SLC7A9 & Cystinuria

What is cystinuria?

- Cystinuria is an inherited cause of kidney stones which develop due to changes in the genes SLC3A1 or SLC7A9.
- Signs and symptoms of kidney stones include pain in your sides (flank pain) and blood in the urine.
- The age at which stones begin to form is very variable, with some as early as infancy.

What is SLC7A9, and how do changes in SLC7A9 affect the kidneys?

- SLC7A9 provides instructions for the reabsorption of amino acids, the building blocks of proteins. This is important, as a build up of these amino acids can have harmful effects.
- A change in this gene means that these amino acids are not reabsorbed and their levels in the urine increase.
- When one of these amino acids, called cystine, accumulates in the urine, it forms crystals.
- These crystals can block the urinary tract and impair the kidneys ability to remove waste products through urine.
- The blockages are also a site for bacteria to cause infections.

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

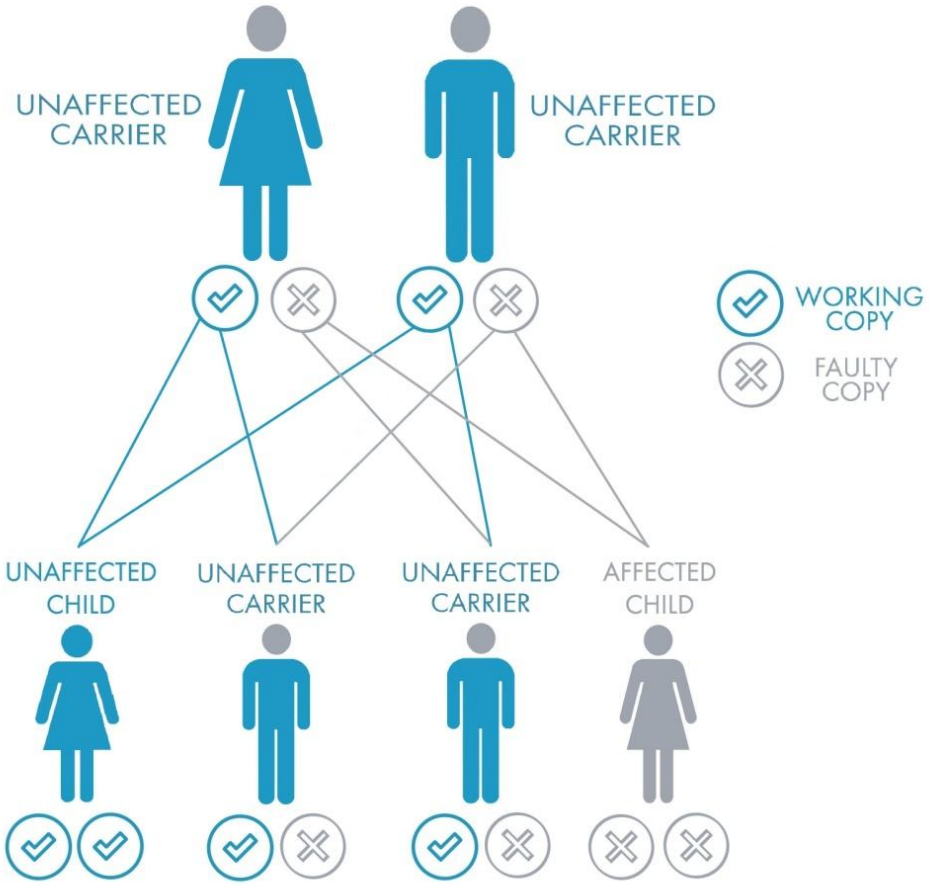
- These changes do not appear to have effects on other parts of the body.

How is cystinuria treated?

- Treatment focuses on lowering the levels of cystine in the urine.
- Initial management involves:
  - High fluid intake.
  - Reducing protein and salt in the diet.
  - Potassium citrate, which is used to reduce the acidity of the urine.
- If these measures are not sufficiently reducing cystine, you may be prescribed a drug called tiopronin which has good efficacy in reducing cystine levels and preventing recurrence of kidney stones.
- If very large stones form and block the urinary tract, surgery may be required.

How is this change passed down through a family?

- You have two copies of SLC7A9.
- To develop cystinuria, two faulty copies of the SLC7A9 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 cystinuria but may pass on that faulty gene to their own offspring.



Should my family members be tested?

- Genetic testing of family members is generally not carried out, other than for research purposes.