

# 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 *PKD1* or *PKD2* gene. This causes a condition known as **autosomal dominant polycystic kidney disease (ADPKD)**.

### About Autosomal Dominant Polycystic Kidney Disease (ADPKD)

ADPKD mainly affects the kidneys, but it can also cause cysts in the liver (and other organs). About half of all people with ADPKD develop kidney failure by the age of 60 – **but not all people do**.

Many things can influence what symptoms develop, and whether or not you will have kidney failure – including which gene is changed (*PKD1* or *PKD2*) and the specific type of change to the gene.

## What does this mean for you?

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

- Management will likely include lifestyle modifications and medications to reduce your blood pressure.

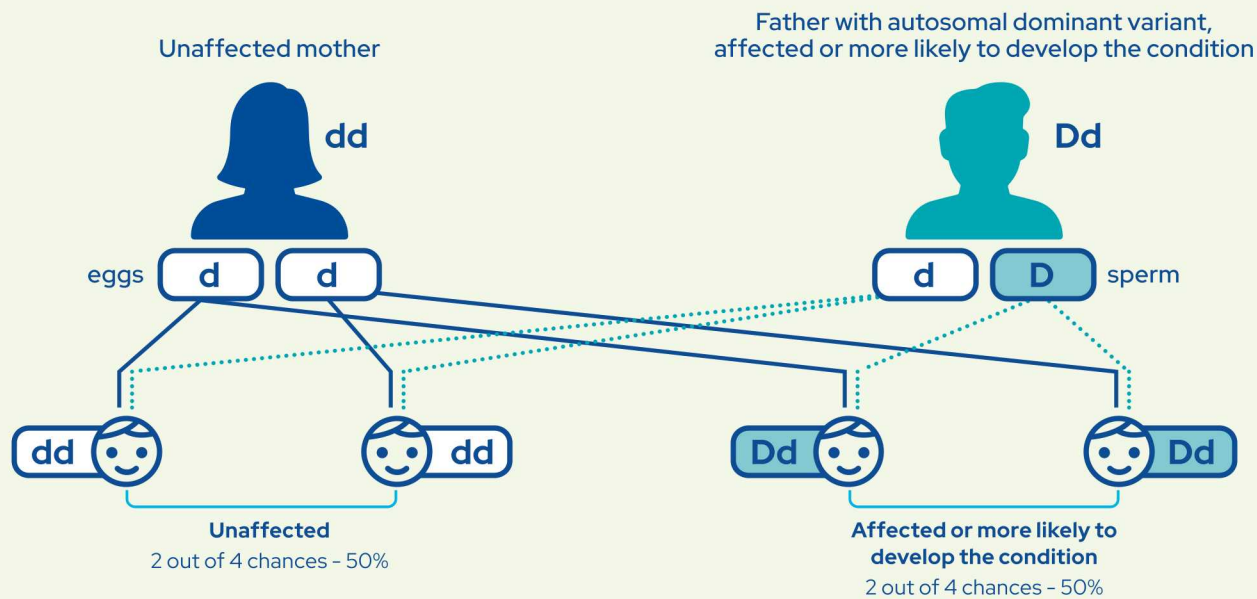
- A medication called *tolvaptan* can sometimes help to slow the progression of kidney disease. Your nephrology team will discuss this if it can benefit you.
- People with ADPKD have an increased risk of having a brain aneurysm. Your nephrologist can discuss screening recommendations with you (this may involve a brain MRI).

## What could this mean for your family?

We all have two copies of the *PKD1* and *PKD2* genes – one copy inherited from each of our parents.

**You have ADPKD because one copy of your *PKD1* or *PKD2* 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.



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

### 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 ADPKD.

- ADPKD is considered an adult condition because most children will not have any symptoms or kidney problems.
- Some children with ADPKD will develop high blood pressure, so your children should have a blood pressure check each year with their GP, paediatrician or nephrologist.
- It is generally recommended to wait until your children are in early adulthood to do genetic testing for ADPKD.
- It is important to discuss your diagnosis with your children as they get older. 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](mailto:kidneygenetics@monashhealth.org).

You may also find these online resources useful:

- [Fact sheet – Polycystic Kidney Disease](#) - on [kidney.org.au](http://kidney.org.au)
- Website for PKD Australia – [pkdaustralia.org](http://pkdaustralia.org)