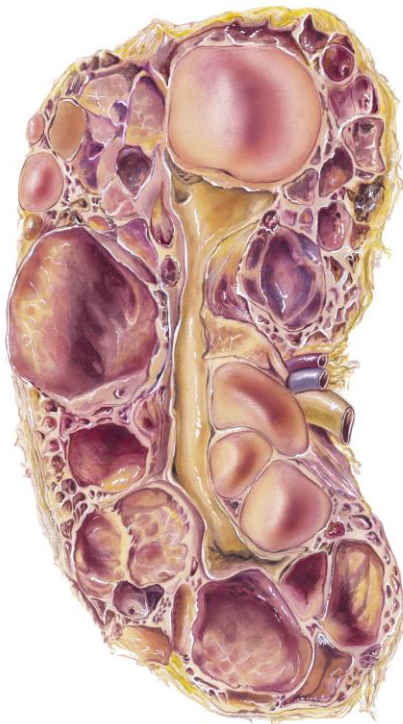


I want to know more about ADPKD

This booklet is about autosomal dominant polycystic kidney disease (ADPKD). It is intended as a general guide for people who have been diagnosed with ADPKD and their families and carers. It explains what causes ADPKD, how it is diagnosed and monitored, its symptoms and complications, and how the disease might progress.

What is ADPKD?

Autosomal dominant polycystic kidney disease (ADPKD) causes cysts - sacs filled with fluid - to develop in the kidneys and liver. The cysts are not cancerous and the fluid inside them is harmless.



Drawing of a polycystic kidney cross section, showing cysts.

In ADPKD, cysts typically begin to form in late childhood or early adulthood. Over time they get bigger and more of them appear, causing the kidneys and liver to enlarge. Most people with ADPKD have numerous cysts in their kidneys and liver by the time they reach old age. In addition, around 10 per cent of people with ADPKD have cysts in their pancreas, although these almost never cause problems [1].

The cysts cause very slow damage to the kidneys that, in some people, results in kidney failure - normally during later adult life. Usually, both kidneys are affected.

How common is ADPKD?

At least one person in every 1,000 has ADPKD, meaning there are more than 60,000 people with the disease in the UK [2]. It affects both sexes and all races equally.

What causes ADPKD?

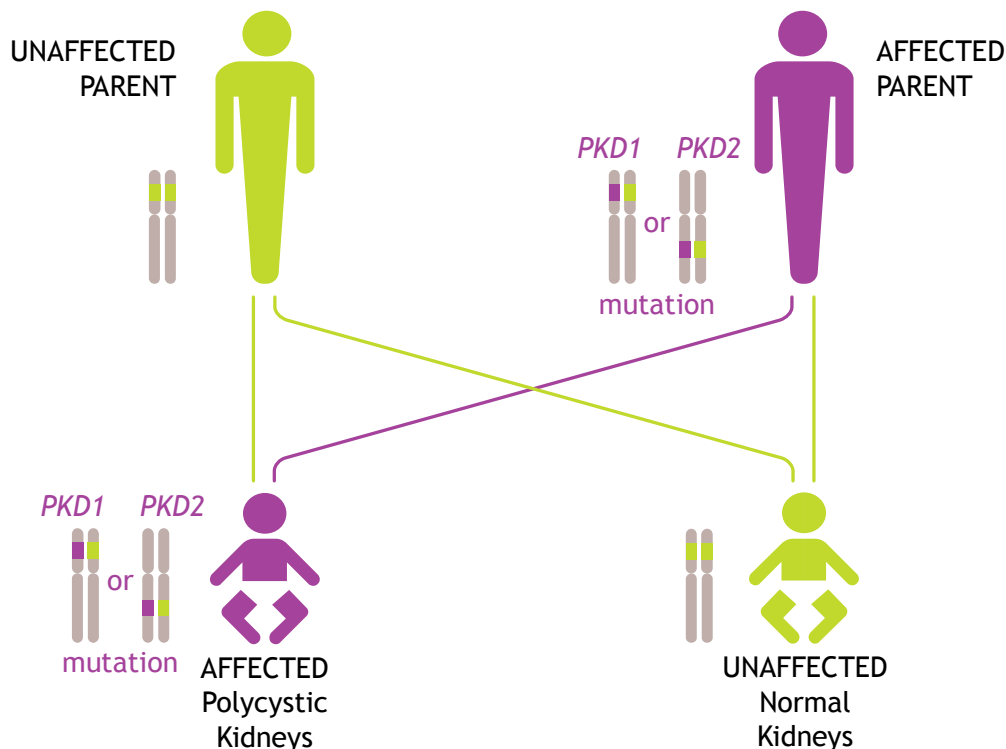
ADPKD is an inherited disease, meaning it is passed on from parents to their children through their genes. Genes are the instructions our cells need to make our body parts and organs. We all have two copies of each gene, one from each of our parents.

ADPKD is caused by an abnormality - often called a mutation - in one of two genes involved in ADPKD. These genes are called *PKD1* and *PKD2*. Most people get the disease when they inherit a faulty copy of one of these from a parent with ADPKD.

Sometimes, though, a new genetic change occurs spontaneously, causing the disease in a person with healthy parents who do not have the mutated gene³. This new mutation can then be passed onto the next generation.

You only need to receive one mutated gene copy to have ADPKD. So, a person with the disease has a one in two (50 per cent) chance of passing the disease on to each of their children, depending on whether they pass on their faulty or normal copy of the gene.

This is known as a dominant pattern of inheritance (see diagram below).



If a child inherits the normal copy from an affected parent, then they (and any children they go on to have) will not have the disease.

What problems does ADPKD cause?

The cysts form at random and can appear anywhere inside your kidneys and liver. As the cysts get bigger they cause the affected organs to enlarge. The larger they get, the more space they take up and as a result, your tummy may become swollen and you may experience abdominal pain.

You may also notice blood in your urine, known as haematuria. This happens when a tiny blood vessel in the wall of a cyst breaks, causing the cyst to bleed. Around 4 in 10 (40 per cent) of people with ADPKD experience pain - usually in their sides or their back - or blood in their urine [4].

ADPKD causes high blood pressure - or hypertension - and this sometimes develops before any cysts have grown. Mild to moderate hypertension rarely causes any symptoms but it increases your risk of heart disease and stroke, and can cause your kidney function to decline more quickly. So, it is very important that high blood pressure is diagnosed and treated early.

Less commonly, kidney cysts can become infected. This is usually very painful and typically accompanied by other symptoms, such as sweating, fever and high temperature. Cyst infections sometimes occur following a bladder or urinary tract infection (UTI). If you suspect you have a UTI, it is important to see your doctor and get treatment as soon as possible. People with ADPKD are also a little more likely than the general population to develop kidney stones [5].

More rarely, ADPKD can cause swelling of one or more of the small arteries in the brain. This can sometimes result in bleeding into the brain (a brain haemorrhage), which can be dangerous. However, because this is so rare, people with ADPKD are only screened for this complication if they or a member of their family have previously had a brain haemorrhage [6].

Liver cysts tend to be larger in women [7] and, although they can be uncomfortable if they grow very large, they almost never cause liver damage.

Will my kidneys stop working?

Our kidneys have several functions, including controlling blood pressure and filtering waste products from the blood and converting them into urine. As kidney cysts grow, they cause gradual damage to your kidneys and affect how well they work, so they cannot perform these functions as efficiently. In its early stages, kidney damage doesn't usually have any symptoms but it's very important that your kidney function is monitored closely.

If your kidney function deteriorates to as low as 30 per cent (an eGFR of around 30ml/min) you may need to take medications to stay healthy. eGFR is explained below. If it falls below around 20 per cent you may begin to experience symptoms such as extreme tiredness, poor sleep and loss of appetite. Because your kidneys do not work properly, the fluid that you would normally pass in your urine can build up in your body, causing swelling around your ankles and shortness of breath.

However, no two people are the same and your disease may progress at a different rate from someone else's. Accordingly, exact symptoms and their severity will vary from person to person.

What is kidney failure?

Kidney failure - sometimes referred to as end-stage renal disease - occurs when your kidneys have lost almost all of their function and your eGFR falls below 15.

If your kidneys fail, you will need a form of treatment to take over their job, known as renal replacement therapy (RRT). This is usually in the form of dialysis (a means of artificially filtering your blood to get rid of waste products), or a transplant (where a healthy kidney is transferred from one person - the donor - into your body). People can live relatively normal, active lives after having an organ transplant and while on dialysis.

When your eGFR reaches between 15 and 20, your doctor should begin to discuss with you a plan for future RRT. If you are a suitable candidate, you may be placed on the transplant waiting list or go ahead with a live kidney donation when your eGFR falls below 15, even though you might still feel well. Dialysis is usually started when your eGFR falls below 10, depending on your symptoms.

Approximately 1 in 2 (50 per cent) of people with ADPKD develop kidney failure before the age of 60 [8], and occasionally this can occur in much younger people. In some people kidney failure can occur later in life, but around 1 in 4 (25 per cent) of people with ADPKD die naturally in old age without ever developing significant kidney problems [9].

How is ADPKD diagnosed and monitored?

ADPKD is typically diagnosed using an ultrasound scan, which detects the presence of cysts. Once the diagnosis has been made, you won't need to have additional routine scans.

You will need to have blood tests to monitor changes in the levels of creatinine and other substances in your body, and your blood pressure will need to be checked regularly so that high blood pressure will need to be checked regularly so that high blood pressure can be treated effectively. You will be under the long-term care of a specialist renal (kidney) doctor, who you will probably need to see about once a year. If blood tests show evidence of kidney damage you may need to be seen more frequently.

Know your numbers

eGFR

Kidney function is often referred to in terms of 'glomerular filtration rate', or GFR - a measure of how many millilitres (ml) of waste fluid your kidneys can filter from your blood in one minute, given in ml/min.

Because GFR is difficult to measure directly, it is usually estimated by testing the levels of a waste product called creatinine in your blood, and taking into account your age, gender and ethnic group.

The result - your 'estimated glomerular filtration rate', or eGFR - is roughly equivalent to your remaining kidney function.

So, an eGFR of 40ml/min means your kidneys are functioning at around 40 per cent.

Blood pressure

Blood pressure is a measure of how strongly blood pushes against your artery walls as your heart pumps it around your body. It is measured in millimetres of mercury (mmHg) and it is recorded as two figures:

Systolic pressure: the pressure of the blood when your heart beats to pump blood out.

Diastolic pressure: the pressure of the blood in between beats when your heart rests.

For example, if your blood pressure is 135/85, your systolic pressure is 135mmHg and your diastolic pressure is 85mmHg.

You are said to have hypertension if your blood pressure is consistently 140/90 or higher on separate occasions.

A blood pressure reading below 130/80 is considered to be normal.

Can ADPKD be treated?

There is no cure for ADPKD, but treatments can help to reduce your symptoms and lower your risk of complications. For example, medications can reduce raised blood pressure, treat infections and help to manage pain. Some people need to have procedures or surgery too, for example to drain a large kidney cyst. If your kidney

function becomes severely reduced or your kidneys fail, kidney specialists will recommend that you have dialysis or a kidney transplant.

A new drug called tolvaptan (Jinarc®) is suitable for some adults with ADPKD - this can slow the progression of ADPKD [10]. Tolvaptan slows the rate at which the kidneys become enlarged by cysts and can help to slow the decline of kidney function. Tolvaptan might be suitable for you if you have mildly or moderately reduced kidney function and your ADPKD is progressing rapidly [10,11].

You'll find more information about treatments for ADPKD, including tolvaptan, on our website: www.pkdcharity.org.uk

How will ADPKD affect my life?

Most people with ADPKD live completely normal lives with no symptoms almost all of the time. A diagnosis of ADPKD does not prevent people from working, having children, going on holiday, taking exercise or drinking alcohol as normal, although you should avoid vigorous contact sports (such as boxing or rugby) if you have large cysts that are visible on a scan.

You do not have to tell your employer that you have ADPKD unless you are off work for long periods, and insurance companies, banks or building societies only need to be informed if they ask for a medical history. It may be more difficult or more expensive to get insurance, such as life and travel insurance or critical illness cover, if you have been diagnosed with ADPKD. If you have an HGV or PSV driving licence you must tell the DVLA that you have ADPKD.

Being diagnosed with any long-term condition, such as ADPKD, can cause shock or upset. There are lots of things to think about and you may feel worried, anxious or depressed. It is important to acknowledge these feelings and to seek support.

You can also call the PKD Charity Helpline to speak to someone with first-hand experience of PKD. Call 0300 111 1234; lines are open weekdays 10.00am to 4.30pm; or email info@pkdcharity.org.uk

What can I do to help my kidneys?

There is currently no cure for ADPKD, but there are things you can do to protect your kidneys and stay as healthy as possible.

Avoid smoking. Smoking damages the blood vessels in the kidneys and accelerates kidney damage [12].

Control your blood pressure. High blood pressure is harmful to the kidneys and increases the risk of stroke and heart disease. Conversely, damage to the kidneys makes high blood pressure worse, so it is vital that your blood pressure is closely

monitored and high blood pressure is treated early. Blood pressure readings taken in a clinic do not always reflect your blood pressure the rest of the time, so it is a good idea to buy a blood pressure machine and check your blood pressure at home as well.

Watch your diet. A balanced diet is important for everyone to stay healthy. It's also important to limit how much salt you have in your diet because eating salt makes high blood pressure worse.

Take exercise. Try to take regular exercise and be active. This helps prevent weight gain and can help to control blood pressure.

Drink plenty of water. There is evidence to suggest that the body's response to dehydration increases cyst growth. This response can be minimised by drinking plenty of water to stay hydrated.

Avoid NSAIDs. Non-steroidal anti-inflammatory drugs (NSAIDs) are a class of medication commonly used to relieve pain, reduce inflammation and bring down a high temperature. They include ibuprofen and diclofenac. NSAIDs can cause scarring in the kidneys so it's best to avoid them [13]. Ask your doctor or pharmacist to suggest alternatives that are safer for you to take.

More information and support

You can find support and more information about ADPKD and living with the disease from the Polycystic Kidney Disease Charity, by visiting www.pkdcharity.org.uk.

You might also find the following organisations and sources useful:

- NHS Choices - general health information
www.nhs.uk
- British Kidney Patient Association
<http://www.britishkidney-pa.co.uk/>
- NKF
<http://www.kidney.org.uk/>
- Kidney Research UK
www.kidneyresearchuk.org/
- Contact a Family
<http://www.cafamily.org.uk/>
- Blood Pressure UK
<http://www.bloodpressureuk.org/>

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We welcome feedback on all our health information. If you would like to give feedback about this information, please email info@pkdcharity.org.uk

For further copies of this information sheet or other PKD Charity information visit www.pkdcharity.org.uk

If you don't have access to a printer and would like a printed version of this information sheet, or any other PKD Charity information, call the PKD Charity Helpline on 0300 111 1234 (weekdays, 10.00am-4.30pm) or email info@pkdcharity.org.uk

The PKD Charity Helpline offers confidential support and information to anyone affected by PKD, including family, friends, carers, newly diagnosed or those who have lived with the condition for many years.

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