LCAT & Complete LCAT Deficiency

What is complete LCAT deficiency?

- Complete LCAT deficiency is an inherited disorder that impairs the body's ability to process cholesterol.
- It has effects on the cornea of the eye, blood cells, arteries and the kidney.
- Most symptoms tend to begin in adulthood.

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

- The LCAT gene is involved in the removal of cholesterol from the blood and tissues.
- When this gene is changed, cholesterol accumulates in the kidneys and impairs kidney function.
- Over time this can lead to kidney failure.

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

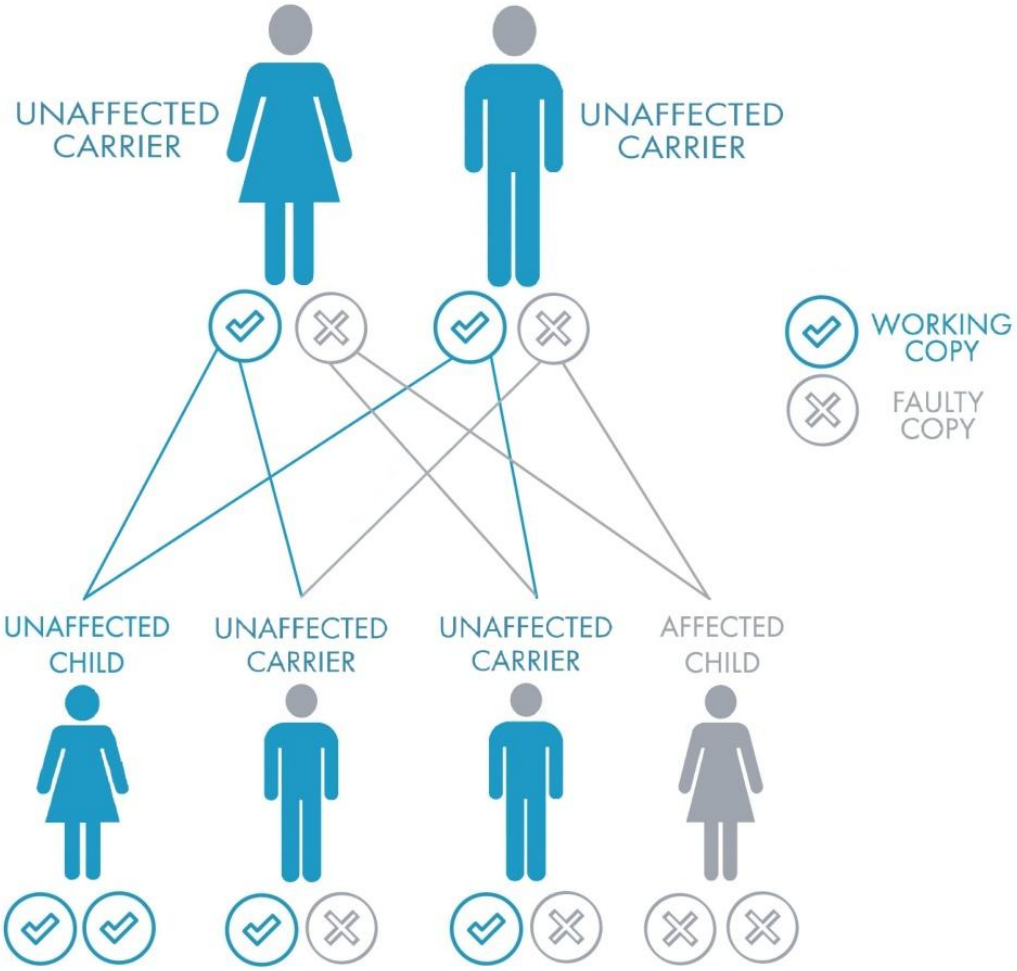
- The excess cholesterol also accumulates in other parts of the body and causes problems.
- Cholesterol can deposit in the eyes, causing the corneas to become cloudy. This generally occurs in childhood and is the earliest symptom of the deficiency. This can cause a significant reduction in vision and will continue to worsen throughout life.
- Due to its effect on red blood cells, some individuals may develop anaemia. This can make you tired, pale, weak, confused or feverish.
- Accumulation of fat deposits in the arteries (atherosclerosis) causes the arteries to narrow. If the arteries become completely blocked, it could potentially result in a stroke or heart attack.
- This deficiency can also cause enlargement of the lymph nodes, spleen and/or liver.

How is complete LCAT deficiency treated?

- There is no specific therapy for Complete LCAT Deficiency, with treatment focused on managing your particular symptoms. This may include:
    - Dialysis or transplant in the event of kidney failure.
    - A corneal transplant if vision is severely reduced.
    - Restricting fat in the diet.
    - Drugs to lower your cholesterol called statins.
    - Blood pressure tablets such as ACE inhibitors or angiotensin receptor blockers.

How is this change passed down through a family?

- You have two copies of LCAT.
- To develop complete LCAT deficiency, two faulty copies of the LCAT 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 complete LCAT deficiency 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.
- It is recommended to discuss this with a genetic counsellor.