

Genetically inherited high cholesterol increases long-term risks of CHD and stroke

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Patients who experience high cholesterol due to an inherited genetic disorder from one of their parents—heterozygous familial hypercholesterolemia—are much more likely than those with average cholesterol levels to have diseases caused by hardening of the arteries, including an accelerated onset of coronary heart disease by up to 30 years, according to new research in the American Heart Association journal *Circulation*.

The inherited form of high [cholesterol](#)—familial hypercholesterolemia—is a genetic disorder that is passed down through families. Individuals with familial hypercholesterolemia have a genetic mutation that prevents the liver from removing excess low-density lipoprotein (LDL), known as "bad" cholesterol, from their blood. Familial hypercholesterolemia is suspected when an individual has an LDL cholesterol level greater or equal to 190 mg/dL (which was considered the familial hypercholesterolemia "phenotype" in this paper) in the setting of a family history of premature cardiovascular events.

Researchers estimate that heterozygous familial hypercholesterolemia affects up to about 1.5 million people in the United States.

Using pooled data from six groups of people in epidemiological studies (with 1.2 million person-years of follow-up), researchers found that patients with the familial hypercholesterolemia phenotype were:

- at five times higher risk for coronary [heart disease](#) over the long term (up to 30 years), compared to those with average levels (less than 130 mg/dL) of LDL cholesterol; and
- more likely to have diseases caused by hardening of the arteries (atherosclerotic cardiovascular disease), including an accelerated onset of coronary heart

disease by up to 20 years earlier in men and 30 years earlier in women.

These increased risks were independent of other risk factors.

Researchers say that their findings may help clinicians communicate the risks of familial hypercholesterolemia more clearly to patients, which is important because familial hypercholesterolemia can be treated with cholesterol-lowering drugs to decrease the risks for coronary heart disease and stroke.

"Clinician-patient discussions about guideline-supported therapies can be informed by this data, as in the following scenario: a 25-year-old woman with newly diagnosed familial hypercholesterolemia can be informed that at her current age, if her cholesterol were to remain untreated, her risk of coronary heart disease death or nonfatal heart attack is comparable to that for a 55-year-old woman. Such an analogy, paired with counseling about how to improve risk, may motivate behavioral changes as well as adoption of and adherence to evidence-based medications," researchers said.

More information: *Circulation*, [DOI: 10.1161/CIRCULATIONAHA.116.022335](#)

Provided by American Heart Association

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