MUC1 & Medullary Cystic Kidney Disease Type 1

What is medullary cystic kidney disease type 1?

- Medullary cystic kidney disease type 1 (MCKD1) is an inherited condition caused by changes in the MUC1 gene.
- It leads to scarring of the kidneys and gradual loss of kidney function.
- This declining kidney function leads to signs and symptoms such as high blood pressure and gout. However, these signs vary, even among family members and may not occur at all.

What is MUC1, and how do changes in MUC1 affect the kidneys?

- The MUC1 gene is believed to play a role in the growth, movement and survival of cells, as well as the normal development of the kidney.
- Changes in the gene result in the production of proteins that do not function properly. It is not yet clear how this change causes disease.
- However, it does result in scarring of the kidneys and loss of function.
- Only about 40% of affected individuals have medullary cysts, which are fluid-filled sacs within the kidney.
- The decline in kidney function generally appears in the teenage years, but the age at which failure occurs is very variable.
    - In some families, failure occurred before 30 years, in others, it occurred as late as 70 years.

Do these changes have effects on other parts of the body?

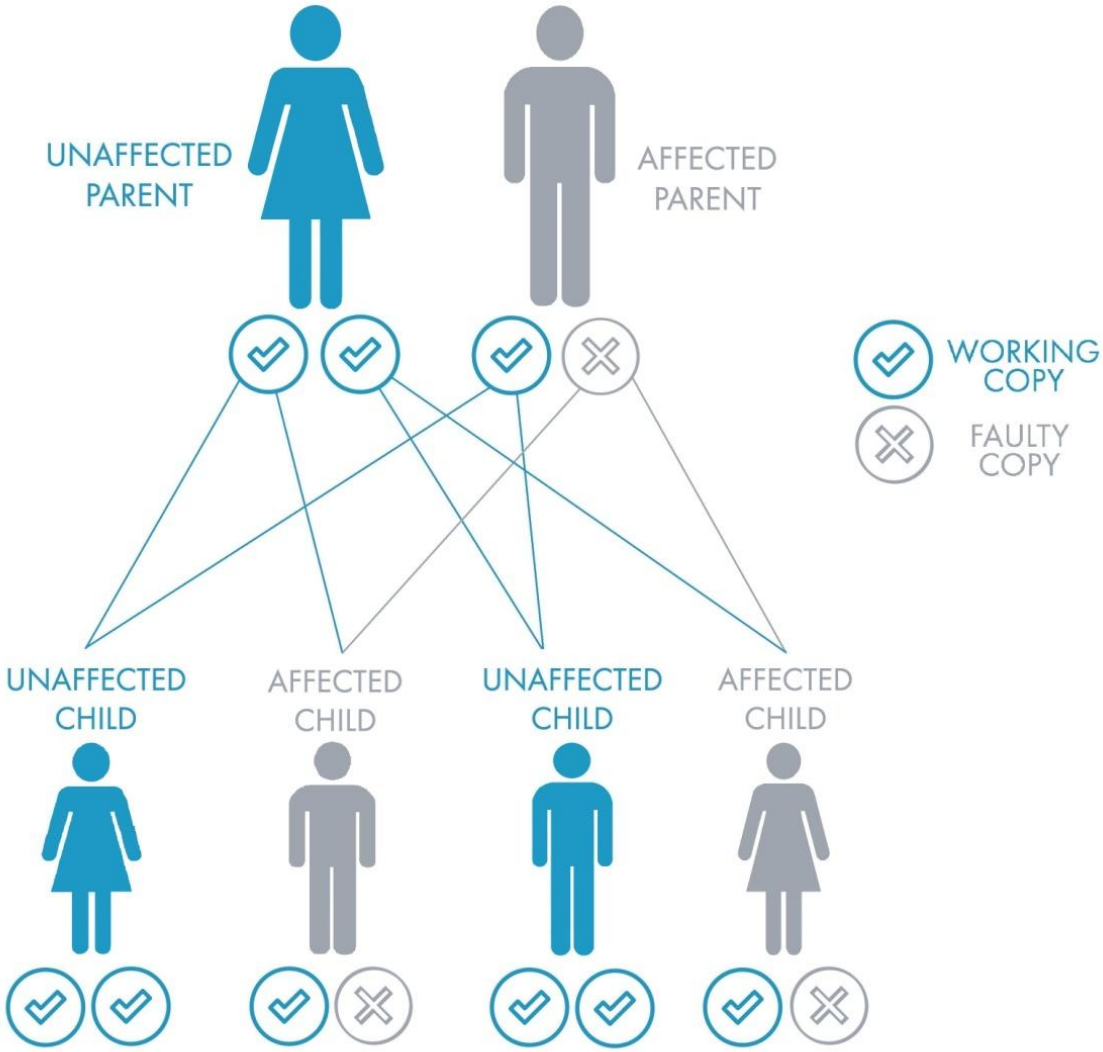
- Changes in MUC1 do not appear to have effects outside the kidney.

How is medullary cystic kidney disease treated?

- There is no specific therapy for Medullary Cystic Kidney Disease. Treatment is focused on managing complications of kidney function loss such as anaemia.
- Outcomes of kidney transplants are generally good as the disease will not develop again in the new kidney.

How is this change passed down through a family?

- You have two copies of MUC1 – one from each of your parents.
- To have MCKD1 you must inherit at least one faulty copy of the MUC1 gene from an affected parent.
- Each child of an affected parent has a 1 in 2 (50%) chance of inheriting the disease.



Should my family members be tested?

- Family members may be advised to undergo genetic testing, especially as early diagnosis and treatment can significantly improve outcomes.
- It is recommended to have a discussion with a genetic counsellor prior to undergoing testing.