

Oxford Kidney Unit

Autosomal Dominant Polycystic Kidney Disease

Information for patients



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 doctor.

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 'erythropoetin' (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 20 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 1 in 2 (50%) chance of

inheriting the condition. A member of an ADPKD family who has not inherited the 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). This way of inheriting a condition is called 'dominant' inheritance.

There are two Autosomal Dominant types of ADPKD:

- Type 1 (17 in 20 people) which tends to cause more problems
- Type 2 (3 in 20 people) which tends to cause less problems.

There is also a rare form of inherited cystic kidney disease, in which a faulty gene has been inherited from each of the parents. However, this condition usually causes problems soon after a baby is born.

How is ADPKD diagnosed?

The best test is an 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.

If the ultrasound does reveal cysts, your kidney doctor will ask for you to have the following tests:

- **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 you are under 30 years of age). However, some people with ADPKD experience the following problems:

- 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.
- 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.
- protein in the urine. You may notice foam when you pass urine.

What will happen to my kidneys?

The cysts can continue to develop (up to several hundred) and grow in size from 2mm to 2cm. They gradually replace the 'normal' kidney tissue, causing a reduction in your kidney function.

By the age of 60, 40-50% of people with ADPKD will develop kidney failure and need dialysis or a kidney transplant.

What other problems could I get?

- high blood pressure (this is common)
- urine infections
- kidney stones – about 1 in 20 people with ADPKD develop these.
- 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 aneurysms (swellings). There is a very small risk that the aneurysm may burst or rupture, causing a brain haemorrhage. This is rare but serious.
- dehydration – some people lose too much salt and water into their urine.
- leaky heart valve (about 1 in 5 people with ADPKD develop this). This rarely requires treatment.
- cysts may also develop in other organs (8 out of 10 people), such as the liver, but they rarely cause any problems.

When should I call for help?

Problem:	What should I do?
Blood in your urine	If this is severe and lasts more than a few days contact your GP.
Symptoms of a urine infection: <ul style="list-style-type: none"> • pain when passing urine • smelly urine • cloudy urine • fever/high temperature (38°C or higher) • increased pain over your kidneys. 	See your GP for a urine test. If the infection is severe it could mean that one of the cysts may be infected. If so, you will need a 4 week course of a particular antibiotic (ciprofloxacin) to get rid of the infection, as this antibiotic is better at getting into the cysts.
Symptoms of a kidney stone: <ul style="list-style-type: none"> • severe pain in your side (over your kidney) moving to the front of your abdomen • pain which comes in waves (colic). 	If you have severe pain you should seek urgent medical advice by phoning 111, to contact the NHS emergency and urgent care services hotline.
Symptoms of a brain haemorrhage (subarachnoid haemorrhage – a rare but serious complication): <ul style="list-style-type: none"> • Sudden and severe headache that may include the following symptoms: <ul style="list-style-type: none"> - neck stiffness - discomfort in bright light - feeling sick (nausea) or vomiting - symptoms of a stroke, i.e. slurred speech, weakness on one side - confusion. 	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.

Your kidney doctor will:

- see you in clinic regularly
- monitor your blood tests. We use a blood test to assess your level of kidney function. The level of kidney function is divided into 5 stages (see the following chart).
- 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).
- ask about your family history. If a member of your family has ever had a bleed in the brain, you will be offered a scan of the blood vessels around your brain using MRI (Magnetic Resonance Imaging). If an aneurysm is found, you may need an operation to prevent it from bursting.
- speak to you about further treatments which may be available (see page 9)
- refer you to a pre-dialysis specialist nurse, if your kidney function is low (at stage 4-5).

Level of kidney function chart

GFR is a measurement of how many millilitres (ml) of fluid your kidneys can filter from your blood in one minute (measured in ml/min).

It is difficult to measure the GFR directly, so it is estimated using a formula. This result is called the estimated GFR or eGFR.

Stage	Kidney function as eGFR	Description	Treatment
Stage G1	More than 90ml/min	Normal kidney function, but other damage found, e.g. protein in the urine.	Blood pressure monitoring Clinic reviews with your kidney doctor
Stage G2	60-89ml/min	Normal to mildly reduced kidney function, but other damage found.	
Stage G3A Stage G3B	30-59ml/min	Mild to moderately reduced kidney function. Moderate to severe reduction.	
Stage G4	15-29ml/min	Severely reduced kidney function.	Planning for dialysis or transplant
Stage G5	Less than 15ml/min	Kidney failure.	Dialysis or transplantation

Kidney function is usually calculated from your blood test result. As the normal value is about 100, it gives an approximate 'score out of 100' for kidney function. However, your normal kidney function will depend on your height, weight and age.

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. Your kidney doctor will tell you if this is something you should do.

- Eat a healthy diet (your kidney doctor will advise you if changes are needed to your diet). Let your kidney doctor know if you would like to speak to a dietitian.
- Avoid anti-inflammatory painkillers (e.g. ibuprofen, aspirin), as they may cause your kidney function to become worse.
- Inform any doctor prescribing you a new medication that you have ADPKD, so that they can check the new medicine is safe for you to take.
- Avoid contact sports, such as rugby and football, as these are more likely to cause injuries to your kidney.

Further treatments for ADPKD

Tolvaptan (Jinarc®) is the first drug specifically for the treatment of ADPKD. In 2015, it was approved for use in the UK by the National Institute of Health and Care Excellence (NICE) for some people with ADPKD. Your doctor will discuss whether this might be the right treatment for you. A separate information leaflet about tolvaptan is available.

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. However, the genes involved in ADPKD are large and complicated. In about 30% (30 in 100) of people who have ADPKD, an abnormal gene can't be found. This is why this test is not used routinely.

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. Your blood pressure and kidney function will need to be monitored closely during your pregnancy.

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.

Information about you

If you have ADPKD, your kidney doctor will talk to you about joining the National Congenital Anomaly and Rare Diseases Registration Service (NCARDRS). This helps scientists to look for better ways to prevent and treat this condition.

Further information

If you need further information about ADPKD, please speak to your kidney doctor. Useful information is also available online:

NHS Choices

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/

Support line: 0300 111 1234

Oxford Kidney Unit

Useful information about the Oxford Kidney unit, for patients and their relatives.

Website: www.ouh.nhs.uk/oku

National Institute Health and Clinical Excellence (NICE)

Information about tolvaptan.

Website: www.nice.org.uk/guidance/ta358

If you have a specific requirement, need an interpreter, a document in Easy Read, another language, large print, Braille or audio version, please call **01865 221 473** or email **PALSJR@ouh.nhs.uk**

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