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Care of Patients with Cardiopulmonary Conditions

The effect of cardiac genetic testing on psychological well-being and illness perceptions

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ABSTRACT

Objective: To assess the effects of positive cardiac genetic diagnoses, ICD discharges, and arrhythmias on measures of psychological well-being.
Methods: Fifty-eight adults with prior cardiac genetic testing were enrolled. Patient well-being was determined using the SF-36 (QoL), HADS-A and HADS-D (anxiety/depression), and IPQ-R (patients' perceptions of illness). Patients with positive and negative cardiac genetic test results were compared using non-parametric statistics.
Results: Genetic testing yielded 76% with a positive diagnosis and 29% reported an ICD shock. QoL assessments ($n = 33$) were within normal ranges (mean of 50) with the exceptions of general health (44.1 ± 12.2, $p < 0.01$) and bodily pain (55.1 ± 9.1, $p < 0.01$) domains, but only the bodily pain domain showed differences between those with positive and negative cardiac genetic test results. Subjects with ICD discharges had higher scores than those without shocks in consequential and emotional IPQR subscales as well as greater perceived risks of experiencing a serious cardiac event, developing additional symptoms, or limitations in daily activities.
Conclusion: Positive genetic results did not negatively impact patient well-being with the exception of the bodily pain domain of the SF-36.

Inherited channelopathies such as Long QT syndrome (LQTS), Brugada (BrS), Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) and the Dilated (DCMs) and Hypertrophic Cardiomyopathies (HCMs) have an underlying genetic basis.[1–3] Cardiac genetic testing may facilitate the identification of the molecular pathogenesis that can place an individual or family at an increased risk of arrhythmias and sudden cardiac death (SCD).[4] Although these conditions are genetically and clinically heterogeneous, they all share an autosomal dominant mode of inheritance, which has significant implications for both patients and family members, including children of mutation carriers.[5] Once a specific cardiac mutation is identified within a family, genetic testing can be used to identify other at risk individuals within a family who may harbor that familial mutation.[5,6] In fact, life-threatening arrhythmias and/or SCD, in otherwise young, healthy individuals can often be the first devastating presentation of an underlying cardiac genetic condition.[4] Thus, it is imperative to identify individuals at risk, so protective therapies such as the initiation of beta blocker medication or placement of an implantable cardioverter defibrillator (ICD) can be initiated.[3,5]

The implantation of the internal ICD is one approach recommended by existing clinical guidelines to prevent SCD in those who have ventricular arrhythmias, or have a clinical/family history of an underlying inherited cardiac condition that places them at increased risk for arrhythmias or SCD.[6,7] ICDs terminate lethal

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ventricular arrhythmias either by overdrive antitachycardia pacing (ATP) or delivering an internal electrical shock to the heart to restore a normal sinus rhythm.[8]

Patients who are diagnosed with a genetic condition develop beliefs about their condition, and these views are key determinants of their illness perception and overall well-being.[9–13] Individuals with the same illness or injury can have widely different perceptions of their condition and these perceptions can lead to very different illness trajectories. This is a dynamic process which changes in response to the patients' perceptions and ideas about their illness. Illness perceptions directly influence the individual's emotional response to the illness.[13] However, despite the importance of the patients' perception of their illness belief this information is rarely sought by healthcare providers in clinical practice. A meta-analysis of quantitative prospective observational studies showed that positive psychological well-being was associated with reduced mortality in both disease and healthy populations, highlighting the importance of evaluating these measures.[14] Additionally, limited published literature exists on overall psychological well-being, illness perception and quality of life (QoL) in a cardiac genetic population.

As cardiac genetic testing becomes even more widely available, and the reporting of cardiac incidental findings becomes integrated into cardiac care,[15] it will become increasingly important to understand the impact of genetic results on physical and psychological well-being. In this study we aimed to assess the relationship between a positive cardiac genetic diagnosis and measures of patient psychological well-being and illness perceptions in order to test the hypothesis that a positive genetic test result may negatively impact these measures. The outcome measures assessed included SF-36 QoL domains, measures of generalized anxiety and depression, perceived risks of experiencing a serious cardiac event or developing additional symptoms or limitations in daily activities, and patients' perceptions regarding the nature of their illness.

Previous studies have shown that patients who experience ICD discharges have increased levels of anxiety, depression and poorer self-reported QoL.[16,17] Therefore, the effects of ICD discharges and cardiac arrhythmias on psychological well-being and illness perceptions were assessed and ICD discharges were also included as a potential confounder in the analyses of the effect of positive cardiac genetic diagnoses.

Methods

Study design

This was a single center, cross-sectional convenience study of patients who had undergone prior clinically indicated cardiac genetics testing between January 2005 and March 2012. Approval to conduct the study was obtained from the Institutional Review Board at Columbia University. The investigation was carried out according to the principles outlined in the Declaration of Helsinki, including written informed consent from all participants.

Participants

The majority of patients who volunteered to participate had a known cardiac genetic diagnosis and had undergone ICD placement under established clinical guidelines. Previously indicated genetic testing was undertaken to determine the underlying genetic basis of their cardiac disease.

Setting

Potential research participants were screened and recruited from the cardiac electrophysiology service and ICD clinic at Columbia University and through the Hypertrophic Cardiomyopathy Association annual meeting.

Inclusion and exclusion criteria

As part of the informed consent process, all patients were asked if they would be willing to participate in a research protocol that would require sharing their cardiac clinical data, cardiac genetic test results, and family history. Eligibility criteria included being age 18 or older and prior cardiac genetic testing for an inherited arrhythmia or cardiomyopathy as well as a willingness to share cardiac clinical data and complete the study questionnaires. Exclusion criteria included age under 18 and unwillingness to have clinical data and genetic test results collected.

Data collection

Clinical data collected included age, gender, self-reported race/ethnicity, results of clinical cardiac genetic testing, ICD placement, and ICD arrhythmia findings (stored electrograms of cardiac arrhythmias recorded in the memory of the ICD). The number of months since cardiac genetic testing was also recorded. Cardiac genetic diagnosis and arrhythmias were confirmed by medical record review.

Instruments

Short form-36 item (SF-36 v2™) quality of life

Quality of life (QoL) was assessed using the Medical Outcomes Study Short Form-36 (SF-36), a widely used, well-known, self-reported measure.[18–20] The SF-36 has been standardized, validated, and used successfully in younger and older patient populations, including those with an ICD.[21] The questionnaire contains 36 items and yields the 8 domain scores of physical functioning, physical role limitations, emotional role limitations, bodily pain, general health perceptions, vitality, social function, and mental health. In addition, physical and mental health summary scores are calculated.[18–20] Psychometric testing of the SF-36 has established construct, predictive, and known-groups validity and good reliability and sensitivity to change have been reported.[18–20] Scores are standardized to population norms using published algorithms, with a mean score set of 50. Higher scores indicate better perceived quality of life.

Hospital anxiety and depression scale (HADS)

Psychological distress was evaluated using the Hospital Anxiety and Depression Scale (HADS).[22] The scale measures generalized anxiety (HADS-A) and depression (HADS-D) with seven items each and have response options range from 0 (not at all) to 3 (very much), adding up to a maximum score of 21 for each subscale (anxiety or depression). A score of 8 indicates elevated distress and a score 11 indicates potentially clinically significant distress for each of the two subscales separately.[22] Cronbach's alphas for HADS-A and HADS-D of 0.84 and 0.83, respectively, have been reported.[9]

Illness perception questionnaire (IPQ-R)

To assess illness perceptions among participants, the revised version of the Illness Perception Questionnaire (IPQ-R) was used.[23] The IPQ-R provides an 18 item assessment of the key components of patients' perceptions of illness based on Leventhal's Self-Regulatory Model and has been utilized in previous studies of hereditary diseases.[23] The questionnaire included the following