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?

- 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

Canadian Cardiovascular Society

Société canadienne de cardiologie

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