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The trajectory of anxiety and depression in people presenting to a cardiac inherited disease service: a longitudinal study

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ABSTRACT

Objective: Little is known about the psychological experiences of individuals being investigated for cardiac inherited diseases (CID). This study aimed to assess the prevalence, trajectory and associated variables of anxiety and depression in this population.

Design: This was a longitudinal study with 116 individuals being investigated for a CID; 85 (73%) completed follow-up.

Main Outcome Measures: Questionnaires were administered at the first appointment, post-disclosure of genetic results, and six-months later. Questionnaires measured demographic and psychological variables. The NZCID Registry provided clinical and genetic information.

Results: Thirty-seven (43%) individuals reported clinical and subclinical levels of anxiety and/or depression at least once. Anxiety and depression at follow up were associated with anxiety ($p < .001$) and depression ($p < .001$) at baseline. Elevated anxiety and depression scores at any point were also associated with more reporting of somatic symptoms ($p < .001$), poorer social support ($p < .01$) and greater intolerance for uncertainty ($p < .001$). There were five different trajectories of anxiety and depression: stable-low, stable-high, increasing, decreasing and fluctuating.

Conclusion: A significant minority of individuals being investigated for a CID experience anxiety and depression. Ongoing screening for anxiety, depression, social support and somatic symptoms could help identify those individuals.

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KEYWORDS
Cardiac inherited diseases; anxiety; depression; clinical and genetic testing

Introduction

People are referred to a cardiac genetic service for many different reasons. Some individuals are referred following the unexplained death of a family member; others may be experiencing minor cardiac symptoms or have experienced a significant cardiac event such as a resuscitated cardiac arrest; and some feel healthy, but are there due

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to another family member's cardiac inherited disease investigation or diagnosis (Earle et al., 2013).

Given the diverse reasons for a referral to the cardiac genetics service it is understandable that people may present in clinic with varied levels of psychological distress. Many individuals attend their first appointment with little to no distress at all, others have a normal level of apprehension in response to the uncertainty of their health, and some can be experiencing clinical levels of psychological distress (Hendriks et al., 2008) with a small proportion in the midst of prolonged grief or post-traumatic stress symptoms (Ingles et al., 2016).

In the last 2 decades the advances in the medical understanding of cardiac inherited diseases has been substantial. There are now clear guidelines for managing many of these conditions (Priori et al., 2013). However, our understanding of the psychological impact of these conditions and any clear guidance around psychologically supporting patients has lagged behind. There are recommendations for integrating psychological screening, assessment and interventions into cardiac care for patients with a congenital heart disease (Warnes et al., 2008), however a similar review and recommendations have not been provided for patients with a cardiac inherited disease.

The lack of psychological guidelines for cardiac inherited diseases may be due to a paucity of research in this population. The research that does exist is relatively heterogeneous, with studies using different timing and methods of psychological measurement. However, what has been consistently found in existing studies is elevated levels of anxiety and depression in a significant minority of individuals living with a cardiac inherited disease. Across five studies, the proportion of patients reporting symptoms of anxiety ranged from 25% to 37% and for depression 14% to 21% (Hamang et al., 2011; Ingles et al., 2013; James et al., 2012; Morgan et al., 2008; Richardson et al., 2018).

There are only two existing studies known to the authors that have looked at psychological distress during the investigation period for these conditions. The first was conducted in the Netherlands with people suspected of having long QT syndrome (Hendriks et al., 2008), it reported no long term psychological distress after the testing process, which supports the general genetic testing literature (Heshka et al., 2008). However a third of participants with an uncertain clinical result still reported significant psychological distress at 18 months (Hendriks et al., 2008). The second study which was conducted in Norway, investigated heart focused anxiety in people suspected of having long QT syndrome or hypertrophic cardiomyopathy (Hamang et al., 2012). This study reported somewhat elevated long term levels of heart focused anxiety, particularly in those with a clinical diagnosis. Given the paucity of research in this area more studies are needed to understand the psychological impact of this investigation phase.

Thus, the current study aimed to understand (1) the psychological impact of engaging with a cardiac genetic service, (2) whether there are common trajectories for individuals in regards to symptoms of anxiety and depression, (3) whether clinical and genetic factors influence the likelihood of anxiety and depression and (4) whether any psychological variables at the first appointment might help to identify individuals who experience ongoing psychological distress.

Materials and methods

Participants

Individuals attending their first appointment at a Cardiac Genetics Clinic in Auckland or Hamilton between 11th July 2017 and 21st August 2018 were recruited for the study. Eligible participants were those being clinically and/or genetically assessed for a cardiac inherited disease, who were 16 years or older and had good English language proficiency. Ethical approval for this study was granted by the Health and Disability Ethics Committee on 11th May 2017 (Ref: 17/NTB/65).

There were 158 people scheduled for appointments during this timeframe. Eight people were excluded because they did not meet eligibility due to needing an interpreter. Of the remaining 150 people scheduled for an appointment, 7 did not attend their appointment and 27 declined to participate (as shown in Table 1). A total of 116 of the 150 eligible people agreed to participate (77% response rate).

Procedure

This was a longitudinal study with three time points. The researcher (CO'D) recruited individuals in the clinic waiting room prior to their first appointment. Potential participants were told about the study, given the information and consent form to read and the researcher returned a few minutes later to answer questions. Those individuals who agreed to take part signed the consent form and were given the first questionnaire (T1) to complete in the waiting room prior to going in to their appointment with the Cardiologist and Genetic Counsellor/Geneticist. In the first questionnaire participants selected how they wanted to complete future questionnaires, either online (via survey monkey), a paper copy (via the post), or completed over the telephone (with the researcher). The second questionnaire (T2) was sent to participants after they had received their genetic results, this was usually around six months (mean 6.25 months, SD 2.75), however it ranged from 2.25 to 13.25 months. The third questionnaire (T3) was sent six months after T2 was returned to the researcher. The second questionnaire was completed by 85 participants, thus 27% of participants dropped out between T1 and T2. The third was completed by 71 participants, with a further 12% dropping out between T2 and T3. The questionnaires measured a number of psychological and demographic variables, as outlined below.

Measures

Depression, anxiety, and somatic symptoms were assessed at each time point with the following measures.

Depression was assessed with the Patient Health Questionnaire – 9 (PHQ-9) (Kroenke et al., 2001). The PHQ-9 has nine symptom items and individuals report how often in the last two weeks they have experienced each of these depression symptoms on a 4-point scale from 0 – 'not at all' to 3 – 'nearly every day. There is a tenth item which asks those individuals reporting symptoms how difficult they had made work, taking care of things at home and getting along with people. Responses ranged

Table 1. Clinical and demographic comparison between participants and non-participants.

	Participants (n = 116)	Non-participants (n = 34)	p
Participated	116 (77)		
Did not participate		34 (23)	
Declined to participate		*27 (18)*	
Did not attend their appointment		*7 (5)*	
Demographic Characteristics:	n (%)	n (%)	
Age: Range (median)	16 – 84 (45)	16 – 79 (46)	ns
Sex: Female	57 (49)	17 (50)	*ns*
Ethnicity:			.003
NZ European	67 (58)	9 (27)	
Māori & Pacific:	27 (24)	11 (33)	
Māori	*22 (19)*	*9 (27)*	
Samoan	*1 (1)*		
Cook Island Maori	*2 (2)*		
Tongan	*2 (2)*	*2 (6)*	
Other:	22 (18)	14 (42)	
Chinese	*3 (2)*		
Indian	*8 (7)*	*6 (18)*	
Other	*11 (9)*	*8 (24)*	
Clinical Characteristics:			
Reason for referral			*.040*
Symptoms of cardiac event not sudden death	19 (17)	9 (27)	
Family member with known CID	46 (40)	6 (18)	
Family member with suspected CID	27 (23)	12 (35)	
Family member with sudden death	22 (19)	5 (15)	
Incidental ECG or Echo abnormality [a]	1 (1)	2 (6)	
Inherited cardiac condition being investigated:			*ns*
Long QT Syndrome	18 (15)	9 (26)	
Hypertrophic Cardiomyopathy	61 (53)	16 (47)	
Sudden Cardiac Death	19 (16)	3 (9)	
Other:	18 (16)	6 (18)	
ARVC	*10 (9)*		
Brugada	*5 (4)*	*4 (12)*	
CPVT	*2 (2)*		
Dilated Cardiomyopathy	*0 (0)*	*1 (3)*	
Resusitated Sudden Cardiac Death	*1 (1)*	*1 (3)*	
Clinic Status			*ns*
Definitely affected	34 (29)	13 (38)	
Probably affected	16 (14)	4 (15)	
Possibly affected	8 (7)	5 (15)	
Unlikely affected	20 (17)	3 (9)	
Not affected	14 (12)	1 (3)	
Unknown (awaiting further clinical tests)	24 (21)	7 (21)	
Genetic Status			*ns*
Genotype Positive	25 (22)	6 (18)	
Testing uninformative	4 (3)	2 (6)	
Unclassified variant	13 (11)	2 (6)	
Genotype Negative	13 (11)	1 (3)	
Not tested/unknown	61 (53)	23 (68)	
Proband			*.001*
True	26 (22)	18 (53)	
False	90 (78)	16 (47)	

[a]This reason was not included in the chi square analysis due to the low frequency.

from 0 – 'Not difficult at all' to 3 – 'Extremely difficult'. The PHQ-9 is widely used and has been validated against clinical interviews, the following cut-off scores: 0 – 4 'normal'; 5 – 9 'mild cases'; 10 – 14 'moderate cases'; 15 – 19 'moderately severe cases'; and $\geq$20 'severe cases' have demonstrated a sensitivity of 88% and specificity of 88% (Kroenke et al., 2001).

The Generalised Anxiety Disorder – 7 (GAD-7) was used to measure anxiety (Spitzer et al., 2006). The GAD-7 follows the same structure as the PHQ-9, with seven items that assess a symptom of anxiety. Individuals respond based on how often they have experienced each symptom during the last two weeks. The eighth item asks how difficult life has been for those reporting symptoms. The GAD-7 has been validated against clinical interviews, the following cut off scores were established: 0 – 4 'normal'; 5 – 9 'mild cases; '10 – 14 'moderate cases'; 15 – 21 'severe cases' with an 89% sensitivity and 82% specificity (Spitzer et al., 2006).

Somatic symptoms were measured with the Physical Health Questionnaire – 15 (PHQ-15) (Kroenke et al., 2002). Individuals were asked to rate how bothered they had been in the last four weeks by a list of somatic symptoms. There are three response options: 0 – 'not bothered at all'; 1 – 'bothered a little'; or 2 – 'bothered a lot'. Scores range from 0 to 30 and scores of 5, 10, and 15 signify low, medium and high somatic symptom severity respectively (Kroenke et al., 2002).

The STOP-D (Young et al., 2007) is a single item screener that has been validated with a cardiac population (Young et al., 2015). The screener has five items which measure depression, anxiety, stress, anger and social support. Users are asked how much they have been bothered by each item over the previous two weeks and they respond on a 10-point scale 0 – 'Not at all' to 9 – 'severely'. The social support item taken at baseline was the only item used in the current analyses, it asks how much they had been bothered by: 'Not having the social support you feel you need?'. Lower scores indicate better perceived social support.

The baseline questionnaire also contained a measure assessing an individual's intolerance for uncertainty. The Intolerance for Uncertainty Scale – 12 (IUS-12) has 12 items (Carleton et al., 2007); seven items measure prospective anxiety e.g. 'I always want to know what the future has in store for me' and five items measure inhibitory anxiety e.g. 'Uncertainty keeps me from living a full life'. Participants respond on a 5-point scale from 1 – 'Not at all characteristic of me' to 5 – 'Entirely characteristic of me'. The IUS-12 has very good internal consistency ($\alpha = .91$), performing as well as the original 27 item IUS (Carleton et al., 2007).

Participants completed demographic questions at baseline including age, gender, ethnicity, education and employment status. Clinical variables, including type of condition being investigated, clinical status, genetic status, and whether participants were a proband (an individual who is the starting point for the genetic study of a family) or family member, were collected from the Cardiac Inherited Diseases Group registry (Earle et al., 2019).

Statistical analysis

Missing data were left out of analysis on a case by case basis. There were 31 individuals excluded from analyses because they only completed the baseline measures. For those individuals remaining the missing data for the outcome variables are outlined in Table 2. There was one individual missing data for anxiety and depression at all time points, so they were excluded from the trajectory and group analyses. For those individuals missing one time point for anxiety and/or depression we assumed no change

Table 2. Frequency table of depression and anxiety scores.

	PHQ – 9 Depression			GAD – 7 Anxiety		
Outcome categories	T1 n (%)	T2 n (%)	T3 n (%)	T1 n (%)	T2 n (%)	T3 n (%)
Normal (0–4)	53 (62.4)	61 (71.8)	46 (64.8)	52 (64.2)	69 (81.1)	48 (69.6)
Sub-clinical (5–9)	18 (22.8)	17 (20)	16 (22.5)	23 (28.4)	10 (11.8)	16 (23.2)
Clinical (10+)	8 (10.1)	7 (8.2)	9 (12.7)	6 (7.4)	6 (7.1)	5 (7.2)
Missing	6	0	14	4	0	16
Total	85	85	85	85	85	85

in score from the second-time point. For the depression scale, there were five cases missing the first-time point, and 13 cases missing the third-time point. For the anxiety scale, there were three cases missing the first-time point, and 15 cases missing the third-time point. The second-time point had no missing cases for either scale.

Analyses were conducted on SPSS version 14. Data was not normally distributed thus non-parametric tests were used and medians reported. The initial analyses used Spearman correlations to assess associations between age and baseline psychological measures with T3 anxiety and depression scores. Mann-Whitney tests were performed to assess whether anxiety and depression differed between gender, proband status and cardiac device status. Kruskal Wallis Tests were performed with ethnicity, education level, condition being investigated, clinical and genetic status, patient type, reason for referral and whether a family member had died.

The trajectories were established by converting scores from the PHQ-9 (depression) and GAD-7 (anxiety) into three discrete groups based on cut-off scores from these scales. Participants were assigned a 1 if their scores fell in the 'normal' category, a 2 for the 'subclinical' category and 3 for the 'clinical' category. Then trajectories were established based on what category participants fell in at each time point. For example, the participants who were assigned a 1 in all three time points were categorised in the 'Scores remain within normal range' group. In the 'Starts elevated but improves with time' group, participants were included if they had been assigned a 2 or a 3 initially and their scores then improved over the remaining two time points with their final time point being in the 'normal' category. In the 'Started normal ended up elevated' group, participants were all assigned a 1 (normal) in their first time point and then a 2 (sub-clinical) or 3 (clinical) in the third-time point (with the majority assigned a 2 or 3 at the second-time point as well). Then for each group the median scores from each time point were then plotted on the graph.

Further analyses were conducted comparing those who reported no symptoms of anxiety or depression at all across the three time points (Group 1) and those who reported at least one instance of symptoms of anxiety and/or depression (Group 2). Chi-squared tests were performed to compare these two groups based on reason for referral, clinical and genetic status, type of condition being investigated, proband status, gender, ethnicity, education level and a sudden death in the family. These groups were further compared using Mann Whitney tests for age, baseline somatic symptoms, patient perceived likelihood and chance of being diagnosed with the condition and the two intolerance of uncertainty subscales. A significance level of .05 was maintained apart from when post-hoc tests were conducted during which a Bonferroni correction was made.

Results

Participants' demographic and clinical variables are outlined in Table 1. Participants significantly differed to non-participants (those who did not take part in the research), by ethnicity, proband status and reason for referral. Europeans were over represented in the participant group ($p < .01$) and the proportion of probands was significantly higher in the non-participant group ($p < .01$). In addition, there were fewer non-participants than expected in the group of people referred for having a family member with a known diagnosis of a cardiac inherited disease ($p < .04$). Participants and non-participants did not significantly differ based on age, gender, clinical status or type of cardiac inherited disease. They also did not differ on genetic status; however, this variable did not meet the chi-squared test assumption of expected frequencies, which causes a loss in power for this analysis, so it should be interpreted with caution.

Rates of clinical, sub-clinical and normal levels of anxiety and depression across the three time points are reported in Table 2. When the whole sample (n = 116) was included at baseline, the clinical levels of anxiety (10%) and depression (16%) were slightly higher than when T2 drop outs were excluded at baseline (n = 85). A Friedman's ANOVA was performed to assess average changes in anxiety and depression scores across the three time points and there was no significant change in average scores for depression χ^2 (2) = 2.48, $p = .29$ or anxiety χ^2 (2) = 2.73, $p = .26$.

Bivariate correlational analyses showed that the final anxiety and depression scores (T3) were not associated with any clinical and demographic factors (Table 3). They were however significantly associated with the psychological measures administered at T1. The psychological variable most strongly associated with depression scores at T3 was depression scores at T1, $r_s = .62$, $p < .001$. The variable most strongly associated with anxiety scores at T3 was anxiety scores at T1, $r_s = .60$, $p < .001$.

Given baseline levels of anxiety and depression appear to be important and the clinical and demographic variables were unrelated, we looked at the group as a whole to determine any consistent patterns of anxiety and depression over the 12–18-month period. As demonstrated in Figure 1a there were five trajectories of anxiety scores and Figure 1b shows six trajectories for depression scores. For both psychological outcomes just over half the participants remained in the normal range across all three time points.

In order to determine what might be unique to the individuals who reported no anxiety or depression at all in the 12-month period, participants were divided into two groups. Those who had not reported elevated levels of anxiety or depression at all in the study period (Group 1 n = 48) and those who had (Group 2 n = 36). Chi squared tests were performed and no significant differences were found for reason for referral χ^2 (3) = 2.42, $p = .49$; genetic status χ^2 (3) = 2.89, $p = .41$; type of condition being investigated (channelopathy, cardiomyopathy or sudden cardiac death) χ^2 (1) = 1.67, $p = .43$; proband versus family member χ^2 (1) = 2.17, $p = .14$; gender χ^2 (1) = .931, $p = .34$; ethnicity χ^2 (2) = 2.13, $p = .35$; education level χ^2 (2) = 3.99, $p = .14$; and death/s in the family due to a cardiac inherited disease χ^2 (2) = 3.74, $p = .15$. The chi squared test for clinical status χ^2 (3) = 7.31, $p = .06$ trended towards significant, and as Table 4 demonstrates there was a significant difference between groups for the

Table 3. Bivariate analyses (Spearman correlations, Mann-Whitney and Kruskal-Wallis tests) of the relationship between clinical, demographic and psychological variables with anxiety (GAD-7) and depression (PHQ-9) scores.

Baseline measures	T3 PHQ-9 - Depression		T3 GAD-7 - Anxiety	
Spearman Correlations	r_s	*P*	r_s	*P*
Age	-.05	.71	-.22	.07
T1 PHQ-9 – Depression	.62	.00	.45	.00
T1 GAD-7 – Anxiety	.41	.00	.60	.00
T1 PHQ-15 – Somatic Symptoms	.60	.00	.51	.00
T1 STOP-D – perceived social support	.25	.04	.35	.00
Intolerance for uncertainty – prospective	.40	.00	.40	.00
Intolerance for uncertainty – inhibitory	.44	.00	.41	.00
Likelihood of getting diagnosed	.20	.09	.18	.13
Chance of getting diagnosed	.22	.07	.25	.04
Time taken for genetic results to be disclosed	.06	.63	.13	.30
Mann Whitney Tests	**Test Statistic *U (z)***	**Significance *p***	**Test Statistic *U (z)***	**Significance *p***
Gender	608.5 (-.25)	.80	574 (-.26)	.80
Proband versus family member	442 (-.47)	.64	520 (.85)	.39
Cardiac device	235.5 (.84)	.41	244.5 (1.21)	.24
Kruskal Wallis Tests	***(df) H***	***p***	***(df) H***	***p***
Ethnicity (Non-European)	(2) .52	.77	(2) .34	.84
Education level	(2) 4.04	.13	(2) 3.30	.19
Cardiomyopathy, Channelopathy or sudden death being investigated	(2) .35	.84	(2) .11	.95
Clinical status	(3) 2.16	.54	(3) 6.01	.11
Genetic status	(3) 5.51	.14	(3) 6.38	.10
Patient type				
Reason for referral	(3) .85	.84	(3) 4.16	.25
Death of a family member	(2) 3.92	.14	(2) 4.58	.10

definitely affected status. Those who were definitely affected were more likely to have experienced anxiety or depression.

A Mann Whitney test was performed to determine if age differed between these two groups, and Group 1 (median = 50.5) did not significantly differ to Group 2 (median = 45.5), U = 744.5, z = −1.16, $p = .24$. When anxiety and depression was analysed separately however, age significantly differed for anxiety alone. In this instance those individuals who did not experience anxiety or depression (median = 51.5) were significantly older than those who did (median = 42.5), U = 594, z = −2.44, $p < .05$. Separate analyses for all other variables did not differ to the combined results as described above.

Mann Whitney tests were also conducted on other baseline measures. Those without depression or anxiety reported significantly fewer somatic symptoms at baseline (median = 2), U = 1,247, z = 5.05, $p < .001$, than those who did (median = 7). They

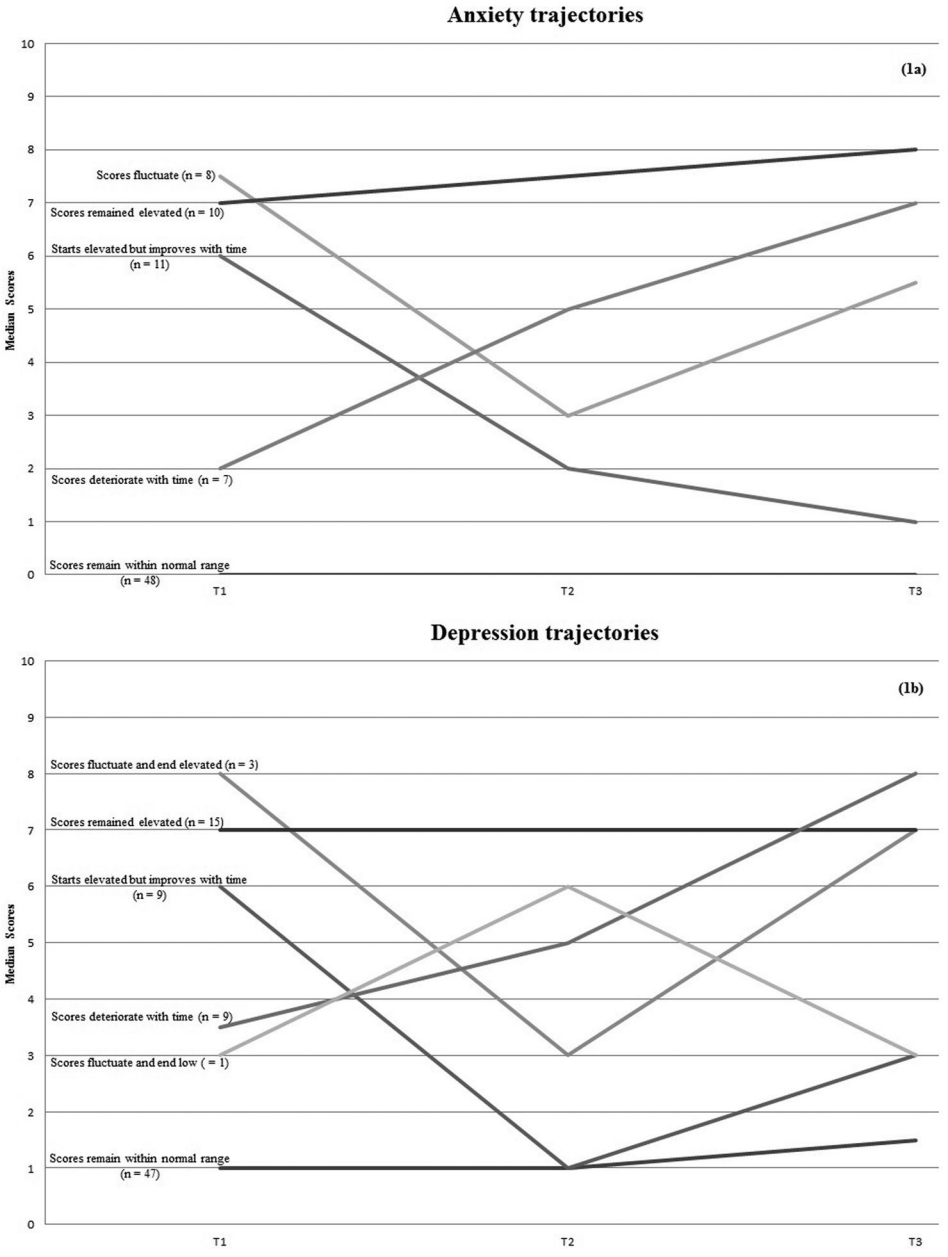

Figure 1. Line graphs showing the different trajectories participants can go on based on anxiety (1a) and depression scores (1b).

also reported significantly less avoidance of uncertainty at baseline (median = 11.5), $U = 1{,}226.5$, $z = 3.18$, $p < .01$, than those who experienced depression or anxiety (median = 16) and a lower tendency to feel unable to act in the face of uncertainty (median = 5.5 versus 8), $U = 1{,}318$, $z = 4.05$, $p < .001$. Finally, those who did not experience depression or anxiety reported significantly better perceived social support, $U = 1{,}148.5$, $z = 3.31$, $p < .01$ than those who did. Both groups had a median of zero

Table 4. Chi square test comparing Group 1 (those who had not reported elevated levels of anxiety or depression at all) and Group 2 (those who had reported at least one instance of elevated levels of anxiety or depression), based on clinical status.

	Anxiety and Depression Symptoms		
Clinical Status	Group 1 No symptoms at all	Group 2 Some symptoms	Total
Definitely Affected	8_a	20_b	28
Probably and Possibly Affected	10_a	7_a	17
Unlikely and not Affected	10_a	14_a	24
Unknown	10_a	5_a	15
Total	38	46	84

Each subscript letter denotes a subset of Group 1 versus Group 2 whose column proportions do not differ significantly from each other at the .05 level.

indicating most participants felt they had good social support. However, the range for Group 1 was $0-5$ and for Group 2 it was $0-8$.

Discussion

Just over half the individuals engaging with the cardiac genetic service for the investigations of a cardiac inherited disease maintained scores within the 'normal' range of anxiety and depression, for approximately 12–18 months following their first appointment. The remaining 44% either had 'elevated' scores for all the time points during the 12 months, or fluctuated between 'normal' and 'elevated'. At the final time point of the study, 10/71 (14%) individuals reported clinical levels of anxiety and/or depression and six of these individuals reported 'elevated' scores at every time point. The strongest association with final anxiety and depression scores were initial anxiety and depression scores. Other significant baseline predictors were higher intolerance of uncertainty, lower perceived social support, and more somatic symptoms. Younger individuals going through the service were more likely to experience anxiety over this period compared to older people. In addition, those definitely affected with a cardiac inherited disease were more likely to experience anxiety and depression at any time point.

Overall these findings broaden our understanding of the psychological experiences of this patient population. The clinical levels of anxiety and depression after 12–18 months in this study were 7.2% (95% CI [2.4, 16.1]) and 12.7% (95% CI [6, 22.7]) respectively, which are similar to the prevalence rates reported in individuals living with a cardiac inherited disease (10% and 13% respectively) (O'Donovan et al., 2019). The prevalence rates of generalised anxiety and clinical depression in the general New Zealand population are 2% (95% CI [1.7, 2.3]) and 5.7% (95% CI [5.2, 6.2]) respectively (Wells et al., 2006). Based on the confidence intervals, these percentages suggest those individuals going through testing for a cardiac inherited disease are at significantly greater risk for anxiety than the general population. There is a slight overlap between confidence intervals for depression, which still suggests higher depression in the testing group than the general population (Field, 2018).

Hendriks and colleagues (2008) combined anxiety and depression scores in their study of patients with long QT syndrome and reported 8% of participants experienced clinical levels of psychological distress 18 months after their first appointment. When

results from the current study are similarly combined, the rate of anxiety and depression (12%) is greater. Hamang et al. (2012) study used a disease specific measure of anxiety that does not have established cut-off thresholds, thus direct comparisons with this study could not be conducted. However, the authors did report 'somewhat' elevated mean scores at 12 months, similar to those found in patients following aortic valve replacement.

It is not immediately clear why Hendriks et al. (2008) found no elevated levels of anxiety and depression at 18 months and the current study and Hamang et al. (2012) did. It could be because Hendriks et al's study (2008) only included long QT syndrome and investigations for long QT syndrome cause less distress. However, Hamang et al. (2011) found no difference at baseline between people being investigated for hypertrophic cardiomyopathy and long QT syndrome regarding distress. Further research is needed to determine if differences do exist between conditions in the experience of psychological distress during this investigation period.

Understanding how people cope psychologically as they move through a health care process provides important information for the most efficient provision of support. Stanton and colleagues (2015) followed 460 women diagnosed with breast cancer for 16 months and found four common trajectories of depressive symptoms. Almost 40% of the group had consistently high levels of depressive symptoms for the full 16 months. A similar proportion either had consistently low or very low levels, and the final group (20%) experienced high levels of depression initially, but recovered with time. The tentative trajectories observed in this study followed similar patterns with the addition of two groups, one that starts with 'normal' levels of anxiety and depression at the initial appointment but becomes elevated with time. The other trajectory begins with elevated levels of anxiety and depression at the initial appointment, these then improve after the disclosure of the genetic results, but become elevated again six months later. These additional trajectories may illustrate the difference between cardiac inherited diseases and breast cancer. In the case of breast cancer, most patients are moving away from the disease and treatment over 16 months, whereas cardiac inherited diseases are lifelong conditions. With time people may develop symptoms of anxiety and depression due to the ongoing burden of living with a chronic condition.

The current study suggests that screening for psychological distress should not just be conducted at initial appointments but should be ongoing. Identifying cases of distress early and intervening could possibly help reduce the development of more significant maladjustment (Hendriks et al., 2008). Further research is needed to confirm an appropriate screening tool, but a simple screen for anxiety, depression, somatic symptoms and social support would likely be sufficient. The STOP-D may be a good candidate as it is a succinct screener that has been used to identify inpatients on a cardiac ward experiencing psychological distress (Young et al., 2015).

Hendriks and colleagues (2008) reported that just under 10% of participants were seen by a psychologist during their study. The current study was based in a service that did not have a psychologist available to patients, however 10% of participants reported they had sought support from a psychological professional and all of them reported that this had been helpful. Of these participants, over half had improvements

in their anxiety and/or depression scores between at least one time period. Although this may have reduced the levels of distress reported in both studies, it is promising in terms of the potential efficacy of psychological intervention.

Finally, this study highlights that some people, going through this process, who had negative or inconclusive test results, were still struggling psychologically 12–18 months after their first appointment. This raises the question of what responsibility a cardiac genetic service should have for those individuals released from immediate medical follow up, but who have potentially been psychologically impacted by the process. Two thirds of those individuals with clinical levels of anxiety and/or depression at follow-up were unaffected non-carriers and individuals with an unknown clinical status (due to long wait times for clinical tests), whose deceased family member had inconclusive genetic results. None of these individuals had any follow up appointments planned with the cardiac genetic service or access to free specialty psychological support. Within a healthcare system that provides these clinical and genetic investigations free of charge, these findings warrant further discussion around the obligation that same healthcare system may have for also making available specialist psychological care for those impacted by that process.

Limitations

Although the response rate for this study was high for the baseline measures (77%), 31 people dropped out of the research before the second time point and another 14 before the third-time point, reducing the response rate overall to 47%. Only two participants wrote asking to be removed from the study, the remaining individuals did not respond to correspondence from the researcher, so it is unclear why they withdrew from the study. This smaller response rate reduces the reliability of the results when generalised beyond the study participants. In addition, given the small sample size and the number of groups in the explorative analyses, statistical power is limited.

There was also large variability in the timing of the second and third measures due to the variability in the time it took for genetic results to be returned. For those individuals having genetic tests for a specific mutation (when another family has received a positive genetic result), their results were returned much faster than those without a specific mutation identified yet (generally probands). However, the time taken to return genetic results was not related to anxiety or depression as reported in Table 3.

Finally, the first measure is not an exact baseline measure of anxiety and depression as people would have been aware of why they were being referred to the cardiac genetics service. It was however an improvement on Hendriks and colleagues' (2008) baseline measure in that their participants attended their appointment with the cardiologist and genetic counsellor prior to completing the baseline measures. Whereas in the current study 73% of participants completed their baseline questionnaire prior to going in to their appointment.

Implications of findings

This longitudinal study suggests that ongoing screening for anxiety and depression in this population could help identify individuals who may need psychological support.

However, at this stage there is no empirical evidence around what that psychological support should be. There is evidence that cognitive behavioural therapy can reduce anxiety and depression and improve quality of life in people living with an implantable cardioverter defibrillator (Pedersen et al., 2007). Future research could investigate whether a similar intervention could benefit people with a cardiac inherited disease.

Conclusion

The first year to 18 months after engaging with a cardiac genetics service for the investigations of a cardiac inherited disease can be difficult for a minority of individuals. Fourteen percent of participants reported clinical levels of anxiety and/or depression at 12–18 months and 16% reported subclinical levels. Elevated scores of anxiety and depression at the first appointment were most strongly associated with follow-up scores of anxiety and depression. This study supports the ongoing screening for anxiety and depression as a way of identifying those individuals likely to experience enduring psychological distress. People who are younger, have a definitely affected clinical status, report more somatic symptoms and tolerate uncertainty poorly are most likely to experience at least some psychological distress during that time period.

Author contributions

This study is part of a PhD being completed by COD who is being supervised by JS and EB. COD designed the study with support from EB, and JS. COD collected the data and performed analyses with support from EB. Interpretation of the results was done by COD with guidance from JS and EB. COD drafted the manuscript which was critically revised by all the other authors. All authors have approved the final version to be published.

Data sharing

As part of this study we collected information on anxiety and depression and risk perceptions and plan to publish papers on these topics. Data was collected with the ethics requirement that patients' data is confidential and will not be shared. However any questions should be directed to the corresponding author.

Disclosure statement

There authors have no conflicts of interest to declare.

Ethics approval

This study was approved by the Health and Disability Ethics Committee New Zealand and local area health board: number: 17/NTB/65.

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