Kidneys - polycystic kidney disease (PKD)

**Summary**

- 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 early detection and treatment can reduce or prevent some complications.

Polycystic kidney disease (PKD) is usually an inherited condition. It belongs to a group of diseases known as 'cystic kidney disease'. Changes in the PKD1, PKD2 or PKHD1 genes cause blisters of fluid (cysts) to grow in the kidneys.

Both kidneys are usually affected, but one may develop cysts earlier than the other. The cysts continue to grow until they compress the healthy tissue and stop the kidneys from working properly.

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**

Autosomal dominant PKD (ADPKD) 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 altered gene (PKD1 or PKD2) and associated condition 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 condition when there is no family history. It is thought that a different inheritance pattern or perhaps a genetic change may be responsible. Like inherited PKD, the affected person has a 50 per cent chance of passing the altered 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 of autosomal dominant PKD 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 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
Autosomal recessive PKD

Autosomal recessive PKD 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's 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 (ADPKD) usually result in a prompt diagnosis. However, in most cases of autosomal dominant PKD, for many years there are no signs that a person has the condition.

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 begin to 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
- urine tests – blood or protein (or both) may be found in the urine
- ultrasound – a simple, non-invasive test that can identify even quite 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.

Genetic counselling for polycystic kidney disease

If you or a family member have been diagnosed with PKD, or if PKD runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand PKD and what causes it, how it is inherited, and what a diagnosis means for your health, lifestyle, and plans for the future. Genetic counsellors are trained provide information and support that is sensitive to your family circumstances, culture and beliefs.

If PKD runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family, to find out your risk of passing that condition on to your child, or to arrange for prenatal tests.

The Genetic Support Network of Victoria (GSNV) is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by MCKD.

Treatment of polycystic kidney disease
Currently there is no cure for PKD. However, early detection and treatment can reduce or prevent some of the complications of PKD.

Common complications and their treatments include:

- high blood pressure – controlling high blood pressure is very important. Antihypertensive medication may be prescribed
- pain – may be due to kidney 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 – fluids, pain-relieving medication, antibiotics and bed rest may be recommended
- urinary tract infections – symptoms may include frequent urination, 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.

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.

From January 2019 a new medication, tolvaptan, was approved and listed on the Pharmaceutical Benefits Scheme (PBS) for the treatment of adults with early stage PKD (stage 2 to 3) and rapidly progressing ADPKD. Tolvaptan 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.

Self-care for polycystic kidney disease

Your doctor or healthcare professional will discuss best healthcare choices in managing this condition. Be guided by your doctor. Self-care suggestions for PKD generally include:

- changing your diet – this may help to manage some symptoms. Dietary changes may include reducing salt, protein, cholesterol (fats) and caffeine. Only make dietary changes after discussion with your doctor or dietitian. Dietary recommendations 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. It is strongly advised that you do not smoke
- avoiding non-steroidal anti-inflammatory drugs (NSAIDs) – do not take NSAIDs without medical advice as they can worsen kidney function
- considering avoiding contact sports if your kidneys, liver, spleen or abdomen are enlarged. A strong blow to the belly could injure affected organs.

Where to get help

- Your GP (doctor)
- Kidney Health Australia Information Line Tel. 1800 454 363
- Genetic Support Network of Victoria (GSNV) Tel. (03) 8341 6315
- Victorian Clinical Genetics Services Tel. 1300 118 247

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