

FAMILIAL HYPERCHOLESTEROLEMIA

Updated April 2024

FH - familial hypercholesterolemia; CCS - Canadian Cardiovascular Society;
CVD- cardiovascular disease; ASCVD - atherosclerosis cardiovascular disease

RISK ASSESSMENT



The use of conventional cardiovascular risk calculators in individuals with FH is not recommended as these greatly underestimate lifetime CVD risk. FH-specific cardiovascular risk calculators (e.g. the FH Risk Score, SAFEHEART) should be considered to assess the risk of ASCVD.

Routine assessment and stratification of the risk of ASCVD in all patients with FH should be used to guide personalized treatment and management.

Those with homozygous status (HoFH, two pathogenic/likely pathogenic (P/LP) variants in an FH gene) should be referred to a specialized lipid centre.

PHARMACEUTICALS (ADULTS)

- Statins are the drug class of choice for individuals with one P/LP variant in a FH gene (heterozygous FH (HeFH)).
- LDL-C should be lowered as fast and as far as possible.
- **The CCS recommends a >50% reduction of LDL-C from baseline beginning at age 18 as primary prevention and that an ideal goal of LDL-C <1.8 mmol/L is recommended for secondary prevention.**
- Observational studies have shown a dramatic decrease in cardiac events in statin-treated individuals with FH.
- Non-fasting lipid profiles should be used to monitor treatment in those whose treatment is stable.



LIFESTYLE

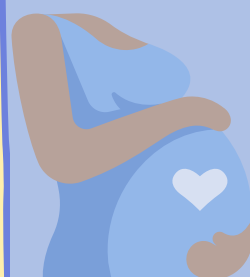
All families with FH (including children and adolescents) should be counselled about the importance of lifestyle modification and heart healthy behaviour such as:

- smoking cessation and avoidance of passive smoking
- diet (e.g. high fibre, low in trans and saturated fatty acids)
- exercise
- stress reduction
- maintenance of ideal body weight



PREGNANCY

- For most persons assigned female at birth who are of reproductive age, an effective birth control method is recommended with discontinuation of statin therapy ideally 3 months prior to planned pregnancy or at the time of a positive pregnancy test.
- A pregnant person with FH and additional risk factors, e.g. established ASCVD, should be referred to a specialty lipid clinic for further treatment advice.



CHILDREN/ADOLESCENTS

- Selective lipid screening (fasting or non-fasting, non-HDL-C or LDL-C) can be considered at any age when there is a positive family history of premature CVD or dyslipidemia, or other cardiovascular risk factors.
- Based on current randomized control trials, the ideal age to begin treatment for FH is between ages 8-12 years.
 - Pharmacological treatment can be considered, incorporating clinical judgement, family and patient preferences.
- Lifestyle modifications as above remain the cornerstone of CVD prevention in both children and adolescents with FH.
- The CCS recommends that children with HoFH are referred to a lipid specialist centre for cholesterol-lowering therapies when >15kg in weight.

