**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)

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 8 mmol/L (or LDL slightly above 6.5 mmol/L).

A cholesterol above 8 mmol/L or LDL above 6.5 mmol/L, a personal or family history of early CVD, and/or clearly suggestive clinical features (especially tendon xanthomas) should prompt suspicion of FH. Perform a **Dutch Lipid Clinic Network Score (DLCNS)** to assess the likelihood for FH:

<https://www.athero.org.au/fh/calculator/>

If the **DLCNS** score is 6 or more, contact the Chemical Pathologist at WDHB laboratory to discuss genetic testing.

A causative mutation in LDLR, APOB 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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