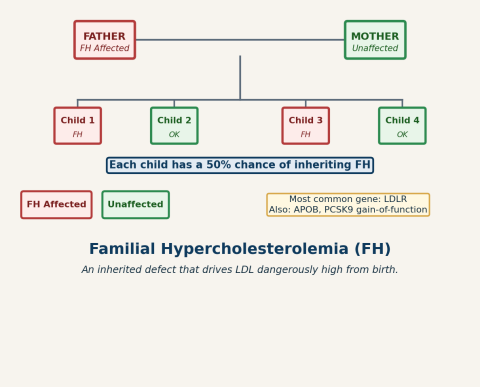
# Familial Hypercholesterolemia (FH)

The Inherited Cholesterol Condition That Can Be Treated — If Found Early

**FH is a genetic disorder that keeps LDL dangerously high from birth. Most people who have it do not know. Early diagnosis and treatment can prevent a heart attack decades before it would otherwise happen.**




**Online:** https://go.riasalimd.com/fh-guide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## Names & Terms You Will Hear

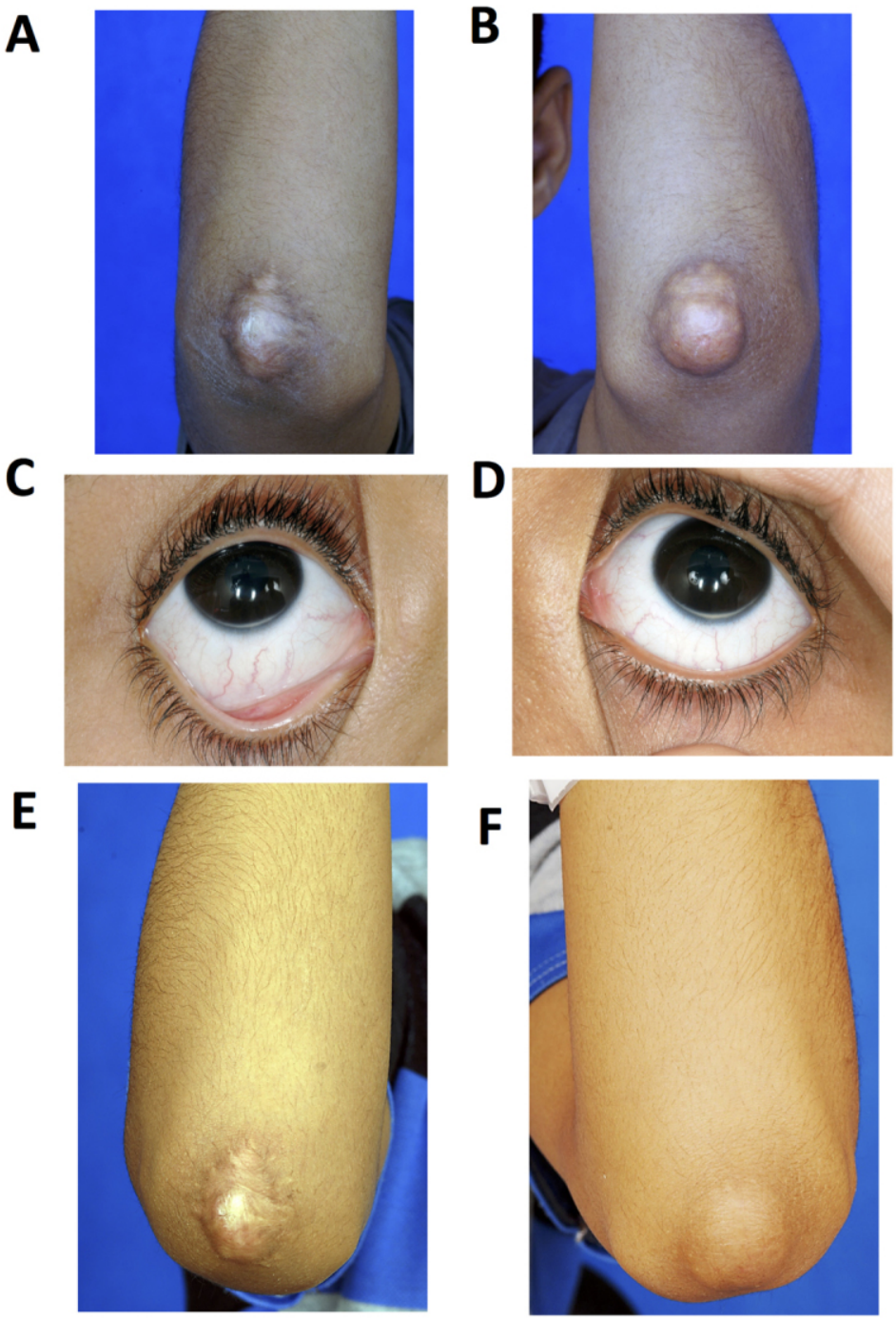
| Term | Meaning |
|---|---|
| Familial Hypercholesterolemia (FH) | A gene problem that stops the liver from clearing LDL. LDL stays very high from birth. Not caused by diet — it is DNA. |
| HeFH — the common form | One copy of the bad gene. Affects about 1 in 250 people. LDL often 190 to 400 mg/dL without treatment. |
| HoFH — the rare form | Two bad gene copies (one from each parent). LDL can go above 500 mg/dL. Heart attacks can happen in childhood. |
| LDLR Gene | The most often mutated gene in FH. It makes LDL receptors. Without them, LDL builds up in the blood. |
| APOB Gene | The second most common FH gene. Makes a protein on LDL that hooks onto receptors. Mutations slow LDL clearance. |
| PCSK9 (gain-of-function) | A less common FH gene. Too much PCSK9 destroys LDL receptors. Fewer receptors means higher LDL. |
| Cascade Screening | Testing your parents, brothers, sisters, and children when you are found to have FH. Each one has a 50% chance. This is the most important step after your own diagnosis. |
| LDL — the BAD cholesterol | In FH, LDL is high from birth. It builds plaque in arteries much faster than normal. |
| Tendon Xanthomas | Cholesterol lumps in tendons — often the Achilles or knuckles. A key clue for FH when LDL is also high. |
| Corneal Arcus Before Age 45 | A pale ring around the eye's cornea in someone under 45. A clue that LDL has been very high for a long time. |
| Heart disease (ASCVD) | Plaque in the arteries. Can cause heart attack, stroke, or leg pain. FH speeds up plaque buildup by decades. |
| PCSK9 Inhibitor | A shot (Repatha or Praluent) given every 2 to 4 weeks. Lowers LDL another 50 to 60% on top of a statin. |

**Three key facts about FH:**

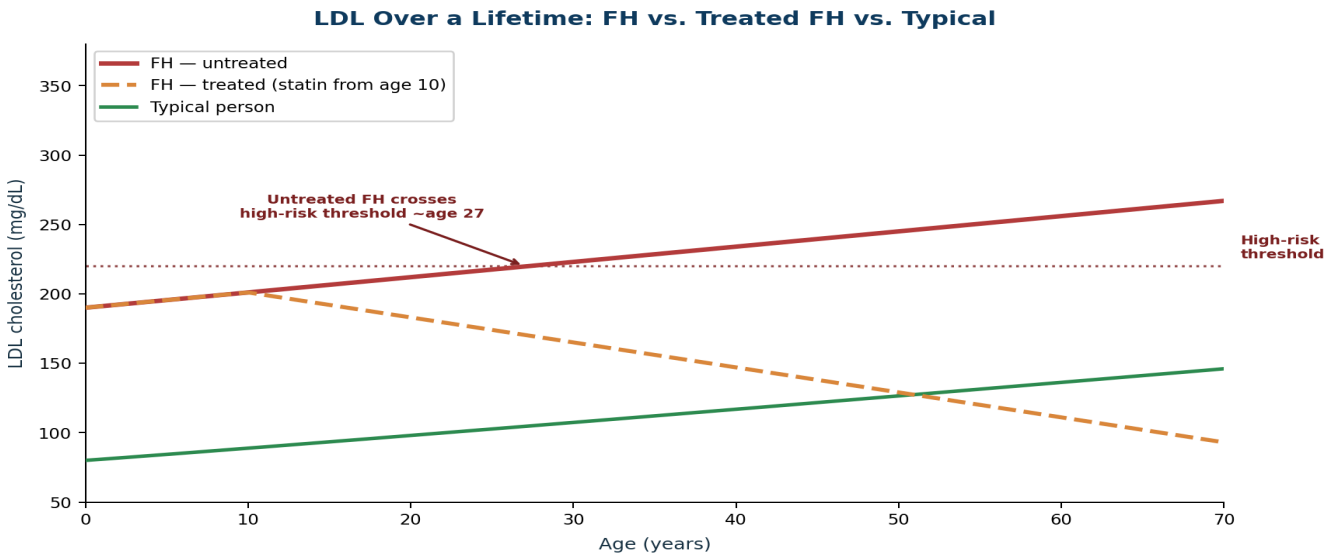
1. **FH is genetic** — diet alone cannot fix it. Medicine is essential.
2. **1 in 250 people have it** — and most do not know. A simple blood test can find it.
3. **Early treatment saves lives.** FH treated from childhood or early adulthood leads to near-normal life expectancy.

## Familial Hypercholesterolemia (FH)?

- FH is a gene problem. It is NOT caused by diet or lifestyle. A broken gene stops the liver from clearing LDL. LDL is high from the day you are born.
- About 1 in 250 people have FH. That is more common than Type 1 diabetes. Yet fewer than 1 in 10 people with FH know they have it. Many find out only after an early heart attack.
- Without treatment, adults with FH often have LDL of 190 to 400 mg/dL. Children with FH often have LDL above 160. Normal LDL for a healthy adult is under 130.
- LDL has been high since birth. So the arteries build up plaque for 30 or 40 years before anyone notices. Heart attacks can hit in the 30s and 40s for men, and the 40s and 50s for women, if not treated.
- The very rare form — HoFH — means you got a bad gene from BOTH parents. LDL can top 500. Heart attacks can happen in childhood without urgent treatment.
- FH is passed down through families. One bad copy is enough. Each child of an affected parent has a 50% chance. It shows up in every generation.
- FH has NO symptoms. You feel normal while plaque builds silently. The only way to find FH is a blood test.
- The great news: FH can be treated. People found and treated early — even as children — can have a near-normal life. Finding it early can save your life.




*Real photos of the two classic physical signs of FH. A-B: tendon xanthomas (firm cholesterol bumps) on the elbow. C-D: arcus lipoides (a pale ring at the cornea's edge), same patient. E-F: the xanthomas shrinking after LDL-apheresis — proof they can regress once LDL comes down. Image: Alnouri et al., Global Heart 2020 (CC BY 4.0).*




*LDL over a lifetime. Red line: untreated FH — high from birth. It crosses the high-risk line at about age 27. Orange dashed: treated FH — statin started at age 10 keeps LDL well below the risk line. Green: a typical person without FH. Every year at a lower LDL is a year of safer arteries. The earlier treatment starts, the better. Source: EAS FH natural history data, 2013.*

## How FH is diagnosed: the clinical clues

| Clue | What it means | Points (DLCN) |
|---|---|---|
| LDL 190 mg/dL or higher (adult) or 160 mg/dL or higher (child) | Primary biochemical criterion for FH evaluation | 1–8 pts |
| Tendon xanthomas (cholesterol deposits in Achilles or knuckle tendons) | Near-diagnostic physical finding for FH | 6 pts |
| Corneal arcus before age 45 | Pale ring around eye cornea — FH clue in younger adults | 4 pts |
| First-degree relative with LDL 190 or higher, or early heart disease | Family history score — parent, sibling, or child affected | 1–6 pts |
| Premature CAD in patient (men < 55, women < 65) | Early personal heart disease raises FH probability | 2 pts |
| Confirmed LDLR / APOB / PCSK9 mutation on genetic testing | Definitive diagnosis — enables precise cascade screening | 8 pts |

## Why It Matters

- Without treatment, FH raises the risk of early heart disease by about 20 times. Half of men with untreated FH have a heart attack by age 50. About 30% of women do by age 60.
- Years of high LDL add up. Plaque builds slowly — for decades. The longer your LDL stays high, the more damage piles up. Starting treatment early saves artery health for life.
- Finding it early changes everything. People treated before their first heart event do much better. Finding it late — after a heart attack — means catching up with arteries that are already damaged.
- Family testing multiplies the benefit. When one person is found to have FH, testing their parents, siblings, and children finds 1 to 2 more people on average. They can start treatment before harm is done.
- FH is widely missed. Of about 1.3 million Americans with FH, fewer than 10% know. This gap costs lives.
- Treatment works. Statins, ezetimibe, and PCSK9 shots can lower LDL by 60 to 80%. The FOURIER and ODYSSEY trials showed this prevents heart attacks and strokes.

## Cascade Screening: Test Your Whole Family

- Once you are found to have FH, every parent, sibling, and child should be tested. Each one has a 50% chance. Most of them have no idea they are at risk.
- For every 1 person found, testing the family finds 1 to 2 more people on average. They can start treatment before any harm is done.
- Children of an FH parent should be tested at age 9 to 11. For the rare double form, test at age 2. Starting treatment early stops plaque from building for decades.
- The test is a simple blood draw (lipid panel). If LDL is very high, a gene test may follow. Many insurers cover gene testing for family members once the first mutation is confirmed.
- A gene test shows the exact flaw. Family members who do NOT have the flaw can be fully reassured. Those who do have it can start medicine right away.
- Free help: the FH Foundation (thefhfoundation.org) and Family Heart Foundation (familyheart.org) both offer free screening kits and guidance for families.
- Share this guide at your next family event. Ask your doctor to write a family letter. Do not wait until a relative has a heart attack to act.

**How FH is scored: Dutch Lipid Clinic Network Criteria.**
Your doctor adds points for LDL level, physical clues, family history, and personal heart history. **Score 8 or more = Definite FH. Score 6–7 = Probable FH. Score 3–5 = Possible FH.** A gene test can confirm any result. See the clue table in the section above.

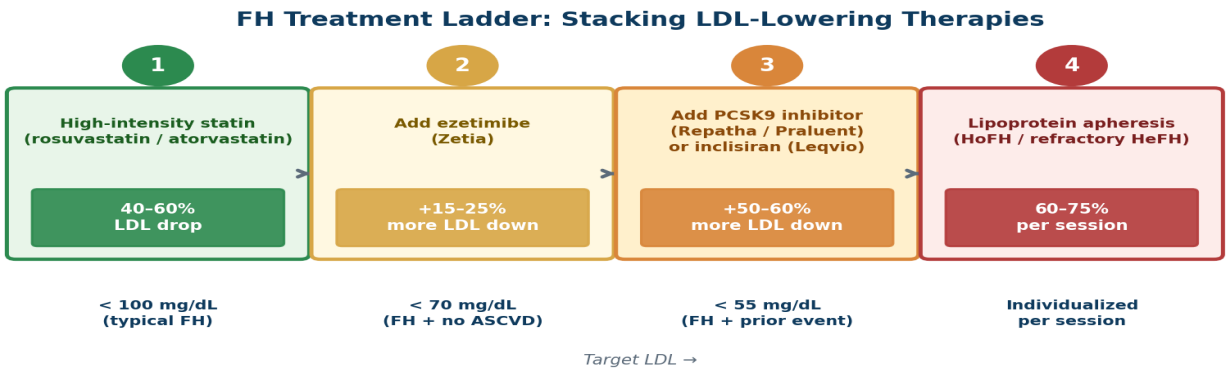
## Risk Factors

Some patients are more likely to develop this condition. Knowing your risk helps your care team take extra precautions.

| Risk Factor | Why It Increases Risk |
|---|---|
| Parent, sibling, or child with FH or early heart disease | Each first-degree relative has a 50% chance of having FH. A heart attack in a male relative before 55, or female before 65, is a warning sign. Get a lipid panel. |
| LDL at or above 190 mg/dL in an adult | The main lab clue for FH in adults. LDL this high almost always has a genetic cause. Even a family history of normal cholesterol does not rule it out. |
| LDL at or above 160 mg/dL in a child | The lab clue for FH in kids. Routine cholesterol testing at ages 9 to 11 and again at 17 to 21 is recommended for all children. |
| Yellow tendon lumps (tendon xanthomas) | Bumps of cholesterol over the Achilles tendon or knuckles. Strong clue for FH. Seen in about 10 to 15% of FH adults. |
| Pale ring around the eye before age 45 | Called corneal arcus. Normal in older adults. In younger adults it points to very high LDL and FH. |
| Yellow patches around the eyelids | Called xanthelasma. Less specific than tendon lumps, but worth checking alongside high LDL. |
| Gene mutation found on genetic testing | A confirmed LDLR, APOB, or PCSK9 mutation is the final word. It allows precise family testing. May also help with drug approval. |
| Both parents have very high LDL | The rare double form (HoFH) needs both parents to carry a gene flaw. LDL above 300 and tendon lumps in childhood are warning signs. |

## Treatment Options

- **Step 1 — High-dose statin, for life.** Rosuvastatin or atorvastatin at the highest safe dose. Lowers LDL by 40 to 60%. Adults start right after diagnosis. Children may start as young as age 8 to 10. Safe and tested in kids. See go.riasalimd.com/statins-guide.
- **Step 2 — Add ezetimibe (Zetia).** A pill that blocks cholesterol from being absorbed. Lowers LDL another 15 to 25%. Taken once a day. Very few side effects. Most people with FH need this on top of a statin.
- **Step 3 — PCSK9 inhibitors: Repatha or Praluent.** A shot given every 2 weeks or once a month. Lowers LDL another 50 to 60% on top of a statin. Approved by the FDA for FH. The FOURIER (2017) and ODYSSEY (2018) trials showed this prevents more heart attacks. See go.riasalimd.com/pcsk9-guide.
- **Inclisiran (Leqvio) — two shots per year.** Works by silencing the gene that breaks LDL receptors. Lowers LDL about 50% on top of a statin. Only 2 injections per year makes it very easy to stay on track.
- **Bempedoic acid (Nexletol) — for statin-intolerant patients.** A non-statin pill. Lowers LDL about 20%. Good choice if muscle pain from statins is a problem. The CLEAR Outcomes trial showed it prevents heart events. See go.riasalimd.com/bempedoic-guide.
- **Evinacumab (Evkeeza) — only for the rare double form (HoFH).** A monthly infusion that lowers LDL by 49%. Works even when LDL receptors do not work at all. The ELIPSE trial (2020) proved this in HoFH patients.
- **Blood filtering (lipoprotein apheresis).** A 3 to 4 hour process that washes LDL out of the blood, like dialysis for cholesterol. Used for severe or double-gene FH when pills alone are not enough. Lowers LDL by 60 to 75% per session.
- **LDL goals in FH:** Under 100 for most adults with FH. Under 70 if you also have diabetes or other risk factors. Under 55 if you have already had a heart attack or stroke.




*FH treatment ladder. Step 1: high-dose statin (40 to 60% LDL drop). Step 2: add ezetimibe (15 to 25% more). Step 3: add a PCSK9 shot or inclisiran (50 to 60% more). Step 4: blood filtering for severe or double-gene FH. LDL goals at bottom: under 100 for most; under 70 or 55 for higher-risk cases. Source: 2018 ACC/AHA guideline.*

## Treatment to Target: What 'Enough' Looks Like

- FH patients need lower LDL goals than most people. This is because LDL has been high since birth. Decades of damage need a very low target to stop more harm.
- Most adults with FH: aim for LDL under 100. If you also have diabetes or high blood pressure: aim for under 70.
- FH plus a prior heart attack or stroke: aim for under 55. This usually needs a statin plus ezetimibe plus a PCSK9 shot.
- Recheck LDL 8 to 12 weeks after any drug change. Once at goal, recheck every 6 to 12 months.
- On max statin plus ezetimibe and still above goal? Tell your heart doctor. A PCSK9 shot or inclisiran is the next step. Insurance often covers it for FH.
- Your goal may be lower than your friends' goals. That is normal for FH. The lower targets are safe and backed by large trials.
- Ask your doctor two questions: What is my LDL goal? Am I there yet? If the answer is no, ask what comes next.

**Statin side effects — what to do.**
About 1 in 10 patients get muscle aches on a statin. Most can keep going after switching brands or cutting the dose. If statins really do not work for you, bempedoic acid plus ezetimibe plus a PCSK9 shot can do most of the same job. Do not stop your medicine without calling us first. See: go.riasalimd.com/sams-guide.

## Comfort Measures at Home (No Medication Needed)

These simple steps support healing and ease symptoms. Use them alongside any medication your doctor prescribes.

- Eat a heart-healthy diet — Mediterranean or DASH style. Diet alone will NOT fix FH. The problem is in your genes. But a good diet can still lower LDL another 5 to 10% and protect your heart in other ways.
- Limit butter, fatty meat, full-fat dairy, coconut oil, and palm oil. Swap to olive oil, nuts, fish, and avocado. These swaps are helpful but modest for FH.
- Eat more soluble fiber. Good sources: oats, beans, lentils, apples, and psyllium (Metamucil). Aim for 10 to 25 grams a day. Lowers LDL 5 to 7% more.
- Move your body 150 minutes a week. Brisk walking, swimming, or cycling all count. Exercise will not normalize FH-level LDL on its own, but it protects your heart in many other ways.
- Do not smoke. Smoking makes FH much worse. It is one of the most important steps you can take.
- Keep blood pressure and blood sugar in check. FH plus high blood pressure or diabetes is a very high-risk mix. Aim for blood pressure below 130/80.
- Tell your family. If you have FH, each parent, sibling, and child has a 50% chance of having it too. Ask them to get tested. This is the most powerful step you can take beyond your own treatment.

## Risks, Benefits, and Alternatives

Every choice has trade-offs. Use this table to start the shared decision conversation.

| Option | Risks | Benefits | Alternatives |
|---|---|---|---|
| High-dose statin | Muscle aches in 5 to 10% (usually mild; try a new brand or lower dose). Small blood-sugar rise in pre-diabetics. Very rare liver-enzyme rise. Stop during pregnancy. | Lowers LDL 40 to 60%. Proven to prevent heart attacks and death in FH. Safe for children from age 8 to 10. | Every-other-day dosing if daily causes aches. Try a different statin. Bempedoic acid if truly statin-intolerant. |
| Ezetimibe added to statin | Very few side effects. Mild stomach upset in some. LDL drop of 15 to 25% on top of a statin. | Lowers LDL another 15 to 25%. IMPROVE-IT trial: cut heart events 6% further after MI. Pill, once daily. Cheap generic available. | Higher statin dose first. Add a PCSK9 shot if LDL is still above goal. |
| PCSK9 inhibitor (Repatha / Praluent) | Shot every 2 weeks or once a month. Sore at the shot site. Costs more — needs insurance approval. Well tolerated. | Lowers LDL 50 to 60% on top of statin. FOURIER trial: 15% fewer heart events. ODYSSEY trial: 15% fewer events in high-risk patients. FDA-approved for FH. | Inclisiran (twice-yearly shots). Bempedoic acid if shots are not an option. |
| Inclisiran (Leqvio) | Two shots per year. Mild soreness at the shot site. Fewer long-term trial data than the PCSK9 shots. | About 50% LDL drop on top of statin. Only 2 shots a year makes it easy to stay on track. Effect lasts months between shots. | PCSK9 inhibitors have more trial data. Higher statin dose plus ezetimibe. |

| Option | Risks | Benefits | Alternatives |
|---|---|---|---|
| Blood filtering (apheresis) | Takes 3 to 4 hours each session, every 1 to 2 weeks. Needs a special center. LDL rises between sessions. | Cuts LDL 60 to 75% per session. For severe FH when pills are not enough. Reduces heart events in this group. | Try all medicines first. Evinacumab (Evkeeza) may reduce the need for apheresis. |
| No treatment | Heart attacks in the 30s to 50s for men, 40s to 50s for women. 20 times higher risk. Plaque builds in silence for decades. | No drug costs or side effects. | Lifestyle changes (diet, exercise) are the gentlest step. They help some, but will not fix FH-level LDL. |

## Common Misconceptions

| Myth | Reality |
|---|---|
| "My cholesterol is high because I eat badly." | FH is in your genes. The broken gene stops the liver from clearing LDL no matter what you eat. A good diet helps a little. But it will never bring an LDL of 250 to a safe level. Medicine is a must. |
| "Kids are too young to have high cholesterol." | FH starts at birth. Kids with FH have high LDL and start building plaque in childhood. Statins are safe for FH children from age 8 to 10. Treating early stops heart attacks from happening later. |
| "I feel fine, so my FH is not dangerous." | FH has no symptoms. You feel normal while plaque builds up. The plaque has been there since childhood. Only a blood test can reveal the risk. |
| "Only one person in my family needs to be tested." | FH is passed from parent to child. Each parent, sibling, and child has a 50% chance. Testing your family finds them BEFORE they have a heart attack. This is the top action after your own diagnosis. |
| "High LDL but no heart attack yet — so I must be OK." | FH is a slow burn. Damage builds for decades. Many people live into their 40s before the first event. No past heart attack does NOT mean you are safe. |
| "Statins cause liver damage." | Serious liver harm from statins is very rare — under 1 in 100,000 patients. Mild liver-enzyme rises are common but almost never need action. The benefit for FH far outweighs this small risk. |
| "PCSK9 shots are too new to trust." | PCSK9 shots have been tested in trials with over 27,000 patients. FOURIER and ODYSSEY both showed fewer heart attacks and strokes. No major safety concerns after 2 or more years. FDA-approved and in the guidelines for FH. |

| Myth | Reality |
| --- | --- |
| "My LDL looks normal on a statin, so I can stop." | LDL looks normal BECAUSE of the statin. Stopping brings it right back to FH levels within weeks. FH treatment is lifelong. |

## Possible Complications

Knowing what can go wrong helps you spot problems early. Most complications are uncommon, especially with treatment.

| Where / What | What Can Happen |
|---|---|
| Early heart attack | Heart attacks in the 30s to 50s from decades of LDL buildup. The top cause of death in untreated FH. Often affects more than one heart artery. |
| Aortic valve disease | LDL deposits on the aortic valve make it stiff and narrow. FH patients need valve repair earlier in life. |
| Stroke | Plaque in the neck arteries can block blood flow to the brain. FH speeds up this process the same way it does in heart arteries. |
| Leg artery disease (PAD) | Plaque in leg arteries causes pain when walking. Can lead to wounds that won't heal and, if untreated, limb loss. |
| Tendon rupture | Cholesterol lumps weaken the tendon. Achilles tendon tears happen more often in FH patients who have these lumps. |
| Second heart events | After a first heart attack, FH patients have higher odds of a second one. The root cause — high LDL — has been active for decades. Aggressive treatment after the first event is a must. |

## FH in Children: Starting Early Saves Arteries

- FH starts at birth. By age 10, kids with FH already have thicker artery walls than kids without it. Plaque builds early and silently.
- All children should have a cholesterol check at age 9 to 11 and again at age 17 to 21. FH is found in most kids with LDL above 160.
- Statins are safe for FH kids from age 8 to 10. Decades of study and the Dutch FH registry show they are safe and cut heart risk in young patients.
- A 2015 review in the New England Journal of Medicine (Wiegman) showed that FH kids treated early had far fewer heart events as adults than those not treated.
- Statins in childhood do NOT hurt growth, puberty, or muscles at the right dose. Regular cholesterol checks are still needed.
- A good diet and exercise are important for FH kids. But they cannot fix the gene flaw. Medicine is needed once LDL is clearly high.
- Tell your child's doctor about your FH diagnosis. Ask for a cholesterol check for your children. Early treatment in kids is the most powerful step in FH care.

## Points to Know

If you remember nothing else, remember these key points.

- FH is genetic. High LDL from birth. Diet alone will not fix it. Medicine is a must.
- 1 in 250 people have FH. Fewer than 1 in 10 know it. Without treatment: heart attacks in the 30s to 50s.
- Key clues: LDL above 190 mg/dL (adults) or 160 mg/dL (kids), tendon lumps, pale ring around the eye before 45, early heart disease in the family.
- Top step after diagnosis: test your family. Each parent, sibling, and child has a 50% chance.
- First-line: high-dose statin. Most FH patients also need ezetimibe. Many need a PCSK9 shot to reach their LDL goal.
- LDL goals are low: under 100 for most; under 70 with added risk; under 55 after a heart attack.
- Statins are safe for FH children from age 8–10. Early treatment normalizes lifetime risk. The earlier, the better.
- Treatment is lifelong. Stopping the statin returns LDL to FH-level within weeks and restarts the clock on plaque buildup.

## When to Call Us - and When to Call 911

If you are not sure, call. We would rather hear from you twice than miss a real problem.

- **Call 911** for sudden chest pain, pressure, or tightness; trouble breathing; weakness on one side; trouble speaking; or fainting. These are heart attack and stroke signs. Call at once. Do not drive yourself.
- **Call our office (727-943-5200)** if your LDL is above 190 mg/dL and you have not been checked for FH. We will review your family history and discuss testing.
- **Call us** if a parent, sibling, or child has been found to have FH or had an early heart attack. We can help set up family testing.
- **Call us** if you have new muscle aches, weakness, or dark urine after starting or changing a statin. We will check labs and adjust your dose.
- **Call us** if you are pregnant or trying to get pregnant. Statins must be stopped before you try. We will plan a safe approach during pregnancy.
- **Call us** if your LDL is still above 100 on a full-dose statin plus ezetimibe. A PCSK9 shot or inclisiran may be the next step. We can handle the insurance paperwork.
- **Call us** if you want to discuss gene testing for yourself or your children. Knowing the exact gene flaw helps your whole family.

**Office: (727) 943-5200**

**See also: companion guides from Dr. Ali.**
Understanding Cholesterol · Statin Therapy · PCSK9 Inhibitors · Lipoprotein(a) · Statin-Associated Muscle Symptoms · Bempedoic Acid · Coronary Artery Disease.

## Trusted Resources

The links below are the trusted resources Dr. Ali used to write this guide and that he recommends you visit for more information. Each is a clickable link in the digital version and a Short.io tracked URL on the printed version.

- FH Foundation — U.S. Patient Organization - Patient-focused nonprofit with FH diagnosis resources, cascade screening toolkit, specialist finder, and support communities for FH patients and families.
- Family Heart Foundation - Advocacy org for FH and Lp(a) — cascade screening guides, testing resources, and patient stories from those living with genetic lipid disorders.
- AHA — Familial Hypercholesterolemia - American Heart Association's patient overview of FH — what it is, how it is diagnosed, and why family testing matters.
- Cleveland Clinic — Familial Hypercholesterolemia - Plain-language guide to FH symptoms, physical signs, diagnosis criteria, and treatment options.
- Mayo Clinic — Familial Hypercholesterolemia - Mayo's patient page — genetic causes, when to see a doctor, and overview of current treatments.
- NIH MedlinePlus — Familial Hypercholesterolemia - Federal resource on FH genetics, inheritance pattern, and links to clinical databases and Spanish-language materials.
- ACC/AHA Cholesterol Guideline (patient summary) - ACC's 10-point plain-language summary of the 2018 guideline — LDL targets, when to add PCSK9, and FH management principles.
- ASCVD Risk Estimator Plus (ACC/AHA) - Free online tool that calculates your 10-year and lifetime heart-attack and stroke risk — helpful for understanding why your LDL target is as low as it is.
- Understanding Cholesterol — Companion Guide - Dr. Ali's full guide to LDL, HDL, triglycerides, Lp(a), and cholesterol management — the foundational companion to this FH guide.
- PCSK9 Inhibitors — Companion Guide - Explains how evolocumab (Repatha) and alirocumab (Praluent) work, who qualifies, how to get prior authorization, and what to expect from the injection.

# Sources Used to Build This Guide

Every recommendation in this guide is grounded in one or more of the studies, guidelines, or trusted resources below. Short links are tracked so Dr. Ali can see which references patients are reading; nothing personally identifying is captured.

- AHA FH Scientific Statement 2015 (Gidding et al.) *[Guideline]*
  AHA's foundational scientific statement on familial hypercholesterolemia — prevalence, diagnosis, cascade screening, treatment recommendations.
- ACC/AHA 2018 Cholesterol Management Guideline (Grundy et al.) *[Guideline]*
  Primary treatment guideline — LDL targets by risk tier, PCSK9 inhibitor use, FH management, statin and add-on therapy recommendations.
- EAS Consensus on Familial Hypercholesterolemia 2013 (Nordestgaard et al.) *[Guideline]*
  European Atherosclerosis Society consensus — global prevalence estimate, Dutch Lipid Clinic Network criteria, treatment thresholds, cascade screening.
- Dutch Lipid Clinic Network (DLCN) Diagnostic Criteria for FH *[Clinical Criteria]*
  The primary validated clinical scoring system for FH diagnosis — family history, clinical signs, LDL level, genetic testing.
- Simon Broome FH Register Diagnostic Criteria *[Clinical Criteria]*
  UK FH register diagnostic criteria — used alongside DLCN for definite vs probable FH classification.
- FOURIER Trial — Sabatine et al. NEJM 2017 *[Trial]*
  Landmark trial: evolocumab (PCSK9 inhibitor) reduced LDL by ~59% and cut cardiovascular events by 15% in high-risk patients — cited for PCSK9 efficacy data.
- ODYSSEY OUTCOMES Trial — Schwartz et al. NEJM 2018 *[Trial]*
  Alirocumab (PCSK9 inhibitor) reduced major cardiovascular events by 15% post-ACS — supports PCSK9 use in FH patients with ASCVD.
- Pediatric FH Screening — Wiegman et al. NEJM 2015 *[Review]*
  Authoritative review of FH in children — early statin use, pediatric screening rationale, long-term safety and efficacy data.
- FH Foundation — Patient Resources *[Patient Org]*
  U.S.-based nonprofit for FH patients — diagnosis resources, cascade screening toolkit, patient stories, specialist finder.
- Family Heart Foundation — FH and Cascade Screening *[Patient Org]*
  Cascade screening resources, FH diagnosis support, Lp(a) testing advocacy. Companion to FH Foundation.
- Cleveland Clinic — Familial Hypercholesterolemia *[Patient Education]*
  Plain-language patient overview used as a readability benchmark for section framing and patient-voice tone.
- Mayo Clinic — Familial Hypercholesterolemia *[Patient Education]*
  Mayo Clinic's patient overview for plain-language framing of symptoms, genetic basis, and treatment ladder.
- NIH MedlinePlus — Familial Hypercholesterolemia *[Patient Education]*
  Federal-level patient resource — genetic mechanism, inheritance pattern, multilingual patient links.
- Evinacumab (ELIPSE HoFH Trial) — Raal et al. NEJM 2020 *[Trial]*
  ANGPTL3 inhibitor evinacumab for homozygous FH — 49% LDL reduction; key data for HoFH therapy section.

- Inclisiran — ORION-10 Trial (Ray et al. NEJM 2020) *[Trial]*
  siRNA-based PCSK9 silencer inclisiran — 52% LDL reduction with twice-yearly dosing; rationale for inclusion in FH treatment ladder.

# About This Guide

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**Reviewer note:** Reviewed Cleveland Clinic, Mayo Clinic, NIH MedlinePlus, FH Foundation, and Family Heart Foundation FH pages. This guide adds: a pedigree diagram showing 50% autosomal-dominant inheritance; a lifetime LDL curve comparing untreated vs. treated FH; a 4-step treatment ladder with percent LDL reductions; dedicated cascade screening, pediatric FH, and bempedoic acid / inclisiran sections absent from most patient pages; and DLCN diagnostic criteria in accessible language.

## Disclaimer

This guide is for educational purposes only and does not replace medical advice from your physician. Recommendations may not apply to every patient. Always follow the specific instructions of Dr. Ali and your care team. In an emergency call 911 or go to the nearest emergency department.

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