

Information about your genetic testing result

Your nephrologist organised a genetic test for you. This information sheet explains your test result and what it might mean for your health and your family members.

This document should not replace the advice of your relevant healthcare professional. Please read it carefully and ask your nephrologist if you have any questions.

Your genetic test result

The test found a disease-causing change (also known as a *pathogenic variant*) in the *COL4A3* or *COL4A4* gene. This causes **Autosomal Dominant Alport Syndrome (ADAS)**.

In the past, ADAS was called 'thin basement membrane nephropathy' or 'benign familial haematuria'.

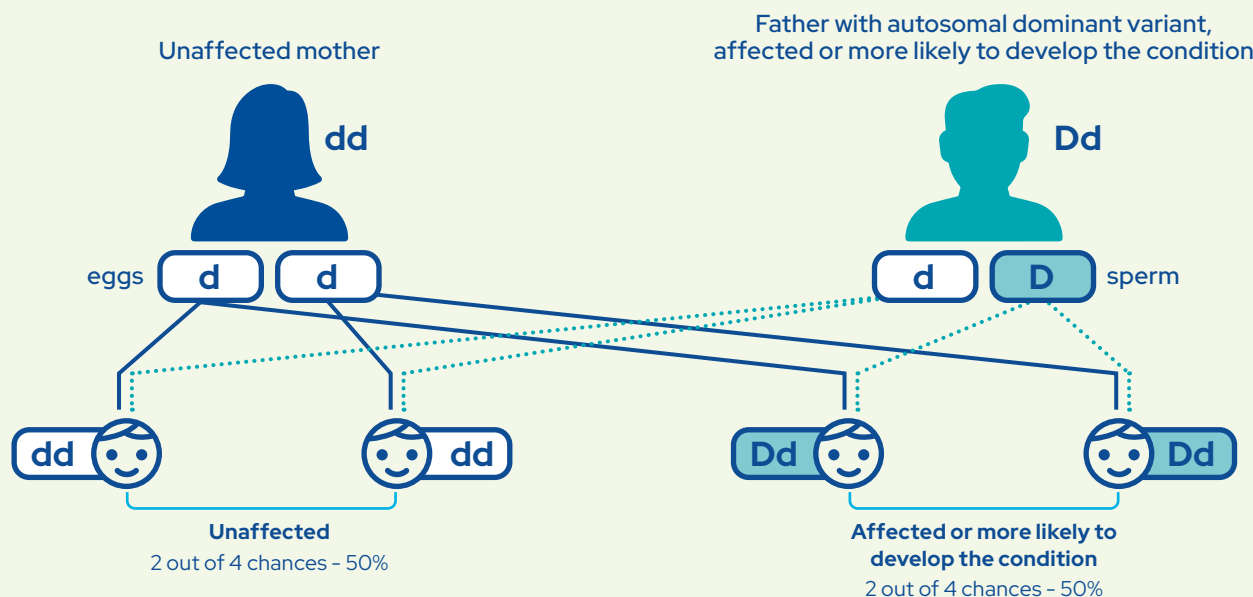
About Autosomal Dominant Alport Syndrome (ADAS)

- In general, kidney involvement in people with ADAS is variable. It can range from no features at all, to isolated blood in the urine that is only visible under the microscope (microscopic haematuria), sometimes with protein in the urine (proteinuria), abnormal kidney function and kidney failure.
- Most people with ADAS will not experience kidney failure, however the risk of this is higher compared to the general population. If kidney failure occurs, it usually develops after the age of 60 years and may be partly due to other medical conditions which can also affect the kidneys.
- ADAS is not typically associated with hearing or vision problems (which can occur with other forms of Alport syndrome). Therefore, you do not usually need screening for these.

What does this mean for you?

Your nephrologist will help you manage this condition and discuss specific treatment based on your genetic test result.

- You should have an annual (yearly) screen that includes monitoring of blood pressure, urine test looking for protein and/or blood in the urine, and blood test to check kidney function.
- If you develop protein in your urine, your nephrologist may prescribe a medication called an *angiotensin-converting enzyme (ACE) inhibitor* or *angiotensin II receptor blocker (ARB)*. These medications also help to control your blood pressure.
- Lifestyle modifications to reduce other risk factors for kidney disease are recommended (such as avoiding smoking, maintaining a healthy weight, and blood pressure control).



This diagram shows how the disease-causing change can be passed from parents to children. The non-working gene copy with an autosomal dominant variant is shown as 'D'; the working copy of the gene by 'd'. Source: genetics.edu.au

What could this mean for your family?

We all have two copies of the COL4A3/4 gene – one copy inherited from each of our parents.

You have ADAS because one copy of your COL4A3/4 genes has a disease-causing change in it. Most people inherit this change from one of their parents, but for some people, it can happen for the first time at conception.

First-degree family members (like your siblings or one of your parents) may also have this gene change. We recommend that you share this information with your family members, so they have the option to get more information on the condition and/or consider genetic testing for themselves.

If you have children (or plan to have children)

Your children have a 1 in 2 (50%) chance of inheriting the gene change that causes ADAS.

- If your partner also carries a COL4A3/4 disease-causing change, there is a chance that you may have a child with autosomal recessive Alport syndrome (ARAS). This is a more severe form of Alport Syndrome, with usual onset in childhood. The chance of this is low (less than 1/1000).
- It is important to discuss your diagnosis with your children as they reach adulthood. This will help

your children to understand the implications for their health management and family planning, and decide whether to get genetic testing.

- If you (or other family members) are **considering having children in the future**, there are options to reduce the risk of passing on a genetic condition. Please refer to the information sheet **Reproductive genetic testing options**.
- If you are **pregnant or you and your partner are currently planning for pregnancy**, please let your nephrologist know so they can arrange an expedited referral to a genetics service if you wish.

Where to get further information

A genetic counsellor may be able to:

- support you to better understand your result
- support you to share your result and this information with your family members
- discuss ways to reduce the risk of passing on a genetic condition to future children

Your doctor or nephrologist can make a referral to a genetics service. You may also be able to find a genetic counsellor in your area by emailing kidneygenetics@monashhealth.org.

You may also find useful information on the Alport Foundation of Australia's website. Visit alport.org.au.