Kidney Health Australia
Prevent, Detect, Support.

Fact sheet

Polycystic Kidney Disease

Polycystic kidney disease (PKD) is group of chronic kidney diseases where thousands of cysts (fluid filled sacs) grow in the kidneys. PKD is the most common inherited kidney disease and is a common cause of Chronic Kidney Disease.

If you have PKD both of your kidneys will be affected but one kidney may develop the cysts earlier than the other. The cysts gradually grow which makes your kidneys larger and reduces the healthy kidney tissue. This makes it harder for your kidneys to work properly.

Some people develop high blood pressure and end stage kidney disease as a result of PKD. PKD affects males and females in equal numbers, and the cysts can appear at any age, depending on the type of PKD.

It is not uncommon for people to develop simple kidney cysts as they become older. Around 50% of people over the age of 50 develop simple cysts. These cysts are not inherited and do not usually require treatment. This fact sheet will discuss PKD which is the inherited type of cyst disease.

The different types of PKD

PKD is an inherited disease. This means it is passed from parents to their children.

However not all people with PKD will have a family history.

There are two different types of PKD:

Autosomal Dominant PKD (ADPKD)
This is the most common form of PKD. People with ADPKD will develop multiple fluid filled cysts in their kidneys, liver, pancreas and other organs. ADPKD is inherited from your parent. There is **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. This is typically a childhood disease which is often diagnosed soon after birth. Both parents must have the faulty gene, with the risk of **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 may have no symptoms in the early stages. The kidney cysts usually start forming in childhood but they are microscopic (and almost invisible to detect) and they grow very slowly so its takes many years before they can be detected by renal ultrasound.

In those who show no symptoms, ADPKD can often be found accidentally while having scans or x-rays for other reasons, through screening those with a positive family history or having early signs of chronic kidney disease such as abnormal urine, high blood pressure or an abnormal kidney function blood test.

Symptoms usually develop when someone is 30-40 years of age, but can be later in some people.

Symptoms may include:

- the need to pass urine more often (polyuria), sometimes during the night (nocturia)
- pain in your back or side
- blood in your urine (haematuria)
- high blood pressure
- reduced kidney function or kidney failure – about half of people with PKD will have kidney failure by 60 years of age, but can be considerably later in some people
- enlarged and painful abdomen (belly area)
- urinary tract infections
- kidney stones
- hernias
- cysts in other organs, most commonly in the liver
- abnormal heart valves
- intracranial aneurysm (weakened spot in the blood vessels of the brain)

Symptoms of Autosomal Recessive PKD

If you have ARPKD the cysts can develop in the early months of life or even before birth. Children with ARPKD may have reduced kidney function, which may lead to end stage kidney disease, or liver problems.

Symptoms and signs of ARPKD in severely affected babies include:

- reduced fluid surrounding the baby in the uterus 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 defects
- kidney failure at birth or in the first few weeks of life

Other complications may include:

- failure to thrive – below average growth and weight gain as a baby
- increased blood pressure in the liver
- blood in the urine
- high blood pressure
- anaemia - low red blood cells

How is PKD diagnosed?

The severe symptoms of ARPKD usually means the condition is diagnosed early. But in less severe PKD, health problems may not be seen for many years. It is often found during medical checks for other problems, such as urinary tract infections or kidney stones. Sometimes it isn't found until high blood pressure or kidney failure develops.

Diagnosing PKD usually considers **age**, having a **positive family history** of PKD combined with an **ultrasound**. To be diagnosed with PKD, there must be a certain number of cysts shown on the ultrasound. Further testing may then be performed, including:

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

Blood tests - used to measure your kidney function.

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

Ultrasound

Blood test

Urine Test

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

How is PKD treated?

Initial treatment for PKD is usually through lifestyle management and modification. 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.

There are several trials being conducted for new medications for the treatment for PKD, these medications may slow the progression of PKD but currently there is no cure for PKD.

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

Diet - Changes to your diet may be required, including reducing dietary salt, protein, cholesterol (fats), alcohol and caffeine. Any dietary changes should be discussed with your doctor or Accredited Practising Dietitian.

See our fact sheets on self-management and nutrition for more information about how to look after your kidneys.

Lifestyle - Undertake physical activity, find something that you enjoy. Not only will regular exercise help you reach and maintain a healthy weight, but it will reduce the risk of other complications.

Smoking - If you smoke, stop. This is an important habit to change to slow the progression of PKD and associated complications. If you need tips to help you quit, call the Quitline on 13 7848 or speak to your doctor.

High blood pressure - Good blood pressure control protects kidney function and can assist in slowing the growth of renal cysts. Medications and a healthy lifestyle can help to lower blood pressure.

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. Cysts can sometimes be drained to relieve extreme back and leg pain.

Blood in the urine - May also be due to kidney stones, bleeding or infection Treatment will depend on the cause. Fluids, pain killers, antibiotics and rest may be needed.

Urinary tract infections - Urinary tract infections can spread to the cysts in the kidneys, so see your doctor immediately about antibiotic treatment.

End Stage Kidney Disease - This usually develops very slowly over many years and may require dialysis or a transplant. PKD does not redevelop in a transplanted kidney.

Medication - In some cases of ADPKD, an oral medication is now available to be used to slow the progression of cyst development and kidney disease in adults.

Support - Many patients diagnosed or living with PKD can experience some feelings of sadness and anxiety due to the diagnosis. There is support available to help patients along their journey and to help them be informed about their disease. Our Kidney Helpline can provide further guidance. Freecall 1800 454 363.

What can I do to prevent PKD complications?

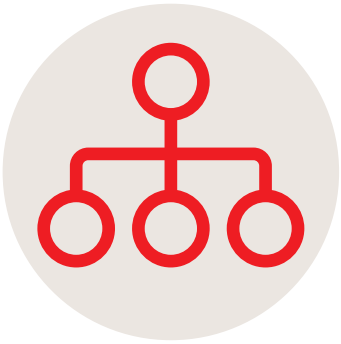
If your kidneys, liver, spleen and abdomen are significantly enlarged, you may need to think about not playing contact sports, as an injury to the belly area can damage your affected organs. All contact sports should be discussed with your doctor.

Non-steroidal anti-inflammatory medications (NSAIDs) should not be taken without medical advice as they can make your kidney function worse.

New treatments

There are several promising developments in slowing or preventing cyst growth and decline in eGFR in PKD. Trials are being done in Australia with medications that may stop the sacs filling up with fluid. Results from worldwide studies have showed promising results for medications which slow or stop cyst growth and slow the drop of kidney function.

A new medication has recently been approved and PBS listed for the treatment of adults with early stage CKD (stage 2 to 3) and rapidly progressing ADPKD. It has been shown to slow the progression of cyst development and kidney disease in ADPKD. Your kidney specialist can keep you updated on new treatments and whether they are right for you.



Should my family members be tested for PKD?

Knowing whether to screen family members is a **complex and personal issue**. If ultrasound findings are unclear and it is necessary to completely rule out PKD, then genetic testing may be offered.

Genetic testing involves a medical test that identifies changes in chromosomes, genes and proteins. Pre-screening counselling is available to provide further information about genetic testing and support to help you make a decision 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.

There are both advantages and disadvantages to screening family members, some of the issues that you may want to consider are:

Advantages	Disadvantages
• PKD can be detected early; • Early detection can lead to better treatment options of PKD; • Early detection can slow the decline of kidney function; • PKD can be considered in family planning.	• A positive diagnosis may cause feelings of sadness, frustration and stress; • A positive diagnosis may present challenges with insurance and employment.

Who should I contact for more information?

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.

Further information can be found on their website **pkdaustralia.org**

Kidney Health Australia – Caring for Australasians with Renal Impairment (KHA-CARI) provides clinical guidelines based on best evidence based practice for patients with kidney disease in Australia and New Zealand. Further information can be found on the website **cari.org.au**

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.
- At the moment there is no cure for PKD, but early detection and treatment can reduce or prevent some complications of PKD.

What does that word mean?

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.

Blood pressure - The pressure of the blood in the arteries as it is pumped around the body by the heart.

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

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

Cyst - A sack full of fluid.

eGFR - An estimation of glomerular filtration rate (GFR). GFR is the best measure of kidney function and helps to determine the stage of kidney disease.

Gene - Each cell in the human body contains about 25,000 to 35,000 genes. Genes carry the information that determines your traits, which are features or characteristics that are passed on to you or inherited from your parents.

Haematuria - The medical term for blood in your urine.

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.

Non-steroidal anti-inflammatory drugs (NSAIDs) - Medications often used to reduce pain and inflammation (swelling and redness). Some commonly used NSAIDs include aspirin (in brands like Disprin), ibuprofen (such as Nurofen), naproxen (such as Naprosyn), diclofenac (such as Voltaren) and celecoxib (such as Celebrex).

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.

For more information about kidney or urinary health, please contact our free call Kidney Helpline on 1800 454 363.

Or visit our website **kidney.org.au** to access free health literature.

This factsheet is intended as a general introduction to this topic and is not meant to substitute for your doctor's or health professional's advice.

All care is taken to ensure that the information is relevant to the reader and applicable to each state in Australia. It should be noted that Kidney Health Australia recognises that each person's experience is individual and that variations do occur in treatment and management due to personal circumstances, the health professional and the state one lives in. Should you require further information always consult your doctor or health professional.

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If you have a **hearing or speech impairment,** contact the National Relay Service on **1800 555 677** or **relayservice.com.au**

For all types of services ask for 1800 454 363