pkd charity
Polycystic Kidney Disease

ADPKD Basics

Autosomal dominant polycystic kidney disease

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I want to know more about ADPKD

This factsheet is for people who have been newly diagnosed with autosomal dominant polycystic kidney disease (ADPKD) and their families and carers. It covers the topics you're likely to want to know more about to understand the disease and how it might affect you.

Getting a diagnosis of ADPKD can be a worrying time. If you'd like emotional support, practical advice and information, ring our confidential PKD Helpline **0300 111 1234** (9.30am–5.00pm Mon to Fri, except bank holidays).

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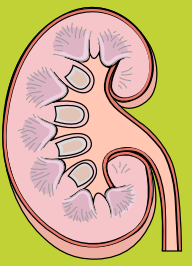

Autosomal dominant polycystic kidney disease (ADPKD) is a genetic condition some people are born with. It causes cysts (balloons of fluid) to develop in your kidneys and sometimes in other organs. Kidney cysts grow over time. Each cyst can grow to a size anywhere between a pea and a grapefruit (Figure 1). Over the years, the cysts can damage much of your kidney tissue and make your kidneys become very large.

With the right care, most people stay in good health for decades after a diagnosis of ADPKD.

Figure 1:

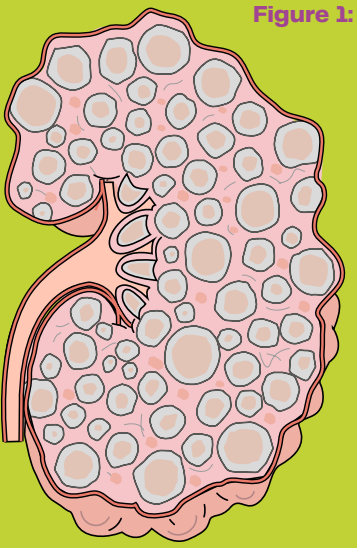

Figure 1: Drawing of the inside of a polycystic kidney (on the right), showing cysts of different sizes, compared with a normal kidney (on the left).

ADPKD is not a disease you can catch, nor is it cancer. Most people with ADPKD get many cysts in their kidneys and liver by their mid-thirties. In addition, around 1 in 10 people get cysts in their pancreas, but these almost never cause problems.

Kidney cysts can stop your kidneys filtering blood properly. Eventually, this can result in kidney failure, which can be treated with dialysis or a kidney transplant. Half of people with ADPKD get kidney failure by the time they turn 60.

Liver cysts can cause problems but it's very unusual for them to cause liver failure. In a few people with ADPKD, the liver grows massively. Rarely, a person may require a liver transplant due to their liver becoming extremely large and related symptoms such as pain and infections.

How common is ADPKD?

In the UK, it's thought that 1 person in every 1,000 to 2,500 has ADPKD. There could be between 30,000 and 70,000 people with the disease in the UK. ADPKD can affect men and women, and people of all ethnic backgrounds.

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How did I get ADPKD?

ADPKD is usually inherited and occurs when gene alterations are passed from a parent to their child. Genes are the instructions our cells need to grow and divide to keep us healthy throughout our lives. We all have two copies of each gene, one from each parent.

ADPKD is most commonly caused by an alteration — often called a mutation — in a gene called PKD1 or a gene called PKD2.

Most people get the disease when they inherit an altered copy of PKD1 or PKD2 from a parent with ADPKD. PKD1 mutations are more common: about 78 out of every 100 people with ADPKD (78%) have an alteration in this gene. About 14 out of every 100 people (14%) have an altered PKD2 gene. Some people have an alteration in a different gene, or the genetic cause is unknown.

Sometimes, a new alteration in a gene occurs by chance in the womb, causing an infant to be born with ADPKD despite having two healthy parents. This is called a 'spontaneous' or 'de novo' mutation and occurs in about 1 in 10 people with ADPKD.

How is ADPKD diagnosed and monitored?

Ultrasound

ADPKD is typically diagnosed with an ultrasound scan at a hospital clinic. Ultrasound uses soundwaves to make an image of the inside of your body. Cysts and enlarged kidneys show up on the scan. After diagnosis, you may have ultrasounds or other scans periodically to check the size of your kidneys. You may also have scans of your liver to check for cysts.

Blood and urine tests

After diagnosis, you'll have periodic blood tests to check how well your kidneys are filtering your blood. These measure your estimated glomerular filtration rate (eGFR), which we explain below. If your kidney function is poor or you're on dialysis, you'll have additional lab tests to check the balance of different substances in your blood that might need correction. A urine test is usually done at the same time to check for other substances such as protein.

eGFR

Kidney function is often assessed using 'estimated glomerular filtration rate', or eGFR. This is a measure of how many millilitres (ml) of waste fluid your kidneys can filter from your blood in one minute. eGFR is usually calculated using a blood test to measure levels of a waste product called creatinine in your blood.

Perfectly functioning kidneys have an eGFR of about 100—110 ml/min. The lower your eGFR is, the poorer your kidney function.

A value between 60 and 89 suggests mild kidney damage. A value between 15 to 60 means you have kidney disease, and a value under 15 means your kidneys are failing.

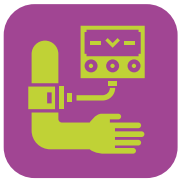
It can be useful to think of eGFR as a percentage. For example, an eGFR of 40 ml/min means your kidneys are working at roughly 40% the level of perfectly functioning kidneys.

eGFR is reasonably accurate but the results can fluctuate, especially if your kidney function is good. eGFR gets lower in all people as they get older. For example, many healthy people have an eGFR of about 80—85 ml/min by the age of 65—74.

Magnetic resonance imaging (MRI)

MRI is a hospital scan that uses a magnet and radio waves to make images of the inside of your body. You might have MRI periodically to check how large your kidneys are and to estimate how much your disease has progressed. Alternatively, an ultrasound or computed tomography (CT) scan might be used.

Blood pressure



Your blood pressure will need to be checked regularly so it can be treated if it becomes too high. A blood pressure test uses an air-filled cuff on your upper arm to measure the strength of your pulse. Many patients choose to check their blood pressure at home but your GP or practice nurse can check it too.

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

- systolic pressure: the pressure of the blood when your heart beats
- diastolic pressure: the pressure of the blood between beats.

For example, if your blood pressure is 135/85, your systolic pressure is 135 mmHg and your diastolic pressure is 85 mmHg. Blood pressure consistently 140/90 mmHg or higher on separate occasions usually indicates high blood pressure.

Genetic testing

Genetic testing is sometimes used for the diagnosis of ADPKD. Not all adults with symptoms of ADPKD need a genetic test to confirm that they have the disease. However, you might have genetic testing if:

- You have symptoms of ADPKD and are below the age of 18 years.
- Your symptoms don't match the classic signs of ADPKD.
- Results of genetic tests could help to plan your health care.

Consultations

You'll be under the long-term care of a specialist kidney doctor (a nephrologist) and will see them for check-ups. Some people may be monitored by a GP if their kidney function is good and they have no complications.

How often you're invited for check-ups depends on:

- how well your kidneys are working
- whether your treatment has recently changed
- any other health problems you have.

Most people with kidney disease have check-ups 1 to 4 times year. A range of other experts will be involved in your care, as needed.

See our website for appointment tips. **www.pkdcharity.org.uk**

What problems does ADPKD cause?

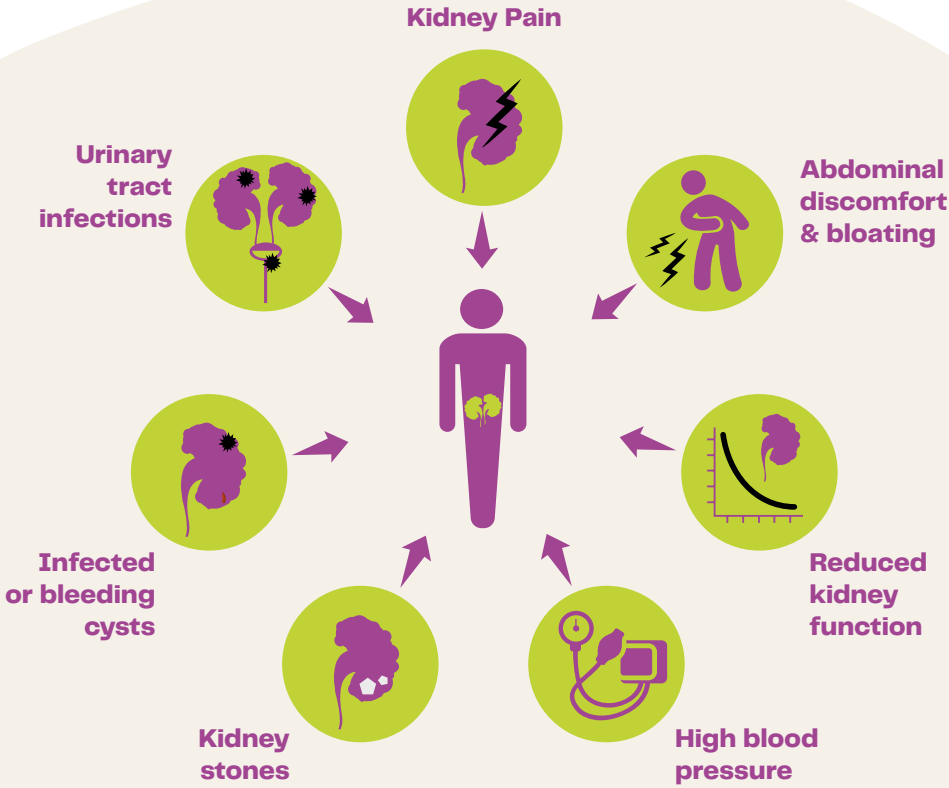


Figure 2: The more common symptoms of ADPKD.

High blood pressure

ADPKD usually causes high blood pressure — known as hypertension. This can start in childhood, but most people with ADPKD get it in their 30s to 40s. Sometimes, it's the first sign of ADPKD. Usually, high blood pressure doesn't cause symptoms and so it's picked up only by blood pressure tests.

It's important to treat high blood pressure because it can increase your risk of heart disease and might increase the speed of kidney damage.

Urinary tract infections

Urinary tract infections are quite common among people with ADPKD; about 3–5 in every 10 people get at least one in their lifetime. They're more common in women than men.

Cyst infections

Kidney and liver cysts can become infected with bacteria, usually from your gut. An infected cyst is usually painful and can cause a high temperature (fever). If you get these symptoms, see your doctor right away.

Kidney stones

People with ADPKD are more likely than the general population to develop kidney stones, which can be painful. These are usually made of uric acid crystals.

Blood in your urine

About 6 in every 10 people with ADPKD get a bleeding cyst or blood in their urine (known as haematuria) at some point. Blood in the urine can happen for a variety of reasons, such as a cyst bleeding, a kidney stone, a cyst infection, or damage to the kidney caused by a big knock. Blood in the urine usually gets better within a week without treatment. Often, the cause is not found.

Let your doctor know if you have blood in your urine. They can check your urine and refer you for further tests if needed.

Abdominal pain and bloating

If you have ADPKD, you'll probably get cysts in your kidneys and liver, making these organs grow over time. The larger they get, the more space they'll take up. Your abdomen (tummy) may become swollen and you may get abdominal or back pain, indigestion or feel full.

Six out of every 10 people with ADPKD have long-term pain (chronic pain). The first step to treating pain is to find the cause. When the cause can't be resolved, medications, psychological approaches and manual therapies can help to manage pain.

Reduced kidney function

Our kidneys have several functions. They filter waste products and extra fluid from our blood and convert them into urine. They also help to control our blood pressure. Having ADPKD can make it harder for your kidneys to do these jobs.

Although ADPKD causes damage to the kidneys, any remaining healthy tissue will continue to work well, usually for decades. People with reduced kidney function usually only get symptoms when their kidney function becomes quite poor. Your kidney specialist will keep a close eye on your kidney function using blood tests and scans.

Kidney failure

A failing kidney can only filter up to a sixth (15%) of the blood that a healthy kidney can in a minute. This is measured as an eGFR of 15 or less ml/min (see earlier for an explanation of eGFR).

Most people with ADPKD find that their kidneys fail between the age of 40 and 70 years old. Before this happens, you and your kidney specialist will plan for you to have a kidney transplant or start dialysis.

Rarer complications

People with ADPKD are more likely to get a swollen small artery in the brain than people without ADPKD. This is called a brain aneurysm. Brain aneurysms may be more likely in people with ADPKD because the gene alterations that cause ADPKD might also affect blood vessels.

There's a risk that these can bleed on the brain, but they can usually be treated before this happens. Treatment reduces the risk of them bleeding.

People with ADPKD who have kidney failure may also develop diverticular disease (where bulges form in the bowel wall) or hernias (where part of the bowel comes through the abdominal muscle).

A failing kidney can only filter up to a sixth (15%) of blood than a healthy kidney can in a minute

How is ADPKD treated?

There is no cure for ADPKD, but treatments can help to reduce your symptoms and lower your risk of complications. For example, different medications can lower your blood pressure, treat infections or help to manage pain.

Some people need to have procedures or surgery, for example to drain and treat a large kidney cyst.

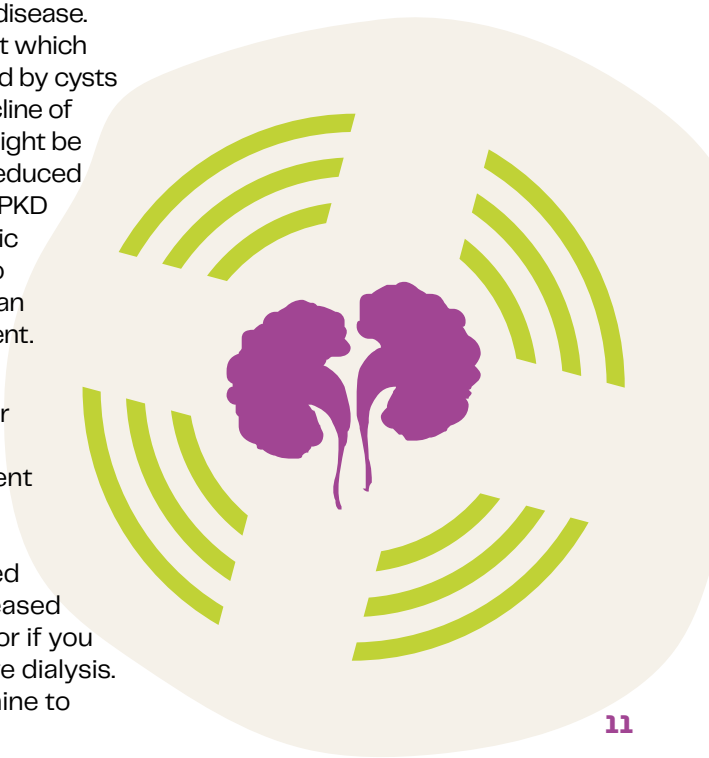
A drug called tolvaptan is suitable for some adults with ADPKD — this can slow the progression of the disease. Tolvaptan reduces the rate at which the kidneys become enlarged by cysts and can help to slow the decline of kidney function. Tolvaptan might be suitable for you if you have reduced kidney function and your ADPKD is progressing rapidly. Genetic testing is sometimes used to help decide whether tolvaptan treatment would help a patient.

If your kidneys fail, you'll need treatment to take over some of their work. This is known as kidney replacement therapy. The best option is usually a kidney transplant, where you receive a donated kidney from a living or deceased donor. If this isn't possible, or if you would prefer, you could have dialysis. Haemodialysis uses a machine to filter your blood, whereas peritoneal dialysis uses a special fluid placed in your abdomen to absorb waste.

A kidney transplant will do about half (50%) the work of a healthy kidney, while dialysis will do about one tenth (10%). Even a 10% increase in function will be enough to improve some of your symptoms.

You can learn more about treatments for ADPKD, including tolvaptan, dialysis and kidney transplants on our website.

www.pkdcharity.org.uk



How will ADPKD affect my life?

Receiving a diagnosis of ADPKD can be a shock but it doesn't mean you need to change your life fundamentally. Most people stay in good health for decades after their diagnosis. They're usually still able to work, have children, go on holiday, keep active and drink alcohol if they wish.

Daily life

Most adults with early ADPKD experience symptoms occasionally, such as back pain and feeling tired. These are often associated with a particular problem (such as an infection) and clear up with treatment. When symptoms occur, they can make it harder to work and keep active.

As your kidney health falls, you may have some restrictions on your diet and other aspects of your life (for example, being advised to avoid rougher sports). Not everybody with ADPKD will experience kidney failure. If your kidneys do fail and you start dialysis, this is likely to impact on your day-to-day life due to the time (and possibly travel) needed for dialysis sessions.

Work

You don't have to tell your employer that you have ADPKD. However, if you explain how your ADPKD affects you, your employer must make changes (known as 'reasonable adjustments') to help you do your job. For more information on how employers should support people with ADPKD, see our factsheet PKD and Employment.

Insurance

Having a family history of ADPKD or a diagnosis of ADPKD could affect the cost of some insurance.

If you or other family members have been diagnosed with ADPKD, you should disclose this if asked. If you withhold the information, this could invalidate your insurance. However, if you (or a family member) have had a genetic test to predict whether you could have ADPKD, the insurance company cannot ask for this information.

Driving

If you drive a car or motorcycle, you don't need to tell the DVLA if you're diagnosed with a kidney problem, such as ADPKD. However, if you drive a bus, coach or lorry, you need to tell the DVLA about your ADPKD.

Rules for related conditions and treatments:

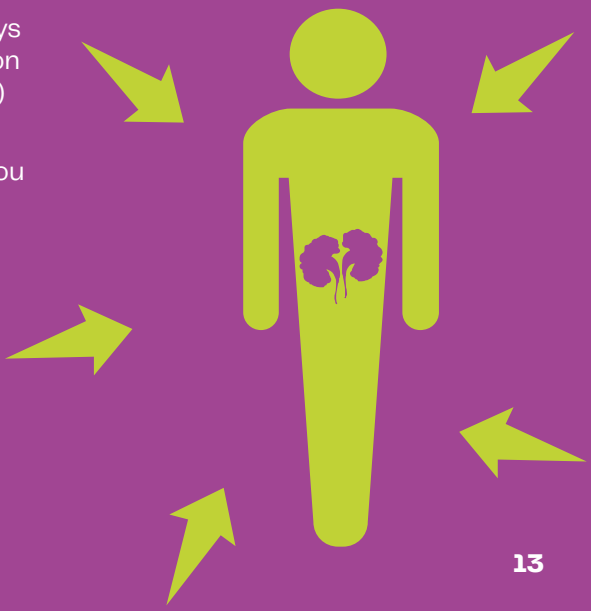
- If you're receiving dialysis, ask your doctor when it's safe to drive after dialysis sessions.
- If you have high blood pressure and you're a bus, coach or lorry driver, you must tell the DVLA.
- Whatever vehicle you drive, you must stop driving if a doctor says you have malignant hypertension (extremely high blood pressure) until it's better controlled.
- If you have a brain aneurysm, you must tell the DVLA.

Ask your doctor for advice and always check with the DVLA for full advice.

Emotions and mental health

People diagnosed with ADPKD often initially feel uncertainty, fear and loss (grief) of the life they expected to have. You may feel anxious or depressed too. Try to acknowledge these feelings and seek support.

The PKD Charity offers a range of support services or you may like to call the PKD Charity Helpline to speak to someone with first-hand experience of PKD. Call **0300 111 1234**; lines are open weekdays 9.30am–5.00pm; or email **info@pkdcharity.org.uk**.



What can I do to help my kidneys?

There are things you can do to help protect your kidneys and stay as healthy as possible:

Avoid smoking

Smoking can increase the speed at which your ADPKD progresses and damages your kidneys. It is also bad for your general health.

Take your blood pressure medication

If you have high blood pressure, it's important to control it. High blood pressure increases the chance of having a heart attack or bleed on the brain (stroke). It can also cause your ADPKD to progress faster. Blood pressure can be treated with lifestyle changes and medications.

Eat healthily

There is no recommended special diet that people with ADPKD should follow, but a balanced diet is important for everyone for controlling weight. Being overweight or obese may lead to a faster decline in your kidney function and can increase your risk of high blood pressure or cardiovascular problems.

Limit how much salt you eat because salt makes high blood pressure worse and may increase the speed that your kidney function declines. Read more about Diet and Lifestyle on our website.

Exercise

Regular exercise is an important part of a healthy lifestyle because it helps to control your weight and blood pressure.

Stay hydrated

There haven't been any studies proving that drinking extra fluid slows the growth of kidney cysts.
For the moment, kidney experts recommend that you drink enough that you don't get thirsty but not excessively.

Use pain medications other than NSAIDs

Non-steroidal anti-inflammatory drugs (NSAIDs), such as ibuprofen, are commonly used for pain and inflammation. However, using them repeatedly or for a long time can damage the kidneys. Long-term use is generally not recommended for people with kidney problems.

Ask your doctor or pharmacist to suggest alternatives that are safer for you.

What is the chance of my child having ADPKD?

Most people with ADPKD have one normal and one altered copy of a PKD gene. Which gene copy your child inherits is down to chance.

If you have ADPKD, there is a one in two (50%) chance you'll pass the altered gene to your child. If they inherit the altered gene, they'll have ADPKD too. This is known as a 'dominant inheritance' (**Figure 3**).

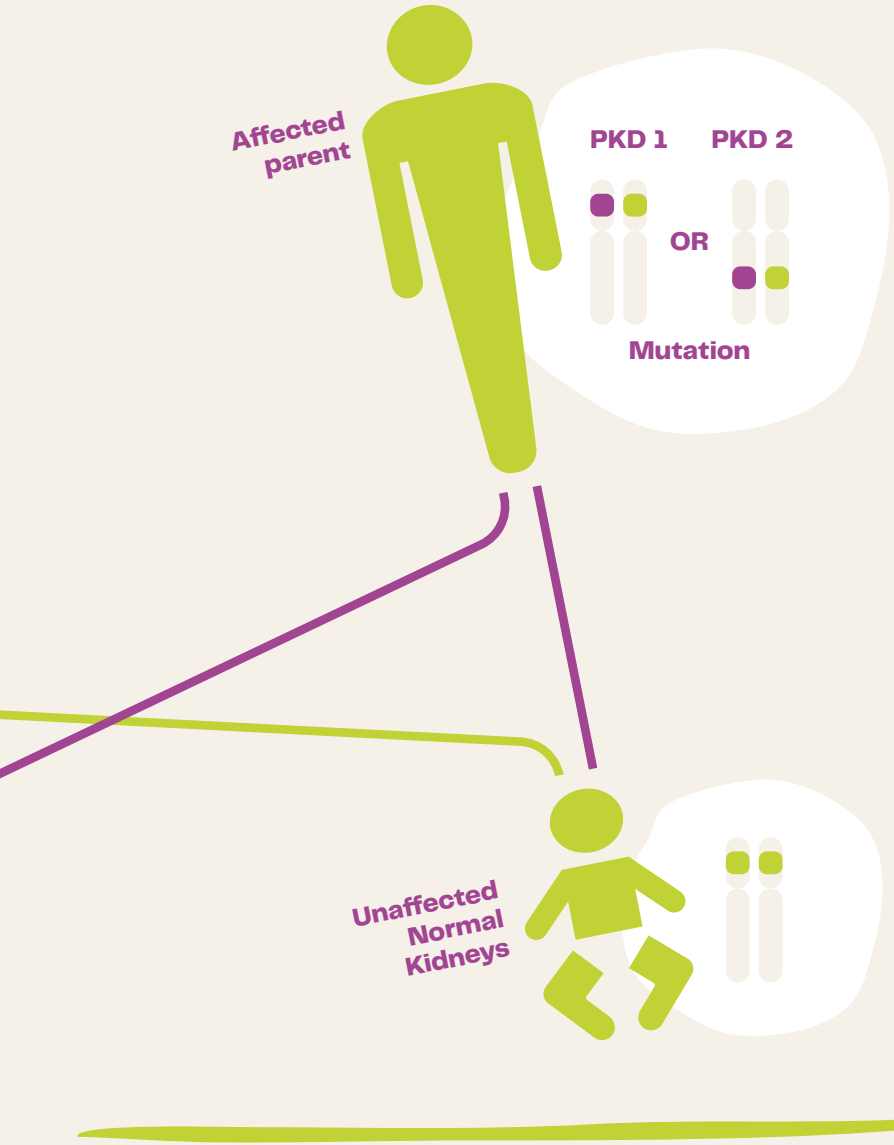


Figure 3: Inheritance of ADPKD. The diagram shows how a PKD gene (either PKD1 or 2) can be passed from a parent to child. The chromosomes that carry the PKD genes are shown. The normal gene in green and the altered gene in purple. The child will inherit one of these genes on a chromosome from the father and one on a chromosome from the mother. The lower half of the diagram shows the different ways that the parents' genes could be passed to their child. In the scenario on the left, the child inherits the altered gene (purple) from the affected parent, and a normal gene from the other parent. This child will have ADPKD. In the scenario on the right, the child inherits a normal gene (green) from each parent and will not have ADPKD.

You can read more about the genes involved in ADPKD and how you can be tested for them in our fact sheet Genetic testing.

Learn more from the PKD Charity

You can find support and further information about ADPKD and living with the disease on the PKD Charity's website. For printed information or to talk to a person with experience of ADPKD, contact our Helpline on **0300 111 1234** (weekdays, 9.30am–5pm, except bank holidays) or email **info@pkdcharity.org.uk**.

Information from other organisations

You might find the following organisations and websites useful:

- Kidney Research UK has information on the latest research into all forms of kidney disease.
- Kidney Care UK provides grants and other financial support to all kidney patients.
- The National Kidney Federation has local kidney patient groups associated with kidney treatment centres around the UK.
- The NHS website has health information on a wide range of topics.
- RaDaR Registry is a UK-wide registry of adults and children with rare kidney diseases including ADPKD. If you're interested to join the registry, please ask your kidney doctor for more information.

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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**.

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, 9.30am–5pm) 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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About PKD Charity

Founded and led by patients, doctors and family members, we're the only UK charity solely dedicated to supporting people affected by PKD.

Since 2000 we've been helping people to live life as fully as possible by providing practical and emotional support, information and advice.

We campaign to raise awareness, improve services, and fund research into discovering better treatments, new treatments, and a cure for PKD.

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Or contact our Support Services Manager at **info@pkdcharity.org.uk** or call/text **07739 632836**

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