Polycystic kidney disease (PKD) is an inherited renal disease that begins with the growth of small cysts in the renal tissue. As these sacs of fluid grow in size and number, they cause the kidneys to swell. Eventually, the cysts take over the healthy, functioning renal tissue that filters wastes. As the disease progresses, the cysts may become so destructive that the kidneys are no longer functional, with dialysis or transplantation required.

It is unclear what causes cysts to form in the kidneys of PKD patients. In most cases, the disorder is inherited when mutated genes are passed down in one of two distinct ways. Autosomal-dominant PKD (ADPKD), the more common form, may be passed down when one parent has the disease and carries a mutated PKD gene; in this case, the child has a 50% chance of developing PKD. The other form, which is far less common, is autosomal-recessive PKD (ARPKD), in which both parents are disease free, but each carries the mutated gene and passes it down to the child; in this case, the child has a 25% chance of developing PKD.

Symptoms of PKD include high blood pressure, frequent kidney infections, hematuria, and lower back pain. ARPKD tends to cause symptoms in young children, whereas ADPKD may not cause symptoms until adulthood.

There is no cure for PKD. Treatment is aimed at controlling pain, high blood pressure, and kidney infections. Early detection is important in order to delay symptoms and preserve renal function for as long as possible. About half of ADPKD patients will eventually develop kidney failure and require either lifelong dialysis or kidney transplantation. Cyst formation will not occur in the transplanted kidney.

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More than half a million people in the United States have polycystic kidney disease (PKD), one of the most frequent causes of kidney failure in this country. In almost all cases, PKD is inherited by the passing down of a mutated gene from one or both parents, although a small percentage of people develop PKD as a result of a spontaneous genetic mutation.

**Symptoms and Disease Progression**

There are two distinct types of PKD: autosomal-dominant (ADPKD) and autosomal-recessive (ARPKD). ADPKD, the more common type, is sometimes referred to as “adult PKD” because the symptoms develop over many years and first occur in early adulthood.

ARPKD, which is relatively rare, is sometimes referred to as “childhood PKD” because the symptoms begin in childhood, sometimes even before birth. Kidney failure may occur more rapidly than with ADPKD.

The symptoms and progression of these disorders are different, but both types cause cysts to form in the part of the kidney that filters wastes. Eventually, the cysts become larger and more numerous, causing the kidneys to enlarge dramatically. Pain in the area of the kidneys may be caused by the cysts themselves, an infection, bleeding, or kidney stones. As the kidneys’ function begins to deteriorate, the blood pressure increases and the cysts develop infections and bleed. The resulting symptoms include lower back pain, headaches, high blood pressure, and urinary tract infections. Complications from PKD include cysts in other organs, such as the pancreas or liver; aneurysms in the blood vessels of the brain; and diverticula (pouches) in the colon.

**Diagnosis and Treatment**

Diagnosis is typically made by ultrasound, CT, or MRI, which allows the doctor to see cysts that are at least one-half inch in diameter. A genetic test to confirm the presence of PKD mutations is available, which may help prospective parents determine their risk of passing down a mutated gene to their child.

Treatment is focused on relieving symptoms and preserving kidney function for as long as possible. Lower back pain or flank pain may be treated with nonprescription or prescription pain medications. Kidney infections should be treated at the first sign of symptoms. Infection or bleeding of a cyst may require drainage. Headaches, which are a common symptom in PKD, may be due to high blood pressure or an aneurysm in a blood vessel in the brain. The cause of headache should be determined before the patient self-treats chronic or severe headaches. Diet, exercise, and antihypertensive medications may be used to normalize blood pressure.

Since the kidneys are important in maintaining the hormonal balance necessary for proper growth, children with PKD may be smaller than average. As a result, growth hormone is sometimes appropriate in the treatment of young children with PKD.

In advanced cases of PKD, surgical removal of one or both kidneys may be necessary. Cyst removal is usually impractical owing to the large number of cysts and the fact that this solution is only temporary.

Your pharmacist can answer questions about medications you may be taking to treat PKD symptoms. He or she also can tell you which medicines to avoid if you have been diagnosed with kidney disease.