

# Critical need for greater understanding into diagnosis of inherited heart disease

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Study author Head of Centenary Institute's Molecular Cardiology Program and leading cardiologist Professor Chris Semsarian. Credit: Centenary Institute

Results of a study carried out by researchers at the Centenary Institute in collaboration with Wisser Healthcare, Royal Prince Alfred Hospital and the University of Sydney, have shown that the use of advanced imaging equipment is driving a significant increase in the diagnosis of a little known inherited heart disease in adults.

Left Ventricular Non-Compaction (LVNC) is a rare form of [heart disease](#), where the walls of the left ventricle (the bottom chamber of the left side of the [heart](#)) do not form properly, with channels forming in the [heart muscle](#) leading to a "spongy appearance". LVNC can be accurately diagnosed in babies and young children, and is very severe in this population group, often requiring a heart transplant to save the child. In adults, approximately two-thirds of individuals with LVNC diagnosed using traditional medical tests will develop heart failure, making early and accurate diagnosis vital.

However, this latest study has highlighted that in adults, the rate and accuracy of diagnosis is being heavily influenced by the introduction of new testing methodologies.

In a comprehensive systematic review of 55 studies of the "disease", the authors of a new study, published today in the highest ranking international cardiovascular journal - *European Heart Journal*, have found that an older heart test, echocardiography (which uses ultrasound) diagnosed the condition in 1% of hospital patients and healthy adults. But using a new cardiac MRI test, 15% of adults were reported to have the condition. In some studies, it was much higher, up to 40%, even in studies of large samples of well people.

Professor Chris Semsarian AM, Head of Centenary's Molecular Cardiology Program and a leading cardiologist at Royal Prince Alfred Hospital, says, "Finding evidence of heart disease in up to 40% of well adults raises significant questions. Many of the studies were done on well

people with no symptoms or signs of heart disease, yet the cardiac MRI showed these abnormal results at alarming levels. This raises questions about the clinical utility of this particular diagnosis. It raises wider concerns too. For example, how should results from new, very sensitive tests be interpreted?"

The study highlights the impact of new technology on what appears to be massive over-diagnosis of the disease. It also provides new insights and guidance on more consistent and accurate [diagnosis](#) of LVNC in [adults](#), to enable a reduction in the number of healthy people undergoing the stress of misdiagnosis, and cost and trauma of unnecessary treatment.

**More information:** *European Heart Journal* (2019). [DOI: 10.1093/eurheartj/ehz317/5506056](#)

Provided by Centenary Institute

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