MEDIA RELEASE

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Concealed cardiomyopathies revealed in cardiac arrest survivors

Centenary Institute researchers have discovered that genetic testing can identify 'concealed cardiomyopathies' in nearly a quarter of sudden cardiac arrest (SCA) survivors who seem to have a normal heart.

The findings will mean improved diagnosis rates and personalised care for SCA survivors as well as guide the screening of family members who may have the same underlying genetic condition.

The study, reported in the 'International Journal of Cardiology', undertook genetic testing and analysis of clinically-idiopathic SCA survivors (individuals where previous clinical investigations had failed to reveal a diagnosis).

The researchers identified a genetic cause of arrest in 22% of the SCA survivors studied. The majority of these newly identified cases had genetic abnormalities associated with cardiomyopathy.

"Cardiomyopathies are diseases of heart muscle. They can impair the heart's ability to pump blood around the body, leading to heart failure but can also cause electrical changes which can lead to dangerous heart rhythms," said lead author of the study, Dr Julia Isbister from the Centenary Institute's Agnes Ginges Centre for Molecular Cardiology.

"These conditions are usually detected on clinical tests such as ultrasound but our findings show that state-of-the-art genetic testing may be useful in revealing cardiomyopathy before structural abnormalities are evident."

Dr Isbister says that identifying the reason for a SCA is critical for both patients and their families.

"If the specific disease can be diagnosed we are better able to implement personalised care for the survivor. If we discover that the SCA is genetically-based we can then screen family members for similar issues. Screening of first-degree relatives is an extremely important aspect of caring for SCA families, aiming to reduce the risk of further cardiac arrests in the family," said Dr Isbister.

Professor Christopher Semsarian AM, Head of the Centenary Institute's Agnes Ginges Centre for Molecular Cardiology and senior author on the study says that the role of genetic testing in the management of SCA survivors requires reappraisal given the results of the team's findings.

"Current guidelines recommend only limited genetic testing of SCA survivors when a specific genetic condition is already suspected. Genetic testing is not generally recommended for those survivors classified as clinically 'unknown'," he said.

“Our study has shown that advances in genetic testing technology and analysis can improve diagnosis rates by revealing heart defects that were previously hidden. A reassessment of guidelines and increased genetic testing may lead to better outcomes for survivors, their families and overall prevention of sudden cardiac death in the young.”

[ENDS]

Publication:
“Concealed cardiomyopathy” as a cause of previously unexplained sudden cardiac arrest.
https://www.sciencedirect.com/science/article/pii/S0167527320338122

Image:
Dr Isbister and Professor Semsarian:
https://drive.google.com/file/d/1eG5nLsl1ad7jPVYgk4tXW84pTWq6FnSK/view?usp=sharing

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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 lies in uncovering disease mechanisms and applying this knowledge to improve diagnostics and treatments for patients.

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