

## Kidneys - polycystic kidney disease

Polycystic kidney disease (PKD) is usually an inherited condition. It belongs to a group of diseases known as 'cystic kidney disease'. Faulty genes cause abnormal blisters of fluid (cysts) to grow in the kidneys.

Both kidneys are usually affected but one may develop the cysts earlier than the other. The cysts continue to grow until they compress the healthy tissue and stop the kidneys from working properly. The kidneys get larger along with the cysts, which can number in the thousands.

Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds. Men usually progress faster to kidney disease, although it is unclear why this occurs. There is currently no cure but the disease can be managed and research into treatment options is ongoing.

The two major inherited forms of polycystic kidney disease are:

- Autosomal dominant PKD
- Autosomal recessive PKD.

### Autosomal dominant PKD

This is the most common inherited form of polycystic kidney disease. A parent with autosomal dominant PKD has a 50 per cent chance of passing the faulty gene and associated disease to each of their children. If a person doesn't inherit the gene, there is no chance of their children inheriting the gene because it never 'skips' a generation.

Occasionally, a person develops the disease when there is no family history. It is thought that a different inheritance pattern or perhaps a genetic mutation may be responsible. Like inherited PKD, the affected person has a 50 per cent chance of passing the faulty gene and associated disease to each of their children. Autosomal dominant PKD can lead to kidney failure.

### Symptoms of autosomal dominant PKD

There may be no symptoms in the early stages. The cysts usually start growing during the teenage years. As the cysts replace healthy tissue, the outline of the kidneys looks irregular or 'moth-eaten'.

Symptoms and signs usually develop between the ages of 30 and 40 (but can begin earlier), and may include:

- High blood pressure (may occur before cysts appear)
- Pain in the back or sides
- Headaches
- Enlarged and painful abdomen
- Blood in the urine (haematuria)
- Urinary tract infections
- Kidney stones
- Liver and pancreatic cysts
- Abnormal heart valves
- Aneurysms in the brain
- Diverticulosis (the development of abnormal pouches in the walls of the large intestine)
- Abdominal wall hernias.

### Autosomal recessive PKD

This is a less common inherited form of polycystic kidney disease. Signs begin in the early months of life or even while the baby is still developing in the uterus (womb). Autosomal recessive PKD is sometimes called 'infantile PKD'. Children born with autosomal recessive PKD often develop kidney failure within a few years of birth and experience liver problems as they grow into adults.

## Symptoms of autosomal recessive PKD

Symptoms and signs in severely affected babies can include:

- Reduced amniotic fluid surrounding the baby in the uterus
- An unusual shape to the face due to the lack of amniotic fluid (Potter facies)
- Enlargement of the child's abdomen due to enlarged kidneys, liver or spleen
- Heart defects
- Underdeveloped lungs
- Kidney failure at birth or in the first few weeks of life.

## Diagnosis of polycystic kidney disease

The severe symptoms of autosomal recessive PKD usually result in a prompt diagnosis. However, in most cases of autosomal dominant PKD, a person's physical condition can appear normal for many years. Physical check-ups or blood and urine tests may not always identify the disease. It is often detected during medical investigations for other health problems, such as urinary tract infections. At other times, the disease isn't discovered until the kidneys fail.

Diagnosis of PKD may involve a number of tests including:

- **Physical examination** – can detect symptoms such as high blood pressure or enlarged kidneys.
- **Blood tests** – to assess kidney function.
- **Urinalysis** – blood or protein (or both) may be found in the urine.
- **Ultrasound** – a simple, non-invasive test that can identify even quite small cysts.
- **Computed tomography (CT) and magnetic resonance (MRI) scans** – these may be required if the results from the ultrasound are inconclusive or if more information is needed. These techniques can detect very small cysts.
- **Genetic testing** – this is not a routine test but may be used for family testing. The presence of the abnormal genetic material can be detected with special blood tests. Genetic counselling is available for affected couples.

## Treatment of polycystic kidney disease

There is no cure for PKD. However, regular monitoring of the kidneys and treatment for the associated complications can help to maintain health and prolong a person's lifespan.

Common complications and their treatments include:

- **Trauma** – consider avoiding contact sports if your kidneys, liver, spleen or abdomen are enlarged. A strong blow to the belly could injure affected organs.
- **High blood pressure** – controlling high blood pressure is very important. Antihypertensive medications may be prescribed.
- **Blood in the urine** – fluids, analgesics, antibiotics and bed rest may be recommended.
- **Urinary tract infections** – symptoms may include frequency to urinate, painful urination and fever. Consult with your doctor immediately about treatment with antibiotics. An untreated urinary tract infection can spread to the kidneys.
- **Kidney failure** – this is treated by dialysis, which is a procedure to remove waste products and extra water from the body by filtering the blood through a special membrane. A kidney transplant is another treatment option. PKD does not redevelop in the transplanted kidney.
- **Liver cysts** – these do not usually affect liver function. Non-surgical management may include avoiding hormone replacement therapy (HRT), also known as hormone therapy (HT). Surgery may occasionally be needed to drain cysts or remove diseased parts of the liver. Rarely, a liver transplant is needed.

Clinical trials have begun in Australia to test medication that alters the production of fluid by the kidney and appears to slow down cyst formation.

## Self-care suggestions for polycystic kidney disease

Be guided by your doctor but self-care suggestions may generally include:

- **Changing your diet** – this may help to manage some symptoms. Dietary changes may include reducing salt, protein, cholesterol (fats) and caffeine. Dietary changes should be made only after discussion with your doctor or dietitian and will depend on your test results.
- **Making healthier lifestyle choices** – for example, participate in regular and moderate physical activity and maintain an appropriate weight for your height and build. Not smoking is strongly advised.
- **Avoiding non-steroidal anti-inflammatory medications (NSAIDs)** – these should not be taken without medical advice as they can worsen kidney function.

Your doctor will give you detailed instructions on how to best take care of yourself. Follow these instructions carefully.

## Where to get help

- Your doctor
- Kidney Health Australia Information Line Tel. 1800 4 KIDNEY (543 639), TTY users phone 1800 555 677 then ask for 1800 454 363
- The National Renal Resource Centre Tel. (02) 9362 3995 or 1800 257 189
- The Polycystic Kidney Disease Association – based at the National Renal Resource Centre.

## Things to remember

- Polycystic kidney disease (PKD) is an inherited condition characterised by the growth of cysts on the kidneys.
- The disease may have no symptoms until it is well advanced.
- There is currently no cure for PKD but treatment can reduce or prevent complications and prolong the person's lifespan.

**This page has been produced in consultation with, and approved by:**

Kidney Health Australia

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