

Gene tests for heart disease risk have limited benefit

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Credit: Imperial College London

Genetic tests to predict a person's risk of heart disease and heart attack have limited benefit over conventional testing.



This is the finding from scientists at Imperial College London, who devised a highly sophisticated test analysing thousands of so-called genetic variants linked to heart health.

The results of the test, published in the *Journal of the American Medical Association*, revealed only modest improvement over the standard method doctors currently use to measure <u>heart disease</u> and <u>heart attack</u> risk—using factors such as cholesterol, blood pressure, smoking habits and presence of diabetes.

The scientists stressed the study doesn't contradict previous findings that heart disease runs in families, or that a person's genes may place them at higher risk of the condition. Rather, the findings suggest that, at present, genetic data adds only modestly to the information we currently gain using conventional testing. This may change in the future as our knowledge of genes affecting heart disease risk improves.

Dr. Ioanna Tzoulaki, lead author of the study from Imperial's School of Public Health, said: "Genetic tests to predict a person's risk of developing a condition are becoming cheaper and cheaper, and will soon become part of routine patient care.

Therefore we needed to evaluate whether these tests can add information to our existing tools of predicting who is at high risk of developing heart disease. Our research suggests that for heart disease these tests do not add much to the information we can gather from assessing factors such as cholesterol levels and blood pressure."

Heart disease is a leading cause of death worldwide and is responsible for around 64,000 deaths in the UK each year. The condition is caused by narrowing of the blood vessels supplying the heart due to the build-up of fatty substances, which then leads to heart attacks.



At the moment, in the UK when doctors want to assess a person's risk of heart disease, they calculate a score called QRISK. This involves analysing factors such as age, sex, cholesterol levels, blood pressure, diabetes and whether a person smokes, to calculate risk of developing heart disease within the next 10 years. If the risk is calculated as above 10 per cent, a patient is recommended treatment such as statins to lower cholesterol.

The researchers behind the current study wanted to see if analysing a person's genetic information could enhance the predictive power of the QRISK score, and a similar score used in the USA.

The team analysed clinical and genetic information from over 350,000 people included in the UK Biobank study. The individuals had no history of cardiovascular disease, and an average age of 55 years old.

The group were tracked for eight years, during which time any heart disease diagnoses or heart attacks were recorded (6,272 of these events occurred in this time frame).

The team then sifted through the genetic data for small DNA changes called single nucleotide polymorphisms (SNPs). These occur when a single nucleotide (building block of DNA) is replaced with another. These changes may cause disease, and the study team used all known SNPs associated with heart disease risk—over one million in total.

The team found that when the genetic results were combined with a patient's QRISK score, approximately 4% of individuals had a more accurate risk assessment compared to QRISK alone, although for some people the prediction was less accurate.

The researchers explained the genetic information was mainly from individuals with European ancestry aged 40-69, and further analyses are



required to confirm the findings among people of different ages and ethnicities.

Dr. Joshua Elliott, first author of the research explained: "Our study suggests that easy-to-collect information such as age, sex, <u>blood pressure</u> and cholesterol levels are still the most powerful tools we have for peering into the future and predicting your risk of heart <u>disease</u> and <u>heart</u> attack."

More information: Predictive Accuracy of a Polygenic Risk Score Enhanced Prediction Model vs. a Clinical Risk Score for Coronary Artery Disease, *Journal of the American Medical Association* (2020).

Provided by Imperial College London

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