

# Familial Hypercholesterolemia Definition & Diagnosis Criteria

AN EXPERT CLINICAL CONSENSUS FROM THE NATIONAL LIPID ASSOCIATION



**Overview:** Familial hypercholesterolemia (FH) is a common genetic disorder inherited in an autosomal semi-dominant pattern, characterized by lifelong elevated low-density lipoprotein cholesterol (LDL-C), leading to a high risk of early onset atherosclerotic cardiovascular disease (ASCVD).

## Key Considerations

- Either clinical or genetic criteria can be used to diagnose FH. The absence of an identifiable pathogenic variant in those genetically tested does not exclude a diagnosis of FH.
- When FH is diagnosed, the affected individual should be assigned the specific ICD-10 code for FH.
- Pharmacologic, diet, and lifestyle interventions are crucial for this patient population to prevent ASCVD.

## FH Diagnosis Criteria

A clinical diagnosis of FH is made based on clinical characteristics, exclusion of secondary causes and phenocopies, severity of LDL-C elevation, and/or genetic testing. Genetic testing is not required to establish a diagnosis of FH, but such testing is appropriate to complement the clinical diagnosis and facilitate cascade screening. Although genetic testing is considered "gold-standard" for diagnosis of FH, the sensitivity of genetic testing for diagnosis of FH may be as low as 60% among individuals who fulfill clinical criteria for a definite diagnosis of FH. Although the diagnosis of FH is straightforward in many cases, there can be considerable nuance in application and interpretation of diagnostic criteria in some patients.

## Clinical characteristics that could indicate FH:

- Personal Patient History of premature CAD and/or Hypercholesterolemia
- Family History of premature CAD and/or hypercholesterolemia
- Elevated LDL-C ( $\geq 190$  mg/dL)
- Tendon Xanthomas
- Corneal arcus (age < 45)
- Genetic mutation\*

*\*Some families with FH may not have an identifiable pathogenic variant known to cause FH.*



## Secondary Causes of Hyperlipidemia

Hyperlipidemia may be present in approximately 20-30% patients with LDL-C  $\geq 190$  mg/dL and could be caused by:

- high-saturated fat
- very-low-carbohydrate diets (e.g., "keto," "carnivore")
- nephrotic syndrome
- hypothyroidism
- obstructive liver disease
- various medications (e.g. cyclosporin)
- menopause

Patients with severe hypercholesterolemia should undergo evaluation for these secondary causes via detailed medical history, physical examination (e.g., thyroid exam), and appropriate laboratory testing (e.g., thyroid stimulating hormone, creatinine, urine protein, and liver function tests).

## Coverage of FH Diagnosis

Possessing the above characteristics is sufficient to substantiate the diagnosis for coverage of appropriate diagnostic evaluation (e.g. genetic testing, advanced imaging), therapeutic procedures (such as lipoprotein apheresis), and the initiation and intensification of lipid-lowering pharmacotherapy.

**ICD-10 codes for insurance coverage:**

**E78.010 - HoFH**

**E78.011 - HeFH**

**E78.019 - FH, unspecified**

**For more information, check out the manuscript:**

Ahmad Z, Agarwala A, Cuchel M, et al. Update on familial hypercholesterolemia: an expert clinical consensus from the NLA. *Journal of Clinical Lipidology*. Published online January 2026. doi:<https://doi.org/10.1016/j.jacl.2026.01.011>

