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Genetic testing identifies overlap of heart failure and arrhythmia conditions

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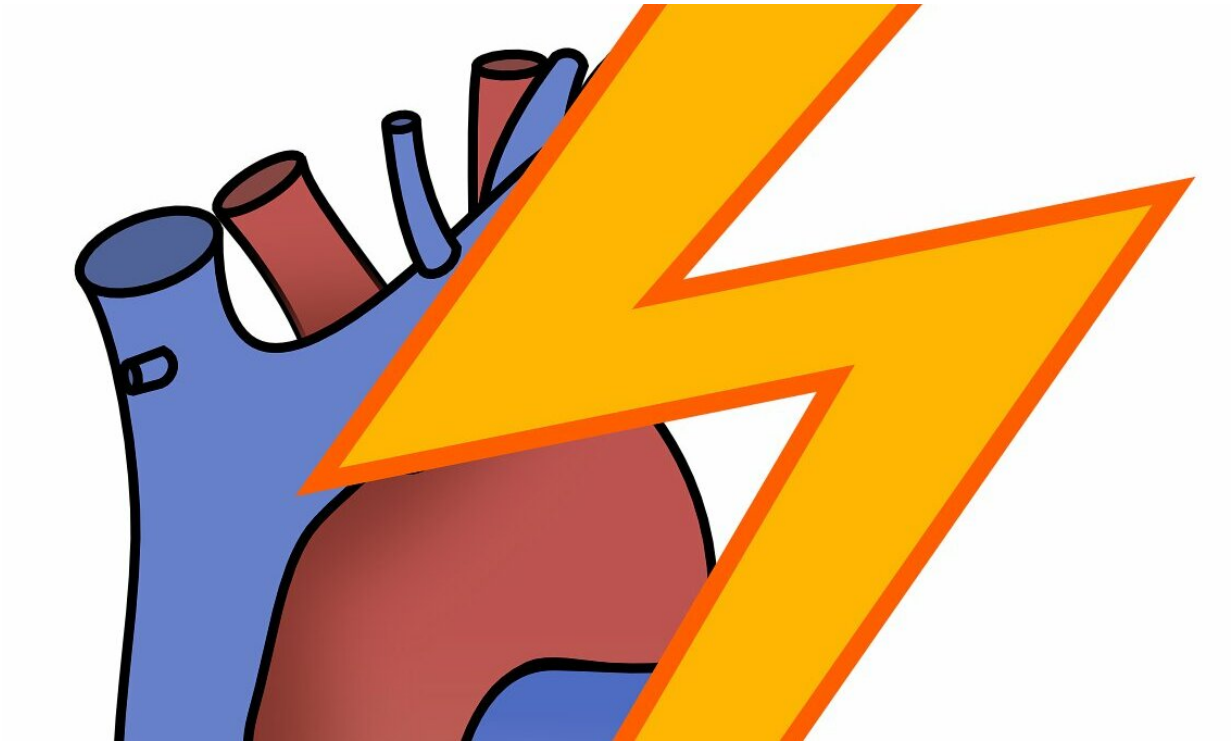

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Heart failure and arrhythmia conditions are often considered as separate disorders, but genetic testing suggests there is much more overlap of these disorders than previously appreciated.

Unfortunately, this potentially life-saving genetic testing is underutilized

in cardiomyopathy patients, reports a new study from Northwestern University Feinberg School of Medicine, Massachusetts General Hospital and Invitae, a medical genetics company. The study found that by screening for variants associated with both cardiomyopathies and arrhythmias simultaneously rather than one condition only, patients' conditions were better detected.

"Diagnosis of these potentially life-threatening conditions holds the key to risk reduction, and genetics plays a big role in identifying who may be at risk and guiding medical care to reduce that risk," said lead study investigator Dr. Elizabeth McNally, director of the Center for Genetic Medicine at Feinberg.

McNally will present the findings Saturday, Nov. 13, at the American Heart Association (AHA) Scientific Sessions.

'Life-threatening in young people'

Heart failure is often caused by cardiomyopathy, and genetic testing is now recommended for cardiomyopathies by cardiac professional societies. Similarly, certain arrhythmia disorders can also have a genetic cause, and genetic testing can identify that cause.

An estimated 1 in 250 people are diagnosed with hypertrophic and dilated cardiomyopathy. Cardiomyopathies can be very serious and lead to compromised lifestyle with activity intolerance and irregular heart rhythms. But the symptoms and disease can be quite variable, with some people having very mild involvement. These disorders can occur in people of all ages, but can also affect young people in which the diagnosis is less likely to be considered since young people are less often thought of as having heart disease, McNally said.

Findings:

The research found that 20% of 4,782 patients had a positive test result. From these positive results, 10.9% of the evaluable patients would have been missed had they only had a condition-specific panel, rather than the combined arrhythmia and myopathy genetic test. Sixty-six percent of those who receive a genetic diagnosis have a result that helps guide their clinical management, such as genes associated with a targeted therapy.

"Many of the genes identified in this testing identify a different disease course with higher risk for atrial fibrillation or heart failure," McNally said. "Some of the genes even have gene-specific therapies available. And there are more drugs in development, so we expect more genes to have specific treatments in the future."

The data from this study stem from the Invitae Detect program, which launched in 2019 to offer sponsored, no-charge genetic testing for conditions in which testing is underutilized and can improve diagnosis and treatment. The study examined data from a one-year interval spanning 2019 to 2020.

Why is genetic testing underutilized?

There are many reasons that contribute to the underuse of genetics, McNally said. Many doctors and nurses have never received any training in how and when to order genetic testing and they may not be confident in interpreting the findings and knowing how to manage the results.

"Cost is much less an issue than it was in the past because costs have come down, and most insurers now cover the cost of genetic testing because this information helps identify risk that can be reduced," McNally said. "Many health care providers may only order the condition-

specific panels because they believe the results may be easier to interpret."

If detected, conditions are manageable, treatable

Many people with cardiomyopathies can be treated with medications and monitoring and treatment for irregular heart rhythms. For the subset of people who have more severe symptoms, there are many advanced options for management, McNally said.

If a patient receives a genetic test result that says they're at risk for arrhythmia, they can monitor their heart rate easily through a skin patch, a device inserted under the skin or even a smartwatch. The devices can detect a life-threatening arrhythmia that could spur early, proactive treatment.

Provided by Northwestern University

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