MEDIA RELEASE

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New insights into why young people die suddenly

A study led by scientists at the Centenary Institute could provide some families with a clearer insight into why a young loved one may have experienced an otherwise unexplained cardiac arrest and in some cases, sudden death.

Arrhythmic right ventricular cardiomyopathy (ARVC) is a poorly understood genetic cardiomyopathy. It occurs if the muscle tissue in the right side of the heart undergoes cell death and is replaced with fat and scar tissue; disrupting the heart's electrical signals and causing an abnormal heart rhythm.

Researchers from the Centenary Institute, Royal Prince Alfred Hospital, Royal North Shore Hospital and the University of Sydney examined four families in which one family member (under the age of 30 years) had ARVC and experienced a cardiac arrest as a result. In two cases, the patients survived, while in the other two cases, the patients died suddenly.

Studies to date suggest patients must exhibit an obvious structural change to their heart before they are at risk of a sudden cardiac episode. This study shows for the first time that this isn't always the case, with none of the four patients' hearts exhibiting any structural changes. In all four cases however, a genetic error in a particular gene called PKP2 was identified.

Head of Centenary's Molecular Cardiology Program, Royal Prince Alfred Hospital cardiologist and University of Sydney Professor Chris Semsarian AM says the findings have several important ramifications.

"This study could force us to re-evaluate how we treat patients with this particular type of cardiomyopathy. For example, should we be more aggressive in treating these patients who carry the PKP2 gene error, but don't show any clinical evidence of having this disease?" says Professor Semsarian.

"It also opens the door for potential gene-targeted therapy further down the track, with the aim of preventing patients with ARVC from experiencing a cardiac arrest."

Lead author and Head of Centenary's Clinical Cardiac Genetics Group within the Molecular Cardiology Program, Dr Jodie Ingles says the results may also help to provide closure for some families.

"In many cases of cardiac arrest or sudden death, it can be difficult to identify the precise cause, particularly if the patient's heart does not show any structural changes. Now, we can attribute a specific gene as to why some people die suddenly, in cases that would otherwise go unexplained," says Dr Ingles.

Concealed arrhythmogenic right ventricular cardiomyopathy in sudden unexplained cardiac death events has been published in 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

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