# Impact of misunderstanding genetic tests for heart conditions

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University of Sydney researchers are raising concerns over the need for informed decision making for genetic testing after a study published today finds patients at risk of inherited heart disease do not always understand test results or the impact results will have on their life.

Genetic testing has become increasingly accessible for many diseases including heart conditions such as hypertrophic cardiomyopathy (HCM). HCM is an inherited heart disease that increases the thickness of the heart wall which can lead to serious consequences including sudden cardiac death in young people. First degree relatives (parents, children, siblings) of an affected person have a 50 per cent chance inheriting the same gene mutation.

"With more and more people getting genetic testing for this condition, there is now a growing population of "silent gene carriers" being identified - these are people whose carry the gene but have no physical signs of disease," said researcher Dr Carissa Bonner from the School of Public Health.

"Little is known about how a positive genetic test result for HCM affects newly diagnosed patients who are at risk but may never develop symptoms.

"The issue facing patients is that genetic test results only indicate that they may be at risk of developing hypertrophic cardiomyopathy, not that they already had the disease.

"The question we're asking is: Would they be better off not knowing?"

Dr Bonner said the interpretation of the HCM genetic test results was variable for silent gene carriers and can lead to psychological and behavioural changes.

"There were a number of psychological and behavioural changes made by patients who received a positive result on their genetic test that they didn't necessarily consider when making the decision about whether or not to be tested.

"This included shock and anxiety about the test result, worry about developing symptoms, worry about children, family planning decisions, and restrictions on physical activity, career options and access to travel insurance.

"We found that when considering genetic testing, most people were motivated by helping their children or grandchildren. They wanted to rule out the possibility for the next generation, which meant they weren't really thinking about the impact on their own lives."

She said negative impacts tended to be more common in patients who misinterpreted the genetic testing results and these impacts could be reduced through clearer communication prior to testing.

"Those who believed they were at very high risk of heart problems described depression and anxiety, reducing the intensity of exercise, changing career, seeking medical advice for fertility and heart palpitations, and difficulty getting insurance because they voluntarily disclosed the test result.

"Those impacts appeared to be more likely when the patient misunderstood the positive genetic test to mean they had a current heart

condition, which is not actually the case.

"The findings illustrate the importance of cardiac genetic counselling prior to the test, and the need for more comprehensive communication around the meaning and implications of the test result so patients can be better informed to make life decisions," Dr Bonner said.

The study was published in the *Journal of Genetic Counseling*.

Provided by University of Sydney