# Study shows most familial hypercholesterolemia patients remain undiagnosed

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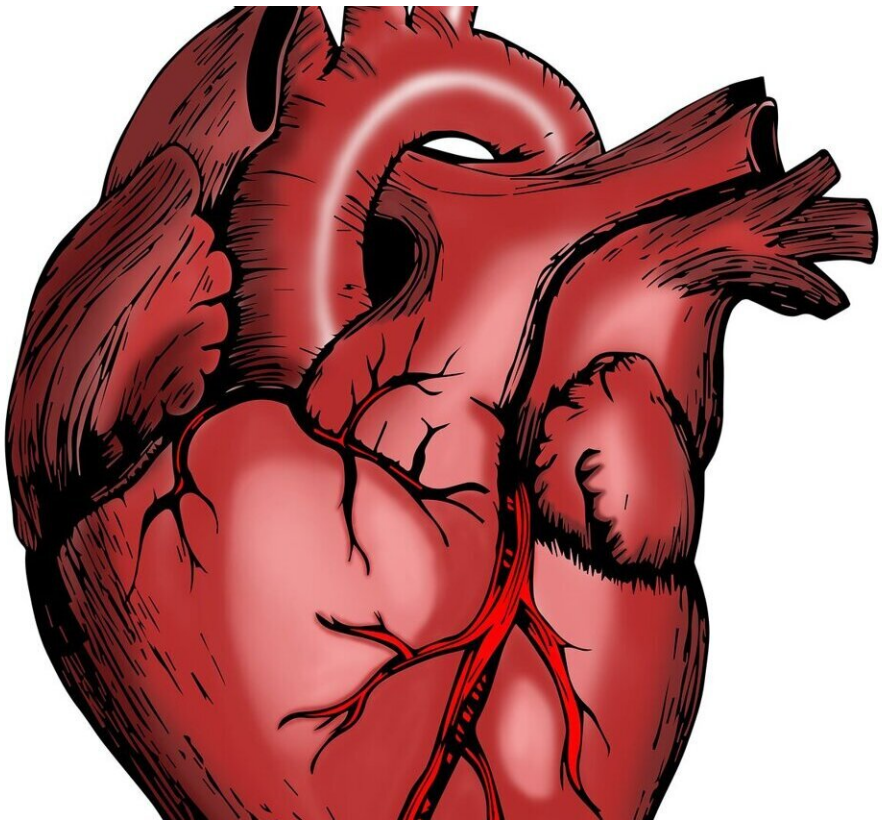

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The Family Heart Foundation, a leading research and advocacy organization, completed an analysis of its large U.S. Family Heart

Database showing that while the percentage of patients diagnosed with familial hypercholesterolemia (FH) has increased substantially since 2016, most remain undiagnosed. The data is being presented in a poster titled, "Diagnosis of Familial Hypercholesterolemia: A Work in Progress," at the American Society for Preventive Cardiology Congress on CVD Prevention on July 29-31 in Louisville, Kentucky.

"Despite clear screening guidelines, classification of FH as a public health threat by the World Health Organization, and the availability of effective therapies, most of the FH population remains undiagnosed, leaving these genetically vulnerable individuals at high risk for premature cardiovascular disease," said Mary P. McGowan, M.D., chief medical officer, Family Heart Foundation, and study co-author. "It is critical that we continue to differentiate FH from other conditions as management in the first two decades of life can substantially reduce the burden of aggressive atherosclerosis. This analysis shows we are moving in the right direction, but there is still more work to be done."

Data was analyzed using the Family Heart Foundation's large, real-world, U.S. healthcare database. The analysis included 197 million people from the U.S. with diagnostic data from claims who were screened or treated for any form of cardiovascular risk from October 2016 through June 2020. The study found that as of June 2020, 31.3% of the estimated 787,886 individuals in the Family Heart Foundation Database who had FH (diagnosed and undiagnosed) had been diagnosed. Patients diagnosed with FH were counted if the diagnostic code was applied for a single in-patient claim or at least twice, more than seven days apart, for an out-patient claim. Published data from 2013 indicated that less than 1% of FH patients in the U.S. were previously diagnosed. In addition, compared with all individuals in the database, those diagnosed with FH were substantially more likely to have ASCVD.

In 2013, the Family Heart Foundation introduced a proposal to the

ICD-10 Coordination and Maintenance Committee to establish an ICD-10 code for FH. The code (E78.01) became effective in October 2016, giving patients with FH a more accurate diagnosis and making it possible to track improvements in the diagnosis rate, as evidenced by this study.

The abstract will be available for viewing throughout the Congress in the Poster Hall.

**More information:** Conference: www.aspconline.org/2022congress/

B. G. Nordestgaard et al, Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease: Consensus Statement of the European Atherosclerosis Society, *European Heart Journal* (2013). DOI: 10.1093/eurheartj/eht273

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