

The psychosocial impact of personalised external aortic root support surgery in Marfan syndrome

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Abstract

Background: Marfan syndrome is a genetic disorder which can result in life-threatening heart defects. A new form of surgery has recently been established and anecdotal reports suggest that it may have a positive psychological impact on its recipients.

Aims: To explore the psychosocial impact of undergoing personalized aortic root support (PEARS) surgery and to examine whether psychometrics are a useful tool in capturing psychological change in future evaluations of PEARS surgery.

Method: Nine people who had PEARS surgery took part in focus groups, which were subsequently thematically analysed. Anxiety and depression were assessed by the Hospital Anxiety and Depression Scale (HADS) and social and occupational functioning was assessed by the Work and Social Adjustment Scale (WSAS).

Results: Participants were generally well-adjusted and positive about the surgery. Concerns were raised over child bearing and adherence to medication regimens. Anxiety significantly decreased from before surgery to after surgery.

Conclusions: People with Marfan syndrome generally lead normal lives, on which the condition has very little impact. However, the impact for female participants was slightly greater due to the implications for child bearing. The requirement for surgery did cause some anxiety on occasion, but most participants approached surgery pragmatically and positively. The study also highlighted the way in which virtually all participants perceived themselves as healthy, normal individuals, and the effects that this could have on their behaviour. The psychometrics used in this study were deemed to be useful methods of capturing change. Future research should examine levels of anxiety and depression in Marfan syndrome and interventions for reducing anxiety about child bearing.

1.0 Introduction

Marfan syndrome is a genetic disorder which affects multiple organ systems, including the cardiovascular system (Loeys et al., 2010). These effects can be life-threatening and usually require corrective surgery (Treasure & Pepper, 2011). At present, several surgical options exist, including a novel operation known as personalised external aortic root support (PEARS) surgery (Treasure, Pepper, Golesworthy, Mohiaddin, & Anderson, 2012). This new approach to treating cardiovascular risk in Marfan syndrome did not come from a surgeon, but from a pioneering individual with the condition himself. A research programme into the efficacy of this new form of surgery is currently underway, with a comparative evaluation of the different surgical options and their timings planned for the near future. This project is part of the research programme and serves to act as a pilot study into the inclusion of psychological measures as patient reported outcomes (PROs) in future research.

This study is a qualitative analysis of the psychological impact of living with Marfan syndrome and undergoing prophylactic personalised external aortic root support surgery. This thesis will first give a brief overview of the aetiology, presentation, and treatment of Marfan syndrome. It will then review the literature on the psychosocial impact of living with congenital heart disease more generally, and Marfan syndrome specifically. The results of a thematic analysis of patient experiences will then be presented, alongside quantitative measures of anxiety, depression, and social and occupational functioning. Finally, the implications of the findings for future service provision and research will be discussed.

1.1 Marfan Syndrome

Marfan syndrome is a genetic condition in which there is a dominant inheritance of a deficiency in the structural protein fibrillin, caused by mutations in the FBN1 gene (Dietz, et al., 1991). Fibrillin is an essential component for the formation of the elastic fibres within connective tissue throughout the body (Handford, 2000). This can manifest in an array of symptoms across the skeletal, ocular, cardiovascular, respiratory, and central nervous systems (Loeys, et al., 2010). The most visible effects of Marfan syndrome are on the skeletal system. People with Marfan syndrome are often above average height and may have long, slender limbs and digits due to elongation of the skeleton (Loeys, et al., 2010). The presence of striae (stretch marks) is also common. Other symptoms can include hypermobility of the

joints, visual problems, and back pain as a result of the weakening of the connective tissue surrounding the spinal cord, which is known as dural ectasia.

The most serious aspect of Marfan syndrome is the effect on the cardiovascular system. People with Marfan syndrome are prone to aortic root aneurysm – that is, an enlargement of the largest blood vessel in the body, the aorta (Milewicz, Dietz, & Miller, 2005). If the aortic root increases beyond a certain size, there is a risk of that it will rupture and cause a catastrophic internal haemorrhage. Without immediate treatment, the outcome is usually death (Murdoch, Walker, Halpern, Kuzma, & McKusick, 1972). Therefore, Marfan syndrome is potentially life-threatening if left untreated.

As a result of this cardiovascular risk, routine measurement of the aortic root is recommended (NICE, 2011). This is assessed by echocardiogram and usually takes place on an annual basis, although the frequency can be increased if there is cause for concern (Milewicz et al., 2005). Common reasons for more frequent assessment include an aortic root measurement nearing the cut-off point of 5 cm, and pregnancy. Once the aortic root measurement approaches or exceeds 5 cm, then surgery is recommended, as at this point the risk of rupture is perceived to be greater than the risks of surgery (Treasure & Pepper, 2011).

At present, three surgical options exist. The Bentall method is a long-established procedure which involves removing aortic tissue and replacing it with a composite mechanical or tissue valve (Bentall & De Bono, 1968). In order to do this, cardiopulmonary bypass is essential. Valve-sparing operations, as the name suggests, replace the aortic root but aim to leave the aortic valve intact (David & Feindel, 1992). Personalised external aortic root support surgery involves placing an external stent around the aortic root (Treasure & Pepper, 2011). Each stent is individually crafted through computer aided design (CAD) modelling based on magnetic resonance imaging (MRI) scans of the individual's aortic root. This operation takes less time than the other forms of surgery and does not require cardiopulmonary bypass.

All these forms of surgery minimise the risk of aortic dissection but they are not without problems. The Bentall method requires recipients to take anti-coagulation medication for the rest of their lives, increasing the risk of bleeding, whilst 50% of people who have had valve-sparing surgery will require another operation at some point in their lives (Treasure &

Pepper, 2011). Anti-coagulation therapy can be unpopular with people with Marfan syndrome (Allen & Pepper, 2010; Golesworthy, 2014), which can lead to some people avoiding the Bentall procedure as a consequence.

Personalised external aortic root support surgery (PEARS) does not require anti-coagulation medication and current evidence shows that recipients of PEARS have not required further surgery in the short term (Treasure, et al., 2012), although it is too soon in the course of this relatively new surgery to say whether this will remain the case in the long term. As a result of this seemingly positive outcome, anecdotal evidence suggests that this form of surgery may have a positive impact on the psychological state of its recipients (NICE, 2011). However, currently no research has been conducted to investigate this.

1.2 Psychological impact of congenital heart conditions

1.2.1 Relevance to Marfan Syndrome

The impact of congenital heart conditions on psychosocial functioning and quality of life has become an increasingly popular research topic. Technological and surgical advances have meant that more people born with heart defects are surviving into adulthood than ever before and consequently the number of people living with congenital heart disease has increased greatly (Perloff, 1991). As a result, measuring outcomes in terms of morbidity and mortality does not provide a satisfactory account of the lived experience of this population.

Marfan syndrome is defined as a syndrome because the fibrillin mutation can cause an array of symptoms, which include congenital heart defects. Although Marfan syndrome accounts for a greater range of symptoms than congenital heart defects alone, findings from research based on the broader definition of congenital heart disease are likely to have some relevance for the Marfan syndrome population due to the common effects of living with a heart condition. In both cases heart defects are present from birth rather than acquired, and as such regular monitoring is required from a young age. Additionally, many heart conditions can place limitations on social activities and life choices. Therefore, it is pertinent to examine the literature on psychosocial functioning in congenital heart disease more generally in order to compare the findings with those in Marfan syndrome specifically.

1.2.2 Psychological impact

The findings on the psychological impact of congenital heart disease are mixed. In a longitudinal follow-up study, there was no difference in the prevalence of psychological problems between those who had undergone surgery for congenital heart disease before the age of 15 and the general population (Utens, Versluis-Den Bieman, Verhulst, Meijboom, Erdman, & Hess, 1998). Similarly, adults with congenital heart disease showed lower levels of psychopathology than a comparison group of orthopaedic outpatients in a British study (Cox, Lewis, Stuart & Murphy, 2002). In a later follow-up study of the same Dutch cohort as Utens et al. (1998), participants who had congenital heart disease were found to be significantly more emotionally well-adjusted than a reference group (Van Rijen, et al., 2003).

However, other studies have reported negative psychosocial effects of living with congenital heart disease. Brandhagen, Feldt, & Williams (1991) reported that adults with congenital heart disease had a greater level of psychological distress than the general population, as assessed by the Symptom Checklist 90-R. In a study of 280 adult congenital heart disease patients, loneliness, fear of negative evaluation, and perceived health status were found to be predictors of depressive symptoms (Kovacs, et al., 2009). Loneliness and fear of negative evaluation were also significant correlates of anxiety symptoms.

Furthermore, a small study of seven participants reported ongoing negative psychological effects of congenital heart disease (Cornett & Simms, 2014). In semi-structured interviews, participants reported that congenital heart disease was a constant and limiting presence which was associated with feelings of low mood, trauma, shame, and lack of control. Coping strategies identified included denial and overcompensation.

In addition to psychological distress, some studies have found that patients with congenital heart disease meet the criteria for psychiatric diagnoses. In a subset of 58 patients randomly selected from the initial sample, 50% met the criteria for at least one lifetime mood or anxiety disorder during clinical interviews (Kovacs et al., 2009). In another study, 35% of patients considered to be 'well adjusted' met the criteria for psychiatric diagnosis (Bromberg, Beasley, D'Angelo, Landzberg, & DeMaso, 2003), compared to 79% of participants in a study by Horner, Lieberthson, and Jellinek (2000).

1.2.3 Quality of life

The quality of life for people with congenital heart disease is generally good, and in some cases has been reported as better than that of the general population. Individuals with congenital heart disease were found to participate more in social activities than their non-affected peers (Van Rijen, et al., 2003). There was also a high level of similarity between the living conditions, marital status, and offspring of the patient group and reference group.

In another Dutch study of mild congenital heart disease, participants reported some social impediments as a result of their condition (Fekkes, Kamphuis, Ottenkamp, Verrips, Vogels, Kamphuis, & Verloove-Vanhorick, 2001). These included effects on schooling (19%), leisure time activities (15%), and choosing (13%) and performing their occupation (9%). However, the health-related quality of life of those experiencing social impediments was comparable to the general population, and those without social impediments reported a better health-related quality of life on 6 of the 12 scales used than the general population. The authors attributed this finding to the coping mechanisms used by participants and suggested that a revision of values could lead to a greater appreciation of health status, as expressed as a higher subjective experience of quality of life.

A recent review concluded that adults with congenital heart disease are generally successfully engaging in adult roles and responsibilities, but there are specific psychosocial challenges faced by this population (Kovacs, Sears, & Saidi, 2005). These include pregnancy considerations and exercise capabilities. The review acknowledged the differences within the literature and cautioned against generalising.

1.2.4 Variation in the literature

There are several possible reasons for the variation in the literature. Firstly, better psychosocial outcomes may be associated with disease severity. Of the studies reporting good psychosocial well-being and quality of life, one included participants with mild congenital heart disease that did not warrant surgical intervention (Fekkes et al., 2001) and another included participants who had all had corrective surgery before the age of 15 (Utens et al., 1998). However, in several studies psychological distress was unrelated to cardiac severity (Brandhagen et al., 1991; Kovacs et al., 2009; Utens et al., 1998), providing little support for this hypothesis.

Alternatively, the method of data collection could also account for the inconsistency. The studies with negative findings nearly all used semi-structured interviews (Bromberg et al., 2003; Cornett & Simms, 2014; Horner et al., 2000; Kovacs et al., 1998), whereas the results for studies using questionnaires were more varied (although this could reflect the different scales used). It might be possible that people with worse psychosocial functioning and quality of life are more likely to volunteer to be interviewed, or that the greater depth of information provided in interviews was more likely to reveal psychopathology.

It has been highlighted that there is a general discrepancy between the findings of North American and European studies (Kovacs et al., 2005). The majority of European studies report equal or superior quality of life in people with congenital heart disease when compared to the general population, although there are some exceptions, (e.g. Cornett & Simms, 2014), whereas North American studies have generally found a reduced quality of life and an increased level of psychopathology. This could be due to differences in health care provision, or broader cultural differences.

Other factors to consider when interpreting the results include the changes in surgical techniques and clinical approaches over time. It is possible that psychosocial outcomes could vary according to different types of surgery and approaches to the management of congenital heart disease, such as providing patients with information and education about their condition. Additionally, the generalisability of results is limited by the heterogeneity of conditions that come under the umbrella term 'congenital heart disease' as each condition can differentially affect quality of life (Saliba, et al., 2001).

1.3 Psychological impact of Marfan Syndrome

Marfan syndrome is distinct from many congenital heart conditions in that its effects are not limited to the cardiovascular system. It is therefore possible that individuals with Marfan syndrome may be affected in different ways to people with other congenital heart conditions. Although the medical implications of Marfan syndrome have been well-researched, the literature on psychosocial functioning in Marfan syndrome is very limited, and therefore poorly understood. Seven studies have been conducted in both Europe and America to date, and an additional prospective trial is currently underway in Belgium (Nobeles, Loeys, De Paepe, & De Backer, 2011).

1.3.1 Self-esteem

Participants in several studies have reported that they have a positive self-image. Peters, Kong, Hanslo, & Biesecker (2002) found that their sample scored highly on a standardised measure of self-esteem, and 91.5% of participants in another study believed that they were as valuable as people without Marfan syndrome (De Bie, De Paepe, Delvaux, Davies, & Hennekam, 2004). However, concerns about body image can negatively affect the lives of people with Marfan syndrome. 53% of one sample reported that they experienced low self-esteem because of their physical appearance (Van Tongerloo & De Paepe, 1998). 50% of participants in another study said that they were not happy with their body image, and 37% reported that they did not view themselves as sexually attractive (De Bie et al., 2004). Participants claimed that this severely hindered them in initiating a sexual relationship. The presence of striae (more commonly known as stretch marks) has been found to be a significant predictor of reduced sex drive in Marfan syndrome (Peters et al., 2002). It is not clear how this compares to the perception of body image in the general population.

1.3.2 Stigma and discrimination

Stigma is the devaluation of an individual based on a particular characteristic, and can be defined as the co-occurrence of the following: labelling, stereotyping, status loss, and discrimination (Link & Phelan, 2001). The distinctive physical characteristics of Marfan syndrome have the potential to leave individuals vulnerable to stigma, as with any visible condition (Joachim & Acorn, 2000). 65% of one sample recalled being teased or bullied during childhood as a result of their physical appearance (Van Tongerloo & De Paepe, 1998). 32% of participants in a study of stigma in adults believed that they had been socially discriminated against or devalued because they had Marfan syndrome (Peters, Apse, Blackford, McHugh, Michalic, & Biesecker, 2005). Endorsement of this belief was significantly associated with depressive symptoms, low self-esteem, the view that Marfan syndrome had significant negative consequences on one's life, striae, and perceptions of workplace discrimination.

However, the direction of this relationship is unclear. Depression is associated with negative cognitive biases (Beck, 2002), including for interpersonal stimuli (Gotlib, Krasnoperova, Yue, & Joormann, 2004), and could therefore lead to depressed individuals with Marfan syndrome perceiving their situation much more negatively than their non-depressed

counterparts. Alternatively, the experience of being discriminated against could lead to depressive symptomology and a reduction in self-esteem. Further research is needed to elucidate this.

1.3.3 Anxiety and depression

Physical conditions can be associated with anxiety and depression, which can sometimes lead to a reduction in health status (Moussavi, Chatterji, Verdes, Tandon, Patel, & Ustun, 2007). Therefore it is important to understand the relationship between mood disorders and Marfan syndrome, in order to minimise any potential effects on people's mental and physical health. The literature on anxiety and depression within Marfan syndrome is incredibly limited. Van Tongerloo and De Paepe (1998) found no significant difference in anxiety and depression between their sample of young adults with Marfan syndrome and the general population, although levels of anxiety and depression were higher in women with Marfan syndrome than in affected men. Depressive symptoms were significantly associated with perceived stigma in another study (Peters et al., 2005). This remains a hugely under-researched area.

1.3.4 Starting a family

The decision to start a family can be a particularly hard one for individuals with Marfan syndrome. In addition to the general considerations associated with having a child, people with Marfan syndrome are faced with the added potential for health complications during pregnancy and childbirth (Mulder & Meijboom, 2012), and the 50% possibility that their child will also have the condition (Dean, 2007). Both of these issues surrounding pregnancy were a major concern for female participants in an early study into psychosocial functioning in Marfan syndrome (Van Tongerloo & De Paepe, 1998). 62% of respondents in another sample said that having Marfan syndrome had affected their reproductive decision making (Peters et al., 2002). Passing on the condition to their children was cited as the main concern, but many also had worries about their personal health.

Prenatal testing is available in the UK (NHS Choices, 2012), although not all people with Marfan syndrome wish to choose this option. 69% of individuals in one study expressed an interest in prenatal genetic testing (Peters et al., 2002). This reduced to 48.5% in another when the possibility of aborting affected foetuses was explicitly raised as a potential

consequence of prenatal genetic testing (De Bie et al., 2004). Prenatal testing is sometimes not an option. Not all affected individuals are aware that they have Marfan syndrome, especially as 25% of people have no familial history of the condition, and therefore some people can pass on the syndrome to their children without knowing it (Dean, 2007). This can be distressing if the condition is later diagnosed.

1.3.5 Limitations on daily life

Marfan syndrome can affect recreational activities and other aspects of day-to-day life. 57% of all participants in a Europe-wide survey said that Marfan syndrome had an influence on several aspects of their daily life (De Bie et al, 2004). Many cited participation in sports as being particularly affected; 34% reported that they were physically unable to play sport, although 41% enjoyed playing at least one sport. Van Tongerloo and De Paepe (1998) also found that the ability to play sport was compromised by the condition, although some participants continued to take part in sporting activities despite being warned against it. Adolescents felt that they would be more physically active if they did not have the disorder (Schneider, Davis, Boxer, Fisher, & Friedman, 1990). Other than sport, the most commonly reported daily life difficulties included finding clothing to fit, driving a car, recognising people in the street, and watching television (Van Tongerloo & De Paepe, 1998).

1.3.6 Quality of life

Measures of quality of life in Marfan syndrome are mixed. Peters et al. (2002) found that participants had an 'adequate' quality of life, albeit with a significant reduction within the spiritual/psychological domain. Similarly, an Italian study found a reduced quality of life in the psychological domain but not the physical domain when compared to a healthy population (Fusar-Poli, Klersy, Stramesi, Callegari, Arbustini, & Politi, 2008). Another study found that their sample had reduced scores on all measures of health-related quality of life, which was comparable to other chronic conditions (Rand-Hendriksen, Johansen, Ove Semb, Geiran, Stanghelle, & Finset, 2010).

It appears that quality of life is related to the subjective perception of illness severity, rather than the actual physical severity of the condition itself (De Bie et al., 2004; Rand-Hendriksen et al., 2010). Therefore, people who perceive their health to be particularly badly affected by Marfan syndrome are more likely to report a worse quality of life than people who do

not perceive their condition to be particularly serious, irrespective of the actual severity of their illness. Clinicians should bear this in mind when treating patients, as individuals with a milder presentation of Marfan syndrome may be wrongly perceived to be coping better with the condition than others with a more complex presentation. This highlights the role of individual differences in coping and adjustment.

1.3.7 The impact of surgery

The researcher is unaware of any studies investigating the psychosocial impact of aortic root surgery on people with Marfan syndrome. The only published account of surgery at the time of writing comes from the ninth person to undergo the PEARS procedure (Allen & Pepper, 2010). Allen describes her experience of surgery positively, although she expressed frustration that the procedure is not well-publicised. PEARS surgery reduced the size of her aorta to a healthy measurement of 3.6 cm, which has remained stable and enabled her to have a second child - something that she had previously been advised against. Allen reports that the procedure has allowed her to return to her 'old self' and described it as the 'ideal solution to my predicament.' She now works as a mentor for other people exploring the option of PEARS surgery. Of course, this account only represents one person's experience of undergoing PEARS surgery but it still contributes to the anecdotal evidence on the possible psychosocial benefits of the procedure.

1.3.8 Summary of findings

Overall, people with Marfan syndrome seem to be relatively well-adjusted (De Bie et al., 2004; Peter et al., 2002; Sneider et al., 1990; Van Tongerloo & De Paepe, 1998). They generally display positive self-esteem (De Bie et al., 2004; Peters et al., 2002) and there does not appear to be any difference in levels of anxiety and depression compared to the general population (Van Tongerloo & De Paepe, 1998). However, there are some specific difficulties faced by individuals with the condition. Physical aspects of Marfan syndrome can lead to negative perceptions of body image (Van Tongerloo & De Paepe, 1998), which can affect relationships (De Bie et al., 2004; Peters et al., 2004). This can be further exacerbated by exposure to stigma from others, which is associated with depressive symptoms (Peters et al., 2005). Other difficulties include limitations to activities of daily life (De Bie et al., 2004; Van Tongerloo & De Paepe, 1998) and the risks associated with starting a family (Mulder & Meijboom, 2012). These difficulties might explain the variation in the findings on quality of

life (Fusar-Poli, et al., 2008; Peters et al., 2002; Rand-Henrikson, et al., 2010); the extent to which individuals experience these scenarios and the way in which they cope with them is likely to affect their overall quality of life.

The generalisability of these findings is limited due to the small body of literature on this topic, sometimes using very small samples. It is also not clear whether it is representative of all people with Marfan syndrome, given that the condition is highly variable in its presentation. There are some similarities with the literature on congenital heart disease, for example in the areas of recreational activities and childbearing, although there are aspects specific to Marfan syndrome, such as stigma based on physical features. It is not possible to compare the findings on psychopathology due to the lack of research in this area on Marfan syndrome.

1.4 Aims of the current study

The literature review highlighted the lack of research into the lived experience of people with Marfan syndrome, particularly with regard to levels of anxiety and depression. None of the studies described investigated the impact of surgery on psychosocial functioning. This is important because heart surgery can be a stressful and potentially life-threatening experience, involving long periods of absence from work and social activities, and a large amount of uncertainty. Time spent on a waiting list for heart surgery is associated with significantly increased levels of anxiety and depression, and significantly impaired social functioning (Underwood, Firmin, & Jehu, 1993). As such, the effect of surgery on patients' well-being should be explored.

Equally, the perspectives of patients are just as important as other outcome measures in determining the efficacy of novel forms of treatment. For example, service user involvement lead to significant insights into the long-term effects of electroconvulsive therapy that may have been missed without their input (Thornicroft & Tansella, 2005). Perhaps more pertinently, PEARS surgery was developed as the result of the idea of a service user, and so it seems important to maintain a high level of service user involvement within its evaluation. Therefore, there is a need to explore additional methods of patient reported outcomes (PROs) as part of research into new treatments.

Based on these findings, the aims of this study were twofold:

1. To explore participants' narrative accounts of their experiences of undergoing prophylactic surgery.
2. To examine the utility of psychometrics and focus groups as PROs in future projects.

2.0 Method

Ethical approval was sought from the National Research Ethics (NRES) Committee London – Hampstead. The application was reviewed by the committee on 15 May 2013 and ethical approval was subsequently granted on 24 May 2013 (REC Ref: 13/LO/0679).

People who had external aortic root support surgery for Marfan syndrome were approached by their surgeon and introduced to the researchers. Of 30 patients, 22 agreed to their details being passed to the research team and received a formal invitation to participate in the research study.

2.1 Service user focus groups

Potential participants were provided with an information sheet containing details of the study, in order to make an informed decision (see Appendix 1). Those who responded were invited to attend a focus group. After being given time to read the information sheet and consent form, written consent was obtained by the researcher.

After written consent has been obtained, participants were asked to complete two HADS and two WSAS assessments; the first relating to how they felt before they had the operation and the second relating to how they felt at the time of the focus group. Participants were given the opportunity to ask questions before completing the questionnaires and were given as much time as they required.

The Hospital Anxiety and Depression Scale (HADS) was used to assess the levels of anxiety and depression experienced by participants at each time point (Zigmond & Snaith, 1983). The HADS is a well-established and widely used 14-point scale, with seven points consisting of statements related to anxiety and seven points relating to depression. Responses to each

item are scored from 0-3 to give a score between 0-21 for each of the two mood states (anxiety and depression). 10 is a clinical threshold for depression and anxiety.

The Work and Social Adjustment Scale (WSAS) was used to assess social and occupational functioning (Mundt, Marks, Shear & Greist, 2002). The WSAS is a five item self-report scale which asks participants to assign a score of 0-8 in response to the five questions, with a score of 0 interpreted as no impairment and a score of 8 representing severe impairment. The scores on each item are then summed to give a total score. A score above 20 suggests moderately severe disturbed functioning. Scores between 10 and 20 are associated with significant functional impairment but less severe clinical symptomology, and scores below 10 are associated with subclinical populations.

Participants then took part in a focus group. Participants were provided with a copy of a series of questions to act as a prompt throughout the focus group (see Appendix 2). These questions were constructed from the results of a thematic analysis of patient accounts of their experiences of surgery taken from the Exostent website (<http://www.marfanaorticrootssupport.org/experiences.html>). The focus groups were recorded using a Dictaphone.

2.2 Data analysis

2.2.1 HADS and WSAS

Participants' responses to the HADS and WSAS were summed to give an overall score for each measure. A Wilcoxon signed rank test was conducted on the data from the HADS and WSAS because the data were paired and normal distribution could not be assumed.

2.2.2 Focus group data

Recordings of the focus group were transcribed by the researcher and analysed using thematic analysis, as described by Braun and Clarke (2006). It was not possible to assess inter-rater reliability as analysis was conducted by the researcher alone due to resourcing constraints. The data from the two focus groups were combined and treated as a single data set.

3.0 Results

3.1 Sample characteristics

Of the 22 potential participants, nine (41%) agreed to take part in this study. A further three people (13.6%) were willing to take part but were unable to attend due to transport problems or other engagements.

The nine people who agreed to take part attended one of two focus groups. The first focus group, held in July 2013, consisted of six participants (three males and three females). The second focus group, held in November 2013, consisted of three participants (two females and one male). All of the participants were unrelated, with the exception of 1D and 1E, who were a father and daughter.

Table 1: Participant characteristics

Participant Identifier	Gender	Focus Group (1/2)
1A	Male	1
1B	Female	1
1C	Male	1
1D	Male	1
1E	Female	1
1F	Female	1
2A	Male	2
2B	Female	2
2C	Female	2

3.2 Thematic analysis

Results from the thematic analysis are presented below. Themes and subthemes are summarised and provided alongside frequencies of occurrence (see Appendix 3 for full table).

3.2.1 Theme 1: Identity

Frequency = 13

Identity was a strong theme in both focus groups. It encompassed both participants' sense of self and how they wished to be perceived by others. This theme was divided into two sub-themes:

Wanting to be normal

Frequency = 3

Participants expressed the view that they wanted to be perceived as normal. They did not want to be defined by their condition or let their condition dictate what they could do.

'I felt strongly that I didn't want it to be a part of my life, I didn't want to be a victim to it all' (1D).

Feeling healthy

Frequency = 10

Participants strongly emphasised that they felt healthy and did not identify with a 'sick' role. Participants explained that they maintained this sense of identity throughout their treatment, including during inpatient stays in hospital and post-surgery.

'I never really thought of myself as having something wrong with me, my friends didn't really either' (1B)

'I felt just a bit like...I don't want to be with...all these sick people...Obviously you are one of them as well but...it doesn't really feel like that' (1C)

3.2.2 Theme 2: Family

Frequency = 14

Family was another frequently occurring theme within the focus groups. This theme was divided into three sub-themes:

Witnessing Marfan syndrome in relatives

Frequency = 4

Participants had mixed experiences of witnessing relatives live with Marfan syndrome. Some participants reported that they had seen the negative effects of the condition in relatives, such as the death of a family member or health complications. Others felt that witnessing the condition in relatives was protective and that it provided a shared experience and support.

'I think the fact that my two brothers have it as well, although it's sad, it's nice that we all three of us have it so we can share the experience of having it...umm...and then there's my dad who I've obviously seen the bad effects of it with...what can happen if...umm...yeah, just what can happen.' (2B)

Being a parent

Frequency = 6

Participants who were parents to children with Marfan syndrome expressed a sense of responsibility and a desire to protect their children. They felt a need to mentally prepare their children for surgery.

[1D and 1E = father and daughter]

1E: *'You always gave us pep talks didn't you?'*

1D: *'Well yeah...I needed to do my job...my job was to prepare you two...'*

1E: *'Yeah I was pretty scared.'*

Starting a family

Frequency = 4

Some female participants expressed concern about the impact of the condition on their ability to start a family. They identified that this was the only time that they felt the condition had impacted on their ability to lead a normal life.

'The only time that I've really felt like it's really impacted on my life is when they told me about the complications that might arise if I were to have children...and...that's always something I'd pictured myself doing so I think I was quite upset when they told me that' (2B)

3.2.3 Theme 3: Routine

Frequency = 8

Other participants did not identify the process of going for an echocardiogram as a stressor and felt that it had become part of their routine. They likened the experience to taking their car for an MOT or attending a dentist appointment.

3.2.4 Theme 4: Denial/Avoidance

Frequency = 6

Some participants expressed a desire to avoid thinking about some aspects of their condition, or reported that they avoided information about it. This included the possibility of aortic dissection or complications arising from surgery. They also identified that they did not want to admit that anything was wrong. In some cases, this led to avoiding behavioural changes that were associated with their condition, such as taking medication.

'I thought I was perfectly healthy, and then...they gave me the beta blockers [...]...and then I decided to take myself off them...'cause I just wanted to be normal' (1A)

'I was prescribed them [beta blockers] for at least three years before the operation and I never got round to taking them. I just didn't want to admit that there was anything wrong...and...umm...I just thought that a tablet every day...that happens when you're ill.'

(1C)

3.2.5 Theme 5: Uncertainty

Frequency = 4

Some participants found the uncertainty of their condition to be a stressful experience. They identified annual echocardiograms as a worrying time. However, it was felt that the results provided some temporary relief by allowing for some certainty.

'That's the psychological impact, because it's a worry...umm... The best description is having the sword of Damocles hanging over you. You don't know what you're going to do...so that's a big concern' (1D)

'The worry always was is this the time that they're going to say oh yes now you need to...so as it went on later on in life then it became a bit more of a worry. I remember feeling quite relieved sometimes when I came back to there and it was all ok and all fine for another year at least. I remember being very happy afterwards!' (1C)

3.2.6 Theme 6: Experience of Personalised External Aortic Root Support (PEARS) surgery

Frequency = 21

In general, participants felt overwhelmingly positive about the surgery. Many said described it as a win-win situation and said that it felt like common sense. Participants expressed a wish to get surgery 'over and done with'. Some younger participants spoke of the convenience of being able to have surgery between gaps in education. Hearing about the possibility of PEARS surgery was a great relief for several participants. Most felt optimistic prior to the surgery, as well as feeling optimistic about their future following surgery. Participants reported high levels of trust in the surgical team.

'When I found out about it... it was the greatest feeling. I thought I've really been saved and something wonderful is happening just when I wanted it to. That was a really massive...umm...good feeling. Obviously a great relief.' (1D)

'I just remember being in a fantastic mood because it was all over and...all your worst fears which you had whilst signing the waiver form...they're all gone...so...yeah, it was just a great feeling that I've come through it and the worst is over now. And this thing that you've been worrying about for the last 15 years is gone. So...um...yeah, a great feeling.' (1C)

'For me, because I had it done between finishing school and starting college over the summer...so for me it was a case of...I wasn't really that anxious about it, it was more just getting it out of the way' (1B)

3.3 HADS and WSAS data

Table 2: HADS and WSAS Data

	HADS-A		HADS-D		WSAS	
	Before	After	Before	After	Before	After
Median	5	1	1	1	0	2
IQR	2-9	1-4	0-2	0-1	0-2	0-2
Range	0-12	0-8	0-8	0-4	0-22	0-25

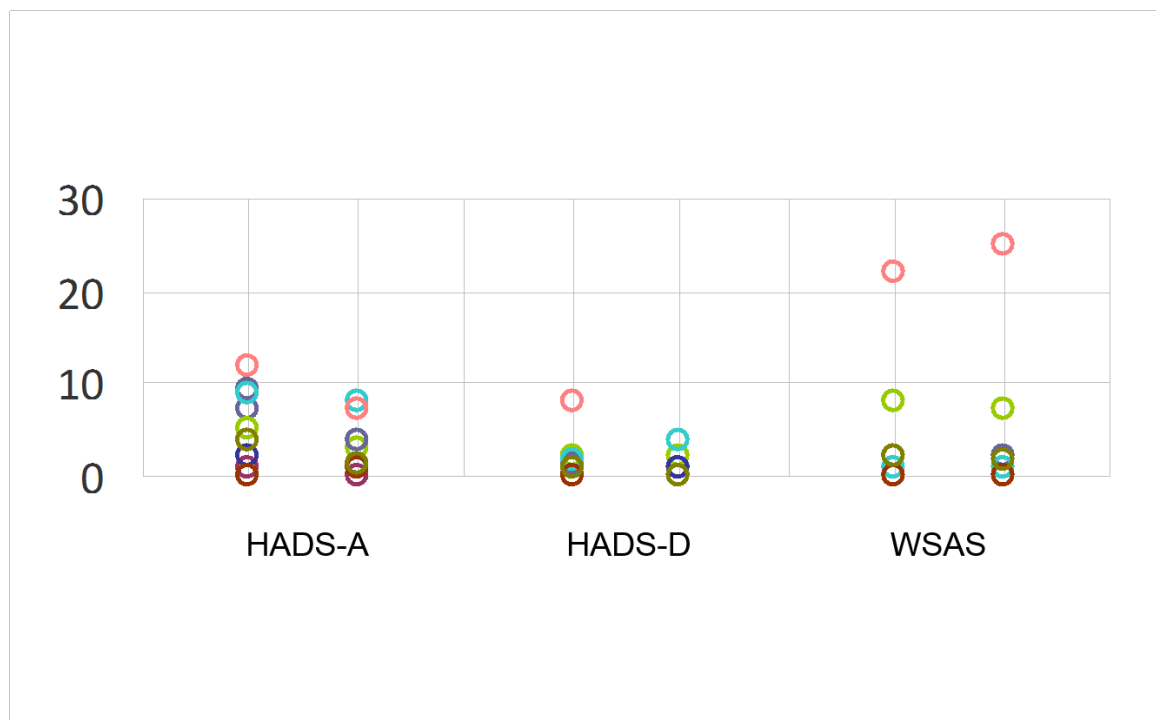


Figure 1: HADS and WSAS scores

Table 3: Analysis of HADS and WSAS scores

Measure	N (excluding tied ranks)	Critical value for W at $p \leq 0.05$	W	Significant at $p \leq 0.05$?
HADS Anxiety	9	5	2.5	Yes
HADS Depression	5*	-	-	-
WSAS	4*	-	-	-

* Too many tied ranks to allow a reliable calculation of the Wilcoxon signed-rank statistic

Anxiety scores as measured by the HADS were significantly higher before surgery than after surgery ($W=2.5$ at $p \leq 0.05$). It was not possible to test the other comparisons as there were too many tied ranks.

4.0 Discussion

This study explored participants' accounts of undergoing prophylactic personalised aortic root support surgery and its impact on their lives. It also explored levels of anxiety and depression, as well as the social and occupational functioning of participants, and the extent to which this had changed after surgery. This was needed as little is known about the impact of prophylactic surgery on the lives of individuals with Marfan syndrome, although anecdotal evidence has suggested that there may be a psychological benefit (NICE, 2011). Additionally, there is a need to investigate the utility of including psychological measures in future research because the assessment of patient related outcomes (PROs) is an important part of the evaluation of this new form of surgery.

Therefore, the aims of this study were to explore participants' narrative accounts of their experiences of undergoing prophylactic surgery, and to examine the utility of psychometrics and focus groups as PROs, with the objective of using these measures in future projects. Participants identified several areas in which Marfan syndrome and surgery had affected their lives, which provide opportunities for potential interventions and future research. The findings are discussed in relation to the aims and the broader implications for individuals and service provision.

4.1 Aim 1: To explore participants' narrative accounts of their experiences of undergoing prophylactic surgery

A number of themes were identified in the analysis of the focus group data. One of the most frequently occurring themes was the strong sense of identity experienced by participants. Participants do not perceive themselves as ill and strongly identify as healthy, 'normal' people. In some cases this is a conscious decision; one participant reported that he was adamant that he would not be defined by his condition or become a victim to it. More commonly, participants simply felt no different to other people. One reason for this could be that none of the sample reported any health effects except undergoing surgery, which in itself was preventative. The lack of any health complications is likely to have contributed towards individuals feeling that they are healthy. The potential of the surgery to allow individuals to live a relatively normal life without further complications may allow participants to maintain this sense of self.

This sense of identity could be a protective factor in some circumstances. Several participants reported maintaining their feelings of good health during the course of surgery, particularly when compared with other people on the ward. This appeared to serve as a coping mechanism; participants felt that their situation was better than other patients, which may have made it easier to cope with. However, it is possible that this could lead to an underestimation of the severity of their condition; some participants reported undertaking activities that they had been recommended against after surgery because they felt that they were fit enough. Clinicians should be aware of this and educate patients accordingly.

In some cases this sense of identity can make going for surgery harder, and can lead to some maladaptive coping strategies, including denial and avoidance. Some participants found that feeling healthy made the thought of major surgery more difficult as it would mean a short-term reduction in health. Similarly, it can be difficult when the realities of the condition require behavioural change, such as taking medication, especially when the behavioural change is incongruent with self-image. Beta blockers are commonly prescribed prophylactically in Marfan syndrome to reduce dilation of the aorta, although their efficacy is disputed (Gersony, McCloughlin, Jin, & Gersony, 2007). Several participants refused to take beta blockers as they did not want to admit that they were ill. This is consistent with the impact of self-identity on medical adherence in other long-term conditions (Adams, Pill, & Jones, 1997) and may provide an opportunity for psychological interventions targeted at improving adherence (Kahana, Drotar, & Frazier, 2008).

Participants' experiences of going for aortic root measurements were mixed. For many, the check-ups were perceived as a normal part of life, comparable to a routine dental appointment or an MOT. They were also viewed positively by some as a way of gaining some certainty about the size of their aorta. However, for others the experience provoked some anxiety, specifically relating to whether they would imminently require surgery or not. It is not clear which factors contribute to feelings of anxiety in this situation and which factors might be protective. Future research could explore this further.

Another frequently occurring theme was the concern about child-bearing raised by female participants. For some, it was the first time that the condition had affected their life choices,

which was difficult in itself. The main concern was with regard to participants' personal health and the potential for obstetric complications. The possibility of passing on the condition to children and genetic testing was only raised by one participant. She had gone through genetic testing with one of her children and described the process as a 'rigmarole'. She also described how she had not had any concerns at the time as she had a positive personal experience of living with Marfan syndrome, but then realised afterwards that her child could have been born with a much more serious presentation than her own.

The finding of concerns about child-bearing is consistent with previous research on the psychosocial impact of Marfan syndrome (Peters et al., 2002; Van Tongerloo & De Paepe, 1998). Given that this concern is expressed in multiple studies, it is important that strategies should be implemented to address this. Participants reported that peer support had helped to alleviate anxieties surrounding childbirth, which could represent a helpful intervention strategy for other women in this position. Mentoring schemes already exist with the Marfan community (Allen & Pepper, 2010), although it is not clear how many people are aware of them or choose to access them. Additionally, one participant said that she felt much more relaxed about the birth of her second child when the team was briefed about Marfan syndrome and a cardiologist was on standby in case of any complications. Making sure that obstetric staff are fully informed of the condition and making appropriate contingency plans should also help to reduce anxiety, especially when the patient is made aware of this.

In terms of participants' opinions of PEARS surgery, the sample was universally positive about the experience. This positivity was the result of several factors. Participants reported high levels of trust in the surgical team, they liked the fact that in some cases the operation could be scheduled to fit in with educational commitments, and they felt that the benefits of surgery vastly outweighed the potential negative consequences. Some of the reasons cited as benefits were the non-invasive nature of the operation, the ability to keep their own organs rather than have artificial replacements, and the opportunity to be drug-free.

Participants were particularly averse to warfarin, with one participant referring to it as 'rat poison'. Previous studies have found that attitudes to warfarin therapy are highly individualistic and based on a cost-benefit analysis of factors important to the patient (Fuller, Dudley, & Blacktop, 2004). Given participants' self-perception as healthy individuals,

the thought of taking potent medication for the rest of their lives is likely to be viewed as a significant cost, and therefore is likely to influence their decision making on their preferred type of surgery. Further research comparing the different surgical options in Marfan syndrome may benefit from exploring the costs and benefits perceived by the service user, as they may differ to the costs and benefits perceived by clinicians. Indeed, research has demonstrated that priorities can significantly differ between clinicians and service users (Perkins, 2001), providing further impetus for research on this topic.

4.2 Aim 2: To examine the utility of psychometrics and focus groups as PROs in future projects

This project trialled the use of two psychological measures in assessing change in psychosocial functioning before and after surgery - the Hospital Anxiety and Depression Scale (HADS) and the Work and Social Adjustment Scale (WSAS). Unfortunately, due to the small sample size, it was not possible to compare results of the depression sub-scale of the HADS, and the WSAS as there were too many tied ranks for accurate analysis in such a small sample size. However, there was a significant reduction in anxiety after surgery, which is consistent with the findings of Underwood et al. (1993). Although there was a reduction, it is important to note that both the mean pre- and post-surgery anxiety scores were within the normal range and therefore did not meet the clinical threshold for anxiety.

Caution should be exercised in interpreting this result as the pre-surgery anxiety score was provided retrospectively; participants' perceptions of their previous anxiety levels could have been minimised in light of the positive outcome post-surgery. Additionally, Underwood's study (1993) was at a time when waiting lists for cardiac surgery were long; 28% of patients in their sample scored within the clinically significant range for anxiety. Participants in the present study reported very short waiting times before surgery and therefore may have experienced a lower level of anxiety than if they had been on the waiting list for a longer period.

Despite this, it is encouraging to see that the mean scores for both anxiety and depression were within the normal range. Combined with the qualitative data, this suggests that participants in this study seem to be experiencing a good quality of life. This is slightly

incongruent with previous research as two studies found that people with Marfan syndrome had a reduced quality of life in the psychological domain (Fusar-Poli et al., 2008; Peters et al., 2002) and one found a reduction in quality of life in all domains (Rand-Hendriksen et al., 2010). This observation is limited in that it is comparing outcomes on standardised measures with a personal interpretation of the current results, but it would be interesting to see if this would be the case if the same quality of life measure was used with the present sample.

Although the ability to analyse the utility of the HADS and WSAS was limited in this study, both measures have been used successfully in other studies investigating the psychosocial impact of surgery (Detroyer, Dobbels, Verfaillie, Meyfroidt, Sergeant, & Milisen, 2008; Porat, Cohen, Schwartz, & Hassin-Baer, 2009). The HADS and WSAS represent useful tools for measuring psychological change in future studies and will have greater validity when used prospectively, as intended with the planned comparative study of the three different forms of surgery.

Compared to quantitative measures such as the HADS and the WSAS, focus groups and the subsequent qualitative analysis could be perceived as a less attractive option for use as a patient reported outcome measure, due to the time commitment involved and the skills that they require. However, focus groups produce much more in-depth data and allow for a greater range of responses than other methods (Barbour, 2005). Although this study used a small sample, the consistency of the themes gives face validity and has provided useful insights into participants' experiences of PEARS surgery. Based on these findings, focus groups could be a useful PRO in future research.

3.3 Limitations

This study has several limitations. Firstly, the sample size of nine participants was small, although this was limited to a certain degree by the relatively small number of people who have had PEARS surgery. With this in mind, the response rate of 41% can be considered quite successful. The small sample size impacted on the ability to conduct inferential statistics and therefore future studies will need to recruit a greater number of participants in order to conduct this sort of analysis. Equally, this meant that only a few people's views

were explored within the focus groups and so they cannot be assumed to be representative of all people with Marfan syndrome.

Secondly, the methodology places some restrictions on the interpretation of this piece of research. Due to the exploratory nature of the study, the quantitative data was collected retrospectively and is therefore open to memory distortions and lapses (Jansen, Stiggelbout, Nooji, Noordijk, & Kievit, 2000). However, in future studies this data will be collected prospectively and therefore will provide a more valid account of participants' experiences. Additionally, only one researcher conducted the thematic analysis of the qualitative data. Ideally, at least two researchers would analyse the data and a percentage of agreement on themes would be included as a measure of inter-rater reliability, with the objective of reducing subjective bias (Braun & Clarke, 2006). This was not possible due to the constraints of a student project, but the researcher was aware of the possibility of bias during the analysis and endeavoured to remain impartial. However, it is possible that there could be some level of bias in the thematic analysis of the data as a result.

The focus group format may have resulted in participants failing to fully disclose information about their experiences. Social desirability bias could have led participants to minimise reports of distress, or, conversely, exaggerate their experiences (Nederhof, 1985).

Alternatively, socially anxious participants may not have felt comfortable speaking about their experiences in front of a group of strangers. Future qualitative research on this topic may benefit from including individual interviews in addition to the focus group format.

Lastly, the inclusion of only those participants to have had PEARS surgery may have resulted in a bias towards certain personality types. Personalised external aortic root support surgery is a novel form of surgery and may have attracted people who had a confident, positive outlook and were accepting of a risk associated with a less tested operation. Equally, not all people with Marfan syndrome are eligible for PEARS surgery, especially those who are particularly unwell. Therefore the sample may have been biased towards confident, healthy individuals, which could have affected the results. It also limits the generalisability of this research for people with Marfan syndrome who have had other forms of surgery.

4.4 Future directions

This study served to act as a pilot study for exploring different methods of assessing the psychosocial impact of Marfan syndrome. The results showed that the HADS and WSAS are suitable measures to quantitatively assess psychological change, and that focus groups can complement the numerical data by providing a more in-depth qualitative account of people's experiences. Therefore, future studies could use these methods as patient reported outcomes. A study is currently being planned which seeks to use the findings from the focus group data to form survey questions (Prof Tom Treasure, personal correspondence).

In terms of expanding the literature base on Marfan syndrome, there are several areas in which new research is required. There is a dearth of studies on anxiety and depression in Marfan syndrome. It is particularly important that this is explored as some studies on congenital heart disease have found an increased level of mood-related symptoms, which in some cases qualify for clinical diagnoses (Bromberg et al., 2003; Horner et al., 2000; Kovacs et al., 2009). Additionally, some of the challenges experienced by people with Marfan syndrome, such as stigma, are significantly associated with depressive symptomology (Peters et al., 2005), but this relationship has not been fully explored. This study has highlighted the unique challenges faced by individuals with Marfan syndrome, and therefore understanding how these may influence mood states is imperative in terms of establishing effective support and interventions for those affected.

Equally, it is important to know if people with Marfan syndrome do not experience heightened levels of anxiety and depression. Understanding factors associated with resilience is just as useful as exploring factors associated with psychopathology, as they can be used to inform interventions and predict those most at risk of anxiety and depression. Research into mood states in Marfan syndrome could therefore help people adapt to other chronic conditions.

This study discovered that some participants failed to take prescribed medication, such as beta blockers, because they did not want to engage in behaviours that they associated with illness. This is worrying, as this medication is prescribed to reduce the pressure on the aorta and therefore individuals may be putting themselves at an increased risk of aortic aneurysm

by not taking it. Research is needed to examine more fully the reasons for non-compliance with medication regimens and to explore the possibility of interventions based on the findings. Participants in this study placed great value on their health status, so an effective strategy might be to frame this behaviour in a health-positive light.

Finally, a consistent finding across research into Marfan syndrome is that women are concerned about the impact of the condition on their ability to have children. As discussed previously, there are already some initiatives available to help with this, including peer mentoring schemes (Allen & Pepper, 2010). However, the availability and efficacy of these services is unclear, and it is also unknown whether there may be other interventions that could help to alleviate these anxieties. Subsequently, there is a real need for research into this area.

5.0 Conclusion

This study has provided one of the first explorations into the psychosocial impact of PEARS surgery on people with Marfan syndrome. It found that individuals with Marfan syndrome generally lead normal lives, on which the condition has very little impact beyond routine appointments. However, the impact for female participants was slightly greater due to the implications for child bearing. The requirement for surgery and the uncertainty surrounding it did cause some anxiety on occasion, but most participants approached surgery pragmatically and positively. The study also highlighted the way in which virtually all participants perceived themselves as healthy, normal individuals, and the effects that this could have on their behaviour. These findings were mostly consistent with the existing literature on the topic, although it could be argued that the sample in this study displayed greater levels of adjustment than others.

There still remains a lack of evidence in some areas of the literature, including explorations of self-identity and adherence, mood states, and interventions for child bearing concerns. The findings of this study add to the understanding of these areas but also highlight the need for further research. Despite its limitations, this study contributes to the understanding of the psychosocial impact of living with Marfan syndrome and undergoing PEARS surgery, and has implications for clinicians, service users and researchers alike.

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7.0 Appendix

7.1 Participant Information Sheet

Participant Information Sheet

When you are thinking about taking part in a research study, it is very important to have all the available information about the study before making your decision. This information includes knowing exactly what will happen before and during the study, and what will happen to the data collected after the research has finished. The purpose of this information sheet is to tell you as much as possible about the study that you have been invited to take part in. Please take your time to read this and ask as many questions as you would like. One of the researchers will discuss this information with you before you fill out the form that tells us whether you would like to take part in the study or not. You are under no obligation to take part in the study if you don't want to.

What is the study about?

You have been invited to take part in this study because you are one of the first group of people with Marfan syndrome to have had personalised external aortic root support (PEARS) surgery, which stops the aorta from expanding by surrounding it with a specially-made sleeve. People have reported that the monitoring checks, and the period waiting for surgery, are stressful experiences, but no studies have looked at this before.

We would like to find out about the psychological impact of waiting for PEARS surgery and what life is like afterwards. Put more simply, we would like to know how your mood and day-to-day life is affected by going for monitoring checks and waiting for surgery, and whether anything has changed since you have had the surgery.

Who is involved in the study?

This study is a research project being undertaken by two Masters students at the Institute of Psychiatry (IoP), King's College London – Judith Fosbraey and Durr-E-Sameen Hashmi. They are being supervised by Dr Kate Tchanturia, a Consultant Clinical Psychologist and Senior Lecturer at the IoP, and Professor Tom Treasure, Cardiothoracic Surgeon.

What will happen if I take part?

If you decide to take part, you will come to the Institute of Psychiatry in London to meet the researchers. We will discuss the study with you and answer any questions you might have, before asking you to fill out a consent form. You will then be asked to fill out four questionnaires. They will ask about your mood before and after surgery, and your day-to-day life before and after surgery.

The second part of the study will be a focus group. This involves getting a small group of people together (in this case, people who have had PEARS surgery) and having an informal discussion. We will ask some questions about going for monitoring checks, waiting for surgery, having surgery and your life after surgery. We will record the focus group using a Dictaphone.

You will then be asked to fill in the same questionnaires as before. This is just in case the focus group has made you want to change your answers in any way. Once you have completed the questionnaires, you have reached the end of the study. You are then welcome to join the research team for lunch, which will be provided as part of the study. We will also cover all your travel costs.

What happens if I change my mind?

You can leave the study at any time, even if you have completed the study – we will simply remove your data from the investigation.

How will my personal data be stored?

Your personal data will be stored on NHS-approved encrypted memory sticks. This means that someone has to have a username and password to access it. The only people who will have access to your data are your direct clinical care team and the research team. Any personal data will also be anonymised by using a code, and any data that could potentially identify you will be excluded from the write-up of the study. Your personal data will be kept for one year, after which it will be securely disposed of.

What happens after the study?

The research team will analyse the questionnaires and the data from the focus group, which will then be written up as an MSc dissertation. The final report may also be published in academic journals and/or presented at conferences. We will be very happy to provide you with a copy of the final report if you would like us to.

Data collected from the study will be securely stored at the Clinical Trials and Evaluation Unit (CTEU) at the Royal Brompton Hospital for 4 years after the study has finished. After this time, it will be destroyed.

We hope this information sheet answers any questions you may have had about the study. However, if there are still things you wish to ask then please do get in touch with the research team using the contact details below. There will also be plenty of opportunities for discussion on the day of the study itself.

Contact Details

Judith Fosbraey: judith.fosbraey@kcl.ac.uk

Durr-E-Sameen Hashmi: durr-e-sameen.hashmi@kcl.ac.uk

7.2 Focus group prompt sheet

Focus Group Questions

1. What is the nature and magnitude of the psychological impact in the watch and wait monitoring phase for patients with Marfan syndrome?

- Before your surgery, how did you feel on a day to day basis?
- What was it like in the week/day/morning before you went for a monitoring echo?
- When the doctors/nurses/technicians talked with you were there any things you remember as making you anxious?
- If so, how did you try to alleviate this anxiety?
- How, if at all, could this stage be made less worrying?
- Have you ever experienced any negative feelings related to the fact that Marfan syndrome is genetic?
- Do you feel you would have benefited from external support to help solve the dilemma of immediately having surgery or not?
- How did you feel when you found out that there was a less intrusive surgery than aortic replacement surgery? Did it help you make a faster decision on whether or not to have surgery as soon as possible?

2. What is the nature and magnitude of the psychological impact of awaiting impending surgery for patients with Marfan syndrome?

- How did you feel when you were told you needed surgery?
- What was it like in the weeks/day/morning before your surgery?
- What was your attitude towards the operation?
- How did you mentally prepare for the operation/how did you try to manage any anxiety?
- How, if at all, could this stage be made less worrying?
- Do you feel personal support available closer to the surgery helped you cope better with the experience?
- Was it helpful to have a whole network of support around you, with other people with Marfan syndrome and professionals willing to share their experiences?

3. Having had external aortic root support surgery. In what direction and how great is any change in psychological state/trait?

- What impact has the surgery had on your life?
- Can you compare before and after (say the year before and the year after) and how you felt?
- Has having the surgery had any effect on how you perceive your future to be?
- If so, how is this different from your outlook on life before the surgery?
- Do you feel that the amount of professional and personal support available at the time, before and during the surgery period helped you cope better with the experience?
- Do you think it would be/ was beneficial to have extra professional and personal support during the time when you were trying to get back into your routine a few days after surgery?

7.3 Focus group themes

Theme	Sub-Theme	Evidence
Uncertainty (4)	-	...before I had the procedure was that it had only been done to Tal so...nothing...I had no expectation...nothing to base...umm...what to expect on... (1D)
		That's the psychological impact, because it's a worry...umm... The best description is having the sword of Damocles hanging over you. You don't know what you're going to do...so that's a big concern (1D)
		The worry always was is this the time that they're going to say oh yes now you need to...so as it went on later on in life then it became a bit more of a worry. I remember feeling quite relieved sometimes when I came back to there and it was all ok and all fine for another year at least. I remember being very happy <i>afterwards!</i> (1C)
		So when I hit 40 it suddenly started to go and time started ticking. (1D)
Routine (8)	-	Well for me it was just something that had to be done...and it just became part of the routine... it was just happened every... six months...and it had to be endured. It was like going to the dentist....so it was part of my normal routine, something that happened to me and I just had to do it (1F)
		Yeah it becomes part of life doesn't it? (1E)
		Yeah, it's like going for an MOT...and another pass...it's not critical, you know? (1D)
		I'd always...like...from when I was very young I had appointments like every six months so it was just something that, like, I did and it didn't...I didn't really know what impact it was having...really... 'cause it was just like an appointment... They'd say the measurements but I didn't really know what that meant...I didn't really have any worries because I didn't really know what was going on, to be fair. (1B)
		...it was just...a regular appointment every six months, have a check-up and then...I wouldn't...really...that would just be it. It was quite fine to be fair. (1A)
		So from my point of view it's never been a big deal. Not really even noticed it to be honest, it's always just been oh, another trip to the doctor. (2A)
		I think it's just sort of all...all part of it and...umm...the fact that I went with my brothers, we all went as a family, we always have our appointment on the same day...umm...we have our echoes, we each have our echoes in the morning and then we all go together to the appointment and yeah, it just sort of seems part of it. (2B)
		...having done it every year it just becomes more and more routine. (2B)
Identity (13)	Wanting to be normal (3)	I felt strongly that I didn't want it to be a part of my life, I didn't want to be a victim to it all (1D)
		Umm...yeah...I was going to say...before I knew I had to have the operation, I was...well, I thought I was perfectly healthy, and then...they gave me the beta blockers. So that made...I was at a tennis academy at the time and then I had to start taking those and then was struggling quite a lot so I found that bit...in my mind...had quite a big thing...and then I decided to take myself off them... 'cause I just wanted to be normal (1A)

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Theme	Sub-Theme	Evidence
Identity (13)	Wanting to be normal (3)	They do the same within their remit as other kids do...you know...I've never stopped them. I've always had a thing...if you want to do something, we'll try it...if you don't want to do it then that's fine...you know. They both ski, we've all been skiing....just sod it, let's go. It's your mind-set as well, isn't it? (1F)
	Feeling healthy (10)	It is strange, being sort of...healthy and then thinking...is the operation going to take anything out of me? (1C)
		Umm...yeah...I was going to say...before I knew I had to have the operation, I was...well, I thought I was perfectly healthy (1A)
		I was prescribed them for at least three years before the operation and I never got round to taking them. I just didn't want to admit that there was anything wrong...and...umm...I just thought that a tablet every day...that happens when you're ill. (1C)
		I never really thought of myself as having something wrong with me, my friends didn't really either (1B)
		...when I was on the ward I...all the other patients were... quite elderly and all with heart...you know, heart issues and...[inaudible]...you know, they all seemed very ill, and after the operation I didn't really class myself as being ill...(1C)
		I felt just a bit like...yeah...I just want to go kind of thing...I don't want to be with, like, all these sick people...Obviously you are one of them as well but...it doesn't really feel...it doesn't really feel like that. (1C)
		<i>[Thoughts on being offered the Bentall procedure]</i> I feel really healthy, I'm not having it done. (1D)
		And you don't feel ill either, do you? We all feel absolutely fine. (2C)
		Also I was in a room of old men, so I was, what, 34 or something at the time, 35, or was I younger than that? 32, 33, something like that, in a room of old men, and actually they seemed to be suffering a lot more than I was, whatever heart operation they'd had, so again, even though I was sick, I wasn't as sick as they were, so again, I didn't feel...and I guess with being young, you still feel much healthier still, even though you are tired and exhausted. (2A)
It's like, I feel fine, why would I want to be pumping Warfarin into me for the rest of my life when I feel absolutely fine at the moment? (2C)		
Denial/Avoidance (6)	-	[I] didn't really...know much about it and didn't really...want to know much about it (1A)
		1C: It was the same for me as well, I...I didn't really know much more about it at all, I just sort of... 1E: Carry on with life... 1C: Yeah...exactly... 1E: Yeah. 1C: But...in the back of my mind... I was prescribed them for at least three years before the operation and I never got round to taking them. I just didn't want to admit that there was anything wrong...and...umm...I just thought that a tablet every day...that happens when you're ill. (1C)

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Theme	Sub-Theme	Evidence	
Denial/Avoidance (6)	-	You have to admit that something might go wrong. (1F)	
		...and when I went for an appointment where it was considered that I have the Bentall graph I can honestly say I became an abject coward and I said I do not want to have this, I'm not going through with this. (1D)	
		Err...I haven't really thought about it, I think I've been slightly oblivious...in that I've gone and had these check-ups and they told me my measurement but I couldn't tell you what my measurement was when I had the operation done or before the operation, it was just...told you that now was the time to do it or it's fine, so it was always it's fine or it's not fine...the detail I'd never really taken much notice of...so no, I haven't really been counting down to it as such. (2A)	
Family (14)	Witnessing Marfan syndrome in relatives (4)	I knew that my grandfather had died of a ruptured aorta at quite a young age so I think there's always that, it can happen. And obviously you read the Marfan trust newsletters and they have dreadful, very sad stories of people dying at young ages so umm...and in our family it's mainly just the heart, we don't have...we aren't affected in many other ways, just the heart, so it does make you aware of it, not anxious I'd say, just aware. (2C)	
		...my dad has it...and so I was aware that he had it [PEARS surgery] (2A)	
		Umm, I think the fact that my two brothers have it as well, although it's sad, it's nice that we all three of us have it so we can share the experience of having it...umm...and then there's my dad who I've obviously seen the bad effects of it with...what can happen if...umm...yeah, just what can happen. (2B)	
		I followed my dad who did it probably a year or two years before mine, so again for me it was very matter of fact, he had it done, I'm having it done, and it wasn't anything more than that, really. (2A)	
	Being a parent of a child with Marfan syndrome (6)		You know, it's only afterwards it starts to sink in that they could have been born with worse side effects than they've had...err...umm...and then when you go into your appointments, the children are aware that you're going in to the appointments...you think oh what is the point, you know? Why are we putting them through this, but...the worry is for the child...less so for yourself. It's something that you don't want them to be...any more put out than they have to...you want them to be covered, so you...you go through it, so...the psychological impact is deferred, as it were, from yourself to the child. So when I was told I had to have the operation, it was...it just hits you, it goes sky high then, 'cause I didn't want...I hoped that they'd be old enough to cope with it, because it was only three years ago...so they were 15 and...16 and 19...so hopefully they're old enough to cope with it. But still...you've got to be aware that they're probably thinking somewhere in their brain that that could be me...that could be me next year...that could be me in ten years' time. How do you...how do you cope with that when you're scared yourself? (1F)
			...my problem was more the fact that I was a parent of two children who have got the same syndrome... [ref Bentall graph] I don't want to put my children through this awful procedure (1D)
I also watched my brother go through the Bentall graph, which is quite a process. I mean, they broke it down, they put him on an artificial this and that...he's on Warfarin for the rest of his life and that's a big deal...when you're thinking about your kid (1D)			

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Theme	Sub-Theme	Evidence
Family (13)	Being a parent of a child with Marfan syndrome (6)	[1D and 1E = father and daughter] 1E: You always gave us pep talks didn't you? 1D: Well yeah...I needed to do my job...my job was to prepare you two... 1E: Yeah I was pretty scared.
		It's just natural as a parent though isn't it? Even more so when you know that you could suffer from the same thing. You just want to protect your child, don't you? (1F)
		In my particular situation it's probably quite different because I'm a single mum, so if anything happens to me, what's happening to my boys? That's my only concern. (1F)
	Having children (4)	...when it was in writing saying that I should definitely stop trying to conceive another child and...umm...have a meeting with your local consultant, that made me...that was quite, that was quite hard because I was really desperate to have a second child and...umm...so it was, you know, that was quite difficult. That was the only aspect of the whole thing about Exostent, the hardest thing just being told to hold off having another child until we figure out what's going on. (2C)
		...and so to be sat down by your consultant who said that you've actually got a ten percent chance of having a ruptured aorta in childbirth was very much like an ooh, ok, that's actually...ten percent's actually quite high. (2C)
		...the only time that I've really felt like it's really impacted on my life is when they told me about the complications that might arise if I were to have children...and, umm, that's always something I'd pictured myself doing so I think I was quite upset when they told me that (2B)
2B to 2F: ...it was really nice to read your patient experience online...umm, before I had the operation. JF: Was it reassuring hearing from someone else? 2B: Very, yeah definitely. JF: What about it did you feel was reassuring? 2B: Umm...just the success of...the success of both of your pregnancies and how the surgery had helped you to have that second pregnancy. Yeah, it was very reassuring.		
Feelings about the Exostent (21)	Relief (3)	When I found out about it and when I heard from Tal and he explained it to me, it was the greatest feeling. I thought I've really been saved and something wonderful is happening just when I wanted it to. That was a really massive...umm...good feeling. Obviously a great relief. (1D)
		I just remember being in a fantastic mood because it was all over and...all your worst fears which you had whilst signing the waiver form...they're all gone...so...yeah, it was just a great feeling that I've come through it and the worst is over now. And this thing that you've been worrying about for the last 15 years is gone. So...um...yeah, a great feeling. (1C)
		Oh, huge relief...huge. But, you know, it just all happened really quickly...it was just great, for me anyway. (2C)
	Optimism (2)	...and as soon as I found out about that procedure [the Exostent] I was...looking forward to it is the wrong word but...But it was just a good feeling of optimism the whole way. (1C)

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Theme	Sub-Theme	Evidence
Feelings towards the Exostent (21)	Optimism (2)	And it's future-proofing it, that's what I'd say (1D)
	Trust (1)	I was only the second one that had it so it will have pay back for future generations and for other people and I knew it was going to be alright, I just knew. I had to give it my best shot and I got as fit as I could and I was confident. That team that was around the project just gave me so much confidence. If you...that's the team you want around you when it happens. What could be the disadvantage about that? (1D)
	Positivity (8)	They explained exactly what it was, how it works, and what the pros and cons were, and there didn't seem to be any cons, it was a win-win situation because, ok, it's a new procedure, but if it didn't work then there was always the fall back of the Bentall's operation...(2C)
		There's nothing to lose. (2A)
		So, in my mind – obviously my parents were a slightly different generation, were like, are you sure, it's a completely new operation – so I'm like, well, somebody's got to be first! And it was a case of, in my mind, it was a win-win situation. It meant, as [2A] said, you didn't have to have Warfarin afterwards, you didn't have to be on a by-pass machine during...just the whole thing was a lot less invasive. (2C)
		Yeah, I feel the same, a bit safer in the things that I'm doing. Yeah it's nice to know that I have it, it's a nice feeling. (2B)
		Umm...I have to say that there's so many positive things about doing it, which we've kind of discussed today since we've been here...so the no drugs thing, the fact that it's not the last resort – there's always something else that you can do if it doesn't work, and so from that point of view, if you are eligible to have it done, and you should have it done and then you should go ahead and do it because you've got nothing to lose really...as far as I can tell, I'm not an expert obviously. From my point of view, I can't see what the downside is. (2A)
		And you can keep all your own organs, that's the beauty of it. (2C)
		I just think it's amazing. It's such a simple idea that saves all that grief of having to have that major surgery - invasive surgery. I do hope that they can use it for other conditions and not just Marfan. (2C)
		I don't get that passionate about a lot of things but I am passionate about this...so I'm really keen that more and more people hear about it, because it has been such a positive experience for all of us. (2C)
	Pragmatism - 'Getting it over and done with' / 'Just getting on with it' (7)	For me, because I had it done between finishing school and starting college over the summer...so for me it was a case of...I wasn't really that anxious about it, it was more just getting it out of the way (1B)
		so it was just a case of having the operation and being able to go to college. I was more, like, wanting to get it done than anxious. (1B)
		Again, wasn't particularly phased by it...umm...it was just kind of part of just getting it over and done with really (2A)
		But I remember thinking I just want to get on with it now, because it was just like treading water waiting. (2C)

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Theme	Sub-Theme	Evidence
Feelings toward the Exostent (21)	Pragmatism - 'Getting it over and done with' / 'Just getting on with it' (7)	I just knew it had to be done so get it over and done with and then I'd be ok. (1E)
		While you're young and fit, have it done while you're young and fit. And then if you're not worried, or if you are worried, it means that you don't have to be worried. It was just about getting it done really. (2A)
		So it was like, let's just do it now and get it over and done with (2A)

