COL4A3 & Alport Syndrome

What is Alport syndrome?

- Alport Syndrome is an inherited condition involving kidney disease, hearing loss and eye abnormalities
- It is a result of changes in forms of the COL4 or COL5 genes.
- The initial symptoms can vary depending on the nature of the change, but generally it is first noticed as recurrent episodes of blood in the urine.
- Over time, protein in the urine, high blood pressure and gradual loss of kidney function occurs until the kidneys fail. The age at which failure occurs depends on which COL4 gene has changed.

What is COL4A3, and how do changes in COL4A3 affect the kidneys?

- In the kidneys, COL4A3 produces proteins that form part of the filtration system to remove water and waste products from the blood.
- A change in this gene prevents the kidney from properly filtering the blood and allows blood and protein to pass into urine. This results in gradual scarring of the kidneys and eventually kidney failure.
- With a COL4A3 change, kidney failure usually occurs between the ages of 16 and 35 years.

Do these changes have effects on other parts of the body?

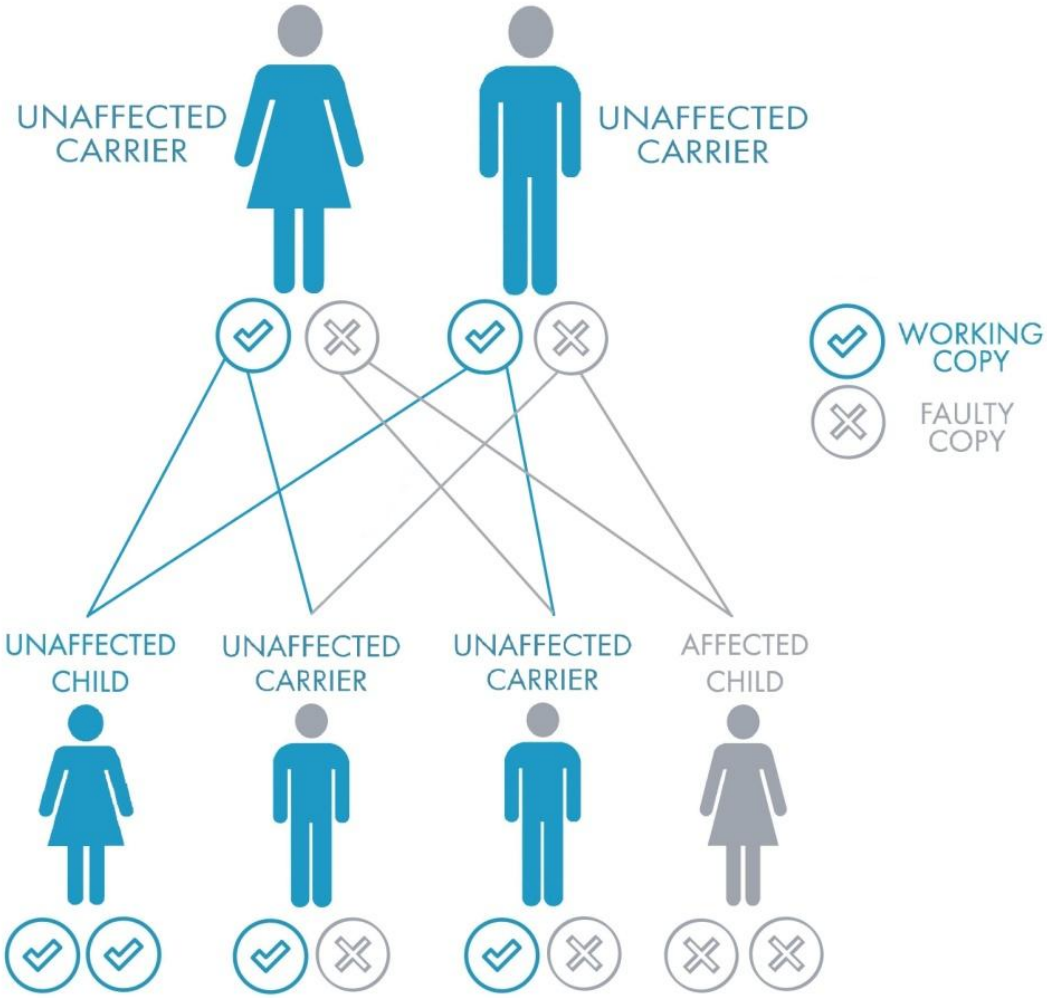
- COL4A3 is also an important part of inner ear structures. Changes in COL4A3 can lead to hearing loss, and this usually occurs during late childhood or early teen years.
- Changes in COL4A3 can also affect the eyes, causing misshapen lenses, eye pain and an abnormally coloured retina. However, these abnormalities do not usually lead to sight loss.

How is Alport syndrome treated?

- There is currently no cure for Alport Syndrome. Treatment mainly focuses on managing complications of kidney function loss.
- Angiotensin blockade drug therapy may be used in some patients.
- A kidney transplant is preferred over dialysis when kidney failure occurs. The disease does not develop again in the new kidney.

How is this change passed down through a family?

- You have two copies of the COL4A3 gene.
- To develop Alport syndrome, two faulty copies of the COL4A3 gene must be inherited, one from each parent – they are “carriers” of the faulty gene and do not have the disease themselves.
- Each child of carrier parents has a 1 in 4 (25%) chance of inheriting the disease.
- If a child receives only one copy of a faulty gene, they themselves become carriers. They will not have Alport syndrome but may pass on that faulty gene to their own offspring.



Should my family members be tested?

- Counselling and genetic testing are generally available to all family members at risk of having the disease.
- At-risk children may be monitored.
- If a living relative wishes to donate a kidney it is important for them to be tested.