MEDIA RELEASE

23rd January 2019

Building the case for a closer look at known heart-disease genes

Centenary Institute scientists have conducted a study which could change how researchers discover the causes of genetic heart disease.

At the moment, the bulk of genetic testing focuses on the **protein-coding** sections of DNA to look for disease-causing variants. However, these protein-coding regions only make up about two-per-cent of our entire DNA sequence.

In a study published in scientific journal *Circulation: Genomic and Precision Medicine*, researchers in Centenary's Molecular Cardiology Program screened 500 families affected by hypertrophic cardiomyopathy – a common genetic heart condition which occurs when the heart muscle thickens, making it difficult to pump blood.

The researchers focused on one of the main disease-causing genes, known as **MYBPC3**, and discovered they were able to attribute the cause of hypertrophic cardiomyopathy in four families to a variant found in the **non-coding region** of the DNA.

First-time Lead Author Emma Singer says while on the surface, it may appear to be a small breakthrough, it's still important for patients affected by genetic heart disease.

"This study makes a major difference for those four families who otherwise would not have known the cause of their heart condition, which in some cases, can be fatal," says Emma.

Senior Researcher Dr Richard Bagnall is hopeful the study will help re-direct the broader focus of genetic heart disease research.

"We would consider this a pilot study, so we are hoping our results will encourage other researchers to undertake a similar approach in larger cohorts of patients with other known disease-causing genes.

"This study demonstrates why we need to be looking at the known genes more closely and more carefully – because we're finding that we're having a lot more success that way, rather than trying to find a new gene altogether that causes disease."

Key value of RNA analysis of MYBPC3 splice site variants in hypertrophic cardiomyopathy has been published in the scientific journal Circulation: Genomic and Precision Medicine.

To arrange an interview, please contact: Centenary Institute Media and Communications Manager, Laura Parr, l.parr@centenary.org.au, 0435 530 537

For more information about Centenary Institute, visit www.centenary.org.au