Cystic kidney disease is a group of diseases that cause abnormal pockets of clear, watery fluid (cysts) to form in the kidneys. In cystic kidney disease multiple cysts form in both kidneys. The cysts slowly replace healthy tissue causing the kidneys to enlarge.

Types of cystic kidney disease include:
- Polycystic Kidney Disease (PKD)
- Medullary Cystic Disease
- Medullary Sponge Kidney

Simple or benign cysts are non-cancerous sacs filled with clear, watery fluid. They range from small blisters to large sacs filled with several litres of fluid. Having one or more cysts is not unusual, particularly in older people. These cysts rarely need treatment and do not mean that you have cystic kidney disease.

Cysts can develop as a result of long-term kidney problems, particularly in people who have kidney failure and have been on dialysis for a long time. This is called Acquired Cystic Disease (ACKD). These cysts are not inherited and do not usually need any specific treatment. Having a cyst on your kidney does not automatically mean that you have cystic kidney disease. Having one or more cysts on your kidney is common in older people and may not require treatment.

POLYCYSTIC KIDNEY DISEASE (PKD)

PKD is the most common inherited cystic kidney disease. It is caused by genetic defects, which lead to the growth of thousands of blisters of fluid (cysts) in the kidneys.

Both kidneys are affected but one kidney may develop the cysts earlier than the other. The cysts gradually grow, making the kidneys larger and reducing healthy kidney tissue. Some people develop high blood pressure and kidney failure as a result of PKD. PKD affect males and females in equal numbers, but men usually progress faster to kidney disease. The cysts may appear at any age.

The two major inherited forms of PKD are:
- **Autosomal Dominant PKD**
  This is the most common inherited form of PKD. A parent with autosomal dominant PKD has a 50% chance in each pregnancy of passing the faulty gene and associated disease to each of their children. If a person doesn’t inherit the gene, there is no chance of their children developing the disease because it never ‘skips’ a generation.

Occasionally, a person develops the disease when there is no family history and it is thought that a different inheritance or a genetic mutation may be responsible. Special blood tests can help to find the abnormal chromosomes linked to PKD and genetic counselling is available for affected couples.

- **Autosomal Recessive PKD**
  This is a less common form of inherited PKD. Both parents have to carry this gene defect for a child to inherit this disease. If both parents are affected, they have a 25% chance of passing it on to their child.

There may have no symptoms of autosomal dominant PKD in the early stages. The cysts can start growing during the teenage years and over time make the outline of the kidneys look irregular or ‘bubbly.’
Complications usually develop at 30 – 40 years of age (but can begin earlier) and include:

- Pain in the back or side
- Blood in the urine (haematuria)
- High blood pressure – occurs early in the disease
- Enlarged and painful abdomen (belly area)
- Urinary tract infections
- Kidney stones
- Hernias
- Liver and pancreatic cysts
- Abnormal heart valves
- Aneurysms in the brain – cysts in the walls of blood vessels
- Colon problems (diverticulosis) – small cysts in the large intestines
- Reduced kidney function or kidney failure - about 50% of people with PKD will have kidney failure by age 60

For more information see ‘Blood in the Urine’, ‘Chronic Kidney Disease’, ‘Heart Disease with Chronic Kidney Disease’ and ‘Kidney Stones’ fact sheets for more information.

### SYMPTOMS OF AUTOSOMAL RECESSIVE PKD

Cysts can develop in the early months of life or even before birth. Children with autosomal recessive PKD may present any time during childhood and teenage years with reduced kidney function, which can eventually lead to kidney failure or liver problems.

Symptoms and signs in severely affected babies include:

- Reduced amniotic fluid surrounding the baby in the uterus
- An unusually shaped face due to the lack of amniotic fluid (Potter facies)
- Delayed or difficult childbirth
- Enlargement of the abdomen (belly area) due to enlarged kidneys, liver and spleen
- Heart defects and underdevelopment of the lungs
- Kidney failure at birth or in the first few weeks of life

Other complications may include:

- Failure to thrive – below average growth and weight gain as a baby/infant
- Increased blood pressure in the liver, with varicose veins around liver and bowel
- Blood in the urine (haematuria)
- High blood pressure
- Anaemia

### MEDULLARY CYSTIC DISEASE

Medullary cystic kidney disease (MCKD) is an inherited kidney disease transmitted in an autosomal dominant pattern. The kidneys gradually lose their ability to work properly because of cysts in the medulla (centre of the kidneys). It often causes kidney failure between 20 and 50 years of age. The childhood disease juvenile nephronopthisis is very similar but usually occurs in young children and is due to autosomal recessive inheritance.

In both diseases, cysts develop in the inner part of the kidney (medulla), the kidneys shrink as the cortex (outer section) thins and kidney failure begins to develop. The kidneys can’t concentrate urine properly, leading to an over production of urine, loss of sodium and dehydration. Other health problems usually follow, including anaemia, kidney bone disease, gout and high blood pressure.
MEDULLARY SPONGE KIDNEY
Medullary Sponge Kidney is a condition where cysts develop in the kidney’s urine collecting ducts and tubules. The exact cause of Medullary Sponge Kidney is not known. One or both kidneys may be affected. Complications include blood in the urine (haematuria), calcium deposits in the kidneys, kidney stones or infections. Kidney failure is rare but can develop as a result of repeated infections or kidney stones.

HOW IS CYSTIC KIDNEY DISEASE DIAGNOSED?
The severe symptoms of autosomal recessive PKD usually result in a prompt diagnosis. However, in most other cases of cystic kidney disease, physical health is not affected for many years. Physical check-ups, blood and urine tests or even ultrasound scans may not pick it up in the early stages. It is often detected during medical investigations for other health problems, such as urinary tract stones or infections. Sometimes it isn’t discovered until high blood pressure or kidney failure develops.

Diagnosing cystic kidney disease can involve a number of tests including:
- Physical examination – may detect high blood pressure or enlarged kidneys
- Blood tests – assess kidney function
- Urinalysis – blood and/or protein may be found in the urine
- Ultrasound – a simple test which uses sound waves to detect cysts in the kidneys. It is good at identifying even quite small cysts
- Computed Tomography (CT) and Magnetic Resonance (MRI) scans can detect very small cysts. They may be required if the results from the ultrasound are inconclusive or more information is needed
- Genetic testing – generally used for family studies and not a routine test

HOW IS CYSTIC KIDNEY DISEASE TREATED?
There is no cure for the three types of cystic kidney disease. However, early detection and proper treatment can reduce or prevent some of the complications and maintain kidney function.

For more information about Kidney or Urinary health, please contact our free call Kidney Health Information Service (KHIS) on 1800 454 363. Alternatively you may wish to email KHIS@kidney.org.au or visit our website www.kidney.org.au to access free health literature.

This is intended as a general introduction to this topic and is not meant to substitute for your doctor’s or Health Professional’s advice. All care is taken to ensure that the information is relevant to the reader and applicable to each state in Australia. It should be noted that Kidney Health Australia recognises that each person’s experience is individual and that variations do occur in treatment and management due to personal circumstances, the health professional and the state one lives in. Should you require further information always consult your doctor or health professional.

Revised Sept 2013