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ORIGINAL ARTICLE

Adjustment and Coping Mechanisms for Individuals with Genetic Aortic Disorders

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Background Advances in diagnosis and management of Genetic Aortic (GA) Disorders have improved prognosis for affected individuals, yet many do not adhere to key management recommendations, and some may experience clinically significant levels of psychological distress. These issues are often not communicated to treating clinicians. Poor adjustment and coping may adversely impact on prognosis, but little is known about the processes contributing to negative outcomes. This study investigated adjustment to GA disorders to determine which processes facilitated or hindered good adherence and psychological outcomes.

Methods Semi-structured interviews involving 21 individuals (12 M, 9 F; age 19-62 years) with a GA Disorder and psychosocial measures of depression/stress/anxiety (DASS), coping (COPE) and involvement in treatment (CPS) were used. Qualitative data were analysed using grounded theory and a model of adjustment was developed.

Results Although most participants adhered to physician management recommendations and experienced minimal emotional distress, a subset reported poor adherence and/or sub/clinical levels of depression/anxiety/stress (29%). Dysfunctional coping mechanisms were infrequent, however 22% participants reported 'little or no' acceptance and 43% avoided life planning in response to a diagnosis of GA disorder. Interviews revealed an overarching theme: *Negotiating perception of self and GA disorder*, supported by five sub-themes: *Restrictions upon Lifestyle, Destabilisation, Future, Support, and Unmet Needs*. Accepting restrictions and having support were conducive to better adherence, whilst destabilisation and loss of control had a negative impact. A model of adjustment is proposed to explain how patients reached one of four outcomes relating to psychological distress and adherence to physician recommendations. The central tenet of the model is founded on how realistically patients appraise their vulnerability to GA threat and whether they are able to integrate their perceptions of illness with their sense of self-identity.

Conclusions This study indicates that individuals with GA are at risk of experiencing psychosocial distress and coping difficulties, even years after diagnosis. Key factors likely to be associated with impaired coping among GA patients include inability to integrate the illness into one's identity/life, or to follow physician recommendations. Potential unmet needs were identified, including the provision of more relevant information and opportunities for peer support. These findings may also be applicable to other inherited cardiac disorders.

Keywords Aorta • Genetics • Marfan syndrome • Patient adherence • Psychological adjustment.

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Introduction

Inherited heart disease is an increasingly important part of modern cardiology practice. The family of Genetic Aortic (GA) disorders includes Loeys-Dietz syndrome (LDS), Marfan syndrome (MFS), familial Thoracic Aortic Aneurysm and Dissection (TAAD) and bicuspid valve aneurysm syndrome (BAV). The incidence of these disorders ranges from 1 in 5000 for MFS to 1 in 300 for BAV in the general population. Individuals with GA disorders are vulnerable to aortic dissection and sudden death [1,2], with mean age of dissection in MFS being 36 years and TAAD being 46 years (Jeremy et al., manuscript under review). Management of GA disorder requires lifetime medical surveillance, lifestyle alterations, pharmaceutical treatment and often multiple surgical procedures [1,3].

Although advances in diagnosis and management have considerably improved prognosis for affected individuals [4], complex treatment decisions are often required, with physical and psychological consequences. For individuals with external physical features, including those with MFS and LDS, diagnosis is more likely to occur at younger age and to adversely impact everyday functioning, with increased stigma and potential isolation from peers [5–8]. For others, diagnosis may be delayed until a family member is identified with the disorder, an abnormal echocardiogram is observed, or a life-threatening complication occurs [1,8]. Uncertainties about optimal management of GA disorders, including limitations on evidence for medication benefit and surgical guidelines, may impede informed decision-making and adversely impact upon patient adjustment to the diagnosis [5,9]. Life-threatening complications can require emergency intervention without prior warning, with increased risk of subsequent morbidity and potential loss of physical functioning [1,3,9]. Accepting and complying with the restrictions required to manage the disorder may also be difficult if significant and permanent lifestyle changes are necessary [10], such as restriction of exercise.

The evidence base on coping and adjustment to diagnosis of GA disorder is contradictory. Some studies indicate the diagnosis can have a significant negative impact on psychological wellbeing, physical functioning and overall quality of life [10–13], whilst other findings are not so consistent [14–16]. Some individuals may engage in risky non-adherent behaviours, such as high impact sport, drug use and lack of adherence to recommended medications [12,14,15,17,18]. Although these findings identify negative outcomes for physical and psychological wellbeing, they provide little insight into the processes contributing to these outcomes.

Several models of adjustment to illness have been described, with the majority focussing on the key construct of coping and attributions (ways in which people appraise life events) [19]. Problem-focussed coping and optimistic, but realistic, appraisals are generally seen to promote positive adjustment, whereas avoidance, emotion-focussed coping and pessimistic attitudes are seen to impede adjustment. Apart from coping and attributions, the way in which an illness may change person's view of themselves / their identity, has been implicated in affecting psychological outcomes, and also the degree to which medical interventions are accepted or rejected (i.e. adherence) [20–24].

This study, therefore, investigated specific relationships between coping, attributions and perception of self for individuals with a GA disorder, and how these factors influence psychological adjustment and adherence to management recommendations. The study sought to develop an integrated model of adjustment for GA disorders and to identify: (i) the facilitators of, and barriers to, management adherence and psychological wellbeing; (ii) coping strategies used by people with GA disorders; and iii) the impact of illness-related changes in self-perception on adjustment.

Methods

This mixed method (qualitative and quantitative) study involved a cross-sectional sample of adults with GA disorder. It was designed to describe reactions to the diagnosis of GA disorder, experiences living with GA conditions, and to identify factors associated with adverse coping and adjustment reactions.

Participants

In order to recruit a sample that was representative of the varied experience of individuals with GA disorders, the research team purposefully sampled from individuals with different aortopathies (i.e. MFS, LDS and TAAD). Participants were recruited through a specialist clinic at a main tertiary hospital. All participants were enrolled in a clinical surveillance program for GA disorders, which included at least annual clinical and cardiac imaging review in the clinic. Participants were eligible if they were over 18 years of age, and had a confirmed diagnosis of one of the three GA disorders at least six months prior to participation in the study. Individuals were not eligible if they were clinically unstable or about to undergo major surgery. Exclusion criteria were insufficient English, and the presence of significant intellectual impairment or a major psychiatric condition, apart from anxiety and depression.

Study Protocol

The study protocol and participation by individuals was approved by the hospital Human Research Ethics Committee (Protocol X10-0246). Eligible patients were identified over a six months period by the attending cardiologist (RJ), according to age at diagnosis, gender balance, experience with GA disorder and presence or absence of previous adverse clinical events. Potential participants were introduced to the study by the cardiologist during annual review and, if interested in participating, were: i) given a study information pack by RJ (n = 16), or ii) mailed a study information pack directly by the researcher (EC) (n = 22). Information packs contained an invitation letter, participant information sheet, consent form, contact preference form, questionnaire and reply paid envelope. Those wishing to participate were asked to return the