Alport syndrome

What is Alport syndrome?

Alport syndrome is a genetic (inherited) condition that affects the kidneys, hearing, and eyesight. It can cause kidneys to lose function over time. People are born with Alport syndrome but may not find out they have it until later in life.

How does Alport syndrome affect the body?

Your body is held together by connective tissue, which contains proteins called collagen. Collagen helps your organs work properly.

The protein known as type IV collagen is found in the filters of your kidneys. Your kidneys need collagen to properly filter waste from your body.

In Alport syndrome, changes in your genes affect how type IV collagen is made. Faulty type IV collagen makes it harder for your body to filter waste. It can also cause problems with hearing and eyesight.

How do I get Alport syndrome?

Alport syndrome is usually inherited through genes passed down from your parents. Genes are like instructions for your body and determine traits - like the colour of your eyes.

Genes are grouped together on chromosomes. Most people have two copies of each chromosome, one passed down from each parent:

- Females have 2 X chromosomes.
- Males have one X and one Y chromosome.

Sometimes, a gene contains a variation – like a spelling mistake – that stops it working like it is meant to. A variation can be passed down from a parent or happen by chance. When a gene does not work as expected, it can lead to genetic conditions like Alport syndrome.

Types of Alport syndrome

There are three main types of Alport syndrome:

X-linked Alport syndrome

This is the most common type, passed down through an X chromosome. 8 of every 10 people with Alport syndrome have this type. Males often have more symptoms than females because they only have one X chromosome. Females may have milder symptoms, but some can still have health problems.

Autosomal Recessive Alport syndrome

This type happens when both parents carry a changed gene and pass it to their child. Males and females are affected in a similar way.

Autosomal Dominant Alport syndrome

Occurs when one parent has the changed gene, which is passed to the child. Symptoms are usually milder and may develop later in life.

It is possible to be a 'carrier' of the Alport gene. This means that you have the faulty gene on one of your X chromosomes, but you may not experience the symptoms. If you are a carrier, you may still pass the Alport gene on to your children.

How is Alport syndrome diagnosed?

If someone in your family has been diagnosed with Alport syndrome, ask your doctor if you should also be tested for the condition. Your doctor will check your kidney function with:

A **urine test** to see if albumin (a type of protein) or blood is present in your urine. Albumin or blood in your urine can be a sign of damage or scarring in the kidneys.

A **blood test** to check your kidney function. This test will measure how well your kidneys are filtering your blood through a result called the estimated glomerular filtration rate (eGFR).

Other tests for Alport syndrome include:

- a **genetic test**, which can help your doctor find out which type of Alport syndrome you have by testing your blood or saliva.
- a **kidney biopsy**, where a small sample of kidney tissue is looked at under a microscope for signs of damage.

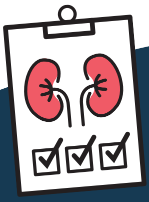
If no one in your family has had Alport syndrome, you may not realise you have it at first. Some people only find out after they develop kidney, hearing or eye problems.

Why is it important to find Alport syndrome early?

If you or a family member has Alport syndrome, your doctor can help identify which other relatives might be at risk. If you are diagnosed with Alport syndrome, talk to your doctor about whether your parents, siblings, or children should also be tested.

You and your healthcare team can plan ahead when Alport syndrome is found early. Your doctor can talk with you about family planning. They may refer you to a genetic counsellor to help you understand the chance of passing Alport syndrome onto your children.

Early diagnosis means your treatment can be started sooner. Starting treatment early can help protect your kidneys and slow down serious problems, like kidney failure.



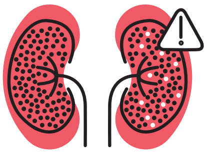
Females may not notice symptoms until later in life. In males, hearing problems may appear before kidney problems. For this reason, it is important that young males with hearing loss have their kidney health checked early.

What are the signs of Alport syndrome?

For **X-linked Alport syndrome**, the first signs of kidney damage are the presence of blood followed by protein in your urine:

- **Blood in your urine** (haematuria) can cause your urine to turn reddish, pink or brown, but small amounts may not be visible. The doctor can order a urine test to find small amounts of blood in the urine.
- **Protein in your urine** (proteinuria) can cause foamy or frothy urine. This symptom is common in teenagers and young adults with Alport syndrome. Proteinuria may lead to swelling of your ankles, wrists or face.

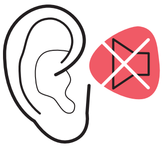
Other common complications include:



Kidney failure -

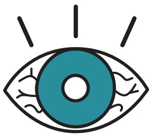
X-linked Alport syndrome can cause kidney failure in males, typically between the ages of 20 and 50 years. Kidney failure is less common in females. The first signs of advancing kidney disease include:

- high blood pressure
- itchy skin
- tiredness or fatigue
- nausea and vomiting
- loss of appetite
- shortness of breath.



Hearing loss -

Not everyone with Alport syndrome will suffer from hearing problems, but those who do generally develop it by the time they are teenagers. Hearing loss in girls tends to be milder than in boys. It often progresses alongside kidney problems.



Eyesight problems -

Alport syndrome can cause problems with the lens and retina of your eyes and may lead to early cataracts (cloudy eyes). An ophthalmologist (eye specialist) can often detect these changes before you notice problems with your eyesight.

Autosomal Recessive Alport syndrome

In this type of Alport syndrome, kidney problems start at an early age. There is a higher chance of kidney failure earlier in life.

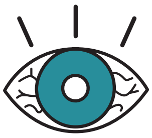
Hearing loss and eyesight problems are common in both males and females. This type of Alport syndrome usually happens faster and may cause more serious symptoms than other types.

Autosomal Dominant Alport syndrome

This type of Alport syndrome develops slower and may have less symptoms. Most people will still have blood in their urine, but kidney failure is less likely. Hearing loss is also less common.

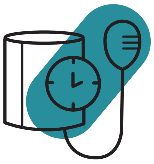
How is Alport syndrome treated?

There is no cure for Alport syndrome, but there are treatments that can help manage the symptoms. These treatments can help protect your kidneys, hearing and eyesight, and support your long term health.



Eyesight problems

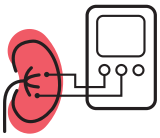
It is important to get regular eye tests. Your doctor may refer you to an ophthalmologist, which is a specialist eye doctor, to help treat any eye problems.



High blood pressure

It is important to control blood pressure if you have Alport syndrome. Good blood pressure control helps your kidneys to work for as long as possible and protects your heart from heart disease. Your doctor may prescribe medicines to control your blood pressure and lower the chance of damage to your kidneys.

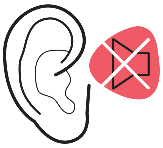
ACE inhibitors and ARBs are types of blood pressure medicines that are commonly to help lower blood pressure in people with Alport syndrome. These medicines have been shown to slow down the need for dialysis or transplant.



Dialysis and transplantation

Most people affected by X-linked or Autosomal Recessive Alport syndrome will experience a loss of kidney function. When your kidneys fail, you will need dialysis or a kidney transplant.

Your kidney specialist (nephrologist) will talk to you about what dialysis and kidney transplant options are suitable for you.



Hearing loss

Hearing aids may be helpful and you should protect your hearing in noisy places. Hearing loss is usually long-term. Your hearing specialist (audiologist) can support you with education, counselling and aids to help you manage day to day.

Protein in your urine

Other medicines have been studied to treat protein in the urine for people with chronic kidney disease (CKD). SGLT2 inhibitors are medicines that have shown benefits for the heart and kidneys in people with CKD.

New trials are studying whether SGLT2 inhibitors are safe and effective for kidney disease caused by Alport syndrome.
If you have protein in your urine and CKD, your doctor may recommend SGLT2 inhibitors.

Healthy lifestyle

Living a full and healthy life is possible with Alport syndrome. Make healthy lifestyle choices such as:

- not smoking
- staying a healthy weight
- eating plenty of fruits and vegetables
- lowering salt in your diet.

Check out Kidney Health Australia's **Kidney Health 4 Life** program, a health and wellness program with modules on healthy diet, exercise, and more. You'll learn practical tips and gain the knowledge you need to manage your kidney disease. Visit **kidneyhealth4life.org.au** to join.

What if I want to start a family?

Early planning helps you and your doctor lower the risk of passing on Alport syndrome. If you or someone in your family has Alport syndrome, you may wish to discuss your situation with a doctor or genetic counsellor before starting a family.

A genetic counsellor can provide information about inherited disease. They can help you and your partner find ways to reduce the chance of passing on Alport syndrome.

Your doctor or genetic counsellor can order a genetic test. It is possible to combine in-vitro fertilisation (IVF) with genetic testing to make sure the Alport gene is not passed on. This is called Pre-implantation Genetic Testing (PGT) and genetic carrier screening. Ask your doctor for more information about referral to a genetic fertility specialist.

What's new with Alport syndrome?

Researchers are currently developing new methods for diagnosing and treating Alport syndrome. The more we learn about the condition, the better we can find and treat it.

Over the past 20 years, there have been several studies looking for new medicines for the treatment of Alport syndrome. Studies have explored using new types of gene therapy and existing diabetes medicines to help treat Alport syndrome.

To learn more about research trials and how you can get involved, visit alportaustralia.com/research. Talk to your nephrologist if you are interested in being involved in a research trial.

Support is available

Being diagnosed with a rare genetic condition can feel overwhelming. Some people or their family members may feel sad, angry, or overwhelmed. It's important to know you are not alone.

The **Alport Foundation of Australia** is a non-profit support group for patients and families affected by Alport syndrome. It promotes research into the treatment of Alport syndrome and related genetic kidney diseases.

Further information can be found on their website **alport.org.au** or by phoning **(02) 4016 0242**.

For crisis support, call **Lifeline** at **13 11 14** or visit **lifeline.org.au**.

If you feel overwhelmed and need mental health support, you can call the **Beyond Blue** support line at **1300 224 636** or visit **beyondblue.org.au**.

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Things to remember:

- ✓ Alport syndrome is a genetic disease that affects your **kidneys, hearing, and eyesight**.
- ✓ There are three main types of Alport syndrome: **X-linked**, **Autosomal Recessive**, and **Autosomal Dominant Alport syndrome**. X-linked is most common and males are more likely to have symptoms.
- ✓ Finding Alport syndrome early means you can start treatment sooner. Some blood pressure medicines can help slow down kidney damage and delay kidney failure.

What does that word mean?

Audiologist – A health care professional who specialises in hearing loss.

Blood pressure – The pressure of the blood in the arteries as it is pumped around the body by the heart.

Carrier – A carrier is a person who has a change in one copy of a gene. Every person carries two copies of most genes (one copy from each biological parent). The carrier does not have the genetic disease related to the abnormal gene, but they can pass this abnormal gene to their child.

Collagen – The main protein in your skin, which gives strength and support for your tendons, cartilage, bone and connective tissue which surround many of your organs.

Dialysis – A treatment for kidney failure that removes waste products and excess fluid from the blood by filtering the blood through a special membrane. There are two types of dialysis: haemodialysis and peritoneal dialysis.

Gene – Each cell in the human body contains about 25,000 to 35,000 genes. Genes carry the information that determines your traits, which are features or characteristics that are passed on to you – or inherited – from your parents.

Genetic Counsellor – Someone trained to help people understand and made decisions around the medical, psychological, family and reproductive effects of your genetic condition. They do not make decisions for you but will help you reach decisions which are right for you and your family. More information can be found from The Australasian Society of Genetic Counsellors (ASGC) at **www.hgsa.org.au/asgc**.

Genetic testing – A type of medical test that identifies changes in chromosomes, genes, or proteins.

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

Haematuria – The medical term for blood in your urine.

Inherited – Passed to you by your parents. This can include personality traits, physical appearance and some health conditions.

Mutation – A gene that is faulty and does not work like it should. This can be inherited or caused by environmental factors including diet and chemicals.

Nephrologist – A medical doctor who specialises in kidney function.

Ophthalmologist – A medical doctor who specialises in diagnosing and managing conditions of the eyes.

Proteinuria – Medical term for too much protein in your urine.

SGLT2 inhibitors – Medicines that help lower blood sugar in people with type 2 diabetes by making them wee out more sugar. They also help protect kidneys in people with kidney disease by reducing the pressure in the kidney filters and helping the body get rid of excess sugar and salt through their wee.

Transplant – Surgically removing an organ from one person and placing into another person.

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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

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