

## RESEARCH SUMMARY

# Update on Familial Hypercholesterolemia: An Expert Clinical Consensus from the National Lipid Association

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**Why this matters:** Familial hypercholesterolemia (FH) is one of the most common inherited conditions, affecting roughly 1 in 311 people worldwide, yet it remains severely underdiagnosed. Left untreated, it causes dangerously high cholesterol from birth and dramatically increases the risk of early heart disease. This updated guidance from leading lipid experts offers the latest recommendations on diagnosis, screening, and treatment—information that could help individuals and families identify the condition early and take action to prevent heart attacks and strokes.

## Overview

This expert consensus from the National Lipid Association updates clinical guidance on familial hypercholesterolemia (FH), a genetic condition that causes lifelong elevated LDL (“bad”) cholesterol due to inherited defects in the body’s ability to clear cholesterol from the blood. The researchers reviewed the latest evidence on how FH is diagnosed, how families can be screened, and what treatments are most effective. The goal is to help clinicians identify FH earlier and treat it more aggressively to reduce the high rates of heart disease and premature death associated with this condition.

## Key findings

- FH affects approximately 1 in 311 people worldwide, but the vast majority remain undiagnosed. Early detection through family screening (testing relatives of known patients) is the most cost-effective way to find new cases.
- Updated treatment goals now recommend lower LDL cholesterol targets than previous guidelines. For adults with FH who have not yet had a heart event, the target is below 70 mg/dL, which is a significant reduction from earlier recommendations of below 100 mg/dL.
- Genetic testing can complement a clinical diagnosis, but is not required to begin treatment. A diagnosis can be made based on high LDL levels, family history, and physical signs.
- New and emerging medications—including PCSK9 inhibitors (**such as alirocumab, evolocumab, and inclisiran**), bempedoic acid, and a newer agent called lerodalcibep can significantly lower LDL cholesterol when statins alone are not enough.
- Treatment should begin as early as possible, ideally in childhood, and continue for life. Starting cholesterol-lowering therapy early in children with FH has been shown to nearly eliminate the excess risk of atherosclerosis (plaque buildup in arteries).
- Universal cholesterol screening is recommended for all children between ages 9 and 11, with earlier screening if there is a family history of high cholesterol or early heart disease.

## **What this means for you**

If you or a family member has very high cholesterol, or if it runs in your family, this research highlights the importance of being evaluated for FH. Ask your doctor whether your cholesterol levels and family history suggest a genetic cause. If you are diagnosed with FH, know that more effective treatments are available now than ever before, and that starting treatment early makes a significant difference. If you have FH and are planning a family, genetic counseling can help you understand the chances of passing it on and how to plan for early screening and treatment of your children.

## **Limitations to keep in mind**

This paper represents an expert clinical consensus rather than a systematic review or randomized trial, so the recommendations reflect the collective judgment of specialists in this field. While the guidance is based on extensive published evidence, some recommendations, particularly the newer, more aggressive treatment targets, have not yet been validated in large-scale outcome trials specific to FH patients.

**Related CarePoint topics:** Heart Health, Chronic Disease Management, Medications