Polycystic Kidney Disease

National Kidney Foundation®

Making Lives Better

What is polycystic kidney disease?

There are several types of polycystic kidney disease (PKD). However, PKD **usually refers to a genetic or inherited disease** that is sometimes called "adult PKD" because it normally appears in adult life. A less common type of PKD occurs primarily in babies and children.

In PKD, **cysts, or fluid-filled pouches, are found primarily in the kidney** but they **can also affect other organs**, including the liver, pancreas, spleen and ovary. Outpouchings may occur in the walls of the large intestine and in the walls of blood vessels in the brain, where they may cause aneurysms. They may also be found in the abdominal wall, causing hernias. In addition, the valves of the heart may be involved, becoming floppy and resulting in a heart murmur in some patients.

How common is PKD?

PKD is the **most common life-threatening genetic disease**, affecting more than 600,000 Americans and an estimated 12.5 million people worldwide. It is found in all races and occurs equally in men and women.

Who is at risk for developing PKD?

The **adult type of PKD** (also called autosomal dominant PKD or ADPKD) is passed from parent to child by an autosomal dominant type of inheritance. This means that only one copy of the abnormal gene is needed to cause the disease. Therefore, if one parent has the disease, **each child has a 50-50 chance of developing the disease.** The risk is the same for every child, regardless of how many children develop the disease. Boys and girls have the same chance of inheriting the disease.

The **less common form of PKD** (also called autosomal recessive PKD or ARPKD) is passed by an autosomal recessive pattern of inheritance. This means that **both parents must carry the abnormal gene**, and both must pass the gene to the child in order for the child to develop the disease. In this situation, **every child has a 25 percent chance of developing the disease** in a family that is at risk.

What are the clues that someone has PKD?

People who have inherited ADPKD often do not notice anything unusual until they are 30 to 40

years old. The **first noticeable symptoms** may include:

- high blood pressure
- back or side pain
- an increase in the size of the abdomen
- blood in the urine
- frequent bladder or kidney infections.

High blood pressure is the most important treatable risk factor in PKD. Occasionally, patients may develop headaches related to high blood pressure or their doctors may detect high blood pressure during a routine physical exam. Finally, **about 25 percent of PKD patients have a floppy heart valve**, and some of these develop a fluttering or pounding in the chest as well as chest pain. These symptoms almost always disappear on their own but may be the first hint that someone has PKD.

How is PKD diagnosed?

Today, **ultrasound** is the most reliable, inexpensive and non-invasive way to diagnose PKD. Someone at risk for PKD who is older than 30

and has a normal ultrasound of the kidneys probably does not have PKD. **Occasionally, a CT scan** (computed tomography scan) may detect smaller cysts that cannot be found by an ultrasound.

At present, PKD cannot be diagnosed by a single blood test. However, in some situations where it is important to have a diagnosis (for example, if a family member wants to donate a kidney to an affected parent or sibling, and ultrasound and CT scans are normal), special blood tests on at least three family members can be done to get a diagnosis in the at-risk individual. This form of testing is called **gene linkage analysis.**

Does everyone with PKD develop kidney failure?

No. About 50 percent of patients with PKD will have kidney failure by age 60, and about 60 percent will have kidney failure by age 70. **Certain people have an increased risk** of kidney failure. They include:

- men
- patients with the most common form of PKD (ADPKD)
- patients with high blood pressure
- patients with protein or blood in their urine
- women with high blood pressure who have had more than three pregnancies.

How is PKD treated?

At present, there is no cure for PKD. However, a lot of research is being done. Many studies suggest that some treatments may slow the rate of kidney disease in PKD, but further research is needed before these treatments can be used with patients. Other studies are improving our understanding of the genetic basis of PKD. It is

hoped that these studies will lead to the development of new therapies to prevent PKD or limit its severity.

In the meantime, many supportive **treatments can be done to help prevent or slow down loss of kidney function** in PKD. These include:

- careful control of blood pressure
- prompt treatment of any bladder or kidney infections
- lots of fluid and bed rest when blood in the urine is first noted
- a healthy lifestyle with regard to smoking, exercise, weight control and salt intake.

Should people with PKD follow a special diet?

At present, **no specific diet** is known to prevent cysts from developing in patients with PKD. Low protein diets have not proved helpful to PKD patients with advanced kidney disease. Nevertheless, it is not wise to eat excessive amounts of protein. **Reducing salt** in the diet helps control blood pressure in PKD patients who have high blood pressure. A diet low in fat and moderate in calories is recommended to maintain a healthy weight.

Is exercise recommended for people with PKD?

Absolutely. Activities such as swimming, walking and biking are always good. However, activities that are potentially harmful to the kidney, such as contact sports, should be avoided if possible. It is important not to become too dehydrated during any physical activity.

Should women with PKD get pregnant?

Most of the women with PKD (80 percent) have successful and uneventful pregnancies. However, some women with PKD have an increased risk for serious complications for themselves and their babies. This includes women with PKD who also have:

- high blood pressure
- decreased kidney function.

Women who have PKD with high blood pressure develop pre-eclampsia (or toxemia) in 40 percent of pregnancies. This is a life-threatening disorder for both the mother and baby, and it can develop suddenly and without warning. Therefore, all women with PKD, particularly those who also have high blood pressure, should be followed closely during their pregnancy by their doctor.

Should people with PKD have children?

Individuals with PKD who are concerned about passing the disease to their children **may want to consult a genetics counselor** to help them with family planning. Many university medical centers have this service.

Questions for My Doctor

What research is being done on PKD?

PKD is a **very active area of research** at present. Some of the questions being investigated are:

- What changes occur in the PKD gene?
- What is the function of polycystin, a protein that may be associated with the development of PKD?
- What makes kidney cysts grow?
- What is the best level of blood pressure control for PKD patients?
- What is the best type of blood pressure medication for PKD patients?
- Do aneurysms in PKD patients grow? How fast do they grow?

What if I have more questions?

If you have more questions, you should speak to your doctor. You can also get more information from:

National Kidney Foundation
30 East 33rd Street
New York, NY 10016
(800) 622-9010
www.kidney.org

Polycystic Kidney Disease Foundation
4901 Main Street, Suite 200
Kansas City, MO 64112-2634
(800) PKD-CURE
www.pkdcure.org

More than 20 million Americans—one in nine adults—have chronic kidney disease, and most don't even know it. More than 20 million others are at increased risk. The National Kidney Foundation, a major voluntary health organization, seeks to prevent kidney and urinary tract diseases, improve the health and well-being of individuals and families affected by these diseases, and increase the availability of all organs for transplantation. Through its 51 affiliates nationwide, the foundation conducts programs in research, professional education, patient and community services, public education and organ donation. The work of the National Kidney Foundation is funded by public donations.

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02-06
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