



POLYCYSTIC KIDNEY DISEASE (PKD)

What is polycystic kidney disease?

Polycystic means “many fluid-filled sacs” or “many cysts”. Polycystic kidney disease (PKD) is a genetic disorder that causes multiple cysts to form in the kidneys. It’s the most common inherited kidney disease and one of the leading causes of kidney failure.

How does PKD affect the kidneys?

Polycystic kidneys have a bumpy surface and become very large over time as the number and size of cysts continues to increase. PKD can be associated with a number of conditions, including:

- High blood pressure
- Urinary and kidney infections
- Kidney stones
- Kidney failure

Kidney failure resulting from PKD is caused by a combination of pressure on normal kidney tissue as the cysts expand, and the scarring of normal support tissue in the kidney.

Who is affected by PKD?

PKD is an inherited disease. This means that it’s passed on from parents to their children through their genes, although sometimes new cases can occur. There are two forms of PKD:

- Autosomal dominant PKD (ADPKD)
- Autosomal recessive PKD (ARPKD)

Autosomal dominant PKD is the most common form of the disease. It affects about 1 in every 500-1,000 births and the symptoms usually appear in midlife. ADPKD follows a dominant inheritance pattern, meaning that if either your mother or your father has the disease, you have a 50% chance of inheriting it. If the gene for ADPKD is not passed on to you, you will neither inherit the disease, nor can you pass it on to your children. There are two types of ADPKD: Type 1 and Type 2.

Autosomal recessive PKD is a very rare form of PKD causing symptoms in infants and young children. Children have a 25% chance of inheriting ARPKD if both parents carry a defective copy of the gene involved. If only one parent carries the defective gene, children cannot inherit the disease.

What are the chances that PKD will cause kidney failure?

Kidney failure occurs when you are left with less than 10-15% of total kidney function. When the kidneys fail, dialysis treatment or a kidney transplant is needed.

Not everyone with PKD will go on to develop kidney failure. On average, half of the people with Type 1 ADPKD will need treatment for kidney failure by age 60. People with Type 2 ADPKD have a lower risk of developing kidney failure. There are treatments available that help to delay progression of the disease.

Children born with ARPKD usually develop kidney failure within a few years.

What are the symptoms of ADPKD?

ADPKD usually progresses very slowly. Early in the disease there are often no symptoms. The most common symptoms include:

- Pain in the back and sides (between your ribs and your hips)
- Headaches

People with ADPKD may also experience:

- High blood pressure (hypertension)
- Urinary tract infections
- Kidney stones
- Blood in the urine (hematuria)
- Liver and pancreatic cysts
- Abnormal heart valves
- Bulges in the walls of blood vessels in the brain (aneurysms)
- Small sacs on the colon (diverticulosis)

How is PKD diagnosed?

PKD is usually diagnosed by ultrasound, CT scan or MRI.

There is another test called gene linkage analysis. With this method, blood tests are used to detect which family members carry the PKD gene. However, this method relies on DNA from multiple affected members of the family. It's not usually done for diagnostic purposes.

Direct genetic testing can be used to diagnose autosomal dominant PKD. With this method, ADPKD can be detected before cysts start to develop.

Before you consider being tested for PKD, it's important to understand the benefits and risks involved. These can include determining the possibility of having a child with PKD, and what the test results could mean to career and insurance discrimination. Speak with your doctor about whether being tested makes sense for you.

What should you do if you have PKD?

Although there is no cure for PKD, there has been significant progress in researching the disease. Doctors are better able to predict how quickly it will progress (Mayo Imaging Classification). There is also some evidence to suggest that lowering your blood pressure can slow the advancement of PKD. Finally, new treatments are available to decrease the growth of the kidney cysts and likely delay the progression to kidney failure (Tolvaptan).

If you have PKD, you should:

- Visit your doctor regularly.
- Learn all you can about the disease.
- Control high blood pressure.
- Have urinary tract infections treated immediately.
- Never take any over-the-counter medication without first checking with your doctor.
- Discuss family planning with your doctor or genetic counselor.

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