A game-changing discovery for Centenary's Molecular Cardiology Program is literally saving lives.

In Australia, up to 200 young people under the age of 35 die from sudden cardiac arrest each year.

The commitment of Professor Chris Semsarian AM and his team in our Molecular Cardiology Program to solving the mysteries behind sudden cardiac death in the young, as well as other cardiovascular disorders caused by underlying genetic abnormalities, has resulted in their most recent ground-breaking discovery.

In Australia, up to 200 young people under the age of 35 die from sudden cardiac arrest each year. That's up to four young lives lost, without warning, each week. And for every young person who dies, there are potentially many more family members at risk.

Hypertrophic cardiomyopathy (HCM) is a common genetic heart disease, which affects an estimated one in 500 Australians. Characterised by a thickening of the heart muscle, it is the most common cause of sudden cardiac death in those under the age of 35.

Professor Semsarian and his team recently uncovered a game changing breakthrough in the diagnosis of genetic heart disease. Their study, which was published in the world-leading scientific journal, the Journal of the American College of Cardiology, showed how whole genome sequencing can boost the diagnostic pick up rate in people with HCM by up to 20 per cent, by allowing scientists to look at regions of patients' genes which were previously overlooked.

The study found changes in introns (formerly referred to as "junk DNA") resulted in changes to the heart muscle, and could lead to disease. The team also discovered how changes to mitochondria (the organelles which generate energy for the cell) produced similar changes.

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

"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," says Professor Semsarian.

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Losing a loved one is heartbreaking and traumatic, but losing a young, healthy loved one, without warning is devastating for the families and friends left behind. In some cases, these young people suddenly drop dead during or soon after exercise; in other cases, he or she will fall asleep and never wake up. It is difficult to even start to understand the terrible impact this must have on surviving family members.

Professor Semsarian tells us that the most difficult part of his job as a Cardiologist is "explaining to the parents of a sudden cardiac death victim that their seemingly healthy, normal child has dropped dead from heart disease".

Every day, the team in our Molecular Cardiology Program continue their important work to discover what triggers the heart to malfunction in people with these abnormal genes.

Direct relatives of people with an inherited heart disease such as a child, brother, sister or parent, have a 50 per cent chance of inheriting the same genetic condition. The work of Professor Semsarian and his team is guiding greater diagnosis of these conditions, but much more is yet to be done. "We really are just scraping the surface of what's possible" says Dr Bagnall.

Michael was fit, healthy and just 35 years old. One evening, he went out to play sport with friends and never came home.

When Michael and Emily found out they were expecting their first child, they were overjoyed.

Just a few weeks later, Michael went out to play volleyball. It was the day before Emily's first scan, when he would have heard his baby's heartbeat for the first time.

"One of Michael's teammates called to say he had collapsed during the game and an ambulance had been called," Emily told us. "I was hysterical, but it didn't even cross my mind that it could be fatal."

By the time Emily arrived at the hospital, a police officer was waiting for her with the devastating news that Michael had died on the playing field. At just 35 he'd had a fatal cardiac arrest.

Research by Centenary's Molecular Cardiology Program has enabled the identification of the genes responsible for Michael's death – information that can be used in future to determine if his children are at risk.

Knowing this has given Emily some peace of mind. Michael and Emily's twin girls who were born on Michael's birthday and whom he never had the chance to meet, have a 50 per cent chance of inheriting Michael's condition.

Up to four Australians under 35 die EACH WEEK from sudden cardiac arrest.

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