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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

potential causes for the illness: stress, heredity, diet, germ/virus, chance, poor medical care, pollution, patient behavior, patient's negative attitude, worry about family problems, overwork, feeling dejected, aging, alcohol, smoking, accident/injury, personality, and altered immunity. Possible responses for each item included "strongly disagree," "disagree," "neither agree nor disagree," "agree," and "strongly agree" and were scored on a scale of 1–5.

The subscales of the IPQ-R are "timeline acute/chronic," "timeline cyclical," "consequences," "personal control," "treatment control," "illness coherence," and "emotional representations." An adjusted mean score (sum of the scale items divided by the number of items) was calculated, with a possible maximum score of 5 for each subscale. Higher scores on these subscales refer to a stronger belief in a chronic/stable course; a stronger belief in serious consequences of having a positive mutation status; a stronger belief that the illness is controllable either by self-care or medical care; a better understanding of the meaning of one's mutation status; and consistency of the subscales has been reported to range from 0.67 to 0.84 in those with and without manifest HCM.[16]

The perceived risks of developing (additional) symptoms and of developing limitations in daily activities were measured with two items for each event, assessing (1) the risk of that event occurring on a scale ranging from 0% to100%, and (2) the perceived severity of the risk of that event occurring on a similar scale; responses were characterized as ranging from "very small" to "very large."[19] For each of the three outcomes a total perceived risk score was calculated by averaging the two item scores.

In addition, the IPQ-R questionnaire asked patients about their symptoms and whether they felt their symptoms were related to their illness. Symptoms queried included: pain, sore throat, nausea, breathlessness, weight loss, fatigue, stiff joints, sore eyes, wheeziness, headaches, upset stomach, sleep difficulties, dizziness, and loss of strength. Any additional cardiac related symptoms were captured at the time of enrollment including palpitations, shortness of breath and chest pain/pressure.

Data analysis

All data were reviewed for accuracy and completeness at the time of collection. Results were expressed as means with standard deviations for continuous variables. For categorical variables, frequencies and percentages were reported. Due to the non-normal distribution of many of the variables, non-parametric statistical tests were used to assess statistical significance. Specifically, the Mann–Whitney *U* test was used to test for differences in measures of well-being and illness perceptions between patients with positive and negative cardiac genetic test results as well as between patients with and without ICD shocks. The potential confounding effects of ICD discharges on the relationship between cardiac genetic test results and measures of well-being and illness perceptions were assessed using a non-parametric analog of an analysis of covariance.[24] In addition, Spearman's correlation was used to test whether outcome measures were related to age or gender while the Kruskal–Wallis test was used to test for differences in outcome measures among cardiac diagnoses. A p-value <0.05 was considered significant for all analyses. Data were analyzed using SAS 9.2 (SAS Institute, Cary, NC).

Results

Population characteristics

Greater than 90% of patients contacted agreed to participate in the study; a total of 4 patients declined participation because of time constraints, one because of no direct benefit for participation, and one did not want to complete the study questionnaires. Two patients did not want to discuss their family history or share their genetic information.

Table 1
Demographic and clinical characteristics of the study population

Demographics	
Number of subjects	58
Age (years, mean ± SD)	41 ± 14
Males	36 (62%)
Race/ethnicity	
Caucasian	48 (83%)
Hispanic	6 (10%)
Asian	2 (3%)
African–American	1 (2%)
Other	1 (2%)
Clinical diagnoses	
ARVD	2 (3%)
Brugada	8 (14%)
Dilated cardiomyopathy	11 (19%)
Hypertrophic cardiomyopathy	24 (41%)
Long QT syndrome	13 (22%)
Electrocardiographic findings	
Atrial fibrillation/flutter	10 (17%)
Non-sustained ventricular tachycardia	11 (19%)
Sustained ventricular tachycardia	2 (3%)
Conduction block	8 (14%)

The demographic and clinical characteristics of the 58 participants in the study are presented in Table 1. The average age of the participants was 41 ± 4 years and 36 (62%) were male. The most frequent diagnosis was Hypertrophic Cardiomyopathy (HCM) (41%), followed by Long QT Syndrome (22%), Dilated Cardiomyopathy (DCM) (19%), Brugada Syndrome (14%), and Arrhythmogenic Right Ventricular Cardiomyopathy (3%). Among participants, 47 had an ICD implanted (81%) and 17 of those 47 (36%) had experienced a discharge from their ICD. Arrhythmias present among participants included VF/VT in 17 (29%), NSVT in 11 (19%), SVT in 2 (3%), and conduction block in 13 (22%). All patients had suffered a cardiac event or underwent cardiac genetic testing because of a family history of SCD with 44 (76%) testing positive for an inherited cardiac genetic condition. The median interval between cardiac genetic testing and tests to assess patient well-being was 18 months (range 3–81 months).

Participants felt strongly that their illness was due to hereditary causes averaging a score of 4.4 ± 0.8 out of 5. All other potential causes averaged <2 with the exception of stress (2.4 ± 1.4) and chance (2.3 ± 1.3). From a list of 14 possible symptoms on the IPQ-R

Table 2a
Norm-based quality of life profiles

	Total cohort ($N = 33$)	Testing positive ($n = 26$)	Testing negative ($n = 7$)
	Mean ± SD	Mean ± SD	Mean ± SD
Component summary measures			
Physical component summary	48.33 ± 11.60	46.30 ± 11.70	55.88 ± 7.91
Mental component summary	50.86 ± 10.11	51.30 ± 10.48	49.22 ± 9.15
Health domain scales			
Physical functioning	47.53 ± 9.72	46.02 ± 10.14	53.12 ± 5.35
Role physical	48.54 ± 13.88	46.68 ± 15.04	55.45 ± 3.70
Body pain	55.06 ± 9.10	53.35 ± 9.56	61.39 ± 1.24*
General health	44.11 ± 12.18	43.51 ± 11.64	46.33 ± 14.81
Vitality	48.87 ± 11.74	47.86 ± 12.58	52.63 ± 7.45
Social functioning	51.23 ± 8.78	50.77 ± 9.04	52.95 ± 8.16
Role emotional	50.23 ± 14.45	50.50 ± 13.87	49.22 ± 17.63
Mental health	50.71 ± 6.80	50.57 ± 7.37	51.21 ± 4.45

*$p = 0.046$ vs. testing positive.

Table 2b
IPQR subscales

	Total cohort ($N = 24$)	Testing positive ($n = 17$)	Testing negative ($n = 7$)
	Mean ± SD	Mean ± SD	Mean ± SD
Timeline acute/chronic	4.26 ± 0.80	4.22 ± 0.80	4.36 ± 0.82
Timeline cyclical	2.53 ± 0.87	2.60 ± 0.89	2.36 ± 0.86
Consequences	3.35 ± 0.87	3.43 ± 0.93	3.14 ± 0.72
Personal control	3.47 ± 0.80	3.40 ± 0.79	3.62 ± 0.86
Treatment control	3.55 ± 0.68	3.70 ± 0.63	3.19 ± 0.72
Emotional representations	2.71 ± 0.86	2.66 ± 0.87	2.83 ± 0.87

questionnaire, participants selected up to 12 as being present with an average of 5.0 ± 3.6 symptoms per patient; the average number of symptoms patients considered to be related to their illness was 3.1 ± 3.4.

Measures of patient well-being and illness perceptions

The mean and standard deviations for the SF-36 QoL domain and summary scores and for IPQ-R subscales are presented in Tables 2a and 2b. Mean scores for all domains and dimensions of the SF-36 were within the normal range (mean of 50) with the exception of the general health dimension which was lower than normal (44.1 ± 12.2, $p < 0.01$) and the bodily pain dimension which was higher than normal (55.1 ± 9.1, $p < 0.01$). Mean scores for IPQ-R subscales ranged from 2.5 ± 0.9 for timeline cyclical to 4.3 ± 0.8 for timeline acute/chronic. The perceived risks of experiencing serious cardiac events or developing additional cardiac symptoms or limitations related to their illness were substantial, averaging 60.5 ± 36.0%, 60.1 ± 31.7%, and 49.8 ± 31.7%, respectively (Table 2c). From the HADS questionnaire, 29.2% exhibited signs of anxiety and 8.3% depression (Table 2d).

Effect of cardiac genetic test results on measures of patient well-being and illness perceptions

Individuals with positive cardiac genetic test results scored significantly lower on the bodily pain dimension of the SF-36 (53.3 ± 9.6 ($n = 26$) vs. 61.4 ± 1.2 ($n = 7$), $p = 0.046$), but did not fall below the value of 50 which is standard for the general population (Table 2a). The other QoL domains (physical functioning, physical role limitations, emotional role limitations general health perceptions, vitality, social function, and mental health) and the physical and mental health summary scores did not differ significantly between those with and without a positive cardiac genetic test result. IPQ-R subscales were not significantly affected by the results of undergoing prior genetic testing (Table 2b). Similarly, perceived risks for experiencing serious cardiac events, developing additional cardiac symptoms, and

Table 2c
Perceived risks

	Total cohort ($N = 24$)	Testing positive ($n = 17$)	Testing negative ($n = 7$)
	Mean ± SD	Mean ± SD	Mean ± SD
Risk of serious cardiac event	60.5 ± 36.0%	58.8 ± 38.5%	64.6 ± 31.5%
Risk of additional symptoms	60.1 ± 31.7%	68.5 ± 29.2%	39.6 ± 29.7%
Risk of limitations in daily life	49.8 ± 31.7%	51.5 ± 34.0%	45.7 ± 27.3%

Table 2d
HADS

	Total cohort ($N = 24$)	Testing positive ($n = 17$)	Testing negative ($n = 7$)
Anxiety (% with score >8)	29.2%	29.4%	28.6%
Depression (% with score >8)	8.3%	11.8%	0%

developing limitations due to their illness did not relate to a positive or negative genetic test result (Table 2c). Furthermore, the percent exhibiting anxiety or depression was not affected by the presence of a positive genetic test result (Table 2d). The time interval between cardiac genetic testing and assessments of patient well-being had no significant effect on the results. Age, gender, and cardiac diagnosis did not relate to any of the measures of patient well-being.

Effect of ICD discharges on measures of patient well-being and illness perceptions

Those with and without ICD discharges did not differ significantly with respect to QoL domains or summary scores. However, the consequences and emotional representations IPQ-R subscales differed significantly between individuals who had experienced shocks from their ICD ($n = 9$) and those who had not had shocks ($n = 15$). Participants who had experienced shocks had higher scores for consequences (3.9 ± 0.8 vs. 3.0 ± 0.7, $p = 0.023$) and for emotional representations (3.2 ± 1.0 vs. 2.4 ± 0.6, $p = 0.040$) subscales (Fig. 1). Significantly higher scores for the emotional representations subscale were also found in patients who had VF/VT (3.4 ± 0.9 ($n = 8$) vs. 2.4 ± 0.6 ($n = 16$), $p = 0.013$) and in those with conduction block (3.6 ± 0.9 ($n = 4$) vs. 2.5 ± 0.8 ($n = 20$), $p = 0.028$). The perceived risks for experiencing serious cardiac events, developing additional cardiac symptoms, and developing limitations related to their illness differed significantly between those who had experienced shocks from their ICD and those without shocks. Patients who had experienced shocks had higher perceived risks for experiencing serious future cardiac events (79.4 ± 28.7% vs. 49.2 ± 35.9%, $p = 0.046$), developing additional cardiac symptoms (78.9 ± 28.0% vs. 48.8 ± 28.9%, $p = 0.017$), and developing limitations related to their illness (70.6 ± 30.5% vs. 37.3 ± 26.1%, $p = 0.012$) (Fig. 2). The percent exhibiting anxiety or depression was not affected by the presence or absence of prior ICD discharges.

No significant confounding effects were observed when the presence of ICD discharges was included in the analyses of the

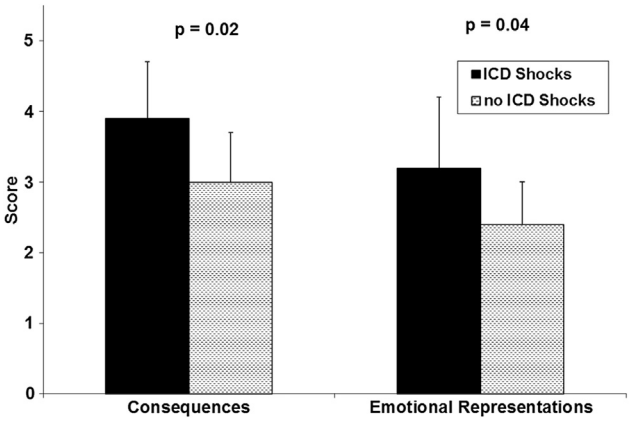

Fig. 1. The comparison between ICD shocks, consequences, and emotional representations from the revised illness perceptions questionnaire (IPQ-R).

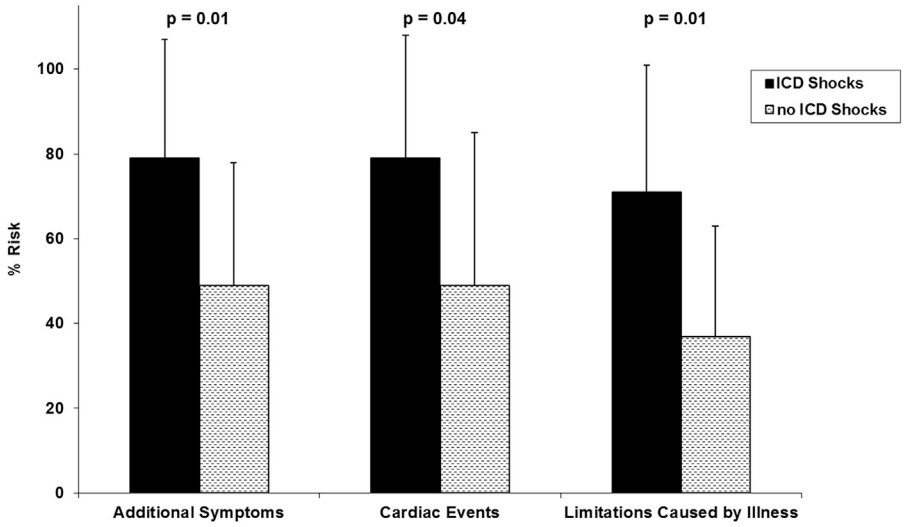

Fig. 2. The comparison between ICD shocks and perceived risks.

effect of cardiac genetic test results on patient well-being and illness perceptions. Only the bodily pain dimension of the SF-36 differed significantly between patients with positive and negative cardiac genetic test results with adjustment for ICD discharges ($p = 0.043$).

Discussion

As cardiac genetic testing becomes more widely available and integrated into cardiac care, it is important for healthcare providers to understand the potential impact of genetic testing on patient well-being. While the diagnostic value of cardiac genetic testing is evident, knowledge of how the results of genetic testing may contribute to emotional responses and perceptions has not been explored. This study aimed to examine the relationship between positive cardiac genetic diagnoses and measures of patient well-being and illness perceptions. Our results indicate that having a positive cardiac genetic diagnose did not negatively affect overall self-reported well-being or illness perceptions with the exception of the bodily pain domain of the SF-36. This finding might be due to the relatively long interval between cardiac genetic testing and assessments of patient well-being (median of 18 months). Alternatively, the fact that 60% of our population was diagnosed with HCM or DCM and were symptomatic may have influenced their sense of well-being and illness perceptions; all of these patients reported frequent symptoms such as chest discomfort, shortness of breath and fatigue that caused them bodily pain/discomfort.

While those who experienced a prior ICD shock did report altered illness perceptions and higher perceived risk, ICD discharges were not found to act as a confounder with respect to the effects of a positive cardiac genetic diagnosis. Our findings that cardiac genetic testing did not affect measures of long term anxiety, depression, and psychological distress are similar to other studies that reported no long-term negative consequences of undergoing genetic testing for other heritable diseases.[17–19] While the results of cardiac genetic testing had no significant effect on patients' perceptions of their illness, patients who experienced ICD shocks had higher scores for the consequences and emotional representations IPQ-R subscales as well as greater perceived risks for developing additional cardiac symptoms, experiencing cardiac events, and developing limitations related to their illness. These results are consistent with previous reports showing that patients who experience ICD discharges have increased levels of anxiety, depression and poorer self-reported QoL.[20,21]

However, while previous studies have found that ICD discharges are associated with diminished QoL[21] the effect may depend upon the time interval between ICD discharges and QoL assessments.[25] Furthermore, other studies have reported that the number of shocks experienced by a patient is a major determinant of the impact on QoL.[26] Regardless of the nature and complexity of these associations, it is important to account for the influence of ICD shocks when assessing patient well-being in those who have undergone cardiac genetic testing.

Limitations

This study has a number of limitations. The study was cross-sectional and conducted at a single center with an unusual sample of patients who had a clinically indicated ICD typically implanted many years prior to the availability of cardiac genetic testing. This limits inferences about causality and generalizability. Moreover, the small sample size limited the power for tests of group differences. In addition, longitudinal data was not obtained (as many older ICD records were not available for review) and changes in outcome measures pre- and post-cardiac genetic testing could not be ascertained. Nevertheless, this is useful preliminary data in a largely unexamined cardiac population where little is known about overall patient well-being.

Conclusions & implications for further research

Positive cardiac genetic results were not associated with diminished patient well-being with the exception of the bodily pain domain of the SF-36 in the overall group. However, patients with ICD discharges had higher scores in consequential and emotional IPQR subscales as well as greater perceived risks of experiencing a serious cardiac event or developing additional symptoms or limitations. However, ICD discharges were not found to be a confounder of the relationships between the results of genetic testing and measures of patient well-being and illness perceptions. Nevertheless, more research will be required to determine the nature of any relationships that may exist between the effect of cardiac genetic testing and ICD discharges.

The effect of genetic findings in relation to specific treatments and conditions will also require further investigation. Future research should address capturing an individual's psychological well-being before and after cardiac genetic testing in a larger

sample of patients. The interrelationship between having a positive cardiac genetic finding and the altered perception of risks following an ICD discharge is of growing relevance. Another potential area of investigation could focus on the time from cardiac genetic diagnosis to subsequent future arrhythmias and/or ICD discharge. Such knowledge is crucial to tailoring individual therapies that seek to ameliorate any deleterious effects that may be associated with cardiac testing in this population.

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