

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 see your nephrologist if you have any questions.

Your genetic test result

The test found two disease-causing changes (also known as *pathogenic variants*) in the *COL4A3* or *COL4A4* genes. This causes a condition known as **Autosomal Recessive Alport Syndrome (ARAS)**.

About Autosomal Recessive Alport Syndrome (ARAS)

Most people with ARAS will develop kidney failure in early adulthood. Many people have symptoms in childhood, such as hearing abnormalities, abnormalities in their urine, and kidney disease.

ARAS can also be associated with eye abnormalities, but these rarely affect vision.

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 regular blood and urine tests and review by a nephrologist.
- You should also start a medication called an *angiotensin-converting enzyme (ACE) inhibitor* or *angiotensin II receptor blocker (ARB)*. This medication delays kidney failure and also helps to control your blood pressure.

- Lifestyle modifications to reduce other risk factors for kidney disease are also recommended (such as avoiding smoking, maintaining a healthy weight, and blood pressure control).
- There may be clinical trials available for people with ARAS, and your nephrologist may discuss if there are any available clinical trials relevant to you.

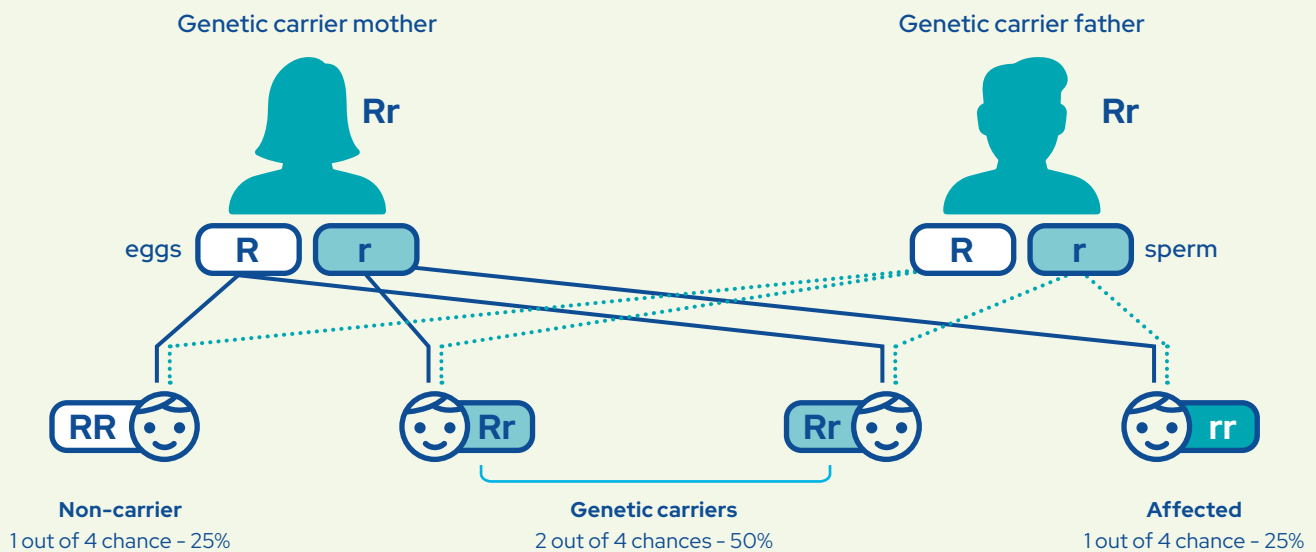
What could this mean for your family?

Your diagnosis of ARAS has important implications for family members. We all have two copies of the *COL4A3/4* gene – one copy inherited from each of our parents.

It is most likely you have ARAS because BOTH copies of your *COL4A3/4* genes have the disease-causing change (or variant). Most people inherit one *COL4A3/4* gene variant from each of their parents, and further testing in your parents or other family members may be organised to confirm this.

All first-degree relatives (like your siblings, parents and children) will be expected to have a variant in at least one copy of the *COL4A3/4* gene. Your siblings may also have ARAS.

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.



This diagram shows how the disease-causing change can be passed from parents to children. The non-working copy of the gene containing a recessive variant is represented by 'r'; the working copy of the gene by 'R'. Source: genetics.edu.au

If you have children (or plan to have children)

All your children will inherit ONE of your COL4A3/4 gene variants.

- Having one copy of the COL4A3/4 gene variant results in a condition called Autosomal Dominant Alport Syndrome (ADAS). However this is a milder form of kidney disease, and symptoms do not appear until later in life.
- If your partner also carries a COL4A3/4 pathogenic variant, there is a chance that you may have a child with ARAS. The chances of this is small (less than 1 in 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 these online resources useful:

- Alport Foundation of Australia – alport.org.au
- [Fact sheet – Autosomal recessive inheritance](http://genetics.edu.au) – at genetics.edu.au