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

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.

Review the full study in the European Heart Journal here

<https://academic.oup.com/eurheartj/advance-article/doi/10.1093/eurheartj/ehz317/5506056?guestAccessKey=54510716-6e62-48e4-954e-f38538dd212f>.

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**To arrange an interview with the Authors, please contact**  
Karen McBrien, Centenary Institute on 0408 601 836.

For more information about Centenary Institute, and the Agnes Ginges Centre for Molecular Cardiology, visit [www.centenary.org.au](http://www.centenary.org.au)

## **About the Centenary Institute**

The Centenary Institute is a world-leading independent medical research institute, closely affiliated to the University of Sydney and the Royal Prince Alfred Hospital. Our research focuses on three key areas: cancer, inflammation and cardiovascular disease. Our strength lays in uncovering disease mechanisms and applying this knowledge to improve diagnostics and treatments for patients.

## **About The University of Sydney**

The University of Sydney is Australia's first university, founded in 1850 on the principle of providing higher education for all. It was one of the first universities in the world to admit students on academic merit, and women on the same basis as men. Today the University of Sydney is ranked in the world's top 50 universities, reflecting their reputation for leadership in research and education. The University aims to create a place where the best researchers and academics and most promising students – whatever their background – can achieve their full potential.

## **About Royal Prince Alfred Hospital**

RPA, co-located with the University of Sydney, is an internationally and nationally respected principal referral hospital which provides a broad range of clinical services including highly specialised services. RPA has a leadership role in the state in complex clinical service development and the rapid translation of research and innovation into patient care. RPA's specialty services are numerous and include the Australian Liver Transplant service, kidney transplantation, cardiovascular and cardiothoracic services, genomics, neurosciences, and neuro-intervention including endovascular clot retrieval, molecular imaging, haematology, cancer care, highly complex maximally invasive surgery including advanced GI surgeries such as pelvic exenteration and peritonectomy, intensive care, neonatal care, maternity and gynaecology.

## **About Wisser Healthcare**

Wisser Healthcare is a research collaboration involving The University of Sydney, Bond University, Monash University, the University of Wollongong, and international colleagues. The research group investigates the cause and extent of the problem of overdiagnosis and overtreatment. It is testing solutions in the areas of cancer, cardiovascular disease and musculoskeletal disorders, with a particular focus on overdiagnosis caused by imaging (like CT scans and MRI scans), testing biomarkers (for example, blood tests like the Prostate Specific Antigen test), and genetic tests.