PAX2 Gene & Renal Coloboma Syndrome

What is renal coloboma syndrome?

- Renal Coloboma Syndrome is an inherited condition which occurs due to changes in the gene PAX2.
- It is associated with kidney and eye abnormalities.

What is PAX2, and how do changes in PAX2 affect the kidneys?

- The PAX2 gene provides instructions for proteins involved in the early development of the eyes, ears, brain, spinal cord, genital tract and kidneys.
- When a change occurs in PAX2, there is an incomplete formation of certain tissues, with the kidneys and eyes being particularly affected.
- One or both kidneys are small and underdeveloped, making it more difficult for them to function. This can lead to kidney failure.
- A less common feature is the formation of fluid-filled sacs called cysts within the kidneys, which can also negatively impact kidney function.

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

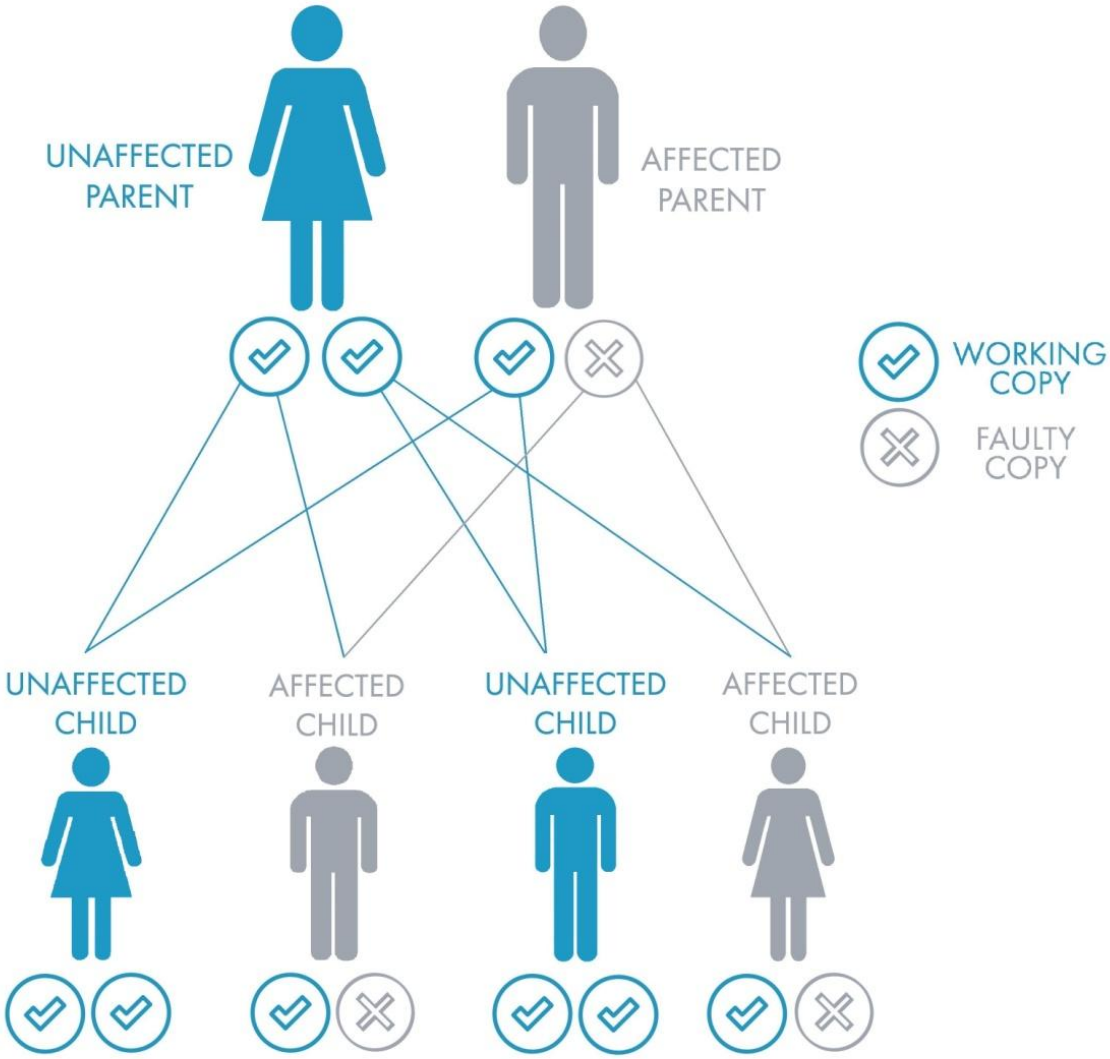
- As mentioned, there may be effects on the eyes. This is due to a malformation in the optic nerve, which is responsible for carrying information from the eye to the brain.
- These malformations are often associated with a gap or hole, called a coloboma, in the retina at the back of the eye.
- The extent of the vision problems caused by these malformations varies widely; some people's vision may be severely impaired, while others may not be affected at all.
- Other less common features include backflow of urine from the bladder, loose joints and mild hearing loss.

How is Renal Coloboma Syndrome treated?

- No specific therapy is available, and the focus of the treatment is managing symptoms.
- In the event of kidney failure, dialysis therapy or transplantation will be required.

How is this change passed down through a family?

- You have two copies of PAX2– one copy from each of your parents.
- To have renal coloboma syndrome you must inherit at least one faulty copy of the PAX2 gene from an affected parent.
- Each child of an affected parent has a 1 in 2 (50%) chance of inheriting the disease.



Should my family members be tested?

- If a family history of kidney disease is identified, it may be advised for family members to undergo genetic testing.
- Before testing is carried out it is recommended that family members discuss this with a genetic counsellor.