

Information about your genetic test result

Your nephrologist organised a genetic test for you. This test analyses genes associated with kidney disease. Please read this document carefully and see your nephrologist if you have any questions.

Your genetic test result

The test **has not found** a disease-causing change (also known as a *pathogenic variant*) in the genes that were tested. This means no genetic diagnosis was made. However, a change was found in the _____ gene. This is currently described as a **variant of uncertain significance (VUS)**.

What is a variant of uncertain significance (VUS) and what does this mean for you?

- Variants (or changes) in genes are common. They are why each person is unique. When a variant is very unlikely to cause disease, it is called a *benign variant*. When it is very likely to cause disease, it is called a *pathogenic variant*.
- A **variant of uncertain significance (VUS)** is a change that medical professionals cannot confidently say is benign or pathogenic. As we learn more about genes over time, many VUS are reclassified as either benign or pathogenic.
- Currently we do not know whether the VUS found in your genetic test is the cause of your kidney disease, or if it is a normal change.
- There is also a chance that your kidney disease is caused by genetic changes that could not be identified with this test. Research helps us identify new genetic causes of kidney disease and ways to test for them.

- **Follow-up:** It is recommended that you ask your nephrologist for a genetics review in 3–5 years. By then, there may be new information about your VUS and/or whether further testing should be considered. In some situations, further testing may be indicated to help clarify your genetic test result. If this is the case, your nephrologist may refer you to a multidisciplinary renal genetics clinic.

What could this mean for your family?

As the VUS currently does not explain your kidney disease, we **do not** recommend your relatives have genetic testing for this variant to inform their medical care. However, if you have relatives with kidney disease, it may still be relevant for them to have genetic testing. They should discuss this with their nephrologist.

Where to get further information

A genetic counsellor may be able to discuss further options or answer questions about genetic testing. Your doctor or nephrologist can make a referral to a genetics service.