XLAS - Male

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 a disease-causing change (also known as a *pathogenic variant*) in the *COL4A5* gene. This causes a condition known as **X-linked Alport Syndrome (XLAS)**.

About X-linked Alport Syndrome (XLAS)

There are different kinds of Alport Syndrome. Your condition (XLAS) affects males and females differently.

- In males, XLAS leads to progressive kidney damage. Most males with XLAS will develop kidney failure in early adulthood.
- Males typically develop symptoms of XLAS in childhood, such as hearing loss, eye abnormalities, blood and/or protein in their urine, and kidney disease. However, symptoms can vary even among members of the same family.

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.
- Your nephrologist may prescribe a medication called an *angiotensin-converting enzyme (ACE) inhibitor or angiotensin II receptor blocker (ARB)*. These medications delay kidney failure and 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).
- There may be clinical trials available for males with X-linked Alport syndrome. Your nephrologist may discuss if there are any available clinical trials relevant to you.

What could this mean for your family?

Your diagnosis of XLAS is likely to have important implications for family members.

XLAS is caused by changes (or variants) in the *COL4A5* gene located on the X chromosome. Females have two X chromosomes (XX) and are typically less severely affected by XLAS than males. Males have an X and a Y chromosome (XY) and typically have more severe symptoms.

It is most likely that you inherited the *COL4A5* gene variant from your mother, but sometimes the variant occurs for the first time in an individual and is not inherited.

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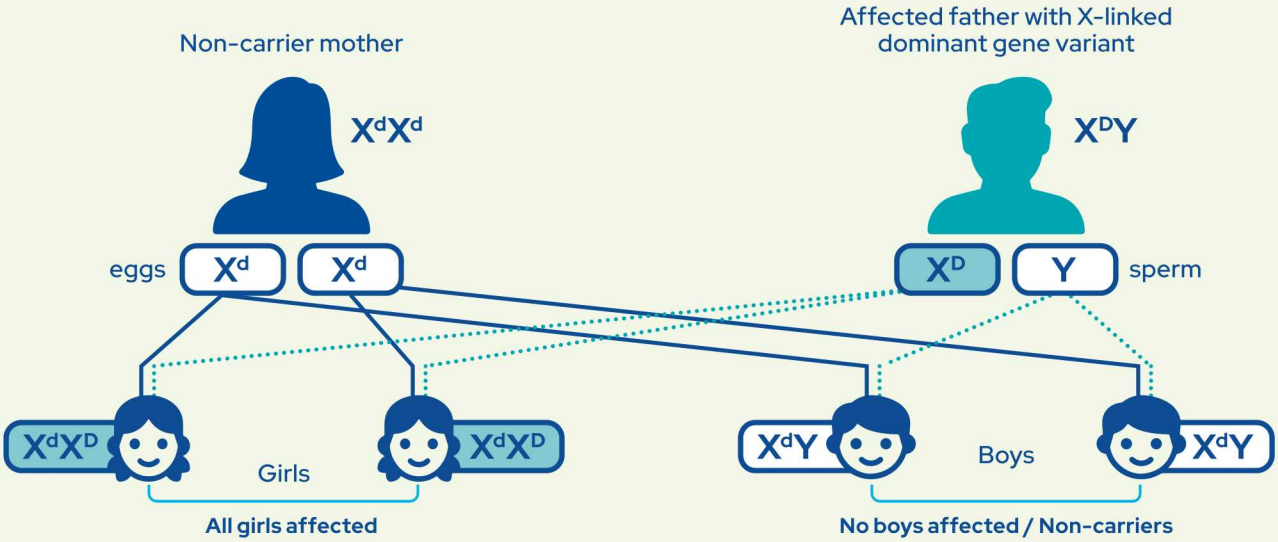
This information sheet was produced through the Scaling Up Genomics in Nephrology project funded by the Melbourne Genomics Health Alliance. Visit **kidneygenomics.org.au** for more information.



This diagram shows how an X-linked dominant variant can be passed from parents to children. Source: genetics.edu.au.

First-degree family members (like your siblings or your mother) may 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 female children will inherit the *COL4A5* variant that causes XLAS, but your male children will not.

- All your daughters will have inherited your *COL4A5* variant. However, females with XLAS are typically less severely affected than males. Many women with XLAS will not experience kidney failure, although the risk of developing kidney failure is still higher compared to the general population.
- It is important to know that females with XLAS can pass the condition to their male children.
- Your sons will not inherit the *COL4A5* variant from you. This is because they inherit their Y chromosome from you, and their X chromosome from their mother. This condition is passed on the X chromosome.
- If you have children, please inform your nephrologist.
- 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 – X-linked dominant inheritance – at **genetics.edu.au**

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