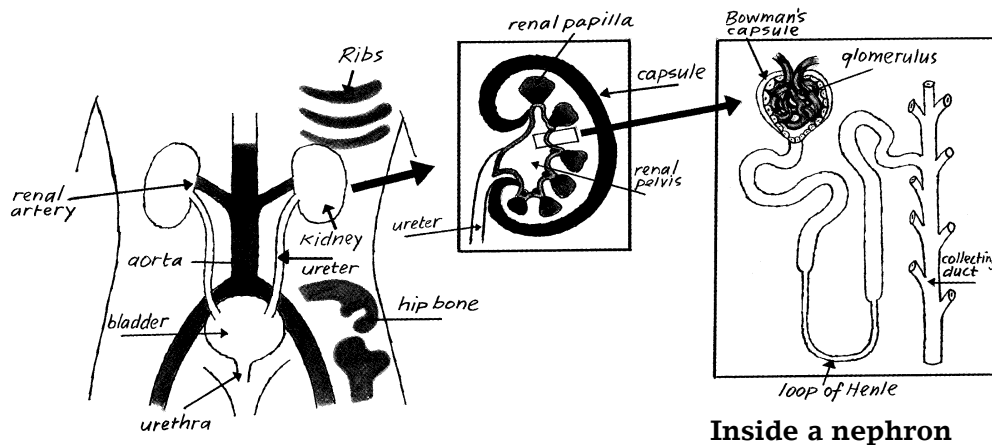


HEREDITARY NEPHRITIS - ALPORT SYNDROME

WHAT IS ALPORT SYNDROME?

Alport Syndrome is a form of hereditary nephritis. Nephritis is a general term used to describe diseases that cause swelling or inflammation of the kidneys. Alport Syndrome affects basement membranes. Basement membranes are fine protective layers of tissue or cells found in many parts of the body. They contain collagen, which is a tough, glue-like protein that shapes the structure of tendons, bones and connective tissues. Alport Syndrome affects those basement membranes that are found in each kidney's nephrons and sometimes those in the eyes and ears.

Each kidney contains up to one million nephrons, the working units of the kidneys. Inside the nephrons are tiny sets of looping blood vessels or capillaries called the glomerulus, which act as filters. Alport Syndrome is caused by faults in the genes that affect type IV collagen. This alters the normal structure and function of each glomerulus. It causes thinning of the collagen-containing membranes and scarring, which reduces their ability to filter waste and extra fluid from the blood.

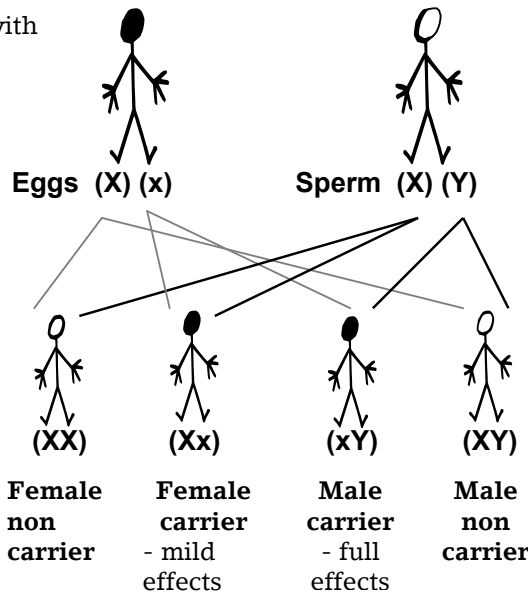


HOW IS ALPORT SYNDROME INHERITED?

The genetics of this syndrome are complicated. The faulty gene is found on the X-chromosome, one of the chromosomes deciding a person's sex. Women have two X-chromosomes and men have an X and Y-chromosome. The most common type of genetic cause is called X-linked recessive inheritance. This means that one of the parents carries a faulty gene on one of their X-chromosomes.

If a mother is the carrier, daughters who inherit the faulty gene have milder effects because they inherit two X-chromosomes. The normal X-chromosome from their father protects them from their mother's faulty gene. Sons have one only X-chromosome. If they inherit a faulty gene from their mother, they do not have a normal gene to protect them and are more severely affected.

Carrier mother with faulty gene (x)

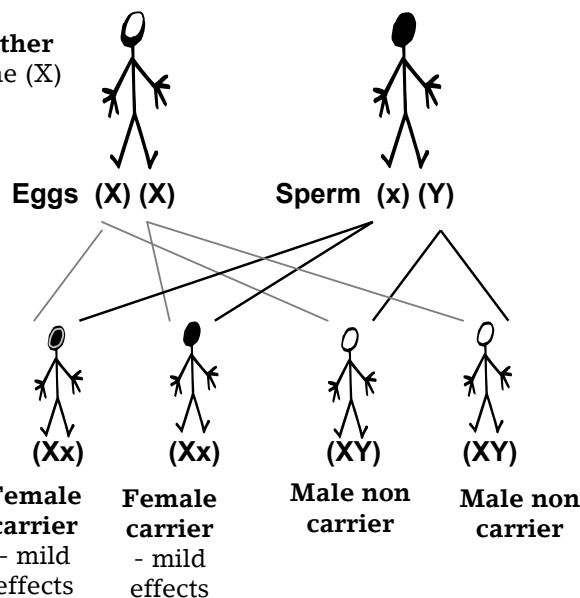


Non carrier father with normal gene (X)

**Alport Syndrome
Female x-linked
Recessive Family
Tree**

The father can also carry the faulty gene. He cannot pass it onto his sons as they inherit his Y-chromosome. Sons can only get Alport Syndrome if they inherit a faulty gene from their mother. The father gives a faulty x-chromosome to his daughters. Both are carriers but protected by their mother's normal X-chromosome so have milder effects.

Non carrier mother with normal gene (X)



Carrier father with faulty gene (x)

**Alport Syndrome
Male x-linked
Recessive Family
Tree**

There are less common forms of Alport Syndrome where both parents carry a faulty gene. Sons and daughters are both severely affected. Some people with Alport Syndrome do not have a family history of this disease. In these cases it occurs as a result of a new genetic mutation.

HOW IS ALPORT SYNDROME DIAGNOSED?

Unless there is a known family history of the condition, it's often difficult to diagnose Alport Syndrome. The symptoms are similar to those of other kidney diseases and final diagnosis is only decided by a kidney biopsy. During a biopsy a small piece of kidney tissue is tested. The sample is examined under a microscope for changes to the basement membrane structure.

WHAT ARE THE COMPLICATIONS OF ALPORT SYNDROME?

The first sign of kidney damage is blood (haematuria) and protein (proteinuria) in the urine:

- Blood in the urine

This is always present in young male children but may be absent in females. Blood in the urine may not always be visible but microscopic amounts can be detected using a urinary dipstick.

- Protein in the urine

Protein may be present in the urine of teenagers and young adults. Large amounts of protein in the urine can cause fluid retention and swelling of body parts, eg. ankles, wrists, face. If males do not have blood and/or protein in their urine after 10 yrs of age, they are unlikely to have Alport Syndrome.

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

Other common complications include:

- Kidney disease

With increasing age boys develop other signs of kidney damage including high blood pressure, reduced appetite and tiredness.

- Kidney failure

Alport Syndrome always causes kidney failure in males but uncommonly and much later in life in females. Males develop kidney failure anywhere from 15 to 50 years of age.

- Deafness

Alport Syndrome can be linked with a slow loss of hearing and may be a clue to diagnosis. Not everyone 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 worsen roughly in step with increasing kidney problems.

- Vision problems

Vision problems can also be linked to Alport Syndrome. It can produce oddly shaped lens, lens changes, cataracts or poor function of the retina in the back of the eye.

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

HOW IS ALPORT SYNDROME TREATED?

There is currently no treatment that can stop or reverse the effects Alport Syndrome. Treatment aims is to control complications and slow the progress of kidney disease:

- High Blood Pressure

Effective control of blood pressure is very important. It helps to preserve kidney function and reduce the risk of cardio vascular disease. Different types of blood pressure tablets work in different ways so it is not unusual for more than one type to be prescribed. In recent years the use of ace inhibitors or angiotensin receptor blockers has proved to be effective treatment for high blood pressure and also helps to reduce proteinuria. Healthy life style choices can also help, eg staying at a healthy weight and being a non-smoker.

- Hearing Loss

Hearing aids may be helpful. People need to protect their hearing in noisy places. Loss of hearing is likely to be permanent so education and counselling to deal with this change and gain new communication skills may be helpful.

Kidney transplantation will not improve hearing problems.

- Vision Problems

If there are severe problems, an intraocular lens implantation or surgery may be needed.

- Dialysis and Transplantation:

When the kidneys fail, dialysis or transplantation is required. Transplantation is usually a successful treatment. However it is important to make sure that a potential family member donor is not a carrier of the faulty gene. In cases where people with Alport Syndrome who have the collagen gene Col4A5 missing, transplantation is not an option because their immune system rejects the new kidney.

See Haemodialysis, Peritoneal Dialysis, Kidney Transplant and Choosing Not To Have Treatment fact sheets for more information.

CAN ALPORT SYNDROME BE PREVENTED?

Couples with a family history of Alport Syndrome may wish to discuss their situation with a doctor or genetic counsellor to identify any risks when starting a family.

WHAT IS THE OUTLOOK FOR ALPORT SYNDROME?

Researchers are currently developing new methods for diagnosing Alport Syndrome and checking for mutations. Identification of faulty genes means that further investigation into the way the gene works can take place and future gene therapy for this condition becomes a possibility.

**For more information about kidneys health or this topic, please contact the
Kidney Health Australia:**

Kidney Information Line (freecall) on 1800 682 531 or visit website

www.kidney.org.au

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.

Kidney Health Australia gratefully acknowledges the valuable contribution of the National Renal Resource Centre and KHA Kidney and Urinary Advisory Group in the development of this material.

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