

Nearly 1 in 5 patients who die from unexplained sudden cardiac death have suspicious gene

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As many as 450,000 Americans die every year from a sudden, fatal heart condition, and in slightly more than one in ten cases the cause remains

unexplained even after an autopsy. Researchers from the University of Maryland School of Medicine (UMSOM) and their colleagues found that nearly 20 percent of patients with unexplained sudden cardiac death—most of whom were under age 50—carried rare genetic variants. These variants likely raised their risk of sudden cardiac death. In some cases, their deaths may have been prevented if their doctors had known about their genetic predisposition to heart disease. The study findings were published last week in *JAMA Cardiology*.

"Genetic screening isn't routinely used in cardiology, and far too many patients still die suddenly from a [heart](#) condition without having any previously established risk factors. We need to do more for them," said study corresponding author Alope Finn, MD, Clinical Associate Professor of Medicine at UMSOM.

To conduct the study, Dr. Finn and his colleagues performed genetic sequencing in 413 patients, who died at age 41 on average of sudden unexplained heart failure. Nearly two-thirds of the group were men, and about half were African American. The study found that 18 percent of patients who experienced sudden [death](#) had previously undetected genes associated with life-threatening arrhythmia or heart failure conditions. None of those who carried these genetic variants had been previously diagnosed with these abnormalities. Their hearts looked normal on autopsies without any signs of heart failure or significant blockages in their coronary arteries.

"What we found opens the door and asks some important questions," said Dr. Finn. "Should we be doing routine genetic screening in those who have a family history of unexplained sudden cardiac death?"

Such screening could have the potential to save lives. It may also leave patients and doctors in a quandary over what to do with such information. There are currently no clear guidelines on how to monitor

or treat patients with these variants in the absence of clinically detectable disease.

Study faculty co-authors from UMSOM include Kristen Maloney, MS, Instructor of Medicine, Libin Wang, BM Ph.D., Assistant Professor of Medicine, Susie Hong, MD, Assistant Professor of Medicine, Anuj Gupta, MD, Associate Professor of Medicine, Linda Jeng, MD, Ph.D., Clinical Associate Professor of Medicine, Braxton Mitchell, Ph.D., Professor of Medicine, and Charles Hong, MD, Ph.D., the Melvin Sharoky, MD, Professor of Medicine.

"This is a fascinating study that provides important new insights into devastating deaths due to unexplained cardiac abnormalities," said E. Albert Reece, MD, Ph.D., MBA, Executive Vice President for Medical Affairs, UM Baltimore, and the John Z. and Akiko K. Bowers Distinguished Professor and Dean, University of Maryland School of Medicine. "It certainly makes the case for more research to address this urgent health need and save lives in the future."

More information: Liang Guo et al, Genetic Variants Associated With Unexplained Sudden Cardiac Death in Adult White and African American Individuals, *JAMA Cardiology* (2021). [DOI: 10.1001/jamacardio.2021.1573](https://doi.org/10.1001/jamacardio.2021.1573)

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