Genetic testing recommended for familial hypercholesterolemia

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"Expected outcomes include greater diagnoses, more effective cascade testing, initiation of therapies at earlier ages, and more accurate risk stratification," the authors write.

Several authors disclosed financial ties to the diagnostics and the pharmaceutical industries.

More information: Abstract/Full Text (subscription or payment may be required)

(HealthDay)—Genetic testing should become the standard of care for patients with definite or probable familial hypercholesterolemia (FH), according to a statement published in the Aug. 7 issue of the *Journal of the American College of Cardiology*.

Amy C. Sturm, from Geisinger in Danville, Pa., and colleagues from the JACC Scientific Expert Panel were convened by the Familial Hypercholesterolemia Foundation to assess the utility of FH genetic testing.

The expert panel recommends that FH genetic testing become the standard of care for patients with definite or probable FH, as well as for their at-risk relatives. Specifically, the panel recommends that testing include the genes encoding the low-density lipoprotein receptor, apolipoprotein B, and proprotein convertase subtilisin/kexin 9. Depending on patient phenotype, other genes may also need to be considered for analysis.