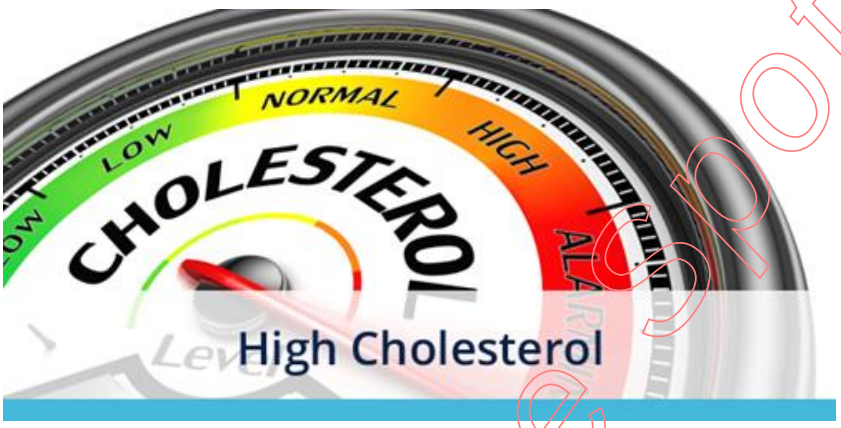
Genetic Science Spotlight

Mass Gen reports, Single-gene Mutations Account for 2% of Cases of Severely Elevated Cholesterol



An international research team led by scientists from Massachusetts General Hospital finds that familial hypercholesterolemia (FH) genetic mutations account for 2% of cases with severely elevated LDL. The researchers sequenced blood samples from 26,025 participants in 12 studies -- seven comparing individuals with coronary artery disease to healthy controls and five long-term epidemiologic studies -- conducted in Europe, North America and South Asia. While the presence of severe hypercholesterolemia without a FH mutation conferred a six-fold greater risk of coronary artery disease than did LDL levels below 130 mg/dl, adding an FH mutation to elevated cholesterol resulted in a 22-fold increased risk. These findings suggest that, beyond simply measuring LDL, gene sequencing may identify individuals at very high risk who need additional preventive therapies. Among people who have the mutation, half of their first-degree relatives are also carriers, indicating a need to advise testing among their family members.

https://www.sciencedaily.com/releases/2016/04/160403195924.htm

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