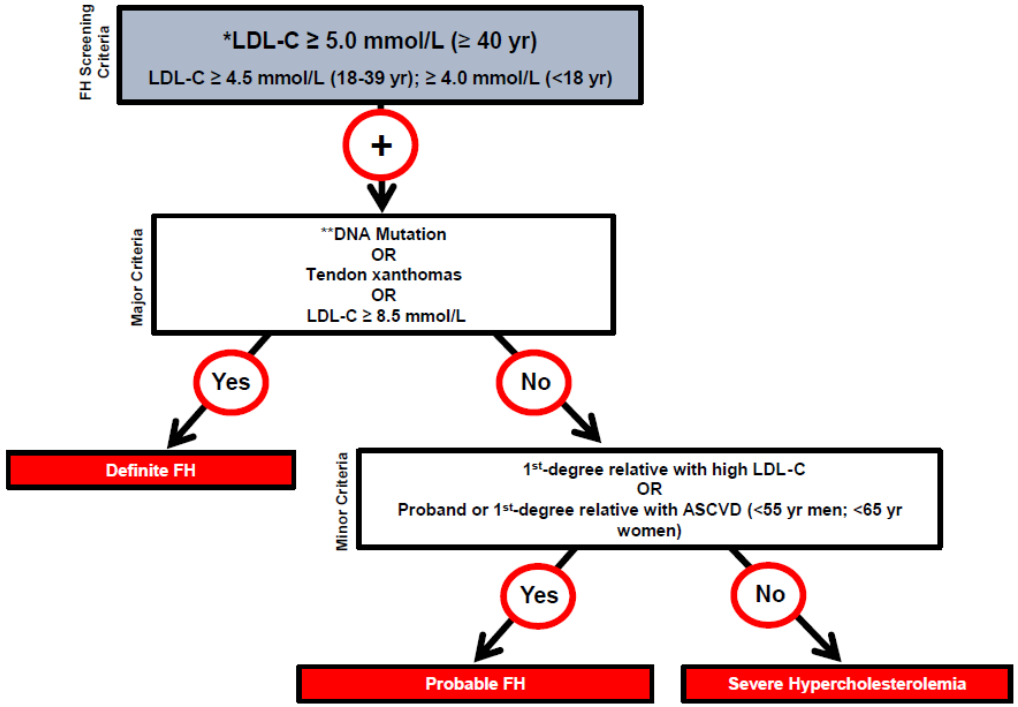
Canadian criteria for the clinical diagnosis of Familial Hypercholesterolemia.

About 1 in 250 Canadians is thought to have the heterozygous (HeFH) form of familial hypercholesterolemia (FH). FH is both under diagnosed and under treated despite the knowledge that early diagnosis and treatment can normalize life expectancy.

Figure 1. Canadian criteria for the clinical diagnosis of FH.



ASCVD: atherosclerotic cardiovascular disease; LDL-C: low-density lipoprotein cholesterol. * Secondary causes of high LDL-C should be ruled out (severe or untreated hypothyroidism, nephrotic syndrome, hepatic disease [biliary cirrhosis], medication, especially antiretroviral agents).

From Ruel *et al.,* 2018 *Can J Cardiol.* Available at www.FHCanada.net.

Table 1. Clinical features of FH in heterozygotes (HeFH) and homozygotes (HoFH).

Clinical features	HeFH	HoFH
Genetics	Mutation in one copy of one FH gene	Mutations in two FH genes, one inherited from each parent
LDL-C levels	≥ 5mmol/L with additional features shown in following boxes	>12 mmol/L *lower LDL-C levels, especially in children or in treated patients, do not exclude HoFH*
Cardiovascular disease (CVD) onset	<55 years of age in men <65 years of age in women	<20 years of age (can be as early as the first year of life)
Other atherosclerotic disease risks	— Stroke or transient ischaemic attack — Peripheral vascular disease	
Physical findings	— Cholesterol deposits in the tendons (xanthomata) and/or around the eyes (xanthelasma) — Arcus cornealis (white, grey, or blue opaque ring in the corneal margin) onset <45years	
Family history	— Early onset CVD — Hyperlipidemia, often requiring treatment	

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