Screening for Familial Hypercholesterolemia (FH)

First exclude secondary causes of elevated Total cholesterol or LDL- Cholesterol

Common secondary causes include:

- Clinical hypothyroidism
- Biliary obstruction
- Nephropathy, especially nephrotic proteinuria
- Poorly controlled diabetes
- Pregnancy
- Drugs include: Glucocorticoids, Isotretinoin, some systemic anti-viral agents and some anti-rejection drugs (e.g. cyclosporine)

The upper limit of "normal" in patients 50 years and older is approximately 8 mmol/L for total cholesterol and 5.3 mmol/L for LDL-cholesterol. Approximately 2.5% of other healthy adults will have TC and/or LDL-C levels above these cut-offs. An estimated 1 in 200 people will have FH. Therefore, even in the absence of one of the above secondary causes, polygenic hypercholesterolaemia is still the most common cause of an elevated cholesterol, especially if cholesterol is only slightly above these levels.

A cholesterol above 8 mmol/L or LDL above 5.0 mmol/L, a personal or family history of early CVD, and/or clearly suggestive clinical features (especially tendon xanthomas) should prompt suspicion of FH. Genetic testing is very expensive, therefore a scoring system is used to increase the pre testing likelihood of identifying a mutation. Perform a **Dutch Lipid Clinic Network Score (DLCNS),** using the "Request for approval of FH gene testing" to assess the likelihood for FH:

If the **DLCNS** score is 6 or more, request FH gene testing and email the DLCNS and consent for genetic testing to Labadmin@waikatodhb.health.nz . Please provide an email address that can be replied to in case additional information is required.

A causative mutation in LDLR, apolipoprotein B or PSCK-9 confirms the diagnosis of FH.

(LDLR = LDL receptor)

FH testing is performed by Canterbury Health Laboratories, for details see:

https://www.chl.co.nz/test/familial-hypercholesterolaemia-fh-ngs-gene-panel/

Genetic testing is negative in 20% of patients with FH.

FH is a clinical diagnosis. Detection of a FH causing mutation is useful to screen close relatives for FH.

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