Galway University Hospitals
Ospidéil na h-Ollscoile Gaillimh
UNIVERSITY HOSPITAL GALWAY
MERLIN PARK UNIVERSITY HOSPITAL

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Patient Information Leaflet

Project Title:
Rare Kidney Disease Registry and Bioresource

Principal Investigator's Name:
Professor Matthew Griffin, Consultant Nephrologist, Merlin Park Hospital, Merlin Park, Galway.

Telephone No. of Principal Investigator: 091-524222 or 091-751131

You are being invited to take part in a research study because you have been diagnosed with a rare kidney disease.

This information sheet will help you understand why we are undertaking the research and what we would like you to do. Please read the sheet carefully and feel free to discuss it with others if you wish. Please ask us if there is anything that is not clear to you or if you would like more information. Please take time to decide whether you wish to take part or not. Thank you for reading this.

Before you decide whether or not you wish to take part, you should read the information provided below carefully and, if you wish, you should discuss it with your family, friends or GP. Take the time to ask questions – do not feel rushed or under any obligation to make a hasty judgement. You should clearly understand the risks and benefits of participating in this study so that you can make a decision that is right for you – this process is known as Informed Consent.

You are **not** obliged to take part in this study and failure to participate will have no effect on your future care.

You may change your mind at any time (before the start of the study or even after you have commenced the study) for whatever reason without having to justify your decision and without any negative impact on the care you will receive from the medical staff.

Why is this study being done?

This study will establish the first patient registry and bioresource for rare kidney disease in Ireland. It will be used to help increase our understanding of such diseases, with the ultimate aim of understanding why certain people are affected and developing better treatments and markers of disease. The study comprises three parts:

1. Collection of medical information in a database in which we will record data about potential disease risk factors (including genetic and environmental factors) and outcomes.
2. Collection of samples of blood and urine and of spare tissue from diagnostic biopsies if available.

3. Long-term storage of the samples in a central location (referred to as a Bioresource or Bio-bank) so that they can be used for future research projects aimed at better understanding, diagnosing and treating rare kidney diseases.

People who have been diagnosed with a rare kidney disease are being asked to participate in these three study parts. Additionally, healthy blood relatives of people with rare kidney diseases as well as unrelated healthy persons are also being asked to participate.

Using information gained from all parts of the study, we have the potential to test for certain risk factors that may help with patient care in the future, for example by increasing medical knowledge so that the most appropriate tests and medicines can be selected.

Some of the information gained will involve genetic testing of people who have been diagnosed with rare kidney disease and of their healthy relatives.

It is important that you understand that the clinical data and patient samples from participants will be stored indefinitely in a Bio-bank for potential use in future novel rare kidney disease research which is not necessarily defined at the time you consent to enrolment in the rare kidney disease registry. For example, the results of current research may be used as the basis for future research. Any future research will be performed after review and approval by an ethics committee.

All information collected for this biobank will be kept strictly confidential.

Who is organising and funding this study?

The study is organised by Professor Griffin, who is a kidney specialist at Galway University Hospitals, along with a network of kidney specialists throughout Ireland. This network is led by Professor Mark Little, a kidney specialist based in Trinity College Dublin. Some of the genetic studies are led by Prof. Peter Conlon of Beaumont Hospital, Dublin. The study is funded by the Science Foundation of Ireland.

How will it be carried out?

There are four types of samples that we may ask your permission to collect and use in the laboratory for research studies of rare kidney disease: 1) Samples of blood; 2) DNA samples (taken from your blood or saliva); 3) Urine samples and 4) Samples of tissue from diagnostic biopsies of kidney and other tissues.

Samples of biopsy tissue will **only** be collected if you have required a biopsy as part of your normal medical care and if there is tissue left over that is not needed for diagnostic testing.

All of these samples will be retained indefinitely in a central Bio-bank located at Trinity College Dublin and will be an on-going resource for future research that may develop in the future.

Details about the different samples are as follows:

1) **Blood samples**. We will need approximately 40mls of blood from you, equivalent to two or three tubes of blood which will be taken by a trained medical professional. We are undertaking the research over five years ask your permission to take up to four separate samples from you during this time. Usually, we will take these extra blood samples at the same time as you are having routine clinic blood tests performed for your medical care. The blood samples will be separated into cell and liquid components and these will be stored in a secure locked freezer using an anonymised code and may be sent to be tested in outside laboratories.

2) **DNA samples** will be extracted from one of your blood samples or will be collected in a saliva sample. The samples will be stored in a secure locked freezer using an anonymous code and may be sent to be tested in outside laboratories.

3) **Samples of your urine.** We will need approximately 100 ml of urine for research studies and will ask you to collect this in a specimen jar up to four times during the research study. The urine samples will be stored in a secure locked freezer using an anonymised code and may be sent to be tested in outside laboratories.

4) **Biopsy samples.** If you are undergoing a biopsy for clinical reasons and you agree to your biopsy sample being used for research purposes, any excess tissue obtained will be collected in a separate container and will be stored in a secure locked freezer using an anonymised code and may be sent to be tested in outside laboratories..

For all of these samples, the data will be completely confidential and no one testing them in a laboratory will have any of your personal details.

At no stage will we be giving you any extra medications during this research. We simply need samples from you that we will store in the laboratory. Samples may be frozen for some months as we may need to repeat laboratory tests or they may be sent to be tested in outside labs. No one working with your samples will know who you are. All the samples we take will be coded and only the study investigators will be able to relate your clinical condition to the results of the laboratory research. The samples you give are purely for research and the results of the research will have no direct benefit for you. However, we hope that the information we obtain from this study will give a better understanding of why and how rare kidney diseases occur and how we may be able to better diagnose and treat them in the future. Some of the research studies may involve testing for genes that are involved in kidney disease and that could potentially affect other members of your family.

Procedures/ What will happen to me if I agree to take part?

You will first be given a chance to ask any questions about the information contained in this leaflet then will be asked by a doctor or nurse to sign a form indicating that you agree to take part.

After this, on the same day or on a later date, you will have additional blood and urine samples and, if relevant, additional biopsy tissue collected by a member of the research team and your medical records will be examined.

Over the next five years you may be asked to provide additional blood, urine and excess biopsy tissue samples on up to 4 more occasions.

What alternative are available to me?

You will have the option of not taking part in the study and of withdrawing from the study before it is completed. The study will have no effect on your treatment.

Risks & Discomforts

Collection of medical information: With any research study that involves collection of medical information, there is a risk that confidential information about you and your health could be seen by people not involved in your medical care. In this study, we will ensure that this risk is minimised by treating your identity and collected medical information as highly confidential and by using an anonymised code on all of the research samples and medical information collected for research purposes. Only Prof. Griffin or members of his research team based at Galway University Hospitals will have access to the key that links your identity to the code.

Blood and urine samples: With all blood sampling there is a risk of some mild discomfort during the blood-taking or mild bruising of the skin afterwards. We will ensure that this risk is minimised by having blood samples taken only by trained medical professionals in a specialised hospital or outpatient clinic environment.

For both blood and urine samples, there is a very small possibility that testing carried out for research purposes will reveal new information about your medical condition or risk of future complications. Although this could have a benefit if it allowed for earlier diagnosis and treatment of a new medical condition than might otherwise have been possible, it could also be associated with unexpected distress and anxiety. If our research results in the discovery of new medical information that could affect your immediate or future health, we will act on this in the following way:

You and your primary care doctor will first be contacted by letter or phone by a member of the research team to provide a basic explanation of the result and its health implications. In a follow-up phone conversation or more detailed letter, advice will be provided, if necessary, about appropriate further evaluation and

follow-up. If needed, the study investigators will provide assistance in arranging suitable medical evaluation and follow-up as dictated by individual situations.

Genetic testing (DNA testing): Testing of your DNA samples or DNA samples taken from any of your blood relatives who have agreed to take part in the study could reveal the presence of a hereditary gene that causes or increases the risk for kidney disease. If you have already been diagnosed with kidney disease, this information may not have significant new medical implications for you personally. However, it could result in one or more of your relatives (including your children) being found to have previously unknown risk for kidney disease. It could also have implications for the risk for kidney disease among your future children. Although this could have benefits for you or your relatives by allowing for early diagnosis and treatment, it could also be associated with unexpected distress and anxiety or with unexpected increased cost for life and health insurance for the individuals found to be affected.

If our research study results in the discovery of new genetic information that could affect your own medical risk and healthcare or those of your relatives, we will act on this in the following way:

A letter will first be written by one of the study investigators to you and to any of the participating members of your family with basic information about the gene identified. You will be given contact information to speak, if you wish, with one of the study investigators who will provide more detail and, if necessary, assist you in making an appointment with a specialist who can provide appropriate follow-up investigation, care and genetic counselling.

Benefits of this Study

You will not benefit directly from taking part in this study. However, it is possible that research carried out using samples and medical information collected for the study will reveal new information about rare kidney disease that will be of value to the future medical care of people suffering from such diseases or to their family members.

Compensation

There is no compensation for participating in this study.

Will there be any additional costs involved?

There will be no additional costs.

Confidentiality

All information collected about you during the research will be kept strictly confidential. Given the rarity of the conditions under study, it will be necessary to combine information across several sites in Ireland and, possibly, abroad. Any

information about you which leaves Galway University Hospitals in this manner, or is published, will be anonymised so that no one from outside the hospital can identify you. Your name and address, hospital number or date of birth will not be published or passed on in any way.

Project Duration

Recruitment for the study is currently planned to run for a period of five years but may continue for a longer duration. Samples and medical information collected for the study will be securely stored for an indefinite period of time.

What if something goes wrong as a result of my participation in this study?

It is very unlikely that anything will go wrong as a result of your participation in this study but, if you have a concern during the course of the study, you should contact Professor Griffin at the numbers provided on this information sheet.

Your responsibility as a participant

Your only responsibility as a participant in the study is to let us know if you do not understand any part of the study procedure.

Our responsibility to you as investigators

We are responsible for explaining the study carefully to you, for answering any questions or concerns you have about the study at any time, for protecting the confidentiality of your medical information and for making sure that your medical care is not affected in any way by your participation.

If you require further information

For additional information now or any future time please contact:

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