JNP

Genetic Knowledge of Parents and Children With Inherited Cardiac Syndromes

Kathleen T. Hickey, EdD, FNP-BC, Teresa C. Riga, BS, Shazia Mitha, MS, BSN, Meghan Reading, BSN, Kate N. Warren, BA, Robert Sciacca, MS, EngScD, and Mary W. Byrne, PhD, DNP

ABSTRACT

Although genetic knowledge related to sudden arrhythmia death syndromes (SADS) has enhanced risk prediction and treatment, it is challenging to assess genetic literacy of individual patients and families. To address this, participants attending the 2015 SADS conference were asked to complete a survey measuring general knowledge of genes and heredity. Correct responses averaged 85 ± 9% with correct responses of ≥ 80% for 17 of 21 questions. However, attitudes toward genetic testing varied markedly. These results suggest that understanding motivations or disincentives to pursue genetic testing is as important as enhancing genetic knowledge for providing effective treatments for SADS.

Keywords: cardiac genetics, genetic knowledge, patients and families

INTRODUCTION

The era of Precision Medicine has had a significant impact on clinical practice and research for those living with an inherited channelopathy or cardiomyopathy.[1,2] Knowledge of specific genetic findings causative for or associated with sudden arrhythmia death syndromes (SADS) has enhanced risk prediction and diagnostic accuracy, and has guided management of genetic testing and genetic counseling in patients with risk of sudden death due to heritable arrhythmias.[3] As members of the interdisciplinary team, geneticists and nurses are often faced with the challenge of assessing the genetic literacy of individual patients and families regarding genes and heredity. Thus, there is a clear need to evaluate the basic genetic literacy of individuals and families living with SADS conditions to address areas where genetic knowledge can be improved. Addressing gaps in genomic knowledge is important to ensure that individuals can make informed decisions related to their personalized health. This pilot study presents quantitative data on knowledge about genes and heredity and attitudes regarding genetic testing collected from individuals and families living with SADS conditions.

BACKGROUND

Completion of the Human Genome Project and improved integration of genetics into health care have ushered in a new era of improvements in personalized health, illness, disease risk, and treatment response, known as Precision Medicine.[4] However, only limited data are available related to the public's basic knowledge of genetics, genomics, and other "omics." Moreover, how individuals and families have begun to integrate omic knowledge into their lives and personal health choices is just beginning to emerge in areas such as oncology, cardiology, and neurology.[5]

The emerging and important field of genomics brings together many disciplines. Genetically trained counselors and nurses are uniquely suited to evaluate the public's genetic literacy and knowledge. They have extensive knowledge of patient, family, and community perspectives; an understanding of biologic and genomic underpinnings; experience with new and evolving genetic technologies; skills in communication; experience in collaboration with individuals, families, and advocacy groups; as well as the public's trust. Despite this, little is known about

the genetic literacy of those living with inherited cardiac conditions such as a channelopathy or cardiomyopathy, conditions that place these individuals at risk for sudden cardiac death (SCD).

SCD afflicts > 400,000 people each year in the United States. The majority of these tragic events occur in otherwise healthy adults without underlying cardiac disease.[3,6] Hypertrophic cardiomyopathy and long QT syndrome (LQTS) are examples of inherited cardiovascular conditions that can lead to lethal arrhythmias and SCD. Many of the inherited cardiac disorders of the heart have known genes and an autosomal dominant pattern of inheritance, meaning all individuals who harbor a genetic mutation will pass the mutation on to half of their children. Cardiac symptoms may be vague or absent. In hypertrophic cardiomyopathy, for example, individuals may experience cardiac symptoms ranging from palpitations and dizziness to more severe symptoms such as chest pain or syncope (loss of consciousness). However, in some asymptomatic individuals, an unexpected, lethal ventricular arrhythmia may be the initial catastrophic presentation.[3,7,8] Many at-risk individuals and family members may decide to undergo genetic testing to guide their treatment decisions. Treatments include medications, placement of an implantable cardioverter-defibrillator for protection against SCD, or "watchful" follow-up with regular cardiac diagnostic testing, such as with echocardiograms, electrocardiograms, or stress testing. The basic knowledge of individuals regarding genetics, especially in those living with a known diagnosis of SADS, remains poorly understood.

The aims of this study were to: (1) evaluate the knowledge about genes and heredity among individuals and families who were diagnosed with a previous SADS; and (2) determine the association between knowledge, sociodemographic factors, and attitudes regarding genetic testing.

METHODS

Participants

Our study sample consisted of individuals and family members attending the 8th annual 2015 International SADS Foundation Conference. The goal of the Conference was to increase awareness of the SADS Foundation; enhance patient and family support; expand education to health professionals; and increase advocacy for research, screening, and medical treatment. The SADS leadership committee reviewed and approved the de-identified questionnaire before the conference and approved access to the attendees, excluding providers. Potential participants were informed at the SADS conference of the voluntary nature of their participation in this research, and their informed consent was obtained.

Instrumentation

The SADS survey was adapted in part from Jallinoja and Aro's questionnaire on knowledge about genes and heredity among Finns,[9] which consisted of 21 structured items. Sixteen items measured general knowledge of genes and heredity, including knowledge of the association between genes and disease and the interplay between genes, chromosomes, and cells within the body. Five additional items measured knowledge regarding the role of genes in susceptibility to disease. For each individual, a summary index of the percentage of correct responses was calculated. The reliability of the survey items (Cronbach alpha has been reported to be 0.86).[9]

The research team added sociodemographic questions and 22 specific questions regarding attitudes toward genetic testing. The latter were evaluated on a Likert scale ranging from totally agree, agree, don't know, disagree, or totally disagree. They included questions regarding experiences with DNA testing and perceptions about the stigma associated with genetic testing.

Procedures

An invitation to participate in the study was conducted in person at the annual SADS Meeting in 2015. Participants consisted of both patients and their family members who attended the conference; providers were excluded from participating in the survey. Participants had the opportunity to ask specific questions in a group or individually related to study participation prior to signing informed consent and completion of the anonymous survey. Participants were instructed to complete the survey and answer all questions during a dedicated block of time at the conference. The survey session was not timed and all