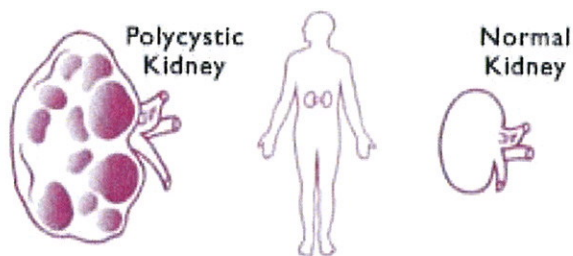


Polycystic kidney disease



AN INTRODUCTION TO POLYCYSTIC KIDNEY DISEASE
This is quite a common type of kidney disease in which cysts (fluid filled sacs) are formed in the kidneys. 'Polycystic' therefore means 'many cysts'.

It affects both kidneys although one may be affected earlier than the other and/or more than the other. Over time the cysts get larger and increase in number, gradually squeezing the normal tissues until the kidneys lose their ability to function.

PKD (polycystic kidney disease) is a hereditary disease (i.e. it is passed from one generation to another) and hence a family disease. Although not all members of the family will inherit it, everyone is affected at least emotionally.

This page is designed to provide factual information about PKD, so that families can understand and cope with the disease.

HISTORY AND PREVALENCE

PKD was first described in the nineteenth century but a comprehensive study describing the disease was not done until 1957 by Dr Dulgaard in Scandinavia. The PKD discussed in this booklet refers to adult PKD also known as Autosomal Dominant PKD – ADPKD (This is not to be confused with the very rare forms of infantile polycystic disease which is unrelated to adult PKD).

PKD is found on all continents and amongst all ethnic groups throughout the world. It is not known just how many people in Australia have the disease because symptoms often don't appear until late in life and many people may not know they have the disease. It is the fourth highest cause of kidney failure in Australia and the approximate frequency of the condition in Caucasians is 1 in 400 to 1 in 1000 people.

SIGNS AND SYMPTOMS

There are no symptoms in early life. However, in adults as the kidneys grow larger, so does the waistline. Some people are unaware that they have the disease until they seek relief for their swollen or sometimes tender abdomens. Other complaints that may lead to the diagnosis include pain in the back or side, development of kidney stones (occasionally), high blood pressure, urinary tract infections or blood in the urine. A doctor may discover the kidneys are enlarged on a routine medical examination. Often the disorder is symptomless until kidney failure develops.

WHO IS AFFECTED?

PKD is a hereditary disease which affects both men and women equally and its effects are seen in adult life.

Each child of a parent with PKD has a 50% chance of inheriting the disease. The prospects are similar to the flip of a coin. So, although 1 in every 2 children is likely to inherit PKD, sometimes both or neither will be affected. This form of inheritance is called Autosomal Dominant – hence PKD is sometimes abbreviated to ADPKD – (Autosomal Dominant PKD).

Around 85% of people with PKD have an abnormal gene on chromosome number 16. This main group is known as ADPKD-1. The remaining 15%, known as ADPKD-2, where the abnormal gene is located on chromosome 4, appear to have a more slowly progressing form of Polycystic Kidney Disease.

PKD does not skip over a generation. Therefore if a parent does not pass on the condition to his or her child, then there is no chance of PKD affecting that child's own children.

Sometimes PKD may appear to skip a generation. For example, a grandmother has the disease, her daughter apparently didn't have it

but the daughter's son has it. Rather than skip a generation, the daughter must have had PKD but never developed symptoms or signs of the condition.

Occasionally a person presenting with PKD is the first in the family with no prior family history of PKD. The disorder is presumed to have arisen by what is called fresh mutation. Brothers and sisters of this person are not at risk but the children of the affected person have the same 50% chance of inheriting the disorder.

AN EXAMPLE OF HOW PKD IS INHERITED

People with PKD often develop kidney failure but this usually does not occur before they reach their 40's. Because cysts do not grow at the same rate in all people, some individuals do not experience kidney failure until their 60's or even later. Only 50% of 60 year olds with PKD require dialysis treatment.

HOW IS IT DIAGNOSED?

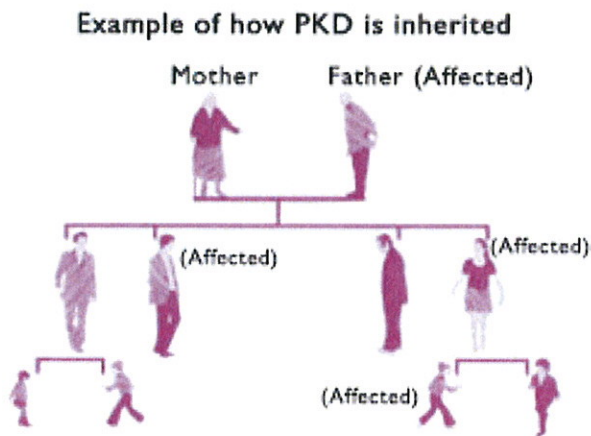
Even though there may be no symptoms, testing for PKD can be done using ultrasound or X-Ray tests including C.T. scans. In all instances, a careful medical examination should be done and only a doctor can tell you for certain if you have PKD. Seeing a doctor experienced in diagnosis and treatment of kidney disease is the best way to resolve any questions and worries you might have about PKD.

WHEN CAN TESTING BE DONE?

PKD cannot be diagnosed at birth. The cysts become detectable during teenage years. The best time to test for PKD is in late teens, after school is completed.

Genetic tests for PKD are now available providing there are 2 other family members with PKD who can attend for a blood test. Your renal physician can arrange referral for these tests.

WHAT CAN YOU DO ABOUT PKD?



Because children of people with this disease have a 50% chance of inheriting it, diagnosis is important at this stage. Although it may not alter the course of the disease, early diagnosis does allow close supervision and prevention of some of the complications so that people stay as healthy as possible. Therefore, regular check-ups with your doctor for blood tests and blood pressure

control are essential.

HOW WILL PKD AFFECT YOU?

Early in the course of the disease, before kidney failure develops, the most important complications are bleeding into the kidneys and high blood pressure. Infection in a cyst can also sometimes occur. The cause of high blood pressure (hypertension) is not well understood but is common in many types of kidney disease. Hypertension may cause further damage to kidneys and hastens the development of kidney failure. However, effective treatment is available for these complications and hypertension can and should be controlled. Bleeding which apparently occurs in the cyst wall and may then be passed in the urine, can be frightening and painful. The bleeding usually stops by itself with rest in a few days. It is not advisable to take aspirin or similar drugs as this may increase likelihood of bleeding.

People with PKD lead a normal and active life until they have lost 90–95% of kidney function. Many people continue to pass urine even at this stage but because the cysts have damaged the kidney's filtering units, the urine passed is only water and salts and does not contain the impurities and toxins that should be removed through the kidneys and passed in urine. It may be helpful to cut down on protein in the diet to avoid overloading the kidneys with protein impurities.

Some people with PKD also develop liver cysts. These rarely cause trouble or pain but the doctor will be alert to signs of liver trouble and manage it if it develops. Very occasionally an aneurysm or weakness occurs in an artery to the brain. Any prolonged, severe headache should be reported to your doctor.

TREATMENT

When kidney function is reduced to less than 10% it becomes necessary to consider artificial kidney (dialysis) treatment and/or a kidney transplant. Should this become necessary, it is important to realise that these are effective life sustaining therapies and help people remain well and active. More information about these treatments can be obtained at renal units of major hospitals or from the Renal Resource Centre.

Sometimes, if the kidneys become too large, are not contributing any useful function or are chronically infected, one or both are removed. If they are very large, one may be removed to make space for a transplant.

PKD AND YOUR FAMILY

PKD, like many other chronic, progressive diseases, can cause concern and stress for the patient and his or her family.

Families are often reluctant to discuss the hereditary aspects of the disease. The parent may feel guilty for passing on the disease and children feel frightened or resentful about the possibility of inheriting PKD. Discussion about PKD and its impact on the family is not easily faced. Everyone reacts differently to the possibility of inheriting the disease. Some people live in fear and anxiety about it. Others respond by ignoring it and denying that it could happen to them, refusing to talk about it or seriously consider it when contemplating marriage and family. However, it is important that it is discussed for a number of reasons:-

1. Early diagnosis can mean better treatment especially for hypertension and infections and therefore may slow down kidney failure.
2. Couples need to consider the possible impact of PKD when considering marriage and planning a family.

Those people who “work through” the facts about PKD and learn to accept and cope with the situation live active and productive lives based on a realistic picture of the future. This approach reduces fear and tension in a family that arises from ignoring or denying the disease and allows for a more positive approach to life.

Pretending “it can’t happen to me” is irresponsible. Each of us has a responsibility to future generations and the full implications of the disease should be considered by everyone contemplating a family. Generally speaking, those who cope best with PKD are those who’ve known about the disease from childhood and discussed it freely and openly with relatives and close friends.

COUNSELLING

If it is difficult to talk about this disease as a family, there are qualified counsellors who understand its inheritance pattern and can help answer the questions common to all PKD patients. Genetic counselling can go a long way towards helping families with PKD retain maximum productivity and acceptance whilst coping with what is admittedly a difficult and challenging situation. Counsellors can help you answer questions like – When should I tell my children? What if they don’t want to know? When should they be tested? It is important that patients discuss this with their doctors who can then refer them to a qualified counsellor.

THE PKD ASSOCIATION

This association has been established to provide:

1. Information and Support to PKD families. Information about qualified counsellors can be provided.
2. Link with International PKD Groups, in particular the US based Polycystic Kidney Research Foundation.
3. Public Education about the disease and its impact on people’s lives.