on 07-3362-0297. You may also use our free call number: 1800-257-179. 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 P3571.

Data Breach Notification

If a data breach occurs that is likely to cause harm, we will notify you and the Queensland Office of the Information Commissioner (OIC) as required by law.

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 Australian Privacy Act.

Your personal details, questionnaire data, biological sample and genetic data will all be stored separately. 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. 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 is severely restricted to members of the research team.

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 relevant Australian privacy and other laws, 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 online 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?

All personal, questionnaire and genetic information collected for the study remains confidential in accordance with the National Health and Medical Research Council (NHMRC) ethical guidelines and the Privacy Act. Personal details, questionnaire data and genetic data will all be stored separately. The only link between personal details and other data is a participant identification number. Linking personal details and other data using this number is restricted to selected members of the QIMR Berghofer research team. All information provided by you will be stored securely, with access restricted to members of the research team.

Any Medicare and PBS data you consent to provide (including the consent form itself) will be used for the purposes of this study only. It cannot be shared with anyone outside the research team for this project without specific Commonwealth Government approval. The original records supplied to the

research team, and any copies, will be deleted from our computer systems 5 years from the publication of the final project report, or after 10 years from the date of supply (whichever is sooner). However, any research findings associated with your Medicare or PBS data will not be able to be destroyed or recalled.

The researchers will store your personal, questionnaire and genetic information indefinitely 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.

It is possible that your genetic information and some of your questionnaire information (but NOT your name, or other personal details) may eventually be put into an international genetics data repository. Access to the information in this database is strictly controlled and will be available only to researchers from around the world who are approved to study how genes cause a variety of health conditions, including epilepsy. 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 epilepsy 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 to only members of the 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.

Future Studies: We would like to store your biological and/or DNA samples for a long time 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 for a long time.

Optional Commercial Research Participation: In addition to academic research, your DNA sample and associated data may also help drive innovation in the private sector. With your separate consent, we may share your de-identified sample and data with commercial researchers working to develop new treatments and technologies for epilepsy and other diseases. Commercial research partnerships can accelerate the development of medications and therapies that may not otherwise be possible through academic channels alone. By choosing to opt in, you contribute to a broader effort to translate genetic discoveries into real-world health solutions. Your identity will never be shared, and all projects must be approved by independent ethics committees. Participation in this optional component is entirely voluntary and will not affect your involvement in the main study.

13. Who are the researchers?

This study is being conducted by the following researchers:

- Professor Dale Nyholt, Queensland University of Technology (and QIMRB Affiliate)
- Professor Nick Martin, QIMR Berghofer Medical Research Institute
- Associate Professor Wendyl J D'Souza, University of Melbourne

14. What if I don't want to participate or what if I change my mind later and want to withdraw from the study?

Participation is voluntary and you can choose not to participate. If you do choose to participate you can withdraw from the study at any time, at any stage, or for any reason for some, part, or all of the research. You can withdraw your consent by contacting the Project Co-Coordinator by phone 1800 257 179 (freecall) or email epilepsy@qimrb.edu.au. These contact details will be listed on your correspondence with the project team. If you do withdraw, you will be asked to complete and sign a 'Withdrawal of Consent' form; the research team will provide this to you.

You can also choose to remove yourself from the Services Australia mailing list by emailing data.requests@humanservices.gov.au. You will need to provide your full name, address, Medicare care number and a contact phone number.

15. What if I have questions?

You can call or email us. Our Free call number is 1800 257 179. Our email address is epilepsy@qimrb.edu.au. We are happy to answer any questions you have before you agree to participate and at any time throughout the study.

If you want any further information concerning this project, you can contact the project coordinator:

Name	Richard Parker
Position	Project Coordinator
Telephone	07 3362 0297
Email	richard.parker@qimrb.edu.au

If you have any complaints about any aspect of the project, the way it is being conducted or any questions about being a research participant in general, then you may contact:

Reviewing HREC name	QIMR Berghofer Medical Research Institute Ethics Committee
HREC Executive Officer	Secretary to the Chairperson of the Ethics Committee
Telephone	07 3362 0117
Email	HREC.Secretariat@qimrb.edu.au

If you have a privacy complaint in relation to the use of your MBS/PBS data, you should contact the Office of the Australian Information Commissioner. You will be able to lodge a complaint with them.

Website: www.oaic.gov.au

Telephone: 1300 363 992

Email: enquiries@oaic.gov.au

Mail: GPO Box 5218, Sydney NSW 2001

If you do not want to participate, thank you for your time. You are not required to respond in any way. You may close the browser window to exit.

I have read this information sheet and have understood it ☐

Consent

If you would like to participate in this study, we need you to tell us below that you have understood what is involved in participating and that you are giving us permission to collect and store the information, biological (saliva) sample and DNA that you provide us.

Clicking on the "agree to participate" button below indicates that:

- I voluntarily give my consent to participate in the research study 'The Genetics of Epilepsy and Medication Response' as described in the Information Sheet, including, if required, the provision of a saliva sample to be used for DNA testing to learn more about how genes and environment affect health and behaviour.
- I acknowledge that the nature, purpose and contemplated effects of this research study, especially as far as they affect me, have been fully described to my satisfaction by the Information Sheet.
- I acknowledge that my saliva sample and DNA, which I choose to provide (but NOT my name or address) may be made available to other medical researchers studying health and behaviour in the future, subject to review by the appropriate research ethics committees.
- I acknowledge that my saliva sample and DNA will be stored indefinitely and may be considered for any use in the future related projects, including uses that are unrelated to this study, subject to review by the appropriate research ethics committees.
- I understand that my involvement in this research study may not be of any direct benefit to me and that I may withdraw my consent at any stage without affecting my rights or the responsibilities of the researchers in any respect.
- I agree to be contacted about future, related studies and understand that I am in no way obligated to participate and can freely withdraw from this request without affecting my rights or the responsibilities of the researchers in any respect.
- I understand all information gathered during this research project will be treated in a strictly confidential manner in accordance with the National Health and Medical Research Council (NHMRC) Guidelines and the Commonwealth Privacy Act.

If you do not want to participate, thank you for your time. You are not required to respond in any way. You may close the browser window to exit.

☐ *Yes - I choose to participate [Date and time automatically recorded]*

☐ *No - I choose not to participate*

Optional Commercial Research Participation

In addition to academic research, your DNA sample and associated data may also help drive innovation in the private sector. With your separate consent, we may share your de-identified sample and data with commercial researchers working to develop new treatments and technologies for epilepsy and other diseases. Commercial research partnerships can accelerate the development of medications and therapies that may not otherwise be possible through academic channels alone. By choosing to opt in, you contribute to a broader effort to translate genetic discoveries into real-world health solutions. Your identity will never be shared, and all projects must be approved by independent ethics committees. Participation in this optional component is entirely voluntary and will not affect your involvement in the main study.

☐ *I agree that my saliva sample and DNA, along with associated data (but NOT my name, address, or any other identifying information), may be shared with approved commercial researchers working to develop new treatments for epilepsy and other diseases. I understand that this research may lead to commercial products, and I will not receive financial compensation or share in any profits. All projects will be reviewed by ethics committees, and my privacy will be fully protected.*

This project has been reviewed and approved by the QIMR Berghofer Medical Research Institute Human Research Ethics Committee (QIMRB-HREC EC00278). If you have any concerns or complaints regarding the conduct of this study, you may contact the Chairperson of the Ethics Committee (QIMRB-HREC) via the Secretary on Tel: 07-3362 0117 and quote reference number P3571.