



Dr Mary N Sheppard

Sudden cardiac death in the young and helping bereaved families

Dr Mary Sheppard has been working as a specialist cardiac pathologist for the past 12 years at the Royal Brompton Hospital. Many people think cardiac pathology is boring, lacking the diagnostic challenges of surgical pathology. Even colleagues have been known to state it is “easy”, based upon ischaemic heart disease where coronary arteries and diseased muscle is all that is needed to be looked at with the naked eye. However, there is also a very personal and moving aspect to the job, which Dr Sheppard shares with us here.

I initially thought that the cardiac side of my work would not take up a lot of my time in a busy cardiothoracic centre where pulmonary surgical cases predominated. How wrong I was. My mentor and teacher was Michael J Davies, who sadly died two years ago. He had a special interest in sudden cardiac death and published extensively on this topic. Working very much as his junior colleague for over four years on two British Heart Foundation (BHF) research projects into sudden cardiac death, with an emphasis on young people where non ischaemic cardiac causes predominate, gave me invaluable experience in cardiac pathology. Through his gift for teaching, he awakened in me what is now my lifetime interest in this topic, which has happily coincided with public and national interest. We published what are now seminal papers on this topic in the UK^{1,2} and established the entity of sudden arrhythmic death syndrome (SADS).³ Our studies also emphasised that many causes of sudden cardiac death in young people are due to cardiomyopathies or channelopathies which are inherited diseases and thereby evolved the concept of expert cardiological family screening to prevent further deaths.⁴

As a result of my work, I came into contact with families who had suffered a traumatic and totally

unexpected death of a child or young partner. The effect on the family, as you can imagine, is total and utter desolation followed by bewilderment and all the stages of bereavement from depression through anger but rarely to resolution.

“Why did my healthy partner/child die?” was a question often put to me by a person, who even by their tone of voice on the phone in the first few stumbling words, I knew was a bereaved human being. These families had to negotiate the bewildering and often slow and archaic coronial system in this country with variable results from the autopsy and subsequent inquest, waiting from months (if they were lucky) to years. From examining post-mortem reports in our initial study,¹ I became aware of the variable quality of autopsies and in particular the way the cardiovascular system was examined. This variable quality in coronial autopsies has since been reiterated by the National Confidential Enquiry into Patient Outcome and Death (NCEPOD) study, which emphasised that sudden cardiac death cases are poorly investigated, especially when the question of cardiomyopathy or cardiac hypertrophy arises.⁵ This is despite published College guidelines and reviews for pathologists investigating sudden death.^{6,7}

The discovery by families that death was “uncertain” or that there was a lack of detail in the autopsy, with little or no tissue being taken, was in many cases utterly devastating. This became a recurring theme with many of the families who contacted me through the BHF research work we published. They usually discovered my name while surfing the internet for answers to their many unanswered questions. Pathologists throughout the UK, as a result of the research, began sending me cases and I had by then built up a database of 700 cases over the past decade which enabled me to develop specific expertise in the diagnosis of the causes of sudden young deaths. Through this work we now know that many of the causes of death in young people are due to inherited conditions such as the cardiomyopathies and channelopathies.



Because of my work I was invited onto the government implementation group responsible for formulating the National Service Framework Chapter 8 document on cardiac arrhythmias and sudden death which we published in 2005.⁸ The coroner on the group, Michael Burgess, and I provided an addendum on the approach to the autopsy which was published on the Department of Health (DH) website. At the working group I became aware that pathology was considered a minor part of the proceedings by the clinicians and managers within the DH who, during discussions, concentrated on family screening and the establishment of inherited cardiac disease and arrhythmia clinics throughout the country. Yet clinicians admit they need a specific diagnosis when somebody dies suddenly, in order to put the family along a specific screening programme and possible genetic investigations.

Working for this group I first met the family groups who had spearheaded this initiative, usually motivated by personal tragedy. SADS (UK) (www.sadsuk.org), The Cardiomyopathy Association (www.cardiomyopathy.org) and Cardiac Risk in the Young (CRY) (www.c-r-y.org.uk/index.htm) were the family groups represented within the implementation group. I met Alison Cox, the chairperson of CRY, who recognised from personal experience with bereaved families the importance of autopsy and pathology. Alison had heard of me through the bereaved families and asked me to talk at their bereavement meetings held annually. Here I was asked to explain my role and emphasise the importance of retention of the heart and tissue in order to come to a specific diagnosis. With careful, sensitive and subtle explanation of exactly what we do in the autopsy, the families can understand the need for retention and what histology, blocks

and slides are. I had to overcome many people's perception of histopathologists as necrophilic, psychopathic, homophobic, desiccated academics surrounded by grisly bits within our own enclosed world (in other words mad, bad and dangerous to know!). Meeting the families and explaining my role to them has opened my eyes to the importance of communication within pathology and with the public. We need to go out there and emphasise the vital role we play in the investigation of sudden cardiac death and other health issues. The bereaved families are looking for answers and need to know more about the conditions that led to the death of their beloved. This role is an important one for us, working in association with the cardiologists who screen the families.

The work I was doing was not funded and carried out by me on a purely voluntary basis. Since then I have obtained funding from CRY to establish a cardiac pathology unit dealing specifically with sudden young cardiac death which will officially open on 7 March 2008. In addition a national network of histopathologists dealing with cases is being developed which has been already highlighted in the *Bulletin* and there is now funding for a national database to be established. These are exciting new developments which will help families enormously in coming to terms with such a traumatic event in their lives. I wish to thank both Royal Brompton and Harefield NHS Trust management and The National Heart and Lung Institute, Imperial College, London for their support.

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