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Interdisciplinary psychosocial care for families with inherited cardiovascular diseases

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ABSTRACT

Inherited cardiovascular diseases pose unique and complex psychosocial challenges for families, including coming to terms with life-long cardiac disease, risk of sudden death, grief related to the sudden death of a loved one, activity restrictions, and inheritance risk to other family members. Psychosocial factors impact not only mental health but also physical health and cooperation with clinical recommendations. We describe an interdisciplinary approach to the care of families with inherited cardiovascular disease, in which psychological care provided by specialized cardiac genetic counselors, nurses, and psychologists is embedded within the cardiovascular care team. We report illustrative cases and the supporting literature to demonstrate common scenarios, as well as practical guidance for clinicians working in the inherited cardiovascular disease setting.

Key words: Psychological wellbeing, Genetic testing, Hypertrophic cardiomyopathy, Quality of life.

Introduction

Cardiovascular genetics has rapidly grown into an important subspecialty of cardiology, covering care of diseases such as inherited cardiomyopathies [e.g., hypertrophic cardiomyopathy (HCM)], primary arrhythmogenic disorders [e.g., catecholaminergic polymorphic ventricular tachycardia (CPVT)], aortopathies (e.g., Marfan syndrome),

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and dyslipidemias [e.g., familial hypercholesterolemia (FH)]. These disorders are common, with an aggregate prevalence greater than 1 in 200. Interdisciplinary specialized cardiovascular genetics clinics have been developed around the world as an ideal model of care, incorporating the expertise of cardiologists, electrophysiologists, geneticists, genetic counselors, nurses, psychologists, and others [1–3].

Inherited cardiovascular diseases pose unique psychological challenges for patients and families, which arise from the hereditary nature of the diseases, the risk of sudden death at a young age, and the potential for symptom onset across the lifespan [4–6]. Diagnoses are often made in childhood, adolescence or early adulthood, sometimes in the setting of a death of a family member prompting evaluation with the need to focus not only on changes in the health status of a young person but also the risk to other family members. Many patients may experience no or only mild symptoms, yet are advised to substantially alter their lifestyle to manage the risk of sudden death. Indeed, exclusion from competitive sports and intense exercise is a major psychosocial challenge for many of these patients [7,8]. In addition to activity restrictions, some patients will be recommended an implantable cardioverter defibrillator (ICD) for sudden death prevention. While ICD therapy is potentially life saving, the psychological consequences of ICDs have been well described, such as the association between multiple shocks and posttraumatic stress [9], as well as body image concerns among many young patients [10]. Correlates of psychological difficulty in patients with ICDs are particularly prevalent in the cardiovascular genetics population, including younger age, female gender, loss of role function (e.g., competitive athletics) and, for some diseases, multiple shocks (e.g., CPVT) [11]. Families who present after the sudden, unexpected death of a young relative may also bear significant psychological burden, including complex grief responses requiring psychological intervention [12,13]. Supporting surviving family members clinically and psychologically is a major challenge in the specialized clinic setting, but one that must be considered if we are to ensure a high-standard of care for these families.

The psychological challenges faced by families with inherited cardiovascular diseases have the potential to affect not only their emotional wellbeing, but also medical outcomes and demands on the clinical team. Among cardiac patients, psychological morbidity has been associated with medication non-adherence, poor self-care, increased mortality, and longer medical visits [14–17]. Given the psychological challenges posed and the influence of psychological wellbeing on health outcomes, the offer of psychological care as a component of the overall management of families is vital. We present case vignettes from our centers that demonstrate the process and impact of psychologically informed care for common issues in inherited cardiovascular disease. The cases highlight an interdisciplinary model, with psychological care provided by specialized genetic counselors, nurses, and psychologists embedded in the cardiovascular care team, and illustrate the unique role of cardiac genetic counselors, going beyond genetic testing to provide specialized support in many other settings. We also provide an overview of the literature supporting this approach to care and provide recommendations for clinicians.

Case 1: Adjustment to diagnosis, exercise restrictions and ICD shocks

After being told 2 years earlier that he had arrhythmogenic right ventricular cardiomyopathy (ARVC), a 54-year-old man presented for the first time to an interdisciplinary cardiovascular genetics center. Prior to diagnosis, cycling had been a major source of self-esteem and stress management, and a central part of his identity. The man reported significant difficulty adapting to his diagnosis, especially the recommended exercise restrictions. The genetic counselor acknowledged this as a major loss and helped him to recognize the need for a grieving process. Having had his emotional response to exercise restrictions previously dismissed, he reported it was helpful to have his experience heard and understood. He also described symptoms of anxiety and avoidance behaviors as a result of multiple appropriate ICD shocks. This included a new hypervigilance to bodily sensations, persistent worries about getting shocked, fear and avoidance of physical activity, and a pervasive sense of threat. He presented to the emergency department repeatedly with concerns about arrhythmias, but was always found to be in normal rhythm. The genetic counselor provided information on the relationship between number of shocks and psychological difficulties, acknowledging his reactions as common and treatable. He also reported depressed mood, a loss of interest in activities he previously enjoyed, and guilt and remorse about how his cycling had contributed to his ARVC. With empathic and supportive counseling from the genetic counselor he was able to work through these feelings while also developing a better understanding of the genetic predisposition. A session with the team psychologist helped him to adjust his thinking and understand his distress in the context of other major life changes. Overall, the man reported that his care at the center improved his mood, alleviated daily fears about his condition, helped him to adapt to the major shift in his lifestyle, and improved his cooperation with exercise recommendations. His repeat visits to the emergency department ceased and he re-engaged in activities he had been avoiding.

For this patient, and many others with inherited cardiovascular disease, the change in his lifestyle and living with an ICD were the most significant concerns in adjusting to his diagnosis. Difficulty accepting and adjusting to recommendations to avoid intense and competitive sport has been consistently reported [7,8]. Patients report that discontinuation of sports and exercise influences self-esteem, body image, ability to cope with stress, family activities and relationships, identity, and social engagement. Many experience this as a true loss and must grieve it as such [7]. Naming the loss, facilitating grieving, validating the psychosocial impact, and providing empathic supportive counseling can all help facilitate adjustment.

Approximately one-third of individuals with an ICD report clinically significant symptoms of anxiety or depression [18,19]. For people with mild levels of distress, the benefits of brief psychological interventions provided by their medical team have been shown [11], while those with marked anxiety, depression or posttraumatic stress warrant referral to a psychologist, ideally one with expertise in ICD therapy [20].