RESEARCH DISSERTATION

The Advanced Human Capital Program, of the National Commission for Scientific and Technological Research (CONICYT), has sponsored this student´s MSc Programme

Mental Health Studies Programme

Student ID: 1350153

Module Title & Code: Research Dissertation, 7PAGMRES

Date: August 25th, 2015

Word count: 10.328

PROJECT TITLE: Relative values placed by people with Marfan syndrome and genetically determined life-threatening aortic root aneurysms on the lifetime implications of time and choice amongst the three currently available forms of prophylactic operation.

ABSTRACT

Background: Marfan syndrome is a heritable disorder of the connective tissue, which affects several biological systems. However, the main hazard for those with this syndrome is that they are prone to aortic dissection. To avoid this risk, prophylactic surgery is suggested. Nonetheless, choosing between available surgery options might be challenging as they have specific flaws. **Aim:** The main objective of this study was to determine relative values placed by people with Marfan syndrome, and genetically determined life-threatening aortic root aneurysms, on the lifetime implications of time and choice amongst the three currently available forms of prophylactic operation. **Methods:** A mixed design was chosen for this work. Then a questionnaire based on the Ottawa Decision Support Framework' values survey was created. In total, 60 participants took part in this study, on a voluntary basis. **Results:** Similar preferences for all questions were found across both genders. However, a significant, yet negative, correlation was found for the same questions among "medical practitioners" and "people with Marfan and/or immediate family", i.e. "people with Marfan and/or immediate family" expressed significantly more polarised preferences than medical practitioners over the choice between "postponing the operation" or "get it on with it and have it behind you". In addition, anticoagulation avoidance was evaluated as to its importance to the group "people with Marfan and/or immediate family", regardless of age and gender differences. Moreover, preferences over the importance of having no noise from the heart valve did differ significantly between the "people with Marfan and/or immediate family" (49 - 60 years old) and those "medical practitioners" of the same age. From the thematic analysis, twenty-two themes, which support the results mentioned above, emerged. **Conclusions:** It is clear that clinicians could have a better insight into the unique preferences of those with Marfan syndrome, especially in the context of medical decision-making between different treatments. If patient's personal values are taken into consideration regarding procedures and screening decisions, it might better meet their individual priorities, needs, and desires.

TABLE OF CONTENTS

1 Introduction

In 1896 a French pediatrician named Antoine Bernard-Jean Marfan was the first who described the Marfan syndrome (Van De Velde , Fillman & Yandow, 2006). This syndrome is an "autosomal dominant, multisystem disease caused by mutations in the FBN1 gene" (Judge & Dietz, 2005). Marfan syndrome can be characterized by ocular, skeletal and cardiovascular difficulties (Castellano , Silvay & Castillo, 2013). Nevertheless, the major risk for those with this syndrome is that they are predisposed to aortic dissection (Chiu , Wu , Chen , Kao, & Huang, 2014). To overcome this threat, prophylactic root replacement is recommended. Notwithstanding, it might be hard to choose the more suitable surgery, as all the available surgery options have their pros and cons (Treasure , Golesworthy , Pepper , Ruiz, & Gallivan, 2011).

In the context of Marfan syndrome, poor systematic studies have been conducted in respect to patient's values as well as their preferences amongst the three treatments available. It can be argued that, this situation is based on the belief that clinicians and patients share the same physical and psychological goals. Unfortunately, sometimes, clinicians can be unskillful judges about patients' preferences (Kennedy et al., 2008). As a result, this research aims to explore: "relative values placed by people with Marfan syndrome and genetically determined life-threatening aortic root aneurysms on the lifetime implications of timing and choice amongst the three currently available forms of prophylactic operations".

This research is divided into seven parts plus an Appendix. The first section discusses the literature regarding Marfan syndrome and the decision-making process in the medical field. The second section covers in detail the methodological perspective used for this study, a mixed design. The third section continues with the main findings from the data analysis and the thematic analysis, respectively. The fourth section consists of the findings discussions. The fifth section presents the strengths and weaknesses of this study. The sixth section provides future directions for practice. The final section presents the conclusion of this study. In addition, the appendix includes tables and graphs, which complement the data analysis.

1.1 Marfan syndrome

Marfan syndrome is "an autosomal dominant multisystem, connective tissue disorder, with primary involvement of the cardiovascular, ocular, and skeletal systems" (Castellano et al., 2013). Indeed, Marfan syndrome is the most prevalent inheritable disorder of the connective tissue found in the population (Loeys, 2014). The principal characteristics displayed by people with Marfan syndrome are: narrowness of the long bones, tall stature, long slender limbs, decreased skeletal muscle mass and scoliosis (Romaniello et al., 2014).

At the same time, people with Marfan syndrome face the challenging and hard decision of starting a family. Apart from general factors related to having a baby, during pregnancy and childbirth, patients with Marfan syndrome tend to deal with additional health complications (Mulder & Meijboom, 2012). Indeed, children born to parents with this syndrome have 50% chance of inheriting the condition (Dean, 2007).

Unfortunately, this group of people is affected by other factors, aside from pregnancy related problems. Social life can be also diminished, especially recreational activities as well as daily routines. In fact, De Bie , De Paepe , Delvaux , Davies, and Hennekam (2004) research sustain that Marfan syndrome impacts diverse aspects of people's everyday life.

Nonetheless, Loeys (2014) sustains that the major difficulty faced by these individuals is the predisposition for aortic root aneurysm and aortic rupture. Aortic root aneurysm is the enlargement of the aorta, which is the largest blood vessel in the body (Milewicz , Dietz & Miller, 2005). Although Marfan syndrome affects several systems, cardiovascular complications are considered the most common cause of death.
Murdoch , Walker , Halpern , Kuzma, and Mckusick (1972) explained that there is a progressive dilatation of the aorta, which can lead to its dissection and rupture. Almost 95% of the people with Marfan syndrome will develop a dilatation of ascending aorta by

the age of 60 and patients under 30 years of age will be affected by other complications (Romaniello et al., 2014).

To prevent this outcome, Romaniello et al. (2014) explain that annual echocardiographic monitoring is suggested to people with Marfan syndrome to detect changes in size of their aorta. If this examination proves that there is aneurysmal dilation of ascending aorta, it might be an appropriate time to consider a prophylactic operation according to established consensus based criteria (Vahanian et al., 2012). Moreover, the recent review, presented by (Treasure , Takkenberg & Pepper, 2014) suggest that the most important life-prolonging treatment for aortic root aneurysm is a prophylactic operation.

1.1.1 Available forms of prophylactic operations for people with Marfan syndrome

In many health care scenarios, two or more medically convenient alternatives are available. Still there is no agreement that one option is definitely more appropriate than the other one (Llewellyn-Thomas & Crump, 2013; Wennberg, 2010). Unfortunately, the medical context of aortic root aneurysm in people with Marfan syndrome is not the exception.

In the case of "effective" medical care options, the advantages far exceed the possible disadvantages. The main goal here is to encourage patient's uptake. However, in the situation of "preference-sensitive" medical care alternatives, the decision between advantages versus harms is valued by the patients themselves (O'connor et al., 2007). Unluckily, sometimes the patients hold non-realistic expectations of treatment gains and losses. Besides, according to O'connor et al. (2007) "clinicians are poor judges of patients' values". As a result, there might be an overestimation of treatment alternatives, which are not valued by informed patients.

Presently, for people with Marfan syndrome, there are three forms of operations available to reduce the risk of sudden deaths, especially in patients with congenital aortic root aneurysms.

One of them is total root replacement (TRR) or the Bentall method, which involves removing and replacing both: the ascending aorta and the aortic valve, now is performed with a composite prosthesis (Bentall & De Bono, 1968). The drawback of this operation, however, is that "patients are committed to a lifelong risk of valve related thromboembolism and an accompanying fear of bleeding from the anticoagulation required to minimise that risk" (Treasure et al., 2011).

The second one is known as the David valve-sparing root replacement (VSRR). The aortic valve is conserved, anticoagulation is not required and, in particular, it allows the patient to go through pregnancy and delivery without any complications regarding anticoagulation. This operation offers freedom from anticoagulation and attendant risks of bleeding. Nevertheless, patients might require another operation at some point in their lives

Lastly, the third option is a personalised external aortic root support (PEARS). This operation involves placing a mesh support around the aortic root (Treasure et al., 2011). Patients having PEARS do not require anticoagulation or another operation in the short-term period. However, the long-term results have not been determined as the average follow-up is under 5 years (Treasure et al., 2014).

The decision upon which prophylactic operation to undergo is complex and determined by a trade-offs between the competing risks of each option. Lee , Low and Ng (2013) sustain that personal values influence patient's decisions but there is "lack of clarity and attention on the concept of patient values in the clinical context''. Moreover, it can be observed that individuals with a given impairment (for example: Marfan syndrome) rate their expected quality of life somewhat higher than those actually living without that impairment. Therefore, physicians should help patients to be capable to form reasonable assumptions about their forthcoming quality of life so that the patients make competently informed decisions (Halpern & Arnold, 2008). Indeed, the fact is that patients usually choose between alternative treatments with similar consequences on mortality, but very distinct impact on their lives (Litwin et al., 2007; Weeks et al., 1998).

1.2 Values and preferences in healthcare

The usage of the word "value" is documented in the Oxford English Dictionary, as "the importance, worth, or usefulness of something" and to the "principles or standards of behaviour; one's judgement of what is important in life" (Stevenson, 2010). In order to fully understand the difference between the meanings of the concept of value, it is necessary to see it as a noun and a verb.

On the one hand, "values" as a ***noun form*** suggests that it should be explored from the viewpoint of the entity being assessed or from the viewpoint of the individual doing the evaluation (Feather, 1995; Rohan, 2000). Indeed, Feather (1992) highlights that values are abstract systems that implicate the beliefs that individuals hold about advantageous behaviour manners or about advantageous outcomes (Feather, 1995). According to this statement, it can be argued that values as ***nouns*** imply "what" is important to the person (Rohan, 2000).

On the other hand, "values" as a ***verb form*** suggests that some appraisal has taken place. For example, when individuals express that they value (verb) a situation or an outcome, they are declaring a profound connection to that matter (Rohan, 2000).

Furthermore, values as ***verbs*** are related to a particular context and to a present lapse of time (Lewin , Heider & Heider, 1936). In addition, values bear a strong connection with emotions. Hence, values would influence the preferences that an individual makes among different options (Feather, 1995).

According to this statement, it can be argued that values as ***verbs*** imply "how" important something is for the individual.

Apart from the usage of values as ***nouns*** or ***verbs***, the Ottawa Decision Support Framework, offers a complementary approach (Edwards & Elwyn, 2009; Stacey et al., 2008). From this point of view, values are not considered as nouns or follow Feather's conceptions. On the contrary, values are referred to as "a person's informed attitudes about the relative desirability/undesirability or individual importance of a health care

option's unique characteristics, which include that option's protocol, possible benefits, and potential harms (Llewellyn-Thomas & Crump, 2013; O'connor, 2012).
Additionally, preferences fall into the "person's overall most-favoured option, after taking into account his or her attitudes toward each option's detailed characteristics" (Llewellyn-Thomas & Crump, 2013).
For the purpose of this research, values will be used as ***verbs*** and according to the Ottawa Decision Support Framework.

1.2.1 Patients´ values

Though it is well known that personal values affect patients' decisions; and that an active attention is placed on patients' values in evidence-based medicine, an absence of certainty and consideration on the idea of patient values in the medical field is still observed (Elwyn et al., 2012; Makoul & Clayman, 2006; Straus , Richardson , Glasziou, & Haynes, 2011).
Certainly, it should be noted that patients' values definitions are usually very ambiguous or too narrow. For example: "patient values are the features that matter most to patients" (Elwyn et al., 2006) or ''the unique preferences, concerns, and expectations each patient brings to a clinical encounter and which must be integrated into clinical decisions if they are to serve the patient''(Straus et al., 2011).

For the moment, most researches on patients´ decision-making have concentrated on perceptible patient outcomes or enhanced patient involvement in the time of consultations (Couët et al., 2013). Therefore, there is a lack of research based on how patients, in fact, choose among given options. As a result, patients´ values and preferences might be omitted from the duologue (Plos-Editors, 2007, 2009). However, it is discovered that patients explain clinical information and make decisions about treatment options using values as filters (Karel, 2000; Reyna, 2008). Indeed, according to Higgins , Idson , Freitas , Spiegel, and Molden (2003) "there is no more important variable in motivation and decision-making than value".

1.2.2 What people with Marfan syndrome value?

Firstly, according to Treasure and Pepper (2015) anticoagulation avoidance is considered an important issue for young patients, as it still gives the possibility of getting pregnant, practicing sports actively, and having a normal lifestyle.

Apart from this, previous studies have shown that anticoagulation using Coumadin/Warfarin was appraised as a "small price to pay" for some patients. Nevertheless, a study carried on by the department of Health Psychology at King's College London (KCL) has discovered that people with Marfan syndrome, strongly and consistently, manifested their interest in avoiding anticoagulation (Fosbraey, 2014). This response to anticoagulation might be explained by the fact that daily medication and permanent blood test procedures emphasise the "impaired" condition of those with Marfan syndrome, while they just want to feel as "normal" as everyone else (Fosbraey, 2014).

Secondly, Fosbraey's study (2014) have established that it is considered an advantage to have a root replacement sooner; mainly because it implies a reduction of the anxiety levels associated with routinely echocardiographic monitoring to detect changes in size of the aorta. Finally, this research also highlighted the great importance of reducing the number of medical appointments and medicaments (Fosbraey, 2014).

1.3 Medical decision-making process

Medical decision-making proceedings have experienced variations as service users and physicians cooperate to make challenging healthcare decisions. Nowadays, a physician does not take exclusive accountability in the process. Moreover, patients should not feel abandoned in making significant decisions about medical treatments (Haward & Janvier, 2015). Despite the fact that current clinical decisions are shared-decisions, substantial differences can be found in the ways of how people make decisions. Therefore, shared-decisions do not imply that patient and physician will share the same decision-making approach.

In health care, personal treatment and screening choices are usually preference-sensitive, containing significant trade-offs. For example, accessible alternatives can be equal in terms of clinical effectiveness; however, it is hard to comprise trade-offs between quality and length of life or between comfort and, potential, effectiveness of the procedures. Moreover, "treatment and screening decisions can be made so that they best fit an individual, with his/her unique situation, needs and desires" if patient's personal values are taken under consideration (Epstein & Peters, 2009; Stiggelbout et al., 2012).

Yet, putting this ideal into practice is challenging because of two main reasons. Firstly, with different diseases, it has been observed that clinicians might be imprecise at appraising patients' values and treatment options regarding health circumstances (Brothers , Cox , Robison , Elliott, & Nietert, 2004; Fraenkel , Bogardus , Concato, & Wittink, 2004). Nonetheless, it is important to mark out that this statement has not been probed specifically in Marfan syndrome. Secondly, although several patients face preference-sensitive decisions full of strong emotions, their personal values and desires are frequently labile. Therefore, it is best to clarify personal values first, in order to improve the decision-making process (Simon , Krawczyk , Bleicher, & Holyoak, 2008).

This clarification procedure can be complex, due to the fact that possible outcomes and risks can be challenging to communicate and visualize. Additionally, available alternatives regularly have trade-offs that makes them incomparable (Epstein & Peters, 2009; Kerstholt , Van Der Zwaard , Bart, & Cremers, 2009).

1.3.1 How do patients make decisions?

In the beginning, cognition was contemplated as a "cold" analytical reasoning process. Hence, it was stated that human beings were capable of making efficient decisions that enhance any given result. This approach was called rational optimisation (Gutenstein, 2014).

However, choosing among different options cannot be achieved in an entirely rational form, by callously comparing evidence. Decisions also compromise emotions and decisions are more than an equation of relative advantages of diverse feasible choices. In fact, it is seen evident steady divergences from rational theory when examining

clinical decisions (Haward & Janvier, 2015). To resolve such discrepancies, (Kahneman & Tversky, 1979; Tversky & Kahneman, 1981) elaborated the *Prospect Theory*. Despite the fact that the *Prospect Theory* was developed in the 70's, it is still used worldwide in different fields. Moreover, in 2002, Kahneman and Tversky were awarded the Nobel Prize (Haward & Janvier, 2015).

As it was stated previously, over a long period of time, academics believed that strong decision-making was a purely logical process. Nonetheless, recent paradigms advocate that complicated and vital decisions are usually taken under circumstances of strong feelings and these feelings should be recognised and accepted as important, advantageous decisional features (Janvier , Lorenz & Lantos, 2012).
The *Prospect Theory* proposes that a result is understood as "a gain or a loss based on the decision-maker's reference point and decisional frame", summing a subjective element to an objective value of a result (Kahneman & Tversky, 1979). In accordance with Tversky and Kahneman (1981), when consequences are contemplated gains, people tend to evade risks. When consequences are contemplated losses, people tend to make riskier alternatives. Without exception, it is known that the judgement of gains or losses is influenced by how information is transmitted or framed (Haward & Janvier, 2015).

1.4 Aims of the current study

Being aware of how values affect individual decision-making is especially significant to preference-sensitive decisions when one single best alternative does not simply exist and where trade-offs are inevitable (Lee et al., 2013). According to Elwyn et al. (2012), clinicians should encourage their patients to be aware of "how they value key aspects of the decision with which they are faced". This suggestion is established in the conviction that, by clarifying patients' values, doctors will be able to offer more thoughtful medical treatments according to each patients' personal choices and treatment targets (De Vries , Fagerlin , Witteman, & Scherer, 2013; Llewellyn-Thomas & Crump, 2013).

However, it has been put under certain amount of a doubt if physicians are capable to make or offer the decisions that service users would like (Zikmund-Fisher , Sarr , Fagerlin, & Ubel, 2006). In fact, "the perspectives of people for whom research on treatment really matters is service users", and it has been shown during the last decade that doctors and patients may differ regarding what is considered as a good result (Rose , Evans , Sweeney, & Wykes, 2011).

Although doctors´ role is to advice patients, so that they can make the "best" choice. However, sometimes clinicians and patients may not share the same decision-making course. Doctors might accentuate possible outcomes because of their role and a lower social connection with the patient. On the contrary, patients might emphasise how results are accomplished (Zikmund-Fisher et al., 2006). Overall, if patients' values and preferences are considered in research, it is more conceivable to produce useful outcomes that can enhance clinical practice (Lee et al., 2013).

Regarding the specific situation of people with Marfan syndrome, what is already known comes from a series of 'focus groups' in a project lead by Fosbraey (2014) and King's College London , which was ran from 2012 to 2014 in order to explore the psychosocial impact of undergoing personalized aortic root support (PEARS). The outcomes from this research stressed the participants desire to just be normal. Apart from a decrease of testing and medicines, people with Marfan syndrome hope for a full, active, and normal life. The results should not have come as a surprise but the force and consistency did.

Nowadays, the aim of this project is to broaden Fosbraey´s research as long as to explore what different people value the most. According to the above, the following goal is pursuited:

- To explore relative values placed by people with Marfan syndrome and genetically determined life-threatening aortic root aneurysms on the lifetime implications of timing and choice amongst the three currently available forms of prophylactic operations.

2 Method

2.1 Methodological Approach

A mixed design was chosen for this work (Plos-Editors, 2007) for two reasons. Firstly, due to the deficit of research on values, expressed by people with Marfan syndrome. Secondly, considering that a mixed design would go along with the exploratory nature of this study.

2.2 Conceptual Framework

The conceptual framework implemented in this survey is the Ottawa Decision Support Framework (ODSF). In the area of health-related decision-making, the benefits of the ODFS is that it allows to identify "decisional needs as values" (Lee et al., 2013) and that it "applies to all participants involved in decision making, including the individual, couple, or family, and their health practitioner" (O'connor, 2012).

This frame of reference is based on fundamental theories and methods, making it possible that the key elements in health care decision-making may be operationalized. Likewise, the ODSF is effective in the "development of interventions supporting health decision making in the context of uncertainty" (Légaré , O'connor , Graham , Wells, & Tremblay, 2006).

The ODSF sustains that contestants´ decisional needs will impact decision outcomes´ quality (e.g. values-based choices), which in return will influence behaviours, emotions and results, and, hence, an adequate use of health assistance programs (O'connor, 2012).

2.3 Sampling

Heterogeneous sampling: The participants were chosen from a group of volunteers who showed their interest in being involved in this survey. In addition, the sample was based on a wide range of various characteristics such as: gender, age, and external aortic root prophylactic operation, among others. A heterogeneous sampling was preferred as it allows to capture as many different perspectives as possible. Indeed, the aim of this approach is "to identify central themes with cut across the variety of cases or people" (Ritchie , Lewis , Mcnaughton-Nicholls, & Ormston, 2014).

2.3.1 Inclusion and exclusion criteria

Group 1:

- People with Marfan syndrome who are aware that a prophylactic aortic root operation may be advised for them.
- Alternatively, people with Marfan syndrome who have had a prophylactic aortic root operation.
- Alternatively, immediate family, of those with Marfan syndrome, who have witnessed the decision making process.

In Group 1, the three options could be mutually inclusive. Meaning, for example, that participants could be people who have had a surgery and, at the same time, they could be members of immediate family of someone with Marfan syndrome. This rule applies only for members of group 1. For the purpose of this research, this group is named **"people with Marfan and/or immediate family".**

Group 2:

- Medical practitioners who advise in specialised clinics.

For the purpose of this research, this group is named **"medical practitioners".**

2.3.2 Participants' characteristics

The sample consists of 78 participants who kindly agreed to take part in this study, on a voluntary basis, and who were, therefore, approached for the exploratory analysis by the head of this research: a supervisor and a medical doctor.
Seventy-eight emails, including a consent form and an information sheet, were distributed, after which seventy-eight participants started the online questionnaire. It is important to mention that the researcher did not have any direct contact with the participants before, during, or after the study. The supervisor of this project sent the emails and provided the data set. Moreover, the whole data collected was anonymous.

However, the data and thematic analysis presented in this study were conducted fully by the researcher, who organized the data, removed the incomplete information, and obtained sixty suitable questionnaires for the analysis.

2.4 Data collection

As a part of this investigation, the researcher designed this survey based on the Ottawa Decision Support Framework ´ values questionnaire.
The questionnaire is made up of 8 questions based on a ranking scale, together with open-ended questions. The ranking scale technique was selected due to its practical applicability. In addition, it was chosen because it is a powerful tool in the medical care field. This scale is also considered to be helpful as it "measures a value function that reflects strength of preference" (Bleichrodt & Johannesson, 1997).

The 8 questions were based not only on previous studies but also on the opinions of people with Marfan syndrome and medical practitioners. This group of experts identified common key patterns that patients had to face before or right after they made the decision of having a prophylactic operation.

The participants were asked to rate each given question on a scale from 0 to 10, where 0 corresponded to "Not at all important to me" and 10 referred to "Extremely important to me". Additionally, each question included an open-ended question as the following: "please write in any thoughts of your own about this question X". It was defined that for the sake of this research only the raking would be mandatory. Therefore, the open-ended questions were optional.

2.4.1 Validity of the questionnaire

Validity indicates if a research tool is measuring what it aims for. Content validity (or face validity) provides with expert judgements; whether the questionnaire exemplify the proposed concepts the way it is expected and the way it should be measured (Ritchie et al., 2014).

As for the quality of the questionnaire, content validity was assessed through a rigorous analysis carried by a group of experts, medical practitioners, and people with Marfan syndrome before the questionnaire was sent.

2.5 Data analysis

The online questionnaire was analysed descriptively with the help of Excel and the Statistical Package for the Social Sciences (SPSS) software. Additionally, as the data was skewed, non-parametric testing was used.

Likewise, the open-ended questions in the data sheet were examined with the Nvivo software version 10.2.1, through a thematic analysis approach. According to Braun and Clarke (2006), this method allows to classify, to evaluate and to address patterns or "themes" within data. Moreover, Braun and Clarke (2006) sustain that the advantage of using thematic analysis in order to complement a quantitative analysis, is that this approach is very flexible. Through its analytical freedom, this technique provides plasticity and utility, which can produce a broad and specific, yet elaborated, report of evidence.

For the purpose of this survey, “themes” are defined as entities that express something meaningful about the data set associated with the research question as well as serve as levels of patterned answers. Therefore, the quality of a theme does not necessarily rely on quantitative frequencies “but rather on whether it captures something important in relation to the overall research question” (Braun & Clarke, 2006).

Following Braun and Clarke (2006) guidelines, the steps for the thematic analysis were:

I. Phase 1: familiarizing with the data
II. Phase 2: generating initial codes
III. Phase 3: searching for themes
IV. Phase 4: reviewing themes
V. Phase 5: defining and naming themes
VI. Phase 6: producing the report

3 Findings

The eligible group of participants included 78 volunteers. Sixty volunteers participated in this study (77%). Eighteen participants started the online questionnaire, however, did not complete it.

As for the whole sample, 73% of participants were "people with Marfan and/or immediate family" and 27% - "medical practitioners", who fully completed the online questionnaire. Additionally, approximately one third of females (27%) and 73% of males participated in the survey altogether.

Table 1 illustrates the participants' ages in this study.

Table 1

Age	People with Marfan and/or immediate family	Medical practitioners
< 37 years old	11%	6%
37 - 48 years old	25%	31%
49 - 60 years old	36%	56%
> 61 years old	27%	6%

3.1 General findings

Questions 1, 2, 3, and 6 exhibit a **mode of 10.** Question 4 reveals **two modes, 0 and 10**, whereas questions 5 and 7 display a **mode of 0**. Meanwhile, question 8 holds a **mode of 5.**

Along with these results, it can be noted that the replies in a range from 10 ("Extremely important to me") to 0 ("Not at all important to me") were provided to most of the questions. Table 2 illustrates the general frequencies.

Table 2

General frequencies

		1. How important is it for you to postpone having an operation on your aorta for as long as the doctors think it is safe to do so?	2. Is taking anticoagulant (blood thinning) medication an important problem for you?	3. How important is it to you, if you need to have an operation on your aorta, to get on with it and have it behind you?	4. Is taking lifelong medication such as beta blockers or losartan an important problem for you?	5. Are repeated visits to the hospital to have echo tests an important problem for you?	6. How important is it to you to have a physically active lifestyle?	7. Would anticoagulation which might complicate pregnancy and make having a baby more difficult be important in your choice of operation?	8. How important is it to you to have no noise from your heart valve?
N		60	60	60	60	60	60	60	60
Median		7	8	8	4	2.5	8	7	5
Mode		10	10	10	1*	0	10	0	5
Minimum		0	0	0	0	0	1	0	0
Maximum		10	10	10	10	10	10	10	10
Percentiles	25	3	7	5	1	1	7	3.25	3
	50	7	8	8	4	2.5	8	7	5
	75	9	10	9	7	7	10	9	8

*Multiple modes exist. The smallest value is shown

Additionally, table nº 3 shows the general frequencies per each question asked.

Table 3

General frequencies per question

	Q1	Q2	Q3	Q4	Q5	Q6	Q7	Q8
	Percent							
0 = "Not at all important to me"	8	2	3	13	18	0	22	13
1	3	0	0	15	13	2	2	0
2	8	5	5	7	18	0	0	7
3	15	5	5	13	8	3	2	8
4	3	3	3	5	2	0	2	8
5	7	7	12	7	8	8	13	17
6	0	0	2	5	5	5	2	5
7	17	13	13	13	5	10	10	8
8	10	17	22	3	15	25	15	15
9	7	15	12	3	2	12	13	5
10 = "Extremely important to me"	22	33	23	15	5	35	20	13
Total	100%	100%	100%	100%	100%	100%	100%	100%

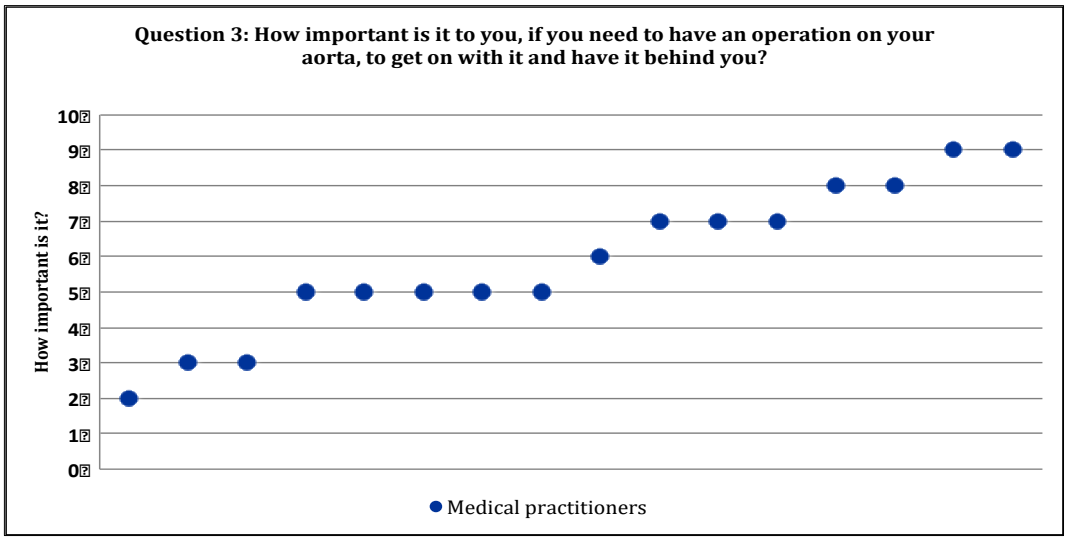
For the results given by the groups of "people with Marfan and/or immediate family" and "medical practitioners" upon the questions, consult **appendix 1.**

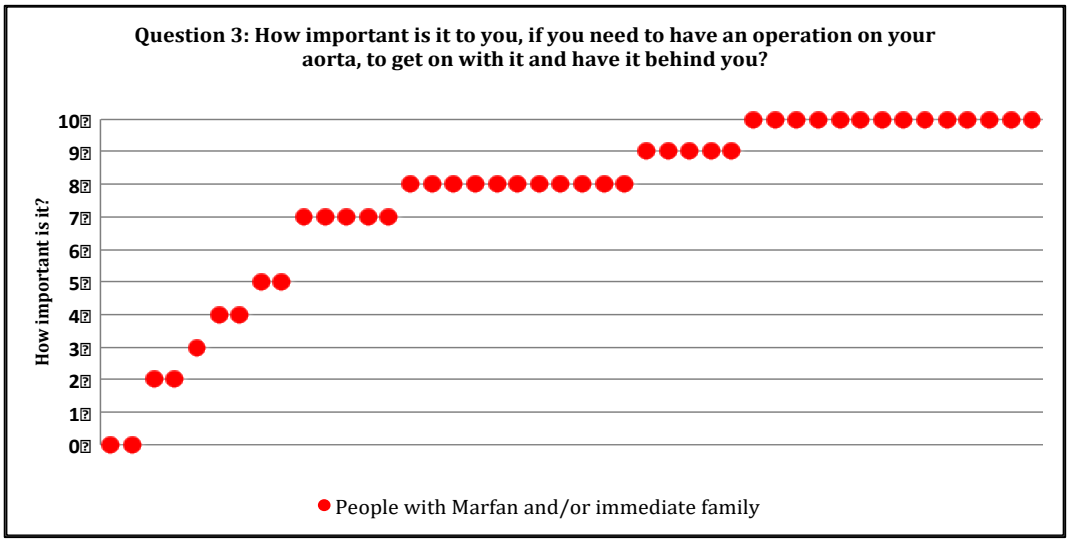
3.1.1 Findings per group: "People with Marfan and/or immediate family" versus "Medical practitioners"

Due to the skewed tendency of the data, Mann-Whitney Test (a non-parametric test) was chosen to compare two independent samples. This test assumes that the responses are ordinal and the alternatives are restrained to a rank. Therefore, the significance can be explained among medians differences (Field, 2013).

According to the Mann-Whitney Test, "people with Marfan and/or immediate family"´ preferences did not differ significantly from "medical practitioners" in questions 1, 2, 4, 5, 6, 7, and 8. However, in **Question 3,** "people with Marfan and/or immediate family"´ preferences (*Mdn*= 8) **do differ significantly** from "medical practitioners" (*Mdn*= 5.5), *U*= 189.5, *z*= -2.756, *Sig. (2-tailed)*= 0.006 ($p < .01$) and *r*= -0.36. This *"r"* means a medium size effect.

In order to support the above analysis, two dot plots were generated to highlight the score distribution in both groups in more detail.



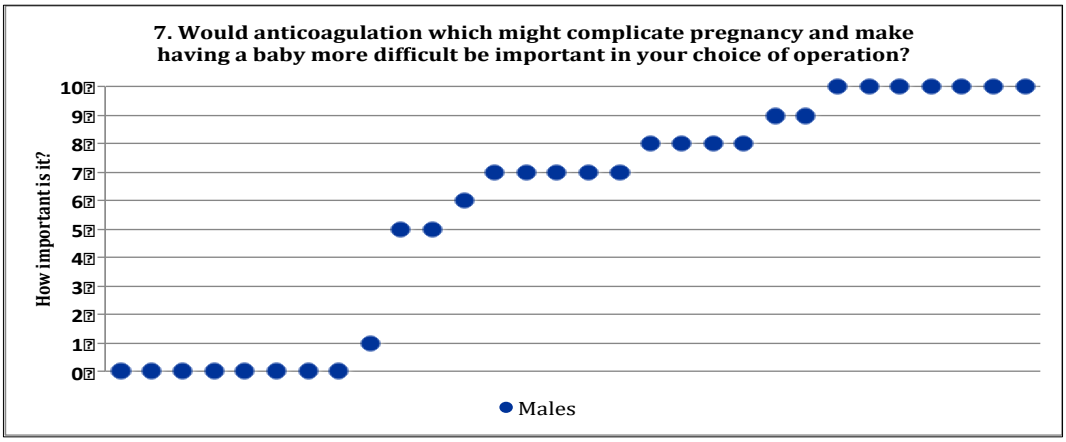


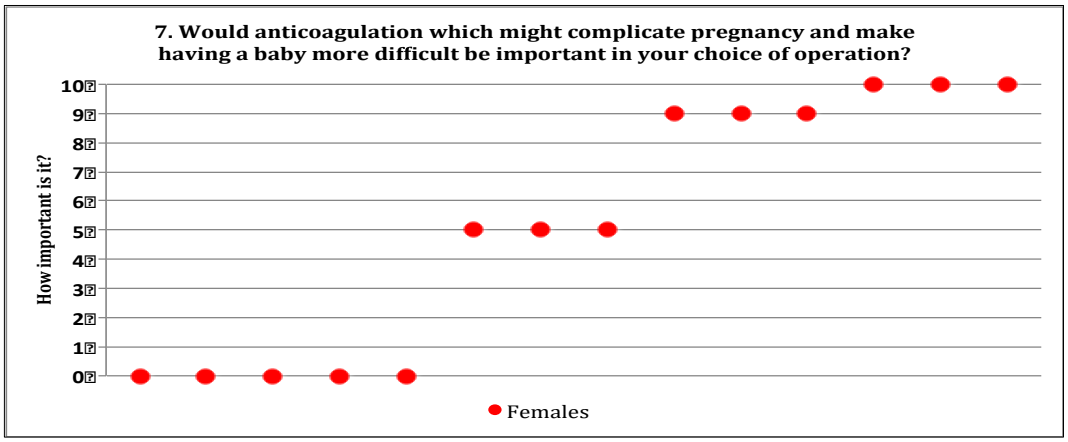
3.1.2 Findings per group: Males versus Females

According to Mann-Whitney Test, the distribution per each question in the group of patients is the same across males and females. *To see the Mann-Whitney Test results per question, check **appendix 2.**

However, and for the purpose of this research, it is important to take under account the distribution of male and female answers to question nº 7. This decision is based on the literature review mentioned previously that suggests that pregnancy might be a problem for women with Marfan syndrome. Apart from being an issue for women with Marfan syndrome, it also could be a problem for their partners and/or immediate family.

In order to support the above analysis, two dot plots were generated to highlight the score distribution between males and females in the group of "people with Marfan and/or immediate family", in more detail. See **appendix 3** for the frequencies between genders.

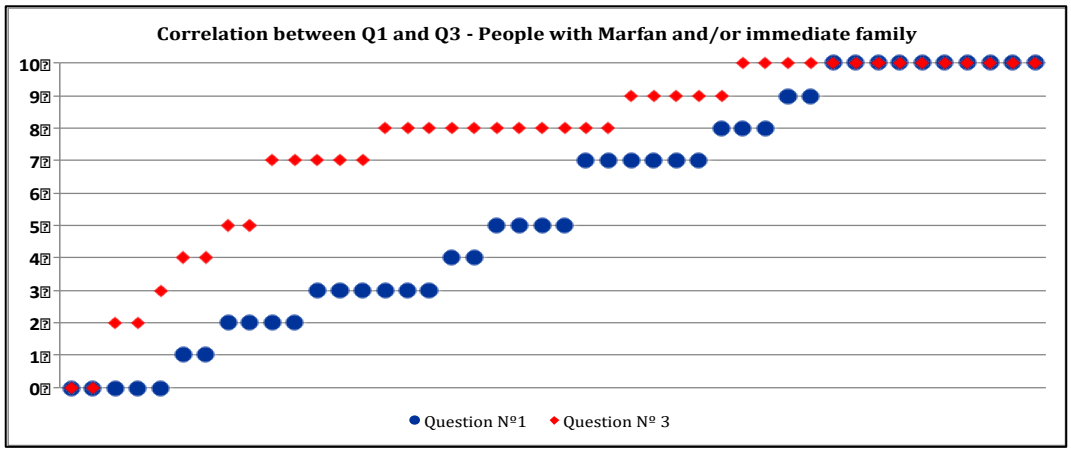


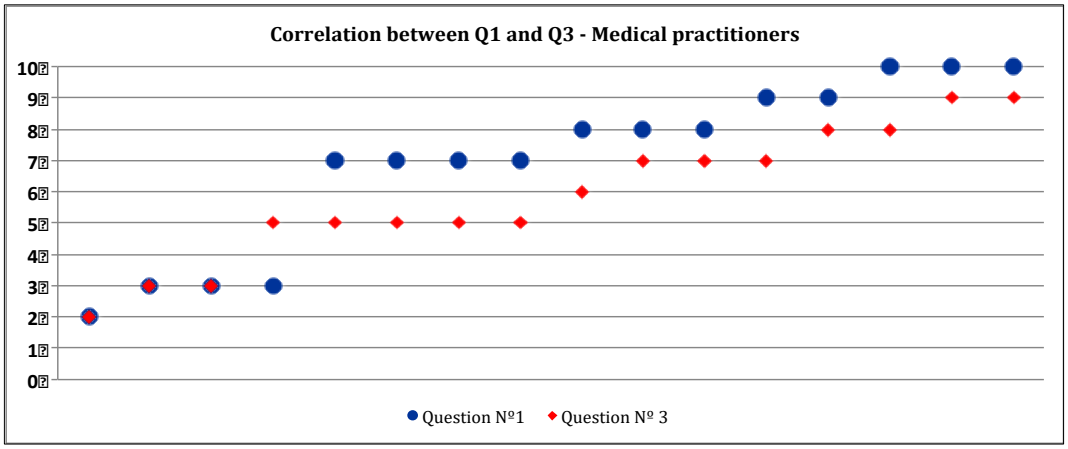


3.1.3 Results obtained by question comparison

In order to achieve a more profound analysis, a Spearman's rank-order correlation was run to determine the relationship between question nº1 and nº3. This was made to explore discrepancies or similarities between the options: to postpone the surgery and to get on with it and have it behind you.

On the one hand, there is a significant, yet negative, correlation between preferences in question nº1 and question nº3 for the group of "people with Marfan and/or immediate family", r_s= -.30, p (2-tailed) < .05. This result means that the higher the preferences for question nº1, the smaller are the preferences for question nº3. However, there is not a significant relationship between preferences in question nº1 and question nº3 in the group of "medical practitioners". In order to support the above analysis, two dot plots were generated.



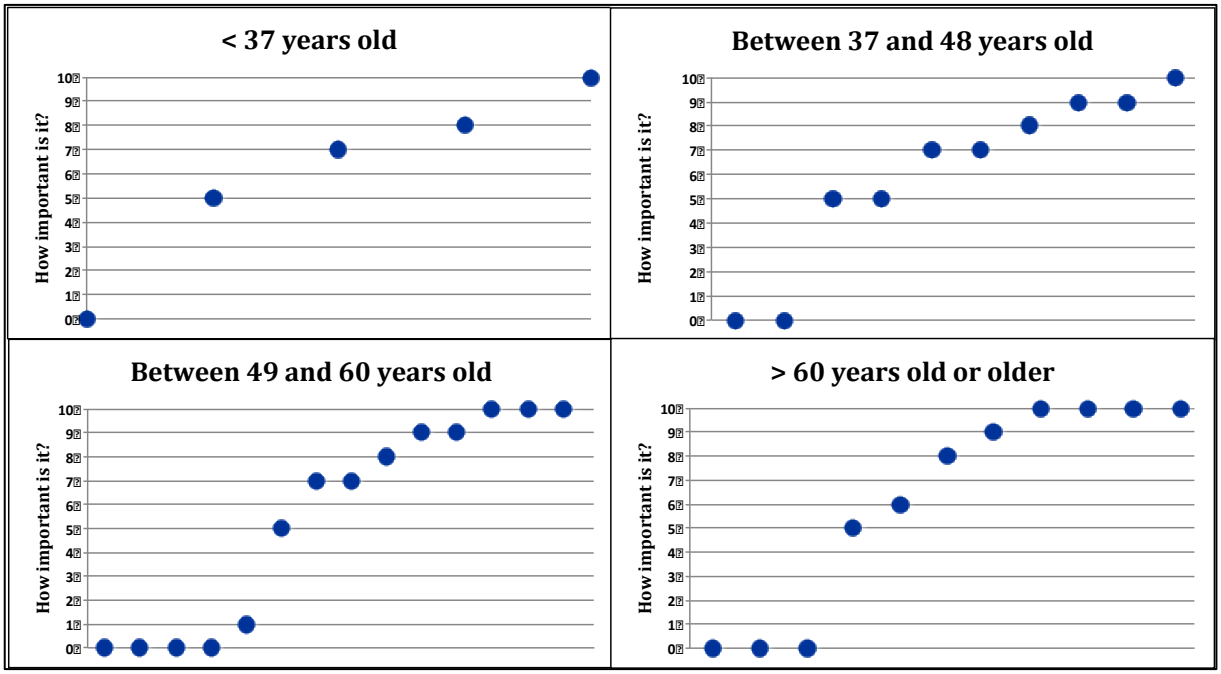


For the positive correlations found in the groups of "people with Marfan and/or immediate family" as well as "medical practitioners" upon the questions, consult **appendix 4.**

3.1.4 Findings per people with Marfan and/or immediate family's ages in question nº 7

Pregnancy, usually, is a matter that has an age limitation. Therefore, question nº7 was analysed taking under account the patient's age as well as focusing on reply distributions.

Question 7: Would anticoagulation, which might complicate pregnancy and make having a baby more difficult, be important in your choice of operation?



In order to explore if there was a statistical relationship between these graphs results, the Kruskal-Wallis Test (a non-parametric test) was chosen to compare several independent samples. In the same line as the Mann-Whitney Test, Kruskal-Wallis Test

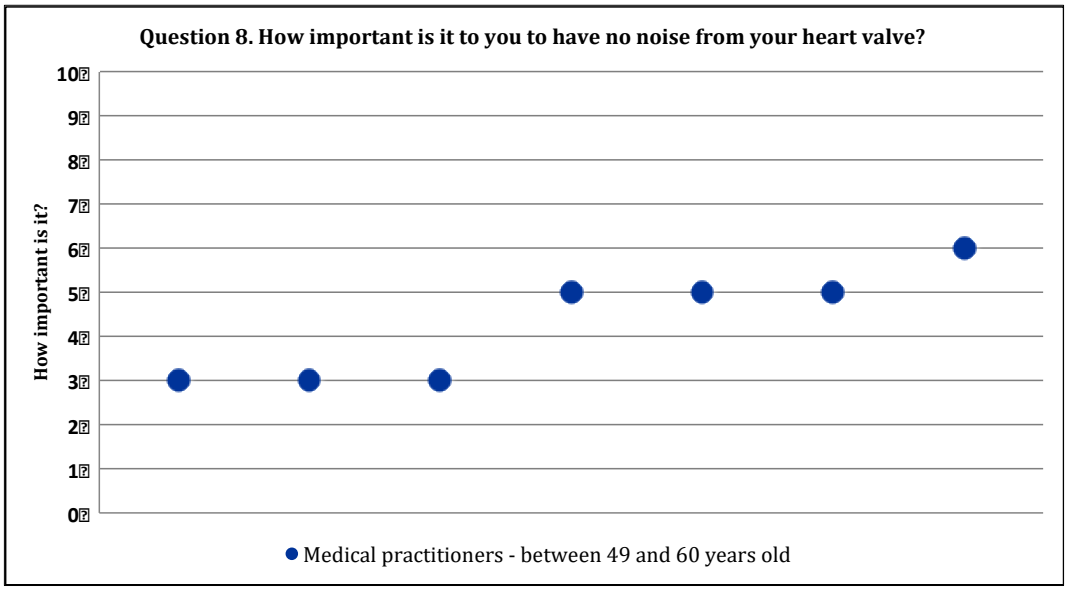
assumes that the responses are ordinal and the alternatives are restrained to a rank. Therefore, the significance can be explained among medians differences (Field, 2013).
The comparison was executed using the four ages' group (less than 37 years old, between 37 and 48 years old, between 49 and 60 years old, and more than 60 years old).
Accordingly to the Kruskal-Wallis Test, scores in question nº7 are not significantly affected by "people with Marfan and/or immediate family" ages, **H:** 0.97, $p > .05$. This result means that this group has similar preferences regarding anticoagulation and pregnancy despite the differences in their ages.

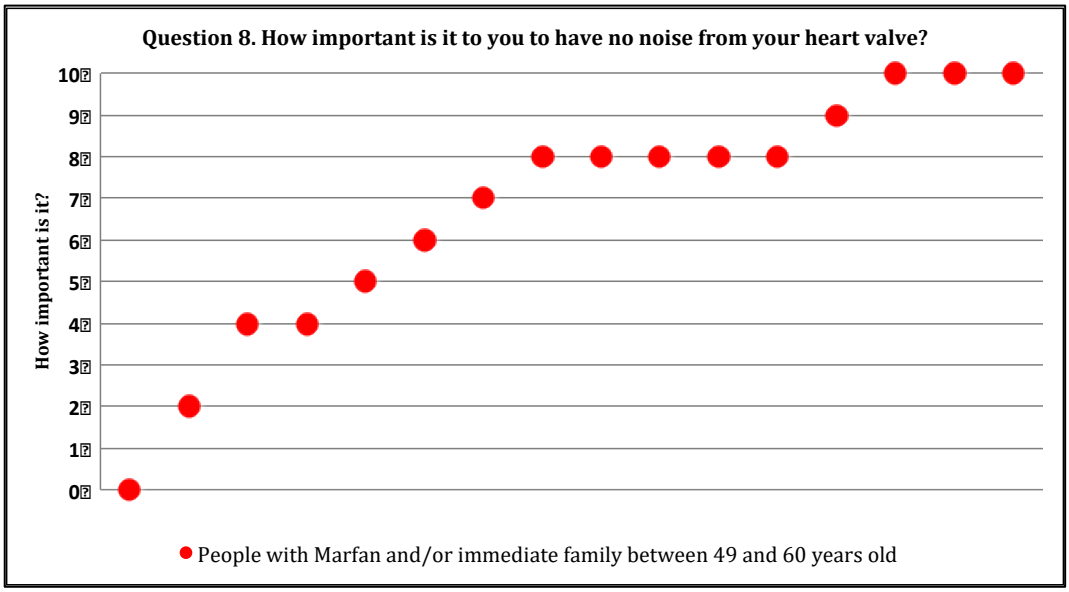
3.1.5 Findings per participant's ages in question nº 8

Once again, Mann-Whitney Test (a non-parametric test) was chosen to compare two independent samples (Field, 2013).

According to the Mann-Whitney Test, "people with Marfan and/or immediate family"´ preferences did not differ significantly from "medical practitioners" in questions 1, 2, 3, 4, 5, 6 and 7, even though they were representing various age groups. However, in **Question 8** (*Mdn*= 5) preferences of "people with Marfan and/or immediate family" between the ages 49 and 60 years old **do differ significantly** from those of "medical practitioners" aged between 49 and 60 years old, *U*= 33, *z*= -2.226, *Sig. (2-tailed)*= 0.027 ($p < .05$) and *r*= -0.45. Again this *"r"* is between Cohen's standard measure of 0.3 and 0.5 for a medium and large effect respectively (Field, 2013).

In order to support the above analysis, two dot plots were generated to highlight the score distribution in both groups in more detail.





3.2 Thematic analysis

Fifty participants, 78% of whom were "people with Marfan and/or immediate family" and 22% - "medical practitioners", completed fully or partly the open-ended questions from the questionnaire. Almost one third of females (29%) and 72% of males participated in the survey (altogether). Two participants were between the ages of 13 – 24 years old; two participants were between the ages of 25 – 36 years old; eleven participants were between the ages of 37 – 48 years old: twenty two participants were between the ages 49 – 60 years old and thirteen participants were 61 years old or older.

The data analysis was dedicated to reviewing themes regarding the coded data per question individually, and contemplating the entire data set. As a result, a themes table was elaborated **(See appendix 5).**

3.2.1 Question 1

How important is it for you to postpone having an operation on your aorta for as long as the doctors think it is safe to do so?

The following three core theme categories were established: 1) I would prefer to avoid the medical risks associated with a surgery, 2) I would prefer to undergo a surgery while I am younger and fitter, and 3) I would prefer to get it over with.

3.2.1.1 Theme I

I would prefer to avoid the medical risks associated with a surgery

Frequency: 19

This theme is defined by the participants´ belief that it is a hazard to postpone the surgery because of the possible medical risks and outcomes. However, medical practitioners offer a mild opinion regarding this point. They support the idea of having a surgery when it is "necessary". See table for examples.

Theme I

I would prefer to avoid the medical risks associated with a surgery			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Male	55 years old	"Any operation carries some risk, so the opportunity to postpone is an obvious option to take"
Medical practitioner	Male	51 years old	"It is a balance - delay surgery but try avoid significantly increased risks"

3.2.1.2 Theme II

I would prefer to undergo a surgery while I am younger and fitter

Frequency: 7

In this case, a common thread can be traced in the various answers provided by the participants (regardless their age). This theme is based on the participants' beliefs that age and fitness are the key elements that help to decide whether or not to go through a surgery. They would also share the opinion that both mentioned elements might improve the recovery process. See table for examples.

Theme II

I would prefer to undergo a surgery while I am younger and fitter			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Male	32 years old	"The younger you are when you have it, the quicker you heal!"
Person with Marfan	Female	60 years old	"If it has to happen anyway, why should you try to postpone it? It is better to have surgery while you are still in good shape"

3.2.1.3 Theme III

I would prefer to get it over with

Frequency: 5

This theme is defined by the interest in having the operation soon, so patients will be able to stop thinking about it. See table for examples.

Theme III

I would prefer to get it over with			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Male	32 years old	"Not so fussed about having the surgery as such. Really just wanted to get it over and done with, as I knew it had to be done at some point anyway"
Person with Marfan	Male	49 years old	"If I am going to have it done at some point lets get on with it"

3.2.2 Question 2

Question 2: Is taking anticoagulant (blood thinning) medication an important problem for you?

The following three core theme categories were established: 1) I would prefer to avoid the risks related to taking anticoagulation, 2) I would prefer to have an active lifestyle, and 3) I would prefer a drugless therapy.

3.2.2.1 Theme IV

I would prefer to avoid the risks related to taking anticoagulation

Frequency: 17

This theme is defined by the participants' agreement that taking anticoagulants might carry some risks. In fact, it can be observed that patients as well as medical practitioners express concern towards medication risks. They are preoccupied with such complications as bleeding. See table for examples.

Theme IV

I would prefer to avoid the risks related to taking anticoagulation

Group	Gender	Age	Response
Person with Marfan	Male	32 years old	"It's just a pain in the backside. More to the point it has other implications, such as if I have a seriousness injury or dissection, I am more likely to bleed out!"
Medical practitioner	Male	57 years old	"It can cause fatal bleeding"

3.2.2.2 Theme V

I would prefer to have an active lifestyle

Frequency: 11

This theme is defined by the shared idea that anticoagulants affect lifestyle in a negative way. See table for examples.

Theme V

I would prefer to have an active lifestyle

Group	Gender	Age	Response
Person with Marfan and immediate family	Female	41 years old	"My father is on Warfarin and it completely affects your daily life"
Medical practitioner	Male	58 years old	"Anticoagulation has complications, needs a disciplined life, and restricts you in your activities and traveling around"

3.2.2.3 Theme VI

I would prefer a drugless therapy

Frequency: 8

This theme is defined by participants ' belief that being permanently on medication is not desirable. See table for examples.

Theme VI

I would prefer a drugless therapy			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Male	69 years old	"A lifetime of anticoagulants is unattractive"
Person with Marfan	Male	59 years old	"I would prefer not to take an anticoagulant over the long term"

3.2.3 Question 3

Question 3: How important is it to you, if you need to have an operation on your aorta, to get on with it and have it behind you?

The following three core theme categories were established: 1) I would prefer to get it over with, 2) I would prefer to avoid the risks associated with a surgery, 3) I would prefer not to feel the anticipatory anxiety and fear before the surgery, and 4) I would prefer to have medical advice.

3.2.3.1 Theme VII

I would prefer to get it over with

Frequency: 9

On the one hand, this theme is defined by participants' belief that having the surgery sooner would mean to have fewer limitations in their lives. Conversely, medical practitioners define this theme as just one more step to improve their quality of life. See table for examples.

Theme VII

I would prefer to get it over with			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Female	24 years old	"Having known I would need surgery from a fairly young age, as the time passed, I just wanted the surgery done so I could get on with my life"
Medical practitioner	Female	55 years old	"It won't be over after aortic root replacement"

3.2.3.2 Theme VIII

I would prefer to avoid the risks associated with the syndrome

Frequency: 11

For some participants, postponing the surgery would mean putting their lives at risk, while for medical practitioners the decision whether to undergo a surgery or not is associated with parameters that are more objective, such as medical exams for example. See table for examples.

Theme VIII

I would prefer to avoid the risks associated with a syndrome			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Female	60 years old	"Why living with the risks of a time-bomb and waiting for it to get worse?"
Medical practitioner	Male	51 years old	"I am reluctant to recommend early intervention if there is not a clear indication (there is a risk of over-treatment)"

3.2.3.3 Theme IX

I would prefer not to feel the anticipatory anxiety and fear before the surgery

Frequency: 6

Participants, especially patients, stated that they would experience moderate to lower level of anxiety before the surgery. Additionally, patients mention feeling apprehensive, stressed, worried, and tired, while waiting for the operation. See table for examples.

Theme IX

I would prefer not to feel the anticipatory anxiety and fear before the surgery			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Male	59 years old	"The anxiety of waiting is not fully apparent until it is removed post op. Those of us yet to be operated on will not understand this until after the event so will not fully appreciate the stress that procrastination is putting on them?"
Person with Marfan	Female	48 years old	"Fear is a dangerous motivator"

3.2.3.4 Theme X

I would prefer to have medical advice

Frequency: 5

Medical advice appears to be an important factor when making decision upon a surgery.

There is a positive attitude regarding doctors' advice. See table for examples.

Theme X

I would prefer to have medical advice			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Female	66 years old	"I always prefer to confront the issue and get it over with! I would, however, always want to be guided by the surgeon"
Person with Marfan	Female	71 years old	"Obviously, I realise how important it is to have the operation, but I would hope to have medical advice as to when and how"

3.2.4 Question 4

Is taking lifelong medication such as beta-blockers or losartan an important problem for you?

The following four core theme categories were established: 1) I would prefer not to have the side effects caused by beta-blockers, 2) I would prefer a drugless therapy, 3) I would prefer to take medications because I want to live, and 4) I would prefer to take medications because I want to live.

3.2.4.1 Theme XI

I would prefer not to have the side effects caused by beta-blockers

Frequency: 15

Both: medical practitioners and patients agree that specific medication might present negative side effects, such as impotence and tiredness. Only two participants stated that they have not had difficulties by the use of beta-blockers. See table for examples.

Theme XI

I would prefer not to have the side effects caused by beta-blockers			
Group	**Gender**	**Age**	**Response**
Medical practitioner	Male	50 years old	"Beta-blockers and impotence"
Person with Marfan	Male	55 years old	"Loss of energy, loss of puff, i.e. heart not beating faster when you want it too, affect on sexual potency and libido"

3.2.4.2 Theme XII

I would prefer a drugless therapy

Frequency: 11

It can be observed that there are discrepancies between the patients and the medical practitioners' beliefs about regular consumption of specific medications. See table for examples.

Theme XII

I would prefer a drugless therapy

Group	Gender	Age	Response
Person with Marfan	Male	39 years old	"I have always been uncomfortable with the thought of regular consumption of pharmaceuticals and have chosen not to do so"
Medical practitioner	Male	58 years old	"I rarely come across patients unwilling to take medication, whether they actually take them is another matter"

3.2.4.3 Theme XIII

I would prefer to take medications because I want to live

Frequency: 7

This theme is defined by the patients' belief that medication might improve their lives and allow them to live actively. See table for examples.

Theme XIII

I would prefer to take medications because I want to live

Group	Gender	Age	Response
Person with Marfan and immediate family	Male	71 years old	"Take the medicine and live a reasonably good life or do without them and be ill"
Person with Marfan	Female	54 years old	"Not a problem if it improves and lengthens my life"

3.2.5 Question 5

Are repeated visits to the hospital to have echo tests an important problem for you?

The following two core theme categories were established: 1) I would prefer not to have this irritating commitment, and 2) I would prefer to have monitoring.

3.2.5.1 Theme XIV

I would prefer not to have this irritating commitment

Frequency: 8

This theme is defined by the patients' belief that frequent hospital visits might be a bothersome activity, which they would prefer to avoid. See table for examples.

Theme XIV

I would prefer not to have this irritating commitment

Group	Gender	Age	Response
Person with Marfan	Male	13 years old	"Any visits to hospital are unpleasant and they take time off other things"
Person with Marfan	Male	69 years old	"I live with that now, but it would rather not"

3.2.5.2 Theme XV

I would prefer to have monitoring

Frequency: 22

This theme is defined by the participants' belief that repeated visits to the hospital are a part of the regular routine for people with Marfan, thus, these visits are not a problem for them. Moreover, patients, immediate family, and medical practitioners agreed that medical appointments are beneficial to clarify questions and to reaffirm the treatment effectiveness. Indeed, regular check-ups at the hospital might bring a sense of relief to the patients. See table for examples.

Theme XV

I would prefer to have monitoring

Group	Gender	Age	Response
Medical practitioner	Male	55 years old	"Repeated visits make the patient confident and offer a place where questions of any type can be answered"
Person with Marfan an	Male	65 years old	"I would be prepared to travel to anywhere to obtain the results that I need and on-going yearly / monthly visits would not be a problem"

3.2.5.3 Theme XVI

I would prefer not to feel the stress of waiting for the medical results

Frequency: 3

Although not all the participants share this opinion, it is worth considering as one of the factors that explains why repeated visits to the hospital might be a problem. See table for examples.

Theme XVI

I would prefer not to feel the stress of waiting for the medical results			
Group	**Gender**	**Age**	**Response**
Person with Marfan	Male	41 years old	"Not a problem in itself, but if you have to be worried about the results, it is certainly not pleasant"
Person with Marfan	Male	39 years old	"Not a problem but perhaps more of a responsibility, with a certain cycle of fear & relief attached to it"

3.2.6 Question 6

How important is it to you to have a physically active lifestyle?

The following two core theme categories were established: 1) I would prefer to keep a normal and active life, and 2) I would prefer to have an active lifestyle but I have limitations.

3.2.6.1 Theme XVII

I would prefer to keep a normal and active life

Frequency: 25

This is the only question where patients, immediate family, and medical practitioners explicitly sustain that having a normal life is important. Moreover, they all agree upon the belief that an active life provides the opportunity to feel “normal”, which seems to be a significant part of patients’ lives.

People with Marfan syndrome just want to live their lives as everybody else. See table for examples.

Theme XVII

I would prefer to keep a normal and active life

Group	Gender	Age	Response
Patient and relative	Female	42 years old	"I want to live a normal life and participate in moderate physical activities, and not feel like a disabled person"
Patient	Male	32 years old	"Life is to be lived, no wrapped in cotton wool"
Patient	Female	49 years old	"This is my life, I would be lost without exercise"

3.2.6.2 Theme XVIII

I would prefer to have an active lifestyle but I have limitations

Frequency: 8

Although patients express their interest in maintaining an active life, they still face several limitations. One restriction lies is the medication and another one is the quality as well as the quantity of physical activity feasible to perform. See table for examples.

Theme XVIII

I would prefer to have an active lifestyle but I have limitations

Group	Gender	Age	Response
Person with Marfan	Male	55 years old	"I have put on weight...perhaps should do more...not always easy when bp tablets and beta-blockers slow you down"
Person with Marfan	Male	25 years old	"I do not run an active sports life, which does not mean that I would not want to. Just me, do not, so I have to be limited to a minimum. I would like to live a normal life like everyone, but health condition does not allow"

3.2.7 Question 7

Would anticoagulation, which might complicate pregnancy and make having a baby more difficult process, be important in your choice of operation? (Men may answer considering the impact on potential fatherhood.)

The following two core theme categories were established: 1) It is not important as I am too old to have children, and 2) I would prefer to avoid the pregnancy risks.

3.2.7.1 Theme XIX

It is not important, as I am too old to have children

Frequency: 4

It can be observed that neither pregnancy nor the avoidance of anticoagulants that might affect it is an important topic for both: women and men after a certain age. See table for examples.

Theme XIX

It is not important, as I am too old to have children

Group	Gender	Age	Response
Person with Marfan	Female	51 years old	"Over 50 very unlikely!!!"
Person with Marfan	Female	58 years old	"Not at all important as I had just about reached the menopause when I had my surgery"

3.2.7.2 Theme XX

I would prefer to avoid the pregnancy risks

Frequency: 13

This theme can be characterised with the belief that, indeed, anticoagulants might complicate pregnancy and having kids. However, if medical practitioners' opinions are compared with the rest of the group, it can be noted that "risk" seems to be a flexible concept. On the one hand, some doctors believe that the risks are possible to manage. Still, other medical practitioners think that it would be better to avoid pregnancy. See table for examples.

Theme XX

I would prefer to avoid the pregnancy risks

Group	Gender	Age	Response
Person with Marfan	Female	54 years old	"I might not have elected to get pregnant if daily shots of heparin required"
Medical practitioner	Male	47 years old	"This is discussed with all female patients at their initial appointments. Most understand that any pregnancy will require close monitoring and changes in medication"
Person with Marfan	Female	42 years old	"If I plan to have a baby, it will be important. But my cardiologist actively discourages people like us to have babies"

3.2.8 Question 8

How important is it to you to have no noise from your heart valve?

The following two core theme categories were established: 1) I would prefer not to have a noise in my heart valve, and 2) Noise is not a problem: I would prefer to have noise as it reassures me that I am alive.

3.2.8.1 Theme XXI

I would prefer not to have a noise in my heart valve

Frequency: 14

Patients would like to avoid it because they do not like it. Moreover, as in some cases, the noise might makes them feel different from the rest in a negative way. See table for examples.

Theme XXI

I would prefer not to have a noise in my heart valve

Group	Gender	Age	Response
Person with Marfan	Female	66 years old	"It can be noticeable but generally not. However, the tilting discs seem to be far noisier and I know from my son and various friends that they often feel embarrassed at having to explain the noise particularly as it often leads to explaining about Marfan as well. No one likes to feel like a freak"
Person with Marfan	Male	69 years old	"I would rather not live with it"

3.2.8.2 Theme XXII

Noise is not a problem: I would prefer to have noise as it reassures me that I am alive

Frequency: 15

Apart from accepting the valve noise as part of their lives, patients acknowledge it as a stimulus that reminds them that they are still alive thanks to the heart valve.

Moreover, medical practitioners zealously affirm that the noise is a problem that affects a minuscule proportion of the population. See table for examples.

Theme XXII

Noise is not a problem: I would prefer to have noise as it reassures me that I am alive

Group	Gender	Age	Response
Medical practitioner	Male	61 years old	"Not an issue in 99% of patients"
Medical practitioner	Male	47 years old	"Never heard this as a problem"
Person with Marfan	Male	62 years old	"Noise from my heart valve does not bother me. It is actually quite reassuring"

4 Discussion

This study aimed to explore relative values on the lifelong implications of timing and choice amongst the three currently available forms of prophylactic operations placed by people with Marfan syndrome and genetically determined life-threatening aortic root aneurysms. On the basis, values were recognized, accordant to the Ottawa Decision Support Framework, as "a person's informed attitudes about the relative desirability/undesirability or individual importance of a health care option's unique characteristics, which include that option's protocol, possible benefits, and potential harms" (Llewellyn-Thomas & Crump, 2013; O'connor, 2012).
Throughout a mixed data analysis, this research identified a range of "people with Marfan and/or immediate family" and medical practitioners' preferences as well as meaningful themes that might influence the decision-making process regarding a prophylactic surgery.

Apart from the quantitative data analysis of the questionnaire, the thematic analysis of the open-ended questions also illustrated that "people with Marfan and/or immediate family"´ preferences might differ or coincide inside their group and/or with medical practitioners. This result goes in accordance with Epstein and Peters (2009) and Stiggelbout et al. (2012) idea that health care treatment and screening choices tend to be preference-sensitive. Furthermore, and thanks to these authors, we know that each individual has his/her own unique situation, needs, and desires. Therefore, discrepancies and similarities between participants support the fluctuations between personal preferences.

Additionally, this survey expands Fosbraey (2014) ´results to cover a broader dimension of patient´s values by expanding the scope of values to those who are in charge of giving advice to people with Marfan syndrome (immediate family or medical practitioners). Through an exhaustive analysis it was highlighted the way such themes as medical risks, time for surgery, medication and side effects, active lifestyle, medical advice, commitments, stress and anxiety, pregnancy and noise of the hart valve are

influential in patient decision making. Thus, the complex association among ”people with Marfan and/or immediate family” and medical practitioners’ preferences, emerged from the data, are more comprehensive, and provide a broader representation of patient’s values.

Firstly, it was mentioned previously that people with Marfan syndrome face difficulties when they want to start a family. Not only patients have to confront pregnancy hazards but also there are high probabilities of having a baby with Marfan syndrome, including all the additional health complications. This point is strongly sustained in this research. Participants, including medical practitioners, evaluated anticoagulation (which might complicate pregnancy and make having babies a more difficult process) as extremely important in their choice of operation (48% scored between 8 and 10). Moreover, according to Mann-Whitney Test, the distribution in question nº7 is the same across the male and female categories in the ”people with Marfan and/or immediate family”’ group. However, and even though there is not a significant statistical difference, it can be observed that males tend to rank their preferences across a wider range, while females tend to have more extremes scores. Although, there is a group of ”people with Marfan and/or immediate family” (30% who scored 0) to whom pregnancy is not important at all as they believe that they are too old to think about it. The results slightly differ from Treasure and Pepper (2015) observations. These authors sustain that anticoagulation is a significant matter for young people, as it still leaves them creating a family option. In this research, it can be observed that preferences regarding anticoagulation and pregnancy are not significantly related to age. However, it can be strongly sustained that it is an important issue for people with Marfan syndrome. In fact, questions nº7 and nº2, both linked to anticoagulation, show 43% of the scores between 8 and 10 and 68% of the scores between 8 and 10, respectively, were the replies provided by the ”people with Marfan and/or immediate family”’ group.

Previous studies sustain that anticoagulation was evaluated as a “small price to pay” for some patients. However, Fosbraey (2014) revealed a robust and persistently tendency to evade medication such as Coumadin/Warfarin. In her research, the main explanation to this decision appears to be the desire for those with Marfan syndrome to feel as

"normal" as everyone else. The current research affirms similar results regarding the tendency to appraise the anticoagulant consumption as an extremely important problem. Notwithstanding, different categories that explain the desire to avoid anticoagulation appeared from the thematic analysis. One of them is the risk associated with anticoagulants, such as bleedings. In addition, the strong desire of leading an active lifestyle and the traumatic idea that medication will not allow it. Lastly, it is patients' motivation to receive a drugless therapy.

Secondly, it is worth considering that not significant differences were found in questions 1, 2, 4, 5, 6, 7, and 8 regarding "people with Marfan and/or immediate family"´ as well as medical practitioners' preferences. These results contradict Brothers et al. (2004) and Fraenkel et al. (2004) presumption that clinicians might be imprecise evaluating patient's values and treatment options. Nevertheless, these results might be influenced by special emphasis patients place on medical advice. According to the thematic analysis, medical advice appears to be an important factor while making decisions upon a surgery. Following this idea, it can be presumed that medical advice has an impact into patient's preferences; as a result, there are similarities among the answers provided by both groups.

Therefore, and as it was mentioned formerly, the *Prospect Theory* establishes that when consequences are contemplated gains, people tend to evade risks (Tversky & Kahneman, 1981). Indeed, "risk" was a theme that emerged across questions number 1, 2, 3, and 7. This topic was raised as "risks" play an important role in people's and medical practitioners´ lives. Firstly, "risks" are the hazards placed on the patients by postponing the surgery; they also are the possible complications coming from the anticoagulation treatment, or the belief that anticoagulants might complicate pregnancy and having kids. However, after comparing medical practitioners' opinions with the once given by the rest of the group, it can be noted that "risk" seems to be a flexible concept. On the one hand, some doctors believe that the risks are possible to manage. Still, other medical practitioners think that it would be better to avoid pregnancy. This sort of flexibility and different points of views in respect to risk might affect what people consider as a gain. In fact, the awareness about benefits is influenced by the way

information is addressed or outlined (Haward & Janvier, 2015). Once again, the role of medical advice emerges as an important theme.

Although not significant differences were found in questions 1, 2, 4, 5, 6, 7, and 8 regarding ”people with Marfan and/or immediate family”´ as well as medical practitioners' preferences. Question nº3 did differ substantially by the answers provided in these two groups, showing a *Sig. (2-tailed)* of 0.006 ($p < .01$) and *r* of -0.36, meaning a medium size effect. This result, is in line with O'connor et al. (2007) who established that clinicians might be limited judges to assay patient's values. Therefore, there might be an underestimation of treatment alternatives, which are extremely valued by informed patients.

Hence, the results occurred in question nº3 might be explained in part analysing the distributions in the graphs. On the one hand, ”people with Marfan and/or immediate family” show a clear tendency to high scores. However, medical practitioners show a moderate tendency between medium scores.

Following the result in question nº 3, it can be observed that there is a significant, yet negative, correlation between preferences in question nº1 and question nº3 for the group of ”people with Marfan and/or immediate family”, $\boldsymbol{r_s}$**=** -.30, $\boldsymbol{p}$ (2-tailed) < .05. However, there is not an important relationship between preferences in question nº1 and question nº3 in the medical practitioners group.

Thirdly, Fosbraey's study (2014) discusses that people with Marfan preferred to have a prophylactic surgery sooner as it is associated with lower levels of anxiety and less medical monitoring. Additionally, (Fosbraey)´s survey also advocates that people with Marfan prefer to cut down the regular visits to the hospital. However, Romaniello et al. (2014) explain that annual echocardiographic monitoring is recommended to detect changes in size of their aorta. The outcomes from this questionnaire support Fosbraey's results. On the one hand, participants sustain that they consider it very important to have an operation on their aorta, to get on with it and have it behind them as they evidence high levels of anticipatory anxiety and fear before the surgery and during the medical appointments. This human reaction is based on current paradigms, which sustain that arduous and critical decisions are made under the influence of stressful

conditions and that those emotional states, related to the mentioned tension, should be considered important if not the key part in the decision making process (Haward & Janvier, 2015; Janvier et al., 2012). As for the hospital visits, this research also broadened Fosbraey's investigation. From the thematic analysis, two main themes emerged. A group of participants believes that hospital visits are an "irritating commitment" and they would prefer to avoid or reduce them. Nonetheless, 39% of the patients' scores were between 0 and 2. This means that almost half of the "people with Marfan and/or immediate family" believe that hospital visits are not at all important to them. In fact, medical appointments are considered necessary as they allow to reassure patients that everything is going well with the treatment or the aorta before or after the surgery. In addition, it is important to mention that "necessary" does not mean without problems. A group of people believes that hospital visits are imperative, however these visits are associated with high levels of stress. Fosbraey arrived to the same conclusion in his findings.

Fourthly, no differences were found while comparing the results collected from both groups, with an exception of question nº3. The same comparison was performed, only this time considering participants and medical practitioners' ages. As a result, preferences of "people with Marfan and/or immediate family" between the ages 49 and 60 years old do differ significantly from those of medical practitioners aged between 49 and 60 years old, *Sig. (2-tailed)*= 0.027 ($p < .05$) and r= -0.45, meaning a medium to large size effect. On the one hand, patients sustain that noise may be a real problem. Moreover, in some cases the noise might make them feel different from the rest in a negative way. However, this group of medical practitioners zealously affirm that the noise is a problem that affects a minuscule proportion of the population. For example, one medical practitioner (61 years old) sustains "not an issue in 99% of the patients".

Finally, based on the results collected from the questionnaire and the thematic analysis, it can be observed that participants, despite their age, would like to avoid anticoagulation. As it was mentioned previously, anticoagulation is a negative consequence of the Bentall method - one of the prophylactic surgeries available (Bentall & De Bono, 1968; Treasure et al., 2011). Indeed, according to the participants, avoiding

anticoagulation means, a safer life, without all the risks and fears associated with this medication. It means freedom from long-term medication treatment as well as a possibility to lead an active lifestyle, which, as we know, could be strongly affected by Marfan syndrome (De Bie et al., 2004).

Regarding the David valve-sparing root replacement (VSRR), the gain here lies in anticoagulation avoidance; however, further surgery might be required at some point (Treasure et al., 2011). In terms of a second surgery, patients and immediate family express mutual agreement; they prefer to postpone surgery if it is possible. Alternatively, they want to have a surgery as soon as possible so that they can feel some relief. In both cases that are correlated, participants sustain they would like to avoid the risks related to the surgery. They believe that they will withstand surgery better while they are still young and in a good shape. Additionally, patients relate time while waiting for a surgery to anxiety and fear. According to the thematic analysis results, as well as to patients and immediate family' preferences, it can be argued that the David valve-sparing root replacement (VSRR) might not be the best alternative in some particular cases. Lastly, the third option is a personalised external aortic root support (PEARS). This option does not involve anticoagulation nor another surgery. However, further long-term results are needed to ensure its reliability. Yet, until now research made supports this method through a positive follow-up less than in 5 years (Treasure et al., 2014). Based on this research, it can be inferred that PEARS not only meets patients and immediate family' preferences in full but also demonstrates advantages over the other two methods.

5 Strengths and Weaknesses of the Study

Firstly, the present study deepens the knowledge in the area of patient's values through a mixed approach implementation. Moreover, it is known that a mixed method analysis provides a more extensive scientific procedure rather than either qualitative or quantitative evidence search individually (Tashakkori &

Teddlie, 2010). The capacity to "get more out of the data" contributes to developing further explanations, thereby increasing the quality of data interpretation (Tashakkori & Teddlie, 2010).

Secondly, the questionnaire was based on a well-know research tool, the Ottawa Decision Support Framework, which specifically explores "decisional needs of patients as values" (Légaré et al., 2006). Therefore, the questionnaire created for this study might contribute to expanding the awareness regarding patient's values as well as enlightening not only a context that might encounter similarities but also discrepancies between medical practitioners and patients. In fact, this scheme has proved to be efficient in comprehending patient's values (Légaré et al., 2006).

Thirdly, it has been argued that often clinical research does not reproduce patient's values properly: the reason lies in focusing on the outcomes described by the scientific community (Bridges & Jones, 2007; Trujols et al., 2013). Certainly, measuring patient's preferences without their participation "might be irrelevant" (Hagell , Reimer & Nyberg, 2009). Consequently, the questionnaire used in this research, apart from offering meaningful data, was developed with the help of experts as well as including people with Marfan syndrome. As a result, the tool is capable of measuring expressions that are important to the patients, immediate family and medical practitioners - all those people who kindly participated in the current investigation.

Last, but not least, this study tend to provide meaningful data to reinforce and expand (Fosbraey)´s study as well as other researchers attracted to work with people with Marfan syndrome.

Apart from the strengths, limitations of the present research should be recognized. Firstly, this study not only presents a small sample of participants but also disproportion in the number of participants in each group, "people with Marfan and/or immediate family" numerically over medical practitioners.

However, Miles and Huberman (1994) sustain that small samples allow to perform case oriented analysis, considering each case "as a whole entity and looking at patterns, relationships and explanations within the case". Future studies will need to recruit a greater and more balanced number of patients versus clinicians in order to perform and deepen the analysis.

Secondly, the themes emerged in this study might not be suitable in other conditions. Patients' values embody social and cultural rules. Therefore, the preferences and beliefs described by this group of participants might differ from participants elsewhere (Lee et al., 2013).

Thirdly, the lack of clear and inclusive guidelines in order to conduct a thematic analysis is a constant critique to qualitative research (Antaki , Billig , Edwards, & Potter, 2003). Unfortunately, it is difficult to know the researcher's exact assumptions while conducting the thematic analysis. Therefore, appraising, measuring, and/or synthesizing the data analysis together with other studies on the same subject might be challenging (Attride-Stirling, 2001). For future research, it is recommended to conduct the thematic analysis between two investigators, in order to include a percentage of agreement regarding the themes proposed. This strategy might work as a measure of inter-rate reliability, aiming to reduce possible biases (Braun & Clarke, 2006). As a student project, only one researcher was in charge of the thematic analysis. The researcher was aware that this technique had its own limitations, therefore it is conceivable that some level of bias might occur in the themes emerged.

Finally, although applying a mixed method might be perceived as a robust option, it might as well be seen as a weak alternative. The research might be questioned as less rigorous in comparison to a multi-method design. In this case, a multi-method design implies that data is not combined as it happens in a mixed method, such as this study. Instead, within a multi-modal study, interrelated projects are planned and aimed to explain a precise sub-question of an overall research question (Tashakkori & Teddlie, 2010).

6 Implications for practice

The most important variables in motivation and decision-making are values (Higgins et al., 2003), which might work as filters to explain clinical information (Karel, 2000; Reyna, 2008).
Taking this statement under consideration, clinicians need to face not only patients' beliefs and feelings while advising them, but also their values towards treatment alternatives. Then, a profound understanding of patients' values is crucial, when making decisions and/or giving recommendations about treatment options (Lee et al., 2013). Indeed, recognizing patient´s values might be remarkably relevant, especially for people with Marfan syndrome. Hence, people who go up against several obstacles, both psychological and physical ones (Fosbraey, 2014).

As a recommendation, values clarifications might be seen as an effective practice to help patients confront difficult decision-making processes (Pieterse , De Vries , Kunneman , Stiggelbout, & Feldman-Stewart, 2013). Apart from the tools that have been created to assist patients, immediate family, and medical practitioners in clarifying values that might influence medical health care decisions, the questionnaire developed for this research is a novel tool designed specifically for people with Marfan syndrome in order to have a better insight on what matters the most to them.

Further studies should be conducted to shed light on other important aspects. For instance, the questionnaire applicability in daily clinical practice as well as the way to implement patient's preferences into the decision-making process in a more systematic way: considering the available treatment options and each case specificity.

7 Conclusion

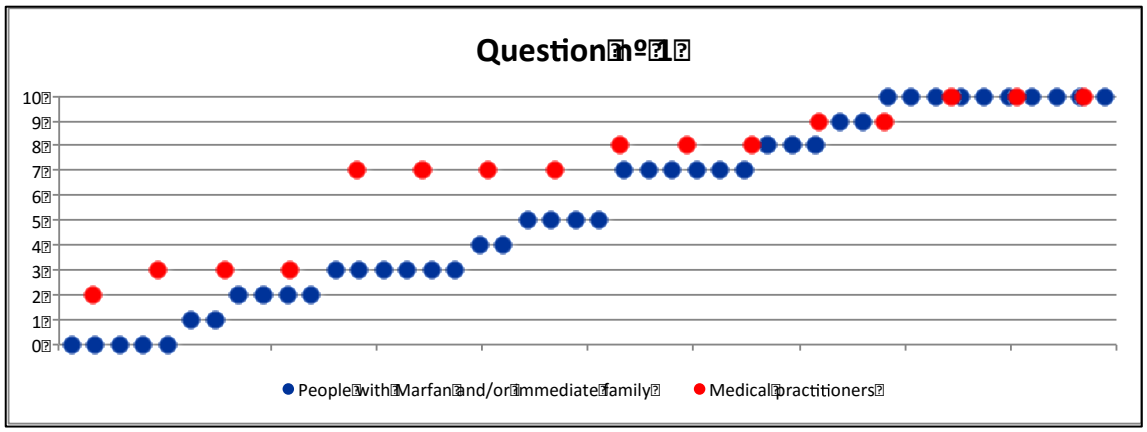
When supporting patients in making decisions, medical practitioners need to address more than just beliefs and feelings regarding the treatment options. A deeper understanding of patients' preferences as well as their background are essential, especially when making decisions concerning treatments. This knowledge will help medical practitioners to communicate and focus on the elements that are important to patients, such as values. According to Kennedy et al. (2008), this sort of "treatment matching" has proved not only to increase patients' satisfaction with the treatment choice but also to enhance medical results.

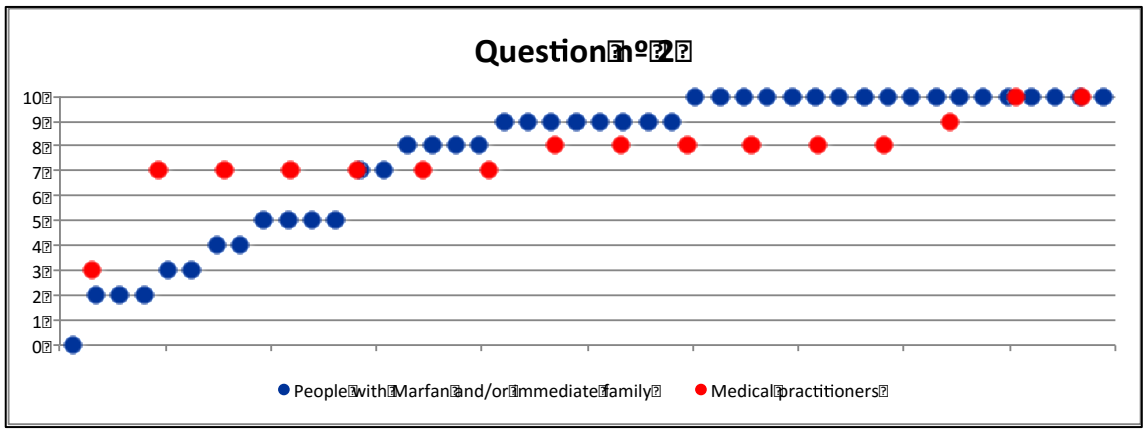
As a summary, if patient's personal values are taken under consideration regarding treatment and screening decisions, it might better meet the patient's individual situation, needs, and desires.

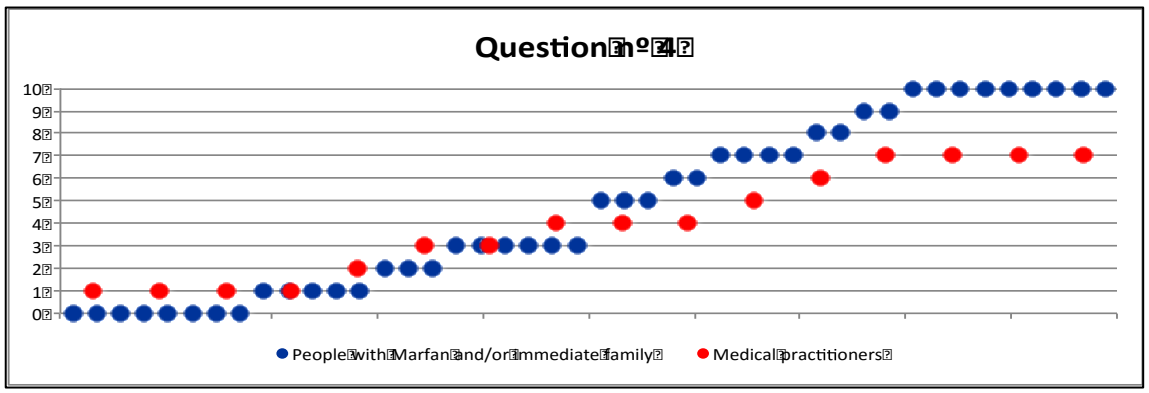
8 Appendix

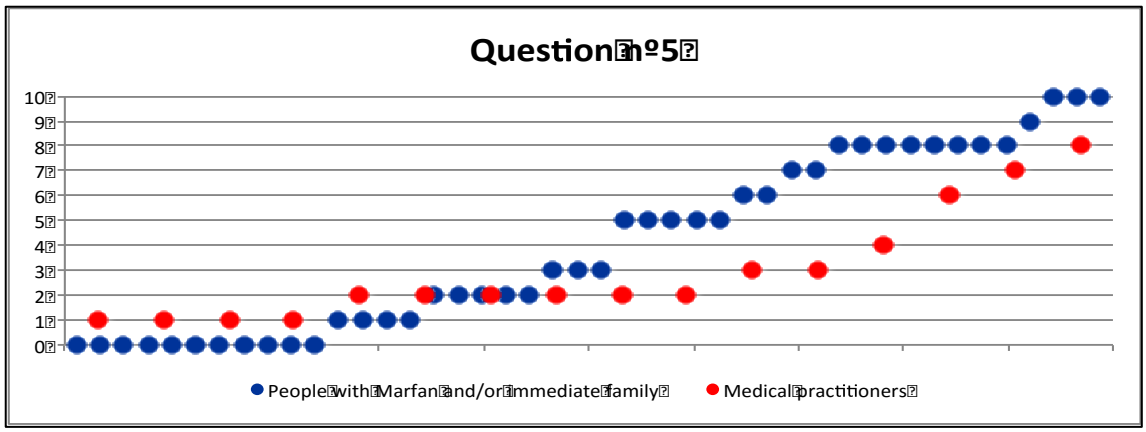
8.1 Appendix 1

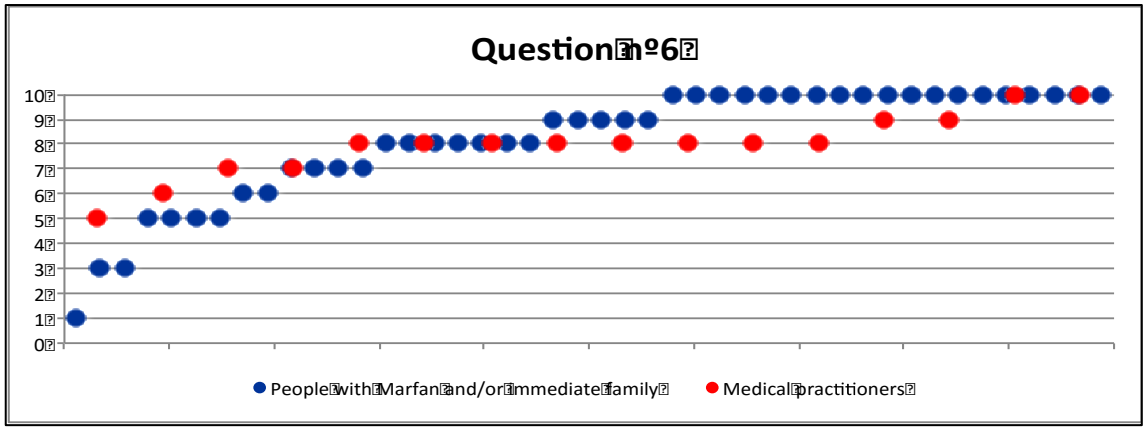
Results given by the groups of "people with Marfan and/or immediate family" and "medical practitioners" upon the questions

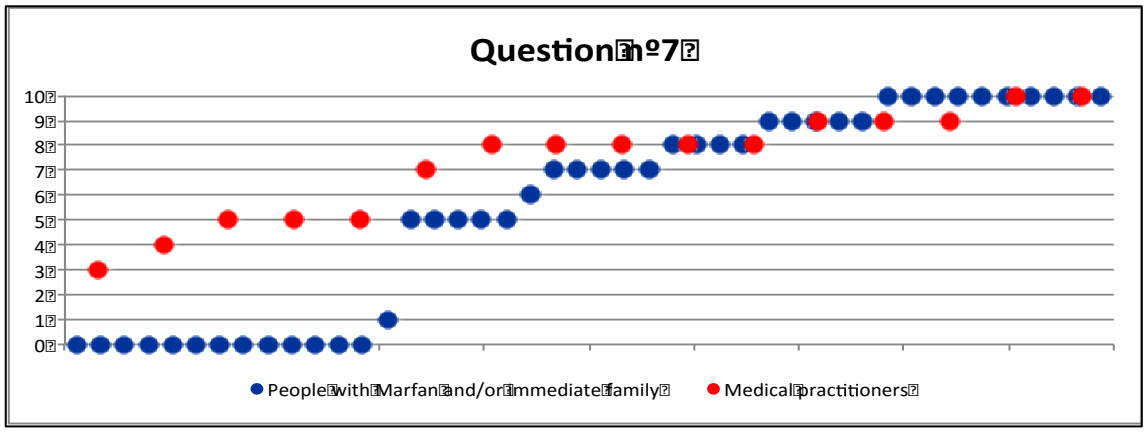


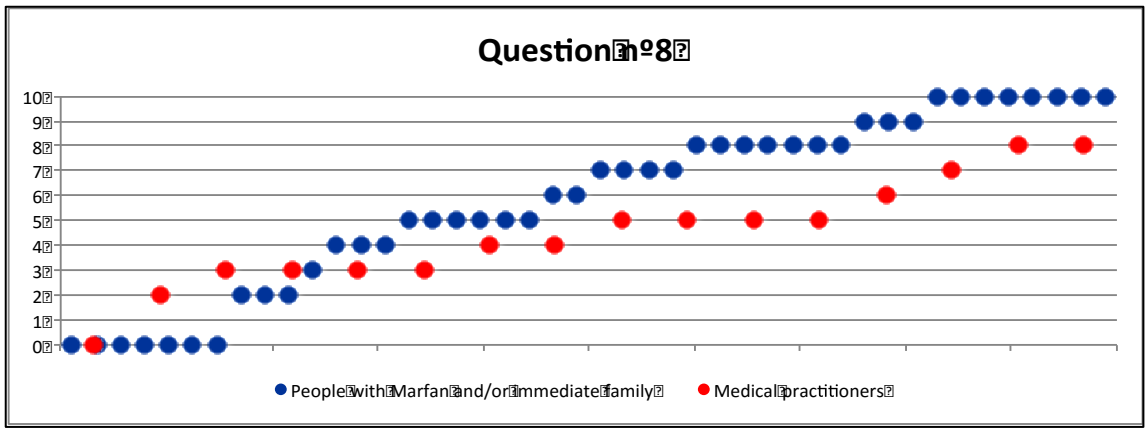





Question nº5
10
9
8
7
6
5
4
3
2
1
0
People with Marfan and/or immediate family
Medical practitioners


Question nº6
10
9
8
7
6
5
4
3
2
1
0
People with Marfan and/or immediate family
Medical practitioners


Question nº7
10
9
8
7
6
5
4
3
2
1
0
People with Marfan and/or immediate family
Medical practitioners



8.2 Appendix 2

Mann-Whitney Test results per question and across gender

Mann-Whitney U per Gender								
	Question 1	Question 2	Question 3	Question 4	Question 5	Question 6	Question 7	Question 8
Mann-Whitney U	334.5	296.5	269.5	312.5	291.5	323	338	305
Z	-0.296	-0.951	-1.399	-0.665	-1.021	-0.5	-0.237	-0.792
Asymp. Sig. (2-tailed)	0.768	0.342	0.162	0.506	0.307	0.617	0.812	0.429

8.3 Appendix 3

Frequencies per Gender in question nº7

Question nº7 - People with Marfan and/or immediate family				
	Males		Females	
	Frequency	Percent	Frequency	Percent
0 = "Not at all important to me"	8	27%	5	36%
1	1	3%	0	0%
2	0	0%	0	0%
3	0	0%	0	0%
4	0	0%	0	0%
5	2	7%	3	21%
6	1	3%	0	0%
7	5	17%	0	0%
8	4	13%	0	0%
9	2	7%	3	21%
10 = "Extremely important to m	7	23%	3	21%
Total	30	100%	14	100%

8.4 Appendix 4

Positive correlations found in the groups of "people with Marfan and/or immediate family" and "medical practitioners" upon the questions.

People with Marfan and/or immediate family						
	Q2 & Q4	**Q2 & Q6**	**Q2 & Q8**	**Q3 & Q6**	**Q4 & Q5**	**Q4 & Q8**
Spearman's rho	0.44	0.31	0.49	0.45	0.36	0.51
p	< 0.01	< 0.05	< 0.01	< 0.01	< 0.05	< 0.01

Medical practitioners			
	Q2 & Q7	**Q4 & Q6**	**Q6 & Q7**
Spearman's rho	0.67	0.59	0.56
p	< 0.01	< 0.05	< 0.05

8.5 Appendix 5

Themes table

Question	Theme	Category	Nº of replies per question	Frequency of occurrence	Percentage total
1	**I**	I would prefer to avoid the medical risks associated with a surgery	**46**	19	**41%**
	II	I would prefer to undergo a surgery while I am younger and fitter		7	**15%**
	III	I would prefer to get it over with		5	**11%**
2	**IV**	I would prefer to avoid the risks related to taking anticoagulation	**43**	17	**40%**
	V	I would prefer to have an active lifestyle		11	**26%**
	VI	I would prefer a drugless therapy		8	**19%**
3	**VII**	I would prefer to get it over with	**44**	9	**20%**
	VIII	I would prefer to avoid the medical risks associated with a surgery		11	**25%**
	IX	I would prefer not to feel the anticipatory anxiety and fear before the surgery		6	**14%**
	X	I would prefer to have medical advice		5	**11%**
4	**XI**	I would prefer not to have the side effects caused by beta-blockers	**44**	15	**34%**
	XII	I would prefer a drugless therapy		11	**25%**
	XIII	I would prefer to take medications because I want to live		7	**16%**
5	**XIV**	I would prefer not to have this irritating commitment	**42**	8	**19%**
	XV	I would prefer to have monitoring		22	**52%**
	XVI	I would prefer not to feel the stress of waiting for the medical results		3	**7%**
6	**XVII**	I would prefer to keep a normal and active life	**41**	25	**61%**
	XVIII	I would prefer to have an active lifestyle but I have limitations		8	**20%**
7	**XIX**	It is not important as I am too old to have children	**35**	4	**11%**
	XX	I would prefer to avoid the pregnancy risks		13	**37%**
8	**XXI**	I would prefer not to have a noise in my heart valve	**35**	14	**40%**
	XXII	Noise is not a problem: I would prefer to have noise as it reassures me that I am alive		15	**43%**

9 References

Antaki, C., Billig, M., Edwards, D., & Potter, J. (2003). Discourse analysis means doing analysis: A critique of six analytic shortcomings. *Discourse Analysis Online*, 1-12.

Attride-Stirling, J. (2001). Thematic networks: an analytic tool for qualitative research. *Qualitative research, 1*(3), 385-405.

Bentall, H., & De Bono, A. (1968). A technique for complete replacement of the ascending aorta. *Thorax, 23*(4), 338-339.

Bleichrodt, H., & Johannesson, M. (1997). An experimental test of a theoretical foundation for rating-scale valuations. *Medical Decision Making, 17*(2), 208-216.

Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology, 3*(2), 77-101.

Bridges, J., & Jones, C. (2007). Patient-based health technology assessment: a vision of the future. *International journal of technology assessment in health care, 23*(01), 30-35.

Brothers, T., Cox, M., Robison, J., Elliott, B., & Nietert, P. (2004). Prospective decision analysis modeling indicates that clinical decisions in vascular surgery often fail to maximize patient expected utility. *Journal of Surgical Research, 120*(2), 278-287.

Castellano, J., Silvay, G., & Castillo, J. (2013). Marfan Syndrome: Clinical, Surgical, and Anesthetic Considerations. *Seminars in Cardiothoracic and Vascular Anesthesia, 18*(3), 260 - 271. doi: 10.1177/1089253213513842

Chiu, H., Wu, M., Chen, H., Kao, F., & Huang, S. (2014). Epidemiological Profile of Marfan Syndrome in a General Population: A National Database Study. *Mayo Clinic Proceedings, 89*(1), 34-42. doi: 10.1016/j.mayocp.2013.08.022

Couët, N., Desroches, S., Robitaille, H., Vaillancourt, H., Leblanc, A., Turcotte, S., Elwyn, G., & Légaré, F. (2013). Assessments of the extent to which health-care providers involve patients in decision making: a systematic review of studies using the option instrument. *Health Expectations, 18*(4), 542 - 561.

De Bie, S., De Paepe, A., Delvaux, I., Davies, S., & Hennekam, R. (2004). Marfan syndrome in Europe. *Public Health Genomics, 7*(4), 216-225.

De Vries, M., Fagerlin, A., Witteman, H., & Scherer, L. (2013). Combining deliberation and intuition in patient decision support. *Patient education and counseling, 91*(2), 154-160.

Dean, J. (2007). Marfan syndrome: clinical diagnosis and management. *European Journal of Human Genetics, 15*(7), 724-733.

Edwards, A., & Elwyn, G. (2009). *Shared decision-making in health care: Achieving evidence-based patient choice*. Oxford: Oxford University Press.

Elwyn, G., Frosch, D., Thomson, R., Joseph-Williams, N., Lloyd, A., Kinnersley, P., Cording, E., Tomson, D., Dodd, C., & Rollnick, S. (2012). Shared decision making: a model for clinical practice. *Journal of General Internal Medicine, 27*(10), 1361-1367.

Elwyn, G., O'Connor, A., Stacey, D., Volk, R., Edwards, A., Coulter, A., Thomson, R., Barratt, A., Barry, M., & Bernstein, S. (2006). Developing a quality criteria framework for patient decision aids: online international Delphi consensus process. *British Medical Journal, 333*(7565), 417.

Epstein, R., & Peters, E. (2009). Beyond information: exploring patients' preferences. *Journal of the American Medical Association, 302*(2), 195-197.

Feather, N. (1992). Values, valences, expectations, and actions. *Journal of Social Issues, 48*(2), 109-124.

Feather, N. (1995). Values, valences, and choice: The influences of values on the perceived attractiveness and choice of alternatives. *Journal of personality and social psychology, 68*(6), 1135.

Field, A. (2013). *Discovering statistics using IBM SPSS statistics*. Lodon: Sage.

Fosbraey, J. (2014). *The psychosocial impact of personalised external aortic root support surgery in Marfan syndrome.* King's College London, London.

Fraenkel, L., Bogardus, S., Concato, J., & Wittink, D. (2004). Treatment options in knee osteoarthritis: the patient's perspective. *Archives of internal medicine, 164*(12), 1299-1304.

Gutenstein, M. (2014). Psychological factors in emergency medicine. *Emergency Medicine Australasia, 26*(3), 295-299. doi: 10.1111/1742-6723.12210

Hagell, P., Reimer, J., & Nyberg, P. (2009). Whose Quality of Life? Ethical Implications in Patient-Reported Health Outcome Measurement. *Value in Health, 12*(4), 613-617.

Halpern, J., & Arnold, R. (2008). Affective Forecasting: An Unrecognized Challenge in Making Serious Health Decisions. *Journal of General Internal Medicine, 23*(10), 1708-1712. doi: 10.1007/s11606-008-0719-5

Haward, M., & Janvier, A. (2015). An introduction to behavioural decision-making theories for paediatricians. *Acta Paediatrica, 104*(4), 340-345. doi: 10.1111/apa.12948

Higgins, T., Idson, L., Freitas, A., Spiegel, S., & Molden, D. (2003). Transfer of value from fit. *Journal of personality and social psychology, 84*(6), 1140.

Janvier, A., Lorenz, J., & Lantos, J. (2012). Antenatal counselling for parents facing an extremely preterm birth: limitations of the medical evidence. *Acta Paediatrica, 101*(8), 800-804.

Judge, D., & Dietz, H. (2005). Marfan's syndrome. *The Lancet, 366*(9501), 1965-1976.

Kahneman, D., & Tversky, A. (1979). Prospect theory: An analysis of decision under risk. *Econometrica: Journal of the Econometric Society, 47*(2), 263-292.

Karel, M. (2000). The assessment of values in medical decision making. *Journal of Aging Studies, 14*(4), 403-422.

Kennedy, E., To, T., Steinhart, H., Detsky, A., Llewellyn-Thomas, H., & McLeod, R. (2008). Do patients consider postoperative maintenance therapy for Crohn's disease worthwhile? *Inflammatory bowel diseases, 14*(2), 224-235.

Kerstholt, J., Van der Zwaard, F., Bart, H., & Cremers, A. (2009). Construction of health preferences: a comparison of direct value assessment and personal narratives. *Medical Decision Making, 29*(4), 513-520.

Lee, Y., Low, W., & Ng, C. (2013). Exploring Patient Values in Medical Decision Making: A Qualitative Study. *PLoS ONE, 8*(11).

Légaré, F., O'Connor, A., Graham, I., Wells, G., & Tremblay, S. (2006). Impact of the Ottawa Decision Support Framework on the Agreement and the Difference between Patients' and Physicians' Decisional Conflict. *Medical Decision Making, 26*(4), 373-390. doi: 10.1177/0272989x06290492

Lewin, K., Heider, F., & Heider, G. (1936). *Principles of topological psychology*. New York: McGraw-Hill.

Litwin, M., Gore, J., Kwan, L., Brandeis, J., Lee, S., Withers, R., & Reiter, R. (2007). Quality of life after surgery, external beam irradiation, or brachytherapy for early-stage prostate cancer. *American Cancer Society, 109*(11), 2239-2247.

Llewellyn-Thomas, H., & Crump, T. (2013). Decision support for patients: values clarification and preference elicitation. *Medical Care Research and Review, 70*(1), 50S - 79S.

Loeys, B. (2014). Angiotensin receptor blockers: a panacea for Marfan syndrome and related disorders? *Drug Discovery Today, 20*(2), 262–266. doi: http://dx.doi.org/10.1016/j.drudis.2014.09.022

Makoul, G., & Clayman, M. (2006). An integrative model of shared decision making in medical encounters. *Patient education and counseling, 60*(3), 301-312.

Miles, M., & Huberman, M. (1994). *Qualitative data analysis: An expanded sourcebook* (2 ed.). Thousand Oaks: Sage.

Milewicz, D., Dietz, H., & Miller, C. (2005). Treatment of Aortic Disease in Patients With Marfan Syndrome. *American Heart Association -Circulation, 111*(11), e150-e157. doi: 10.1161/01.cir.0000155243.70456.f4

Mulder, B., & Meijboom, L. (2012). Pregnancy and Marfan syndrome: an ongoing discussion. *Journal of the American College of Cardiology, 60*(3), 230-231.

Murdoch, L., Walker, B., Halpern, B., Kuzma, J., & McKusick, V. (1972). Life expectancy and causes of death in the Marfan syndrome. *New England Journal of Medicine, 286*(15), 804-808.

O'Connor, A. (2012). Ottawa Decision Support Framework to address decisional conflict. URL: www. ohri. ca/decision aid

O'Connor, A., Wennberg, J., Legare, F., Llewellyn-Thomas, H., Moulton, B., Sepucha, K., Sodano, A., & King, J. (2007). Toward the 'tipping point': decision aids and informed patient choice. *Health Affairs, 26*(3), 716-725.

Pieterse, A., De Vries, M., Kunneman, M., Stiggelbout, A., & Feldman-Stewart, D. (2013). Theory-informed design of values clarification methods: A cognitive psychological perspective on patient treatment decision making. *Social Science and Medicine, 77*, 156 - 163.

PLoS-editors. (2007). Qualitative Research: Understanding patients' needs and experiences. *PLoS Medicine, 4*(8), e258. doi: DOI: 10.1371/journal.pmed.0040258

PLoS-editors. (2009). Making the “right” health care decisions: why values matter. *PLoS Medicine, 6*(8).

Reyna, V. (2008). A theory of medical decision making and health: fuzzy trace theory. *Medical Decision Making, 28*(6), 850 - 865.

Ritchie, J., Lewis, J., McNaughton-Nicholls, C., & Ormston, R. (2014). *Qualitative Research Practice: A guide for social science students & researchers* (2 ed.). London: SAGE Publications.

Rohan, M. (2000). A rose by any name? The values construct. *Personality and social psychology review, 4*(3), 255-277.

Romaniello, F., Mazzaglia, D., Pellegrino, A., Grego, S., Fiorito, R., Ferlosio, A., Chiariello, L., & Orlandi, A. (2014). Aortopathy in Marfan syndrome: an update. *Cardiovascular Pathology, 23*(5), 261-266. doi: http://dx.doi.org/10.1016/j.carpath.2014.04.007

Rose, D., Evans, J., Sweeney, A., & Wykes, T. (2011). A model for developing outcome measures from the perspectives of mental health service users. *International Review of Psychiatry, 23*(1), 41-46.

Simon, D., Krawczyk, D., Bleicher, A., & Holyoak, K. (2008). The transience of constructed preferences. *Journal of Behavioral Decision Making, 21*(1), 1-14.

Stacey, D., Murray, M., Legare, F., Sandy, D., Menard, P., & O'Connor, A. (2008). Decision coaching to support shared decision making: a framework, evidence, and implications for nursing practice, education, and policy. *Worldviews on Evidence-Based Nursing, 5*(1), 25-35.

Stevenson, A. (2010). *Oxford dictionary of English*. London: Oxford University Press.

Stiggelbout, A., Van der Weijden, T., De Wit, M., Frosch, D., Légaré, F., Montori, V., Trevena, L., & Elwyn, G. (2012). Shared decision making: really putting patients at the centre of healthcare. *British Medical Journal, 344*(e256), 1-6.

Straus, S., Richardson, S., Glasziou, P., & Haynes, B. (2011). *Evidence-based medicine: how to practice and teach it* (4 ed.). London: Churchill Livingstone Elsevier.

Tashakkori, A., & Teddlie, C. (2010). *Sage handbook of mixed methods in social & behavioral research*. London: Sage.

Treasure, T., Golesworthy, T., Pepper, J., Ruiz, F., & Gallivan, S. (2011). Prophylactic surgery of the aortic root in Marfan Syndrome: reconsideration of the decision making process in the era of customised external aortic root support. *The italian journal of vascular and endovascular surgery, 18*(3), 215-223.

Treasure, T., & Pepper, J. (2015). Personalised External Aortic Root Support (PEARS) compared with alternatives for people with life-threatening genetically determined aneurysms of the aortic root. *Diseases, 3*(1), 2-14.

Treasure, T., Takkenberg, J. J. M., & Pepper, J. (2014). Surgical management of aortic root disease in Marfan syndrome and other congenital disorders associated with aortic root aneurysms. *Heart, 100*(20), 1571-1576. doi: 10.1136/heartjnl-2013-305132

Trujols, J., Portella, M., Iraurgi, I., Campins, J., Siñol, N., & Pérez de los Cobos, J. (2013). Patient-reported outcome measures: Are they patient-generated, patient-centred or patient-valued? *Journal of Mental Health, 22*(6), 555-562.

Tversky, A., & Kahneman, D. (1981). The framing of decisions and the psychology of choice. *Science, 211*(4481), 453-458.

Vahanian, A., Alfieri, O., Andreotti, F., Antunes, M., Barón-Esquivias, G., Baumgartner, H., Borger, M., Carrel, T., De Bonis, M., & Evangelista, A. (2012). Guidelines on the management of valvular heart disease. *European heart journal, 33*(19), 2451-2496.

Van de Velde, S., Fillman, R., & Yandow, S. (2006). Protrusio acetabuli in Marfan syndrome. *The Journal of Bone & Joint Surgery, 88*(3), 639-646.

Weeks, J., Cook, F., O'Day, S., Peterson, L., Wenger, N., Reding, D., Harrell, F., Kussin, P., Dawson, N., & Connors Jr, A. (1998). Relationship between cancer patients' predictions of prognosis and their treatment preferences. *Journal of the American Medical Association, 279*(21), 1709-1714.

Wennberg, J. (2010). *Tracking medicine: a researcher's quest to understand health care*. London: Oxford University Press.

Zikmund-Fisher, B., Sarr, B., Fagerlin, A., & Ubel, P. (2006). A matter of perspective: choosing for others differs from choosing for yourself in making treatment decisions. *Journal of General Internal Medicine, 21*(6), 618-622.