Polycystic kidney disease (PKD)

What is PKD?

PKD is the most common inherited (genetic) disorder of the kidneys. In families with PKD, there is a 50% chance that a person with PKD will pass it on to a child, but it can occur spontaneously in people with no family history of PKD. In these cases, neither of the parents will have had the disease.

Many people with PKD live for several decades without any symptoms, and unless it is known that another family member has PKD, there may be no reason to look for it in people without symptoms.

A polycystic kidney is the same shape as a normal healthy kidney but over time, often becomes much larger due to the development of thousands of cysts on the kidney surface. High blood pressure is common and develops in most patients by age 20 or 30. Kidney failure can occur.

How do people know they have PKD?

The most common symptom is pain in the back or sides, or between the ribs and hips.

Pain may be mild or severe, temporary or persistent.

People with PKD can also experience the following complications:

- High blood pressure
- Persistent or recurrent urinary tract infections
- Blood in the urine (haematuria)
- Cysts on the liver or pancreas
- Abnormal heart valves
- Kidney stones
- Aneurysms (bulges in the walls of blood vessels) in the brain, which can
- Bleed causing a stroke
- Diverticulosis (small pouches that bulge outward through the colon).

What causes PKD?

PKD is caused by a genetic abnormality in special genes called PKD1 and PKD2. Many types of genetic abnormality can occur, so genetic testing is not simple and the tests can be difficult to interpret. Anyone considering genetic testing should receive genetic counselling to understand the implications of the tests and their results.

How is PKD diagnosed?

PKD is usually diagnosed with an ultrasound of the kidneys. Ultrasound findings are variable and partly depend on a patient's age, with younger patients usually having fewer, smaller cysts. Doctors have developed specific criteria for diagnosing the disease with an ultrasound depending on a patient's age. For example, the presence of at least three cysts in each kidney by age 30 in a patient with a family history of the disease can confirm the diagnosis of PKD. There is no point in doing an ultrasound on the children of a patient with PKD until they are 18, as cysts will not be big enough to see.

How is PKD treated?

Although there is no cure for PKD, treatment can ease symptoms, delay dialysis and prolong life.

Pain: Pain in the area of the kidneys can be caused by cyst infection, expansion or bursting, bleeding into cysts or kidney stones. If there is severe pain from cyst enlargement, surgery to shrink cysts can sometimes help; but may only provide temporary relief as it will not slow the disease progression or development of further cysts.

Urinary tract infections: People with PKD tend to have frequent urinary tract infections, which should be treated with antibiotics. Cyst infections are difficult to treat because many antibiotics do not penetrate to the cysts.

High blood pressure: An important complication of PKD is high blood pressure (hypertension). Keeping blood pressure under control can slow the effects of PKD. Two types of blood pressure tablet – called angiotensin-converting enzyme inhibitors (ACE-i) and angiotensin receptor blockers (ARBs) – may protect kidney function and are possibly superior to other blood pressure tablets in PKD. Whatever is used, the blood pressure should always be under 130/80mmHg.

End-stage renal disease (ESRD): After many years, PKD can cause the kidneys to fail, and dialysis and/or a transplant may become necessary. Kidneys transplanted into patients with PKD do not develop cysts.

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