

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

l in 250 individuals.

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



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



Definite FH



First degree relative with high LDL-C **OR** Index patient **OR** first degree relative with ASCVD (<55 yr men; <65 yr women)



Probable FH



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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