British Heart Foundation

Factfile 04/2002

SUDDEN CARDIAC DEATH IN YOUNG PEOPLE

Background
Surveys of sudden death in England suggest that 3,500 unexpected sudden deaths occur per annum in the 16-64 age group. Of these the majority are due to coronary artery disease but in 4.1% a coroner's post-mortem is unable to find evidence of structural cardiac disease or significant drug exposure. The cause of death is therefore unascertainable. These cases have been referred to as sudden adult death syndrome or sudden unexpected death syndrome. More recently the term sudden arrhythmic death syndrome (SADS) has been suggested because those affected range in age from 4-65 years.

Aetiology
The underlying causes of sudden death in individuals with morphologically normal hearts include the cardiac ion channelopathies:

- Long QT Syndrome (LQTS) - a disorder of cardiac repolarisation causing QT prolongation on the ECG with a risk of ventricular arrhythmias (Torsade de Pointes)
- Brugada Syndrome (BS) - a disorder of cardiac activation causing right bundle branch block and ST elevation in leads V1-3 on the ECG with a risk of polymorphic ventricular tachycardia
- Progressive Cardiac Conduction Disease - a disorder of cardiac conduction causing variable degrees of heart block predisposing to cardiac arrest
- Catecholaminergic Polymorphic Ventricular tachycardia - a disorder of cellular calcium handling causing exertional ventricular arrhythmias (typically bi-directional)

These are potentially inheritable conditions and, as such, the relatives of the victim may also be at risk of dying suddenly from a treatable condition. Sometimes the underlying diagnosis may only be identified by detecting abnormalities in relatives. This necessitates cardiological evaluation of family members.

Other rare genetic conditions that cause structural cardiac disease, which have been identified in relatives of SADS victims even though post-mortems have proved negative or equivocal include:

- Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)
- Hypertrophic Cardiomyopathy (HOCM)
- Dilated Cardiomyopathy
- Dystrophica Myotonica
- Wolff-Parkinson-White Syndrome (WPW)
- Mitral Valve Prolapse

This highlights the diagnostic limitations of standard autopsies when potentially diagnostic features are not obvious. Consequently, coroners' pathologists often refer hearts and/or slides for an expert cardiac assessment.

Family Evaluation
This consists of two parts. First, the background history of the proband (victim of sudden death) should be elicited including preceding symptoms, previous medical history and circumstances of death. Second, the family history must be established including any histories of unexplained syncope, sudden death, muscle weakness or congenital deafness. Ideally each first-degree relative should have their personal history taken and undergo an examination. The following investigations are often undertaken:

- 12 lead ECG (with signal averaging)
- Echocardiogram
- Holter monitor (24 hours or longer)
- 12 lead ECG (with signal averaging)
- Cardio-pulmonary exercise ECG test

Depending on these results, further specialist tests are undertaken relating to ARVC, Brugada Syndrome and WPW.

- Cardiac Magnetic Resonance Imaging
- Ajmaline provocation testing
- Adenosine provocation testing

Genetic Testing
Genotyping of families with LQTS, BS and HOCM is possible allowing confirmation of diagnosis, clarification of carrier status and even guidance of therapy. The limited knowledge of the full genetics of these disorders makes a negative result unhelpful.

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Factfile *is produced by the British Heart Foundation in association with the British Cardiac Society and is compiled with the advice of a wide spectrum of doctors, including general practitioners. It reflects a consensus of opinion.*

Further Management
It is reasonable to review those families without a diagnosis in order to detect conditions that have labile ECG appearances (for example LQTS and Brugada Syndrome). If a diagnosis has been made then advice on testing of other relatives is appropriate. Further investigation may be necessary (for example electrophysiological study in Brugada Syndrome) and appropriate treatments made available (for example betablockers and/or implantable cardioverter defibrillators [Factfile 5/99] in LQTS). There is evidence that such interventions in these conditions can prevent sudden death. Regular follow-up will then be required in affected individuals.

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