Study: Informational booklet improves family screening for inherited heart muscle disease

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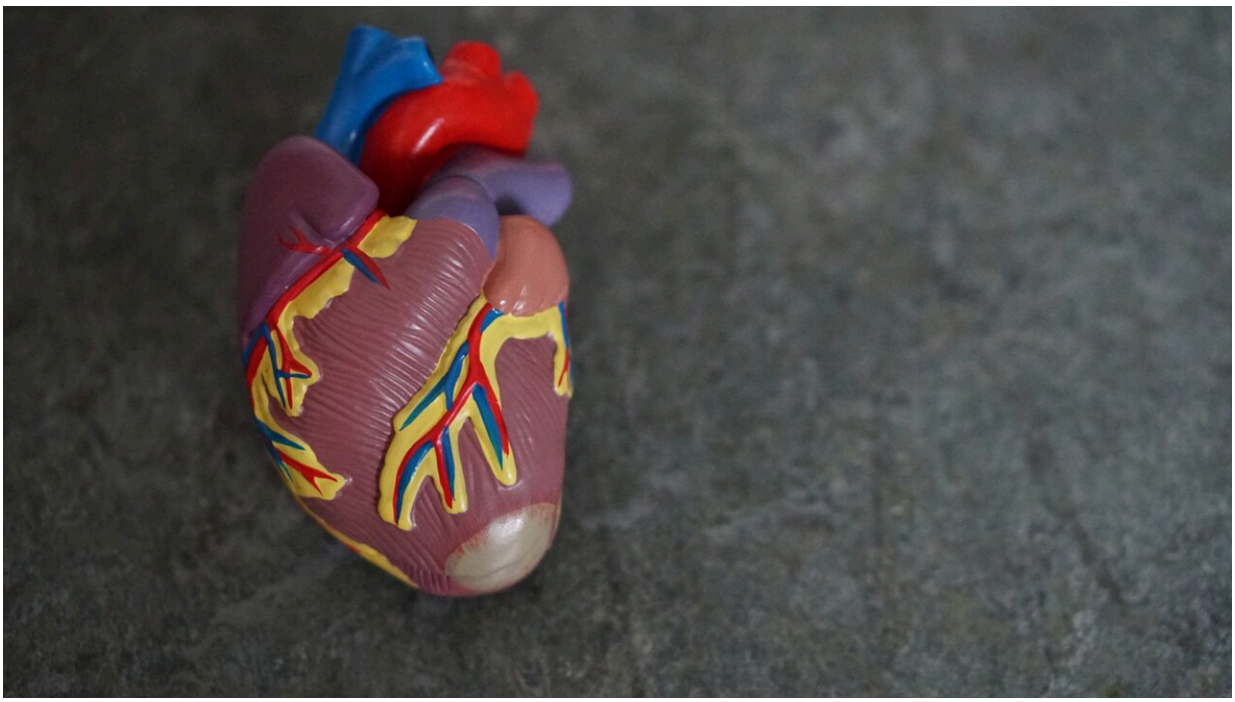

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A booklet developed by cardiovascular and genetics experts and provided to patients with dilated cardiomyopathy (DCM) of unknown cause was effective in increasing screening of relatives at risk for the heart muscle disease, according to a study led by researchers at The Ohio

State University Wexner Medical Center and College of Medicine.

DCM is a condition in which the heart muscle weakens and the left ventricle enlarges. It's 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.

Researchers believe most of idiopathic, or unknown cause, DCM has a genetic basis. Because some patients are asymptomatic for months or years until severe disease like heart failure develops, experts recommend cardiovascular screening of first-degree family members (children, siblings or parents) of patients with DCM. Medical treatment has been shown to mitigate asymptomatic DCM. About 1 in 5 first-degree family members of patients with idiopathic DCM are at risk of getting the condition during their lifetime.

Studies have shown that patients are often ill-equipped to share genetic risk information that motivates at-risk relatives to be screened. Because patients' medical information is confidential, providers can't directly contact family members.

"It's been challenging to find an effective and easy method to educate family members about their risk. Barriers include emotional or geographic distance among relatives, low health literacy, lack of confidence to explain genetics and reluctance to share personal information. This is the only large, randomized study ever conducted of a communication tool developed for DCM patients to help them explain the need for screening to their family members. The booklet that was developed showed promising results," said Dr. Ray Hershberger, a cardiologist and division director of human genetics at the Ohio State Wexner Medical Center and a researcher at the Dorothy M. Davis Heart and Lung Research Institute.

Hershberger is senior author of the study, which was published today in *Circulation*.

The booklet included visuals and simple explanations of the evaluation and care of individuals with DCM as well as how cardiac screening of asymptomatic family members can detect the disease at the earliest possible stage. It also provided guidance on how to talk with family members about DCM risk and included sample emails.

Between June 2016 and March 2020, 1,241 DCM patients were randomized to receive the booklet or not at 25 leading academic U.S. heart failure/heart transplant programs that are part of the DCM Consortium, led by Hershberger. The trial was designed and overseen by investigators and Ohio State co-first authors Daniel Kinnamon and Elizabeth Jordan. Researchers found that DCM patients who received the booklet had a higher percentage of eligible first-degree relatives complete screening (19.5%) than those who did not get the booklet (16%).

"DCM can occur in family members at almost any age but the typical onset is mid 40s," said Jordan, a cardiovascular genetic counselor and associate professor of clinical internal medicine. "The severity of the condition can vary within families, with some family members exhibiting minor or no symptoms. This makes clinical and genetic screening for DCM important because it usually presents with advanced disease such as heart failure or an arrhythmia causing sudden cardiac death."

Other symptoms include shortness of breath with exertion, fatigue, edema of the legs and feet or an irregular heartbeat.

"Managing disease risk among first-degree relatives of DCM patients is central to precision medicine. The "Family Heart Talk' booklet is an

effective tool that is low cost and requires a minimal time investment to implement into clinical care," said Kinnamon, director of Human Genetics Research Informatics and a research assistant professor who planned and oversaw the study's analysis.

Researchers found no evidence that the effect of the booklet differed among first-degree relatives of Black, Hispanic and white DCM patients. In a previous study by the DCM Consortium, researchers found that first-degree relatives of Black patients with DCM are more likely to develop DCM than those of white patients.

More information: Daniel D. Kinnamon et al, Effectiveness of the Family Heart Talk Communication Tool in Improving Family Member Screening for Dilated Cardiomyopathy: Results of a Randomized Trial, *Circulation* (2023). DOI: 10.1161/CIRCULATIONAHA.122.062507. www.ahajournals.org/doi/10.116 … LATIONAHA.122.062507

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