Polycystic kidney disease (PKD) is the most common inherited cystic kidney disease. It leads to the thousands of cysts (fluid filled sacs) growing in your kidneys. Polycystic means ‘many cysts’. 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 kidney failure as a result of PKD. PKD affects males and females in equal numbers, and the cysts can appear at any age.

**The different types of PKD**

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

The two types of PKD are:

**Autosomal Dominant PKD** - This is the most common inherited form of PKD. If you have autosomal dominant PKD you have a one in two chance of passing the faulty gene and associated disease to each of your children.

**Autosomal Recessive PKD** - This is a less common form of inherited PKD. If you have autosomal recessive PKD your children may inherit PKD only if your partner also has the faulty gene.

It is possible to develop PKD when there is no family history. It is thought that a different inheritance or a genetic mutation may be responsible.
Symptoms of Autosomal Dominant PKD

If you have autosomal dominant PKD you may have no symptoms in the early stages. The cysts can start growing when you are a teenager. Complications frequently develop at 30-40 years of age, but can be later in some people:
- pain in your back or side
- blood in your urine (haematuria)
- high blood pressure
- enlarged and painful abdomen (belly area)
- urinary tract infections
- kidney stones
- hernias
- cysts in other organs such as your liver, pancreas, intestine or brain
- abnormal heart valves
- 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

Symptoms of Autosomal Recessive PKD

If you have autosomal recessive PKD the cysts can develop in the early months of life or even before birth. Children with autosomal recessive PKD may have reduced kidney function, which may lead to kidney failure, or liver problems.

Symptoms and signs of autosomal recessive PKD 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

How is PKD diagnosed?

The severe symptoms of autosomal recessive PKD 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 stones or infections. Sometimes it isn't found until high blood pressure or kidney failure develops.

Diagnosing PKD can require a number of tests 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** - an imaging procedure which uses sound waves to detect cysts in your kidneys.
PKD will be diagnosed in at-risk people with a family history of PKD if:

<table>
<thead>
<tr>
<th>Age</th>
<th>Number of cysts shown on ultrasound</th>
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<tbody>
<tr>
<td>15-39 years</td>
<td>At least 3 in total</td>
</tr>
<tr>
<td>Aged 40-59 years</td>
<td>At least 2 in each kidney</td>
</tr>
<tr>
<td>Aged 60 years or older</td>
<td>At least 4 in each kidney</td>
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Computed Tomography (CT) and Magnetic Resonance Imaging (MRI) scans - an imaging procedure which can detect very small cysts in your kidneys. These scans may be needed if more information is needed after an ultrasound.

Genetic testing - a type of medical test that identifies changes in chromosomes, genes or proteins. If ultrasound findings are unclear, and it is necessary to completely rule out PKD, then genetic testing may be offered. A genetic counsellor can help with information about genetic testing.

**How is PKD treated?**

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

Some of the common complications of PKD, and recommended treatments are outlined here.

**High blood pressure** - Good blood pressure control may protect kidney function. 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.

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

**What else 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.

Changes to your diet may also help to treat PKD. These may include reducing salt, protein, cholesterol (fats) and caffeine. Any dietary changes should be made after discussion with your doctor or Accredited Practicing Dietitian.

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

**New developments**

There are several promising developments in slowing or preventing cyst growth 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. These medications are expensive and have side-effects, and they are not yet available in Australia.
Who should I contact for more information?

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.

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.

**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.

**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.

**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.

This 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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For more information about kidney or urinary health, please contact our free call Kidney Health Information Service (KHIS) on 1800 454 363.

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

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.