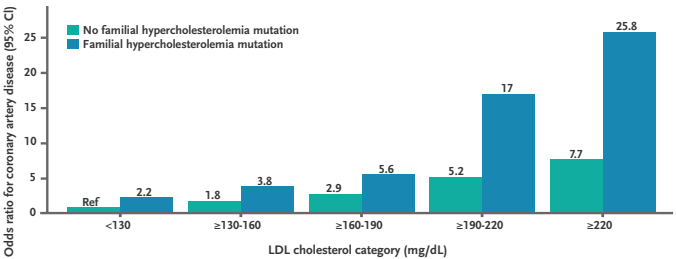
A better indicator of risk: Genetic testing for familial hypercholesterolemia

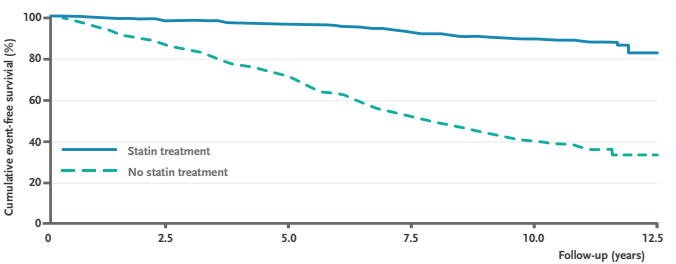
Familial hypercholesterolemia (FH) affects approximately 1 in 220 people, and more than a million people in the US have FH and are undiagnosed.

An established genetic diagnosis of FH indicates a **substantially higher** risk of coronary artery disease (CAD) than clinical diagnosis alone.



Impact of familial hypercholesterolemia mutation status on coronary artery disease according to LDL cholesterol level. Adapted from Khera AV et al. *2016.*

If FH is identified early and treated aggressively, **morbidity and mortality are reduced by 80%**.



Kaplan-Meier curve estimates of cumulative coronary heart disease-free survival among patients with FH according to statin treatment (P<0.001 for difference). Adapted from Versmissen J et al. *2008.*

Genetic testing also enables **life-saving early interventions for at-risk family members**. Family members could be at increased risk even if their cholesterol levels are normal. Cascade genetic testing of family members is recommended by JACC scientific expert panel and the CDC after a patient receives a genetic diagnosis of FH.

WHO TO TEST

JACC scientific expert panel and CDC recommendations:

Genetic testing for FH **should be offered** when there is:

- No apparent secondary cause of hypercholesterolemia and persistent levels are
  - LDL-C ≥190 mg/dL in adults
  - LDL-C ≥160 mg/dL in children
- Known FH-causing genetic variant present in family

Genetic testing **enables life-saving early interventions for at-risk family members;** even family members with normal cholesterol levels could be at increased risk.

The likelihood of FH increases when the following are present:

- Cholesterol deposits on the
  - skin (xanthalasma, tuberous xanthomas)
  - tendons (tendon xanthomas)
  - cornea (arcus corneae)
- First-degree relative with
  - LDL-C ≥190 mg/dL
  - premature CAD (<55 years for males, <65 years for females)

Questions? Call Invitae Clinical Consult at 800-436-3037. Learn more about Invitae's high-quality, affordable genetic testing at www.invitae.com/cardiology.

References:

1. Sturm AC *et al.* Clinical Genetic Testing for Familial Hypercholesterolemia: JACC Scientific Expert Panel. *J Am Coll Cardiol.* 2018;72(6):662-680.
2. Goldberg A, *et al.* Familial hypercholesterolemia: screening, diagnosis and management of pediatric and adult patients. *J Clin Lipidol.* 2011;5(3 Suppl):S1-8.
3. Khera AV *et al.* Diagnostic yield and clinical utility of sequencing familial hypercholesterolemia genes in patients with severe hypercholesterolemia. *J Am Coll Cardiol.* 2016;67(22):2578-89.
4. Versmissen J *et al.* Efficacy of statins in familial hypercholesterolemia: a long term cohort study. *BMJ.* 2008;337:a2423.
5. Centers for Disease Control and Prevention. Public Health Genomics Database. https://phgkb.cdc.gov/PHGKB/topicFinder.action. Accessed December 2017.

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