Progress in Pediatric Cardiology xxx (2017) xxx–xxx

Contents lists available at ScienceDirect

Progress in Pediatric Cardiology

journal homepage: www.elsevier.com/locate/ppedcard

Review

Psychosocial care and cardiac genetic counseling following sudden cardiac death in the young☆

Jodie Ingles [a,b,c,*], Cynthia James [d]

[a] Agnes Ginges Centre for Molecular Cardiology, Centenary Institute, Sydney, Australia
[b] Sydney Medical School, University of Sydney, Sydney, Australia
[c] Department of Cardiology, Royal Prince Alfred Hospital, Sydney, Australia
[d] Division of Cardiology, Department of Medicine, Johns Hopkins University, Baltimore, MD, United States

ARTICLE INFO

Article history:
Received 15 December 2016
Received in revised form 28 February 2017
Accepted 1 March 2017
Available online xxxx

Keywords:
Sudden cardiac death in the young
Cardiac genetic counseling
Genetic testing
Psychological wellbeing

ABSTRACT

Sudden cardiac death is a devastating event and in up to 40% of cases in the young (<35 years) a cause of death will remain elusive. Dealing with grief over the loss of a loved one in uncertain and unexpected circumstances poses significant psychosocial burden. It is not surprising then that poor psychological sequelae including anxiety, prolonged grief and posttraumatic stress symptoms will occur in at least 1 in 2 family members. Furthermore, ongoing worry around the suspected familial nature of the disease, risk to surviving children, need to understand and act on complex genetic information and a lack of closure relating to the cause of death make the psychosocial and clinical needs of this population unique. Cardiac genetic counselors have evolved to play a key role in the care of families following a sudden cardiac death, possessing the skills, resources and time to develop a strong rapport. While the challenges this population face are vast, there are many commonalities that exist. Here we present a case that represents many of the difficulties experienced by sudden cardiac death families, providing a review of the literature and more practical recommendations based on clinical expertise. While a family will never get over the loss of a loved one, learning to find new meaning in life and minimizing psychological difficulties should be a key consideration for any clinicians involved in the care of these families.

1. Introduction

Sudden cardiac death in the young is a devastating event. In many cases it occurs without prior warning, often in healthy, active young people with no premorbid diagnoses of heart disease. In up to 40% of young sudden deaths, no cause of death will be identified at postmortem evaluation, leaving an already grief stricken family at a loss to understand why the death occurred [1]. Cardiac genetic diseases are a presumed cause of sudden unexplained death, and given the familial risk family members are advised to undergo clinical surveillance. For the families, this poses an additional stressor at a time of intense grief, as they come to terms with potential risk to themselves, to other children, and to extended family. Furthermore, with the increasing availability of postmortem genetic testing (i.e. "molecular autopsy") [2,3] and inevitable return of uncertain genetic findings, we are finding ourselves more often having complex genetic discussions with these families. Psychologically, we know family members are at increased risk of anxiety, prolonged grief and posttraumatic stress symptoms [4,5].

Given the unique challenges, management and care in a specialized multidisciplinary clinic that incorporates expertise of cardiac genetic counselors is critical. Here we describe some common challenges involved in family care. Using an illustrative case, we will give practical advice about psychosocial care and the evolving cardiac genetic counseling needs for families following sudden cardiac death in the young.

A mother calls the cardiogenetics clinic reporting that her 15 year old son suffered a cardiac arrest while playing sport. Despite prompt resuscitation efforts, including defibrillation, he was unable to revived. She has just received the postmortem evaluation, which reports some non-specific features of arrhythmogenic right ventricular cardiomyopathy (ARVC), though these are non-diagnostic and the final cause of death is unascertained. The family have been informed that the death may be familial and are advised to undergo clinical assessment with a specialist multidisciplinary clinic. The family are understandably distraught by the sudden death, reporting that the decedent had not been previously unwell, nor had he any prior cardiac diagnoses that

☆ No conflicts of interest.

* Corresponding author at: Agnes Ginges Centre for Molecular Cardiology, Centenary Institute, Locked Bag 6, Newtown, NSW 2042, Australia.
E-mail address: j.ingles@centenary.org.au (J. Ingles).

http://dx.doi.org/10.1016/j.ppedcard.2017.03.001
1058-9813/

would put him at risk of sudden death. In fact, they described him as a healthy and athletic child. The uncertainty of not knowing the cause as well as whether other children would suffer the same fate has been very problematic. The mother, who witnessed the death and resuscitation efforts, reveals she has been unable to drive her car due to uncontrolled shaking of her hands and persistent flashbacks to the event disturb her sleep.

The parents and young siblings of the decedent attend the cardiogenetics clinic for general discussion, cardiac genetic counseling and clinical screening. They are understandably anxious and very upset, though are happy to share their story with the clinic team. While they have a support network of concerned family and friends, many avoid discussion of the decedent. The mother tells us she is glad to have the opportunity to speak openly about her son to us. There is detailed discussion of the events leading up to the death, the decedent's premorbid clinical history, and the circumstances of the death. The family reveals there had been some speculation in the community about whether drugs were a factor in the death. The postmortem findings are explained, as is the rationale for why a cardiac genetic disease is suspected. Reassuring the parents that a virus he had 2 months earlier, or the headache he complained about 1 week before, were not likely related helped them remove some self-blame that they missed some important symptom that might have prevented the death.

Options for genetic testing are discussed, first noting the importance of testing a stored DNA sample (5–10 mL whole blood in an EDTA tube). The potential outcomes of this test (i.e. no variants, uncertain variants, or a likely pathogenic/pathogenic variant) are explained, as are the follow-on implications for the family. Genetic testing including a broad panel of genes associated with inherited cardiomyopathy and arrhythmias is ordered and a plan for disclosure of genetic test results put in place.

There is considerable anxiety regarding the clinical evaluation of the surviving children and fortunately both siblings (13 year old brother and 16 year old sister) and parents have no evidence of disease. A plan is made to repeat clinical screening in 12 months time, and contact details of the clinic coordinator/cardiac genetic counselor are shared to provide additional support if needed. The family is reminded that this plan may be altered by the results of genetic testing and resulting potential availability of cascade genetic testing. This uncertainty is difficult for the family to take in. Nonetheless, the parents report being relieved to know there are doctors and researchers dedicated to better understanding sudden death in the young. They are reassured that their surviving children have no evidence of disease at present.

Several months later, results of genetic testing come back from the laboratory showing a known pathogenic mutation in the gene encoding desmoplakin (DSP) and a variant of uncertain significance (VUS) in MYH7. The family is counseled that the DSP variant is likely associated with the sudden death, but there is uncertainty regarding whether the MYH7 variant also contributed to his disease. Following guidelines [6], both siblings and parents are offered cascade screening for the DSP pathogenic variant, but not the MYH7 VUS. The mother and older sibling are found to be DSP mutation carriers. They are counseled to reduce vigorous exercise to reduce likelihood of disease expression [7–9]. Ongoing cardiac screening is recommended for the father and younger sibling until further data allows improved adjudication of the MYH7 variant. The family is encouraged to share the DSP result and recommendations for cardiac screening with the mother's relatives. The ongoing uncertainty regarding 1) the likelihood the child who is a DSP carrier will eventually develop disease, 2) the likelihood the MYH7 VUS played a role in the SCD and 3) whether the children should be tested for the MYH7 VUS is difficult for this family. The father expresses frustration the data is not able to give a more definitive answer. The mother reveals differences in grieving have also proven to be a point of contention in the family, with the mother's open coping style vastly different from her husband's need to grieve more privately. Making meaning of the death is something both parents are having overwhelming difficulty with.

The family returns to clinic yearly for cardiac evaluation of the DSP carriers. They struggle with exercise restrictions recommended. They continue to grieve for their son/brother, but have made progress, forming a foundation in his memory and remaining involved in the community of children and families that were his peers.

1.1. First Contact With the Family

There are many pathways families take to the cardiogenetics clinic following a young sudden death. They may be notified of the possibility of a cardiac genetic disease following the postmortem examination, or many years may pass before finding their way to a specialist center. The clinic coordinator or cardiac genetic counselor may take the initial phone call, and the skills in being able to sensitively gather important clinical and family information are invaluable. At this point in time, determining whether the referral to the specialist center is appropriate is vital. Given the pressing time and resource demands of offering such highly skilled clinical services, triage of referrals ensures those families who are truly at risk of a cardiac genetic disease are seen promptly. Likewise, those families where the death is unlikely to be familial can be somewhat reassured.

First contact can be a variable experience, though common elements often exist and these are discussed below.

(1) Lack of understanding of the death: There is little awareness in the community that young, healthy individuals can die suddenly. When explaining the causes of sudden cardiac death and the frequency it occurs, many family members express relief to know there is some understanding of what has occurred in their family. While there is disbelief that young people can “drop dead” for no apparent reason, knowing there are others in the same situation is reassuring. Many families receive intense scrutiny about the cause of death, with some finding those in the community speculating about suicide or drug overdose. Police involvement is not infrequent. Not surprisingly, this can be very upsetting for the family.

(2) Acute grief: Grief is a natural psychological response to the loss of a loved one. The psychology of grief is well studied, and while most believe there is not one ‘right’ way to grieve, common elements include disbelief, yearning, anger, depression and acceptance, peaking within 6-12 months post-death [10]. Whilst one may never truly recover from the sudden death of a significant other, over time many people are able find a way to accept the loss without clinical intervention, to create a revised but meaningful life without the deceased [11]. In approximately 7% of bereaved people, this process does not progress and is often referred to as prolonged grief, or persistent, complex bereavement disorder in the Diagnostic and Statistical Manual of Mental Disorders 5th Edition (DSM-5) [12,13]. When managing a family soon after a death, care should be taken to acknowledge the high-level of psychosocial stress, which can manifest as anger, irritability, depression, guilt, insomnia, reduced appetite and weight loss. Some data also suggest early bereavement may impact cognition [14], an important consideration when complex and emotive discussions around inheritance and genetic testing options may be necessary. Evidence from elderly bereaved populations show most psychosocial impairments will normalize over time, however sub-groups at increased risk of unresolved grief exist. Failure to move beyond the acute grief stage is an indicator of prolonged or complicated grief and will be discussed further below.

(3) Collection of family history and decedent details: Understanding the cause of a sudden cardiac death is likened to detective work, where pieces of evidence are investigated and eventually put together to give an overall impression. In this process, information gathered from a detailed 3-generation family history has

Please cite this article as: Ingles J, James C, Psychosocial care and cardiac genetic counseling following sudden cardiac death in the young, Prog Pediatr Cardiol (2017), http://dx.doi.org/10.1016/j.ppedcard.2017.03.001