Medical Xpress

Study shows benefit of routine genetic testing of dilated cardiomyopathy patients with advanced heart disease

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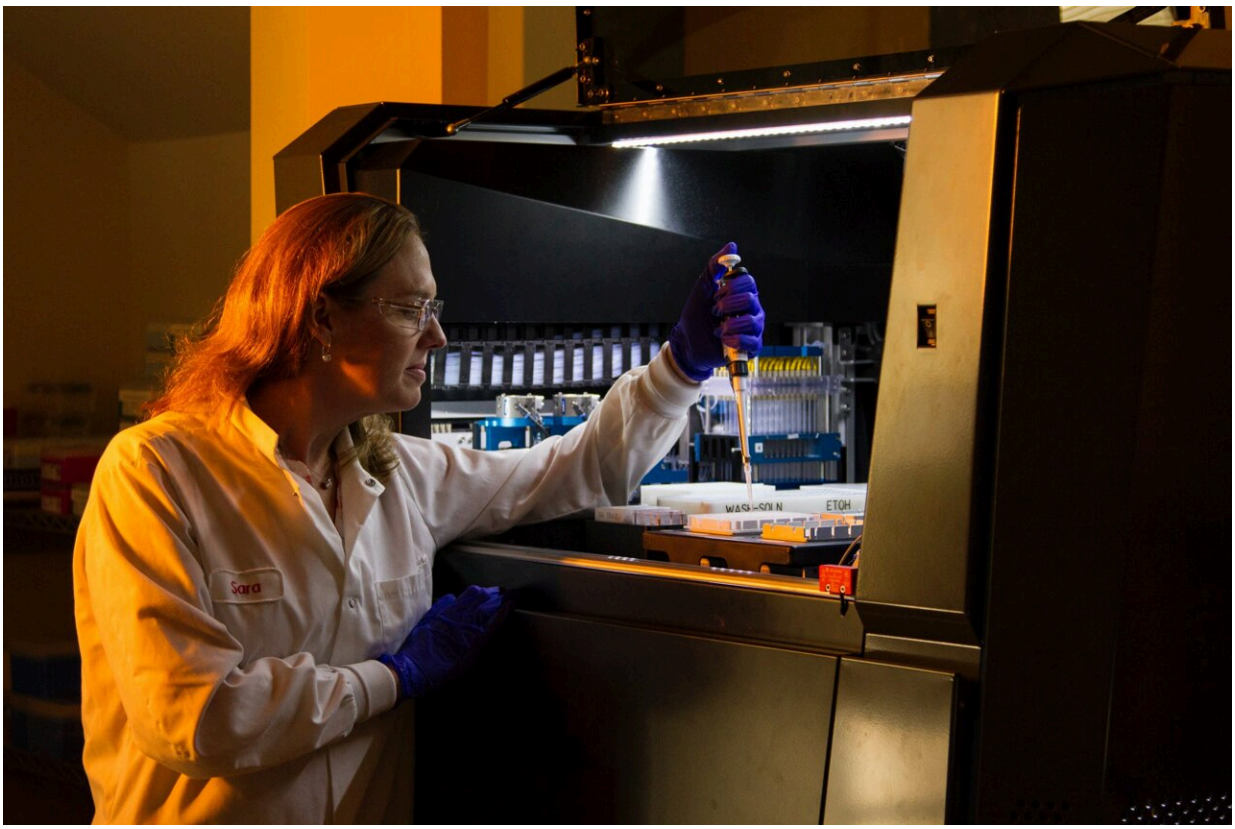

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A new study led by researchers at The Ohio State University Wexner Medical Center and College of Medicine shows the benefit of having

patients with advanced disease from dilated cardiomyopathy (DCM) undergo genetic testing to help identify the inherited heart muscle disease in family members.

Researchers found that DCM patients with advanced heart disease, defined as having a heart transplant or left ventricular assist device (LVAD), are nearly 2 ½ times more likely to have a mutation in a relevant DCM gene than those with less severe heart disease.

"These findings are highly relevant. The ultimate goal is to prevent advanced DCM, when a heart transplant or LVAD may be needed. Even though dilated cardiomyopathy is known to run in families, genetic testing is seldom done on patients who had have heart transplants or LVADs. If genetic testing were done routinely on these patients, we could identify their family members who carry the same gene mutations and start treatment with early signs of disease before the individual has advanced disease," said senior author Ray Hershberger, MD, a cardiologist and division director of Human Genetics at the College of Medicine.

Results of the study were published in *Circulation*.

DCM happens when the heart muscle weakens and the left ventricle enlarges. It can occur in family members at almost any age but the typical onset is mid 40s. Severity can vary within families with some family members exhibiting minor symptoms while others may die of heart failure or an arrhythmia causing sudden cardiac death.

Over a five-year period, researchers analyzed clinical and genetic sequence data from more than 1,200 patients at 25 leading academic U.S. heart failure/heart transplant programs that are part of the Dilated Cardiomyopathy Consortium led by Hershberger.

"This is the first time that the association of rare variant genetics with advanced DCM has been systematically studied. The study's results provide additional insight for clinicians who help DCM patients with disease management and outcome assessment," said Mark Hofmeyer, MD, first author and medical director of advanced heart failure at MedStar Washington Hospital Center in Washington, D.C.

Dilated cardiomyopathy is the most common cause of patients needing a heart transplant and is responsible for up to half of the heart failure cases that result from a weakened left ventricle. Symptoms include shortness of breath with exertion, fatigue, edema of the legs and feet, an irregular heartbeat and lethal arrhythmias.

"This study and others from the DCM Consortium can help improve genetic evaluation and testing guidelines for patients who have DCM and their first-degree relatives, which are children, siblings or parents," said co-author Garrie Haas, MD, professor of clinical medicine and a senior clinician in the Advanced Heart Failure and Cardiac Transplant Program at Ohio State. He reviewed clinical data from all study participants to ensure all DCM diagnoses were applied in a systematic and rigorous manner for the study.

Patrice Desvigne-Nickens, M.D., a medical officer at the National Heart, Lung, and Blood Institute (NHLBI), part of the National Institutes of Health, agreed. She noted that the study also sheds light on racial disparities associated with DCM, as Black patients suffer a disproportionate disease burden compared to white patients.

"African Americans are too often underrepresented in research, especially in cohorts investigating genetics," Nickens said. "This study is uniquely designed to consider a biracial cohort investigating genetic underpinnings of dilated cardiomyopathy. The findings and conclusions strongly underscore the importance of genetics in DCM, predicting

disease severity and risk of family members."

More information: Mark Hofmeyer et al, Rare Variant Genetics and Dilated Cardiomyopathy Severity: The DCM Precision Medicine Study, *Circulation* (2023). DOI: 10.1161/CIRCULATIONAHA.123.064847

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