NPHP1 & Nephronophthisis

What is nephronophthisis?

- Nephronophthisis is a disorder characterised by inflammation and scarring of the kidneys, eventually resulting in kidney failure.
- There are several genetic causes, and each gene is associated with different types of the disease.

What is NPHP1, and how do changes in NPHP1 affect the kidneys?

- The NPHP1 gene is involved in maintaining the normal function of cell structures that allow cells to communicate with each other and their environment.
- Maintaining this communication is very important for the normal growth and function of cells.
- When this gene becomes faulty, abnormal cell growth occurs, resulting in the formation of fluid-filled sacs called cysts.
- The growth of these cysts causes the kidney to become scarred and inflamed, and the kidney is unable to function correctly.
- Early signs and symptoms include increased urination and thirst. Later, anaemia, extreme tiredness, nausea and weakness may develop.
- Kidney failure will eventually occur, usually around 13 years of age.

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

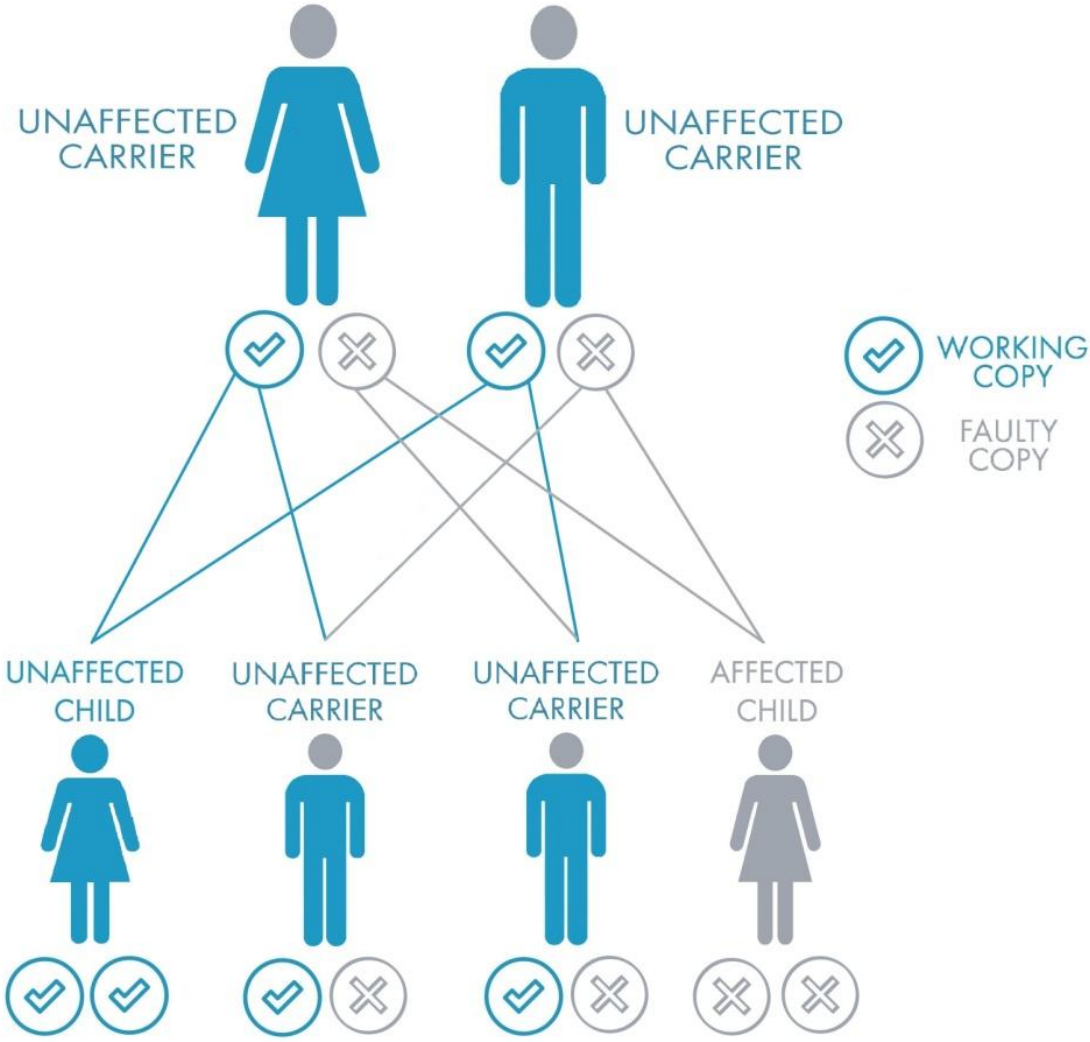
- Changes in the NPHP1 gene can be associated with a number of different disorders, which may also have nephronophthisis as a feature.
  - Senior-Løken syndrome is a combination of nephronophthisis and sight impairment.
  - Joubert syndrome involves neurological problems and can include nephronophthisis, eye abnormalities and liver disease.

How is nephronophthisis treated?

- There is currently no specific therapy for nephronophthisis.
- Treatment involves managing symptoms and includes:
  - Replacing lost water and salt
  - Treating anaemia with supplements
- Kidney transplantation is associated with very good outcomes as the disease does not recur in the new kidney.

How is this change passed down through a family?

- You have two copies of NPHP1.
- To develop nephronopthisis, two faulty copies of the NPHP1 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 nephronopthisis but may pass on that faulty gene to their own offspring.



Should my family members be tested?

- Screening for NPHP1 changes is not generally carried out unless an individual is showing symptoms.