

The Agnes Ginges Centre for Molecular Cardiology

Sudden Cardiac Death Information Sheet

Sudden Death

Sudden death in a young person is a devastating event for any family. Unfortunately, in many cases this can be the first presentation of an underlying heart condition in an otherwise healthy person. A range of inherited (genetic) heart conditions can cause sudden death and therefore it is important to investigate family members following such an event. There are two main types of inherited heart conditions, i.e. those causing *electrical* or *structural* problems in the heart. Electrical (arrhythmogenic) problems include long QT and Brugada syndromes, while structural problems, include hypertrophic cardiomyopathy and arrhythmogenic right ventricular dysplasia.

When a person dies suddenly, a post-mortem is often performed to determine why the particular individual died, i.e. the cause of death. Some heart conditions are easily diagnosed at post-mortem. In particular, structural causes of sudden death can be determined at post-mortem since the heart may be thickened, enlarged or structures within the heart, such as blood vessels and valves, may be abnormal. Unfortunately, the heart often appears “normal” in those who died because of electrical problems in the heart, such as in long QT syndrome. The abnormalities in the heart are often at the cellular level and cannot be seen at post-mortem. These post-mortems, where no abnormality is found in the heart or other structures in the body, are often called “unascertained”, meaning no direct cause of death was identified.

The Family

As mentioned, many inherited heart conditions can lead to sudden death, particularly in the young. The majority of these conditions are termed *autosomal dominant* meaning children of an affected person have a 1 in 2 (or 50% chance) of inheriting the gene mutation. Males and females are affected equally. In addition, most inherited heart conditions show wide variability in symptoms amongst family members (i.e. one person in a family may have severe disease, while another from the same family may only have very mild symptoms).

Due to the risk to family members, we advise all immediate relatives of someone who has died suddenly to be assessed by a cardiologist. A visit to the cardiologist must include a clinical history being taken, physical examination, electrocardiogram and an echocardiogram. Further tests may need to be performed including electrophysiological testing, exercise studies and MRI scans of the heart.

Genetic Testing

Genetic testing is the process of screening genes for a particular DNA alteration (mutation) that is the cause of disease. It is likened to searching through a whole bookshelf full of books to find one spelling mistake, and as a result is a time consuming and costly process. In addition, as many different genes have been identified to cause different types of inherited heart diseases, it is important to try and determine, on a case by case basis, which genes should be looked at to narrow down the search and more importantly, improve the chances of identifying exactly which gene is defective. Information from the post mortem can assist in working out what genes are more likely to carry a mutation.

At present, genetic testing for the more common inherited heart diseases is now available commercially through private testing centres. This means that a family would need to pay for such a service (likely to be in the order of a few thousand dollars). It should be taken in to consideration that genetic testing of stored tissue samples from a deceased family member can be very difficult as the DNA quality is very poor, particularly if it has been stored for many years. This may mean that

there is a lower chance of identifying a gene mutation. Alternatively, if a living family member is identified to have the heart condition, initial gene screening can be carried out on that person's blood sample.

Before deciding to undergo genetic testing, it is important to discuss as a family what you would do if a gene mutation were identified. For example, if a laboratory identified the causative mutation would other family members want to have a predictive genetic test to determine if they too carry the mutation? It is important to know that there are no cures for inherited heart conditions, however there are ways of managing the symptoms and reducing the likelihood of sudden death.

Sudden Death Research at the Agnes Ginges Centre for Molecular Cardiology

The Agnes Ginges Centre for Molecular Cardiology carries out research to look at the causes and prevention of sudden death. In addition to the laboratory work, we collect information about families where a sudden death has occurred. This includes the post mortem report, any pre-death medical records and information about family members such as a detailed family history and notes from visits to the cardiologist.

If you would like to make an appointment to see Professor Chris Semsarian, *Hypertrophic Cardiomyopathy* and *Genetic Heart Disease* clinics are run on alternate weeks at the Sydney Heart Centre, RPA Hospital Medical Centre. All cardiac evaluation and investigations can be performed on family members and the genetic implications of the heart disease in the family, including genetic counselling, are fully discussed and explained.

The Agnes Ginges Centre for Molecular Cardiology carries out important research investigating the causes and prevention of sudden death in the young. Any donations towards this research would be gratefully received. Please contact Professor Christopher Semsarian or Ms Laura Yeates if you would like to know more.

Contacts:

If you or any family members have any questions, please call:

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Sudden Arrhythmic Death Syndromes (SADS) Foundation

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The Cardiomyopathy Association of Australia (CMAA)

For details of your nearest contact person, please contact: 1300 552 622
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