TRPC6 & Focal Segmental Glomerulosclerosis

What is Focal Segmental Glomerulosclerosis?

- Focal Segmental Glomerulosclerosis (FSGS) is a type of kidney lesion which occurs due to inflammation and scarring of kidney cells.
- It presents as steroid-resistant nephrotic syndrome in most affected individuals, and is a leading cause of kidney failure in adults.
- A number of causes have been identified and include genetic changes, viruses and certain drugs and toxins.

What is TRPC6, and how do changes in TRPC6 affect the kidneys?

- The TRPC6 gene is believed to help maintain the proper structure and function of kidney cells involved in filtering the blood.
- When there are changes in this gene, these cells become damaged and scarred. This can eventually result in kidney failure.
- Signs and symptoms can include protein in the urine, swelling, high blood pressure and high cholesterol.

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

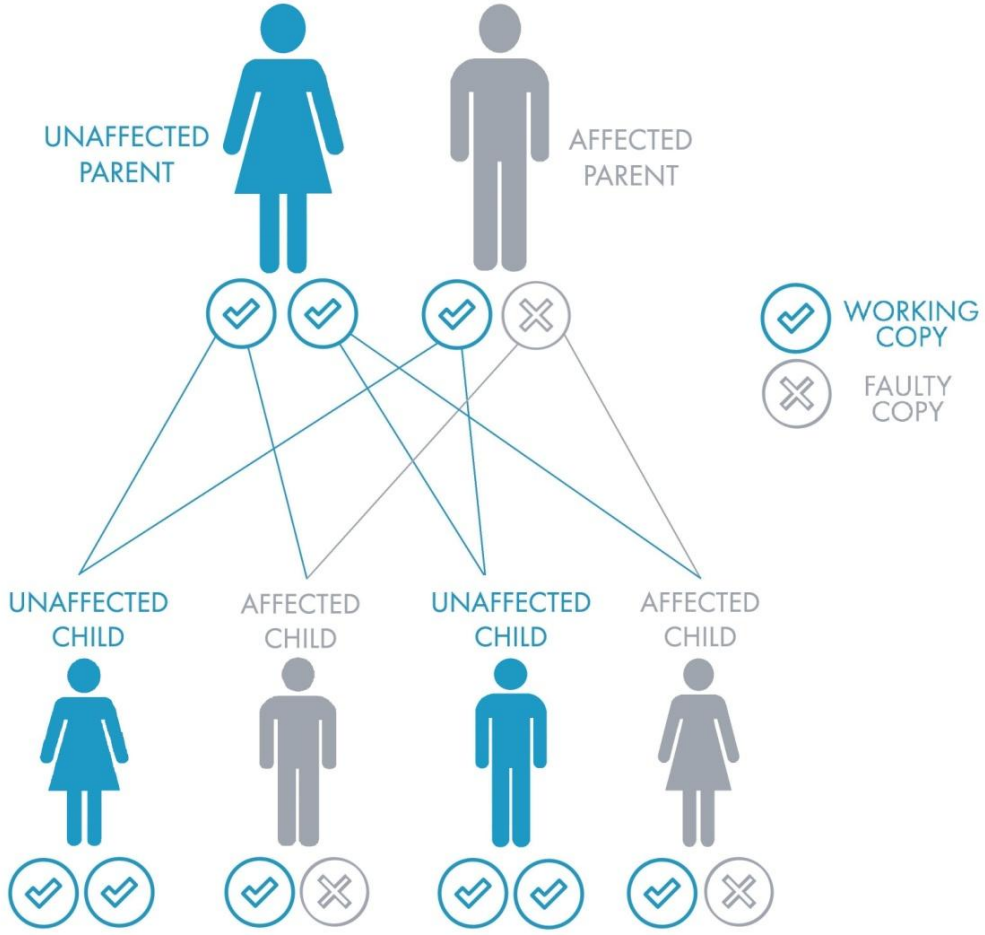
- Changes in TRPC6 do not appear to have effects on other parts of the body.

How is Focal Segmental Glomerulosclerosis treated?

- A number of treatments may be recommended.
- Medication to suppress the immune system.
- Diuretics and a low salt diet to control swelling.
- ACE inhibitors to control blood pressure or lower the amount of protein in the urine.
- If there is a progression to kidney failure, dialysis therapy or transplantation would be required.

How is this change passed down through a family?

- You have two copies of TRPC6 – one copy from each of your parents.
- For FSGS to occur you must inherit at least one faulty copy of the TRPC6 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 there is a family history of kidney disease, it may be advisable for family members to undergo genetic testing.
- It is recommended that before this testing is carried out, it is discussed with a genetic counsellor.