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 elevated cholesterol, especially if cholesterol is only slightly above 8 mmol/L (or LDL slightly above 6.5 mmol/L).

A cholesterol level above 8.0 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/wp-content/uploads/Dutch-Lipid-Clinic-Network-Score2.pdf

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 PCSK-9 confirms the diagnosis of FH.

(LDLR = LDL receptor)

In New Zealand, genetic testing is currently available for LDLR and APOB but not for PCSK-9 mutations. APOB mutation testing is routinely performed when LDLR mutation testing is requested.

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

Stephen du Toit,
Chemical Pathologist
4 April 2019