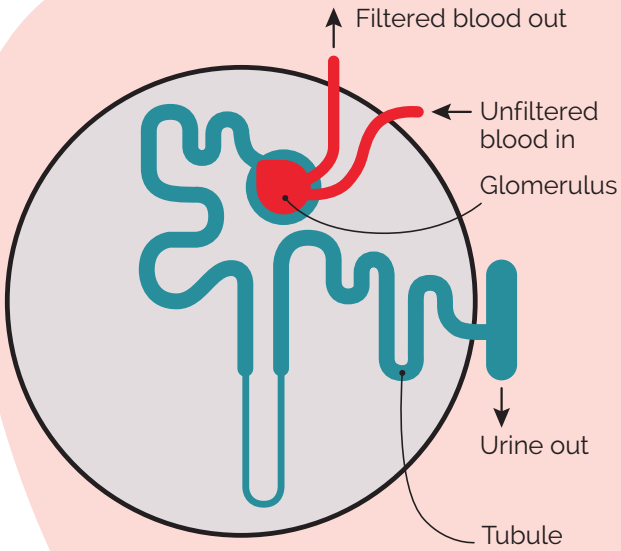
Kidney Health® Australia

Rare kidney disease

What are the kidneys?

The kidneys are the unsung heroes of your body. Each kidney is about the size of a fist, bean-shaped, and located under the rib cage on either side of your spine.

Each kidney contains up to one million tiny filtering units, called nephrons. Inside a nephron, there is a tiny set of blood vessels called the glomerulus. The glomerulus filters your blood allowing extra fluid and waste to pass into your urine (wee). Clean blood flows back to the rest of your body.



What are rare kidney diseases?

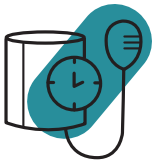
A disease is rare if it affects less than 5 in 10,000 people in Australia. Because they are rare, it may take a while for a doctor to discover that you have a rare disease.

Most rare kidney diseases are genetic or inherited, which can be passed down from your parents through their genes. Some rare diseases affect your immune system - these are called autoimmune diseases.

There are many rare diseases that can affect your kidneys, making it harder for them to filter waste and extra fluid from your blood.

How does my doctor find out if I have a rare kidney disease?

Your doctor will order certain tests to determine if you have a rare kidney disease. First, the doctor will check for kidney disease. A **Kidney Health Check** is a quick and simple way to check for kidney disease. It includes:



A **blood pressure check** to see if you have high blood pressure. Kidney disease can cause your blood pressure to increase.

A **urine test**, to look for blood or a type of protein, called albumin, in your urine. Albumin or blood in the urine can be a sign of kidney damage.

A **blood test** to check your kidney function. The estimated glomerular filtration rate (eGFR) measures how well your kidneys are filtering blood.

The doctor may order other tests to find out if kidney damage is due to a rare disease. These tests may include:

- A **kidney biopsy** to take a small sample of your kidney tissue for examination under a microscope.
- A **genetic test** to screen for changes in your genes. The laboratory takes a sample of your spit (also called saliva) or blood to test for genetic mistakes.
- An **ultrasound imaging test** to scan the inside structures of your kidneys.

Symptoms of kidney disease

Rare kidney diseases can have different symptoms for different people. Some may affect other organs, like the skin, eyes, lungs, or heart.

If the disease slowly affects the kidneys, you may not notice symptoms. You can **lose up to 90 per cent** of your kidney function before you experience symptoms.

Some symptoms of kidney disease include:

Low energy

Swelling of the feet or ankles

Trouble sleeping

Feeling sick

Itchy skin

Trouble breathing

Constipation or diarrhoea

Depressed mood

What are genetic kidney diseases?

Genes are the instructions for your body's traits, such as eye colour. Your genes are passed down (inherited) from your parents.

Genetic kidney diseases are caused by changes (mutations) in your genes. When a gene contains a mutation, it stops working like it is meant to. Gene mutations can be inherited or happen by chance.

Alport syndrome

Alport syndrome is a genetic (inherited) condition that affects the kidneys, hearing, and eyesight. Alport syndrome affects how the body makes type IV collagen, an important protein found in the filters of your kidneys.

While there is no cure for Alport syndrome, the doctor may recommend medicines to help control blood pressure and protect the kidneys. You may need regular eye checks and hearing tests depending on the type of Alport syndrome.

Download our **Alport syndrome** factsheet to learn more.

Fabry disease

Fabry disease affects an enzyme that breaks down fat in the body. When this enzyme does not work properly, it can cause certain fats to build up in the kidneys, heart, and nerves. Symptoms include nerve pain, stomach pain, and less sweating. Some people develop chronic kidney disease and/or heart disease.

Your doctor can give special medicines to replace the enzyme affected by Fabry disease. These medicines help to protect the kidneys and slow down kidney disease and failure. In addition, your doctor can prescribe treatments to help control the symptoms of Fabry disease.

Polycystic kidney disease

Polycystic kidney disease (PKD) is the most common type of genetic kidney disease. PKD causes fluid-filled sacs, called cysts, to grow in the kidneys. The cysts grow slowly and make it harder for your kidneys to do their job. There are two different types:

- **Autosomal Dominant PKD (ADPKD)** affects 1 in 1000 people and therefore is a common genetic kidney disease. ADPKD may cause cysts to form in the kidneys, liver, and pancreas. Many people do not find out they have ADPKD until adulthood unless they know the condition runs in their family.
- **Autosomal Recessive PKD** is rare and is often discovered in childhood or even at birth. It can affect how well the kidneys work and may affect the liver.

Check out our factsheet, **Polycystic kidney disease** for more information.

What are autoimmune kidney diseases?

Your immune system is your body's defense against illness. Your immune system protects your body against infections by making antibodies, which fight germs.

Autoimmune disease occurs when your immune system mistakenly attacks your own healthy tissues. The immune system does not work properly, and the antibodies cause damage to your own cells.

Some autoimmune diseases affect the kidney. Antibodies attack healthy kidney tissue leading to swelling (inflammation) and damage. This makes it harder for the kidneys to filter waste and clean your blood.

Most autoimmune diseases happen when your body overreacts to germs. Some of the autoimmune diseases affecting the kidney can be linked to genetic factors.

IgA nephropathy

IgA nephropathy leads to kidney damage when IgA antibodies get caught in the glomeruli (filters) of the kidneys.

IgA nephropathy is a type of chronic kidney disease that may slowly worsen over 10 to 20 years.

Check out our factsheet on IgA nephropathy.

Lupus nephritis

Systemic Lupus Erythematosus, often called lupus, is a disease that causes inflammation in many different parts of your body, including your skin, joints, kidneys, lungs and brain.

Lupus nephritis is when lupus affects the kidneys, causing swelling and scarring. While there is no cure, medicines can help treat the symptoms of lupus.

Learn more by downloading the Lupus nephritis factsheet.

Complement 3 glomerulopathy

Complement 3 glomerulopathy (C3G) causes a protein called C3 to clog the filters of the kidneys. This makes it harder for the kidneys to do their job. It can cause kidney failure in about half of people within 10 years of diagnosis.

While there is no cure for C3G, the doctor may prescribe blood pressure medicines and cholesterol-lowering medicines to help protect the kidneys. Scientists are studying new medicines to help treat C3G.

Focal segmental glomerulosclerosis

Focal segmental glomerulosclerosis (FSGS) is a kidney condition that causes scarring (called sclerosis) on small sections of the glomerulus. This affects the ability of the kidneys to work as they should. FSGS can affect both adults and children. It is slightly more common in males than females.

Primary FSGS is when the disease happens on its own without an obvious cause. Secondary FSGS is when the disease is caused by another disease, most commonly diabetes, an infection, or a drug. Familial FSGS is a rare form caused by genetics.

How can I look after myself with a rare kidney disease?

The general principles of a healthy lifestyle can help your kidneys when you have a rare disease. Taking care of your health can improve your overall wellbeing. Follow these tips:

Take prescribed medicines. Follow your doctors' instructions and take your medicines as prescribed.

Limit salt. Ditch your saltshaker and limit processed foods.

Eat a healthy diet. Choose whole grains, lean proteins, vegetables and fruits over processed and fast food.

Choose water. Limit sugar-sweetened cola, juices, and lattes – drink water instead.

Exercise. Aim for 150 - 300 minutes of exercise per week.

Be a non-smoker. If you smoke or vape, you can call the **Quitline** at **13 78 48**.

Keep blood pressure under control. If you have high blood pressure, you may need medicines or lifestyle changes to control your blood pressure.

Support is available

Being diagnosed with rare disease can feel overwhelming. You or your family members may feel sad, angry, or hopeless. Help is available. Speaking to a trusted healthcare professional can help, such as your doctor or a social worker.

Know that you're not alone on your kidney health journey. **Kidney Health 4 Life**, a free wellness program and support service, can help connect you to others with kidney disease who may understand what you are going through.

Whether you're newly diagnosed or struggling through your treatments, our peer support services offer a safe space to share experiences, ask questions, and find encouragement.

Visit **kidneyhealth4life.org.au** to learn more.

Feeling overwhelmed and need immediate help?
Call **Beyond Blue** support line at **1300 224 636**
or call **Lifeline** at **13 11 14**.

Things to remember:

- ✓ **Rare kidney disease affects less than 5 in 10,000 people.**
- ✓ **Genetic kidney disease can be passed down from your parents. Autoimmune kidney disease is due to an overreaction of your immune system.**
- ✓ **Your doctor can order a Kidney Health Check and other tests, like genetics tests or a kidney biopsy, to help find rare kidney diseases.**

What does that word mean?

ACE inhibitors – A common medication prescribed by doctors to manage blood pressure.

Albuminuria – Occurs when albumin is present in the urine. There are filters in the kidneys that prevent large molecules, such as albumin, from passing through. If these filters are damaged, albumin passes from the blood into the urine.

Antibody – A protein molecule made by your immune system to attack tissue that is not normally part of your body (e.g. viruses and bacteria).

Angiotensin receptor blockers (ARBs) – A common medication prescribed by doctors to control blood pressure.

Glomerulus – One of the key structures that make up the nephron which is the filtering unit of the kidney.

Kidney biopsy – A small piece of kidney tissue is removed for testing and examined under a microscope.

Nephrologist – A doctor who specialises in treating conditions of the kidney.

Nephron – The tiny parts of the kidney that filter blood to make urine. There are over one million filters in each kidney.

Special Thanks! **This educational resource is supported by a sponsorship provided by Novartis**

Kidney Health Australia

Free Kidney Helpline 1800 454 363
kidney.org.au

If you have a hearing or speech impairment, contact the National Relay Service on 1800 555 677 or relayservice.com.au
Have them connect you to the Free Kidney Helpline - 1800 454 363

KIDNEY HEALTH 4 LIFE®

WANT TO LEARN MORE?

Kidney Health 4 Life is a health and wellbeing program equipping people, and those that care for them, with the knowledge and resources to take more active management of their kidney health or kidney disease.

Join Kidney Health 4 Life by scanning the QR code