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## Kidneys - medullary cystic kidney disease

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### Summary

- Medullary cystic kidney disease (MCKD) is an inherited condition that causes the growth of 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.
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### What is medullary cystic kidney disease (MCKD)?

Medullary cystic kidney disease (MCKD) is an inherited condition. It belongs to a group of diseases known as cystic kidney disease. Genetic changes cause 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.

### What causes MCKD?

Medullary cystic kidney disease is caused by changes in the MUC1 gene. This gene provides instructions for making a protein called mucin 1. Changes in the gene lead to the production of an altered mucin protein. It is not known how this change causes MCKD.

### Pattern of inheritance of MCKD

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

### Symptoms of MCKD

Symptoms of MCKD usually begin before the age of 20, but they vary greatly. Some 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
  - 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
  - growth delay in children.
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## 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.

## 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. Talk with your doctor or dietitian before making any dietary changes
- avoiding contact sports – this is recommended if your kidneys are very enlarged, as a strong knock to your stomach area could cause damage.

Currently, everyone with MCKD will eventually need treatment for kidney failure. Treatment options include:

- dialysis
- kidney transplantation
- supportive care.

## Genetic counselling and support for MCKD

If you or a family member have been diagnosed with MCKD, or if MCKD runs in your family, it can be helpful to

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**[betterhealth.vic.gov.au](http://betterhealth.vic.gov.au)**

Speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand MCKD and what causes it, how it is inherited, and what a diagnosis means for your or your child's health, development and lifestyle. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

A genetic counsellor can explain what genetic testing options are available to you and other family members thinking about being tested for MCKD.

You may also choose to visit a genetic counsellor if you are planning a family, to find out your risk of passing on MCKD to your child, or to arrange for prenatal tests.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by MCKD.

### **Where to get help**

- Your **GP (doctor)**
- **Kidney Health Australia** Kidney Helpline Tel. 1800 454 363
- **Victorian Clinical Genetics Services (VCGS)**, Royal Children's Hospital, Tel. 1300 118 247
- **Genetic Support Network of Victoria (GSNV)** Tel. (03) 8341 6315

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Kidney Health Australia

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