When an elite athlete drops dead on the sporting field, the community wonders why a young, seemingly healthy person can die so suddenly. Sudden cardiac death is a tragic and devastating complication of a number of cardiovascular diseases. The death is most often unexpected and has major implications for the surviving family and the community. In fact the first presentation of an underlying cardiac disease in up to 50% of people is cardiac death. In children and young adults, genetic disorders, such as inherited heart muscle and electrical rhythm diseases are a major cause. In those older than 35 years, coronary artery disease and myocardial infarction together comprise over 90% of sudden cardiac death cases.

Understanding the causes and arrhythmogenic substrates underpinning sudden cardiac death and elucidating what may trigger malignant arrhythmias is crucial for the development of treatment and prevention strategies in our communities.

Most recently, significant advances in genetic technologies have impacted on the number of cardiac genes that can be interrogated in the setting of families who have experienced sudden cardiac death. While the original molecular autopsy tested only 4-5 genes and took 6-12 months to perform, we can now use amazing new genetic technologies that can test all 22,000 genes that we have, to look for the genetic causes of SCD in the young. This next generation sequencing technology has led to whole exome and whole genome sequencing which can be performed in less than 6 weeks. Most recently, our group reported the first series of young sudden death victims where this latest whole exome sequencing approach was used. This evolves as a crucial factor in both establishing an underlying cause of death and in screening at-risk family relatives in the hope that we can prevent tragic and unnecessary deaths. In cases where no definitive cause is identified at postmortem, i.e. sudden unexpected or unexplained death, the “molecular autopsy” has emerged as a key process in the investigation of the cause of death in young victims.

This involves obtaining a blood sample at postmortem, extracting DNA from the sample, and then performing genetic testing for key cardiac genes to identify a cause of death. The molecular autopsy, in conjunction with clinical screening of relatives of the deceased has two major implications. First, the testing can reveal the cause of death in the victim, and second, provides an additional diagnostic tool in identifying other family members who may be at risk of SCD. The combination of clinical and genetic evaluation of families provides a life-saving platform for early initiation of therapeutic and prevention strategies, such as lifestyle changes, medications, and therapy with an implantable cardioverter defibrillator (ICD).

Major advances have been made in our understanding of the causes of sudden cardiac death in the young and in the identification of at-risk family members. However this focus should be part of a broader initiative to prevent sudden cardiac death in the community. Simply, education programs need to be developed to raise community awareness about the importance of a family history of sudden death or heart disease in a young relative; understanding of the important clinical symptoms such as exercise-induced dizziness or an unexplained collapse; and a greater education about the importance of cardiopulmonary resuscitation (CPR) skills. Public access defibrillators need to be more widely distributed, in all major public schools including sporting clubs and schools. It is only through such a multifaceted public health approach that we have the best chance a successfully preventing SCD amongst the young in our communities.

References:

Chris Semsarian is Professor of Medicine in Sydney Medical School, cardiologist at RPAH, NHMRC Practitioner Fellow, and leads the molecular cardiology research program at Centenary Institute.
GETTING THE COMMUNITY ON BOARD

Professor Chris Semsarian is on a mission. He believes defibrillators should be made available in every NSW public school. He also wants mandatory CPR training to be delivered to all.

Up to four Australians under the age of 35 die every week of sudden cardiac death. Professor Semsarian says targeting the community in terms of training is key if we are to prevent sudden cardiac death in the young.

“All the evidence suggests that when someone has a cardiac arrest, I could do as much as I could with CPR and try to resuscitate you, but if you’ve had a true cardiac arrest the only thing that will actually save you is a shock by the defibrillator and every minute that passes the survival drops by about 10 to 15 per cent.”

Without early CPR and defibrillation, after four minutes brain damage is caused and after eight minutes there is little chance of survival.

Professor Semsarian says workplace and some health regulations are preventing the defibrillators being placed in public schools. However he won’t be deterred. “The only way we are going to do this is by legislation change. My understanding is there is this fear that if a person who has never used a defibrillator in a school setting uses one they are going to cause some sort of harm or damage.”

Professor Semsarian says this simply isn’t the case and they’re virtually foolproof. He’s dedicated to his campaign for change, appearing in the media and online to help raise awareness. His latest efforts include a YouTube video, which has been seen by nearly 4000 people, and a change.org petition which already has over 53,000 signatures.

Here, Professor Semsarian reflects on how his research program at the Centenary Institute and Royal Prince Alfred Hospital has identified new genes involved in hypertrophic cardiomyopathy, and in turn provided improved diagnosis, therapies to prevent complications, and better risk management to prevent heart failure and sudden death in patients with genetic cardiovascular disease.

Thea Manning

Simply, education programs need to be developed to raise community awareness about the importance of a family history of sudden death or heart disease in a young relative...