
INF2 & 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 also a leading cause of kidney failure in adults.
- Signs and symptoms can include protein in the urine, swelling, high blood pressure and high cholesterol.
- A number of causes have been identified and include genetic changes, viruses and certain drugs and toxins.

What is INF2, and how do changes in INF2 affect the kidneys?

- INF2 is a gene that plays a role in maintaining cell structure.
- When this gene is changed, kidney cells become damaged and scarred, and are unable to filter the blood properly. This eventually leads to kidney failure.
- INF2-associated FSGS usually begins in adolescence or early adulthood.

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

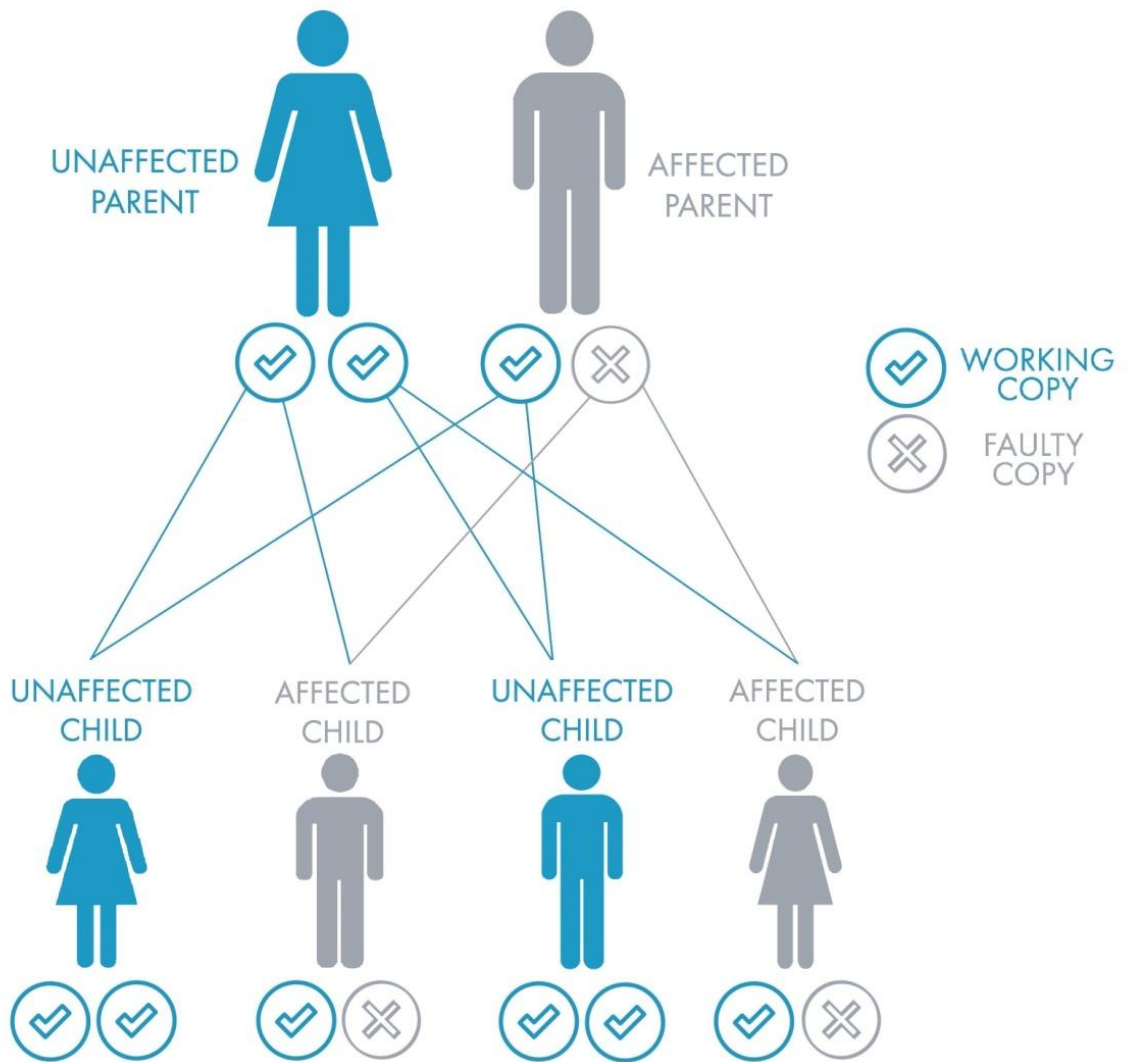
- Changes in INF2 do not have effects outside of the kidney.

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.
- In the event of kidney failure, dialysis therapy or transplantation is required.

How is this change passed down through a family?

- You have two copies of INF2 – one copy from each of your parents.
- To develop focal segmental glomerulosclerosis you must inherit at least one faulty copy of the INF2 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 that it is discussed with a genetic counsellor.