A game-changing discovery in diagnosing genetic heart disease

Scientists at the Centenary Institute and Royal Prince Alfred Hospital have used state-of-the-art technology to significantly improve the diagnosis rate of a potentially deadly heart condition by up to 20 per cent.

Hypertrophic cardiomyopathy is a common genetic heart condition, which can affect both men and women at any age. It occurs when the heart muscle thickens - making it difficult for the heart to pump blood, and in some cases, results in sudden cardiac death.

Funded by NSW Health, Centenary scientists have used *whole genome sequencing* to explore hypertrophic cardiomyopathy in 58 Australian families, who are directly affected by the disease. *Whole genome sequencing* allowed the scientists to look at regions of patients' genes which were previously not considered important.

The study, published in world-leading scientific journal *The Journal of the American College of Cardiology*, found changes in *introns* (previously referred to as "junk DNA") resulted in changes to the heart muscle, and could lead to disease. The scientists also discovered how changes to *mitochondria* (the organelles which generate energy for the cell) generated similar changes.

Ultimately, the study showed for the first time how *whole genome sequencing* can boost the diagnostic pick-up rate in people with cardiomyopathy by up to 20 per cent.

Senior Researcher for Centenary Institute's Molecular Cardiology Program and the University of Sydney, Dr Richard Bagnall is lead author of the paper, and believes it's a game-changer in the field.

"This study shows we can use this incredible technology to diagnose hypertrophic cardiomyopathy in 2-out-of-10 more families. But there is a lot more information within those whole genomes we haven't even had a look at yet, so this is really just scraping the surface of what's possible," Dr Bagnall said.

Head of Centenary's Molecular Cardiology Program, Royal Prince Alfred Hospital cardiologist and University of Sydney Professor Chris Semsarian AM, has led the study, which he describes as a much more robust approach to genetic diagnosis in cardiomyopathy.

"This is precision medicine – finding the exact genetic change which is leading to disease in individual patients. The next step is to then link those genetic changes with the clinical features of these patients," Professor Semsarian said

"While this study shows how we can use whole genome sequencing in diagnosis, the technique is also crucial for guiding more effective and targeted therapies in the future."

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