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.

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

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 $\geq 190$ mg/dL in adults
  - LDL-C $\geq 160$ 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 (xanthelasma, tuberous xanthomas)
  - tendons (tendon xanthomas)
  - cornea (arcus corneae)
- First-degree relative with
  - LDL-C $\geq 190$ mg/dL
  - premature CAD (<55 years for males, <65 years for females)


References: