

## Identification of genes involved in renal, electrolyte and urinary tract disorders

### An Information Leaflet

You are invited to participate in a research study because you or a relative of yours is thought to suffer from an inherited condition affecting the kidney (renal), the blood salts (electrolytes) or the urinary tract. This study aims to identify the gene(s) causing this disorder. Before you decide if you want to participate, we would like to give you some information about this study.

#### First we want you to know, that:

Taking part is entirely voluntary.

You may choose not to take part, or you may withdraw from the study at any time. In either case, this will not affect your/your relative's ability to receive care here at Great Ormond Street Hospital (GOSH).

You will be offered no benefits for taking part. The research may give us knowledge that may help people in the future.

Now we describe this research study. Before you decide about taking part, please take as much time as you need to ask any questions and discuss this study with anyone at GOSH, or with family, friends or your GP or other health professional.

#### Why is this study being done?

We are asking to collect a blood and urine sample because you or a relative of yours is thought to have a genetic (inherited) condition affecting the kidney and/or blood salts. In addition, if you had had a kidney biopsy, we ask your consent to use for research the remainder of the tissue not needed by the pathologist for clinical diagnosis. The purpose of this study is to allow laboratory evaluation by scientists of your samples in order to identify the gene that is altered (mutated) in the affected family members and identify the consequences of this in the kidney. We hope that identification of the gene will help understand your/your relative's condition. Participation in the study may lead to the diagnosis of a genetic condition in you or your relative or may just contribute to research in genetics without any direct benefits to you or your family.

#### What is involved in this study?

We may take a detailed family history. We may perform a physical examination including standard measurements such as blood pressure, height and weight. We will collect a blood sample from you and send it for laboratory investigations and to genetic scientists for analysis. About 2-4 teaspoons of blood may be drawn from an adult and 1-2 teaspoons from children (depending on their size).

We may ask also for a urine sample for specific laboratory investigations. We may ask you to have an ultrasound performed to get an impression of the appearance of the kidneys. If you/your child had a kidney biopsy, we may ask the pathologist holding this sample to provide unused material for research investigations.

Similarly, if you/your child had tissue removed during an operation, we may want to use for research investigations part of this tissue, which is not needed for diagnostic purposes by the pathologist.

We may ask to use some of the sample we collect to establish a “cell line”. These cells will contain your genes and will allow us to study them in the future without the need to take another sample.

If you agree, we may also share your/your child’s samples with other organisations conducting research into your/your child’s disease. These organisations might include other hospitals/universities and also commercial collaborators and/or service providers. We will not share any personally identifiable data (such as your/your child’s name, address or date of birth) with any other organisation, meaning that you/your child will not be able to be identified in any way. Neither we nor you/your child will receive any payment for these samples.

#### What are the risks of this study?

There may be some discomfort due to sample collection. During blood collection you may experience some discomfort at the needle site and there is a risk of a bruise forming at the site of collection. There is also a small risk of an infection or fainting. Issues of adoption and paternity (fatherhood) may be discovered in the course of this research project. It is our policy that such information will not be disclosed or discussed with you. If biopsy tissue is used for research investigations, less or no tissue may be left, in case the pathologist wanted to do further clinical investigations at a later time.

#### Are there benefits in taking part in the study?

You will not be offered any payment or other compensation for taking part in this study. In fact, there may be no direct benefits to you or your family. However, we hope that this study will increase our understanding of specific genetic conditions, including the one affecting you/your relative and thus contribute to the medical care, treatment or prevention in the future.

#### What are my options?

You may choose not to participate in the study. You can specify, what samples (blood, urine, biopsy) could be used for research. By agreeing to participate you do not waive any rights that you may have regarding access to and disclosure of your medical notes. For further information on these rights contact Prof Robert Kleta, the principal investigator at 0207 317 7554.

#### What if I change my mind?

You may stop participating in this study at any time. In this case, the sample will be destroyed so it cannot be studied any further.

#### Data protection

GOSH is the sponsor for this study based in the United Kingdom. We will be using information from you and/or your medical records in order to undertake this study and will act as the data controller for this study. This means that we are responsible for looking after your information and using it properly. GOSH will keep identifiable information about you for over 3 years. Your rights to access, change or move your information are limited, as we need to manage your information in specific ways in order for the research to be reliable and accurate. To safeguard your rights, we will use the minimum personally-identifiable information possible.

You can find out more about how we use your information at <https://www.gosh.nhs.uk/our-research/our-research-infrastructure/joint-rd-office/gdpr-and-research>. Our Data Protection Officer is Anna Ferrant and you can contact them at [your.data@gosh.nhs.uk](mailto:your.data@gosh.nhs.uk)

Where can I get more information?

If this information leaflet does not adequately answer your questions, please ask the member of the research team asking for your consent or contact the Principal Investigator, Prof Robert Kleta at 0207 317 7554.