CFHR & C3 Glomerulopathy

What is C3 Glomerulopathy?

- C3 Glomerulopathy is a condition associated with excessive deposition of proteins and progressive kidney failure.
- Some of the major features include blood and protein in the urine, reduced amounts of urine, low levels of protein in the blood, high blood pressure and swelling.
- Two forms of the disease have been identified – dense deposit disease and C3 glomerulonephritis.
    - The conditions cause similar kidney problems but differ by the age at which symptoms begin – dense deposit disease generally begins earlier.
- Inherited forms of this condition may be caused by mutations in the CFHR genes (1 – 5).

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

- CFHR genes regulate proteins called complement which are involved in the immune system.
- When this gene changes, it results in overactive complement proteins which become deposited in kidney cells.
- This interferes with the cells' ability to filter blood and eventually leads to kidney failure. This is usually within 10 years of diagnosis.

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

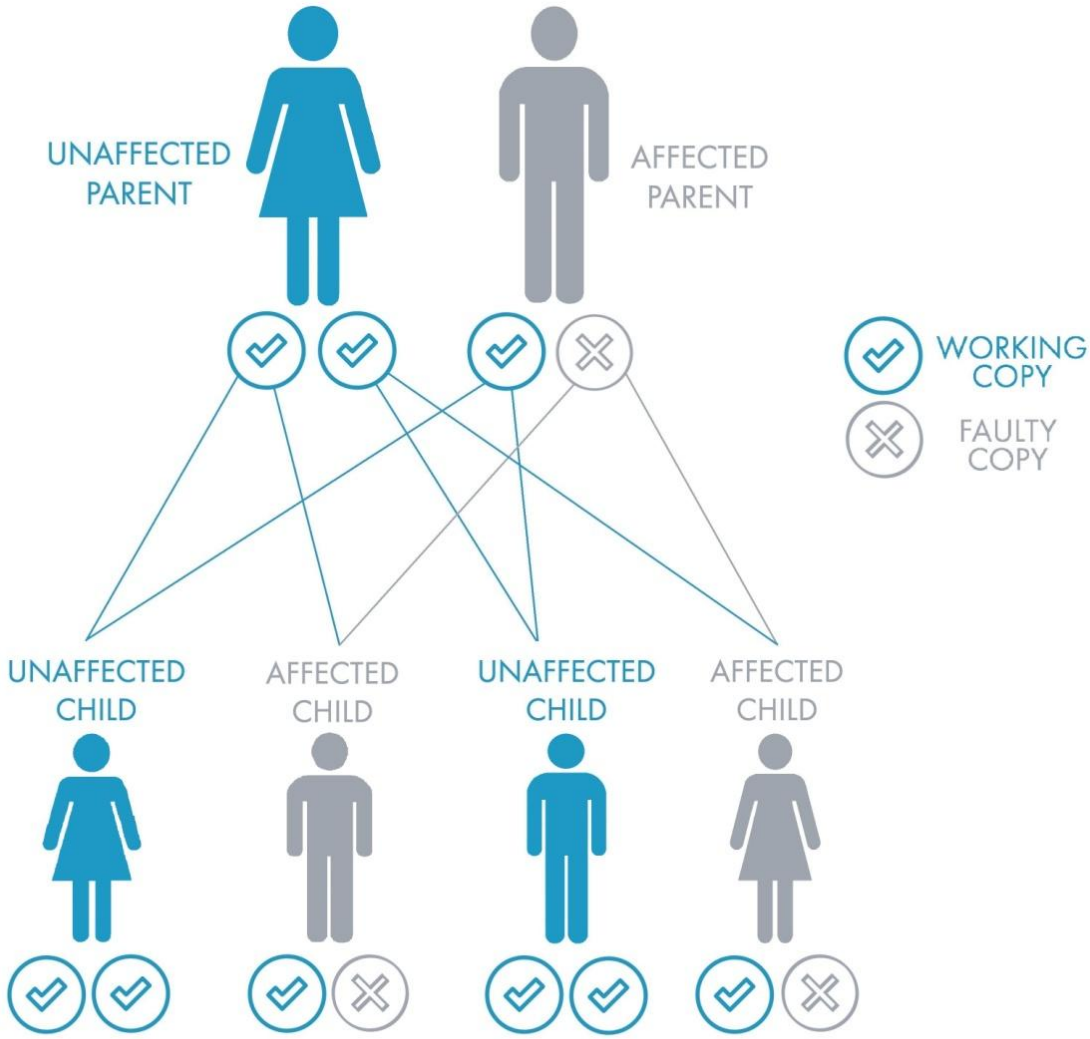
- The dense deposit disease form can also be associated with effects outside of the kidney.
    - Vision loss can occur due to deposition of proteins in the eyes.
    - A rare metabolic condition called acquired partial lipodystrophy, where fat is lost from the lower limbs and torso and builds up in the upper limbs, face and neck.

How is C3 Glomerulopathy treated?

- As C3 Glomerulopathy is so uncommon, there is limited evidence on the best way to approach treatment.
- Your doctor will make a judgement based on your individual symptoms and the severity of the condition.
- Some examples of treatments include:
    - ACE inhibitors for high blood pressure or protein in the urine.
    - Immunosuppressive drugs such as steroids.
    - Drugs called monoclonal antibodies which help control the damage caused by deposits.
- Transplantation may be an option, but the disease tends to recur in the new kidney.

How is this change passed down through a family?

- You have two copies of CFHR – one copy from each of your parents.
- To have C3 Glomerulopathy you must inherit at least one faulty copy of the CFHR 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 a family history of kidney disease is identified, it may be advised for family members to undergo genetic testing.
- Before testing is carried out it is recommended that family members have a discussion with a genetic counsellor.