

Fact sheet

Alport Syndrome

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What is alport syndrome?

Alport syndrome is a genetic (inherited) disease of the kidneys which can also lead to problems with your hearing and eyes. It affects at least 1 in 5,000 people. Males are usually more severely affected than females.

Your body is held together by connective tissue. This connective tissue is made up of collagen. The collagen known as type IV collagen is in the filtering part of your kidneys called the glomerulus. This collagen is needed to make sure your kidneys are

able to do their filtering job properly. In Alport syndrome the collagen in the glomerulus is affected. Your kidneys become weak and damaged, and the amount of waste that is filtered from your blood is reduced.

How is alport syndrome inherited?

Genes are a part of chromosomes, which you inherit from your parents. If you are female you have two X chromosomes, while if you are a man you have an X and Y chromosome. There are many genes located on the X chromosome, but only a few on the Y chromosome.

Sometimes, a gene contains a variation – like a spelling mistake – that stops it working like it is meant to. A variation can also happen with no known cause. Variations in the coding that make a gene not work properly (faulty) are called mutations and can lead to a wide range of conditions.

The most common way that Alport syndrome is inherited is by a mutated gene carried on your X chromosome. This is called X Linked Alport syndrome. It accounts for around 80 percent of people with Alport syndrome. Other types of Alport syndrome are called Autosomal Dominant Alport Syndrome and Autosomal Recessive Alport Syndrome.

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 do not have Alport syndrome itself. You can still pass the Alport gene on to your children.

Please see the Alport Foundation website www.alport.org.au for more detailed information about how Alport syndrome is inherited.

How is alport syndrome diagnosed?

If you, or someone in your family has been diagnosed with Alport syndrome, it is important for all family members to also be checked for the condition. This may involve a urine test to look for small amounts of blood. A test called a kidney biopsy may also be recommended. This is a fairly simple process where some kidney cells are removed to look for changes. Further genetic testing may also be needed before the diagnosis of Alport syndrome is made.

Unless there is a known family history of Alport syndrome, a diagnosis may not be made until kidney, hearing or eye problems develop. It is very important for young males with hearing loss to have their kidney function checked, as hearing problems usually show much earlier than kidney problems.



Why is early identification of alport syndrome important?

Alport syndrome is an inherited condition. It is possible to not know that you have Alport syndrome, or are a 'carrier' for the Alport syndrome gene, even when you are aged in your 30s. By then you may have started your family and already passed the Alport gene on to your children. Diagnosis in females is very important. If you are a female with Alport

syndrome you have a 50% chance of passing the gene to your sons and daughters.

If it is known that people in your family have Alport syndrome then it is possible to determine who else in the family may be at risk. Early detection of Alport syndrome means that your treatment can be started sooner.

It also means that you can discuss with health professionals ways of reducing the risk of passing the mutation on to your children.

What are the complications of alport syndrome?

The first signs of kidney damage are usually blood (haematuria) and then protein (proteinuria) in your urine:

- **Blood in your urine** is always present in young male children with Alport syndrome, but may not be present in young females. Blood in your urine may not always be noticed. Small amounts can be detected using a urine test.
- **Protein in your urine** may be present in the urine of teenagers and young adults with Alport syndrome. Large amounts of protein in your urine can cause fluid retention and swelling of body parts such as your ankles, wrists, or face. If males do not have blood and/or protein in their urine after 10 years of age, they are unlikely to have Alport syndrome.

See the 'Albuminuria' and 'Blood in Urine' fact sheets for more information.

Other common complications include:

- **Kidney failure** - Alport syndrome always causes kidney failure in males, which usually happens between the ages of 15 and 50 years. Kidney failure is not as common in females with Alport syndrome.
- **Deafness** - Not everyone with Alport syndrome will suffer from deafness, but those who do generally develop it by the time they are teenagers. Loss of hearing in girls tends to be milder than in boys. Hearing loss seems to increase at the same stage as kidney problems.
- **Eye sight problems** - Alport syndrome can cause problems with your eye lens, retina or it can cause cataracts. An ophthalmologist (eye specialist) may be able to diagnose this before you even notice any problems with your eye sight.

See 'Chronic Kidney Disease' as well as 'Heart Disease and Chronic Kidney Disease' fact sheets for more information.

How is alport syndrome treated?

Eye sight problems

If you have severe problems with your eye sight, your eye specialist (ophthalmologist) will talk to you about suitable treatment options.

Hearing Loss

Hearing aids may be helpful, and you should protect your hearing in noisy places. Loss of hearing is likely to be permanent, so seek education and counselling with your hearing specialist (audiologist).

Dialysis and transplantation

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.



High blood pressure

Good control of blood pressure is very important. It helps to keep your kidneys functioning for as long as possible and reduces your risk of cardiovascular disease (heart disease or stroke). Different types of blood pressure tablets work in different ways so usually more than one type is prescribed. Talk to your doctor about what your target blood pressure should be.

Healthy lifestyle

Choices can also help, such as not smoking, staying at a healthy weight, eating a healthy diet which is low in salt and having protein intake checked (an Accredited Practising Dietitian can help with this).

Can alport syndrome be prevented?

If you have a family history of Alport syndrome, you may wish to discuss your situation with a doctor or genetic counsellor before starting a family. It is possible to combine in-vitro fertilisation (IVF) with genetic testing to make sure the Alport gene is not passed on.

What is the outlook for alport syndrome?

Researchers are currently developing new methods for diagnosing and treating Alport syndrome.

If you have Alport syndrome, or know someone who does, you may want to learn about the ATHENA study which is now recruiting patients around the world. The ATHENA study is designed to learn more about the progression of Alport syndrome. Information

obtained from this study will help in the design of future clinical trials to test a new drug in people with Alport syndrome. More information can be found at www.alport.org.au/RESEARCH.

Who should I contact for more information?

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 renal diseases.

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



THINGS TO REMEMBER

- Alport syndrome is a genetic disease which affects your kidneys, hearing and eye sight
- Early identification means your treatment can start sooner
- Genetic testing can reduce the risk of passing the Alport gene on to your children

What does that word mean?

Audiologist - A medical doctor 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 the mother and one from the father). 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 make 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.

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 - The medical term for too much protein in your urine.

Transplant - An organ transplant involves surgically removing an organ from one person and placing into another person.

For more information about kidney or urinary health, please contact our free call Kidney Health Information Service (KHIS) on 1800 454 363.

Or visit our website kidney.org.au to access free health literature.

This is intended as a general introduction to this topic and is not meant to substitute for your doctor's or Health Professional's advice. All care is taken to ensure that the information is relevant to the reader and applicable to each state in Australia. It should be noted that Kidney Health Australia recognises that each person's experience is individual and that variations do occur in treatment and management due to personal circumstances, the health professional and the state one lives in. Should you require further information always consult your doctor or health professional.

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If you have a **hearing or speech impairment**, contact the National Relay Service on **1800 555 677** or **relayservice.com.au**

For all types of services ask for 1800 454 363