FAMILIAL HYPERCHOLESTEROLEMIA (FH)

Familial hypercholesterolemia is an inherited genetic disorder that causes dangerously high levels of LDL or "bad" cholesterol.

Heterozygous FH is the most common monogenic disorder encountered in clinical practice, affecting **1 in 250 individuals**.

It is estimated that only about **10% of patients** with FH in Canada have been diagnosed.

Patients with FH are at high risk of early cardiac events

Men with untreated FH have **a 50% chance of having a cardiac event by age 50**

Women with untreated FH have **a 30% chance of having a cardiac event by age 60**

Patients with untreated FH have approximately **a 20 times greater risk of developing early heart disease and atherosclerosis**

When should a patient be screened for FH?

If LDL-C ≥5.0 mmol/L (40 yrs or older)
If LDL-C ≥4.5 mmol/L (If 18-39 yrs old)
If LDL-C ≥4.0 mmol/L (If 18 yrs or younger)

YES

DNA mutation **OR**
Tendon xanthomas **OR**
LDL-C ≥ 8.5 mmol/L

YES → Definite FH

NO → First degree relative with high LDL-C **OR** Index patient **OR** first degree relative with ASCVD (<55 yr men; <65 yr women)

YES → Probable FH

NO → Severe hypercholesterolemia

If a patient's initial treatment with cholesterol-lowering medications fails to reach target LDL-C levels, it could be an indication of FH.

We recommend that genetic testing be offered, when available, to complement/confirm a clinical diagnosis of FH and to enable cascade screening. Fifty percent of an index patient's first degree relatives will also be affected, making family-based cascade testing vital.

We recommend that initial screening should include a fasting or non-fasting lipid profile and LDL-C calculated by the Friedewald formula.

How Is FH treated?

We recommend a personalized treatment plan using a combination of therapies to reach target LDL-C levels of 2.5 or 2.0 mmol/L if vascular disease is absent or present, respectively.

- Diet, exercise and lifestyle changes
- Pharmacological therapy (statins as a primary therapy with secondary agents as required, including ezetimibe and PCSK9 inhibitors)
- Specialized treatments are required for rare patients with the very severe homozygous form

To learn more about FH visit us at **CCS.CA** and **FHCanada.net**

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This infographic is an informative knowledge translation tool that features information based on the *Canadian Cardiovascular Society Position Statement on Familial Hypercholestrolemia*: Update 2018