

KIDNEYMATTERS

Information from the National Kidney Federation - supporting kidney patients, their friends & family

ADULT POLYCYSTIC KIDNEY DISEASE

- What is polycystic disease?
- What is the cause of Adult Polycystic Kidney Disease?
- What happens to the kidneys in polycystic disease?
- How is it transmitted?
- Should a family member be screened for polycystic disease?
- What are the other complications of polycystic disease?
- What treatment is available for PCKD?
- Can cysts be removed or operated on?
- Can a polycystic patient lead a normal life?
- Other information/Help groups



www.kidney.org.uk

HELPLINE 0800 169 0936

What is polycystic disease (PKD)?

In polycystic kidney disease abnormal cysts form in the kidneys and can also form in other organs. Cysts are round swellings of fluid filled with fluid. There are two main forms of polycystic kidney disease, Autosomal Dominant Polycystic Kidney disease and Autosomal Recessive Polycystic kidney disease.

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is the most common genetic kidney disease. This information is focused on this disease.

Autosomal Recessive Polycystic Kidney Disease is diagnosed in childhood

What is the cause of Adult Polycystic Kidney Disease?

ADPKD is caused by a mistake in the DNA, the code which determines the nature of all tissues in the body. Larger sections of DNA are called genes, so ADPKD is called a genetic condition. There are several genetic abnormalities that can lead to PCKD. This makes it difficult to perform genetic testing to diagnose ADPKD and may also be one of the explanations for the different outcomes in people with ADPKD.

In ADPKD, the commonest genetic abnormality is in the gene that codes for a protein that is called polycystin. This affects the function of tiny hair-like structures on the surface of many cells in the body, including the kidney.

What happens to the kidneys in Polycystic Disease?

Multiple cysts grow in the kidneys. A cyst is a small circular ball filled with watery fluid. They vary in size from a pinhead to a table tennis ball (2cm). Each cyst is a ballooning up of tiny tubes that are normally found in the kidneys. Eventually the kidneys may become very enlarged, looking as though they consist of a mass of small balls stuck together. This is not a malignant or cancerous change. They can be 3-4 times the size of normal kidneys.

Kidney failure often develops in late middle age. However there are some families where kidney failure is delayed or never occurs, and others in which kidney failure occurs earlier (age 40-45 years).

Normally the function gets worse only very gradually but it is possible to predict from the blood tests how much the kidneys will last. When kidney failure occurs, dialysis may be necessary and a kidney transplant should be possible.

How is it transmitted?

Note: this explanation applies only to ADPKD (transmission of the childhood disease is quite different). Furthermore, some people with ADPKD do not seem to have a family history of the condition or to develop kidney failure and the inheritance could be different in such cases.

ADPKD is a genetic condition. This means that it is caused by a mutation, which makes a mistake or error in the genetic code which makes important proteins in the body. There are two copies of the gene which carries the code for ADPKD. Our genes are inherited and therefore it is possible to pass on the gene to children.

The two important points that determine transmission in classical PCKD are:-

1. If you have the polycystic gene, you get the disease. There are no hidden carriers, unlike some other inherited diseases;
2. You have 2 copies of each gene and only one needs to be abnormal for you to have Polycystic Kidney Disease. If you have Polycystic Kidney Disease, you have one normal gene and abnormal gene.

Your parents

If you have polycystic disease, one of your parents had polycystic disease, unless the abnormality in the genes started with you (in which case you have a “new mutation”).

Your brothers and sisters

If you have polycystic disease, each of your brothers or sisters has a 50% chance of having polycystic disease. Men and women are equally

affected. If a family member has the disease but you have been told you do not have the disease, it can't be passed on to their children.

Your children

If you have polycystic disease, each of your children has a 50% chance of getting your polycystic gene, and a 50% chance of getting your normal copy of this gene. Boys and girls are equally affected.

Should a family member be screened for polycystic disease?

The screening test is an ultrasound (sound wave) scan of the kidneys. This is painless and simple to do.

Scans performed of a baby before birth may show some cysts in the kidneys. Even if just a couple of cysts are visible, this is highly suggestive of ADPKD in the baby if the mother also has ADPKD

Until a few years ago, children were not screened for ADPKD This was partly because cysts may not be seen in the kidneys until someone is 25-30 years of age, and doctors did not want to cause anxiety by giving a child with a diagnosis of ADPKD if there was no treatment available. However, this view has changed considerably in the last few years. First, modern ultrasound machines can detect small cysts in the kidney, and these may be visible in many children, making the diagnosis more certain. Secondly, there is increased attention to the treatment of high blood pressure in children with kidney disease, so that there may be considerable long term benefit in early diagnosis.

Exact advice may vary in different parts of the country, but many specialists advise getting an ultrasound scan of the kidneys in children at risk between the ages of 7 and 10, repeating every 5 years up to the age of 30 if cysts are not seen.

Of course it is not essential to have a scan, and quite a few people decide against scans. It might be prudent, though, to have a blood pressure check from time to time even if a scan is not performed. Some people at risk of ADPKD think that by not having a scan they will avoid problems with life insurance and health insurance.

This is not the case. In fact, the situation is quite the reverse, if someone has concealed the possibility of ADPKD from an insurance company, they are at risk of invalidating all their insurance.

What are the other complications of Polycystic Disease?

- Pain can occur in the kidneys, due either to an infection or a small bleed into a cyst. Normally, simple painkillers will treat the symptoms which should get better in a few days. A sample of urine may need to be tested to see if there is an infection.
- Occasionally some bleeding into a cyst may cause blood in the urine.
- There may be cysts in the liver, which would be seen on the ultrasound scan when the kidney disease is diagnosed. It is very unusual for these to cause liver failure.
- High blood pressure commonly occurs, and should be treated at an early stage to help protect the kidneys and the rest of the circulation from possible damage. Even at an early stage when the kidney function is normal, the blood pressure should be checked every 6-12 months.
- A rare complication of APCKD is stroke. This makes it even more important to ensure that the blood pressure is controlled. If any members of your family with polycystic disease have had premature stroke, discuss this with your doctor.
- The large bowel can develop outpouchings in the wall, called diverticulae. These are very common in the general population, and are often not a problem. However they can cause variation in bowel habit with discomfort (diarrhoea and constipation). Diverticular disease is not bowel cancer and does not cause bowel cancer.

What treatment is available for ADPKD?

Treatment of blood pressure seems very important in any type of kidney disease, and this probably includes ADPKD. If the blood pressure rises to

140/80, treatment should be given, with a target level of 130/80. High blood pressure may appear in early stages of the disease, in children with normal renal function. Some kidney specialists believe there is value in starting treatment of the blood pressure as early as possible, and as a result are seeing some people whose kidneys are working much better than their parents' and grandparents' kidneys were at the same age.

Drugs have been developed to specifically treat the formation of cysts in ADPKD. Some have been less successful for example sirolimus (a drug also used to prevent kidney transplant rejection).

More recently, a medicine called Tolvaptan has been approved for use in ADPKD and can be used in some patients. This medicine works by reducing the pumping of water into cysts and therefore, slows their increase in size. It is unlikely that these drugs will help people with advanced kidney failure but have the potential to prevent or delay kidney failure when started early in the disease from getting worse quite quickly. There are strict criteria for the medicine and the criteria is different in Scotland from England and Wales. People with polycystic disease should ask their kidney specialist whether they should be considered for the drug.

Can cysts be removed or operated on?

It is rare for cysts to require drainage. If so, this can be done with a needle, with some local anaesthetic to dull the pain and an X-ray (ultrasound) machine for guidance. However, polycystic kidneys contain so many cysts that drainage of a single cyst cannot cure the condition. Even if one cyst is painful, there may be hundreds of cysts in each kidney, and needling the one causing the problem can be very hard. However, even though cyst drainage remains rather experimental, it can sometimes help.

In extreme cases, particularly if severe infection has been a problem, the whole kidney can be removed at an operation. The kidney is often very enlarged, so this is not a minor operation, but it can be very helpful in some situations. In some cases, the kidney is so large that it would be impossible to put a transplant kidney in place, so removal of a polycystic kidney is necessary before going on the transplant list.

Can people with polycystic disease lead a normal life?

It is usually possible to lead a normal active life with this condition. Kidney failure does not normally cause any symptoms until it is very advanced. Dialysis and transplantation should allow you to carry on with work and do most of the things you want to. Some patients, however, do have times when the kidneys are quite painful, and this can restrict heavy work and some daily activities.

The condition will have to be mentioned when applying for mortgages or life insurance. Nowadays it should still be possible to get cover in the normal way, though there may need to be some loading of the policy. The insurance company may ask for special medical reports, so do plan for a little delay when any policies are needed. If you are refused by one company, do try again, and ask the National Kidney Federation for advice about helpful companies.

Other Information/Help Groups

PKD Charity Helpline 0300 111 123

E-mail : info@pkdcharity.org.uk

The National Kidney Federation cannot accept responsibility for information provided. The above is for guidance only. Patients are advised to seek further information from their own doctor.

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The NKF - the largest Kidney Patient charity in the UK

A federation of more than fifty patient charities, supporting kidney patients & carers

NKF(National Kidney Federation)

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