Polycystic kidney disease

Polycystic kidney disease (PKD) is a group of genetic diseases where thousands of cysts (fluid filled sacs) grow in the kidneys. If you have PKD, both kidneys are affected, but one may form cysts sooner than the other.

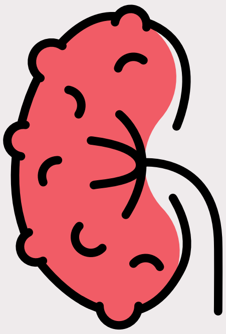
In PKD, the cysts grow slowly and make your kidneys larger. This leaves less healthy kidney tissue to do its job, making it harder for your kidneys to filter your blood. PKD affects males and females in equal numbers, and the cysts can appear at any age, depending on the type of PKD.

PKD is different from simple kidney cysts, which are common in most people as they age and are often harmless. This factsheet covers PKD, an inherited condition that causes many cysts and has risks to your kidney health.

The different types of PKD

PKD is the most common type of inherited kidney disease. Your parents can pass down inherited disease through their genes. Genes are the instructions for your body's traits, such as eye colour.

Sometimes, a gene contains a variation – like a spelling mistake – that stops it working like it is meant to. A variation can be **inherited or happen by chance**, causing a gene to be faulty. These mutations can lead to a wide range of genetic conditions, including PKD.



There are two different types of PKD:

Autosomal Dominant PKD (ADPKD)

This is the most common form of PKD. People with ADPKD can get cysts in their kidneys, liver, pancreas, and other organs.

ADPKD is passed down from the parent. There is a **one in two** chance of passing the faulty gene onto your children. Cysts can often be detected between the ages of 18-35 years. This affects both males and females equally and can sometimes lead to kidney failure later in life.

Autosomal Recessive PKD (ARPKD)

This is a much less common form of PKD. ARPKD is typically a childhood disease diagnosed soon after birth.

Both parents must have the faulty gene, with the risk of a **one in four** chance of passing the gene onto each of their children. ARPKD can lead to kidney failure and/or liver problems later in life.

Symptoms of Autosomal Dominant PKD

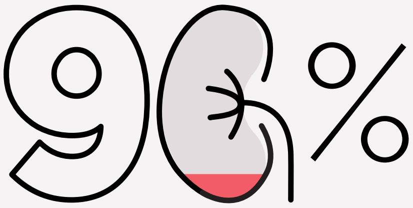
If you have ADPKD, you might not have any symptoms at first. The kidney cysts usually start forming in childhood, but they are microscopic (and almost invisible to detect).

They grow very slowly and it may be many years before they can be seen on an ultrasound. In those who show no symptoms, ADPKD can often be found accidentally while having scans or x-rays for other reasons.

Your doctor may suspect ADPKD if you have signs of chronic kidney disease such as bloody or frothy urine (wee), high blood pressure, and an **abnormal kidney function blood test**. Your doctor may recommend testing if a relative is diagnosed with ADPKD.

Symptoms usually develop when someone is 30-40 years of age but can occur later in some people. You may lose 90% of kidney function before showing symptoms. Symptoms may include:

- **high blood pressure**
- **the need to pass urine more often, sometimes during the night**
- **pain in your abdomen (belly area)**
- **pain in your back or side**
- **blood in your urine (haematuria)**



of kidney function can be lost without experiencing any symptoms

If you notice these symptoms, be sure to talk to your doctor!

ADPKD may get worse over time. About half the people with PKD will develop kidney failure by age 60. Possible complications include:

- **kidneys that do not work as well**
- **urinary tract infections**
- **kidney stones**
- **bulges in your stomach area (called hernias)**
- **heart valve problems**
- **cysts in other organs, most commonly in the liver**
- **kidney failure**
- **brain bleeds due to weakened blood vessels in the brain**

If you experience a sudden, intense headache, call 0-0-0 or seek emergency services. This could be a sign of a serious complication of ADPKD causing a brain bleed.

Symptoms of Autosomal Recessive PKD (ARPKD)

If you have ARPKD the cysts can develop in the early months of life or even before birth. Children with ARPKD may have poorly working kidneys, which may lead to kidney failure or liver problems.

Signs of ARPKD:

- **reduced fluid surrounding the baby during pregnancy which can lead to the baby having an unusually shaped face**
- **delayed or difficult childbirth**
- **high blood pressure**
- **swelling of the abdomen (belly area) due to enlarged kidneys, liver and spleen**
- **heart and lung problems**
- **kidney failure at birth or in the first few weeks of life**

Other problems may include:

- **a lower birth weight**
- **increased blood pressure in the liver**
- **blood in the urine**
- **high blood pressure**
- **anaemia (low red blood cells)**

Sometimes, ARPKD can be found before a baby is born during a routine ultrasound.

How is PKD diagnosed?

ARPKD is typically diagnosed early because symptoms are often noticeable at birth. In less severe PKD, health problems may not be seen for many years.

PKD may be discovered during medical checks for other problems, such as urinary tract infections or kidney stones. Sometimes it is not found until high blood pressure or kidney failure develops.

Your doctor considers **age**, a **positive family history** of PKD, and **ultrasound** results when diagnosing PKD. Other tests to diagnose PKD involve:

Physical examination - used to look for high blood pressure or enlarged kidneys.

Urine tests - used to look for blood and/or protein in your urine.

Blood tests - used to measure your kidney function.

Computed Tomography (CT) or Magnetic Resonance Imaging (MRI) - used to help your doctor see an image of your kidneys. These imaging tests are used when the ultrasound does not provide enough information.

PKD will be diagnosed in at-risk people with a family history of PKD if:

Age	Number of cysts shown on ultrasound
15-39 years	At least 3 in total
Aged 40-59 years	At least 2 in each kidney
Aged 60 years or older	At least 4 in each kidney

Should my family members be tested for PKD?

Knowing whether to screen family members is a complex and personal issue. PKD can differ in severity, even between family members. If it's unclear if you or your family members has PKD, your doctor may offer genetic testing.

Genetic testing involves a blood test that **identifies changes in your genes.** Pre-screening counselling is available to provide further information about genetic testing and support to help you decide about testing of PKD.

A genetic counsellor will be able to talk to you about the pros and cons of screening and help you to make the best decision for you and your family.

Genetic testing gives you and your family members early answers and peace of mind.

Planning for a family when you have PKD

If you are planning to start a family, speak to your nephrologist about the risks and benefits of pregnancy in PKD. Your medicines used to treat PKD may change if you are trying to conceive. You will need to closely monitor your blood pressure if you have PKD and are pregnant.

How is PKD treated?

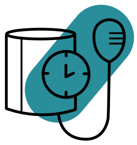
While there is no cure for PKD, lifestyle changes, good blood pressure control, and medicines can help.

Lifestyle changes together with good blood pressure control have been shown to slow the growth of kidney cysts. For many people this may be the only treatment they require.

A new medicine, called Tolvaptan, is available for the treatment of adults with stage 1-3 kidney disease and certain types of ADPKD. This medicine has been shown to slow down cyst growth and delay kidney failure. Your nephrologist will tell you if this medicine is right for you.

Managing the symptoms of PKD

Achieving and maintaining a healthy weight and blood pressure is extremely important in the treatment and management of PKD.

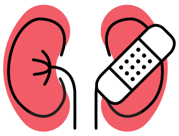


High blood pressure - good blood pressure control protects kidney function and can assist in slowing the growth of kidney cysts.

→ Blood pressure medicines such as ACE inhibitors (medicines ending in -PRIL) or ARBs (medicines ending in -SARTAN) can help to reduce blood pressure and prevent worsening of kidney disease. Following a healthy lifestyle, limiting salt in your diet, and getting enough exercise can also help to lower blood pressures.

Pain - may be due to stones, bleeding, or infection. Treatment will depend on the cause. Talk to your doctor if you are getting repeated or severe back and kidney pain or headaches. Your doctor may drain the cysts to relieve pain.

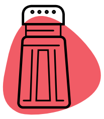
Urinary tract infections (UTIs) - UTIs can spread to the cysts in the kidneys, so see your doctor immediately if you notice signs of infection such as foul-smelling urine or painful urination. Your doctor will prescribe you antibiotics to treat your UTI. See our Urinary Tract Infections factsheet to learn more.



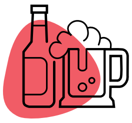
Chronic kidney disease (CKD) - if your kidney disease progresses to kidney failure, you will require dialysis or kidney transplantation. Medicines and a healthy lifestyle can help with managing the symptoms of CKD.

Healthy habits to support your kidneys

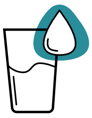
In addition to following up with your doctor and taking any prescribed medicines, you can follow these healthy lifestyle choices to manage your kidney disease:



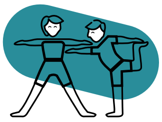
Reduce salt in your diet. Instead of salt, use herbs and spices to flavour your food.



Limit alcohol. The less you drink alcohol, the better.

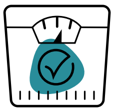


Drink enough water. Aim for more than two litres a day, especially if you get kidney stones.



Exercise regularly. Aim for 2.5 - 5 hours of moderate intensity exercise per week.

Stop smoking or vaping. If you need tips to help you quit, call the Quitline on 13 7848 or speak to your doctor.



Maintain a healthy weight. Exercise and a healthy diet can help.

New treatments

There are several promising developments in preventing cyst growth and slowing the decline in your kidney function. In Australia, scientists are exploring medicines may stop the sacs from filling up with fluid.

Results from worldwide studies have shown promising results for new medicines and treatments. To learn more about the studies happening for PKD, check out **pkdaustralia.org/clinical-trials**.

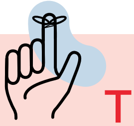
While there is no specific PKD diet, you may be instructed to reduce dietary salt, protein, cholesterol (fats), alcohol, and caffeine. Discuss any dietary changes with your doctor or Accredited Practising Dietitian.

What if I am having trouble coping?

Receiving a diagnosis of PKD can feel overwhelming. You may feel down, depressed, or hopeless. Know that you are not alone. Speak to your doctor, a family member, or a social worker if you are having trouble coping.

Kidney Health Australia operates a free call kidney helpline for people seeking more information about kidney disease and related conditions. You can contact the helpline on **1800 454 363**.

The **PKD Foundation of Australia** connects, supports, and provides education for Australians and their families affected by PKD.
The PKD Foundation also raises funds specifically to find a cure for PKD.
Visit **pkdaustralia.org** for more information.



Things to remember:

✓ Polycystic kidney disease (PKD) is an **inherited condition** which leads to the growth of cysts on the kidneys.

✓ You may have **no symptoms or health problems** for many years.

✓ The treatments for PKD include **good blood pressure control,** new medicines to slow down cysts, and a healthy lifestyle.

What does that word mean?

Accredited Practising Dietitian (ADP) – A person who is professionally qualified to give practical diet and nutrition advice.

Anaemia – When there are only a small number of red blood cells in the blood or the blood cells are not working properly. Red blood cells carry oxygen, so if you have anaemia, you can feel weak, tired and short of breath.

Chromosomes – The structures that carry your DNA. We usually have 23 pairs.

Computed tomography (CT) – An imaging procedure that uses special x-ray equipment to create a series of detailed pictures or scans of areas inside your body.

Hernias – When parts of tissue or organ exit the weak parts of the abdomen.

Genetic counsellor – Someone trained to help people understand and make decisions around the medical, psychological and reproductive effects of genetic conditions. See The Australian Society of Genetic Counsellors hgsa.org.au/asgc for more information.

Inherited – Passed to you by your parents. This can include personality traits, physical appearance and some health conditions.

Magnetic resonance imaging (MRI) – An imaging procedure that uses a magnetic field and radio wave energy to make detailed pictures or scans of areas inside your body.

Ultrasound – An imaging procedure where an instrument is moved over the skin, sending and receiving signals to make pictures of your kidneys and bladder.

Urinary tract infection – Causes symptoms like needing to urinate frequently or pain when urinating. It is caused by bacteria and may need to be treated.

Kidney Health Australia

Free Kidney Helpline 1800 454 363
kidney.org.au

If you have a hearing or speech impairment, contact the National Relay Service on 1800 555 677 or relayservice.com.au Have them connect you to the Free Kidney Helpline - 1800 454 363

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