# GRHPR & Primary Hyperoxaluria

## What is primary hyperoxaluria?

- Primary hyperoxaluria is an inherited condition caused by changes in one of three genes – AGXT, GRHPR or HOGA1.
- These changes result in recurrent kidney and bladder stones. Depending on which gene has been affected, this can progress to kidney failure.
- Signs and symptoms of primary hyperoxaluria include blood in the urine, urinary tract infections and abdominal pain.

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

- The GRHPR gene provides instructions for the production of enzymes which help remove harmful waste products from the body.
- When this gene is changed, there is a build-up of a substance called oxalate, which combines with calcium to form stones. These stones get stuck in the kidney and reduce its ability to function. Over time this may result in kidney failure.
- However, changes in GRHPR are generally associated with less severe disease, and progression to kidney failure is more gradual and less frequent overall compared to other forms of the disease.

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

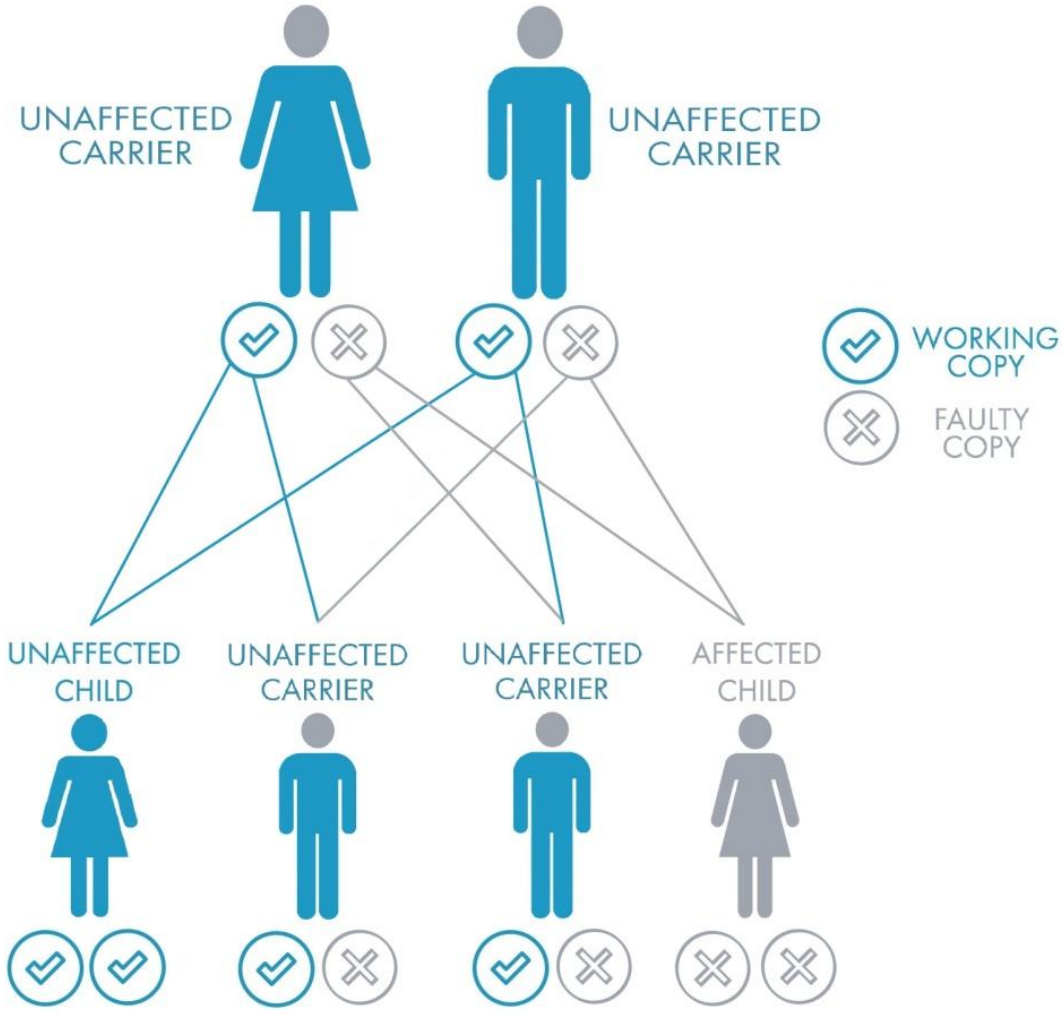
- Excess oxalate can also deposit in the heart, blood vessels, joints, bone and eyes and have a number of effects:
  - Disruption of electric signals in the heart.
  - Reduced blood circulation in the legs that can result in gangrene.
  - Joint pain and reduced mobility.
  - Impaired vision.
  - Dental problems such as tooth pain.
  - Bone pain, anaemia and an increased risk of bone fracture.

## How is primary hyperoxaluria treated?

- Treatment focuses on reducing oxalate levels. This may include:
  - High fluid intake.
  - Orthophosphate, potassium citrate-citric acid and/or magnesium oxide therapy.
- In the rare circumstance of kidney failure, a kidney transplant may be required.

## How is this change passed down through a family?

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




## Should my family members be tested?

- Family members may be advised to undergo genetic testing, especially as early diagnosis and treatment can help delay or reduce kidney impairment.
- It is recommended to have a discussion with a genetic counsellor prior to undergoing testing.