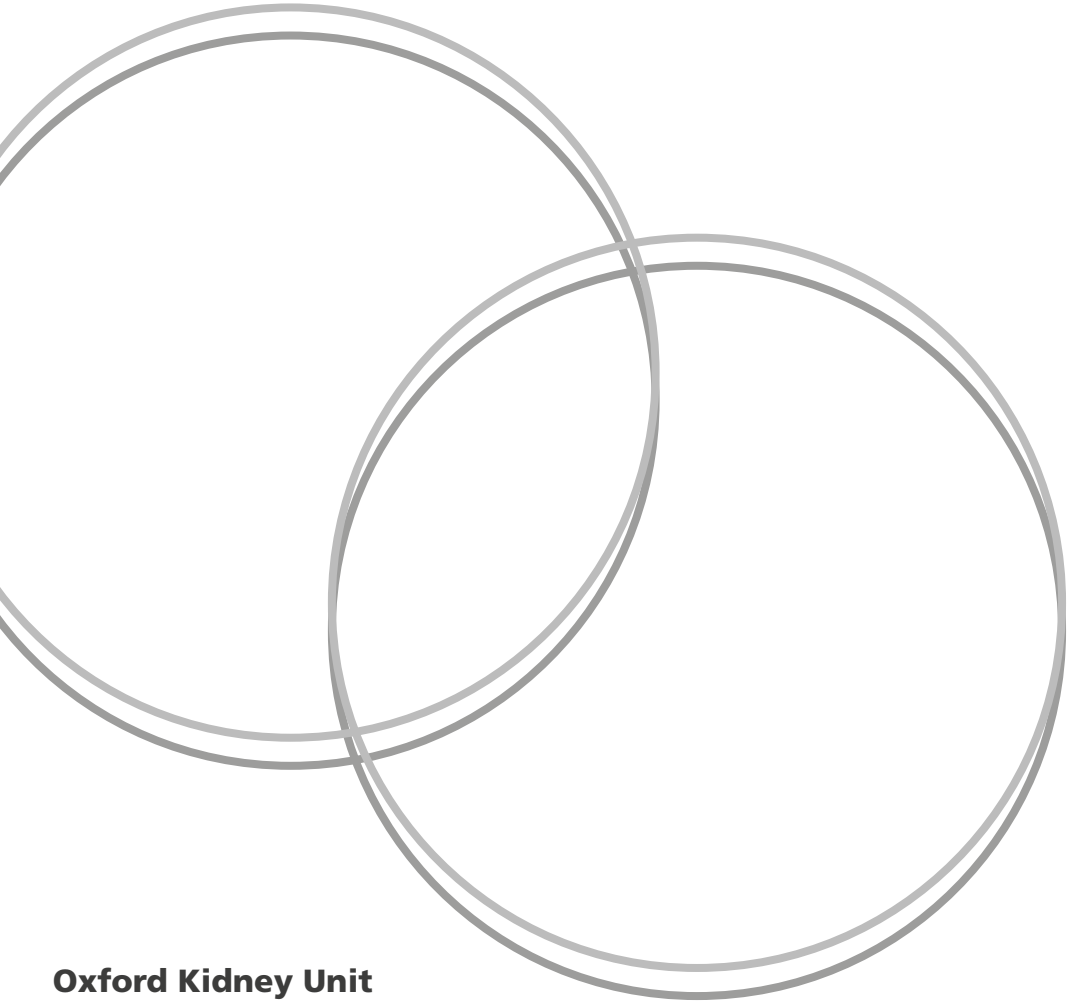
NHS
Oxford University Hospitals
NHS Foundation Trust

Autosomal Dominant Polycystic Kidney Disease

Information about ADPKD



Oxford Kidney Unit

We have written this leaflet to provide you with information about Autosomal Dominant Polycystic Kidney Disease (ADPKD). This is also known as Adult Polycystic Kidney Disease (APKD). If there is anything else you feel you need to know after reading the leaflet, please speak to your kidney team.

What does a kidney do?

Most people have two kidneys, each about the size of a fist, located on either side of the spine at the bottom of the ribcage. The kidneys are responsible for a number of functions. The most important function is removing waste products and balancing fluid levels in the body.

Other functions include regulating your blood pressure, production of the hormone 'erythropoietin' (which controls the production of red blood cells) and converting vitamin D from sunlight, which helps to make strong and healthy bones.

What is Autosomal Dominant Polycystic Kidney Disease (ADPKD)?

ADPKD is an inherited (genetic) condition, in which fluid-filled cysts develop and grow in both kidneys. People without ADPKD may develop a few cysts, but if you have ADPKD you are likely to develop multiple cysts (sometimes hundreds). 1 in every 800 people is affected by ADPKD.

Most people with ADPKD have inherited a faulty gene from one parent and a normal gene from the other parent. However, 1 in 10 people with ADPKD have no family history of the condition, as the fault in the gene has developed 'out of the blue'. Unfortunately, this means the faulty gene can now be passed on to their children.

The child of a parent with ADPKD has a 50% (1 in 2) chance of inheriting this condition. The risk of inheriting this condition is the same for men and women. A member of an ADPKD family who has not inherited the faulty gene cannot pass the condition on and will have completely normal kidneys.

The risk of inheriting ADPKD is the same for women and men (but men may develop problems at a slightly earlier age).

Most cases of ADPKD are caused by one of two faulty genes. The different genes and the type of mutation can influence how severe the disease is. We now know that several other genes can cause cystic kidney disease, although less is known about how those conditions progress.

How is ADPKD diagnosed?

- **Ultrasound** – of your kidneys; this is a simple test that usually reveals cysts of a size greater than 1cm. Special jelly is placed onto your back and a scanner that carries ultrasound waves is rolled back and forth over the area where your kidneys are. The sound waves create an image of your kidneys on a screen.
- **blood test** – to check the level of your kidney function.
- **urine test** – to check for blood or protein.

What are the symptoms of ADPKD?

Many people with ADPKD have no symptoms (especially if they are under 30 years of age). However, some people with ADPKD experience the following problems:

- Passing blood in the urine (haematuria). This happens in about half of people with ADPKD and usually stops without treatment.
- Abdominal pain or a swollen abdomen. As more cysts develop and grow larger, your kidneys become larger. You may be able to easily feel them on either side of your abdomen.
- High blood pressure (proteinuria not generally an issue).
- Low back pain (may also be called loin pain).
- Sudden worsening of pain over the kidneys. This is usually because of bleeding from a cyst.

What will happen to my kidneys?

The cysts can continue to develop (up to several hundred) and grow in size. They gradually replace the 'normal' kidney tissue, causing a reduction in your kidney function.

By the age of 60, around half of the people with ADPKD will develop kidney failure and need dialysis or a kidney transplant.

What other problems could I get?

- High blood pressure (common).
- An increased risk of developing cardiovascular disease.
- Kidney stones – about 1 in 20 people.
- Weakness of the large bowel wall, leading to finger-like pockets that push out from the bowel wall (colonic diverticulosis). This is also common in people without ADPKD.
- Weakness of blood vessels in the brain, which can develop into aneurisms (swellings - about 1 in 10). There is a very small risk that the aneurism can burst or rupture, causing a brain haemorrhage. These can be detected by a brain scan before they grow large enough to cause a problem.
- Leaky heart valve (about 1 in 5 people). This rarely requires treatment.
- Cysts commonly develop in other organs (such as the liver) but they rarely cause problems.

When should I call for help?

Problem:
Blood in your urine.

What should I do?
If this is severe and lasts more than a few days contact your GP.

Problem:
Symptoms of a kidney stone:

- severe pain in your side (over your kidney) moving to the front of your abdomen
- pain which comes in waves (colic).

What should I do?
If you have severe pain you should seek urgent medical advice by phoning 111, to contact the NHS emergency and urgent care services hotline.

Problem:
Symptoms of a brain haemorrhage (subarachnoid haemorrhage – a rare but serious complication):

- Sudden and severe headache that may include the following symptoms:
  - neck stiffness
  - discomfort in bright light
  - feeling sick or vomiting
  - confusion
  - symptoms of a stroke, i.e. slurred speech, weakness on one side.

What should I do?
You should seek urgent medical advice by phoning 111, to contact the NHS emergency and urgent care services hotline.

What will my kidney doctor do?

There is no cure for ADPKD. The aim of treatment is to control any symptoms and delay the decline of your kidney function.

You kidney doctor will:

- See you in clinic regularly.
- Monitor your kidney function.
- Measure your blood pressure and recommend treatment if it is too high.
- Listen to your heart. If it is suspected you may have a leaky heart valve, your kidney doctor will organise for you to have an ultrasound of your heart (echocardiogram or ECHO).
- Discuss whether it would be helpful to arrange a brain scan. This is particularly important if a member of your family has ever had a bleed in the blood vessels around your brain.
- Speak to you about further treatments which may be available (page 9).
- Refer you to a pre-dialysis specialist nurse, if your kidney function is declining and we estimate you are around 18 months from needing dialysis or a transplant.

What can I do to help my condition?

- If you smoke, then you should stop.
- Have your blood pressure measured regularly, as good blood pressure control is very important in protecting your kidneys and your heart. You won't know you have high blood pressure unless you have it tested, as it rarely causes any symptoms. Most people with ADPKD develop high blood pressure at some stage in their life.
- Your kidney doctor will give you a blood pressure target and advise you what to do if your blood pressure is too high. Some people measure their own blood pressure at home.
- Eat a healthy diet (your kidney team will advise you if changes are needed to your diet). Let your kidney team know if you would like to speak to a dietitian.
- Avoid anti-inflammatory painkillers (e.g. ibuprofen), as they may cause your kidney function to become worse.
- Inform any clinician prescribing you a new medication that you have ADPKD, so that they can check the new medicine is safe for you to take.
- Exercise as recommended by the NHS. At least 150 minutes of moderate intensity activity a week or 75 minutes of vigorous intensity activity a week.

Further treatments for ADPKD

Tolvaptan is a medication that is recommended by the National Institute for Health and Care Excellence (NICE) to treat ADPKD in adults.

It is used to slow down the growth of cysts, reducing overall kidney growth and preserving kidney function for longer.

Tolvaptan is only available in certain situations.

Your kidney team will talk with you about whether this may be the right treatment for you. We also have a separate leaflet about Tolvaptan.

Should my family be tested for ADPKD?

You may wish to let your family know that you have ADPKD, as they might choose to be tested to see whether they also have it.

If a member of your family has any of the following symptoms they should have an ultrasound examination of their kidneys to find out whether they have ADPKD.

- raised blood pressure
- more than one urine infection as an adult
- pain in their side (over their kidney)
- blood in their urine.

Any family members who do not have symptoms of ADPKD may still want to consider having the following checks:

- yearly blood pressure checks
- yearly blood tests to check their kidney function
- an ultrasound examination of their kidneys to see if they have ADPKD.

If you take out a new critical illness, life or travel insurance policy you will need to inform the insurer that you have ADPKD.

Some people choose not to have the ultrasound screening test if their blood pressure and kidney function are normal, as being diagnosed with ADPKD may have implications for health insurance and or some particular jobs, such as the armed forces. However, you will still need to advise your insurer that you have ADPKD in your family. They will then decide whether they want you to have a medical check.

Screening family members for ADPKD using an ultrasound test is not helpful until they are over 20 years of age. This is because the cysts do not usually appear until this age.

Any family members between the age of 20 and 30 years old may want to consider having an ultrasound scan. If the scan shows two or more cysts it is likely that they have ADPKD. If the scan does not show any cysts we would suggest a further scan at 30 years of age, as in some people the cysts do not develop until they are in their 30's.

What about genetic testing?

Genetic testing uses your own DNA to identify whether you have inherited a particular condition or gene. Genetic testing can identify the mutation responsible in 90% of cases. It is particularly useful when the ultrasound exam is not clear-cut or if you are the first person in your family to have this condition. Even with the latest technology, genetic testing may not give a clear answer in 10% people with ADPKD.

If a member of your family wishes to donate a kidney to you, genetic testing may be needed, as it is important to know whether your family member has ADPKD before donating their kidney.

What if I am planning a family?

If you are a woman who is planning to have a baby, it is really important that you speak to your kidney doctor, as you may need some changes to your medicines. Your kidney doctor and obstetrician (pregnancy specialist) will be able to provide you with information and support, so that your pregnancy is as safe as possible.

If you have ADPKD, there is a 1 in 2 (50%) chance of your child inheriting the condition. The condition cannot be detected in pregnancy using ultrasound scans. Antenatal genetic testing (before a baby is born) can sometimes be done, if you would like this. Further details about this should be discussed with your kidney doctor.

Can women with ADPKD have a normal pregnancy?

Women who have normal or near-normal kidney function (stage 1-2) and become pregnant, usually have a fairly normal pregnancy, but you should tell your obstetrician and midwife that you have ADPKD.

Pregnant women with reduced kidney function (stage 3-4) have an increased risk of developing pre-eclampsia (a particular kind of high blood pressure which can cause problems in pregnancy). They may also have an early delivery and a further decline in their kidney function. You should discuss these risks with your kidney doctor who may refer you for more specialist assessment. Your blood pressure and kidney function will need to be monitored closely during your pregnancy.

Information about you

If you have ADPKD, your kidney doctor will talk to you about joining the RADAR registry. This helps scientists to look for better ways to prevent and treat this condition.

Useful websites

NHS Website

NHS website which provides information on specific conditions.

Website: www.nhs.uk/conditions/Autosomal-dominant-polycystic-kidney-disease/Pages/Introduction.aspx

PKD Charity

Providing information to people with ADPKD and their family/carers.

Website: www.pkdcharity.org.uk

Oxford Kidney Unit

Lots of information about the Oxford Kidney Unit for patients and carers.

Website: www.ouh.nhs.uk/oku

National Institute Health and Clinical Excellence (NICE)

Information about tolvaptan.

Website: www.nice.org.uk/guidance/ta358/resources/tolvaptan-for-treating-autosomal-dominant-polycystic-kidney-disease-82602675026629

RareRenal (The Renal Association)

Run by UK based kidney doctors promoting research. There are links from the website to join the register and be the first to know about potential new treatments and clinical trials.

Register: www.rarerenal.org/radar-registry

UK Kidney Association

Patient information leaflets and advice.

Website: www.ukkidney.org/patients/information-resources/patient-information-leaflets

Kidney Patient Guide

Information for patients with kidney failure and those who care for them.

Website: www.kidneypatientguide.org.uk

Kidney Care UK

A charity which has lots of practical support and information for people with kidney disease.

Website: www.kidneycareuk.org

Six Counties Kidney Patients Association

The SCKPA is run for patients by patients or family members.

They offer support to people suffering from kidney disease or who are on dialysis. They work closely with the Oxford Kidney Unit and have branches in Oxfordshire, Northamptonshire, Buckinghamshire, and Milton Keynes, and parts of Wiltshire, Gloucestershire and Berkshire.

Website: www.sixcountieskpa.org.uk

OUH Patient Portal Health for Me

Please ask a member of the renal team to sign you up to the patient portal.

Website: www.ouh.nhs.uk/patient-guide/patient-portal

Further information

If you would like an interpreter, please speak to the department where you are being seen.

Please also tell them if you would like this information in another format, such as:

- Easy Read
- large print
- braille
- audio
- electronic
- another language.

We have tried to make the information in this leaflet meet your needs. If it does not meet your individual needs or situation, please speak to your healthcare team. They are happy to help.

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