Polycystic Kidney Disease
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This is quite a common kidney disease, in which cysts (fluid filled sacs) are formed in the kidneys. ‘Poly cystic’ 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 grow 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 booklet is designed to provide factual information about PKD, so that families can understand and best manage the disease.
History and Prevalence

PKD was first described in the nineteenth century European medical literature. A comprehensive study describing the disease was published in 1957 by Dr O Z Dalgaard in his classic doctoral thesis on the inheritance of PKD as an autosomal dominant trait.

The PKD discussed in this booklet refers to autosomal dominant PKD (ADPKD) or adult PKD. This form of PKD is not to be confused with autosomal recessive PKD (ARPKD) or infantile PKD. This rare form of PKD is usually diagnosed at birth.

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 later in life and many people do not know they have the disease. In Australia, PKD accounts for 6% of all people diagnosed with kidney failure. The approximate frequency of PKD in Caucasians is 1 in 400 to 1 in 1000. Because of its inheritance pattern, the children of PKD patients will have a 50% chance of inheriting the disorder.

Signs and Symptoms?

There are no symptoms in early life. However, in adulthood, 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 and sometimes tender abdomens. More common complaints that may lead to a diagnosis of PKD, include high blood pressure, blood in the urine or pain in the back or side. The kidneys may be found to be enlarged on a routine medical examination or on ultrasound done for other reasons, e.g. pregnancy, gall stones. In many cases, the disorder is symptomless until substantial kidney function is lost.
Who is affected?

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is a hereditary disease which affects both men and women equally, generally in the fourth, fifth and sixth decades of life.

Each child of a parent with ADPKD has a 50% chance of inheriting the disease, because the gene for ADPKD is dominant. The prospects are similar to a flip of a coin. So, although 1 in every 2 children has a chance of inheriting ADPKD, sometimes both or neither will be affected. This form of inheritance is called Autosomal Dominant inheritance.

About 85% of people with ADPKD have an abnormal gene on chromosome 16. This form of ADPKD is called ADPKD-1. Those with an abnormal gene located on chromosome 4 have ADPKD-2. This appears to be a more slowly progressing form of PKD. It is believed that there may be a small percentage of people with yet another PKD gene.
ADPKD does not skip a generation. If the gene is not inherited by an individual from one of his/her parents, then it is impossible for the disease to be passed on by that person to the next generation. Of course, brothers or sisters who may have inherited the gene from that same parent, have a 50% chance of passing the gene onto their children.

Sometimes ADPKD may appear to skip a generation. For example, a grandmother has the disease, her daughter apparently didn’t have it during her lifetime, but her daughter’s son has it. Rather than skip a generation, the daughter must have had ADPKD but never developed symptoms or signs of the condition.

Occasionally, a person presenting with ADPKD is the first in the family with no prior family history of ADPKD. The disorder is presumed to have arisen by what is called a 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.

People with ADPKD often develop kidney failure but this does not usually 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 ADPKD require some form of renal replacement therapy - either dialysis treatment or a kidney transplant.
How is ADPKD diagnosed?

Even though there may be no symptoms, testing for PKD can be done using ultrasound imaging, CT scans, MRI scans and genetic testing. A diagnosis is made where three or more cysts are detected and is strengthened by a family history of ADPKD and in some cases, the presence of cysts in the liver. Seeing a doctor experienced in the diagnosis and treatment of kidney disease is the best way to resolve any questions and worries you may have about PKD. Your GP can arrange a referral to a renal physician (nephrologist).

When can testing be done?

ADPKD cannot be diagnosed at birth. The cysts become detectable during teenage years and can sometimes appear up to the age of 30. The best time to test for ADPKD is the late teens or early twenties after schooling and studies are completed.

Your physician can arrange genetic testing but these tests cannot predict the severity or onset of the disease.

What can you do about ADPKD?

Because children of people with ADPKD have a 50% chance of inheriting it, diagnosis in early adulthood is important. Although early diagnosis may not alter the course of the disease, it does allow close supervision and management of kidney function, blood pressure and diet. The prolongation of kidney function and the prevention of the complications of kidney disease are therefore much more likely. Additionally, in the event of breakthroughs in the management of ADPKD, a renal physician will be aware of these and be able to recommend them to you.
How will ADPKD affect you?

Early in the course of the disease, the most important complications are bleeding from the cysts 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. If untreated, hypertension may cause further damage to kidneys and thus hasten the progression of pre-existing kidney disease.

Effective treatment is available for these complications and good control of hypertension is essential to slow the progression of kidney disease. Bleeding, which occurs in the cyst wall and may then be passed in the urine as bright red blood, can be frightening and sometimes painful. The bleeding usually stops by itself with rest in a few days. It is not advisable to take aspirin as this may increase the likelihood of bleeding. Paracetamol is recommended for pain relief in preference to non-steroidal anti-inflammatory drugs, which, although good pain relievers, can be damaging to kidneys.

People with ADPKD lead normal and active lives until they have lost around 90% of kidney function. Although the kidneys continue to produce urine, because cysts have damaged the kidneys’ filtering units, the urine passed is largely water and salts. It does not contain the waste products, which should also be removed by the kidneys. An accumulation of these waste products leads to a variety of symptoms, severely affecting your health and well being. As kidney function deteriorates, your physician will prescribe medications and perhaps some dietary changes to relieve these symptoms.

Some people with ADPKD may develop liver cysts. This is more common in women, develops at an earlier age and is more extensive than in men. It appears to be more severe in women exposed to the oral contraceptive pill and who have had three or more pregnancies. It is very rare for liver cystic disease to affect liver function but it can cause enlargement of the liver leading to some discomfort and decreased appetite. Recent research has suggested that postmenopausal use of
oestrogen can increase the number and size of liver cysts. These changes were mild and did not affect liver function or symptoms. To minimise the large delivery of oestrogen to the liver from ingested medication, it is recommended that oestrogen patches be applied to the skin if a postmenopausal woman with ADPKD wishes to use oestrogen therapy.

People with ADPKD have a slightly increased chance of cerebral aneurysms (a weakness in an artery in the brain). This can run in families. Any prolonged, severe headache should be reported to your physician. Should an aneurysm be suspected, screening will be performed. Your physician will advise on the best management approach.

**Treatment**

When kidney function is reduced to less than 15%, dialysis treatment and / or a kidney transplant should be considered. These are very effective life sustaining therapies and help people remain well and active, able to continue full time work, participation in sport and other leisure activities. More information about these treatments, can be obtained from your renal physician, your hospital’s renal unit or the Renal Resource Centre (see back page).

Sometimes, if polycystic kidneys are very large, cause a great deal of discomfort, bleed frequently or are chronically infected and are not contributing any useful function, one or both may be removed. Occasionally, one may be removed prior to transplantation, to create adequate space in the lower abdomen for the new kidney. However, native kidneys are not routinely removed prior to transplantation. Your physician will advise if such surgery is likely.
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ADPKD and Your Family

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

Families can often be reluctant to discuss the hereditary aspects of the disease. It is common for parents with ADPKD to feel guilty about passing on the disease. Children may feel frightened and resentful about the possibility of inheriting ADPKD. Some people become very anxious about it. Others respond by ignoring the possibility of inheritance, refuse to talk about it, be tested 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. This can slow down kidney failure.

2. Couples need to consider the possible impact of PKD when considering marriage and planning a family. They may decide to limit the size of their family. A diagnosis of ADPKD does not mean that having children should be entirely avoided. Your physician and social worker can provide advice about genetic counselling services and the possibility of genetic screening.

3. Open discussion, screening and treatment allow for a more positive approach to life. Those who learn about ADPKD and are screened and diagnosed with ADPKD, eventually mange to accept and cope with their diagnosis. Based on a realistic picture of the future, people learn that living an active and productive life is entirely possible. Adopting this approach reduces the fear and tension that can arise in families when ADPKD is not faced.
Counselling

If it is difficult to talk about this disease as a family, there are qualified counsellors who understand its inheritance pattern and the implications of such a diagnosis. They can help answer the questions common to all ADPKD patients.

Counselling can assist families to retain maximum productivity and acceptance whilst coping with what is a difficult and challenging situation. Counsellors can help you answer questions such as - What should I tell my partner? When should I tell my children? What if they don’t want to know? When should children be tested? Should I have a family?

It is important that patients discuss these concerns with their doctors, who can refer them to a renal social worker and/or a genetic counsellor, particularly if genetic screening is to be explored.
The Renal Resource Centre provides information and educational materials on kidney disease, dialysis and transplantation for patients and health professionals.

The primary objective of the Centre is to ensure that patients have easy access to such information, are well informed and can actively participate in their own health care. The Renal Resource Centre is committed to providing education and service to the renal community.
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