Kidneys - medullary cystic kidney disease

Summary

- Medullary cystic kidney disease (MCKD) is an inherited condition that causes the growth of abnormal blisters of fluid (cysts) in the kidneys.
- MCKD often causes kidney failure in people between 20 and 50 years of age.
- There is no cure – treatment aims to manage the symptoms and reduce the risk of complications.

Medullary cystic kidney disease (MCKD) is an inherited condition. It belongs to a group of diseases known as cystic kidney disease. Genetic changes cause abnormal blisters of fluid (cysts) to grow in the kidneys.

The cysts develop in the inner part (medulla) of the kidney. Scar tissue forms and the kidneys shrink as the outer section (cortex) thins. The kidneys lose their ability to remove enough fluid from filtered wastes and pass great amounts of undiluted urine. There is no cure. This condition often causes kidney failure in people between 20 and 50 years of age.

Symptoms of MCKD

Symptoms usually begin before the age of 20, but they vary greatly. A few people do not have any symptoms until much later in life.

Symptoms of MCKD may include:

- excessive urination, caused by the reduced ability of the diseased kidneys to respond to antidiuretic (fluid-holding) hormones
- getting up in the night to urinate
- low blood pressure
- unexplained weight loss
- generally feeling unwell
- nausea and vomiting
- headache
- gout
- skin changes, such as pale or yellowish skin tones and easy bruising
- muscle cramps or twitches
- retarded growth in children.

Complications of MCKD

Some of the complications associated with MCKD include:

- anaemia
- ulcers and bleeding of the gastrointestinal tract
- loss of bone density and increased risk of fractures
- infertility
- end-stage kidney disease
- high blood pressure (caused by the failing kidneys)
- heart problems, including pericarditis or congestive heart failure
- haemorrhage (excessive bleeding)
- changes in glucose metabolism
- infertility
- menstrual problems
- miscarriage
- peripheral neuropathy (damage to the nerves of the peripheral nervous system)
- easy bruising (which can signify platelet dysfunction)
- skin colour changes.

**Pattern of inheritance of MCKD**

MCKD is an inherited disease transmitted in an 'autosomal dominant' pattern, which means that the condition will occur even if the genetic changes are inherited from only one parent. The condition can occasionally occur when there is no family history of the disease.

The childhood disease 'juvenile nephronophthisis' is similar to MCKD, but usually occurs in young children and is due to an inherited recessive genetic condition. This means that the child must receive the genetic changes from both parents in order to develop the condition.

**Diagnosis of MCKD**

Tests to diagnose MCKD may include:

- physical checks – your doctor may detect high blood pressure or enlarged kidneys
- blood tests – to check how your kidneys are working to filter your blood and remove wastes
- urine tests – may detect blood or protein in the urine, or both
- x-rays – to check the size of the kidneys
- ultrasound – this method is good at identifying even quite small cysts
- computed tomography (CT) and magnetic resonance imaging (MRI) scans – can detect very small cysts. They may be required if the results from the ultrasound are not clear or more information is needed
- renal biopsy – a small piece of kidney tissue is surgically removed and sent to a laboratory for analysis
- genetic testing – generally only used if cystic kidney disease runs in your family.

**Treatment for MCKD**

MCKD is a slow, progressive disease. There is no cure. Treatment aims to manage the symptoms and decrease the risk of complications. Treatment options include:

- medication and lifestyle changes to control high blood pressure and manage pain
- dietary changes – these may include staying adequately hydrated and reducing salt, protein, cholesterol (fats) and caffeine. Any changes should be made after talking with your doctor or dietitian
- avoiding contact sports – if your kidneys are very enlarged, as a strong knock to your stomach area can cause damage.

Everyone with MCKD will eventually need treatment for kidney failure. Treatment options include:

- dialysis
- kidney transplantation
- supportive care.

**Where to get help**

- Your doctor
- **Kidney Health Australia Information Service. Tel. 1800 454 363**