

Supplementary Data

Patient Information Sheet: Inherited Renal Disease: Attitudes to Prenatal Diagnosis and Preimplantation Genetic Diagnosis

We would like to invite you to take part in our research study. Before you decide, we would like you to understand why the research is being done and what it would involve for you. One of our team members will go through the information sheet with you and answer any questions you have. We'd suggest this should take about 10 min. Talk to others about the study if you wish. (Part 1 tells you the purpose of this study and what will happen to you if you take part. Part 2 gives you more information about the conduct of the study). Ask us if there is anything that is not clear.

Part 1

What is the purpose of this study?

No standard currently exists for interest and/or intended uptake of preimplantation genetic diagnosis (PGD) for inherited kidney diseases such as autosomal dominant polycystic kidney disease (ADPKD). This is a baseline study that will inform clinicians working in this field regarding whether there is interest in making prenatal genetic counseling available to patients with ADPKD. A previous study at Royal Free Hospital has looked at attitudes in patients with CKD stages I–IV toward prenatal diagnosis and PGD; the remit of this study is to evaluate attitudes in ADPKD patients with end-stage renal failure receiving dialysis.

ADPKD is the most prevalent potentially lethal monogenic disorder worldwide, affecting 1 in 800 live births (4–6 million people) globally and accounting for 10% of patients receiving renal replacement therapy in the UK.

Why have I been invited to take part?

You are a patient with ADPKD with end-stage renal failure receiving dialysis. Therefore, we would be interested to hear what your attitudes for prenatal diagnosis and PGD are.

Do I have to take part?

Involvement in this study is completely voluntary. We will describe the study and go through this information sheet. If you agree to take part, we will then ask you to sign a consent form. You are free to withdraw at any time, without giving a reason. If you decide not to participate, your clinical care will not be influenced.

What will happen to me if I take part?

As a part of the study, you will be asked to complete a consent form and a questionnaire asking you about your attitudes to prenatal diagnosis and PGD. No aspects of your clinical care will be altered as a result of taking part in the study. The questionnaire will take about 10–15 min to fill out and can be done while you are attending dialysis.

What are the potential benefits of taking part?

The intended benefits of this study are to assess attitudes toward prenatal diagnosis and PGD, and whether patients with end-stage renal failure receiving dialysis as a result of ADPKD would be interested in having prenatal diagnosis or PGD.

Below is some background on prenatal diagnosis and PGD.

Prenatal diagnosis

Prenatal diagnosis is available to families who have a significant genetic condition, and there is a risk of passing this onto their children. We need to find the exact gene change associated with the disease in a family, to offer prenatal diagnosis. When the woman becomes pregnant, she can then be offered a prenatal test. This involves taking a sample of the placenta or amniotic fluid (the fluid that surrounds the baby) and carrying out a gene test on cells from the baby. These tests can be done at around 11–15 weeks of pregnancy. Parents can then be offered the option of terminating an affected pregnancy.

Preimplantation genetic diagnosis

PGD is an alternative to prenatal diagnosis. In PGD, we can test an embryo for a genetic abnormality before pregnancy. Again, we need to know the exact gene change associated with the disease in a family, to offer PGD. Only embryos that do not carry the gene change are placed in the mother's womb. This enables couples with a genetic disease to have an unaffected child without needing to consider whether they would terminate an affected pregnancy.

For PGD, the couple undergoes *in-vitro* fertilization (IVF) to create a number of embryos. One or two cells from each embryo are removed and analyzed to see if it is affected with the change in the gene. One or two unaffected embryos are then transferred to the woman's womb. In some instances, all the embryos may be affected with the disorder, and so none are suitable for transfer.

Background on PGD

PGD was developed in the UK and the first cases were performed in 1989. It is available for many different genetic conditions, including ADPKD. It is technically difficult to perform a genetic test on a single cell and unfortunately some wrong diagnoses have been reported worldwide. Success depends on a number of factors, including the woman's age and whether a couple has had any problems with their fertility. In 2010 (the year for which the most recent data are available), 311 women received 383 cycles of PGD. This resulted in 121 live births (live birth rate of 31.6% per cycle started).

Part 2

Will my taking part in this study be kept confidential?

All information, which is collected about you during the course of the research, will be kept strictly confidential, and any information about you, which leaves the hospital/dialysis unit, will have your name and address removed, so that you cannot be recognized (if it is applicable to your research).

What will happen with the results of the research study?

The data, once collated and analyzed, may be published in journals or conferences to help enhance understanding of attitudes in ADPKD patients with end-stage renal failure receiving dialysis.

Who has reviewed the study?

All research in the NHS is looked at by independent group of people, called a Research Ethics Committee, to protect your interests. This study has been reviewed and given favorable opinion by South Yorkshire Research Ethics Committee.

Attitudes Toward Prenatal Diagnosis and Preimplantation Genetic Diagnosis (PGD) for Autosomal Dominant Polycystic Kidney Disease (ADPKD)

Please answer as many questions as you can. There is room at the end for any comments you would like to add.

The first questions are to find out some information about you.

1. Age: _____ years
2. Gender:
 Female
 Male
3. Marital status:
 Single
 Married
 Living with partner
 Divorced
 Widowed

The next questions are to find out about your experiences with ADPKD.

4. At what age were you diagnosed with ADPKD? _____ years
5. What were the circumstances that led to your diagnosis of ADPKD?
 I have a family history of ADPKD and was diagnosed by ultrasound
 I have a family history of ADPKD and was diagnosed by genetic testing
 I was diagnosed by a scan following investigation for kidney problems
 I was diagnosed by a scan following investigation for other health problems
6. Have any members of your family experienced any of the following? If **yes**, please indicate how many family members.
Dialysis Yes _____ No
Kidney transplant Yes _____ No
Subarachnoid haemorrhage Yes _____ No
7. Have you ever seen a genetics doctor or genetic counsellor?
 Yes No Unsure
8. Have you ever had genetic testing for ADPKD?
 Yes No Unsure

These next questions are to find out your views on having children.

9. Do you have children?
 Yes (please indicate how many) _____ No

10. Do you intend to have (more) children in the future?
 Yes No Unsure
11. If you intend to have children in the future, are you concerned that they may also have ADPKD?
 Yes No Unsure
12. Have you already made the decision not to have children because of the concern they might develop ADPKD?
 Yes No

These next questions are to find out your views on prenatal diagnosis and termination of pregnancy.

13. Do you think that a termination is acceptable if a couple or woman decides that she does not want to be pregnant in an apparently normal pregnancy?
 Yes No Unsure
14. In general, do you think that a termination is acceptable if the child has a serious genetic abnormality, such as Down syndrome, cystic fibrosis, etc?
 Yes No Unsure
15. Would you consider prenatal diagnosis and termination of pregnancy because the foetus is found to have ADPKD?
 Yes No Unsure

These next questions are to find out your views on preimplantation genetic diagnosis (PGD).

16. If you have already completed your family, do you think you would have opted for PGD if it had been available at the time?
 Yes No Unsure
17. If you are still going to have children, do you think that you might consider preimplantation genetic diagnosis as a reproductive option?
 Yes No Unsure
18. If your answer to questions 13 or 14 was **yes**, would you be willing to pay for PGD? The cost is about £10,000.
 Yes No Unsure
19. If your answer to questions 13 or 14 was **no**, what factors have made you decide against considering PGD?
 Cost Inconvenience
 Having to go through IVF treatment The small chance of misdiagnosis
 Low success rate Other – please specify

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20. Even if you would not consider PGD for yourself, do you think that this procedure should be offered to other people with ADPKD?
 Yes No Unsure
 21. Given that ADPKD can be passed onto children, if prenatal diagnosis and PGD were available to you on the NHS, which of the following option would you choose? (Please only tick one option)
 I/we would not have more children
 I/we would have more children and not have any prenatal tests done
 I/we would have more children, but I/we would request prenatal diagnosis
 I/we would have more children, but I/we would request PGD
 I/we would consider adoption if I/we wanted (more) children
 I/we are not able to have more children
 I/we do not want any children

Comments – please feel free to write any comments here.

Thank you very much for completing this questionnaire.
Your views will be extremely helpful in making decisions about need for PGD.