

# FAMILIAL HYPERCHOLESTEROLEMIA

## CLINICAL FEATURES OF FH

Updated July 2026

| Clinical Features                                                                                                                                                                    | Heterozygotes (HeFH)                                                                                                                                                                                                                                      | Homozygotes (HoFH)                                                                                            |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------|
| <b>Genetics</b><br>pathogenic/likely pathogenic (P/LP) variants (what used to be called mutations)                                                                                   | P/LP variant in one copy of one FH gene                                                                                                                                                                                                                   | P/LP variant in two FH genes, one inherited from each parent                                                  |
| <b>Untreated LDL-C levels</b><br> CardioRisk App<br>to impute baseline value in those on treatment | $\geq 5\text{mmol/L}$ at age 40 years or older<br>$\geq 4.5\text{mmol/L}$ at age 18-39 years<br>$\geq 4\text{mmol/L}$ at age younger than 18 years<br><u>with</u> additional features shown in following boxes                                            | $>12\text{ mmol/L}$<br>lower LDL-C levels, especially in children or in treated patients, do not exclude HoFH |
| <b>Cardiovascular disease (CVD) onset</b>                                                                                                                                            | $<55$ years of age in men<br>$<65$ years of age in women                                                                                                                                                                                                  | $<20$ years of age<br>can be as early as the first year of life                                               |
| <b>Other atherosclerotic disease risks</b>                                                                                                                                           | <ul style="list-style-type: none"> <li>Stroke or transient ischaemic attack</li> <li>Peripheral vascular disease</li> </ul>                                                                                                                               |                                                                                                               |
| <b>Physical findings</b>                                                                                                                                                             | <ul style="list-style-type: none"> <li>Cholesterol deposits in the tendons (xanthomata) and/or around the eyes (xanthelasma)</li> <li>Arcus cornealis (white, grey, or blue opaque ring in the corneal margin) onset <math>&lt;45</math> years</li> </ul> |                                                                                                               |
| <b>Family history</b>                                                                                                                                                                | <ul style="list-style-type: none"> <li>Early onset cardiovascular disease</li> <li>Hyperlipidemia, often requiring treatment</li> </ul>                                                                                                                   |                                                                                                               |

## RESOURCES

- [Canadian Lipid Clinics \(June 2026\)](#)
- [Genome BC has curated resources and links on FH.](#)
- [National Human Genome Research Institute has more on FH and genomics for the public and clinicians](#)
- [Infographic for individuals and families by the Canadian Cardiovascular Society and FHC Canada](#)