Polycystic Kidney Disease Information

Prevention Support Research

Kidney Health New Zealand
www.kidneys.co.nz
0800 KIDNEY 0800-543639

What is polycystic kidney disease (PKD)?

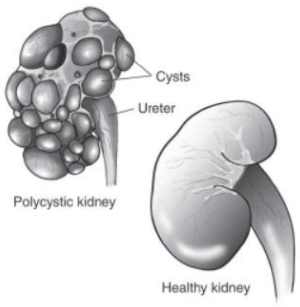
Polycystic kidney disease (PKD) is a condition that affects the kidneys. When people have PKD, abnormal fluid-filled sacs called "cysts" grow in the kidneys.

The cysts cause the kidneys to get bigger than normal. The cysts can also keep the kidneys from working normally. This can lead to problems, such as high blood pressure, kidney infections, and reduced kidney performance, leading to kidney failure. Kidney failure is when the kidneys are working so poorly that you need dialysis or a kidney transplant to continue living. Besides kidney problems, PKD can cause problems in other parts of the body.

What causes PKD:

The development of cysts happens because of a genetic mutation in your DNA .Sometimes the cyst may be present in other organs like your liver, pancreas, spleen, ovaries, and large bowel. Cysts in these organs do not usually cause serious problems. People of all genders, ages, races, ethnicities, and nationalities can have PKD.

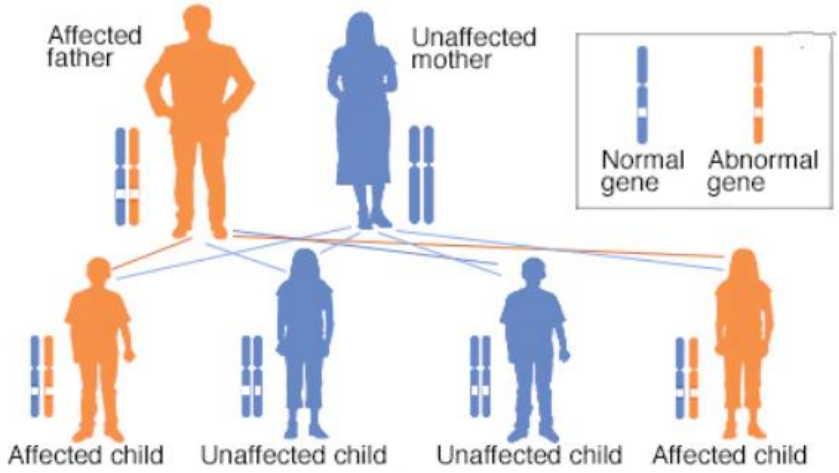
If you have PKD both of your kidneys will be affected but one kidney may develop the cysts earlier than the other. Over time, kidneys become larger and change their shape. When the kidneys become very large, they can press on other organs causing discomfort and a feeling of abdominal bloating.



There are 2 different types of PKD:

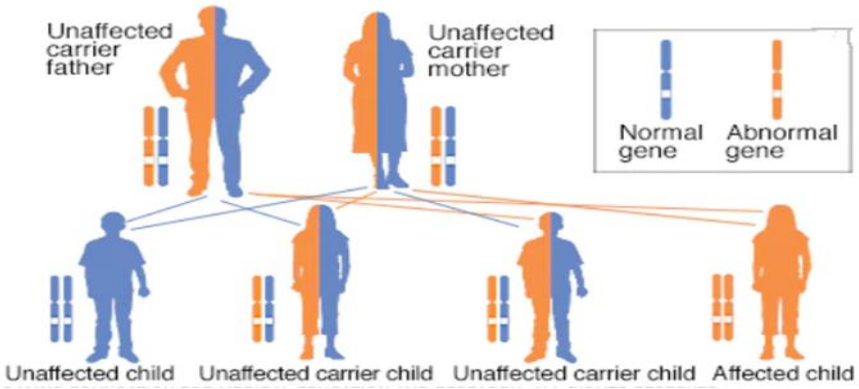
Autosomal **D**ominant **P**olycystic **K**idney **D**isease **(ADPKD)** is one of the most common genetic diseases. is caused by two well-described genetic mutations: PKD1 on chromosome 16 and PKD2 on chromosome 4. Among all PKD cases, PKD1 mutations are more common (71 -85 %) than PKD2 mutations (15 %). However, in approximately 6.8 percent of patients, it is not possible to determine which gene mutation is causing the disease. PKD2 disease is milder; therefore, it often presents later in life and is sometimes not diagnosed at all.

In ADPKD, fluid-filled cysts develop and enlarge in both kidneys, eventually leading to kidney failure. Unlike some genetic diseases, ADPKD does not skip a generation meaning it often affects many people in one family. This affects both males and females. PKD varies greatly in its severity signs and symptoms often develop between the ages of 30 and 40. Each of the children of an affected parent will have a 1 in 2 (50%) chance of inheriting the gene for ADPKD.



Autosomal **R**ecessive **P**olycystic **K**idney **D**isease **(ARPKD)**

This is a much less common form of PKD. This is typically a childhood disease and **t**he signs and symptoms often appear shortly after birth. It affects boys and girls equally. Sometimes, symptoms do not appear until later in childhood or during adolescence Both parents must have the faulty gene. No family history except in siblings sometimes. ARPKD is different disease process causing early liver failure as well as kidney failure. For more information: watch a video: https://pkdcure.org/what-is-arpkd/



"Note: working with your Doctor
In addition to your general practitioner (GP), you should also see a doctor who specializes in kidneys. A nephrologist (kidney specialist) will be able to advise you best on how to care for your polycystic kidneys and the other related symptoms."

How will I know if I have PKD and what does it mean?

There are usually few or no symptoms early in the disease. Sometimes PKD gets detected by accident. As it progresses, you may experience:

- High blood pressure
  Is a common feature of ADPKD, occurring in 60 to 70 percent of patients with normal kidney function by the age of 29 years. Over 90 percent of patients will have high blood pressure by the time they reach kidney failure. Men have higher blood pressures than women, and high blood pressure is associated with bigger kidneys and faster rates of kidney growth
- Urinary tract infections
- Hematuria (blood in the urine)
  occurs in 35 to 50 percent of people with ADPKD and may be the first sign of the disease. With hematuria, the urine may be a pink or red color. Repeated episodes of hematuria are common. Hematuria is associated with increased kidney size and a faster rate of kidney growth.

  Hematuria related to bleeding cysts generally stops within two to seven days. The usual treatment includes bed rest and increasing fluids until the bleeding stops. If bleeding does not stop with bed rest and increased fluids, a procedure to stop the bleeding may be required. Bleeding into a cyst that does not communicate with the urinary system may not produce visible blood; in this setting, patients typically have localized flank pain and, in some cases, a low-grade fever.
- The need to pass urine more often
- Kidney pain
  Pain is the most common feature of ADPKD. People with ADPKD often develop flank and abdominal pain that is not related to infection, bleeding into a cyst, or a kidney stone. The pain is often dull and persistent and is thought to be caused by stretching of the wall of a cyst or pressure on other organs when the kidneys and/or liver are very large. This is the most common symptom in ADPKD patients. In contrast, pain that begins suddenly is more likely to be caused by bleeding into or infection of a cyst, or a kidney stone.
- Kidney stones

These are small, stone-like objects that form inside the kidneys. Kidney stones occur in up to 25 percent of people with ADPKD. Kidney stones may cause pain, or sometimes they can block the flow of urine without symptoms.

Treating kidney stones that block urine flow is extremely important in patients with ADPKD. If the blockage is not relieved, the function in that kidney may be lost. Relieving the blockage may require the help of a urologist. The cysts make it harder to surgically remove the stone or use shock waves to break up the stone (extracorporeal shock-wave lithotripsy [ESWL]). blood in the urine.

- Kidney failure

  Normally, the kidneys filter out excess toxic and waste substances and fluid from the blood. In people with polycystic kidney disease (PKD), the kidneys become enlarged with cysts that and kidneys don't function well. Kidney (renal) failure that is severe enough to require dialysis or kidney transplantation is called end-stage kidney disease. https://www.kidney.health.nz/Patient-Information/Information-on-Kidney-Disease/

Does everyone with PKD develop kidney failure?

No. ADPKD rarely leads to end-stage kidney disease in early childhood; it most commonly occurs in middle age or later in life. The likelihood of requiring dialysis in people with ADPKD is estimated at less than 2 percent in people under age 40 years, increasing to 50 to 75 percent by age 70 to 75 years

Certain people have an increased risk of kidney failure including:

- patients with high blood pressure
- patients with protein or blood in their urine

PKD can also cause problems in other parts of the body, such as:

- A bulging blood vessel in the brain (aneurysm)‑ If the blood vessel bursts, it can cause a sudden, severe headache and nausea and vomiting. A burst blood vessel can lead to brain damage and even death. Aneurysm rupture occurs most often in people with larger aneurysms (>10 mm). **Early detection (before symptoms occur) of intracranial aneurysms is recommended in people who are at high risk.(a family member has also suffered aneurysm) Screening (looking for an aneurysm before it ruptures) is generally performed with magnetic resonance angiography (MRA) or, if not available, a computed tomography (CT) scan*
- Cysts in the liver ‑ These can cause belly(abdominal)
- pain.

  Liver cysts occur commonly in people with ADPKD. More than 85 percent of patients will have liver cysts by age 30 years. Most people with liver cysts have no symptoms and have normal or near-normal liver function. However, some people develop abdominal fullness, early satiety, difficulty breathing, or, less commonly, pain

- A weak area in the belly muscles (called a "hernia") – This can cause an area of the belly to bulge out.
  Abdominal wall hernias are relatively frequent, affecting approximately 45 percent of people with ADPKD. Abdominal wall hernias are usually located on the midline just above the belly button or in the groin areas.
  Surgery is the best treatment for abdominal wall hernias that cause pain, but not all hernias require surgical repair. Small hernias may be monitored over time
- Heart problems
  Abnormalities of a heart valves (mitral valve) are detected in up to 25 percent of patients with ADPKD. Most patients with this form of heart valve disease have no symptoms and require no treatment. However, in very rare cases, the valve disease may progress over time and become severe enough to require valve replacement. Some patients with valve disease (mitral valve prolapse) may present with palpitations and may need specific medications (beta blockers).

If you have any of these symptoms, contact your health care provider. He or she may want to test for kidney problems. If your health care provider thinks ADPKD may be causing your symptoms, he or she will check the following:

- Family history of PKD
- Imaging tests, such as ultrasound, sometimes a CT scans or MRI scans (may detect smaller cysts that cannot be found by an ultrasound) diagnosis may not be clear on imaging of children and young people. However, by age 30 if you have PKD it will be identifiable on imaging
- Physical examination to check for enlarged kidneys and blood pressure check
- Blood test
- Urine test

A positive family history is known in about 75 percent of patients with PKD. This is helpful to identify other at-risk family members. In general, the signs and symptoms of PKD are not specific enough by themselves for your doctor to make the diagnosis
The imaging tests may be done to look for visible signs of cysts in your kidneys. Blood test will look for signs of kidney damage. Urine test will look for blood and protein in your urine.

I have family members with PKD, should I be tested?

It is an important and impactful decision. Things to consider before being tested:

- What will I do with the information once I have it?
- Am I better off knowing or not knowing?

An ultrasound of the kidneys is the best screening test. This test can be undertaken at any age but in children and those under the age of 30 an ultrasound may sometimes not confirm the diagnosis even though it will develop later. If your ultrasound is normal and you are older then 30; then you will not develop PKD.

Some people choose to remain undiagnosed but live a healthy lifestyle, eating well with blood pressure monitoring. They see the doctor yearly often to monitor kidney function. If or when they have symptoms, lose kidney function, or develop high blood pressure, an ultrasound should be undertaken.

How to take care of yourself:

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. Treating high blood pressure can have a number of benefits in people with PKD because it can help prevent heart disease and strokes as well as reducing the likelihood of developing kidney failure

Make sure you take the medication prescribed by your doctor

Diet for living a healthy lifestyle including weight loss, exercise, and a low-salt diet and controlling your cholesterol are all an important part of controlling your blood pressure and staying as healthy as possible. You should do your best to reach these goals and work with your family doctor and your Kidney specialist to achieve the best outcomes.

If you have other symptoms or problems, you might need other treatments, too.

For example, doctors can:

- Treat kidney infections with antibiotic medicines
- Treat pain with pain-relieving medicines The use of nonsteroidal anti-inflammatory drugs (NSAIDs), such as ibuprofen or naproxen, is not recommended, although people with ADPKD may, on occasion, benefit safely from these medications in short courses .You should speak to your health care provider about the risks and benefits of NSAIDs before using them. NSAIDs are not recommended for patients with ADPKD when kidney function is reduced or when they need to be used on a long-term basis to treat chronic pain basis
- Undertake surgery when safe and appropriate to fix a bulging blood vessel (aneurysm) in the brain
- Undertake surgery to fix a hernia

How much fluid should I drink each day:

Avoiding dehydration is very important when you have PKD. generous water intake:2 or more liters a day when there is no kidney failure helps maintain a dilute urine and decreases the risk of kidney stones, which are seen at increased frequency in PKD patients.

In addition, with severe kidney disease, generous water intake can sometimes be problematic and even dangerous. Thus, it is important to discuss appropriate water intake with your doctor.

Who should I contact for more information or support?

Kidney Health NZ 0800 543 639 www.kidneys.co.nz

https://pkdaustralia.org/# or www.PKD.org

Kidney Health NZ 0800-KIDNEYS (0800 543639) www.kidneys.co.nz