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**‘WHO’S LEFT HOLDING THE BABY?’  
EXPLORING COUPLES’ DECISION-MAKING TO  
HAVE A BIOLOGICAL BABY FOLLOWING A  
WOMAN’S DIAGNOSIS OF MARFAN  
SYNDROME**

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*Submitted in partial fulfillment of the requirements for the  
Doctorate in Counselling Psychology and Psychotherapy  
by Professional Studies (DCPsych).*

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University*

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**Thank you**

## **ABSTRACT**

Research into the psychological effects of reproductive decision-making when an individual has a diagnosis of Marfan syndrome, a genetically inherited and potentially life-threatening condition, is rare and mainly quantitative in nature. In response, this study has investigated the experience of couples' decision-making to have a baby when a woman has been diagnosed with Marfan syndrome. The study was conducted using unstructured interview data, analysed using the qualitative methodology of Interpretative Phenomenological Analysis (IPA). The participants were six couples with a female diagnosis of Marfan syndrome. Five couples with children provided retrospective data and one couple making the decision to have a baby provided live data. Four superordinate themes emerged: her decision is already made; creating an informed decision; finding psychosocial support; existing with the fallout of the decision. The experience of decision-making was complex and multidimensional for the males, females and couples. The women demonstrated a strong drive to have a baby despite the 50/50 odds of having a child with Marfan syndrome, whereas the men described the tension between wanting a child and coping with the potentially life-threatening risks for a woman in pregnancy and childbirth. A compelling account of the couples' difficulties in exploring their options and assessing the risks with medical professionals emerged. It is argued that this research provides important insights for counselling psychologists and other professionals when working with individuals and couples affected by Marfan syndrome and other genetically inherited conditions at the critical time of coping with reproductive decision-making.

# 1. INTRODUCTION

## 1.1 What is Marfan syndrome?

Marfan syndrome is a complex syndrome named after a French paediatrician, Antoine Marfan. In 1896 he described a young girl of five years old called Gabrielle as having disproportionately long, thin limbs, fingers and toes. Her spine curved abnormally and she had poor muscle development. By the beginning of the Twentieth Century, other patients were described with similar characteristics and some reported eye problems as well. It was referred to as a syndrome as there were a group of symptoms that frequently occur together in a pattern that doctors were able to recognise.

Marfan syndrome is an inheritable condition caused by a single abnormal, mutant gene on Chromosome 15. It is a disorder of the connective tissue, which can be described as 'the glue and the scaffolding of the body' (Pyeritz & Gasner, 1999, p. 2). All the organs of the body contain connective tissue that holds the body together and is responsible for providing a structure for how bodies develop and grow. In Marfan syndrome the defective connective tissue does not act as it should. This is because the production of one of the elastic proteins in the connective tissue called fibrillin, located on Chromosome 15, is reduced and therefore many parts of the body do not have the strength or stretch required during growth and development. As connective tissue is found throughout the body, the effects of Marfan syndrome can be found in the bones and ligaments, the eyes, the heart and blood vessels, the lungs, skin, tendons, joints and the nervous system.

When an individual inherits Marfan syndrome, regardless of whether they are male or female, there is a 50/50 chance that their children will inherit the condition. However, in 25% of new cases, spontaneous new mutations can occur when the abnormal gene appears in an egg or a sperm (Marfan Association UK, 2006). This means that 75% of individuals who are affected with Marfan syndrome have a family history of the condition.

Marfan syndrome is classed by the medical profession as a common rare disease due to its prevalence in the UK. It is estimated that Marfan syndrome affects 1 in 5,000 individuals worldwide (Marfan Association, 2006). Both males and females of any race or ethnic group can be affected. It affects males and females in equal numbers. The symptoms of Marfan syndrome vary in severity and many people may not display all the signs. This can make Marfan syndrome quite difficult to diagnose. Blood tests to identify the abnormal gene are becoming more widely available, but generally a specialist physician makes a diagnosis after a judicious physical examination, together with a family history.

The most serious potentially life-threatening problems for patients with Marfan syndrome involve the cardiovascular system. These problems may lead to heart failure, irregular heart rhythm, aneurysm or complications with the main artery in the body, the aorta, becoming enlarged. These problems can generally be controlled by medication, or in some cases where the aorta becomes too enlarged surgical repair may be necessary. However,

compared with healthy females, there is a higher risk of cardiovascular complications for women with Marfan syndrome in pregnancy and childbirth.

There is no cure for Marfan syndrome, but the literature emphasises that with careful medical and life-style management the potentially life-threatening risks can be reduced. New drugs and surgical procedures are constantly improving and the prognosis is promising.

## **1.2 Background to the research topic**

I had never heard of Marfan syndrome until it was diagnosed in my family in 2004. There were Marfan-related heart complications and two members of the family underwent a pioneering operation to fit a recently developed stent around the aorta to protect it from rupture. We were advised that everyone in the family had to seek guidance and genetic testing. I felt that we coped using a mix of practicality, connectedness and love.

So what does it mean now to be a member of a Marfan family? Naturally each family member has their own thoughts and feelings, but for me, although it has been quite challenging in many ways, it is also life-enhancing and has given me a heightened awareness of the strength and resilience we have as a family. In the past few years I have followed my family through the trauma of the diagnosis and heart operations, sharing concerns and watching them thrive. I believe it has brought us even closer together. One member of my family was unaware that she had Marfan syndrome when she became pregnant and gave birth to her children. Therefore coping with a genetically

inherited condition was not a consideration when she and her husband were deciding to have their children.

My experience of Marfan syndrome progressed from the personal to the professional. I became increasingly interested in how other families diagnosed with Marfan syndrome cope with this potentially life-threatening condition. In particular, I wondered about the decision to have a baby when a woman has been diagnosed with Marfan syndrome. How do couples make a decision to have a baby when a woman has been diagnosed with Marfan syndrome?

Making a decision to have a baby can play a crucial role in the meaning of human experience. When that decision is complicated by the challenge of coping with a potentially life-threatening condition, it has the capacity to impact on relationships, emotions, thoughts, behaviours and overall quality of life. A further significant and important influence on the decision is when a woman is at risk during pregnancy and childbirth as in the case of females with Marfan syndrome. With this in mind, I was surprised to learn that few studies have specifically examined decision-making to have a baby in known carriers of a genetic condition (Kay & Kingston, 2002).

This research explores how couples make a decision to have a baby when a woman has been diagnosed with Marfan syndrome, a genetically inherited condition. When Marfan syndrome was diagnosed in my family it was a traumatic time. I was naturally concerned, but it also eventually piqued my

professional interest sufficiently to search the psychological databases for significant literature. I had read extensively about the condition from a medical perspective, but quite simply I was inspired and wanted to know more from a psychological perspective.

Apart from wanting to be more informed about the condition for personal reasons, I also became progressively curious to know more from a multi-dimensional professional viewpoint. On making preliminary enquiries in libraries and databases there had appeared to be a poverty of psychological literature on Marfan syndrome. Further investigation revealed that not only was there a poverty of psychological literature, but also limited or practically no knowledge of Marfan syndrome among many of the healthcare professionals with whom I discussed the condition. These ranged from GPs and other doctors, nurses, physiotherapists, osteopaths, chiropractors, opticians and dentists. In my opinion, all these professionals should have had some working knowledge of Marfan syndrome due to the various physical effects of the condition.

The next stage of my investigations involved locating and contacting a wide range of professionals involved specifically in the care of patients with Marfan syndrome and other genetically inherited conditions. Their knowledge and help has been invaluable in writing about not only the medical aspects of Marfan syndrome, but also some of the psychological factors.

### **1.3 Research aims and objectives**

Considering that Marfan syndrome is one of the most common heritable connective tissue disorders (Rantamaki, Raghunath, Karttunen, Lonnqvist, Child and Peltonen, 1995) and that there are 10,000 people in the UK with Marfan syndrome, studies concentrating on the psychological effects of Marfan syndrome are rare. Furthermore, studies concentrating explicitly on exploring the personal meaning for couples deciding to have a baby appear to be non-existent. The central concern in the present research is to address how and why a couple's thoughts, feelings, beliefs and behaviours are constructed and maintained in the experience of making a decision to have a baby, following a woman's diagnosis of Marfan syndrome.

This research aims to explore the ways in which counselling psychology and psychotherapy could be effective in meeting the needs of a couple's decision-making when the mother has been diagnosed with Marfan syndrome. Specific objectives are to: understand the meaning of a couple's psychological and social world, in particular the decision to have a baby when a woman has been diagnosed with Marfan syndrome; generally assess the implications for counselling psychology and psychotherapy in working with patients coping with this genetically inherited, potentially life-threatening, condition; generate new knowledge of the psychological needs of couples in the management of Marfan syndrome throughout their decision to start a family together and beyond; know how counselling psychology may address these needs with a view to improving the present care and support system.

#### **1.4 Research question**

Marfan syndrome affects both males and females in equal numbers. However, the present research will focus on the psychological implications for couples in deciding to have a child when a woman has been diagnosed with Marfan syndrome. The main reason for focusing on couples with a female Marfan diagnosis is that, aside from the normal physical differences between males and females, there is a fundamental gender difference for women in pregnancy. Special precautions have to be taken as the complications and risks associated with pregnancy and breastfeeding for women with Marfan syndrome are increased. This raises several important questions:

- What it is like for couples making a decision to have a baby when the mother has been diagnosed with Marfan syndrome, a genetically inherited and potentially life-threatening condition?
- How is this decision managed in the present healthcare system?
- Who is responsible for the application of psychological care?
- Where are the potential implications for counselling psychology and psychotherapy in the current system?

#### **1.5 Contribution of the present study to the field**

The present research seeks to advance the understanding and awareness of Marfan syndrome and provide a non-medicalised perspective in line with the philosophy and aims of counselling psychology. This is expected to be of particular significance to couples and all those involved with helping them in making the decision to have a baby.

It is envisaged that it will be important to contextualise the findings of the research within a multidisciplinary team of healthcare professionals in caring for the needs of patients and their families affected by Marfan syndrome. It is predicted that the findings will imply suggestions for the teaching and training of counselling psychologists and psychotherapists, essentially helping those providing counselling services to Marfan patients to be more informed and therefore more confident and competent.

It appears that the gateway to psychological support is confined to the field of medical professionals, but curiously it seems there is a lack of understanding of the psychological implications of Marfan syndrome within the medical world. I consider that some knowledge and training regarding the psychological perspectives of Marfan syndrome is imperative for all healthcare professionals in the holistic care of patients.

In a wider context of the field, the present study may have significant implications for counselling patients with other genetically inherited conditions. A psychological counselling service may be needed, running alongside genetic counselling, providing an integrated service for patients making the decision whether or not to have a baby when there is a diagnosis of a genetically inherited disorder.

The present study seeks to provide a psychological perspective on genetically inherited conditions, thereby offering a contribution to extant psychological literature. Further, it will aim to highlight the potentially significant role of

counselling psychology when working with patients affected by genetically inherited disorders, particularly within the realm of research literature of other professionals.

### **1.6 Uniqueness of the contribution**

There are over 200 different disorders of the connective tissue. However, Marfan syndrome is unique in several ways. First, it is the most common of the disorders of the connective tissue. Second, many people with Marfan syndrome have heart and blood vessel problems such as weakness in the aorta or heart valves that leak. They may also have problems with their lungs, skin, bones, eyes and nervous system. Third, mutations are largely unique and therefore prenatal diagnosis is not an option for most families. This study is unique in that, to my knowledge, there has not been a research study carried out that has explored in depth the meaning of what it is like for a couple to make a decision to have a baby when a woman has a diagnosis of Marfan syndrome and is potentially at risk in childbirth, and that in the absence of prenatal diagnosis the genetic condition may be passed on to their child.

Potentially this research could offer a major contribution to providing psychological support for couples in their decision to have a baby when there is a female diagnosis of a genetically inherited condition. It would appear that a unique condition like Marfan syndrome could require a unique approach. Therefore, the implications for service development are considerable.

Additional professional psychological support could be offered to patients alongside the information and educational services of genetic counselling.

### **1.7 Personal reflexivity**

One of the essential criteria for this project was to follow the process of my position in relation to the research from its conception through to completion. Finlay (2009) highlights that in phenomenological research attention should be paid to the researcher's journey alongside the research process of how the data emerges in the encounter between the researcher and the participant. I have already stated how I came to choose the topic and in this section I aim to outline some assumptions and challenges that I encountered throughout the process.

I was aware from the outset that the decision to research a topic related to Marfan syndrome was perhaps a challenging choice. I knew that I was enthusiastic and motivated to know more about the psychological impact of this condition and my dedication would sustain me throughout the project to the end. My dedication to my family, interviewees, all those affected by Marfan syndrome and of course my profession as a counselling psychologist, were powerful motivators. It was also challenging because of my connection with Marfan and the responsibility of researching a sensitive subject.

I had to seriously consider my own safety and that of my family in researching a topic so close to my heart. My family followed me through the research and we talked on a regular basis to discuss any thoughts or feelings. My mother,

who does not have Marfan syndrome, had learned about the family diagnosis of Marfan syndrome when she was in her eighties and my weekly visits to take her out for coffee or lunch were therapeutic for us both, as we often discussed the research and family issues. Peer support was invaluable for encouragement and considering research-related pressures. My clinical supervisor provided a regular space where I could reflect on my safety and fitness to practice.

Following the decision to choose Marfan syndrome as the topic on which to write my doctorate, I was faced with the challenge of being an 'insider researcher' as I was already involved and associated within the field of the research topic. I was on the outside looking in and on the inside looking out; I had a dual role and this was not to be underestimated. I was mindful that as an insider, objectivity may not be achievable and therefore this could threaten the trustworthiness of the research. The reflexive methodological considerations of this are discussed in more detail in the methodology section. However, on a personal note, I was aware that being an insider researcher would need to be explored on a more intimate level. I realised that my attitude and my concerns regarding Marfan syndrome in my own family were very much part of my everyday life, in the background at times, but nonetheless a constant influence on my daily existence. I suppose that since Marfan syndrome was diagnosed in my family in 2004 I have just accepted that I do not have the condition, but when I considered this particular area to research, I came to realise how important my thinking and feeling regarding the condition of my own health would be. What did it mean for me not to have

a diagnosis of Marfan syndrome? In considering this, I came to understand that it had been lurking on the very periphery of my awareness for some time, an aspect of my family's diagnosis that I hadn't really accepted as being that significant. We tend to ignore so much of the information that comes our way, and with this in mind I noticed a heightened awareness in myself of every encounter with Marfan syndrome both professionally and personally.

I spent a considerable amount of time pondering on what a diagnosis of Marfan syndrome would mean to me and how it might be to experience such a meaningful change to my existence. However, this was not a productive exercise for the purposes of this research, not least because my standpoint as such is as one who has shared and observed members of my family's experience over the past nine years. For this reason, I focused on how my perceptions of that experience might affect the process of the interviews, my analysis of the transcripts and the research as a whole. This led me to wonder whether I should share the fact that there is Marfan syndrome in my family, and if I did, how this would affect the couples I would be interviewing.

It was inevitable that I would want to share the fact that there was a diagnosis of Marfan syndrome in my family. It was critical that I could be with the participants both emotionally and physically, thereby listening to them earnestly (Finlay, 2009). Moreover, I envisaged that it would prepare my participants to engage in an interview that would ultimately provide fruitful and truthful data, as well as introducing myself as not only a researcher, but also as someone holding a personal interest. I hoped they would feel more at

ease talking with me about Marfan syndrome as I had a good working knowledge of the condition and I would understand the medical terminology and any procedures they wished to share with me.

I am a mother, which was another consideration in carrying out this research project. My husband and I made the decision to have a baby fairly early on in our relationship. From my own experience, the decision-making process was a very private matter. However, it was necessary not to confuse the boundary between what was my experience and that of the participants. In recognising my own process, where I had the optimism to have a baby and the tenacity to see it through, whatever the circumstances, there was a clear need for caution in what I would be open to hearing, or not, during the interviews and the analysis. Therefore, it was crucial to be open to hearing multiple possibilities and not allow my own experience to shape the lens of the participants' narratives.

How would I have felt making the decision to have a baby if I had Marfan syndrome? Based on the experience of Marfan in my family, I think my decision to have a biological child would have been the same. I am not sure how my husband would have felt. Like many prospective parents we had concerns that the pregnancy and birth would go well and that the baby would be healthy. I have no doubt there would have been additional concerns if I had had Marfan syndrome. However, I believe my own tendency would have been to see the situation from an optimistic perspective and that decision-making would be in a positive light. Therefore, it was necessary to be aware

of how I would hear and interpret the participants' narratives and be open to the possibility that their experience might not be quite so positive or hopeful.

I felt that disclosing the fact that I was a mother might have had the potential to shift the focus of the participants' narratives during the interview, from their experience to mine. If they referred to it before or during the interview I would acknowledge that I did have a child. I intended to share these personal circumstances after the interviews if it was deemed appropriate.

At the beginning of the project, my starting point was that I assumed that the couples would go through some sort of decision-making process. On reflection, the foundation of my assumptions was that the pregnancy would be planned. This was casually challenged at a Marfan Association meeting in London when I described my research to one of the professionals there. He listened very carefully nodding and then concluded 'Do people make a decision? Don't they just drink a bottle of wine and it happens?' We both laughed, but the consideration of whether a pregnancy was planned or unplanned would have to be factored into the research. One female member of my family did not have to make a decision whether or not to have a baby as her diagnosis came several years after she had given birth to her children. I felt that this would be an advantage for me as an insider researcher; it would not be potentially problematic, as I would have no prior experience of decision-making that may affect the interview or the subsequent analysis of the data.

The medical literature on Marfan syndrome generally tends to be information giving and very often has a negative perspective of the condition. However, in sharing my family's world I have come to understand that it is not necessarily a totally negative experience. Of course there are numerous problems that have to be overcome, and the impact of a diagnosis of Marfan syndrome is never to be underestimated. However, it would be wrong to blindly assume that a woman with Marfan syndrome and her partner would be negatively affected in their decision-making to have a baby.

I was mindful that during my Masters research into women's experiences of breast cancer (Hinchliff, 2006) I had diligently read the medical literature. Prior to the interviews with the women, my perception of breast cancer was as a devastating and totally negative event, but this was to be challenged. I had wrongly assumed that losing a breast would be one of the worst things that could happen to a woman, as I perceived it would be for me. During the first interview I was sharply reminded of the rule of *epochè* (Husserl, 1970) as a participant created a fresh perspective on this experience for me. It was more devastating for her husband regarding her sexuality, while she compared it favourably to losing an arm or a leg.

Naturally, I cannot aim to be unbiased in the research process and acknowledge that my subjectivity will form part of the research process, but in assuming a reflexive stance I offer myself and the reader an insight into my motivation for exploring the inevitable assumptions and challenges surrounding the chosen topic from the outset. I believe this transparency is

one of the most profound aspects of the research process and reflexivity is considered in more depth in the methodology section and in the reflexivity section following the discussion.

### **1.8 Considering the role of counselling psychology in healthcare**

This is my second research project exploring a health-related topic, and it was probably inevitable that I would choose to research another health-related topic for my doctorate, as there has always been some considerable focus on health issues in my work with patients.

Although I now only see patients privately, my work for seven years as a counselling psychologist was mainly based in NHS hospitals and GP surgeries. In both the private and NHS work I provided therapy for a high number of patients with health-related issues. Sometimes the patient was the person suffering and coping directly with the illness, other times it could be a member of the family, a friend or colleague, who was helping or caring for the patient. There were many illnesses I had never heard of, but without exception I endeavoured to educate myself by talking to medical professionals and reading about the condition until I had a working knowledge of the illness. I was often frustrated to find that there was a dearth of information or literature from which I could learn more about a particular condition from a psychological perspective.

The discipline of health psychology focuses on the psychological impact of illness on individuals and their families. The interventions aim to promote

efficient self-management and effective coping abilities. The unique approach as a counselling psychologist is concerned with the underlying issues. Counselling psychology as a profession has the skills and potential to offer a different perspective than that of health psychology. For example, the patient and the counselling psychologist can form a collaborative relationship to consider psychosocial issues relevant to the presenting problem, thereby providing the ability and a constructive environment to explore the psychological impact of health-related issues in a wider context.

Counselling psychology is a comparatively new discipline in the UK. It was introduced as a section in the British Psychological Society in 1982 and evolved into chartered status in 1994 for students who wanted to specialise as an applied counselling psychologist. The role of counselling psychology has rapidly adapted and developed into a discipline that can be used in a wide range of healthcare settings. Corney (2006) highlights that this used to be the domain of clinical psychologists, but the unique skills of counselling psychologists are now valued in areas such as pain management and cardiac rehabilitation. Patients who are coping with health-related issues are not necessarily mentally ill, and both the counselling psychologist's and the clinical psychologist's training can help patients to understand their illness, develop coping strategies and improve their quality of life by making any required behavioural changes.

As a counselling psychologist, I have always welcomed the opportunity to work with patients with health problems. There are certain aspects of my

training and ever-evolving professional development that I have found to be invaluable in working with this particular population, their GPs, and other healthcare professionals. Based on the framework of the role of counselling psychologists in healthcare (Roth-Roemer, Robinson Kurpius & Carmin, 1998) these include:

- The importance of not losing my identity and core values as a counselling psychologist when working with other healthcare professionals
- The ability to create an environment, regardless of being in a clinical setting, that facilitates a good quality collaborative relationship with the patient to solve any problems
- The ability to be with the patient as a thinking, feeling, person with unique values who is experiencing a particular health problem, and not define them by their illness, condition or disease.
- Grounding the patient and their situation in the wider context of family, societal factors and healthcare professionals
- Identifying the main problems and the patient's strengths required for coping or enabling change
- Designing efficient and focused interventions to enhance the patient's confidence in, and awareness of, their ability to cope

- Respecting the differences and aiming to create a balance between me, as the counselling psychologist, the patient, and individuals of the healthcare team
- Identifying resources and other healthcare professionals who may be helpful in my work with a patient
- Competency to consult with medical and other healthcare professionals with the primary interest of my patient's well being
- Insight into how a particular healthcare professional and patient may work more effectively with each other and the capability to facilitate that change

In addition to our role working with patients, there is also the possibility that counselling psychologists have an important and potentially influential role to play in the recruitment, selection and training of healthcare professionals (Roth-Roemer et al, 1998). How the discipline of counselling psychology differs from clinical psychology is considered later in this project under the heading 'The practice of counselling psychology and psychotherapy'.

As the medical world has evolved, there is an increasing need for a biopsychosocial health model (Alcorn, 1998; Schwartz, 1982). Goals for the treatment of patients include prevention, effective coping for chronic conditions, and enhancing patients' quality of life (White, 1988).

Consequently, the roles for non-medical professionals such as counselling psychologists have increased within the healthcare environment. However, Alcorn (1998) highlights that additional courses and workplace experience may well be beneficial, if not necessary, for students intending to work within the multidisciplinary healthcare system. He suggests that a selection of modules or courses should be made available for counselling psychologists such as pharmacology, medical terminology, neuropsychology, health psychology, medical aspects of disability and community health. The experience of working in a GP surgery and my enthusiasm for my work certainly provided me with a working knowledge of all of the above. However, I would have welcomed the opportunity of this optional additional training. On further investigation, and to the best of my knowledge, supplementary learning of the above modules or inclusion in post-graduate counselling psychology training programmes, is currently either extremely limited or else not available in some parts of the UK. I make this proposal not with the intention of becoming more 'clinically minded', but providing our profession with speciality training that would enable an enhanced proficiency and understanding within a multidisciplinary team.

Defining the role of counselling psychology in healthcare settings is potentially problematic. With the relatively recent shift in the Health and Care Professions Council (HCPC) to include counselling psychologists, we have a health remit and it is clearly important that we retain our identity. This is an issue I have discussed with many colleagues, but one conversation resonated

with my thinking, and that is that “we are not health professionals, we are professionals in health” (Rawson, 2012).

### **1.9 Marfan in the Media**

Here I present some of the people in the public eye with Marfan syndrome to help the reader place the condition in a social context.

Since archaeologists first discovered the ancient Egyptian Pharaoh King Akhenaten (Tutankhamun) in the early nineteenth century, historians wondered what could have caused his quite distinct features; he is described as having a long, slender neck, a long face, almond-shaped eyes, long arms and fingers and a tall slender build (Lorenz, 1996). It was eventually considered that he most probably had Marfan syndrome (Burrige, 1995). Interestingly, some of his relatives had similar features.

The American President Abraham Lincoln is said to have had Marfan syndrome, based on his long limbs and joint hypermobility. The composer Sir John Tavener, whose cello piece, ‘Song for Athene’, was played at Princess Diana’s funeral, had life-saving emergency heart surgery in 2011 due to Marfan complications. Other musicians with Marfan syndrome include Nicolo Paganini and Sergei Rachmaninov; naturally the artistic long fingers associated with the condition are an obvious advantage when playing the piano (Stuttaford, 2004).

Jonathan Larson, who wrote the Broadway show *Rent*, had Marfan syndrome, but died from untreated aortic complications. Russ Hexter, director of the film *Dadatown*, also died from an undiagnosed aortic condition at the age of 27 years. Flo Hyman, an Olympic Volleyball player had an undiagnosed case of Marfan syndrome and died from a ruptured aortic aneurism. The basketball player, Chris Patton, also died undiagnosed in 1976. All the above tragic cases were undiagnosed, but on a positive note their fame has highlighted the need for an early diagnosis of Marfan syndrome, thereby leading to more effective life-saving treatment. In particular, the death of Flo Hyman caused widespread attention on the syndrome (Pyeritz, 1991).

Other well-known people who it is speculated may have had or have Marfan syndrome include Mary Queen of Scots, Joey Ramone from the punk rock band The Ramones, Bradford Cox from the bands Deerhunter and Atlas Sound, the actor and author Vincent Schiavelli, Robert Johnson the blues singer and guitarist, the swimmer Duncan Goodhew, Osama Bin Laden, and authors Jennie Logsdon Martin, Roanne Weisman and Mathew Rudes.

In addition to high profile individuals, there are also two films associated with Marfan syndrome. It features in the 1997 science fiction film, *Gattaca*, which is about eradicating genetically inherited conditions. The 2007 film, *Mo*, features a teenage boy with Marfan syndrome who lives on Long Island New York. It follows his journey through the challenge of coping with the condition in a physical and social context.

Marfan syndrome has a relatively low profile in the media despite the examples quoted here. It is often through a family diagnosis, as in the case of my family, or some connection to an individual with the condition, that the public are introduced to the condition. It is considered that a higher profile through the media would highlight the condition in both the public and professional domains, thereby leading to more effective diagnosis and treatment. I hope that this research may assist in raising the profile of Marfan syndrome.

## **2. LITERATURE REVIEW**

The aim of this review is to explore the extant literature relating to the overarching theme of couples' decision-making to have a baby when a woman has been diagnosed with the potentially life-threatening condition of Marfan syndrome. The review will first outline the implications of Marfan syndrome and pregnancy. Next, the role of genetic counselling is considered, its identity, strengths and potential limitations. The following section examines the research on how individuals adapt to a potentially life-threatening event and the implications for the care and treatment of patients. A critical review of the research literature on reproductive decision-making concerning people with a genetic condition is then explored, with a view to grounding the research topic in the context in which it occurs. With particular reference to counselling psychology and therapy in general, the couple relationship is explored as an important aspect of the current research. Finally, the current research is considered in the light of the reviewed extant literature.

### **2.1 Marfan syndrome and pregnancy**

The National Marfan Foundation, Inc. in the United States, the Marfan Association here in the United Kingdom, Marfan syndrome Support Group Ireland, and other support networks around the world are dedicated to educating and supporting both the general public and medical professionals. This, in part, has led to Marfan syndrome being recognized and diagnosed in early life. There have also been numerous important medical advances that have improved the diagnosis, medical care and life expectancy of individuals

with Marfan syndrome (Pyeritz, 1991; Nienaber & Von Kodolitsch, 1999). Earlier diagnosis has meant that affected individuals can have the opportunity to care for their health appropriately with healthcare professionals.

It has also meant that individuals or couples may think about planning a pregnancy in the light of their diagnosis (Peters, Kong, Hanslo & Biesecker, 2002b). As a result, couples may consider the risks and make a more informed decision regarding the 50/50 chance of having a child with Marfan syndrome. The clinical symptoms of Marfan syndrome vary in severity and therefore it is thought that the psychological impact of the condition on the couples and their future children should be a consideration.

Predictive screening tests, such as prenatal genetic diagnosis and preimplantation diagnosis, are available for some individuals with Marfan syndrome (Blaszczyk, Tang & Dietz, 1998). Prenatal genetic diagnosis aims to detect birth defects such as genetic diseases or conditions by testing the embryo or foetus before it is born. This is possible when the affected parent has been diagnosed with Marfan syndrome, as the test has to be carried out prior to 11 weeks gestation. However, as Marfan syndrome is such a complex and variable disorder the process of screening is considerably hindered. Furthermore, a positive diagnosis of Marfan syndrome may present additional concerns as to whether to terminate the pregnancy or not.

Preimplantation genetic diagnosis (PIGD) is where the embryo is screened for specific genetic diseases prior to implantation. This is usually under licensed

arrangements at specific hospitals, is a specialised process and normally available on a private basis, but there is some limited NHS funding available.

In making a decision to have a baby, couples have to consider a number of additional risks that pregnancy carries for women with Marfan syndrome. All pregnancies for women with Marfan syndrome are considered 'high risk' and it is recommended that women with Marfan syndrome have children earlier rather than later in life. A pregnant woman with Marfan syndrome will be referred to a hospital with specialist facilities in the monitoring and care of 'high-risk' pregnancies. A multidisciplinary approach is usually recommended for the care of these patients and their infants (Lind & Wallenburg, (2001).

Pregnancy and labour increase the stress put on the cardiovascular system, the heart and blood vessels. In any pregnancy there is increased stress due to the need for the baby and placenta to be supplied with blood. But extra precautions have to be taken in the case of 'high risk' women with Marfan syndrome throughout the pregnancy and beyond as there can be serious complications. Frequent blood pressure checks and echocardiograms are performed during the pregnancy and at least six months after the birth. High blood pressure is treated with urgency, and when the aorta becomes dilated more than 4cm there is a potentially life-threatening risk that it may rupture or cause regurgitation where the blood leaks back through a faulty heart valve.

There are also less serious complications for pregnant women in connection with Marfan syndrome. There may be some changes in eyesight and there

may be more stretch marks than usual which can be quite distressing. There is a risk of prolapse of the womb that may need to be treated, but pelvic floor exercises are usually suggested to help prevent this. Women with Marfan can also suffer from more aches and joint pain. Scoliosis (curvature of the spine) can be another problem for individuals with Marfan syndrome and this can cause respiratory complications, particularly in pregnancy. Backache is common in women with Marfan, but during pregnancy this can become quite severe and debilitating.

Until 1980 it was estimated that more than 50% of pregnancies for women with Marfan had significant Marfan-related cardiac problems (Pyeritz, 1991). As a result many health care professionals advised women against becoming pregnant (Shabetai, 1994). However, there is now a more optimistic outlook for couples wishing to have children when there is a female diagnosis of Marfan. Encouragingly, a significant number of women with Marfan syndrome successfully complete pregnancy without serious complications (Lind & Wallenberg, 2001).

## **2.2 The role of genetic counselling**

A family history of a genetic disorder means that couples go into pregnancy with an increased risk of conceiving a baby with that particular condition. Marfan syndrome is considered to be among the most common inherited disorders of the connective tissue.

The decision to have a child is usually a very personal one and it is accepted practice that the couples who are considering parenthood should make it themselves. But, when there is a diagnosis of Marfan syndrome in either partner, it is important that they recognize and understand any potential risks. As shown in the previous section, 'Marfan syndrome and pregnancy', it is particularly important when there is a female diagnosis.

Genetic counselling helps couples assess the risks so that they can then make an informed decision about whether or not they wish to have a baby. In addition there are, of course, alternative options to consider such as adoption, surrogate motherhood or donor artificial insemination.

Genetic counselling is often offered to Marfan patients, but it has been reported as providing technical and medical information with little time to discuss social and emotional needs (Sorenson, Scotch & Swazey, 1981; Kessler & Jacopini, 1982). However, it is difficult to define genetic counselling, as the perspectives, medical practices, goals and ethics vary according to the interests of the individuals defining it (Resta, 2006). However, genetic counsellors usually stay within their area of expertise of technical and medical information in order to avoid emotional issues (Bosk, 1992).

In attempting to understand the role of genetic counselling, it seems that at its heart is the process of communicating knowledge and a philosophical aim to be non-directive. My understanding is that genetic counselling is an umbrella

term for multiple activities such as diagnostics, facilitating decision-making, medical management of a condition, information-giving and how to break news or talk to friends and family about a genetically inherited condition.

One consideration that should be discussed with prospective parents is prenatal diagnosis, where the condition may be detected at an early stage in the embryo or foetus. The mutations in Marfan syndrome are largely unique and therefore prenatal diagnosis can only be offered to families where a previous mutation has been established (Rantamaki, 1995). Decisions involving prenatal diagnosis, which may lead to terminate the pregnancy or cope with any other psychological consequences, need to be considered. There is a wealth of literature on decision-making and termination in prenatal diagnosis of foetal complications, which outlines the clinical outcomes for parents and health professionals, but there is a dearth of research into the practical and psychological issues (Statham, 2002).

It is thought that for parents making such an important decision, a non-judgemental and non-directive approach is critical within the field of genetic counselling. However, it is debatable whether non-directive counselling is an achievable goal for trained genetic counsellors (Clarke, 1991; Mitchie, Bron, Bobrow & Marteau, 1997). Furthermore, patients may be seeking an answer from the clinical professionals and feel frustrated that an opinion is not forthcoming (Karp, 1983). There are many health professionals, untrained in a non-directive approach, who are involved in the decision-making, but as such these health professionals reported 'a variety of circumstances when

non-directive counselling did not seem to them to be possible, or to be the most appropriate response to the situation' (Williams, Alderson, & Farsides, 2002, p. 340)

Information management for families with genetic conditions is critical so that they understand the condition and realise the implications of genetic risk for other family members (Gallo, Knafl, & Angst, 2009). The term 'information management' is described as '...a complex process that involves the interplay of beliefs and behaviours related to accessing and interpreting genetic information, as well as making decisions and taking action based on information' (Gallo et al, 2009, p. 194). Genetic counselling provides information sessions and individual counselling to help patients accept, understand and evaluate their condition. Information management could therefore be an influential part of reproductive decision-making.

Prospective parents can access genetic counselling to assist with reproductive decision-making. However, Timmerman (2005) found that individuals often receive and understand genetic information incorrectly. They invariably found it difficult to understand or accurately perceive the risks and probabilities so as to be able to use them effectively in reproductive decision-making. Timmerman (2005) suggests that healthcare providers do not provide sufficient or clear information to individuals regarding their genetic condition.

It is difficult to assess genetic counselling and it has been suggested that there should be some method of assessment of retrospective client satisfaction with the service provided (Clarke, Parsons & Williams, 2008). Encouragingly, it appears that this lack of assessment is currently being addressed. McAllister, Wood, Dunn, Shiloh & Todd (2011) reported on a patient-reported outcome measure (PROM) for clinical genetics services, using a questionnaire Genetic Counselling Outcome Scale (GCOS-24). They found that the GCOS-24 had potential as a PROM.

I was interested to bring the role of genetic counselling more alive, so I contacted the clinical genetics department of several UK hospitals. I spoke with various professionals, who were most generous with their time and patience with my limited medical knowledge of genetics.

One clinical genetics department at the Royal Devon and Exeter Hospital invited me to present my research proposal. They were most interested in the research and it was an opportunity not only to share my psychological interest in the subject, but to also have a constructive critique from the 12 or so consultants and genetic counsellors who attended the presentation. They were most generous with their knowledge and I learned a considerable amount of up to date medical information about Marfan syndrome.

In addition to this meeting at the Royal Devon and Exeter Hospital, all the contact with different professionals involved in the care of Marfan was recorded in my personal journal. The following paragraph is a summary of the

main points of interest and information gathered from telephone, email and face-to-face conversations over three years.

Although the genetic departments received referrals of individuals and families affected with Marfan syndrome, they told me that there were not a large number of couples with Marfan, going through the decision to have a baby, referred to their departments. The role of genetic counselling was described as giving psychosocial support and care, and as specialising in helping patients in adapting and living with a diagnosis of a genetic disorder and bereavement. It was considered that it was a scientific and educational role incorporating psychological counselling skills when needed. If a patient became very distressed, my understanding of the procedure was that they were not allowed to make direct referrals for psychological counselling, so patients were referred back to their GP for a referral to a psychiatrist. Although there were psychology departments at the hospitals, Marfan syndrome patients were not routinely under their remit. Marfan syndrome was not at the top of the list and they felt that patients with Marfan syndrome and their families were not particularly distressed by their condition unless there was a death. Pregnancy was not reported to bother women with Marfan syndrome. The medical team needed to understand the complications, but in general there was not much difficulty except in the case of high risk of heart complications. Patients were not advised against going ahead with a pregnancy. When I asked about the lack of referrals of couples making a decision to have a baby with a female diagnosis of Marfan syndrome, I was

surprised to learn that, in general, they were not contacted when couples were thinking about starting a family.

Following up on this last point, Aalfs, Smets, de Haes & Leschot (2003) suggested that 10-20% of the women who attend a clinical genetics department are already pregnant when they attend for reproductive counselling. They explored the role of GPs in a genetic counselling referral and found that the main reason GPs referred a woman during her pregnancy rather than before was that the GP was not aware of any potential risks before the pregnancy, and as a result they did not have the opportunity to discuss a referral. Pregnant women were referred when they consulted their GP regarding concerns about the unborn child or they asked for a referral. Although the majority of GPs agreed that genetic counselling would be more beneficial before pregnancy rather than during pregnancy, they reported that their knowledge of genetic counselling was somewhat limited. Aalfs et al (2003) concluded that, although the patients not presenting before a pregnancy was confirmed undermined the GP's gatekeeper role, there was also a lack of knowledge, alertness and awareness among GPs regarding referrals for genetic counselling. They advocated that, as a result of their research, GPs should implement routine family history taking as part of good general practice.

It is considered that genetic counselling may not be sufficient to meet certain needs of people affected by Marfan syndrome. Smith, Michie, Allanson & Elwy (2000) examined the communication that takes place during genetic

counselling sessions. They analysed nine consultations with a single member of the clinical genetics team, finding that the clinician adhered to a clinician-centred rather than a patient-centred style of communication. These findings are consistent with research by Van Zuuren (1997) that although genetic counselling aspires to be neutral and non-directive, practice does not always reflect this. These findings have important implications for patients' needs, but I wonder what these needs are and how the profession of counselling psychology can help.

I would argue that, if this is the current level of psychological care offered to couples in their decision-making in living with Marfan syndrome, there may be a pressing need for counselling psychology to provide non-directive, non-judgemental, patient-centred counselling within the present system.

### **2.3 Research into psychological adjustment to a potentially life-threatening event**

#### ***Responding to a diagnosis***

How an individual with a potentially life-threatening condition responds and adapts to their illness is of interest to the current research due to the impact it may have on their psychological well-being and reproductive decision-making. Physical conditions can be associated with anxiety and depression, which can affect psychological health (Moussavi, Chatterji, Verdes, Tandon, Patel & Ustan, 2007). The literature on anxiety and depression within Marfan

syndrome is very limited. Given the mixed results of the extant research below I consider this to be a significantly under-researched area.

A diagnosis of Marfan syndrome may involve different types of coping styles such as denial, stoic acceptance, helplessness, hopelessness, anxiety or depression. Psychological distress, assessed using the General Health Questionnaire (GHQ), was found to be higher than expected in individuals with Marfan syndrome compared with the general population at large (Rand-Hendriksen, Sorenson, Holmstrom, Anderson & Finset, 2007). This finding suggests that perhaps there should be a routine assessment of psychological health following a diagnosis of Marfan syndrome and beyond, in order to address patients' psychological needs.

Fatigue and issues related to body-image and self-concept can be particularly problematic for individuals with Marfan syndrome. I would consider this to be a significant physical and psychological issue for individuals coping and adapting to the condition. Peters, Kong, Horne, Francomano & Biesecker (2001) explored the cognitive perceptions of the physical aspects of Marfan syndrome in 174 adults diagnosed with the condition. They found that in respect of the physical views of their condition 44% of their cohort reported significant symptoms of depression. They suggested that genetic counselling to address patients' perceptions of Marfan syndrome could improve their adaptation to the condition. I would suggest that psychological counselling might be more appropriate to meet their needs.

Congenital heart disease (CHD) shares many aspects of the physical and psychological characteristics of the cardiovascular problems associated with Marfan syndrome. The challenges for children and adults in coping and adapting to a diagnosis mirror many of the physical and psychological features of Marfan syndrome outlined above. In fact, Marfan syndrome is often included as a form of CHD in newborns for cardiology research due to the genetically determined defects that can be present at birth, but Marfan-related diagnoses later in life are rarely included (Hoffman & Kaplan, 2002).

Horner, Lithbertson & Jellinek (2000) highlighted that adolescents and young adults with CHD experienced problems with self-concept and body image, often feeling different from their peers as they coped with physical health restrictions, relationships and uncertainty regarding the future. Shearer, Rempel, Norris & Magill-Evans (2012) found that although adolescents reported many of the challenging life events and emotional problems associated with CHD, they didn't see themselves as any different from their peers. I cautiously summarise from these studies that although adolescents and young adults with CHD may face many psychosocial challenges as they grow up, which seemingly could present psychological concerns, there may be other variables such as demographics, personality and support systems that most likely play a part in adapting to the challenges of a diagnosis of CHD.

The psychological functioning of adults with CHD presents a mixed picture. Brandhagen, Feldt & Williams (1991) found that adults with CHD had higher

levels of psychological distress than the general population. Another study analysed data from interviews with adults with CHD and found that they perceived themselves as normal and that their psychological health compared favourably with the general population (Rijen, Utens, Roos-Hesselink, Meijboom, van Domberg, Roelandt, Bogers & Verhulst, 2003). One longitudinal study found both positive and negative outcomes regarding the psychological well-being of adult patients with CHD (Shampaine, Nadelman & Rosenthal, 1989).

### ***The importance of considering changes over time and gender***

In considering the outcomes of the above research there is probably a need for caution in assessing the psychological needs of children, adolescents and adults with CHD. Given the mixed positive and negative picture, a one-size-fits-all approach is most likely not appropriate but I believe it is important to acknowledge that at certain times in the lifecycle, such as decision-making to have a baby as discussed below, there may well be commonalities from a psychological perspective. However, I would argue that further research into the psychological impact of a diagnosis of Marfan syndrome might present a similarly mixed picture to the research studies outlined above, mainly due to the wide scope of demographic variables.

The age of an individual when they are first diagnosed with Marfan syndrome and indeed other genetic conditions could be a critical factor in how the condition affects them over the course of time. An early diagnosis in childhood of a potentially life-threatening illness may seem particularly

challenging. A diagnosis in childhood accompanied by medical tests and experiences may be traumatic for the child. There may be echoes of these early experiences later in life following other similar medical events in adulthood, and this could cause post traumatic stress (Kazak, Schneider & Kassam Adams, 2009; Manne, 2009).

McCabe (2011), a clinical psychologist, described how during the course of psychotherapy with a female patient aged 25 years, with a childhood diagnosis of a genetically inherited cancer, the patient reported that her traumatic memories of hospital from childhood impacted on a course of current medical treatment. However, this may not be typical behaviour of individuals diagnosed with other genetic conditions. McCabe (2011) acknowledges that her patient's childhood diagnosis of the illness was particularly extensive and required multiple medical interventions. She found that, although her patient's developmental journey from childhood to emerging adulthood in the face of life-threatening illness was characterised by all the same features of transition such as love, work, education, home and self-focus (Arnett, 2004) as other individuals without illness, she also had an over-developed stoicism, did see herself as different from her peers, but described herself as a better person and had learned to take care of herself and navigate her way assertively round the medical world (McCabe, 2011). The above study is a single case study and therefore we cannot generalise from it. However, a childhood diagnosis of a chronic illness can have the potential to normalise the condition in adolescents among their peers (Shearer et al, 2012).

The psychological understanding of adjustment to a life-threatening illness over the course of time is not well understood. Some cross-sectional studies have considered various phases or aspects of a specific life-threatening chronic illness, but the length of the course of time that an individual has lived with the diagnosis of a condition varies considerably (Rolland, 1984). A recent qualitative study using semi-structured interviews examined the experience of 32 individuals living with the uncertainty of a chronic life-threatening condition concluded that over time it produces complex and paradoxical experiences (Bruce, Sheilds, Molzahn, Beuthin, Schick & Shermak, 2014). They found an overarching theme of pervasive liminality where the participants' narratives moved between the experience of feeling fear and feeling fearless, being alive but not living, certainty and uncertainty, and the invisibility and visibility of the condition (Bruce et al, 2014). It seems to me that individuals living with the enduring nature of a potentially life-threatening condition like Marfan syndrome might experience a similar liminal position.

Similar to Marfan syndrome, each child of an individual with genetically inherited breast cancer has a 50% chance of inheriting it (Werner-Lin, Rubin, Doyle, Stern, Savin, Hurley & Sagi, 2012). In previously researching the counselling and psychotherapy needs of breast cancer patients, I found that there is a need for mental health professionals to have a working knowledge of the medical aspects and psychological factors involved in breast cancer and its treatment in order to support women going through the experience of coping with a potentially life-threatening condition (Hinchliff, 2006). Similarly,

Ward, Hogan, Stuart & Singleton (2008) found that patients with myalgic encephalitis (ME) experienced negative reactions to counsellors who did not understand their condition. I believe these studies may have significant implications for the psychological care of patients affected by Marfan syndrome.

Psychotherapy research suggests that, in general, clinically relevant differences in treatment needs exist according to women's different social and cultural circumstances in life (Bankoff, 1994). Van Tongerloo and De Paepe (1998) investigated the psychological effects of Marfan syndrome in 17 patients aged between 16 and 35 years and found that anxiety and depression levels in female patients were higher than the male group. Indeed, women score higher than men on self-report measures of symptoms and distress in illness (Verbrugge, 1980). Wingard (1982) highlights that women deal more effectively than men with health problems as they are more likely to seek social support from others. However, it is important to emphasise that in general there are gender differences in distress (Verma, Balhara & Gupta, 2011)

Rolland (1984, p. 245) writes, "One's concern is the interaction of a disease within an individual, family or other biopsychosocial systems". Therefore medical or physical problems have significant implications for the relationships among a person's family members, their healthcare providers and work associates. Furthermore, it has been suggested that patients suffering from chronic or life-threatening conditions should be viewed in the context of all

other relationships in any particular setting of their lives (Altschuler, 1997). In order to fully understand how possible problems associated with coping with a life-threatening event are defined, there may be a need to consider various possible contexts in which any of the problems may have occurred (Van Trommel, 1983).

When coping with a diagnosis of a life-threatening event, a woman's self-construct can be attacked (Keitel and Kopola, 2000). The needs of the family are often overlooked as everyone is concentrating on the needs of the patient (Fallowfield, 1992). Froma Walsh (1998) presents a contextual framework in understanding family resilience in facing the challenges of a potentially life-threatening genetically inherited condition. She believes that the keys to family resilience are: 'family belief systems' - being positive and making meaning out of adversity; 'organisational patterns' - being flexible and connected, and having adequate social and economic resources; 'communication processes' - clear and emotional expressions, and the ability to solve problems collaboratively. I would argue that these are particularly important points to consider for families with a genetic condition such as Marfan syndrome, as there is most likely the very real issue of uncertainty surrounding transmission of the condition.

Reibstein (2004) suggests that it is not always easy coping with the reactions of some family members, friends and work colleagues; some relationships may deteriorate and others may become stronger. She suggests that when others are supportive, in particular the woman's partner, they can offer

emotional and practical help in coping with any adverse consequences that may arise. She adds that, conversely, relationships could be problematic if people are avoidant. Interestingly, there is a correlation between secure adult attachment styles and positive psychological adjustment to illness (Rolland, 1984), which I believe is an important factor in psychological adjustment.

I would suggest that an adult female diagnosis of Marfan syndrome would have familial implications for her subsequent care and treatment, and that a woman would have to cope with significant psychological and social changes to her life, in particular due to the potentially life-threatening problem associated with Marfan syndrome in connection with the cardiovascular system.

Adapting to a life-threatening situation involves a sequence of adjustments. Taylor (1983) proposed a theory of cognitive adaptation to threatening events, maintaining that cognitive adaptation was a response by an individual attempting to restore or enhance their previous level of psychological functioning. One could argue that some elements of the findings of pervasive liminality in the study cited above by Bruce et al (2014) may have some of the characteristics of Taylor's (1983) theory of cognitive adaptation.

It has been suggested that as human beings we are sensitive to a threatening environment. We have a tendency not to like imbalance and can react to it by focusing on maintaining homeostasis. It is through constant changing cognitive processes and behaviours that an individual copes effectively with external and internal threats or demands (Lazarus & Folkman, 1984).

Attributions are thought to be important in adjusting to a threat or change in one's environment as they help an individual to understand, predict and control the threat (Kelley, 1967). However, one research study found that, although social cognitions and attributions might be important to the adjustment process, cancer patients' attributions for their cancer were generally unrelated to their adjustment (Taylor, Lichtman, & Wood, 1984).

These studies present a complex picture of women's experiences of a life-threatening condition and demonstrate that different and specific contexts should be considered alongside how women manage their experience. It seems to me that the intrapsychic processes that shape the perception of how the patient views the self should also be considered. In my opinion it is necessary to address the interface between how either partner in a relationship thinks about the self, and how they are in relation to others. Maybe there is a need for counselling, in addition to genetic counselling, for couples at risk with Marfan to help support and maintain their relationships through pregnancy and beyond, thereby facilitating the care and support their families may need.

It seems that psychological adjustment to a potentially life-threatening condition such as Marfan syndrome is a complex process and the above research presents a mixed picture. There may be a need for appropriately trained counselling psychologists and psychotherapists, with a working knowledge of genetic conditions, to become involved in the care of patients of

all ages by identifying and managing psychological and psychosocial problems.

#### **2.4 Reproductive decision-making with a genetically inherited condition**

Apart from Peters, Kong, Hanslo & Bieskecker (2002b) who specifically considered quality of life and reproductive planning in individuals living with Marfan syndrome, it appears that there is a dearth of research papers aimed at the psychological experience of reproductive decision-making of individuals or couples affected by a genetically inherited condition like Marfan syndrome. Also, I found that research into reproductive decision-making for women with CHD was clearly lacking and apparently very little is known about this aspect of the condition (Kilpatrick & Purden, 2007). However, there are a number of papers that have explored and considered reproductive decision-making in other genetically inherited disorders (i.e. Kessler, 1989; Frets, Buivenvoorden, Verhage, Niermeijer, van de Berge & Galjaard, 1990; Evers-Kiebooms, 1995; Lippman-Hand & Fraser, 1999). More recently, other studies have addressed the psychological processes involved in making a decision to have a baby when an individual has a known diagnosis of a genetically inherited condition (i.e. Kay & Kingston, 2002; Gallo, Wilkie, Suarez, Labotka, Molokie, Thompson, Hershberger & Johnson, 2010).

Research to date presents a mixed picture regarding decision-making with a genetic condition. Kay & Kingston (2002) examined qualitative data collected from interviews with 14 Caucasian women aged between 20-39 years who did not have an affected child and were known carriers of an X-linked condition

that, similar to Marfan syndrome, causes a faulty gene in both males and females. There is a range of X-linked conditions, but Duchenne muscular dystrophy (DMD), Menkes syndrome and Lesch-Nyhan syndrome are comparable to Marfan syndrome as they are progressive and potentially life-threatening conditions. Also similar to Marfan syndrome, carriers of this condition have a 50% risk of passing the faulty gene to their children.

A complex picture emerged in their study of the women's feelings regarding reproductive decision-making, in particular that it was dependent on their personal experience of the condition. Interestingly, having a male sibling with the condition meant that the women were more determined to avoid having a child affected with the same condition. The women experienced a certain amount of guilt as they considered and imagined the consequences of having an affected child with the condition. Decision-making was influenced by historical family patterns of inheritance of the condition, and the women used the patterns to predict the chances of having an affected child. In one case this led to one woman having a termination without evidence that the child may be affected. The women felt responsible for taking control of the reproductive decision-making (Kay & Kingston, 2002).

There were some limitations to the study. Although none of the women had children affected by the condition, there was quite a wide age range (20-39) in the women who were interviewed and it was unclear from the study which of the women already had children. Another issue that may have affected the findings was that not all the participants had a positive diagnosis of an X-

linked condition; they were either known or highly likely carriers. There is a possibility that these factors could have affected the trustworthiness of the research due to the lack of homogeneity of the sample.

The Kay & Kingston study is of interest to the current research project as it raises the question of what particular psychological and situational factors potentially determine and predict the reproductive decision-making process not only for a woman with Marfan syndrome but also her partner. This research could potentially generate new knowledge for counselling psychologists and all professionals concerned with helping couples making the decision to have a baby.

Another investigation that has contributed to reproductive decision-making with a genetically inherited condition is that of Gallo et al (2010). They interviewed a focus group of 15 African American participants with sickle cell disease (SCD), an inherited condition most commonly found in people of African, Mediterranean, Middle Eastern and Indian origin. The majority of the participants were women and their ages ranged between 36-63 years. The findings included coping with health-related issues, choosing a partner and sharing the experience of the condition with that partner, but it is the multifaceted findings related to making an informed decision regarding reproductive options that is of particular interest to the present study. Some of the women quite clearly expressed a very strong desire to have biological children despite the risks involved. Other women confessed that, retrospectively, they would have reconsidered their decision to have a child

now knowing how severe and complicated the condition could be. A couple of the participants shared that they wouldn't even take the risk and would consider adoption or other methods. Some of the participants thought that prenatal diagnosis was a good idea so that if the foetus was affected they could psychologically prepare for the future. Conversely, however, the participants who had been offered an amniocentesis test had turned down the opportunity, fearing that the foetus would be harmed. Some of the participants considered that they would terminate a pregnancy if the mother or foetus were at risk, whereas others would not terminate a pregnancy on the grounds of personal or religious beliefs. Curiously, considering the date and current nature of the research, without exception, all of the participants were unaware of *in vitro* fertilisation (IVF) or preimplantation genetic diagnosis (PGD) as reproductive options. The overwhelming belief of the participants was that the desire to have biological children was so strong that the other options were not even considered.

The reliability of this study could be questionable as the reports of the participants were retrospective and, with such a vast age range of 36-63 years, there will undoubtedly be some considerable variations in knowledge of the condition, options available, societal expectations and personal attitudes in the process of reproductive decision-making over that period of time. Furthermore, the study questionnaires lacked data collection on demographic information on the number of children in the participants' family or their choices to have or not have children.

Nevertheless, the study by Gallo et al (2010) is significant for the present research study as it highlights the complex and multidimensional aspects of decision-making. This suggests that there is a need to appreciate individual variation in context and that there can be no universal answer to the dilemma of decision-making. The value of the current study is the homogeneity of the sample and the in-depth exploration of the experience of decision-making for couples, primarily with a focus on improving the psychological care of patients within a multidisciplinary team.

An investigation using quantitative and qualitative methodology that has contributed to the knowledge of reproductive decision-making specifically in adults affected with Marfan syndrome is that of Peters et al (2002b). They studied the factors that influenced reproductive planning and quality of life in 174 individuals aged 18 years or older with Marfan syndrome. The majority of the participants were reported as being women (101 participants).

Approximately 62% of the participants agreed that having Marfan syndrome was a significant consideration in their reproductive decision-making, in particular for individuals diagnosed at the age of 15 years or younger. There were several suggested possible reasons for this correlation. First, an early diagnosis could bring an increased awareness of the implications of pregnancy and childbirth and therefore these issues were considered from an early stage in the reproductive decision-making process. Another explanation could be that participants who were diagnosed early may be more physically affected and therefore pregnancy and childbirth may be viewed as more risky,

especially for the female participants. One further explanation could be that Marfan is considered more life-changing when there is an early diagnosis than when it is diagnosed later in life.

The value of this study is that it has increased our awareness, understanding and knowledge of the clinical and psychosocial issues for individuals with Marfan syndrome in the reproductive decision-making process. Further it has provided valuable insights into both attitudes and behaviours in relation to reproductive decision-making. One clear advantage of this research is that it has highlighted the need for a psychological component alongside the educational approach for genetics professionals.

The authors acknowledge that there are several limitations to the study such as bias due to self-reported data and self-selecting sample. There was no evidence that the respondents were clinically affected with Marfan syndrome. Also they were mainly Caucasian and therefore the authors emphasised that the findings should be viewed as exploratory and may not be generalisable to the wider population.

I would add that further information on contextual factors in the participants' lives, such as the strength of the marital relationship and ages of their children should have been investigated and reported as these may have influenced the reliability of the study. Furthermore, it would have been interesting to know the ages of the participants and whether they are male or female, as these are important considerations when researching reproductive decision-

making. Lastly, aside from the lack of generalisability due to sample demographics, there is also the undefined age range of the sample aged 18 years or older, which is an overwhelmingly significant factor in self-reports of reproductive decision-making.

Although there are some significant and relevant psychological findings in the Peters et al (2002b) study, many of them are speculative due to the undefined age range and they lack understanding of the underlying processes involved in couples decision-making. The current research aims to provide a much broader and richer picture of the experience of decision-making within the coupledness, using purely qualitative research.

## **2.5 The couple relationship**

The aim of this section is to define and make explicit the importance of the couple relationship for this and other research involving decision-making to have a baby when a woman has a potentially life-threatening condition. Furthermore, it seeks to consider the implications for counselling psychology and psychotherapy.

Defining the couple relationship is multifaceted, but there is an overwhelming agreement among the major theorists that this and other close relationships include an element of interdependence (Campbell & Rubin, 2012). Bowlby (1969) incorporated this in his theory of attachment and Kelley and Thibaut (1978) described the interdependent nature of close relationships whereby an individual's thoughts, feelings and behaviours are inextricably linked with the

other person. Therefore, the couple relationship could be defined as a collaborative and shared experience of the dynamics that exist between two people.

In creating a close couple relationship, individuals generally make a considerable psychological investment involving commitment (Rusbult, 1980), building trust (Rempel, Holmes & Zanna, 1985), and the ability to grow as an individual within the relationship (Aron, Aron, Tudor & Nelson, 1991). But, as discussed, it is the interconnections existing between the individuals as such that define the couple relationship as 'greater than the sum of its parts'. It has been suggested that this relationship cannot truly be observed, as it is ultimately invisible, like the powerful forces of nature such as electricity, the four winds and gravity (Bersheid, 1999).

My understanding is that defining a couple in the psychology of society has not been heavily explored as a research area. The definitions outlined above suggest that a couple should be considered as a separate entity in its own right as the couple quality overrides the individual. I believe that the concept of the couple is important and worthy of investigation. Although there has been an increase in the study of close relationships over the past two decades (Campbell & Rubin, 2012), the extant literature is scant and most research focuses on the individual processes. In particular, an extensive literature search revealed that there was an over-dominance of research focused on the individual in psychology and psychotherapy. Methodologies have invariably reflected the emphasis on researching the individual but in the

last decade researchers have become more creative in devising methods to accommodate relationships as suggested by Bersheid (1999).

For a couple, having a child is considered by society as a natural progression in the life cycle (Daniluk, 2001). It is suggested that for some men it is regarded as a major contribution to a fulfilling family life, providing status and companionship over the years (Mason, 1993; Owens, 1982). Whereas women, who generally have a slightly stronger desire than men to attain the goal of having a child (Brase & Brase, 2012), are often thrown into confusion about their identity and what they really want from life when they eventually become a mother (de Marneffe, 2004). Studies have shown that following the birth of a child, women reported a decline in their levels of fulfilment within their marital relationship, while men's levels of marital satisfaction remained relatively stable (Lawrence, Nylén & Cobb, 2007; Shapiro, Gottman & Carrere, 2000).

Predictably, research has focused on the individual experience, but there have been some interesting findings, in particular in the field of infertility. Research has reported that women cope and accept childlessness with less distress than men (Koropecky-Cox & Pendell, 2007; Dalzell, 2007). However, it has been suggested that there is a higher risk of illness, depression, mortality, isolation and exclusion in both men and women (Dykstra & Hagestad, 2007; Kendig, Dykstra, van Gaalen & Melkas, 2007). It would appear from the above studies on infertility that much is at stake for couples in their decision-making to have a baby. The challenges of making

that decision when a woman has a potentially life-threatening health issue such as Marfan syndrome may outweigh the prospect of being childless. These studies and others focusing on the intrapsychic processes neglect the overarching interactive space between the individuals, thereby losing a sense of the effects of 'other' in the self. Essentially, it is considered that this is an important aspect of couples' research, with far reaching implications for counselling psychology and psychotherapy.

Interestingly, other studies have found that when there is a stressful life event, partners within the coupledom may respond in many different ways emotionally to maintain a homeostatic state in the relationship (La Guardia, 2009). They may seek closeness and reassurance from the other and form a collaboratively supportive environment (Mikulincer, Florian & Hirschberger, 2004). Alternatively, they may become less open and responsive to each other (Randall & Bodenmann, 2009), withdrawing and distancing themselves from the other in order to reduce feelings of distress, vulnerability or fear (Rafaeli & Gleason, 2009).

Kilpatrick & Purden (2007) found that women with CHD considering pregnancy, resolved their internal decision-making issues before discussions with medical health professionals or their partners. Although the women had concerns about their own health, the health of the baby and the risk of passing CHD to their child were of particular concern. The women gathered as much information as they could before they discussed the subject with their husband, as they wanted to make sure they were as fully informed as

possible to answer any questions he may have and be in a strong position to reassure him concerning any health risks in pregnancy and for the baby.

Couples based interventions for significant life stressors such as health issues are currently limited or not available and typically involve short-term individual psychological therapy or stress management educational programmes (La Guardia, 2012). These are usually aimed at the patient (Linden, Philips & Leclerc, 2007) and the success rate in reducing psychological symptoms such as anxiety and depression is low (Manne, Rubin, Edelson, Rosenblum, Bergman, Hernandez, 2007). Couples often only seek therapy when their relationship has broken down and there is considerable unhappiness, whereas preventative intervention would help couples to cope with challenges and life's transitions (Cowan & Cowan, 2012).

Understanding couples' interactions under a significant life stressor is perhaps critical to understanding their associated emotional and psychological health. In better understanding the interpersonal dynamics of couples at times of challenging life events, we may have the opportunity to identify potential psychological consequences, thereby providing an opportunity to promote timely and appropriate referrals for couples-based interventions.

## **2.6 The current research**

The cited research studies above indicate that individuals are indeed influenced by factors relating to a genetically inherited condition, and highlight that there is a need for an increased understanding of the processes and

influences that may impact on couples in the decision to become parents when there is a genetic risk involved. Although the studies have attempted to research and theorise on the factors that may influence decision-making, they present a rather simplistic overview and neglect the complexity and diversity of an individual's experience. What is lacking is an increased understanding of the interpersonal and intrapersonal relationships that would be of considerable value and interest in the care of patients. The rarity of such studies has inspired and informed the current research.

The current study emphasises the need for an in-depth exploration of the lived experience of reproductive decision-making by interviewing individual couples to gain meaningful insights into both their interpersonal and intrapersonal worlds. Particular consideration will be given to the meaning and implications of the impact of the genetically inherited condition Marfan syndrome. An in depth understanding of what it means to experience such a phenomena, in personal, social and world view realms, for a couple, is one of the aims and objectives of the current research project. Recognising their needs and the potential for counselling psychology to address these needs and enhance the existing care and support system are the ultimate aims of this research.

### **3. METHODOLOGY**

In this section I consider the process of deciding on a research design and methodology. I offer an explanation of why I chose a qualitative approach and a phenomenological method and how this influenced my eventual choice of using interpretative phenomenological analysis (IPA) for the project. Participant recruitment and design are discussed. The process of gathering the data in the interviews follows this. I then offer a detailed explanation of how the data were analysed. Trustworthiness is considered and I outline the ethical issues and give an account of how these were addressed. The important issue of being an insider researcher is carefully examined. I explain my epistemological stance and finally there is a section on reflexivity, which is considered as imperative in good qualitative research.

#### **3.1 Why qualitative methods**

Much thought and consideration was given to deciding upon the most appropriate methodological design to explore and understand the process of how the participants made their decision to have a baby. The overall process of data gathering and analysis quite clearly required an approach that would encompass not only the subjective experience of the participants, but also the underlying meaning of the experience and how they made sense of it. I was mindful that different research methods have their strengths and limitations (McLeod, 2002), but quantitative methods were considered unsuitable for the present study, as it is difficult to measure thoughts, feelings or underlying

meanings (Smith, Flowers & Larkin, 2010). Therefore, I considered a qualitative approach for this piece of research was appropriate.

While quantitative research functions mainly on a macro level and could highlight the dynamics, and the negative and positive outcomes of couples experience of decision-making to have a baby when a woman has Marfan syndrome, qualitative research has the ability to function on a micro level and explore the underlying processes by studying the human being as an individual. I concluded that research functioning on a micro level would be potentially helpful to counselling psychology and psychotherapy for understanding the psychological processes of people affected by Marfan.

Qualitative research has frequently been viewed as not being as scientific as quantitative methods. I am aware that qualitative research is often criticised as unavoidably biased due to its subjective nature, lacking in reliability, mere storytelling and intuition, being based on assumptions (McLeod, 2002). Furthermore, qualitative research methods could be challenged as being vague, shrouded in mystery, lacking in clear comprehensible guidelines and using inaccessible language (Brocki & Wearden, 2006). I believe this is a misinterpretation of qualitative methods that are valued in social psychology, education and ethnographic studies.

Mishler (1984) highlights the importance of producing a piece of qualitative clinical research in a language that is accessible to both patients and practitioners, claiming there is a need for researchers to listen to the real world

and to be able to communicate effectively without the use of 'jargon'. Therefore, I considered that choosing an accessible, transparent and well-defined qualitative method for the current research was fundamental and of the utmost importance.

In qualitative research the hypothesis evolves as the research progresses, and the analysis usually starts at the same time as the data is being collected. However, I was mindful that I would have some unavoidable preconceptions despite attempts to bracket and suspend them, and therefore these formed both strengths and weaknesses of the final thesis. Relationships between people are complex and require an intimate analysis in order to comprehend what is going on. In comparison, quantitative research requires simplified measurement procedures and elements of control that Walker (1985, p. 11) cautions might 'lead one to ignore the fundamental complexity of certain social phenomena'. The study of people, in particular counselling psychology and psychotherapy research, requires a methodology to understand what is going on from within.

### **3.2 Considering approach and methodology**

Exploring the various qualitative approaches within psychological research methods to find an ideal choice can be confusing. A one-size-fits-all approach would clearly be inadequate to study the rich diversity of human beings (Ashworth, 2009). Having explored various qualitative approaches for the current study, I deemed two approaches suitable: grounded theory and interpretative phenomenological analysis (IPA).

Grounded theory was a serious contender as it is well understood and respected in a wide range of research literature. There are many similarities between IPA and grounded theory; in particular they both share similar terms and techniques used for analysis (Giorgi & Giorgi, 2009; Smith, Flowers & Larkin, 2010). Traditional grounded theory (Glaser & Strauss, 1967) and its more recent derivatives (Strauss & Corbin, 1998; Charmaz, 2003) would have developed and provided a theory of the process of decision-making. However, I did not merely intend to consider the social processes (Willig, 2001) or the various issues influencing their decision-making (Smith, Flowers & Larkin, 2010), which would establish a theory just to explain the reasons for the participants' experience. I was seeking to understand the meaning and underlying processes of their experience of decision-making. Additionally, I rejected grounded theory on the basis of pragmatics; the sample for this project would be drawn from a relatively small population of 10,000 people of which only 5,000 would be female. Grounded theory typically requires interviews with 20-30 participants in order to 'saturate' the categories and develop the theory. I therefore concluded that it was not ideally suited to the aim of this research, and thus turned to phenomenological approaches.

### **3.3 Why phenomenological research methods**

Phenomenological research can use powerful methodologies for understanding subjective experiences and gaining insight into an individual's thoughts, feelings and behaviours. Phenomenology is one of the basic tools of qualitative research (Osborne, 1994). Phenomenological research is still based on the processes originally identified by the philosopher Husserl (1931):

bracketing-off assumptions, horizontalisation (no one meaning is thought to be more important than another), thorough description and searching for the essence of the phenomenon. It seeks to describe rather than to explain experiences and perceptions, without the bias of preconceptions or hypotheses (Husserl, 1970).

My understanding is that a phenomenological approach is concerned with studying experiences from the participant's perspective while acknowledging that interpretation of the interview material is through the lens of the interviewer, who is another human being with their own experience of the world. Colaizzi (1978) highlights that despite every effort to bracket off prior assumptions and perceptions (Husserl, 1931), phenomenologists acknowledge that this is impossible and some bias will remain. I agree that a researcher's competent engagement with participants will naturally facilitate some bracketing (Smith, Flowers & Larkin, 2010). I acknowledge that as a researcher I should attempt, as far as it is possible, to suspend beliefs and suppositions, and focus on events and experiences pertinent to the research process as they unfold and evolve, while acknowledging their potential role.

Phenomenological research is based on the assumption of intentionality. All meanings are construed in relation. It is not possible to separate the subjective and the objective, the observer and the observed or the 'I' from the 'not I'. Husserl's (1931) notion of the lifeworld (*Lebenswelt*), encompasses an individual's experience of their physical body, the self and their relationships with others, together with their perception of the world around them and how

they relate objectively in that world. The lifeworld is the inter-relational social world of an individual's everyday lived experience, and not the introspective inner world. It is pre-reflective (Finlay, 2008); a world that happens before we are even aware of it or can find the words to describe it.

Mearns and McLeod (1984) suggested that qualitative researchers required the facilitative therapist qualities identified by Carl Rogers (1961) - empathy, congruence and acceptance. Although McLeod (2002, p. 198) states that 'Any discussion of the personal qualities of researchers is highly embarrassing and goes against the grain of scientific research', I do believe that certain therapeutic qualities are necessary in order to carry out phenomenological research. The phenomenological researcher shares similarities with Spinelli's idea of the therapist as 'the potential co-author of a new, less problematic, narrative' for the client (Spinelli, 2001, p. 6).

I was mindful that it was important not to make definite what is not definite (Dahlberg, Dahlberg, & Nystrom, 2008), and I did not want to lose the essential phenomena of experience, such as insightful emotions, perceptions and social relationships in interviewing the research participants. Employing the fundamental stance of naivety or un-knowing (Spinelli, 2006) exposes cavernous channels of potential access to an individual's world with countless possibilities. As Merleau-Ponty (1992, p. 112) states, 'the psychologist does not know what he is dealing with', enticingly leaving the research phenomena to 'speak for itself' (Morley, 2012, p. 586).

Phenomenological research requires organisation and care with the data collection, analysis and beyond. The aim of phenomenological research is to clarify the underlying structure of any lived-in experience, and to identify the meaningful aspects of the complexities, ambiguities, paradoxes and uncertainties of an individual's experience. For the purposes of research into counselling and psychotherapy with Marfan-related patients, I would argue that a phenomenological approach is required. The lived experience is important in phenomenological enquiry and this is important for Marfan patients because of the impact a diagnosis of Marfan syndrome may have on their world.

Phenomenological research is an opportunity to explore how any particular couple responds to the experience without losing any details in the process, thereby gaining a more general understanding of the experience of Marfan syndrome on a couple's world in their decision to have a baby. I considered that Interpretative phenomenological analysis was an appropriate choice.

### **3.4 Interpretative phenomenological analysis**

Interpretative phenomenological analysis (IPA) has multidimensional foundations: phenomenology focuses on an individual's perceptions and experiences of their world; hermeneutics is the theory of interpretation; idiography is concerned with the experience of the individual; and symbolic-interactionism is a two-way process which relies on reciprocal social interaction between the co-researcher and the researcher, resulting in a dynamic process of production and interpretation of meanings (Smith, Flowers

& Larkin (2010). Essentially, I as the researcher sought to understand the world of the participant, from their individual perspective, as they experienced their particular circumstances.

IPA has roots in Husserlian philosophy, and incorporates the work of several major phenomenological philosophers, namely Heidegger, Merleau-Ponty and Sartre. It also shares the theory of hermeneutics from an interpretative perspective, drawing on the ideas of theorists such as Schleiermacher, Heidegger and Gadamer. Heidegger's radical shift towards an existential and hermeneutic perspective contributes some crucial features to the theory and methodology. Essentially, that a human being is a *Dasein* or 'being there' (Heidegger, 1962), or as it has been translated 'Being-in-the-world' (Spinelli, 1989). Rejecting the Cartesian notion of division between mind and body; object and subject; person and world, human beings are literally Beings-in-the-world interwoven with other people and the things which surround them (Eatough & Smith, 2008). Merleau-Ponty eloquently describes this as 'man is in the world, and only in the world does he know himself' (1962, xi). To explain further, my Being-in-the-world or *Dasein* can be referred to as 'a human existence' and my body will sense and reveal for me a world that is unique to me and different from the world that another individual's body will reveal to them (Eatough & Smith, 2008). This has particular relevance for IPA and its focus on an individual's intersubjective experience (Eatough & Smith, 2008).

In my opinion IPA was an ideal choice as a qualitative approach, which is theoretically rooted in phenomenology, hermeneutics, symbolic interactionism and idiography (Smith, 2011a). IPA has been used in a wide range of research into the experience of illness (Arroll & Senior, 2008; Borkoles, Nicholls, Bell, Butterfly & Polman, 2008; Bramley & Eatough, 2005). Furthermore, it is commensurate with the National Health Service's concerns with service-users' experiences of the NHS and its recent surge of patient-centred research (Brocki & Wearden, 2006).

IPA is considered to be of significant value in health-related psychological research, especially where professionals are not within their particular field of expertise (Biggerstaff & Thompson, 2008). Approximately a quarter of IPA studies are health-related topics (Smith, 2011a). IPA's unique approach allows for considerable 'freedom and creativity' (Willig, 2001, p. 69), and I consider these qualities as relevant in researching a novel situation or exceptional population as in the present research. I believe it is a suitable methodology, as it would illuminate the meaning of the lived experience of decision-making for the participants and at the same time explore the language they use to explain how they make sense of that experience cognitively, emotionally, and physically.

I was mindful that IPA has been criticised for its use of the term 'cognitions' as in the sense of scientific information processing and therefore not phenomenological, since genuine phenomenological philosophy rejects the mind and body divide (Langdrige, 2007; Willig, 2001). In response, Eatough

& Smith (2008) clarify their conceptualisation of cognitions as not the subject and object divide suggested by the information processing accounts of cognitive theory, but as a central aspect of how human beings make sense and meaning of lived experience and existence. They argue that these are an essential feature of being-in-the-world. This is consistent with the original concept of cognitions in cognitive psychology as understanding meaning and meaning-making, prior to the information processing approach (Smith & Osborn, 2008; Smith, 2004).

Heavily influenced by hermeneutics, the theory of interpretation, analysis in IPA is a constant process of making sense of the spoken and written word. I believe it is through the intensive engagement of me as the researcher that the rich dialogue from the participants' accounts will shine forth. Analysis will be an iterative process that will take place on several levels. It is concerned with moving between parts such as words or texts, and the sum of those parts, the whole, such as the sentence where words are embedded, or the research project from where the texts are derived (Smith, Flowers & Larkin, 2010). The relationship between the researcher's interpretative meanings derived from the analysis of the data and its iterative process could be described as a 'hermeneutic circle' (Eatough & Smith, 2008). The hermeneutic circle facilitates a free-flowing channel where new potential understandings of the data can unfold over time.

The researcher's position as a human being with their own experience of the world, endeavouring to interpret, make sense of and reflect the true value of

the participant's experience, has been referred to as a 'double hermeneutic' (Smith & Osborn, 2003). This demonstrates the duality and iterative qualities of the process where 'the researcher is making sense of the participant, who is making sense of x' (Smith, Flowers & Larkin, 2010, p. 35).

Compared to other phenomenological research methods, IPA is non-prescriptive, auditable and adaptable according to the aims of the research (Smith et al, 2010). It is a user-friendly and intricately subtle methodology, but researchers need awareness, detailed knowledge and appreciation of the philosophical underpinnings to fully capture the subtleties (Smith et al, 2010). IPA emphasises interpretation and hermeneutic enquiry, and seeks to encapsulate convergence and divergence in the data analysis (Smith et al, 2010). It is important to be mindful that IPA is a complex and time-consuming process, not merely collecting, classifying, explaining and understanding data.

To explain further, IPA typically follows a traceable link of a participant's embodied experience, the verbal experience, how they make sense of that experience and how they react emotionally to it (Smith, Jarman & Osbourne, 1999). For example, in the current research my interest would be to interview the couples and aim to answer the question 'How have the couples made sense of decision-making with a female diagnosis of Marfan syndrome?' In the analysis of the narratives I would aim to understand the personal meaning and thereby make sense of their experience of decision-making in that particular context, while other phenomenological approaches would focus on the question 'What were the main factors that influenced the couple's decision-

making process?' Analysis of the narratives would be focused primarily on commonalities of decision-making. I considered this as a rather loose, questions and answers distinction between IPA and other phenomenological methods. However, for the purposes of defining IPA as an appropriate methodology for the current research question, it creates a persuasive and salient argument for choosing it as 'the right tool for the job' (Smith et al, 2010).

Marfan syndrome is a subjective experience and this has important implications for understanding the different aspects of the psychological world of a man and a woman in their decision to have a baby. Therefore, I would emphasise that if the meaning of Marfan syndrome to a man and a woman where there is an increased potential risk in pregnancy, childbirth and beyond is to be fully explored, then a qualitative phenomenological approach such as IPA is essential.

### **3.5 Design**

The qualitative phenomenological approach of interpretative phenomenological analysis (IPA) was adopted as analysis of the data required a methodology to describe the meanings of the participants' experiences and beyond. It is idiographic and therefore looked at the unique characteristics of the individuals within their relationship.

It was thought that not only was IPA well-suited to the aims of the research, but also it meshed into the context of the philosophy and practice of

counselling psychology, in particular from the human science perspective (Strawbridge & Woolfe, 2003).

IPA is a dynamic process, which constantly moves and changes as it attempts to enter and understand the participants' personal way in which they view the world. Interpretation and analysis of the participants' accounts was a repetitive process and the cycle continued, through to and during the writing of the report, as new insights were likely to emerge with each stage of the analysis. The analysis is both complicated by, and dependent upon, the researcher's own interpretations that are required to access and understand the participant's personal world (Smith, 1996). The analysis is not a standardised process, it is creative, and ultimately IPA attempts to achieve a phenomenological 'insiders perspective' (Conrad, 1987) of a participant's account of a particular situation.

Each of the couples was interviewed as a couple to emphasise the coupledness. I am aware that the use of IPA to analyse transcripts from couples, as opposed to individuals, may be challenging the purist perspective of some researchers in the field. Smith (2010) argues that IPA is idiographic, but a couple has a shared identity and this idiographic approach could apply as much to couples as individuals. Similarly, Tomkins & Eatough (2010) argue that we can do justice to research using IPA and focus groups if we can take a step back from traditional oppositions while respecting their classic foundations. Therefore, in order to tap into the essential experience of making a decision to have a baby for a couple, it was necessary to capture the

inner experience of the individuals and also look at the shared experience. There can be no single truth. Mead (1934, p. 164) highlights the importance of a relational self, 'Selves can only exist in definite relations to other selves.' Therefore, the inner experience of the participants was considered as mutually interdependent with their relational experience.

### **3.6 Participant recruitment and selection**

Recruiting participants proved to be quite a challenge. It was originally proposed to interview six couples going through the decision-making process to have a baby when a woman has been diagnosed with Marfan syndrome. It was thought that this would provide a more 'live' or real-time reflection of the decision-making process. The participants would literally 'talk aloud' as they described an on-going process (Aanstoos, 1985).

However, despite advertisements posted in Marfan support groups, only one couple contacted me and volunteered to be interviewed. Following an exhaustive search throughout the year for further couples, it was decided to broaden the search to couples with a female diagnosis of Marfan syndrome who had already made the decision to have children. Five couples came forward and volunteered to be interviewed. Each of the couples had a child or children under the age of twelve years. The age range of the children was purely a function of the couples that came forward for interview. The criterion of couples having received genetic counselling was originally included as I thought it would maximise the homogeneity of the sample given that Marfan syndrome is a genetic condition. However, this was later dropped as the

research was concerned with the experience of decision-making, and genetic counselling was not essentially a part of that experience. Therefore, it was deemed unnecessary for the purposes of the research.

Obviously, interviews with the five couples would provide retrospective data and the possibility of different accounts, given the passage of time, were issues that needed consideration. But, notwithstanding these limitations, the narratives would still be subjective and for the purposes of the research would reflect how the couples remembered the experience. The retrospective data would still be psychologically revealing, but as a researcher I was mindful that it would also be their memory of the situation. Retrospective descriptions are not necessarily problematic, but they should certainly make the researcher cautious and aware of the above issues (Giorgi & Giorgi, 2009). Besides, phenomenological research is most often based on peoples' past experiences and their retrospective descriptions can provide trustworthy data (Giorgi & Giorgi, 2009; Flowers, 2008).

The data from the first couple going through the process of decision-making served to check the trustworthiness for the analysis of the data from the five other couples and were included in the final analysis. It was thought that this was an important and psychologically interesting feature of the research. It not only explored the process as it was lived through for one couple, but also provided an opportunity to compare that with the retrospectively reconstructed accounts of the other five couples. One clear advantage of this method of checking the trustworthiness is that it has the potential to reveal similarities

and differences in constructive processes over time (Smith, 2003). I acknowledged the difference of obtaining the retrospective and current accounts and was particularly vigilant in seeking convergence and divergence between the data.

The participants were recruited from the United Kingdom and Northern Ireland. They were contacted through the Marfan Association UK, which has over 1000 members, and through the Marfan support network group. A poster advertising the research and requesting participants (Appendix A) was distributed via the website and advertisements in their bi-annual magazine 'Marfan in-touch'.

The participants were approached by the author and were asked to read and sign a consent form, and to complete a questionnaire with general personal information.

### **3.7 Participants**

Purposive sampling was used to recruit six couples: Belle and Adrian, Clare and James, Jack and Joan, Theresa and Tom, Carl and Cathy, Lorraine and Bob. All the women had a diagnosis of Marfan syndrome. The names have been changed to protect their confidentiality. As previously mentioned, one couple, Belle and Adrian, were in the process of making the decision whether or not to have a baby and the five other couples had already made the decision and had children aged between two and eleven years of age. They had been together as a couple between seven and seventeen years. All the

participants were in paid work except one who described herself as a housewife. All the women were under fourteen years of age when they learned they had Marfan syndrome. The ages of the women ranged from 29 years to 37 years and the men from 29 to 49 years. They all had English as a first language. Four of the couples had one child and one couple had two children. Two of the couples had one child with the diagnosis of Marfan. The demographics of the participants are presented in Table 1 below:

Name	Age	Length of time as a couple	Female age of MFS diagnosis	Employment Status	Number and age of children without MFS	Number and age of children with MFS
Belle & Adrian	27 years 29 years	7 years	12 years	Employed Employed	0	0
Clare & James	32 years 30 years	12 years	5 years	Employed Employed	2 3 & 5 years	0
Joan & Jack	37 years 49 years	14 years	14 years	Housewife Employed	1 11 years	0
Theresa & Tom	30 years 33 years	9 years	2 years	Employed Employed	0	1 3 years
Cathy & Carl	36 years 39 years	17 years	3.5 years	Employed Employed	1 3 years	0
Lorraine & Bob	26 years 36 years	8 years	7 years	Employed Employed	1 1 year	1 7 years

**Table 1.** Demographics of participants

### 3.8 Data gathering

The data for this project was derived from an unstructured interview with six couples as participants. The individual couples were interviewed together throughout the ninety-minute interview. Four couples were interviewed face-to-face in their own homes and two couples were interviewed via Skype from my consulting room in Hertfordshire.

Each individual couple was interviewed together to explore the essence of the research question. It was considered important, as it would reflect the dynamics and interaction of the coupledom and the individuals within the relationship as they went through the experience of decision-making. The focus on both interpersonal and intrapsychic processes would endeavour to provide a complete and coherent account of what it was like for them to make that decision. However, the differences between an individual interview and a 'being in relation' interview had to be considered.

The quality of the relationship was a significant factor: would they only disclose what they wanted the other to hear; would a dominant partner override the other; how would they respond as a couple to me as a female; would they argue and shift the focus of the research question? But, the interviews were well balanced and provided intense data offering a valuable means of understanding the individual and relational experience. The joint interview essentially presented the psychological reality of the themes that arose from the decision-making, including personal, relational and social perspectives. This is commensurate with counselling psychologists' view of the relationship in context (Street, 2006).

The option of Skype interviews was a practical one regarding the distance of two of the couples. Technologies such as Skype have become an increasingly realistic and practical research interview medium providing an efficient and effectively interpersonal method of communication between researchers and their participants (Evans, Elford & Wiggins, 2008) and in

particular in IPA (Flowers, 2008). Moreover, the very nature of the visual aspects of Skype mean that the interview retains many of the features of face-to-face interviewing such as physical interaction, real-time synchronous interaction, empathetic and active listening and other subtleties (Flick, 2009). Hanna (2012) claims that these arguments overcome any potential criticisms of using Skype, such as being perceived as a barrier for interviewer and interviewee, feeling uncomfortable or self-conscious, technical issues, ethical and confidentiality considerations and researchers' and participants' familiarity with Skype, and argues that it is a viable research medium. I would argue that the Skype interviews in the current research were a flexible, efficient and practical alternative to face-to-face interviews, with good quality audio for recording and later transcription.

The Skype interviews followed the same procedure as the other interviews. There was an initial introduction where we chatted and built a good foundation for an effective working relationship. Couples were reminded that they had an opportunity to withdraw. With the couples' agreement the interview began. The Skype interviews went smoothly and were remarkably similar to the other interviews. The focus was on the interview and not the environment. They produced rich data and the depth of analysis was comparable with the other interviews.

For all the interviews a single open-ended question acted as a guide for the interview: 'Can you tell me what it's like for you in making a decision to have a baby?', leaving opportunities to follow up any interesting issues that emerged

throughout the interview. The interviews lasted approximately ninety minutes and were tape-recorded using a Sony WS-311M digital voice recorder. A back-up Phillips voice recorder was also used as a precaution. Verbatim transcripts were made from the tape recordings.

Two private secretaries transcribed the tape-recorded interviews and crosschecked the transcripts for accuracy by verifying them with the taped interviews. Both signed a confidentiality agreement. The verbatim transcripts of the interviews served as the data.

### **3.9 Data analysis**

The transcripts were analysed by the author using interpretative phenomenological analysis (IPA) (Smith, 1996). A detailed account of the method is necessary to enable the reader to fully appreciate and comprehend the meaning of the research analysis. The analytic process used five main stages as follows:

First, the tape-recorded interviews were listened to and the interview transcripts were read several times to achieve a visual familiarity with the text, and to allow the author to make sense of each of the participant's accounts, using her experience of the interview. During the readings, anything of interest or significance in the text was noted in the left hand column. These first initial notes are sometimes associations or connections and at other times they are an attempt at interpretations.

Second, the text was reread and any emergent themes were identified and noted in the right hand column. The emergent themes were then organised into a **preliminary list of emerging themes** (Appendix B) with page and line references to where they appeared in the transcript.

Third, the emergent themes were arranged in **clusters** (Appendix C) of connected material and the content became more detailed and psychological. This stage involved looking for connections between the themes, searching for similarities and differences in what the participants were saying. This was done partly on the computer using 'cut and paste' and then using a large white tablecloth to further arrange themes by cutting and gluing the colour coded text themes on to white A3 cards. Some of the themes naturally clustered together, and during the process new superordinate themes emerged which helped to pull together many of the emerging themes on the preliminary list. It was necessary to continually keep checking back to the script to make sure the connections and my interpretations actually reflected what the participants were saying. This became clearer when the clusters were arranged into **gender clusters** (Appendix D)

Fourth, a master table of **superordinate themes** (Appendix E) or general categories designed to capture the meaning and essence of the participants' experiences was produced; repeatedly comparing and contrasting the clusters of themes from all the participants achieved this. The themes were selected on the basis of prevalence, together with the extent to which they illuminated the richness of the account. Some of the themes may have been selected

because certain extracts powerfully illustrated the common themes. Other themes may have been selected because they helped to clarify different aspects of the participants' accounts.

Finally, the verbatim extracts for each theme were highlighted and read and reread in the context in which they occurred. Key extracts were selected to support the **master list of themes** (Appendix F), which was then translated into a narrative account using the verbatim extracts from the transcripts. Analysis continued throughout the process to allow richer interpretations to emerge.

It was important to return to the audio recordings from time to time throughout the analysis in order to remain as close as possible with the original medium of spoken word. This enabled me to draw out further information that may have remained hidden in the text.

Another important aspect of the analysis worthy of mention was my 'antenna' for identifying as many as possible potentially powerful and illuminating extracts of text from the narratives. As I listened to or read particular passages of the participants' narratives, I was intrigued and often alerted to various questions such as: how am I feeling right now; what does this *really* mean; how can I make sense of this; what other issues or influences are involved; where does this particular narrative fit in with what they are telling me? This experience of engaging with the narratives allowed special 'gems' to

transparently 'shine through', or to be 'seductively suggestive' in their meaning or to be 'secretive' chinks of light in the darkness (Smith, 2011b).

Finally, two procedures were employed following the analysis in order to enhance trustworthiness. First, a second reader, a psychologist familiar with IPA, was asked to look over and check the quotes and themes in the emerging findings. Second, a summary of the emerging findings (Appendix G) was sent to the participants to ask if they were commensurate with their thinking.

### **3.10 Ethical issues and confidentiality**

A protocol for the study and a completed research ethics approval form were submitted to Metanoia Institute. This outlined the proposed procedures and precautions to be used in order to overcome any potential risk or harm to the participants, myself or my family and to ensure that ethical questions were properly addressed and considered. A letter of approval from Metanoia Institute was received giving permission to carry out the research (Appendix H).

As a registered counselling psychologist with the Health and Care Professions Council (HCPC) and chartered member of the British Psychological Society (BPS), I confirm that I work in accordance with the organisation's Code of Ethics and Practice (2009). In addition, I read, gave due consideration and used as guidance all the appropriate codes that I abide by outlined in the British Psychological Society's Code of Human Research Ethics (2011). The ethical implications in this research project have been considered at all times.

The study of Marfan syndrome is a sensitive subject and therefore I was judiciously mindful of the ethical issues outlined by McLeod (1994, p. 174).

*Informed consent* – participants should understand the purpose and nature of the study and what participation in the exercise requires.

*No research should cause harm* - a judgement needs to be made as to whether a particular procedure is likely to affect the well-being of participants. Procedures must be justified and alternative approaches involving less risk explored.

*Confidentiality and anonymity should be maintained* – cautions should be taken to protect confidentiality of participants and data.

Following the interviews, couples were reminded that they were able to contact me with any concerns they might have. They were reminded of the independent source of therapeutic support offered in the project information sheet, should it be required.

The sensitive nature of this research could have been upsetting for the participants and therefore every possible care was taken to ensure they were fully informed of the objectives of the research. I was aware that painful issues could emerge for the participants as a result of the interview, and this was considered from the outset of this project. The interview may have created tensions or differences between the couple and stirred up disharmony,

emotions or regrets regarding their decision-making. Therefore the procedures, outlined in the methods section, were strictly adhered to.

I felt it was important to take a while before the interview to establish a good rapport with the couples from the outset. Prior to the tape recorder being switched on I checked whether they still felt comfortable about giving the interview. I also reminded participants, making it clear that they did not have to answer any questions with which they did not feel comfortable and it was fine if they wanted to stop at any time.

They had received written details outlining the purpose of the study. They all signed a consent form giving permission to use the anonymised material from the interviews in the final report. All signed material was kept in a locked filing cabinet in my consulting room at home. They were informed that they were free to withdraw from the research at any time without giving any reason.

Before the interview date, participants were contacted with a follow-up telephone call confirming that the arrangements were convenient, and to give them an opportunity to withdraw. I remained with the participants following the interview and they were given the opportunity to talk about how they experienced the interview and to discuss any issues. I ensured that each person had my contact details and emphasised that should they have any concerns they would like to discuss with me they could contact me in confidence. Finally, I checked whether they still felt comfortable with me using the data for research purposes.

As previously mentioned, participants were informed that in the event of any distress they could discuss this with me in confidence. They were also offered contact details for British Psychological Society (BPS), United Kingdom Council for Psychotherapy (UKCP) and British Association for Counselling and Psychotherapy (BACP).

The interviews were recorded and the tape recordings were stored in a locked filing cabinet in my consulting room at home. The confidentiality of the participants was respected at every stage of the research, and every attempt was made to keep their identity confidential. Any identifying information such as names, places and occupations were removed from the transcripts and do not appear in any material used in the final report. I gave full and due consideration to confidentiality and in accordance with the Data Protection Act (1998) all pertinent data relating to the participants was treated accordingly. In the final presentation, only material relevant to the study has been used, anonymity has been preserved, and at the end of the study the recordings will be destroyed.

I acknowledged that issues might have arisen that could have caused distress to my family or myself. My family was aware and supportive of my research into Marfan syndrome, and this was agreed from the outset. Throughout the research and beyond, their thoughts and feelings were discussed and monitored via meetings and personal communications. They were offered support and the opportunity to discuss any issues with me at any time. I had the support of my clinical supervisor and from time to time it was helpful to

discuss any concerns. Mindful of a duty of care for this research (Levinas, 1985), my personal safety and the safety of others were considered at all times.

Following the interview the participants described the interview as therapeutic. I was aware that it was important to manage the boundaries of my position as a researcher during the interviews. The traditional positivist format of research assumes a tradition of neutrality in the context of the interview as a method of data collection. However, in line with my epistemological stance I considered the interviews to be therapeutic with a small 't'. I believe that by the very nature of my profession as a counselling psychologist, the interview had to be in some way therapeutic, as it had the potential to stir up the bottom of the pond or open up a 'Pandora's box'. Essentially, the primary function of the interview was for data gathering, but philosophically and ethically I maintained the necessary and sufficient conditions (Rogers, 1961). I was aware of the risks concerning boundaries and navigated the interviews accordingly.

### **3.11 Trustworthiness**

Since its conception in 1990's IPA has constantly evolved and, in line with trustworthiness in qualitative research in general, it has been surrounded by much debate regarding trustworthiness (Yardley, 2008). Using the four core principle guidelines for evaluating trustworthiness in IPA research suggested by Yardley (2008), I aim to demonstrate that the current study has the necessary commitment to the rigorous and reliable qualities of a worthy research project.

### *Sensitivity to context*

The rationale for the research question is outlined in the literature review, and this research aims to contribute a unique psychological perspective to the extant literature regarding reproductive decision-making for people with Marfan syndrome and other related genetic conditions.

The interviews all took place with the participants in familiar surroundings of their own homes. Participants' socio-cultural background was respected at all times and care was taken to ensure that the participants were able to talk openly and express themselves freely. As the researcher, I endeavoured to build an effective working relationship with the participants. The interview used a single open-ended question and there was considerable flexibility to promote the participants' willingness to share their experience with minimal intervention from me.

Sensitivity to context continued throughout the data collection, the process of analysis and in the writing-up of the thesis. It was envisaged that these considerations would aim to achieve a trustworthy research study.

### *Commitment and rigour*

The rationale for the choice of the purposive sample is clearly outlined. It was homogenous in nature and appropriate for the research project. Considerable care was taken with the data collection, and the extensive in-depth analysis of the data demonstrates good practice. My personal investment and

assiduousness have aimed to produce robust, reliable and trustworthy research.

### *Transparency and coherence*

This research aims to produce a coherent presentation of the design, analysis, findings and discussion. I hope that my diligence to provide clear, comprehensive and explicit details of the methodology will enable the reader to fully understand the research process and the reasons behind it.

To check the trustworthiness of this research a 'paper trail' of the analysis (Yin, 1989; Flick, 1998) was made transparent. A reader familiar with IPA was invited to view the paper trail and verify that the findings were coherent and credible. This procedure recognised the limitations of interpretative freedom (Smith, Flowers & Larkin, 2010). In addition, participants were sent a summary of the findings and invited to make comments as suggested by Silverman (1993). It is debatable whether this is an appropriate approach to trustworthiness in IPA as evaluation of the data is concerned with the researcher's analysis. However, in my opinion it is an ethical and worthwhile part of research process. The participant feedback is outlined at the end of the methodological reflexivity section.

Reflexivity was considered an essential part of enhancing transparency (Brocki & Wearden, 2006) and this has been demonstrated in the report. My epistemological standpoint was defined in the methodological reflexivity

section, as this too was deemed an important factor in assessing how my own experience may have influenced the research.

I was mindful of the implications of conducting research as an insider researcher. There were advantages, such as my knowledge of Marfan syndrome and its language. But, there were also disadvantages, such as my knowledge unwittingly causing assumptions, misinterpretations, distortions and biases (Rooney, 2005). The potential problems associated with insider research, and the experience of researching as an insider were explored and considered throughout the research. My status as a researcher on the inside and how that could potentially compromise the trustworthiness of the research are discussed later in the report.

#### *Impact and importance*

It is believed that the quality of this research was enhanced by the application of myself as a practitioner researcher. As a counselling psychologist I had a desire to discover more about the research question. My personal involvement in Marfan increased my passion of wanting to know. It is envisaged that a deeper and more meaningful understanding of these couples' experience of decision-making will facilitate a more effective clinical practice within a multidisciplinary team. Furthermore, it is thought that by applying a reflexive approach, commensurate with my professional practice, a trustworthy piece of qualitative research has been produced for counselling psychologists and other professionals.

### **3.12 Establishing trustworthiness as an insider-researcher**

In this section I consider the role of being an insider-researcher and the complications that could have arisen regarding objectivity, subjectivity and potential threats to trustworthiness. Insider-researcher is defined by being in some way personally connected or involved with the research topic (Robson, 2002) and contrasts with the researcher as an outsider, objectively studying participants from an external perspective (Denzin & Lincoln, 2000).

As I planned the research I became increasingly aware of my identity as a counselling psychologist. I had chosen a topic where I would be an insider-researcher and being personally affected with Marfan syndrome in my family was a significant factor that needed to be explored. This led me to pay special attention to the roles of myself as a counselling psychologist and an insider-researcher. I needed to explore my personal beliefs about Marfan syndrome and to consider how that could have affected the relationship with the participants and ultimately the process and trustworthiness of the research.

As I explored the concept of being an insider-researcher I was mindful that I was challenging the traditional positivist notion that a researcher should be an objective outsider searching for an objective reality (Denzin and Lincoln, 2000) or truth (Hammersley & Gomm, 2000). However, my beliefs are firmly grounded in qualitative research that is not 'contained within a set of standardised research procedures' and I consider that being an insider-researcher in this project, although potentially risky, was an advantage as it ultimately gave the research an 'edge' (McLeod, 2002, p. 10). Furthermore,

from a postmodernist perspective, it could be argued that my subjectivity was central to the research process and complete objective reality or truth were unobtainable goals (Rooney, 2005). But it was necessary to guard against any self-deception (Edwards, 2002) leading to 'hermeneutics of suspicion' (Salner, 1999, p. 2), whereby the researcher as an insider may distort the participants' truth to provide a more palatable analysis of the narratives.

The aim in this research was to learn more about the essence of the underlying processes of couples' decision-making to have a baby and to develop an understanding of their experiences. My goal as a researcher was to produce new knowledge that would make a difference to the world of Marfan syndrome. With this in mind, I needed to consider how I would successfully accomplish a trustworthy research project. Kvale (1995) rejects the general notion that there is only one truth or reality, but while I agree and acknowledge that pure objectivity is impossible I was concerned that insider-researcher bias could have threatened the trustworthiness of the research on various levels.

I acknowledged that my involvement with Marfan could have affected the research. I was aware that I would have to set aside, as much as is realistically possible, any preconceived ideas I may have held of my participants' experiences. But, for the interview, engaging in the process of exploring the participants' personal experiences was more important, as this naturally enhanced the process of bracketing during the analysis (Smith, Flowers & Larkin, 2010). Congruent with my philosophical beliefs in hermeneutics I assumed the stance that I, together with the participants, would

co-create a narrative that would represent the experience of the decision-making process and have the potential of informing other populations. But how would this translate into findings that would provide a satisfactory and trustworthy foundation for change within the world of Marfan syndrome and other similar genetically inherited conditions?

Although I had not considered the aspect of decision-making to have a baby before embarking on this research, I did have extensive knowledge of the medical aspects of Marfan syndrome and I had valuable experience of the complications and implications of the condition. But this knowledge could have been problematic without my due awareness and attention, as familiarity could potentially lead to loss of subtle nuances in the data collection and analysis (Edwards, 2002). I believe that insider knowledge and experience contributed to obtaining a richer data (Coghlan, 2003) than if I had been an outsider. As an insider-researcher there is the advantage of considering the participants' views and the specific aims of the research when choosing responses and interventions in the interviews (Reay, 1996, p. 62). Moreover, it is thought that my involvement with Marfan syndrome was crucial (Marshall & Rossman, 1989) to the overall aim of a successful research project.

Having concluded that my neutrality was not achievable or, indeed, desirable for IPA, I felt cautiously confident that participants would feel more able to talk to me freely in a language using medical terminology in an area with which I am familiar. Spradley (1979) suggests that insider-researchers have the advantage of understanding the distinctive language that external researchers

have to establish. However, I did have to consider whether this could have had a detrimental effect on the research process. I shared with participants the fact that Marfan syndrome affects my family. This was an important consideration and I ensured that it was mentioned in advertisements, all contact with participants, and in the information sheet. The interviews were carried out in the informal and familiar setting of the participants' homes either via Skype or by invitation to their homes. As a counselling psychologist there was an emphasis on the relationship in the interview, which I believe emanated trust, confidence and good rapport that enabled the participants to share their story. Being an insider-researcher helps to establish the relationship between participants and the researcher (Glesne, 1989). These were important factors that provided a safe environment for gathering reliable, honest and thereby rich data (Rooney, 2005).

Finally, understanding the role of the insider-researcher and how it may have affected the research processes has provided invaluable insights (Hammersley & Atkinson, 1995) for the study as a whole. Importantly, Zavella, 1996, p. 141) reminds us that as insider-researchers '...we should realise that we are simultaneously insiders and outsiders'. It is envisaged that this project will be an ongoing process for me, potentially having an enduring influence in the world of Marfan syndrome and impacting on my personal relationships (Rowan & Reason, 1981).

### **3.13 Methodological reflexivity**

Reflexivity is crucial within a qualitative project as it is impossible to achieve researcher partiality or detachment from the research topic and thereby an objective stance. It is essential in understanding the nature of human beings in psychological research (Smith, 2003). I have considered reflexivity throughout the project from the perspective of my own personal thoughts, emotions and sociocultural environment and considered the influences of my epistemological and ontological assumptions. My understanding of who I am and how that relates to the project creates a link between research and practice (Etherington, 2000; Heron, 1996; Reason, 1994), and facilitates an awareness of my contribution to the research and findings (Stiles, 1993). Moreover, it is considered a key concept in qualitative research, such as IPA, involving hermeneutics and symbolic interactionism (Bannister, Burman, Parker, Taylor & Tindall, 1994).

Reflexivity in the current project explored the researcher and the researched, and reflected on the interviews where the narratives and meanings were constructed and essentially co-created. The encounter was inevitably multidimensional, individual, shared, unconscious, conscious, and I, as the researcher, was both subject and object (Mead, 1934). Further, it looked at the process of analysis, my understanding of the couples' experiences of decision-making and some of the ethical dilemmas encountered during the research.

My epistemological standpoint rejects Cartesian dualism and embraces Heideggerian existential thinking that we are essentially beings-in-the-world. As such we are inextricably intertwined with other people and the world around us (Eatough & Smith, 2008). Inevitably we are always involved in the world in some context or other, through which we develop a sense of self, others and meaning-making. My stance is heavily influenced by phenomenology; as a psychologist I am also a human being studying other human beings, and the consideration of my subjectivity is an on-going challenge. In line with IPA I am committed to 'the exploration of personal lived experience' (Smith, 2004, p. 41).

Turning to the interviews I have to admit that, although I was excited at the prospect of meeting with the participants and gathering data for the project, I also felt quite anxious. I had to allow a while to focus, relax and locate myself prior to each of the interviews. During the interviews it was quite evident that the couples were trying to make sense of the decision-making process, perhaps for the first time as they recounted what it was like for them. They often revealed new information to each other and I realised that my presence may have had an effect on how safe they felt to disclose vulnerable thoughts and emotions. As a researcher I had entered into their world for the purpose of generating new knowledge, but the interview had a therapeutic quality whereby their private world as an individual was shared within the relationship.

Following the interviews I found it necessary to 'come down' from them and the practice of being 'creatively indifferent' was invaluable in order to take care

of myself. As I reflected upon the dynamics of the interview and questioned my role in this shared experience, I believe that my training as a counselling psychologist provided me with the skills and theory for the research interview. But most importantly, my being and my curiosity to know more, fostered a fertile environment for change not only for the participants, but also myself (Etherington, 2001a). Interestingly, I was aware of an increased need to spend more time on my own throughout the interview process and welcomed the opportunity for long walks and swimming.

The therapeutic nature of the interview led me to consider what other influences I may have had on the couples in the interview. I wondered what the possible impact of my gender as a female may have been. The couples were very open, honest and eager to share their stories with me. The men exposed their vulnerabilities and engaged on an emotional level in the interviews. I suspect that if I had been a male researcher they may have been more reluctant to show this side. I had a distinct feeling in the interviews that the women assumed that I had experienced pregnancy and childbirth and I wondered if the women would have been so passionate in their accounts regarding their longing for a child if I had been a male. I am at least twenty years older than the participants, but I have been told that my appearance is younger than my years. However, in my opinion I look quite “motherly”, and it’s difficult to say how these factors may have influenced the interview. Overall, it is very difficult to speculate on the intersubjective dynamics at play during the interviews or to consider how the encounters may have changed if my appearance or gender had been different. But suffice to say that gender of

interviewers and interviewees has been found to influence research interviews because '...the interview takes place within the cultural boundaries of a paternalistic social system in which masculine identities are differentiated from feminine ones' (Fontana & Frey, 2000, p. 658).

My sense was that meeting with couples as opposed to individuals offered the distinctive advantage of maximising the participants' dialogue. Interviews with the couples generated a jointly constructed picture of decision-making. As a researcher I have always aimed to make minimal interventions in the interviews and would describe my role as a passive prompt, but also as an active listener (Smith, Flowers & Larkin, 2010). It is acknowledged that the research process is a two-way street between the researcher and the researched (Alvesson & Skoldberg, 2010), but I would argue that my interviews with the couples were a four-way process: researcher and male; researcher and female; the coupledom; the researcher and coupledom. Through my work with couples as a counselling psychologist, I could not help but be aware of these complex dynamics throughout the interviews and beyond.

I suppose I had tacit expectations of my meetings with the couples through my own experience with Marfan syndrome. Despite my best efforts to set any prior assumptions aside, I now believe that I had assumed that maybe the women's awareness of their diagnosis of Marfan syndrome would have somehow affected their decision-making in a negative manner, and I am not sure how this may have silently impacted on the data collection. This was the only assumption that came to light in the aftermath of the interviews, and it

was only through my subsequent understanding of the couple's experience that it emerged (Smith, 2007). However, I remember in the interviews feeling quite in awe at the sense of power, strength and resilience that the women portrayed.

Turning to the analysis of the interviews, I found that this was more demanding and time-consuming than my previous experiences with IPA. At times I doubted whether an analysis of the vast amount of data was actually achievable. Various thoughts pervaded my thinking, but mainly I seriously considered that the data from six couples was overwhelming. I questioned the extent of the data and wondered whether a single case study of the couple actively going through the decision, and a cross-comparison of a single case study of a couple's reflective accounts would have produced the same findings. Nevertheless, each interview produced rich data and I did not want to lose any of the narratives that the couples had provided. Their importance to the research presented valuable material and I was determined that it would not be lost by my lack of commitment or enthusiasm for the daunting task ahead.

Immersing myself in the research, I began to understand the intricate details of the couples' decision-making. I reflected back on the title of the research project, 'Who's Left Holding the Baby? ...', and found it intriguing but quite uncomfortable as I realised that this broad conceptualisation was true, mirroring the couples' accounts and my understanding and interpretation of them. Time and time again I moved between the couples' accounts and my interpretations. Many months passed and it proved to be a worthwhile

challenge as themes emerged. I felt quite isolated as I explored their worlds and needed reflective activities such as walking and swimming to sustain my physical and mental energy.

One theory that relates to my experience of IPA analysis is that of 'flow' (Csikszentmihalyi, 1990). I needed to withdraw from the world to focus on mastering the challenging and changing demands of tackling the participants' narratives. It is suggested that maybe the huge process of analysis motivated me to such an extent, that I entered the zone of flow. In the state of flow, I could concentrate and perform the task at hand at an optimal level, whereby thoughts of the self, others, success or failure were effectively contained. The function of flow is to facilitate challenging tasks, using the minimum of mental energy and thereby a less active brain. Therefore the focus becomes external and reduces the internal existential anxiety. I would argue that flow is an important application of theory in the passionate desire to achieve clarity from the chaos of such complex data.

One further consideration was that of sharing the findings with the participants. I sent a summary of the findings to them inviting comments or feedback. With their written permission these appear in the methodological reflexivity section. However, given the sensitivity of the research topic, I was confronted with wondering how they would receive the interpretations. The summary of the findings was not designed to appear more palatable to the couples, it represented the true and honest analysis. Was I being ethical in releasing my interpretations of the interviews in such a manner? Having pondered this

dilemma for a while, I concluded that it would be unethical to withhold or dilute any of the interpretations. I aimed to be as transparent as possible throughout the research, and this would be disrespectful of the open and honest interviews they had shared with me. Moreover it would not give them the full opportunity to comment on the findings. Sharing the research analysis with participants is important as it gives them the chance to share their reactions with the researcher (Etherington, 2004). However, Josselson (1996) warns that researchers must remain aware of the powerful impact of participants viewing their own words in print. With this in mind, I welcomed receiving all the feedback from the couples discussed below.

Following the analysis, a summary of the emergent findings was sent to the participants for their comments. Below are extracts from the feedback:

Theresa and Tom: *'The only comment I would have personally for us having had two children the genetic risk in having a third would have a far greater impact as part of the decision to have any more children'*

Clare and James: *'Thank you for the report I found it very interesting and also nice to learn others felt the same way as me'*

Cathy and Carl: *'Thank you for giving us the opportunity to give our point of view...'*

Jack and Joan: *'It's a difficult time...it was good to talk about it'*

Belle and Adrian: *'Thank you for the report if you need any further information please call us'*

Theresa and Tom make an interesting point in highlighting the perceived increase in risk after the birth of a second child and I wondered whether over time this might be the case for other couples. Overall the feedback suggests the couples' willingness to share their story. But, I was mindful that given the nature of the research, there might have been more behind these comments. I also wondered what the impact might have been on the couples talking about their experiences of decision-making. For example, had the couples talking retrospectively had any negative thoughts about their decision or had the interview highlighted the significance of their situation? They had talked quite openly, so perhaps the interview had led to further discussions between them.

I was certainly enthusiastic about the research when we met for the interview and there may have been an element of wanting to please me with favourable comments in the feedback. I had considered this in the debriefing with the participants following the interviews and told them that I would be inviting them to comment on a summary of the findings. I emphasised that I would welcome both negative and positive comments on the feedback. Similarly, I suggested that in the meantime they were welcome to contact me with any thoughts they may have.

As mentioned, the participants had been invited to call me if they had any concerns and they had been offered the contact details for counselling support outlined in the ethics section. Following the feedback I took the opportunity of Belle and Adrian's offer to call them, as their distress at times during the interview was quite evident. I spoke to Adrian and asked him how they had been since the interview. Belle had had the operation she was waiting for at the time of the interview and they were keen to conceive a baby as soon as possible. Although we were on the telephone for about 20 minutes, I gleaned no other information except being thanked for doing the research and how helpful it had been to talk about everything. I invited him to contact me if he had any concerns, but I was still left wondering about the impact of the research and what other thoughts or feelings they might have.

In the absence of further research interviews with the participants, I remain uncertain as to what impact the research interviews had on the couples. It is possible that having opened the 'Pandora's box' in the interview, they decided to place it back in the cupboard, and in the process of locking it away the feedback they offered to me was in their best interest.

## 4. FINDINGS

This research focused on couples' experiences of decision-making to have a baby when a woman has the genetically inherited condition of Marfan syndrome. Analysis of the data, verbatim in-depth interview transcripts, identified four main superordinate themes, each of which is supported by the master themes as shown in Table 2 below.

<p><b>A. Her decision is already made</b></p> <ul style="list-style-type: none"> <li>A predetermined decision</li> <li>Female drive and identity</li> <li>Urgency of timing and age</li> <li>MFS as strength and resilience</li> <li>Comparison with past self</li> </ul>	<p><b>B. Creating an informed decision</b></p> <ul style="list-style-type: none"> <li>She wants him to know the facts</li> <li>Sharing responsibility for decision</li> <li>Coping with doctors</li> <li>The meaning of genetic counselling</li> <li>The role of the Internet</li> <li>Exploring options</li> <li>Assessing the risks</li> <li>Optimism of medical advancements</li> </ul>
<p><b>C. Finding psychosocial support</b></p> <ul style="list-style-type: none"> <li>The effects of her mother's experience</li> <li>The strength of the relationship</li> <li>Temporary parallel lives</li> <li>Limitations of friends' support</li> <li>The role of support groups</li> <li>Psychological counselling as potentially helpful</li> <li>The interview as a therapeutic experience</li> </ul>	<p><b>D. Existing with the fallout of the decision</b></p> <ul style="list-style-type: none"> <li>Coping with high-risk pregnancy</li> <li>Her concerns for child</li> <li>He fears losing her</li> <li>He copes and cries alone</li> <li>Feeling responsible, selfish, guilty</li> <li>Acceptance of gamble, uncertainty, fate</li> </ul>

**Table 2.** Four superordinate themes supported by master themes

Data extracts were used from the participants' interviews to support and illustrate the superordinate and master themes, and to help the reader capture the essence of the participants' experiences. Also, most importantly, direct quotations '...retain the voice of the participant's personal experience' (Shinebourne & Smith, 2009, p. 155). Each section begins with an overview of the superordinate theme, and then the master themes are described in detail with supporting quotes from the participants.

#### **4.1 A. HER DECISION IS ALREADY MADE**

The first superordinate theme, 'her decision is already made' shows how five of the female participants have given some considerable thought at an early age to the prospect of having children later in life. There was some considerable focus during the interviews on the notion that the women believe so strongly that they must have a baby, there is not really a decision to be made for them. From the women's narratives, it appears that their decisions were largely based on what they considered to be a physically driven phenomenon to have their own baby and fulfil their personal role as a woman. Over time their age and state of health was 'a huge factor' (Lorraine). Their personal experience of adjusting and adapting to Marfan syndrome seemed to give them strength and resilience to cope with the future. In making their decision they made comparisons with themselves as a child. These were physical and psychological comparisons and provided a measure for them when evaluating the risks for their own child.

### **A predetermined decision**

Five out of the six women described how they had given some considerable thought to having children later in life. The following extracts capture the determination with which the women achieved their ultimate goal of having a child through natural childbirth.

Belle mentioned it several times during the interview, and her determination to have children shone through as she described her thought processes during her early years. She acknowledged that she does have some worries, but they would not stand in her way of having a baby. She liberally used the words “I’ve always been set” and in this particular extract she said it twice to stress her point:

*Belle: I’ve always been very set, I’ve never wavered on having children, (turning to her husband) have I? I’ve always been set on having children and, although there are things I do worry about, nothing could really affect my decision.*

Similarly, turning to Clare, her decision was already made at an early age too.

*Clare: I knew from an early age that I always wanted to be a mum by hook or crook. I didn’t mind how we got there that’s what I was saying to you, if we can’t by any means would you consider adoption, which you then said you would.*

Belle and Clare used the interview to emphasise that the decision to have a baby was a 'fait accompli' to their partners. By using the phrases 'I've always been set' and 'by hook or crook' they reaffirmed that their decision was set in stone and they would stop at nothing to succeed in the quest to have a child.

Joan echoed the knowing from an early age, but added that she had never considered that it might not be possible physically for her to have a child. Her own sense of risk was diminished as she considered her physical condition as irrelevant:

*Joan: I knew I always wanted to have a child even from a very young age, even from ten/eleven I was baby-sitting and minding children and babies up through my teenage years. I think I was quite maternal. I never thought that physically I wouldn't be able to have a child.*

In contrast with four of the women, Lorraine talked of the decision of not wanting children when she was younger. This was an interesting turn of events in the interview with the final couple:

*Lorraine: After I left school I started going (to the clinic) on my own and they started speaking to me a bit more – 'you shouldn't be smoking, you shouldn't do this and then it was have you looked at having babies?' I was like 'no, I don't really want kids!' (laughing)*

However, she made her decision to have a baby when she met her partner, but never discussed it with him. She told me 'I spoke to the hospital about things like that (getting pregnant), they spoke to me, but between us it never really come into it'.

### **Female drive and identity**

Although this theme does not apply to females universally, many women will identify with the longing that the participants portrayed in their accounts of the passion with which they fought their way through the minefield of potential threats to their goal of having a child.

Belle described how she feels that the desire to have a child is out of her control, a seemingly physically driven phenomenon; she talked of 'the broody nerve', which suggested an anxious edginess in her thought processes:

*Belle: ...carrying, being pregnant, it's just a very female desire isn't it? A bit broody (laughs), the broody nerve has taken over somewhere.*

Theresa talked of her initial drive to have a child as being 'high' as she remembered making the decision to have their first child. Although she was aware of the risks of Marfan syndrome she regarded it as a manageable condition. She then explained that when the powerful female drive or desire was fulfilled by having a child the perception of the risks associated with Marfan syndrome in considering further children was increased:

*Theresa: The biggest factor is, it's almost like you say your desire to have kids in the beginning is high so you, you don't downplay the risks of Marfan, but you think OK we can deal with that. So as when you have kids your desire to have kids has obviously decreased because you've got two already, so the risk of Marfan becomes more important almost. Does that explain it any better?*

Her perception of risk-status intensified as her desire to conceive diminished. Compared to her previous understanding of the risks involved, her interpretation demonstrated contradictory thought processes compared to her previous understanding of risk when making a decision with their first child.

For Cathy, the value of herself as a woman would have been diminished considerably if she had been unable to produce the child she had decided she wanted. She made a short but powerful statement leaving her audience in no doubt as to how she felt:

*Cathy: I can't have a child, so I feel pretty useless.*

It appeared that she would have felt inadequate as a woman and that having a child placed a practical value on her and made her effective as a female. Not becoming pregnant and giving birth to her child may have meant a life unfulfilled. Her sense of self and her femininity threatened, she may have perceived herself as 'useless', an unused commodity and not as nature intended.

### **Urgency of timing and age**

Having made their decision, the women wanted to become pregnant sooner rather than later. The decision was concrete for these women who had lived through the experience of coping with Marfan syndrome and they resented having to wait. There was a striking difference between the females' determination and, as we shall see later, their partners' caution. The women had made their decision and there was a sense of anxiety and frustration as they recounted their readiness to go ahead and cope with the practicalities of their situation.

Belle was keen to get pregnant as soon as possible. She stressed that if they left the decision any longer the risks to her health in pregnancy and childbirth would increase. She used the term 'a bit of a leeway there', which emphasised the narrow margin of opportunity for her to become pregnant:

*Belle: I have got a stretching of the aorta and they are at the point at the moment where going into a pregnancy there is a bit of a leeway there. It's not been an immediate risk for me at this point in time, but looking at that fact I think it's important for us to go ahead now rather than waiting any longer because it is a progressive disorder.*

Similarly Cathy described the urgency of reaching a successful conclusion in her decision to have a baby. She considered age and the lengthy process of following the various options to have a baby. She used 'we' not 'I' to include her husband in the decision-making, but as we have seen, she would have

felt useless if she couldn't have a baby. Her mind was made up, so it seemed that the process of exploring the options was to satisfy her husband. The tone of her voice and the speed with which she poured out her words portrayed her memory of that urgency. I listened to this passage several times and it struck me how Cathy sounded somewhat like the momentum of a train as it pulls out of a station! There was an emphasis on certain words (highlighted) illustrating that her need was to eliminate the other options as quickly as possible so that she could proceed with a natural pregnancy:

*Cathy: We were in our early thirties so, we were saying like adoption it could take **ten** years or **eight** years and then if we **did** go ahead and have a child ourselves we knew also that we would be waiting on every echo (echocardiogram) to make sure that my heart aorta hadn't increased, because at **that stage** that option would go out of the window, so you do have a bit of **pressure** because the adoption could take so long, as each measurement on the heart could change at any time and if we did look at that time frame change then...also the **financial** situation if we did go down the American route seeing if we could have the Marfan gene removed from an egg and so on, if that was an option. There was just that little bit of **extra push** because we knew we had a **timeframe** I suppose that we kind of had to **push on**.*

Clare too remembered that having made her decision there was a feeling that 'time's dragging on'. The following extract of an exchange between Clare and her husband James demonstrated that becoming pregnant was more Clare's

choice than James's! Her use of the words 'pulled' and 'pushed' highlighted her need to stress that it was a joint decision:

*Clare: ...so I think I pulled it to a head to say let's make a decision now. This is what I think, what do you think? So I probably pulled it to a head, but I wouldn't say I pushed you into it.*

James responded:

*James: No I wouldn't say you pushed, I think you made the decision and I sort of went along with it!*

### **MFS as strength and resilience**

The women emphasised how their decisions were partly based on the strength and resilience they had acquired over the years as they coped with MFS. This suggested that they had already thought through the issues when considering having a baby. This was possibly the quality that pulled them through the decision-making with their husbands.

Belle was trying to convince Adrian, who had expressed his extreme concerns for his wife's safety, that she felt not only able to cope with the risks to her in having a child, but also with anything that might have stood in the way of achieving her objective:

*Belle: You focus more on what it's going to do to me whereas I'm not bothered by that. I know I should be, maybe not bothered is the wrong word, I am bothered, I do worry about it, but not to the extent that it would stop me from wanting to have a child.*

Belle's contradiction about being 'bothered' emanated from her direct and personal experience of Marfan syndrome as a potentially life-threatening condition. She did not want to focus on the risks to her 'I'm not bothered', but reality and common sense prevailed 'I know I should be'. She then questioned her use of the word bothered, 'the wrong word' and finally accepted and admitted that she was 'bothered'.

James was talking about his wife Clare as being very 'single minded', and indeed Clare demonstrated how her strength and resilience emerged as she was growing up:

*Clare: I was always told when I was little that I wasn't allowed to join in and do stuff. I wasn't allowed to compete. So, I **have** got this, I have **got** to prove myself.*

As we have seen earlier, Joan made it quite clear that her mind had always been made up to have a child. She then explained where her strength and resilience had come from:

*Joan: If you are a child with Marfan you can be limited and if someone says to you 'you can't do this', you want to do it...as children, going from reasonably happy children to 'you're not doing that, you are not going to get pregnant, you're not going to be a nurse and have a physical job', nothing that you had planned all the way up until you were sixteen, nothing is going to happen for you. That is what makes me determined in adult life.*

Although her husband Jack made a comment about her being a rebel, Joan continued her theme to emphasise her strength and resilience as a woman faced with the challenges of Marfan by saying 'if I'm told I can't do it I will find a different way to do it', leaving her audience in no doubt as to how she had achieved her goal of having their son.

### **Comparison with past self**

The women drew on their own experience of being a child with Marfan as a benchmark to prove that in making their decision they had thought through the issues that may arise for their children.

Belle explained that she thought quite positively about her decision and the risk of having a child with Marfan and told us 'I have had a relatively well childhood'. Therefore, one was left with the strong sense that she felt her own child would have a similar experience.

Lorraine felt that her childhood experience was 'fine'. 'Fine' is a word she used freely throughout the interview when talking about herself and her

health. However, in comparing her experience with that of her daughter who has Marfan she told me:

*Lorraine: It has never affected me myself whereas now when we go back for our daughter's scan they are going to decide on what medication to put her on if it's (the aorta) grown again. If I had known that before, it would have made me doubly think about things.*

This suggested that when Lorraine made her decision it was partly based on her own experience of Marfan as a child and being 'fine'. In making reference to her own childhood when she made her decision she now realised that her beliefs about the outcome of having a child were erroneous. Retrospectively, she would have thought more about the decision as she struggled to come to terms with her daughter's diagnosis.

Clare had a realistic personal view of the implications of being a child with Marfan. Although she knew from an early age that she wanted children, having made that decision she checked out how prepared she would be if their children were to have Marfan. At the same time she was trying to objectify her thinking by cross-checking her husband's understanding of the implications of having a child with Marfan:

*Clare: I had to be sure that I knew I could cope with it if our children had Marfan, because I have been through it. I tried to explain what it would be*

*like, all the appointments, all the heartache all the stuff that comes with it really.*

#### **4.2 B. CREATING AN INFORMED DECISION**

In the second superordinate theme the participants described how they used different sources of information to help them in an attempt to create some certainty as they weighed up the pros and cons in considering childbirth. The women expressed the need to share the facts of Marfan syndrome with their partners from early on in their relationship and wanted them to understand the risks and implications of having a baby. Having made every effort to ensure that their partners knew the facts, they then encouraged them to share the decision-making, as they didn't want to take sole responsibility for the outcome. They sought advice from various doctors and, although there was some reliance on the doctor's 'all clear' to go ahead, there was evidence of a poverty of understanding of Marfan syndrome among the doctors and medical professionals in general. The meaning of genetic counselling was considered and four out of the five couples described their experience. The Internet was a source of information on Marfan syndrome, but the descriptions and implications of the condition also provided a cause for concern. Five out of the six couples described how they explored the different options of having a baby including adoption and IVF. However, this was more for the men's benefit than the women's. They generally assessed the 50/50 risk of having a child affected by Marfan and were comforted by their optimism that medical knowledge of Marfan was advancing at a steady and encouraging rate.

**She wants him to know the facts**

The women wanted their husbands to be aware of the implications of Marfan syndrome. They encouraged them to understand the meaning of coping with a potentially life-threatening condition and what that may have meant in pregnancy and childbirth.

Belle had said that she had educated herself to a point where she had a fairly good understanding of Marfan syndrome and what to expect in pregnancy and childbirth. Adrian told us that he wanted to have kids and although he had attended a doctor's appointment with Belle at her request, he didn't really get much out of it and Belle's attempt to make sure he knew the facts was rejected. He told us how he felt about making the decision:

*Adrian: And you've got to make your own mind up about that based on your own morals or standing, whatever. Not based on what the doctor told you, it's all got to be heart and head.*

Belle then made a further attempt to check that Adrian understood what Marfan is and the implications of it. She said to him 'you always read through the Marfan syndrome books and things.' It was a statement, but she said it as if it was a question and she was checking. Adrian responded:

*Adrian: Yeah you read through it and I know what I'm up against.*

In this short powerful statement Adrian moved from the objective 'you' to describe the action of passively reading the information, to the subjective 'I' to explain his perception of the challenge of what might lie ahead. However, he confirmed that he had understood the literature and it was as if Adrian was telling us that he was now preparing for battle and resigned to his fate.

Clare had gone to great lengths to ensure that James was fully aware of the implications of Marfan syndrome when they married, but she was concerned that James did not fully understand the facts before they had children 'he just dismisses everything'. However, James valued Clare's direct approach to her condition and contrary to Clare's perception that she was not being heard, his understanding and knowledge of Marfan was respectfully evident:

*James: I know my life is with Clare, she is very open and honest, I think that's the best thing she did from the outset, be very honest about her condition and everything else, so you go in with your eyes wide open, so there are no skeletons in the cupboard.*

Jack's opening line addressed to his wife in the interview was *'you were 100% honest with me, when we first met you told me you had a genetic illness. Admittedly I didn't understand a lot about the condition...'*

As her relationship with Tom became more serious Theresa wanted Tom to be more aware of the genetic implications of Marfan syndrome within a relationship:

*Theresa: I really wanted to make him aware that this was a genetic syndrome and the implications of that. I have a very mild case so if he looked at me and went 'well', and go 'well you're OK'...(pause) I tried to emphasise to him that it could be worse and there are a lot of people out there in a worse condition. Then I think he went and did a bit of research himself.*

As we have seen, Lorraine had already made her decision to have a baby, but she was concerned that Bob did not know the facts. She had tried to explain to him what she knew about Marfan, but it appears that his understanding was still limited:

*Lorraine: The other thing is that he doesn't really have a clue what it is because it's not been explained to him properly. I mean I can tell him what I know and he can read the notes, even when reading the notes it's all in terms you don't really understand.*

This was in contrast with the other couples. Lorraine was telling us that Bob did not know the facts and implied that her knowledge of Marfan was somewhat limited as she found it difficult to understand the language in the available information resources.

### **Sharing responsibility for decision**

The women's inclination to encourage their husbands to share in the responsibility for the decision was a significant consideration. The women either didn't want to take sole responsibility as they didn't want to take the

blame for any consequences, or conversely one couple felt that it was acceptable for the woman to take control of the situation and to keep her husband informed. The men's reluctance to share the responsibility for what was essentially the women's decision is evident.

Adrian was finding it difficult to make a decision and shared his dilemma by saying 'Damned if you do, damned if you don't kind of thing'. He was reluctant to share or take responsibility for Belle's decision to have a baby. He would rather have considered a less risky alternative and tells us:

*Adrian: I would straight away go for adoption or fostering without a second of doubt because we would make good parents. There are plenty of kids out there.*

He then acknowledged and agreed with Belle that they both wanted children. However his use of the word '**you**' demonstrated a reluctance to own his part in the decision-making process:

*Adrian: Yes, **you** want your name to carry forward, **you** want to have kids, **you** learn about yourselves when they are growing up.*

Clare talked about researching every option and aspect of having a baby. She wanted to 'explore every avenue' because she felt it was 'only fair' to 'cover all the aspects' for her husband. She was cautious that she may have

ended up taking the blame for the consequences of her decision to have a baby:

*Clare: It was just I didn't want him to turn around to me and say you created this whatever. I just didn't want him to blame me for anything. (Turning to face her husband) I just wanted to explore it all for you more than for me.*

Cathy was trying to include her husband, Carl, in her decision by using the word 'we'. For example 'we didn't realise it would be so risky to have a family'; 'we got married and realised how serious it was'; 'we didn't have heart complaints...' However, Carl was totally honest about how he felt about his reluctance in sharing the responsibility:

*Carl: I was totally selfish here and say it wasn't an issue to me I suppose. I know it's going to sound terrible and I know I'm going to sound like the devil himself sitting here, but at the end of the day it wasn't something I was going to have to deal with, so I suppose I was happy that I never sat down and consciously made a decision, we are going to have a child and damn the consequences you know. It was always in my mind that our child would be born with Marfan, well it wasn't me who gave the marfan so in that sense it was never something I was going to have to deal with. Obviously I will have to deal with my child if it was going to have a life-threatening disease.*

Lorraine felt that her approach 'as long as I know all about it' worked in their relationship. This was confirmed by Bob's response:

*Bob: I know it sounds a bit selfish, if there was anything wrong she would tell me.*

### **Coping with doctors**

The couples had positive and negative experiences in visiting doctors. However, they all recounted the various difficulties they were faced with from time to time as they sought medical advice and information to help them with making their decision.

Belle and Adrian told me about a visit to the doctor at the hospital for Belle to have a check-up and to discuss what to expect from a pregnancy, the risks, giving birth and how Belle could be afterwards:

*Belle: He explained that we would have a team right from the go and we would be cared for and I would be monitored a lot more closely and things like that. And yeah that was all we really covered. I mean we were there for a good hour or so. But yeah, it wasn't, we didn't come away feeling like, right it was enough to make the decision on purely what they had given us there. But I suppose it was only one session, so we have done a lot more research ourselves really and I don't know if we did ever want to go back we could, but we really don't see what the benefit of it really was.*

As Belle was speaking she was trying to make sense of the visit as if it was the first time she had considered what their needs were to help them make a decision.

Jack and Joan felt that they made their decision without considering the implications for giving birth when a woman has Marfan. She told me that she would have found it helpful to have more information about what to expect:

*Joan: I think it would definitely have been helpful if they could have explained a little more of you know, you cannot have a natural birth and then at the end that's not going to happen you are going to have an epidural, forceps delivery to reduce the pressure, I didn't really realise that still afterwards in my ignorance. I just thought they would give me a caesarean. I didn't have that information beforehand, but that was just my own ignorance.*

Joan was blaming herself for not understanding more about what to have expected. Her belief was based on her assumption that it was her responsibility to obtain the information about childbirth.

Theresa and Tom had had some frustrating experiences with doctors. They mentioned the 'lack of knowledge' of Marfan generally among the doctors. They told me that over the years they had learned to try and work differently with the doctors when the system wasn't working for them. Tom had learned that if you get frustrated with the doctors you 'get nowhere', so they had learned to 'pull back and come at it from a different angle'. He could see the lack of knowledge from both their perspective and the doctors' and he understood that the doctors 'don't come across Marfan that often, so they are not going to know about it'.

*Tom: It's just being basically unaware and lack of knowledge and I suppose for ourselves we have to acknowledge that fact and say look these guys are just not aware of this and when we go into see certain medical professionals it's like we are teaching them, you know, they are taking the book out and referencing. So then the relationship builds up over time and they become aware and it gets easier. I suppose from the time we have been together it has become easier when we do talk to doctors now, I think in the last five years I think it has improved dramatically, but I think there is a huge gap there in the knowledge base amongst the medical professionals, definitely.*

Carl and Cathy had tried to find out some more information before they decided to have a baby. At their first visit to the local clinic at the hospital they had a meeting with a gynaecologist and a cardiologist who both agreed that Carl and Cathy should not have children. By chance they were introduced to a professor through a Marfan support group who subsequently helped them decide to go ahead with their decision:

*Carl: When the gynaecologist and the cardiologist initially said no, no you shouldn't have any children, we were pretty upset and disappointed, but I think we kind of...let's call it off and see what the professor has to say before we take it to be gospel. In fairness to the local cardiologist, he did admit it himself that he did not remember Marfan syndrome, so from the little he knew of Marfan, he told us we shouldn't have any children, but that was just from his understanding of it. And we said we were going to the clinic anyway to*

*see the professor and he said 'well, do that and see what he has to say, but I say you shouldn't have them anyway'.*

### **The meaning of genetic counselling**

The role of genetic counselling was considered as the participants described how they experienced this service. Four out of the six couples attended an appointment for genetic counselling, it wasn't offered to one couple and another couple were unaware that it existed.

Belle had to ask her GP for a referral to a genetic counsellor to help them with their decision. She found the meeting 'fairly helpful' as he confirmed the 50/50 risk and answered some of her concerns about having a baby. However, she then highlighted the lack of emotional support:

*Belle: I think about it more matter of fact than emotionally, I try to keep my emotions separate. But then I think that when we did go to the counselling (genetic) it was more medical, matter of fact. There wasn't much emotional support there.*

Adrian did not find the genetic counselling appointment helpful and described the meeting from his perspective:

*Adrian: The doctor sat down with a notepad as big as a bible, just flicking through pages of notes and barely making eye contact and talking under his*

*voice 'blah de blah de blah' and just talking to you (to Belle) and that wasn't counselling at all.*

Adrian did not feel included in the meeting and told me that he did not understand what the doctor had said to them. The meeting was impersonal for him and there was a sense that he felt invisible and superfluous.

Clare and James also visited a genetic counsellor when they were making their decision. Clare remembered the appointment 'I don't think she really counselled us she just gave us our options and what was available'.

Jack and Joan recall their visit:

*Joan: The genetic guy just stood there all men and women thing and did all these circles and squares, and obviously the medical tests they do alongside which was already confirmed what I had...I don't know if the genetic counsellor helped me with the decision, that's just me personally. I knew the best what I was going to do. We just went through the whole process, at least then at the end of the process we could have said that we have all the information, we didn't go into it that blindly, but really from a medical point of view we probably did. (Jack: humming agreement throughout)*

The account of this visit to the genetic counsellor served to confirm that Joan's mind was already made up to have a child 'I knew best what I was

going to do', and this was just a routine process that attempted to gather more medical information.

Theresa and Tom described genetic counselling as 'it's quite clinical and these are the facts'. Lorraine and Bob told me that it was 'not offered'.

### **The role of the Internet**

The participants described how they searched the Internet for information but generally they did not find it helpful in making their decision. Belle and Adrian did not have access to the Internet but other participants told me their reactions to the various websites they visited.

Joan felt that the information could 'be quite scary' and 'very upsetting reading that, you know the worst case scenario'.

Theresa put it in perspective when she stressed that she would prefer to 'talk to a person affected with Marfan syndrome and hear that they have a quality of life. That says a lot more than reading all these scary lines'.

Similarly Cathy remembers reading up on pregnancy and Marfan syndrome:

*Cathy: I read up a bit on the Internet and then I would stop doing that because it would start me worrying, because it said about all the complications, so I stopped doing that.*

Bob felt the Internet '*panics people*' and that included NHS direct.

### **Exploring options**

Five out of the six couples explained how they explored all the options available to have a baby before they decided on the natural route. They had considered adoption, in vitro fertilisation (IVF), prenatal genetic diagnosis (PGD) and pre-implantation genetic diagnosis (PIGD). Aborting a foetus was ruled out.

Belle and Adrian were discussing the risks to Belle if she went ahead with the pregnancy. Adrian had already told us that he was informed about Marfan syndrome and he was not persuaded to agree to the decision for Belle to become pregnant. There was frustration in his voice and a strong sense of responsibility to find another way to have a baby came through as he said:

*Adrian: ...personally I would rather adopt than have Belle destroyed by pregnancy.*

By using the very powerful adjective '*destroyed*' to describe his image of how he felt his wife would be affected by pregnancy Adrian was fantasising about the devastating consequences he feared pregnancy might have on her body. This had the desired effect of making himself heard to Belle.

Interestingly Belle had been quite upbeat and positive during the first part of the interview and on hearing Adrian's comment she became quite pensive as

she described the fairly extensive physical Marfan-related problems she was experiencing at that moment. This confirmed that she had indeed heard what Adrian was saying. She concluded by saying 'How much use am I going to be afterwards kind of thing after putting your body through that'?

However, Adrian responded by playing down the risk of natural childbirth by saying:

*Adrian: The only part they worry about is labour as far as your heart goes, but then they've cut that out by saying that it will only ever be a caesarean for you isn't it?*

Belle became upbeat again as they discussed a positive meeting she had with her cardiologist regarding pregnancy and childbirth.

James and Clare remembered what it was like as they were trying to decide which route to take:

*James: You were very emotional at the time; you were upset a lot about what route to take.*

*Clare: I think once I'd made my mind up I thought, if we didn't naturally then we could have gone IVF as a second route or we talked about adoption as well didn't we? We even spoke about adoption as a preference to IVF*

*because of the risk involved to me. So that was...we did talk seriously about that.*

Clare's opening line 'once I'd made my mind up' demonstrated her expectations of having a child naturally. Initially she used the first person 'I' and then changed to 'we' when she was talking about considering the alternatives to a natural childbirth. This was further evidence that her mind was made up to become pregnant and give birth naturally and that the options were being explored solely as a routine process to satisfy her husband.

### **Assessing the risks**

The participants considered the risks involved as they tried to make their decision. The dominant risk they considered was 50/50 risk of their child inheriting the gene. The 50/50 risk was generally acceptable, but it was the males who could be more cautious and consider the risks as more risky than the females. This was further confirmation that the females had already decided that they were going ahead with the pregnancy, regardless of risk.

Belle and Adrian saw the risks quite differently. Adrian was assessing how the severity of Belle's condition would affect the severity of their child's Marfan if they decided to have a child:

*Adrian: If Belle has Marfan syndrome on the scale of 1 to 10, she is about an 8 in severity and a child born from someone with Marfan syndrome has a 1 in 10 chance of that being a severe case, absolute severe, 10 out of 10, walking,*

*twisting, skeleton, skinny, not going to last very long and I think of that, it does feel incredibly selfish to say that I want kids. But I do want to have kids you know.*

Belle responded:

*Belle: But you have also got to think that there is a 50/50 chance, that they will be born healthy.*

Clare and James followed the trail of events as they made their decision to go down the natural route. It was a lengthy process waiting to make that ultimate decision to go ahead. James picked up the story following Clare's heart operation and their subsequent investigations into having a baby:

*James: We had all these questions that we possibly imagined, so what's the risks, 50%, what's the risk to Clare, well that's more than average which was 1% so we are looking at 10% risk to Clare, a risk of aneurism, but with the aorta root replaced...it must have been a good few months...*

Clare completed the trail of events:

*Clare: It took about a year and a half to get confirmation from the hospital that they would be happy for me to try.*

Jack and Joan became emotional as they made some adjustments to the odds, which they felt would have made a difference to their decision to have their son:

*Joan: ...even though the chances were 50/50, if the chances were 80/20 it might have been a different situation.*

Jack followed up the confirmation of how this might have affected their decision to have their much-loved son who did not have MFS.

*Jack: Yes it would have been difficult. It wouldn't have taken a lot the other way I think if it was 55% I would have been really hesitant. Oh dear...(they both become upset).*

It seemed as if this was the first time they had realised the consequences of their decision-making.

When Lorraine and Bob were considering having their son they assessed the risks to the child based on the severity of Lorraine's condition. This was commensurate with how Adrian was considering the severity of Belle's condition as they were trying to make an informed decision. Lorraine explained:

*Lorraine: When it came to having our son they told us that as I have got Marfan syndrome that would be the same, they won't be any worse than what*

*I have it will be exactly the same. If I have something the matter with my heart, but my eyes are fine, then it's more than likely the kids will not be any different. If it was a really bad strain of Marfan syndrome it's not going to get any worse for the kids. I was fine, so the reason we chose to have our son after that was, I was fine...'*

### **Optimism of medical advancements**

Medical advancements were a major consideration while the participants were considering the outcome of their proposed decision. They expressed how they felt optimistic that the medical knowledge of MFS would continue to improve as it had done since the women were younger.

Adrian was expressing to Belle that he was concerned about how Marfan would affect her health in pregnancy and childbirth. Belle was trying to convince him that she would overcome any difficulties and she argued that '...they can fix things anything these days'. Her optimistic statement dismissed Adrian's concerns and gave finality to the situation as she projected the problem over to healthcare professionals.

James told me that he and Clare were optimistic about the care of their daughter and that medical advancements were a consideration when they were deciding to have a baby:

*James: 'In the decision process as well the way the medical advancements have gone over the last thirty years...it has progressively improved over the last thirty years since Clare's been alive.*

Similarly Joan tells me *'we are not as ignorant as we were 20 years ago you know when we were growing up. The more you learn about it the more you can make informed decisions really.*

Theresa shared her optimism in the interview:

*Theresa: What am I now thirty-two, twenty-eight years ago what people knew about Marfan was pretty serious. If you were facing that you would be more inclined to think I won't have a child, but we know more about it you have a good quality of life. You know one of the big actual points I was reading before all of it was the life expectancy of a Marfan patient was I think two years below the normal life expectancy. I thought that's pretty normal and the advances in the heart surgery, you are talking about you are not going to die at a young age.*

In normalising Marfan syndrome as a non life-threatening condition in today's advanced medical conditions Theresa was mentally raising the odds in favour of making a decision to have a child. She had a realistic sense of the risk being lower in the present medical environment than it was when she was a child; this was confidently backed up by providing 'evidence' of advances in

heart surgery, quality of life and life expectancy marginally below others without the condition.

As we turn to Carl and Cathy, their thinking was on similar lines to Theresa's reasoning. At the beginning of the interview Carl and Cathy discussed the whirlwind of events they went through as they were trying to decide whether to have a baby. Carl enthusiastically took the lead in the conversation as he explained how medical advancements affected their decision-making. He explained how this was a consideration and helped to substantiate their decision:

*Carl: Where it was a life-threatening disease I don't think it's such a life-threatening as it was, so we kind of use that to justify our decision a little bit if you follow what I am saying...not like if you go back twenty years there was no monitoring there was nothing. That was twenty years ago. Can you imagine what it would be like in another twenty years?'*

#### **4.3 C. FINDING PSYCHOSOCIAL SUPPORT**

The participants described the different sources of support they experienced during the decision-making. The women's mothers had an influence on the situation for both the women and their partners. Love and the strength of the couples' own relationship was an overwhelming factor in their support structure. However, for a while they mentally led parallel lives as they employed their individual coping strategies. There was a limitation to the extent to which friends provided support. A support group with other couples

who had successfully been through the experience of decision-making was considered as potentially helpful. The role of psychological counselling was explored and viewed as potentially helpful. Interestingly, they described the interview as a therapeutic experience. This section concludes with the couples giving positive and negative evaluations of the health professionals and what they considered to be the essence of helpful support.

### **The effects of her mother's experience**

I asked the participants about their relationship with their family and how it was when they were going through the decision to have a baby. They generally did not share the decision with his parents or her father, but the women's mothers all had some effect on the decision-making process.

I asked Belle and Adrian if they had shared making a decision with their family. Belle did not really discuss the Marfan with her mother. However, they were both aware of how she felt and I noticed that as they told me their voices became a little depressed:

*Belle: My mum's terrified.*

*Adrian: Yeah your mum is terrified.*

*Belle: She doesn't even want us to try really...*

In that brief moment as they considered Belle's mother, Belle's mood appeared to be deflated. Belle quickly moved on in the same sentence to talk about her sister as being supportive and her voice became upbeat again. It

was obvious that she did not want to linger on her mother's negative response to her considering becoming pregnant. Her mother was not mentioned again in the interview.

In contrast Clare and James found Clare's mum to be quite supportive and James recalled *'we consulted your mum because she had more knowledge about it'*. Clare responded:

*Clare: My mum just says I wouldn't change you for the world and it's not that bad a disease. She kind of focused on all the positives.*

Theresa's mother supported Tom and encouraged him to attend one of the Marfan information meetings when they became serious about starting a family:

*Tom: I think myself and my wife started to get shall we say serious maybe four or five years in. My wife's mum definitely pushed me to go to one of the meetings.*

Tom felt that his mother-in-law wanted to make sure that he was 'fully aware' of the implications of their decision. He respected her thoughtfulness in wanting him to know the facts before they make a decision.

Lorraine told me how she was able to talk with her mother about Marfan because she had it too. But Cathy was more stoic, and kept things from her mother so as *'not to make her worry extra and unnecessary'*.

### **The strength of the relationship**

On a number of occasions in the interviews the couples demonstrated the extent to which they loved and respected each other and wanted to be together. Marfan syndrome was part of the relationship and this provided a secure base from which they could make a decision to have a baby.

At the beginning of the interview with Belle and Adrian I asked my opening question, 'what has it been like for you making the decision to have a baby?' Following Belle's response I turned to Adrian to ask him how he felt:

*Adrian: Terrified. It goes completely against the grain doesn't it? You don't want your kids ever to be ill or whatnot and now it's a 50/50 chance. Do you know what I mean? Especially for a bloke it's against evolution. You don't look for a mate, you look for a healthy mate don't you? This is the opposite. Love's a very powerful thing I guess. It doesn't matter as much, but it does matter hugely.*

Adrian's description of his decision-making process here was consonant with his previous comment about making a decision using 'heart and head'. The extract demonstrated the complexity of his thoughts and feelings and explained the fear and frustration of his dilemma. Finally, he summarised by

verbalising the contradictory nature of the conflict he was experiencing both emotionally and cognitively.

Clare told me that she was honest about her condition from the beginning of their relationship and tested James's love for her by giving him the opportunity to 'run for the hills' if he wanted. James proved his love for her by resolutely standing by her in the relationship.

Jack explained how the strength of their relationship intensified their ability to manage the situation and isolated them from needing others to cope:

*Jack: There were times when it was, us against the world, we knew what we were going through.*

Tom described how he shared Marfan with Theresa as part of their relationship. He explained that he did not define her as an illness or condition; her identity was as a woman and as his wife:

*Tom: So I suppose my knowledge of Marfan has been progressing through the years, so when we made that decision to get married I suppose I wouldn't even have thought about the Marfan at that stage because it would have been part and parcel of our relationship for a long time...I don't see it as Marfan I just see my wife.*

**Temporary parallel lives**

There was evidence that the participants coped in their own individual world at times and did not share their thoughts or concerns with their partners. This was because they did not want to show any vulnerability, become another problem for their partner to deal with because they needed to conserve their strength to cope with their situation. It could be difficult for them to talk to each other about certain things and temporarily withdrawing may have given them the opportunity to process any problems and work out the best coping strategy. Honesty was not always the best policy as they temporarily felt that the relationship would benefit from a modicum of independence.

Adrian explained how he kept things to himself sometimes rather than share his thoughts with Belle:

*Adrian: A lot of things you can't talk to Belle about because how can you, it will just end up in a row! (all laugh)*

*Belle: Thanks!*

*Interviewer: Well that's honest (all laugh)*

*Adrian: It's true because you can't be honest sometimes to the people closest to you because it can hurt, they have their own problems, and they don't need to hear your problems. Do you know what I mean?'*

*Interviewer: And you wouldn't really be able to talk about what is really, really on your mind; you wouldn't be able to say it.*

*(Both nod in agreement)*

Interestingly Adrian talked using the third person and he generalised to other people instead of keeping the experience as his own. Throughout the interview he often asked me 'do you know what I mean?' Here again he asked for confirmation that I had understood what he was saying to me and I responded by reflecting back my understanding in order to check it out. Their silent nod in agreement confirmed not only that I had understood, but also that Belle felt the same way as Adrian.

Similarly, Clare and James had been considering whom they turned to for support and concluded that at times they coped in their own worlds:

*James: Most of the time I just muddle through myself. That's just the way I am.*

*Clare: I think you would have benefitted from talking to somebody probably on your own at times because then you wouldn't have...*

*James: I think that's probably true, I could talk things through without having to worry what your... you knew your own mind, I guess so...*

*Clare: I knew I was fine myself, that's what I kept saying...I didn't though, I was really scared as well, but I kept saying I was fine.*

If we turn to Joan, she offered a powerful explanation of the function of the parallel lives that evolved. Although she was considering her own experience, she talked objectively on behalf of all women with Marfan and their partners as she explained the need and the purpose of withdrawing to conserve the necessary energy required. She described the mutual empathy that emerged from the situation:

*Joan: It's always harder for people on the outside because as a Marfan patient you kind of have to use your strength to deal with what you are doing. Whereas those on the outside, it's always harder because they are actually seeing it, they have to be strong and they really can't understand that it's always harder looking in than it is actually being there. It's difficult, I have only realised that in the last few years, how difficult it is for the partner who cares because they love us you know and they don't want to see us in that situation. It's like if your child is hurt, you cry with them.*

Cathy also explained how she and Carl had always been very open, but following the decision to have a baby they needed to cope individually to maximise their strengths. Here, Cathy expanded on the meaning of briefly leading parallel lives:

*Cathy: Before it (the decision) we went through everything and our choices together. And then a little bit of a lull in our communication, and after we seemed to explode with all our emotions. And I suppose the time I needed him to be strong he was strong and I didn't know he had all these worries. So I don't know if there would have been any point in him sharing with me when I was pregnant because maybe I just wouldn't have handled my pregnancy so well.*

### **Limitations of friends' support**

I wondered if the couples talked to their friends when they were making the decision. And if they did, what was it like? Although four out of the six couples talked to friends, including close friends, they found that the amount of support friends could offer at that time was limited. Friends found it difficult to fully understand Marfan syndrome and the implications of what it was like to make the decision to have a child. Essentially the decision to have a baby lay within the privacy of the coupledom.

Belle and Adrian were discussing their various friendships and it was clear that all their friends already had children which left Belle and Adrian as the odd ones out in their group of friends. Belle told me:

*Belle: They are all expecting us to have, or at least be trying at the minute, and I don't think any of them really realise, that although they know I've got Marfan, that it can affect pregnancy. And I think they thought it can be passed*

*on and things, I don't think that even enters their mind when they ask us when are you going to have a baby then and things like that.*

Belle and Adrian were coping with the social expectations of others.

Jack and Joan understood that it was difficult for others to understand about Marfan and felt that friends would not be able to help them make a decision. Jack explained why they made their decision as a couple and did not share it with friends:

*Jack: It was the worry of what could happen and it was the thought that if I talked to people about this we could have gone off at a tangent and made a bad decision, anyway **we** were going to have the child. If our child had turned out to be handicapped, I would always remember those conversations I had had with my friends. It could have been one of those cases of 'I told you so.'*

Jack was concerned about the repercussions of sharing the decision-making with friends. 'Off at a tangent' suggested that talking to friends would have been a distraction. There was an element of shame as he considered how they might have reacted if they had a handicapped child. He emphasised the word '**we**' as he wanted to share the decision with his wife. Whatever the outcome of the 50/50 chance might have been he would only have to cope with that and not his perception of how friends may react.

Theresa had never really discussed the Marfan with her friends and explained her reasoning. It is worth noting how she used both 'I' and 'we' as she considered whether she would share their decision-making with friends:

*Theresa: I don't think I would have talked to people about whether to have children or not. It would have been a case, these are the decisions we are making and talking about the situations that we were dealing with. From being pregnant and meeting the doctors, I would probably have spoken about it at that stage. Before I decided, we decided, to have children I don't think I would have chatted to friends.*

### **The role of support groups**

Talking to other people in the same situation as them was something they felt would be helpful. The women, in particular, considered sharing other couples' experiences of making the decision and going through pregnancy and childbirth as a significant potential source of support.

Belle and Adrian both felt that other peoples' experiences would help them as they went through trying to make a decision. Adrian highlighted the lack of support:

*Adrian: You don't hear any stories of others, you feel isolated to be honest. This is a great study because I'm sure there are other people going through it and it's not accessible for us to find them.*

Carl and Cathy went along to a Marfan syndrome group meeting:

*Cathy: The one thing I found helpful was the Marfan syndrome group. We meet people in the same situation so we feel like we can discuss it with them. The one girl we talked with earlier on who also wanted children, she was great and she was like, she is very personable and she shared with us what she went through and how she went through it and she told us as much as we needed to know.*

Theresa and Tom both felt so strongly about the need for a support group that they wanted to provide a support structure for other couples that they would have appreciated themselves:

*Theresa: ...from my point of view one of the biggest things I want to do is put that personal touch out there and go 'I am a Marfan person and I have kids and here's my story' kind of situation, so that someone in my book would be able to go and find me so that they would have the sort of necessary support that I didn't have...*

### **Psychological counselling as potentially helpful**

Having established that none of the couples were offered or attended psychological counselling I asked them if they would have found it helpful.

Belle responded:

*Belle: Yeah, it would have been a big benefit emotionally.*

Adrian continued:

*Adrian: 'I've had counselling in the past...counselling has always helped and I've always been open to it...'*

Belle and Adrian had been coping alone with trying to make their decision. Adrian had already told us how isolated he feels and Belle now stressed that at no time during any of the appointments with healthcare professionals had any help been offered to them:

*Belle: In terms of Marfan we have never once been offered any psychological help which I think would have been a great help to be honest.*

Clare didn't feel that she would want to talk with anyone 'specialised', but James felt differently. He was reluctant to admit that he may have found it helpful:

*James: In hindsight I could probably see the benefit of it but the thing is we muddled our way through and got where we wanted to be, so I don't think it would have been a major...I wouldn't change anything, but in hindsight it could have been a benefit for us to have talked through things, it was quite an emotional time.*

They remembered some of the difficulties they experienced when they were making the decision and James realised that he 'just muddled through'. Clare said that she thinks he 'would have benefitted from talking to somebody probably on your own at times'. James agreed:

*James: I think that's probably true, I could talk things through without having to worry about your...you knew your own mind I guess.*

Jack initially told me that talking to someone would be a weakness, but he then reconsidered who he would find it helpful to talk to, 'I don't know, a counsellor maybe'. Joan agreed as she said to him, 'that way you could off-load'. He then confessed that there was a time just after they had had their son when he would maybe have appreciated talking to someone:

*Jack: I think I could certainly have done with counselling when you were ill leading up to your operation. That was a very, very bad time for me. It was like the whole world was crumbling around me. (Joan agrees with him as she nods her head and says 'mm'). It's terrible looking at your loved one and there is nothing you can do.*

Theresa and Tom didn't see any need for a third party. However, Carl considered whether counselling would have been helpful for him. He had been used to coping stoically and I asked him whether he might have found it helpful:

*Carl: I am not really...I find it...I wouldn't sit down and talk to complete strangers much...(I nod and we all end up laughing as we realise he is talking to me, a complete stranger!)...with regards counselling, even if it had been available, would I have went, probably not, if it was available, but it wasn't though. I think it's right that it should be available for people who are asking for it, like I say for me no...when all this was going I just kept it inside.*

### **The interview as a therapeutic experience**

At the end of each interview I gave the couples the opportunity to share what it was like for them talking about their experiences with me in the interview. They all found the interview to be a positive experience.

Adrian and Belle explained that it had been a space to hear what each other had to say and to try and make sense of their thoughts about the decision. Belle told me, *'it's been good'* and Adrian agreed, *'it's been nice'*. Belle then continued:

*Belle: It's actually been nice just to sit down and talk and think about it properly for a while. It's always really in the back of your mind and if you have discussions about it, but when you sit down and talk about it with someone else you start to understand the situation. I got quite upset in some places it gets to me (she starts crying). Give me a hug (to husband)!*

*Adrian: It's nice to be able to have an intermediate in the room so we hear things that we haven't necessarily said to each other yet.*

Clare turned to her husband James and said *'It's quite refreshing for me to hear what you think'*. James responded:

*James: To be honest half the things I had forgotten about. And you questioned me on Clare and my memories so in all honesty I had never thought about anything that had happened until this, until today. So it's like I didn't realise that perhaps when we were asked to do the interview I was very much of questions, questions, that will be it and we made our decision for research. But actually now I am thinking about it there is a lot more emotion there than I realised. In terms of conversation, yes we had bits and pieces, but the thought-processes in the back of my own mind about life without Clare or holding a baby are things, which were there. So I mean it's like you said it's been eye-opening. So I have to say I hadn't thought about it.*

When I asked Cathy and Carl what it had been like for them talking about the decision in the interview it became evident that Carl had been thinking about it and had a preconceived idea of what the interview might be like for him:

*Carl: Not as emotionally draining as I thought it would be. I was thinking I should have a box of tissues at the ready just in case, but no it was ok to talk about stuff that I had clearly forgotten myself and you remind me.*

*Cathy: I think Carl really likes this counselling, I think I am going to get him a couple of sessions for Christmas! (All laughing)*

#### **4.4 D. EXISTING WITH THE FALLOUT OF THE DECISION**

The final superordinate theme is a powerful exploration of the final stages in the decision-making process. The participants described their thoughts as the focus of their attention shifted from the hypothetical to the practical. In the first sub-theme, living with the physical and psychological consequences of coping with a high-risk pregnancy are explored. The women expressed their concerns for their child's well-being, which overrode the risks to themselves. The men had an overwhelming fear of losing the woman they love in childbirth and talked about how they coped and often cried alone. The men considered the reality of 'Who's left holding the baby?' as they struggled with the ongoing uncertainty of the risks to their wives in pregnancy and childbirth. Five out of the six couples reported how they sometimes felt responsible, selfish or guilty for putting a child at risk of inheriting Marfan syndrome. However, there was also an acceptance of the 50/50 gamble, uncertainty and fate.

##### **Coping with high-risk pregnancy**

The participants described the impact of coping with a high-risk pregnancy. It was the men who defined the tension between wanting a child and coping with the risk of their wives being pregnant and giving birth.

Belle and Adrian were talking about the decision as being 'a very measured decision' and Adrian became quite animated as he said:

*Adrian: It always feels like we are waiting for something as well, waiting for this to happen, waiting for your medication to come down and it's never going*

*to happen. When do you start? They have also said you know it needs to be measured, we have got to have a team on you from day one. As what do you do ring the doctor and say right I am taking my clothes off now and she's lying on the bed and we are getting started! (All laughing) It takes some of the magic out of it you know. It's like those guys whose wives are having trouble conceiving. They have to make the charts up, and then on the clock, beep, beep, beep, oh they have to go home!*

Adrian was seemingly comparing a high-risk pregnancy to infertility. In talking about the decision as '*waiting for something*', '*measured*' and '*takes some of the magic out*' Adrian illustrated how adapting to a high-risk pregnancy required a careful balance of accepting a more cautious and considered pregnancy and remaining focused on the purpose of reducing the risks. He mistakenly believed that the conception had to be '*measured*' as with infertility, provoking a somewhat gloomier forecast of what might lie ahead.

Carl and Cathy remembered how they felt when they made their decision to have a child. Carl had been doing most of the talking and became quite emotional. The impact of high-risk pregnancy was causing Carl to agonise over the decision as he took the blame for putting his wife at risk:

*Carl: The thought of putting Cathy's life at risk even asking her to do this. I remember it was just so, so tough for me to ask my wife to have a child for us and me putting her at risk and the child at risk as well. I just remember thinking it's so tough, it's really horrible.*

Bob told me how he felt the impact of Marfan became serious for him when Lorraine was getting near the time to give birth:

*Bob: The only time it came into the equation for me was when it was getting near the time for him (son) to be born. They was always monitoring Lorraine's heart, she can't push for too long. This was the only time it came into the equation for me, I was like wow bloody hell... (silence)*

*Interviewer: So what did you actually think at that time, 'bloody hell'?*

*Bob: You go like she's got all these tests and then someone said she can't push for more than half an hour. I am like what is this, is this going to get complicated.*

So, Lorraine played down the risks of Marfan syndrome, Bob had limited knowledge of the condition and left it up to her to tell him if anything was wrong. However, the practical reality and proof of the risk had shocked him and evoked a contradictory response to pregnancy and childbirth for his wife with Marfan syndrome.

### **Her concerns for child**

The women appeared to be more concerned about their babies than the risks to themselves. They were concerned that they could have given the child Marfan syndrome and expressed their wishes to put the baby first if faced with a life-threatening situation during the birth. There were concerns regarding

their fears of who would care for the child if they were unable to do so, and it was important to ensure a safe and secure future for their unborn child:

Belle told us some of her concerns for the child she and Adrian proposed to have:

*Belle: I understand there is a lot of variable factors in the Marfan syndrome condition, one person is never like the next. Even though I have been relatively well in my childhood other than joint problems, our baby could be a lot worse than me, it could be affected in a lot of different ways. It could have a really bad heart and I think having gone through it and seeing Dave and Joan (family) go through it with their problems, it's made it hit home a bit I suppose...I don't really think about the risks to myself, where you do I suppose (to Adrian).*

In considering her decision to have a baby Belle seemed to be focusing on the situation of her own baby being found to be affected with Marfan syndrome and tried to imagine the consequences. In thinking about the possible outcome she compared her own situation with that of other affected relatives. But her personal experience of coping with the condition and her concerns for the child diminished her perception of the risks to her.

Clare captured the essence of what it was like for her when she had made a decision to have a baby as she demonstrated her understanding of potentially being in a no-win situation:

*Clare: ...I was always 'save the baby'. Then I would get upset wouldn't I, what if I bring this baby into the world and then I die, it's not fair on that child, they would never have a mum. And then I go through the other side of things, and even when I had our daughter I had really bad hallucinations every night that I'd lost her.*

Clare too considered preparing her family for the worst, as she was concerned about providing a secure future for her unborn child:

*Clare: Looking back I did throw a lot at everyone really. I didn't realise I was upsetting everybody, but when I was saying things like 'mum if anything happens to me will you help look after them' I was trying to plan for the baby's future if I was not around, because that was my biggest fear because if I'm dead, I'm dead you know, don't worry about me it's the baby that needs looking after. I guess that's the maternal instinct really...*

Joan put their decision-making to have a child into perspective as she considered who actually made the decision. She then continued and normalised her concern for the child as she reasoned what it was like for a woman with or without Marfan to have a baby:

*Joan: I have to admit, I mean we both made the decision, but I think as a woman it is ultimately my decision whether I do as a woman. As a mum I would be more concerned about the child and not myself, but then again I think that would be the same for anyone if you had Marfan or not.*

### **He fears losing her**

While the women were altruistically concerned with the risks to their child, all the men expressed a fear of losing their wives. The immense risks to a woman with Marfan syndrome in pregnancy and childbirth were not to be underestimated as the men consider the 'worst-case scenario'. They imagined what the possible outcome of their decision to have a baby could be, and they were apprehensive about how they would cope if they were left to care for their child:

Adrian was concerned about how Belle would manage in childbirth. Adrian was considering the worst-case scenario of being left to care for the baby. He questioned his ability to cope and tended to feel pessimistic about the outcome of their decision. Interestingly, even though his hard-hitting remarks were aimed at Belle, his focus was very much on directing his point across to me to make himself heard:

*Adrian: And then how am I going to be around everyone if it's put you down, if you're bedridden for a few months afterwards? How am I going to be around everyone when I'm getting up in the night to look after baby, getting up at six in the morning, going to work and then coming home and doing any housework or anything that needed doing and looking after the baby, making sure that everyone has eaten blah, blah, blah...and go to bed, an hour's sleep then back up and look after the baby. You know, how am I going to be because I'm the sort of person that needs time to himself, I really do need... that little bit of time once a week to myself keeps me sane. I would never run*

*away from this situation, but I would feel like it hell of a lot in that situation and that scares me.*

James explained how he felt about Clare being pregnant and he was able to see the situation from her perspective of 'save the baby' as well. He empathised with her physical and emotional attachment to their unborn child:

*James: Actually the biggest thing that happened I was more concerned about you and you were more concerned about the baby. I guess it was because I didn't have the emotional attachment because obviously the baby is not growing inside me. I saw the scans and saw I really wanted a child. I guess I feel that living thing inside me as Clare would. I was more worried about Clare than anything else.*

James then told us it would have been good to have a 'sounding board', someone he could talk to as he struggled to cope with the emotional side of the decision:

*James: I know all the medical, it's more about the emotional side of things, my fears I guess of losing you and what then if I was left holding a baby by myself, those sorts of things. You and I had those discussions.*

Tom wanted to emphasise how his concerns for Theresa were so strong that he wanted her to know that he would have put his wife's life before the baby:

*Tom: ...I told my wife this; if anything happened she would be the priority. I said to my wife if there was any complications, any decisions to be made, I suppose I just needed to tell her that she would be...that if there were any complications I was going to make her the priority that was the big thing for me. I said it a few times; from my wife's point of view, she heard me but she wasn't hearing me, so I may have pushed that a little bit.*

Tom's experience of not being heard by his wife was commensurate with Adrian's account of his concerns for Belle and having to stress his point in order to make himself heard.

Carl told me how he questioned the decision they had made before and after Cathy had given birth. Here, as his wife was experiencing complications with the birth, he realised that he had previously been either unaware or in denial of the full impact of the risks to his wife in childbirth. He responded by assuming responsibility for the decision and was full of self-blame and self-recrimination:

*Carl: The morning after our baby was born I questioned the decisions we had made, I questioned the decisions that I had made really...I was looking into my heart and asked what the fuck have you done you idiot, you know what I mean. Especially when our daughter was born there were so many complications which you half expect, though I suppose it was not what I was expecting...it was all very scary, let's just...and I remember I just went to the*

*toilet and looked at myself in the mirror and thinking, what have you done you fucking moron, you know what I mean...*

### **He copes and cries alone**

Generally the men 'bottled up' their emotions as their wives coped with the practicalities of pregnancy and childbirth. The purpose of not sharing their feelings with their wives was to protect her from having any additional problems to cope with. This sub-theme contained some of the most heartfelt comments from the men as they struggled to cope with the decision:

Belle had just said to Adrian that she felt he was 'preparing for the worst'. I ask Adrian what it was like hearing Belle say that:

*Adrian: It makes you cry at times. You have got to go and have five minutes or a long shower or just have a good cry and let it out; I don't like anyone to see that.*

The following conversation demonstrated the strength of the men's emotions as Jack described how he struggled to cope with Joan's Marfan syndrome and her determination to have a baby:

*Interviewer: So you didn't like sharing the emotional side with you wife.*

*Jack: No, no.*

*Interviewer: So what did you do with that?*

*Jack: Just dealt with it, just had to bottle it up inside me. I just had to reassure myself that in the end everything was going to be all right (he looks emotional and his eyes moisten)*

*Interviewer: A few tears?*

*Jack: Yes plenty of tears, floods of tears.*

*Interviewer: Where did you shed them?*

*Jack: At home mostly, while my wife was asleep I would go through to the lounge and sitting on my own. I was just thinking of it all, the worry and the stress and what was going to become of it all. Many times I would just get up and go and have a quiet time in the lounge just thinking about it all.*

As we have seen, the men bottled up their emotions. Bob told me what it was like for him and explained how he wanted to protect his wife from having to cope with his concerns:

*Bob: I tend to bottle things up a bit. If I think that she (Lorraine) thinks I am worried then she will get more worried. So I try to keep it inside of me more.*

Similarly Carl talked about making the decision and hiding his emotions:

*Carl: I was petrified, I was absolutely terrified to the core and the only thing I knew to do was to keep that inside of me. The last thing I wanted was for Cathy to see I was afraid for her. I didn't want to show any of those things to Cathy, certainly not during the pregnancy and I suppose certainly not before she got pregnant, in case she might change her mind (laughing). But certainly during the pregnancy I was terrified and worried and everything else, but I didn't show anything to Cathy, I barely showed anything, just kept my fingers crossed and hoped for the best. As I said I am an optimistic person you know, 'the glass is half full' I just thought let's get on with it.*

### **Feeling responsible, selfish and guilty**

The participants all experienced feeling responsible, selfish and guilty.

Adrian was very keen to have a family and would have found it hard not to have children. However he also felt that the decision was somewhat selfish:

*Adrian: It would seem like a hollow existence, it's what we're here for. It seems selfish even, and hollow even if there are matters out of our hands like. It may stop at that, but still each experiences things in life and you want to share them.*

Clare and James were discussing what it was like for them when their daughter was diagnosed with Mafan's:

*Clare: That's why I was so upset when she was diagnosed because I felt real guilt for you and I really I remember that's why I kept saying I am so sorry.*

*James: Yes you were crying and saying 'I am so sorry'. But that was never an issue with me...*

Jack was telling us about what it was like feeling the responsibility of making the decision to have a baby, and was describing the associated feeling of guilt if his child had had Marfan:

*Jack: I wanted to have a child; I couldn't imagine not having a child you know, the worry was there. Then if the child has got Marfan then you know it's my fault, I'd feel bad you know that I had let it happen. Terrible thoughts go through my mind that I wanted a child and pledged whatever happened we would love that child and nurture it and always put the child first sort of thing.*

Carl and Cathy were thinking about what it was like when they were making their decision regarding passing on the Marfan gene to their child. Carl told us that he felt selfish, as he did not regard passing on the Marfan gene to his child as his responsibility. As we have seen, Carl had left the responsibility of coping with Marfan syndrome, pregnancy and childbirth to Cathy. Although Cathy had been agreeable to this arrangement, she quietly confirmed how very difficult 'tough' she would have found it if she had passed on the gene to her child:

*Carl: I think your instinct is to have one child. We would probably still have gone through with it now. We find we are a little bit blinded with the realities of what might have happened, then again it sounds so selfish I know it's going*

*to sound terrible, but if it was our child who had all these terrible Marfan things that she should have had, again it's not us that's going to have them, it's the child. I know that sounds absolutely horrible, but that's the way it is.*

*Cathy: I think I would have found it very tough if I had found out that I had given my child Marfan syndrome, if I had passed on the gene, the Marfan syndrome gene (silence).*

### **Acceptance of gamble, uncertainty and fate**

Having decided to go ahead and have a baby the couples recognised the limitations of making a fully informed and accurate decision. It made sense to define their situation with a generally positive view of the prospective outcome. They acknowledged that the future may not be perfect, but their choice was partly based on what they thought they could cope with at the time. They had assessed the personal, social and emotional consequences of the risks and demonstrated that the burden of making the decision is not to be underestimated. Although the women were the leaders in the decision-making, both parents were intent on sharing their life with a child. The couples accepted the opportunity to make the decision, regardless of the risks.

Belle and Adrian were discussing whom they would consider talking to in the family about making the decision and tried to assess to what extent others understood their dilemma. Belle concluded the discussion by highlighting how she felt about the decision:

*Belle: It's the unknown isn't it? You don't know exactly what's going to happen, so you are just making a decision really on what? I mean people can advise you on...(shrugs), best they can, but at the end of the day you don't know what the outcome will be, you don't know how it's going to affect you, it's a big step into the unknown really.*

Clare and James had met a couple where the man had Marfan and he and his wife had decided not to have children. Clare remembered the discussion they had after their meeting and how, even though she felt 'selfish' in going ahead with their decision to have a baby, she was determined to stand by her choice to go ahead and get pregnant, embracing the uncertainty and fate of what lay ahead and she recognised the impossibility of making a fully informed decision:

*Clare: ...I think if you have some certainties in life you might actually, you might actually be able to make an informed decision, but I did say to you I want to be pregnant, I want to have...*

James then interrupted and brought himself into the decision by using the word 'we'.

*James: You were very clear once we made the decision.*

This comment then prompted Clare to recall a conversation she had that helped her to cope with gambling, uncertainty and the fate of what lay ahead:

*Clare: My sister's ex boyfriend, he said such a weird thing, he said James was the luckiest bloke I know, so it will be fine. So, because it's a fifty/fifty chance... (pause), so even that soothed me. I don't know why he was so wrong (their child has Marfan). I think that's why I was so shocked, but he said James was lucky!*

Interestingly Jack and Joan used her family history as a significant factor in their decision to finally accept the gamble of whether they would produce a child with Marfan. They had visited a genetic counsellor where they had mapped out the genetic history of her family. Jack commented on the possible effects that a more historical diagnosis of Marfan syndrome would have had on the decision and Joan's response rejected the need for certainty, summarising her total acceptance of whatever fate lay in store for them in the future:

*Jack: That could have made a big difference in our decision-making as well. You know, the further back they went.*

*Joan: Yes the further back they went, but when you're already sitting there with a genetic counsellor you know there is an issue, you know there is a problem. I don't know, it's difficult. If you're going to have a child, you're going to have a child. Women all over the world have children and they will accept those children no matter what's wrong with them.*

## 5. DISCUSSION

The purpose of this research was to consider the process of couples' decision-making to have a baby when a woman has Marfan syndrome. In this section I aim to consider and reflect on the findings from data derived during interviews with the six couples.

Four main superordinate themes were revealed in the analysis: Her decision is already made; Creating an informed decision; Finding psychosocial support; Existing with the fallout of the decision. The themes demonstrate a distinction between the individual male and female perspectives, and the perspective of the individuals as a couple. In particular I was interested in how the couples' accounts illustrate that when a woman has Marfan syndrome, it does not belong to her specifically because it was quite evident that it plays out in the dynamics of the couples' relationship. To this end, these perspectives are represented in the superordinate themes and the supporting master themes of the findings. These are now considered and discussed according to how they were presented in the findings section. Finally, there is a critique of the current research and suggestions for further research.

### 5.1 A. HER DECISION IS ALREADY MADE

#### **A predetermined decision**

The women were aged between 2 to 14 years when they were first diagnosed and knew they would like to have children when they became adults. They

had given some considerable thought to having children later in life and, despite the clear risks involved for mother and baby, the participants demonstrated their determination to have a biological child. Extant literature on reproductive decision-making for women with a genetically inherited condition presents a similar picture (Shiloh, 1996; Lippman-Hand & Fraser, 1979; Minnich, 1997; Peters et al, 2002). The age at diagnosis is a significant factor according to Peters et al (2002). They found that participants diagnosed at age 15 years or younger were able to assimilate any issues associated with Marfan syndrome in their decision-making at an early age. This was largely due to their heightened awareness of their condition and the implications of having a child from an early starting point.

The positive experience of living with Marfan syndrome as a child played an enormous part in the women's decision-making process. There is a very real sense that their child will be 'fine' whatever the outcome, as they had managed to cope with Marfan syndrome in childhood themselves. For Lorraine and Clare there is the realisation that this may not be the same scenario for their own child. Personal and subjective experience was empowering for them, but conversely created a dilemma when they considered the meaning of having a child with Marfan syndrome. This dimension recedes into the shadow as the focus on strength and resilience is brought to the fore. It is very clear from all the narratives, that they have acquired these qualities over the years in coping with the condition, and this gives them a more positive stance when weighing up the risks of having an affected child or coping with the risks to themselves in pregnancy and

childbirth. This is consistent with the findings of other research (Peters et al, 2002; Gallo et al, 2010).

Generally, many women can be inclined to think about having children from an early age, often long before they are in a relationship with a partner (Rijken & Knijn, 2009). Therefore, they enter into the relationship with this insight and the decision-making process starts with many women already knowing their wishes regarding parenthood (Scanzoni & Szinovacz, 1980).

### **Female drive and identity**

The women conveyed a great sense of their strong identity and psychological fulfilment due to the opportunity of assimilating Marfan syndrome into their lifeworld over time. A study by Williams & Schutte (1997) emphasised that the age at which an individual learns that they have a genetically inherited condition is an important consideration in reproductive expectations. They found that women who learned that they had a genetic condition in adulthood 'experienced feelings of loss of hope to have children or grandchildren who would be free of the condition' (p. 75).

The women who participated in my research expressed an overwhelming desire to carry a child and give birth naturally. Essentially it is powerfully driven by their identity as a female, which negates the risks involved in pregnancy and childbirth. This is consistent with research findings by Hill (2003) and Asgharian & Anie (2003), and resonates strongly with the study carried out by Gallo et al (2010) who found that the women, in their response

to both questionnaires and the focus group they attended, demonstrated a strong desire to have a biologically related child, despite the risks. This suggests that genetic counselling for women in understanding the risks with respect to having a child may be experienced as mere fact giving for them, as the longing to have that child clearly overrides any potential hazards. Indeed, studies have shown that individuals generally have a poor recall rate over time for the genetic risks discussed in genetic counselling sessions (Hallowell & Richards, 1997; Sorenson et al, 1981; Seidenfield & Antley, 1981).

Undoubtedly there are other possible reasons for the women's overwhelming desire to have a biologically related child, despite the risks to themselves and the child. One possible explanation may be the socially constructed role of what it means to be a female. Despite changes in society, motherhood is seen by some as the ultimate fulfilment for women; they do not need to be taught it, it is seen as natural, and women who experience a negative response to motherhood or choose not to have children are generally not regarded as the norm (Segal, 1999). Historically, women in their role as a mother ensure that their daughters retain their feminine traits through nurturing and meaningful communication; it is an intimate relationship that defines women only as in relation to others (Chodorow, 1984). With the rise of sociobiology and a new evolutionary theory following the influence of Darwinian theory, motherhood and its associated skills were assumed to be innate (Dawkins, 1989). Some psychologists (eg. Archer, 1996; Buss, 1994) were keen to emphasise that the theories of evolutionary psychology made more sense than the socially constructed notions of traditional male and

female differences. However, it would perhaps be impossible to set aside the prescriptive female gender role of pregnancy and childbirth from the expectations of society, despite women's changing role and attitudes towards pursuing a career and their overall identity.

It is evident from the narratives that the women attach a great importance to becoming pregnant, and what is clear in their accounts is the need to maintain their identity as a female. For these women, a successful pregnancy is not only the norm for social reasons, it can also be seen as crucial in nurturing both psychological and biological self-esteem, thereby, to a certain extent, maintaining a sense of personal and social competence as a woman. The female body in relation to pregnancy and childbirth has been considered as inextricably entwined with female identity, which may lead to a sense of loss, inadequacy and failure when a woman does not become pregnant (Butler, 1996). Existentially, motherhood could be considered as an embodied, meaningful and rewarding symbol of femininity (Radley, 2000).

### **Urgency of timing and age**

Severity of their condition at the time of decision-making varied in each of the women, from mild to the occasional need for the assistance of a wheelchair. De Bie, De Paepe, Delvaux, Davies, & Hennekam (2004) found that decision-making to become pregnant for individuals with Marfan syndrome was not influenced by severity of their condition. Regardless, there is a very powerful sense of the women's urgency to become pregnant as soon as possible, despite having to overcome their partner's caution regarding the risks.

Werner-Lin et al (2012) found that participants with the breast cancer gene expressed a preference for biologically related children, despite being aware that their partner would assume the burden of a genetically inherited condition via their children. The high level of self-awareness revealed in the women's accounts of their age and the condition of their health was evident. They were clearly aware that age and timing were critical, and there was frustration and anxiety regarding the numerous obstacles they had to overcome before they could proceed with a natural pregnancy.

Although the women's accounts indicate that they wanted it to be a joint decision with their partner, they are essentially claiming their authority in the situation. They emphasise the considerable importance of their ability to cope. It is likely that, as a result of their urgency to become pregnant, there may be less focus on the self and more focus on the persuasive techniques required to convince their partners that this is the right decision. Furthermore, as they strive for control in the situation, there is no doubt a continuing awareness for the women that there is a decision to be made as a couple. The women invest a considerable amount of energy in generating a positive argument for natural pregnancy and childbirth and promoting an optimistic environment. This may entail an additional psychological problem for them causing further complications in the decision-making process. The psychological implications of an increased awareness in decision-making to have a baby, in the context of genetic risk, could be viewed as an extra pressure for some individuals with Marfan syndrome (Shiloh, 1996).

The consequences of the women taking authority over the decision and yet wanting to share the responsibility with the men should be considered in the light of timing and age. Assuming that the men and the women have 'linked lives' (Elder, 1994) means that they each have to weigh up their own values of the costs and benefits from an interdependent perspective. For the men, there was the pressure from the women to agree to the decision sooner rather than later, a decision which was not an entirely attractive one that the men may regret in the future. However, the women appeared impatient to fulfil the desire to become a biological mother, a decision they had made when they were younger. In some ways this parallels the different ways in which men and women can be eager to become a parent generally. Townsend (2002) found that many women have the ability to separate out the decision to have a baby from their relationship and a family perspective, having made their decision early in life, whereas for many men the decision to have a child is part of being married, becoming a father and creating a family. Regardless of age, there is often an awareness of the biological clock ticking in the background for would-be parents (Rijken & Knijn, 2009). Consequently, these factors suggest that for the relationship to survive there is an urgent need for a modicum of 'give and take' to maintain the status quo. Considering that 'her decision is already made', it remains that if the relationship is important to the men, they will go along with the decision despite all. This could possibly set a benchmark for the dynamics regarding parenting overall for the future. Moreover, it appears that the men possibly carry the regrets and resentment of potentially being left to care for the baby silently, benefitting from not discussing the issue in order to maintain the status quo. These factors

resemble the patterns of behaviour in how partners generally have the ability to implicitly influence each other through latent power mechanisms unless one partner is willing to raise and discuss an issue (Hochschild, 1989; Komter, 1989; Rijken & Knijn, 2009).

It is important to note that the urgency of timing and age is common in all women who want to have a baby because, in contrast to males, they have to use different strategies to achieve the goal of having a baby due to their limited reproductive life (Buss & Schmidt, 1993). The focus on these critical factors for females exists simply to maximise the chances of reproductive success and survival (Hollway, Cooper, Johnston & Stevens, 2007).

### **MFS as strength and resilience / Comparison with past self**

There was a deep sense that the women adopted an attitude full of hopefulness and a willingness to cope with whatever the future may hold. They would literally 'Feel the Fear and Do it Anyway', (Jeffers, 2007). Certainly hope and fear do work alongside each other (Hunniche, 2009, p. 98). Hunniche's study looked at existential concerns in individuals with the progressive genetic condition Huntington's disease and found that in a life at risk 'there is a deeper dimension to the experiences of risk, chance, adversity, and fear.'(p. 98). Hunniche (2009) suggests that these dimensions of a life at risk contribute towards the emotional evaluations of the personal implications of the disease. She further suggests that 'there is a connection between hoping and fearing and making decisions' (p. 98). Interestingly, she draws on theories of science and philosophy to assess how people may react when

they feel overwhelmed by their circumstances. Rollo May (1967) considers hope and fear as he reflects on the insights of the philosopher Spinoza who argued that through using reason and courage we can overcome doubt. Zournazi, (2002, p. 245) gives an alternative argument, quoting the philosopher Isabelle Stengers, who describes hope as 'the difference between probability and possibility' and suggests that hope and fear are based on uncertainty. In other words, it could be said that the women can calculate the probability of risk in making the decision at any particular given time, but the possibility of risk cannot be calculated due to their ever-changing lifeworld, the life that is lived and experienced by them as an individual embedded in their environment.

The differing views of Spinoza and Stengers both offer a framework that may account for the women's readiness to go ahead with pregnancy and their perceived ability to cope with the practicalities of their situation. According to Spinoza's argument, the women certainly had the courage to pursue their longing for a biological child and, over time, had formed justifiable reasons for following it through. Stengers framework inviting us to consider the concepts of probability and possibility also supports the findings of the current research. It would seem that the women in their need and desire to become pregnant have weighed up the risks, considered the probability of any mishaps and concluded that the possibility of risk cannot be ruled out, thereby offering them the opportunity to proceed. These dual perspectives offer an explanation for the consistency and persistence revealed in the women's narratives.

It is thought that the women's experience of making the decision to become pregnant is significantly positive, but this does not negate concerns and issues regarding having children. There is still evidence of the women's awareness of the risks as they struggle to reach their goal of becoming pregnant and giving birth, but this is not as prominent as the positive attitudes of the women toward themselves, and what seems to be the strength and resilience that are common between the women. It is quite likely that these factors, together with their perceived ability to cope with the future, were important factors and essentially enabled them to challenge the negative aspects of making a decision. This strongly resonates with the findings of Peters et al (2002) where adult individuals with Marfan syndrome who scored highly on the Rosenberg Self-Esteem Scale demonstrating positive self-esteem, viewed Marfan syndrome as not particularly life-changing.

## **5.2 B. CREATING AN INFORMED DECISION**

### **She wants him to know the facts**

The women considered that sharing Marfan syndrome with their partners was important. They emphasised that it was crucial that their partners knew that they carried a genetic disorder and understood the meaning of Marfan syndrome as a potentially life-threatening condition before they committed to marriage. Despite the women sharing their family history, the men continued with the relationship, suggesting unconditional love and acceptance. Indeed, it could be considered that not sharing this information could have become a source of long-standing resentment. Moreover, it is rare for a marriage not to proceed when a potential partner becomes aware of a genetic condition

(Richards, 1999). In contrast to these findings Acharya, Lang & Ross (2009) found that women had great difficulty in discussing their genetic condition with their partners, because of the potential stigma or concerns that it assumed the relationship to be more serious. However, in the current research it is very clear from the women's narratives that the genetic implications of Marfan syndrome were discussed as the couples' relationships became more established. Furthermore, it could be argued that sharing their condition was symbolic in establishing a meaningful relationship. For some women being in a relationship with a genetically inherited condition can be a time of considerable concern and requires a sensitive, cautious and gradual approach to confronting the issue with their partner (Huniche, 2009).

### **Sharing responsibility for decision**

The women's experiences of sharing their condition and the choices regarding having children with their partner were by no means straightforward. The dilemma of making a joint decision is reflected in their conflicting needs (Huniche, 2009) in wanting to have a baby. The women want a natural child, but their partners are more concerned about the potential outcome. Achieving a joint decision is a complex multidimensional process encompassing differing views and variable consequences for each individual (Shiloh, 1999). As the driving force behind gathering information, the women played a key role in ensuring that their partners agreed to undertake joint responsibility for the decision. They provided opportunities to identify and explore their partners' thoughts and feelings, which encompassed the need to share and manage the process of collecting facts, figures and evidence from the various sources

available. This notion of the female partner taking a lead role has been described as 'genetic housekeeping' (Richards, 1999; Shakespeare, 1992; Tyler & Harper, 1983). In the present study it is considered that the women generally act as a genetic housekeeper in the relationship, initiating contact with others and encouraging their partners to understand Marfan syndrome and thereby create an informed decision.

However, despite the females' persistence in attempting to engage their partner's commitment to the decision-making process, there was a marked reluctance from the men to own their part in sharing the responsibility. It may be that the men adopt a relatively passive role due to their concern that over-emphasising the disadvantages of a natural pregnancy may worry their partners more or lead to negative effects on their relationship. This could be argued as consistent with the findings and reports of other studies, where even when men are affected with a genetic condition, it is the women who retain the role of genetic and emotional housekeepers (Shiloh, 1999; Snowdon & Green, 1994).

It is not easy to unravel the framework of responsibility in this theme. It seems as if the men changed over time in order to accommodate the women, in response to the women having already made their decision. It also appeared that the women resolutely continued in their plight to persuade the men to share the decision they had made alone. The notion of 'sharing responsibility for the decision' does not fall neatly into a conclusion as ultimately it could be considered that the men cooperated with the women.

The image I had of a bridge being co-created between them appeared to serve more as a compromise than a clear pathway forward. It could be argued that sharing the responsibility for the decision could be detrimental to the relationship as relationships can rely on the individual thoughts, feelings and behaviours when faced with challenging situations (Tran & Simpson, 2009).

Understanding the complexity of how relationships are maintained may shed some more light on this theme. It could be said that the couples had the common goal of wanting to stay together and to become parents. Research shows that this involves the crucial element of commitment in maintaining the relationship and adapting to the changing needs and goals within the relationship over time in order to keep the relationship going (Guerrero & Chavez, 2005). Considering that all the couples had been together for a minimum of seven years suggests that they had probably experienced a shift in their needs and goals as their relationship evolved.

The extensive research to date has demonstrated that there are two common factors that define whether couples stay together; the rewards from the relationship and the issues that discourage them from leaving (Kurdek, 2006). The women demonstrated that they wanted to claim their authority in making the decision to have a biological baby, but they also wanted the men to share the responsibility for that decision. The men agreed to go along with the decision to have a biological baby, but they did not agree to take responsibility for the decision due to the potential risks to their wife or child. Essentially,

they are both weighing up not only the rewards, but also the costs (Sedikides, Oliver & Campbell, 1994). The couples were in a well-established relationship and apparently there is a tendency for couples to become closer as the rewards and costs increase over time (Argyle, 1996).

The rewards for the women in making their decision included the experience of having a biological baby, becoming a mother and maintaining their relationship with their husband. The costs were the risks to themselves in producing that baby and to some degree the 50/50 risk that the baby would be born with Marfan. There was the also the chance that if they were not successful in achieving shared responsibility, the men might have felt resentful and blamed them for having made that decision. On the other hand, the rewards for the men were becoming a father and maintaining their relationship. It is quite likely that they also felt that there was compensation in safeguarding themselves against agreeing to share the responsibility for the decision. The costs were the 50/50 risk that their child would be born with Marfan and the possibility of there being complications in the pregnancy and birth. They had a common goal to become parents and maintain their relationship. However, the perceptions of the costs and consequences of attaining that goal were greater for the men inasmuch that they may have been 'left holding the baby' if their wife had been unable to care for that child, regardless of whether it had been born with or without Marfan syndrome.

### **Coping with doctors**

Associations with doctors, consultants and other medically trained staff were significant in coping with the multiple needs of the couples as they gathered medical information and advice. There were reports of understanding, kindly and informative doctors and consultants, but also evident from the data are the various difficulties the couples invariably encountered. This is demonstrated in their descriptions of the meetings as being quite frustrating for several reasons: having to educate the professionals due to their poverty of knowledge regarding Marfan syndrome; receiving conflicting information regarding natural pregnancy and childbirth; considering their needs and these not being met in the consultation; learning to develop strategies in order to communicate more effectively with the medical professionals. These findings are consistent with Budych, Helms & Schultz's (2012) qualitative research of patient-physician interaction with patients suffering from six rare diseases, including Marfan syndrome. They found that the expertise of many healthcare providers is insufficient and proves to be a major problem in what should be highly specialised treatment. The patient often had to become an expert in the field of their illness, and the interactions and patterns of communication in the consultations were inclined to be patient-directed, which had a major impact on the role of both the patients and physician. The traditional role of the physician, controlling decision-making processes and providing information has changed in recent years, resulting in a need for healthcare providers to acknowledge the patient as an informed, involved and interactive partner in the consultations. However, it is questionable whether a patient facing the challenges of Marfan syndrome or any other disease should have

to cope with the additional challenges of healthcare providers' poverty of understanding, or an insensitive approach to the individual needs of their patients.

Budysh et al's (2012) research, and the relevance of the data in the current study, suggest that there is clearly a need for improvement in this area, as the emotional impact and the amount and kind of information the couples need appear to be underestimated among the medical profession. Although I understand that it is impossible to have detailed knowledge of every illness, it seems that there is a need for all professionals to make every effort to educate themselves in the patient's presenting condition, whether a disease is common, rare or, as in the case of Marfan syndrome, a common-rare condition. An atmosphere that is tolerant of the different responses and reactions of patients, one that could sensitively establish the couples' preferences and accommodate their needs, may facilitate a more productive experience than the physician as a 'student' in Marfan syndrome or a perceived detached expert. Doctors and consultants play a vital role, as the couples are dependent on them at this time, and it is evident from the couples' narratives that, as patients, they were more inclined to be compliant and less inclined to be assertive at a time when assertiveness and expressing their needs would have been of considerable help.

There are, no doubt, many possible reasons for the couples' negative experiences in consultations. Carl and Cathy received conflicting advice from specialists in Marfan syndrome in seeking to make an informed decision, one

consultant advising them against becoming pregnant and another advising them to the contrary. There may be a problem concerning a mismatch between the perceptions of physicians, the women and their partners. De Bie et al (2004) found that Marfan syndrome patients perceive their condition differently from the way that professionals do. Patients remarkably underestimate life-threatening conditions, such as cardiac problems, if they have no physical evidence of the severity. This may illustrate a lack of communication in consultations or reflect patients' denial of the implications of their condition (De Bie et al, 2004; Grigoni et al, 2003).

### **The meaning of genetic counselling**

Further, also evident from the data, is how the couples' expectations are not fully met when they attend for genetic counselling appointments. They do not always understand the complex genetic concepts, or are unclear about the information they need, or there is a lack of emotional support. Belle and Adrian had requested a referral from their GP and described their one visit to help them make a decision whether or not to have a baby. Belle found the appointment fairly helpful, but would have appreciated some emotional support. This strongly resonates with the findings of Sorenson et al (1981); Kessler & Jacopini (1982); Bosk (1992); Clarke et al (2008). Adrian didn't feel included in the meeting and didn't understand the information the genetic doctor gave them.

The couples' narratives illustrate how each have their own agenda in the decision-making process. Given that the women aim to ensure their partners

know the facts and share responsibility for the decision, it is thought that professionals should assess both the women's and men's understanding of the condition as discrepancies are not uncommon (Gallo et al, 2009).

The genetic appointments were focused on giving medical information, and while this was acceptable for the participants, an opening to discuss the situation from an emotional perspective would have been helpful. Other research has found that the process of genetic counselling was essentially a clinical and routine process (Van Zuuren, 1997), which lacked an empathetic patient-led consultation (Smith et al, 2000). It is clear from the couples' narratives that they were not given the opportunity to discuss any concerns regarding emotional issues and would have welcomed a chance to do so. Despite research (Biasecker & Peters, 2001) highlighting patients' needs to voice emotional concerns, it appears these needs are not being met with regards to Marfan. Further, if the genetic counsellors or other genetic consultants were open to emotional issues, they could assume the role of gatekeepers in referring patients to the appropriate psychological services (Peters et al, 2002).

According to geneticists, couples with a Marfan diagnosis are not routinely referred to hospitals for genetic counselling when they are thinking about starting a family. In the current research, Belle and Adrian had to request a referral, it was not offered to Lorraine and Bob, and Carl and Cathy were unaware it even existed. It appears there are many gaps in this service, which is potentially a valuable resource for couples making a decision to have

a baby. There is a lack of research exploring the needs of individuals with genetic conditions (Chapman & Smith, 2002), which assesses their quality of life and their understanding of medical terminology (Genetic Interest Group, 1999).

In this light, it is not surprising that the experience of genetic counselling was perceived as not particularly useful and there was little evidence that it was influential in the decision-making process. It is likely that this was due to the women having made their decision or the couples being unsure about what to expect and uncertain how to prepare for the appointment. These latter points are consistent with the findings of others (Hallowell, Murton, Statham, Green & Richards, 1997; Bernhardt, Biesecker & Mastromarino, 2000; Macleod, Craufurd & Booth, 2002).

### **The role of the Internet**

Another source of information that came into play for the participants was the Internet. But they found most of the information 'quite scary', 'worrying' or 'upsetting'. It is clear that the Internet provides a wealth of information for those with genetic conditions, which ranges from simple explanations of a condition to quite complex accounts that would benefit from further clarification. While the Internet can serve a valuable educational and social role, it is apparent from the participants' accounts that talking to a person could provide a more balanced perspective. It is most probable that reading the facts about Marfan syndrome increases consciousness of the awareness of the risks involved and the women in particular did not want to be reminded

of the complications. Indeed, in browsing some of the web pages, it appears that many official websites tend to be quite depressing, symptom-specific and fail to emphasise the value of taking special precautions and careful monitoring in pregnancy, unlike the helpful websites and booklets provided by the Marfan Association UK and the Marfan Trust.

The quality of health information on the Internet is of concern, and research has identified that there is a considerable amount of unsatisfactory information, and little is known about how people use the information (Powell, Lowe, Griffiths, & Thorogood, 2005). People who turn to the Internet for information are generally aware that it may not be accurate (Ziebland, 2004), and although people report that they only search appropriate sources where the authorship is respectable and credible, in practice this is not always the case (Eysenbach & Kohler, 2002).

Another aspect of turning to the Internet for information may be as a result of the interaction with medical professionals, which as we have seen is less than satisfactory. Timmerman (2005) found that patients were given unclear or limited information from healthcare providers. Furthermore, many patients are dissatisfied with the information they receive in consultations (Starke & Moller, 2002), and turn to the Internet for information (Bernhardt & Felter, 2004). Given that there is an increasing demand for online health resources (Fox & Fallows, 2003; Cotton & Gupta, 2004; Gallo, Angst, Knafi, Hadley & Smith, 2005) there seems to be scant information and guidance on how internet

research and communication could be most efficacious for both patients and professionals.

Interestingly, it appears that the couples in their decision-making did not use Internet support groups or forums. This would suggest the very personal nature of the decision as one that belongs to the individuals and their relationship. Furthermore, discussing their situation or sharing the decision-making with others in chat rooms may expose them to not only positive and successful stories of childbirth, but also to any negative experiences or narratives. The role of internet support groups in problematic pregnancy has been found to be helpful for women in terms of sharing their experiences and understanding their emotional feelings with a like-minded community (Lowe, Powell, Griffiths, Thorogood, & Locock, 2009), but the findings in the current study suggest that it is doubtful whether a Web-based support group would be particularly valuable for couples in their decision-making. Internet support groups have been considered as not only beneficial but also harmful (Lowe et al, 2009). It would appear that the Internet has the potential to provide a source of beneficial psychoeducational and social benefits through Internet bulletin boards, video-conferencing and chat rooms (Troster, Paolo, Glatt, & Hubble, 1995), but needs to be treated with caution as the information is not necessarily factually accurate.

### **Exploring options**

In considering the negative aspects of natural pregnancy and childbirth the couples explored alternative options to have a child, such as adoption, in vitro

fertilisation (IVF), prenatal genetic diagnosis (PGD) and pre-implantation genetic diagnosis (PIGD). They unanimously agreed that they would not consider abortion if the foetus was affected by Marfan syndrome. Given that the women had already made their decision to have a child naturally, this latter point is hardly surprising and their partners agreed that they too would not consider termination. Therefore PGD was one option that could be ruled out of the decision-making, as there would be no need to have a prenatal diagnosis. The decision not to have PGD or terminate a child is understandable in the circumstances. The women have lived with Marfan syndrome for a long time and their partners have chosen to share their lives with them. To terminate an affected child may imply that people with Marfan syndrome are not acceptable or less than perfect. Moreover, termination of affected pregnancies could be viewed as discriminating against individuals living with a genetic condition (Chapman, 2002; Gillam, 1999; Kaplan, 1999).

The narratives as they discussed the process of exploring the different options illuminated a wide spectrum of thinking, ranging from stark reality of the risks of natural pregnancy and childbirth to the mother, to a mood portraying more upbeat hope and expectations of taking the natural route. It was an emotional time where the couples became more conscious of the dilemma of their situation and made a deliberate effort to consider and discuss the options. This increased awareness in reproductive decision-making has been documented in other research (Rapp, 2000; Peters, Kong, Hanslo & Biesecker, 2002b). Although the couples discussed the options, it is evident from the exchanges in the interviews that the women are exploring the various

alternatives in order to satisfy their partner as their stance remained on sharing responsibility for the decision.

Interestingly, Belle and Adrian's very 'live' discussion of currently making the decision was reflected in the accounts of the other couples as they recalled what it was like for them retrospectively. Considering that this was such an emotional time for the couples, it could be argued that there is a pressing need for psychological support at this time for the couples or as individuals.

### **Assessing the risks**

In considering the 50/50 risk of having a child with Marfan syndrome there were varying levels of concern between the males and the females. Having ruled out PGD and the choice of termination that would have introduced predictability into the decision-making, the couples assessed the risks of their child inheriting the Marfan gene before agreeing to proceed with a natural pregnancy.

In terms of the 50% risk of their child inheriting the Marfan gene, the males were the most cautious and spoke of the dilemma of making the decision to go ahead with a natural pregnancy. Throughout the interviews the males demonstrated their recurring attempts to make a decision in an analytic way of weighing up the pros and cons, while concurrently, on another level, the longing for a child within their relationship blurred their perception of the risks involved. It was impossible to calculate the right answer to the problem. As they employed the prospectively efficient strategies of weighing up the pros

and cons in attempting to create an informed decision, significant and powerful emotional signals changed their perspective and actively played a part in the decision-making process. There was a tension between needing to weigh up the risks involved and desiring the favourable conclusion of creating a biological family unit. This supports findings showing that although motivational reasoning is generally rational (Kunda, 1990), health issues present a different picture where an individual wrestles between seeking an accurate assessment of the risks and their desire to achieve a favourable goal (Croyle, 1992). Moreover, Timmerman (2005) suggests that people invariably find it difficult to understand the risks or the probabilities when making reproductive decisions and have inaccurate perceptions of the risks to be able to use them effectively in decision-making.

These findings are supported by research showing that, in decision-making with a genetic condition, people have an inclination to interpret a 50/50 risk as one that will result in a positive outcome for them as individuals (Parsons & Atkinson, 1992). Here, in the present research, there was evidence that the decision to have a baby could have been different if the risks were increased. This highlights the intricate nature of weighing up the risks involved, and resonates strongly with the research of Smith, Michie, Stephenson, & Quarrel (2002) who found that patients with Huntington's disease were inclined to favour an uneven risk in decision-making, and sought additional information in order to convert the risk accordingly.

Even though the women's experience of decision-making was significantly less of a dilemma, this did not negate the emotional rollercoaster of events leading up to the final decision. They shared information with their partners about the risks until they reached a 'tipping-point' whereby they passed the threshold of making the decision that having a baby naturally was the best choice for them. Despite acknowledging the risks, the women in particular were optimistic in the face of reality. There was a sense that the women wilfully ignored or successfully filtered out negative information in estimating the risks. It was as if they viewed the world through rose-coloured spectacles, creating an illusion whereby they could ingeniously ensure the survival of their longing for natural pregnancy and the birth of a child. In terms of the extant literature, these findings could be argued to be consistent with the conclusions of Smeltzer (2002) that women with multiple sclerosis proceeded to become pregnant, despite their fears of the risks of adverse effects in pregnancy and childbirth. Moreover, they continued to perceive their decision to become pregnant as risky both during the pregnancy and beyond. In addition to this, Kay & Kingston's (2002) qualitative investigation of women who were carriers of an X linked condition found that the women purposefully avoided overloading themselves with information to avoid a more difficult and burdensome decision-making process.

As the couples shared the decision-making process they eventually reached the consensus of a democratic decision, but it is clear from the findings that there are individual differences delineating the male and female perspectives in making that decision. These findings resonate strongly with Harman's

(1995) study, which reported male and female differences when making reproductive decisions from the perspective of gender. It could also be argued that the couples agreed on the riskier decision to have a biologically related child as opposed to a less risky option as a result of it being a joint decision and not an individual choice. This is consistent with the findings of Stoner (1961) who found that individuals making a decision on their own choose a less risky option with a secure reward, as opposed to choosing a more risky option even when it gives a possibly greater reward. Moreover, social and cultural influences cannot be ruled out of the risk assessment for the couples, as by nature we are sociocentric beings (Paul & Elder, 2002) wanting to conform to our lifeworld (Ashworth, 2003).

It is evident from the narratives that the decision-making process cannot be completely understood from the normative psychological theories and models of decision-making and risk perception. Much of the existing literature on decision-making gives a quantitative viewpoint on how individuals make a decision under uncertainty. The study of cognitive models of decision-making (Ranyard, Crozier & Svenson, 1997), heuristics, biases and probabilities (Kahneman, Slovic & Tversky, 1982) or measurements of the relationship between attitudes and behaviour (Ajzen & Fishbein, 1980) are inadequate explanations for the complex and multi-faceted predicament presented in the current study.

In order to understand further the processes involved in assessing the risks for the men and the women, the social intuitionist model (Haidt, 2001) may

offer an alternative explanation. It may be that the men as well as the women had already made an emotionally-driven decision. Then, following their decisions, they each assessed the risks by putting forward an argument to justify that decision. Understanding the risk assessment from this perspective, the couple's initial intuitive processes fundamentally had to run their course before they could reason with each other that the decision they had made was the right one (Haidt, 2007). The social intuitionist approach (Haidt, 2001) offers a plausible explanation for the couple's searching for evidence to convince each other that the individual decision they had made was the right one.

### **Optimism of medical advancements**

The role of optimism in the decision-making process has been isolated as a sub theme in this research due to its unanimous significance for the couples in making their decision. It is widely believed that medical advancements are a hugely important factor for individuals and families in making choices and coping with Marfan syndrome now and in the future. Exciting and encouraging fast moving developments such as identifying and mapping the Marfan gene, improved surgical techniques, more efficient diagnosis, prenatal and pre-implantation diagnosis mean that the perceived and actual risks associated with Marfan syndrome are reduced and the couples can view it as a non-threatening condition. Indeed, medical advancements in Marfan syndrome have resulted in many healthcare professionals viewing the condition as treatable (Peters, Horne, Kong, Francomano & Biesecker, 2002a). Furthermore, increased availability of treatments that improve quality

of life and longevity have been found to be an important feature of decision-making in people with sickle cell and thalassaemia disorders (Atkin, Ahmed, Hewison & Green, 2008). The couples, in particular the women, were empowered by the optimistic outlook for themselves and their children. The women considered the medical advancements that had taken place since they were children, and felt that these would progress at a similar rate over time. Individuals seek optimistic information to maximise certainty and to help them feel less vulnerable, thereby perceiving that something positive can be done, either now or in the future (Macleod, Craufurd & Booth, 2002).

### **5.3 C. FINDING PSYCHOSOCIAL SUPPORT**

#### **The effects of her mother's experience**

Aside from seeking helpful support from the medical profession, psychosocial support was important too, the former has been described as formal support (provided by healthcare professionals) and the latter as informal support (provided by family and friends) (Cutrona & Russell, 1990). The data in the present study suggest that social support came from limited sources as they were making their decision, but of interest was the women's mother's experience of coping with Marfan syndrome in their children, which had a significant effect on the ease of the couples' decision-making. Two of the women found it difficult to talk to their mothers about having children. Cathy didn't discuss it with her mother, as she didn't want to cause her any unnecessary extra worry, whereas Belle's mother did not approve of her even becoming pregnant. The other four couples appreciated and welcomed the support that the mothers offered.

The difference in their mother's attitudes may be explained by their mother's own self-esteem, perceived levels of stress and generally the quality of experience in coping with Marfan syndrome in their daughter or other members of the family (Cohen, Hettler & Park, 1997). These details are not documented in the current study, but parents' lives are touched by a child's chronic illness and their style of parenting can be influenced by a wide range of emotions associated with their child's condition, such as guilt, helplessness, anxiety, shame, anger and low-mood (Pitts & Phillips, 1998). Parents who cope effectively with such emotions have been found to have a positive impact on how the children cope themselves (Mattsson, 1972).

It is not surprising that four of the couples' narratives reveal that it is the women's mothers who have a supportive role in the couples' decision-making. Generally, females have more access to information about emotions as mothers are inclined to discuss emotional feelings with their daughters, but not so much with their sons (Goleman, 1996). As a result, males have a tendency to become unconscious of emotional states in both themselves and others (Brody & Hall, 1993). Historically, mothers traditionally are expected to take primary responsibility for the care of children, the home, and their husbands, and later in life their own ageing parents (McGoldrick, 1999). Despite society challenging this traditional role, the rapidly changing expectations of women's responsibility in family life is at odds with the reality and their role remains in many ways the same (Carter, 1996; Garcia Coll, Surrey & Weingarten, 1998).

Interestingly, when Belle discussed her mother's disapproval of her becoming pregnant it had quite a significant effect on her mood in the interview. She became quite low and thoughtful for a while, demonstrating that her mother's adverse reaction to her becoming pregnant presented a further dilemma. Cathy found that she had to be stoic and not share the experience with her mother, as it would worry her unnecessarily. Additionally, it is thought that if Cathy did share her decision-making she would then have to cope with her mother's worries and concerns. This finding is supported by research showing that during personal trauma, such as coping with a life-threatening illness, it is not always easy coping with the reactions of relatives (Hinchliff, 2006; Haber, 1997), and in particular for couples coping with a genetic condition, during the reproductive decision-making process (Frets, Duivenvoorden, Verhage, Peters-Romeyn & Niermeijer, 1991; Lippmann-Hand & Fraser, 1979; Rangell, 1971). The findings in the current study strongly suggest that the support and reactions of not only mothers, but also other close family members are an important point for professionals to factor in when considering the care and support needs of couples as they go through the experience of decision-making to have a child. Furthermore, the adverse reactions of close relatives can cause persistent reproductive uncertainty and doubts both during and after decision-making (Frets et al, 1991).

### **The strength of the relationship**

There is a very real sense of the empowering dimension in the strength of the couples' relationships with each other prevailing throughout the data. There is

evidence on the one hand that the men viewed the women as their wives, not defining them as a woman with Marfan syndrome, and also what is clear from their accounts is that the decision to have a baby is both desired and feared. It could be argued that to experience such tensions and contradictions the strength of the relationship is fundamental. The importance of this relational aspect of the decision-making is evident from the narratives. This finding is consistent with research that has shown that a supportive marriage is an important contributor to positive psychological health (Beach, Fincham, Katz, & Bradbury, 1996; Dunkel-Schetter, Sagrestano, Feldman, & Killingsworth, 1996). Also Peters et al (2002b) found a correlation between marital satisfaction and psychological well-being in individuals with Marfan syndrome.

Although it is considered that their experience of making the decision is embedded in their lifeworld, it is the strength of their relationship with their partners that is crucial at this time. Making the decision intensified the relationship and, in many ways, they isolated themselves from the external social realm of their lifeworld. This latter point resonates strongly with Carter's (1999) suggestion that when couples are experiencing the fear of infertility they isolate themselves socially. This is also consonant with the research suggesting that a potentially life-threatening event for a female should always be considered as a family event, especially in the context of her relationship with her partner (Reibstein, 2004; Walsh, 1998)

It is evident from the couples' accounts that their negotiations in making a decision are complex, intricate and inextricably intertwined with the way in

which they relate to themselves as an individual through their relationship with each other. Tantam and Van Deurzen (2005) suggest that we need a better understanding of how couples relate to each other, 'Research needs to be carried out on how personal and interpersonal disposition, mood and decision-making interact with people's values and beliefs' (p. 132). It is thought that the value of the current study is enhanced through the phenomenological and hermeneutic nature of analysis in line with IPA. Furthermore, an approach that uses an effective phenomenological methodology has the potential to emphasise 'the complexity, ambiguity and ambivalence of participants' experiences' (Finlay, 2008, p. 7). Analysis of the couples' narratives in the present research could be argued to demonstrate such complexities, ambiguities and ambivalence in suggesting an underlying struggle in the balance of power within the relationship. For example, the women have made their minds up to have a biologically related child, and they have to go through the process of convincing their partners that this is the right decision. The men, in turn, have to decide whether or not they agree to father a child under the circumstances. Thereby to achieve a biologically related child it has to be a joint decision by force of nature! The balance of power waxes and wanes as the couples strive to reach an acceptable conclusion to their decision-making for both the individuals and their relationship. It could be said that they have a foot in both worlds, the self as an individual and the self in the relationship.

One theory that might account for this is the dynamics of decision-making within a couple's relationship when they are coping with male infertility. This

is a perspective that proposes that, even when it is the male who is infertile, it is the woman who facilitates the decision-making (Lorber & Bandlamudi, 1993). But without him she cannot use his sperm or make other choices to conceive, therefore her power to negotiate is perhaps decreased (Rosen, 1999). However, it may be that it is the men who perceive the women to have all the power, the one who wants the child, and therefore it is possible that they might consider their male role as the one which is willing to cooperate (Rosen, 1999).

### **Temporary parallel lives**

Interestingly, analysis of the data revealed that at times the couples needed to lead temporary parallel lives and cope in their own individual worlds. This was one commonality in the couples' experiences that clearly demonstrated both their strengths and vulnerabilities in the decision-making process. They were reluctant to share their concerns, suggesting that temporarily withdrawing was one strategy that would protect their partners from additional problems, and thereby ensure the survival of their relationship and the goal of a biologically related child. This modicum of independence could be explained in general terms by women's ability to be necessarily emotionally self-sufficient within a marriage, whereas males are considered to need the emotional support of their wives (Bernard, 1982). This finding has important implications for the men's needs for psychological support at this time.

Furthermore, although the couples' accounts demonstrate that there are lulls in their communication, these are fleeting and in the background of an

otherwise seemingly successful and communicative relationship. Patterns of parallel verbal and non-verbal communication in a marriage regarding assumptions of oneself and ones partner have been shown to affect the health of a relationship (Beck, 1988). However, one speculative hypothesis could be that the temporary parallel lives serve to establish and maintain an optimistic outlook in the face of coping with the dilemma of their situation. There is a striking parallel here with research that considers the role of hope and optimism in harnessing emotions that are perceived to be out of control (e.g. Seligman, 1991; Whalen, Henker, O'Neil, Hollingshead, Holman & Moore, 1994). While Seligman (1991) found that an optimistic outlook was an encouraging feature when faced with a dispiriting situation, Whalen et al (1994) found that optimism in individuals generates hope that everything will be alright in the end, despite all the challenges.

Moreover, in psychoanalytic thinking such as that of Winnicott (1975) we have been taught that we can be alone when it comes to providing the comfort essential to our emotional needs. Indeed, simply finding a sense of intimacy in just being together or apart is thought to be an essential part of the marital relationship. But nevertheless, the couples' accounts in the present study suggest that although both males and females are aware of their partner's emotions, the signals are possibly insufficient to openly discuss the issues that are concerning them. One reason for this lack of sensitivity to their partners' needs, in particular for the men, may be their lack of skill when it comes to reading their partner's visual cues of emotion. Women are more sensitive and adept at reading visual emotional cues in a man (Sternberg,

1988), but this does not fully explain the dynamic from the women's perspective or, indeed, why the couples need to temporarily withdraw and lead parallel lives.

The difference in perspectives and the conjoint decision may represent too large a threat to the status quo of the couple. The decision could be said to be contentious, thereby creating contradictions that might threaten the coupledness. Sweet denial and disconnecting from Marfan syndrome may probably be the best socio-emotional glue that hangs them together as a couple. Parking the decision to one side and leaving it in the background may be the best strategy for keeping them together as a couple. Possibly, leading separate lives conserves the energy needed in the relationship at this time. Ultimately, it might be said that it is essentially functional, in that it sustains the couple and thereby does not pose a threat to the bond between them.

One framework that might account for this change in their dynamics is cognitive dissonance theory (Festinger, 1957). This is a perspective proposing that individuals desire and seek cognitive balance. The women in the current study have already made their minds up to have a child despite the risks to her own life and their child. The women therefore hold two psychologically opposing views: one: 'I am going to have a biologically related child.' and two: 'I will have to cope with the risks'. The emotional discomfort of coping with these two opposing cognitions leads her to irrationally alter her beliefs and avoid any exposure to information that is inconsistent with her

positive decision to have a baby. She therefore withdraws from discussing the situation, thereby maintaining a manageable psychological state.

In addition to this, as discussed earlier, the data in the present study indicates that the women had already decided to have a biologically related baby, and the men are striving to make sense of the situation, both of which would entail an element of independent planning in the relationship. Street (2006) suggests that when people are planning a life-event they have a need to psychologically withdraw from the relationship, allowing time for reflexivity. This temporarily removes any external input and the individual can then focus on their particular viewpoint and thus create a plan of action. There is no doubt that, in becoming a couple, individuals have to readjust to define themselves (Gergen, 1991). Choosing to have a child is a major event and the couples have to change and develop their relationship in order to respond to their circumstances (Street, 2006).

Perhaps a combination of tactics and strategies are at play here, and all are worthy of consideration. Perhaps the women simply needed to withdraw to focus on mastering the challenging and changing demands of pregnancy and childbirth. Maybe the prospect of having a biologically related baby motivates the women to such an extent that in the passionate desire to procreate there is no need for the other. As in the cognitive dissonance theory mentioned earlier in this section, the distance may serve to maintain their desire and protect them from exposure to their partner's rational perspective. The men may well respond to this by maintaining their own distance.

### **Limitations of friends' support**

Another aspect of the couples' experiences that emerged from the data was that friends could only offer a limited amount of support. Their accounts illustrate some of the difficulties of sharing decision-making and Marfan syndrome in general with friends. It is clear in reading the accounts that talking to friends and considering their input would be an additional factor if they shared it with them. Besides the finding that they wanted to make the decision as a couple, talking to friends proved to be more of a psychological and emotional hindrance rather than a support. Friends found it difficult to understand Marfan syndrome and there was their perceived expectation to have children as a natural part of their relationship as a couple. The couples' tendency not to share the experience with friends may be accounted for through research investigating why social support can be problematic in coping with a medical condition.

Individuals with Marfan syndrome restrict talking about their condition to their partner, parents, close relatives or friends (De Bie, De Paepe, Delvaux, Davies, & Hennekam, 2004). Although the research findings of Peters, Kong, Hanslo, & Biesecker (2002b) highlighted that patients with Marfan syndrome generally had a positive attitude towards themselves, other research has demonstrated that some individuals withdraw from certain social situations, choose to keep their condition from friends, and have to educate others about Marfan syndrome in order to avoid feelings of perceived stigma (Peters, Apse, Blackford, McHugh, Michalic, & Biesecker, 2005). Having positive and

favourable feelings about the condition correlates with an individual's perception of available social support (Peters & Pettrill, 2011).

Social stigma is indeed an issue that is suggested from the data in the current study, but this is more in connection with the pressure to conform to societal expectations that they will have children. Belle and Adrian feel the odd ones out when they meet with their friends who have children, and perhaps the decision not to share Marfan syndrome and their decision-making is part of the perceived stigma reported by Peters et al (2005). Certainly, individuals do use social comparison with others they know socially in defining themselves (Strahan, Wilson, Cressman, & Buote, 2006). A condition such as Marfan syndrome could be said to give individuals a heightened awareness of themselves physically (Radley, 2000), and, moreover, comparison to others they perceive to be more fortunate than them tends to negatively effect self-esteem and mood (Wheeler & Miyake, 1992). I would argue that this is consistent with the women's accounts in the present study that demonstrate the urgency of making a decision with their physical and biological clock ticking away. They demonstrated a desperate need to conceive and avoid the social onus of childlessness. Nature is a powerful determinant of behaviour, but societal expectations have a strong influence on our emotions.

### **The role of support groups**

Marfan support groups were deemed as either helpful for the couples who had attended them or potentially helpful for those who hadn't. Interestingly, research on support groups has demonstrated a negative picture of their

efficacy. De Bie et al (2004) found that talking about Marfan syndrome with other patients or outsiders was reported as difficult or non-existent. They suggested that support groups needed to foster a more positive approach towards Marfan syndrome, and thereby a more productive environment that would facilitate psychosocial well-being. Likewise, the experience of support groups has been reported as unhelpful in coping and adapting to breast cancer, defining it as an additional social or psychological problem (Hinchliff, 2006). Peters & Petrill (2011) found that patients with Marfan syndrome were more likely to seek and consider support groups helpful if they felt uncertain or had negative feelings about their condition. Considering that Belle and Adrian feel 'isolated' in their decision-making, and Theresa and Tom feel so strongly that there is a need for a support group that they themselves would have appreciated at this time, it is surprising that this fundamental source of support is scarce. Particularly when Carl and Cathy found that sharing their decision-making with a woman at a Marfan support group meeting who had been through the experience successfully, was invaluable in finding out everything they needed to know. These findings clearly have important universal implications for the social needs of people with Marfan syndrome and other genetic disorders in coping with reproductive decision-making.

### **Psychological counselling as potentially helpful**

The findings suggest that the men would benefit from emotional support. It may be that formal group psychotherapy would be helpful for the men to attend. Group psychotherapy has been found to be psychoeducational and supportive for patients and their families in coping with health-related issues

(McRae & Smith, 1998; Lorig, Holman, Sobel, Laurent, Gonzales, & Minor, 1993), offering a space to express emotions (Forester, Kornfield, Fleiss, & Thompson, 1993). It is evident from the narratives that psychological support would be helpful for the men as they wrestled with the dilemma of the decision and beyond, but professional psychological help was not forthcoming. It may be that a more informal support group shared with other couples going through the decision-making process, or couples who had experienced the process in the past, would be more effective in providing support for couples.

Although Theresa and Tom didn't feel the need for outside help, the other couples' accounts reveal that, in hindsight, psychological counselling might have been beneficial for the men. There is evidence on the one hand that some participants, in particular the men, struggle with the confusion in coping with the interface of assimilating the medical facts alongside their emotional reactions to the situation. It is also very clear from the narratives that the women display a stoic coping ability. It could be argued that the women's stoic approach serves to reduce vulnerability by masking any chinks in their armour, and thereby establishing a firm sense of their ability to cope efficiently with the risks of pregnancy and childbirth. Further, the women's stoic approach resonates strongly with Lewis-Herman's (2001) suggestion that '...survivors must invent their own methods, drawing on their individual strengths and the supportive relationships naturally available to them in their communities' (p. 241).

It is apparent from the findings that there are generally both psychological and emotional problems in coping with the decision to have a baby. The narratives highlight that the couples not only have to cope with the medical aspects and associated risks, but also the psychological turmoil throughout the process.

Considering that research has advocated the importance of social and professional support in alleviating or preventing depression and anxiety (De Bie et al, 2004), and that a lack of on-going supportive counselling is associated with problems in the reproductive decision-making process (Frets et al, 1991), it is remarkable that in practice there is a poverty of such services. This especially, when the impact of psychological counselling has been found to be an important intervention for couples' relationships when coping with an illness (McRae & Smith, 1998), and specifically for young adults between 16 and 35 years of age diagnosed with Marfan syndrome (Van Tongerloo & De Paepe, 1998).

### **The interview as a therapeutic experience**

Interestingly, the couples reported that the experience of the research interview gave them a welcome opportunity to talk about what it was like for them going through the decision-making. The interview could be described as therapeutic for the participants, as it was evident that they shared certain information that they hadn't necessarily discussed within their relationship. Both male and female participants were quite tearful at times as they discovered that there were more emotional aspects of the decision-making

than they had admitted to themselves or each other at the time. The process of the interview generated a significant volume of disclosure at the interpersonal level. It emerged that an intermediate in the room was a catalyst for effective communication with each other.

This finding has important implications for all researchers studying couples and their close relationships, and provides support for a longitudinal study of dating couples whereby the couples researcher unwittingly had the effect of either strengthening a couple's relationship or, in some cases, facilitating its dissolution (Rubin & Mitchell, 1976). They acknowledged that this raises important methodological and ethical implications for couples' research, while offering useful implications for the practice of couples counselling.

These findings strongly suggest that there is an overwhelming argument for the provision of psychological services alongside the general medical teams. Counselling for couples or individuals, in particular for the men, should be made available throughout the decision-making process and beyond. To the best of my knowledge, this is not the case and there are no indications as to how many psychologists are involved in the care of patients with genetic concerns. Shiloh (1996) has suggested that there is a pressing need for psychologists within the field of genetic counselling, to not only provide psychological counselling to patients, but also to train and consult with genetic counsellors, and carry out genetic counselling research.

#### **5.4 D. EXISTING WITH THE FALLOUT OF THE DECISION**

The participants' accounts of the process of decision-making demonstrate the enduring complexity and fundamental multiple dimensions of the lived experience for them as individuals and as a couple within their relationship. Therefore the next section focuses on specific psychological, emotional and practical issues in the decision-making and, in particular, on existing with the fallout of making the decision to have a biologically related baby.

##### **Coping with high-risk pregnancy**

Aside from the process of assessing the risks, the couples talked about coping with a high-risk pregnancy. Evident from the narratives is the extent of monitoring of the pregnancy from the outset and the impact of living day-to-day with a high-risk pregnancy. Increased awareness and the practical realities of becoming pregnant permeated the course of events from pre-conception to birth and beyond. These factors tended to diminish the spontaneity and joy of the experience of natural pregnancy and childbirth. Shiloh (1996) suggests that a heightened awareness of having to make a decision to have a baby when an individual has a genetic condition might also cause an increase in psychological problems.

The data in the current study demonstrates that the practical reality and proof of the risks affected each of the participants, causing degrees of anxiety at varying times. It is uncertain whether the variable extent of the anxiety reflects denial, perception of risk or a lack of awareness at any given time.

However, the couples' accounts revealed a continuum of concerns relating to conception, pregnancy and childbirth.

In particular, anxiety-related events even include planning conception, with Belle and Adrian likening it to infertility. This suggests that in planning to conceive with the knowledge of a high-risk pregnancy, cognitive processes that may affect mood and functioning are involved. Marfan syndrome has been found to have a negative effect on sexual relationships (De Bie et al, 2004; Peters et al, 2002b), but these studies are mainly related to negative body image and lack of sex drive. To the best of my knowledge there are no studies specifically aimed at the cause-effect relationship between awareness of a high-risk pregnancy and sexual functioning.

However, the narratives of the couples throughout the data imply there might be some parallels with the factors relating to infertility. These include the women's urgency to become pregnant and couple relationship problems in the light of a medicalised perspective on pregnancy. A sense of urgency can seriously impact on a couple's intimacy and sexual fulfilment (Robinson Kurpui & Maresh, 1998). The couple's relationship may be tested as they cope with the emotional and physical drain associated with pressures to conceive (Covington, 1988). Therefore, it could be said that the demands of coping with a high-risk pregnancy have far reaching consequences and implications for the psychological health of the couples, not only during decision-making, but also as they attempt to conceive.

### **Her concerns for child**

There is a marked distinction between the male and female perspectives of the risks involved. Namely, the women are altruistically more concerned for their child's well-being, whereas the men's accounts emphasise their fear of losing their wives and having to cope if they were left to care for the baby. These distinctive differences heralded the separateness of the decision as individual versus joint, thereby minimising coupleness and maximising individuality. The parallel lives discussed earlier, re-establish who the men and the women are as individuals. Sharing the profound differences in thinking could possibly stir up doubts about the decision, and the benefits of putting doubts to one side are clearly demonstrated within the context of the relationship. Furthermore, as the data suggests, the couples do not share the same concerns regarding the pregnancy. From the outset of decision-making it is evident that, while the men's agenda is more focused on the fear of potential loss of their wife, the women are preoccupied with the survival of their child.

The image of mother and child is a familiar one, with its roots steeped in history. A special prenatal bond can exist between a mother and her much wanted unborn child, although this is not always the case. However, it could be argued that the women in the current study are preoccupied with pregnancy and childbirth, as it defines them as a female, suggesting the importance of acknowledging the embodied nature of the experience. Finlay (2003a) argues that such an experience is both subjective (reflective) and objective, and therefore cannot be separated from an individual's lifeworld, the

lived and relational world. The women's accounts acknowledge the importance of their mothers' and their partners' support, but it is evident that there is a powerful and fundamental emotional attachment between the women and their unborn child. The women do not consider the risks to themselves; the health, safety and security of the baby are their priority. There is a striking parallel here with the findings of MacDonald, & Jonas-Simpson (2009) where women with a high-risk pregnancy were positive, hopeful, remained connected with their unborn children, placed minimum emphasis on their own well-being, and were steadfast in their resolve that the pregnancy should focus on the safety of the baby.

Understanding a high-risk pregnancy for women with Marfan syndrome entails expert medical advice, but the bodily experience of pregnancy could be considered as a valuable source of embodied knowledge and information (Lowe, 2005). The data in the current study suggests that the meaning of pregnancy for the women and their preoccupation with the survival of their child is at odds with the priorities of the world around them. This resonates strongly with Stainton's (1992) research, revealing that healthcare professionals focused on the medical aspects of a woman's high-risk pregnancy, whereas the women were more preoccupied with becoming a mother, and moreover disliked being considered as high-risk. If this is the case for women with high-risk reproductive choices, the subjective meaning of a high-risk pregnancy for a woman could be valuable in understanding their experiences. Further research is needed to expand our understanding of a mother's embodied experience of a high-risk pregnancy to identify and meet

her needs (Stainton, 1992), as traditional sources of professional and social support are often insufficient in meeting the educational or emotional needs of women with a high-risk pregnancy (Lowe, 2005).

### **He fears losing her**

In stark contrast, the men's narratives demonstrate their fears and concerns of losing their wives as a result of a high-risk pregnancy. There is evidence on the one hand that the men are concerned with the emotional consequences of coping with an untimely loss, but it is also very clear, on another dimension, that the meaning of losing their wives has a practical significance: the prospect of having to look after the baby. The men's accounts could be argued to challenge the notion of changing gender roles of men and women in today's society, suggesting that little has changed regarding the societal expectations of males and females within the transition to parenthood. Although men do show a natural ability to become a mother figure to children in the absence of women, this relies on how competent they consider themselves to be in undertaking the task (Gerson, 1993). However, the data in the present study strongly suggests that women are still considered to be the primary caregivers, and that traditional roles are embedded in our cognitions and emotions (Carter, 1999).

The importance of recognising a man's fears of losing the woman he loves in pregnancy or childbirth is not to be underestimated. In Western society, the typical masculine role is still evident on many levels and in particular in connection with fatherhood (Hadley & Hanley, 2011). The values associated

with male identity still encompass virility, bravery, rationality, objectivity, emotional distance, macho characteristics and control (Nelson-Jones, 2006). It may be that the societal norms and expectations of the male as 'protector' project an anticipated fear of failure or helplessness as he faces the possibility of coping after the loss of his wife, the woman he loves. Interestingly, research has shown that males experience the same emotions as women, but the opportunities to access and verbalise their thoughts and feelings are limited (Wong & Rochlen, 2005). This suggests that appropriate referrals and resources are absent from current service provision.

### **He copes and cries alone**

Adrian's concerns as he and Belle embark upon pregnancy are well-founded, as they echo the accounts of the other men. From the beginning, the men's narratives demonstrate that, for them, agreeing to the decision to have a baby entails coping with a high-risk pregnancy, and that means the prospect of loss. The emotional factors of losing their wives and thereby their relationship, cannot be separated from the emotional and practical implications of caring for their child. The men bottled up their emotions and did not share their feelings with their wives. The majority of the men admitted to crying when they were on their own. Coping with the situation was an ever-present demanding process where verbalising their emotions was not considered by the men to be appropriate. Besides, males are thought to be adept at concealing their emotions and generally it is more difficult to read emotional feelings from a man's face than a woman's (Brody & Hall, 1993).

The experience of pregnancy for any first-time father can cause psychological distress (Condon, Boyce, & Corkindale, 2004) and can be a trigger for depression and anxiety (Boyce, Condon, Barton, & Corkindale, 2007), but in particular, high-risk pregnancy has been shown to lead to feelings of uncertainty and stress in fathers, to the extent that it overshadowed the experience of pregnancy (McCain & Deatruck, 1994). In the present study there are, no doubt, many possible reasons for the men's distress. One explanation might be that their marital relationship has been significantly changing during the decision-making to become pregnant, and in many ways, the men have lost a certain amount of companionship within the coupledom by leading temporary separate lives. The men's narratives demonstrate awareness that there is a third party from day one of the decision-making, the unborn child, in the relationship. Moreover, it is one that is forming a strong bond with their wives. This in turn may lead the men to feel left-out and possibly redundant. These findings resonate with Boyce et al's (2007) questionnaire study of first-time fathers, where it was found that participants suffered considerable amounts of distress when struggling with certain aspects of becoming an expectant father. In the current study, it could be argued that the men's identity and lifestyle are challenged from the first moment of decision-making to have a baby, especially when a high-risk pregnancy requires such an intensive level of professional care.

These findings, highlighting the striking differences between the men and women, hold important implications for the psychological care of couples within a wide range of healthcare settings. It is only too easy to place males

and females into a particular stereotypical biologically or socially determined domain, but to do so ignores the role of free will. The majority of children develop a strong sense of male or female identity from an early age (Smith, Nolen-Hoeksema, Fredrickson & Loftus, 2003), but gender definition is a complex process and there is much ongoing debate regarding the roles of nature versus nurture. The men in the current study acknowledged the pressures of decision-making, demonstrating their vulnerability. Interestingly, research suggests that even before social interactions, infant males reveal more vulnerability than females (Kraemer, 2000). But the societal expectations of men as the strong, tough, self-reliant sex are prescriptive, and both men and boys are pressurised into behaving accordingly (Courtenay, 2000; Martin, 1994). Most importantly, there must be caution in making stereotypical assumptions regarding the emotional needs of males and females in coping with decision-making to have a baby with a genetic condition.

### **Feeling responsible, selfish, guilty**

Both the men and the women communicated that at times they felt responsible, selfish and guilty. It was apparent from the women's narratives that their history of Marfan syndrome, the relationship with their partners and passing on the gene held great significance in all three emotions. Although it is clear from the women's accounts that they endeavoured to create an environment of joint informed decision-making and shared responsibility, the couples' accounts reflect the women as sometimes less positive, and maybe protective, towards their partners and the proposed child. One reason for this

could be that it is perhaps related to the women's perceived control over the decision. It is possible that following the initial tendency to underestimate the risks and thereby enhance the goal of a baby, elements of responsibility become burdensome. Reproductive decision-making in the context of genetic risk emphasises the freedom to choose, but at the cost of responsibility and guilt (Miller, 1983; Lippman, 1992). Rodriguez (1998) highlights that healthcare providers need to be sensitive in genetic reproductive decision-making to the changing attitudes and emotions over time of individuals towards issues such as guilt, anxiety, desire to have children and taking risks.

There were interesting differences in the men's narratives. Although their instincts were to have one biologically related child despite feeling selfish and guilty, feelings of responsibility were largely focused towards their wives. They feel responsible for not only agreeing to the risk of having a child with Marfan syndrome, but also placing their partner's life at risk. However, there was a notable difference between Carl's account and that of the other individuals. Carl acknowledges responsibility and guilt for the decision, as the reality and meaning of risk to his wife emerges, but interestingly he feels selfish for *not* taking responsibility for passing the Marfan gene to his child, regarding it as the child's potential problem and not his. It is difficult to speculate on the reason for this difference between Carl and the other men, but it could well be related to denial. Perhaps more detail relating to the quality of his relationship with his partner and his personal history may reveal more reasons that could account for this factor.

Notwithstanding these overall differences, it appears that as the decision is being made, associated feelings of responsibility, guilt, or selfishness are assimilated without long-standing detriment to their marital relationship. It is possible that the women's personal experience of living with the condition strongly influenced their relatively high levels of responsibility and guilt, but conversely at the same time reduced these emotions to manageable rational proportions. This may be, as Beeson & Goldbus (1985) suggest, that women with personal experience of a genetic condition are more concrete in their thinking in considering having a child at risk of inheriting the condition than women who have not had this experience. On the other hand, the men's emotional feelings indicate that not having prior experience of Marfan syndrome is more confusing in the fallout of the decision-making process, and thereby they demonstrate vagueness in sharing the responsibility. This is further complicated with guilt at putting their partner's life at risk, the responsibility of agreeing to the decision and apprehension about the burden of having to care for a child. This latter point could be described as selfish doubt. This theory therefore suggests that the women's motives for feeling responsible, guilty, and selfish in the fallout of the decision, differ significantly from the male's motives. An individual's emotional response to a given risk can vary enormously. The responsibility of a situation may appear daunting to one person but inspiring to another. Carrying a genetic condition can mean tragedy or challenge (Fraser, 1974).

It is important to note that the feelings of responsibility, selfishness and guilt for the women may have more universal implications than just specifically for

women with Marfan. Generally, some women can feel the desire to have a biologically related child more than some men, and the investment a woman makes in conceiving and giving birth are considerably greater than that of a male whose role in the process could potentially end following conception (Buss & Schmidt, 1993). Having established a long-term relationship, the women in the current research may feel responsible and guilty that they have made the decision despite the risks, and that their partners have gone along with their decision.

### **Acceptance of gamble, uncertainty and fate**

The couples' accounts clearly demonstrate that they are prepared to accept the challenge of the 50/50 gamble and, embracing uncertainty, leave the prospective outcome to fate. However, the men's narratives, in particular, clearly reveal that in the absence of finding an optimal solution they are faced with the uncertainty of worst-case scenario. They have both assessed the risks and, regardless of the potential problematic consequences, agree to take a leap into the unknown.

Rationally, there are the multiple risks involved, but emotionally the overwhelming desire to have a biologically related child dominates for the women. In Platonic thinking the rational mind should take over here, but the women's emotional longing overrides reason. The women rely more on trusting their instincts and therefore take an emotional short cut to making the decision. While the men also demonstrate an emotional motive, they are rather more cautious in their decision-making. However, the men agree to the

women's predetermined decision, which may tell us more about their desire to maintain the relationship than the decision-making process.

Following on from these last points, a further exploration of the gender differences from a social intuitionist perspective (Haidt, 2001) might offer some additional explanations of the dynamics involved in their decision-making, prior to their acceptance of what lay ahead. Both the males and females appear to invest a considerable amount of effort in persuading the other that their decision is correct and it could be said that their motives were selfishly led. Considering that the social intuitionism model encompasses intuition, reasoning and a social component to reach a morally satisfactory decision, (Haidt & Kesebir, 2010) suggests that there are gender variations in that process. Many of the studies of gender and emotions rely on self-reported responses and are therefore vulnerable to biases such as stereotypes and retrospective data (Gross, 2007). When exposed to an emotionally-laden experience, women show more response in the amygdala, the area in the brain associated with emotions, than men. In re-evaluating an emotional experience, men show less increase in the prefrontal areas of the brain, associated with cognitive reassessment, suggesting that they need to use less effort than women and their emotions appear to be automatically regulated (McRae, Ochsner, Mauss, Gabrieli & Gross, 2008). Therefore, in the light of social intuitionism, the women's initial emotionally-driven decision appears to require more effort to rationalise the situation than the men's, from a social and cognitive reasoning perspective. The findings from the current study suggest that the women's emotional drive to have a biological child was

greater than the men's, whereas the men focussed less on the emotions and presented what could be interpreted as a more rational perspective.

The outcome eventually reaches the same conclusion of facing the uncertainty of naturally conceiving a child. As the philosopher and psychologist William James (1950) proposed, decisiveness requires both rational and emotional qualities, but successful decision-making involves the skill of trusting the right one.

Nevertheless, the decision could be described as a 'non-decision' (Lippman-Hand & Fraser, 1979) as they have ultimately chosen not to choose by accepting fate. After all, having ruled out alternative methods of having a child such as adoption, making the decision not to have a child as nature intended would involve not only the emotional consequences, but also the social stigma of being childless. Childlessness or infertility has been shown to be as distressing as the death of a child or spouse (Kedem, Mikulincer, Nathanson & Bartov, 1990). The data in the current study suggest that it was not the rational information gathering at genetic counselling that influenced their decision, but the role of optimism and feeling hopeful alongside acceptance of fear that was a major factor in the final choice of which path to take. These findings are commensurate with research that has found that genetic counselling does not eliminate reproductive uncertainty (Wertz, Sorenson, & Heeren, 1984) and that generally, reproductive decisions are already made before couples even attend an appointment for genetic counselling (Kessler, 1989).

The analysis of the present study revealed that the odds of 50/50 played an integral part for the men in the couples' willingness to leave the situation to fate. The data suggest that the couples were aware of the odds from the outset. For the women, the decision is clear-cut from the outset, but it is unclear at which point in the decision-making process that the men's thinking accepts the uncertain conditions of leaving events to fate. The women's process resonates strongly with Smith et al's (2002) research, which found that women with a 50% genetic risk of passing on their condition had made their minds up before they even approached clinics. Their research and the findings in the current study challenge normative models of decision-making where the professionals provide information for the patients to assimilate over time, and are consistent with real-life decision-making processes using the least effort and time (Broadstock & Mitchie, 2000).

For the men, it is not apparent whether their decision changed over time from cautious to the comparative acceptance of fate. It is difficult to estimate the extent of the men's fluctuating beliefs and attitudes regarding the risks. Although their narratives demonstrate an optimistic approach that undoubtedly contributed to their acceptance of fate in the decision, this was not necessarily an enduring feature throughout the process. Their approach was malleable depending on the circumstances, often appraising the situation as they considered the worst-case scenario. There is a strong sense of the men as passive bystanders in the situation, and perhaps one reason for the men's choice of acceptance of the 50/50 gamble is due to the fact they cannot control the risks. The very nature of the reality of having a child with Marfan

syndrome and the related risks to the women in pregnancy and childbirth could be considered as out of their control. Therefore, it is probable that the men have tended to accept a high-risk gamble, as events are perceived as externally controlled (Liverant & Scodel, 1960). Furthermore, the men's decision-making is pressurised due to their wives' biological clock. Perhaps this leads them either consciously or unconsciously to make a fast economically viable solution to a complex life-changing decision (Gigerenzer, 2008).

## 6. BEING A THERAPIST AND A RESEARCHER

It has been acknowledged that research can blur the boundaries between research interviews and psychological therapy (e.g. Etherington, 2001c; Rosenthal, 2003; Skinner, 1998, Wosket, 1999). In the current research this played a crucial role in the on-going discourse between the couples, opening up endless opportunities, similar to a therapeutic counselling session (Skinner, 1998). During the interviews and throughout the research, I had a deep sense that the participants, and I as the researcher, experienced a significant transformation, which left an indelible mark on our lives. The potential for this interactional phenomenon has also been recognised by Denzin (1989). But as a researcher it was important not to lose sight of the primary purpose of the interview as a method of data collection for the research study (Etherington, 2004).

I have explored my identity of being a counselling psychologist and being a researcher. This will continue to be an on-going process throughout my career. The two roles naturally have similarities, but there are differences. As a therapist, I do not know what my clients take with them when they leave the room. However, there is the knowledge that they come to me for change and there is usually an opportunity for further appointments to follow through that process of change. As a researcher, I am seeking to expand the knowledge of my chosen profession with the informed consent of my participants, aiming to improve the service we provide. When I leave my participants after the

interviews I do not really know what impact the interview may have had on them, despite comprehensive debriefing at the end.

Gates & Hinds (2000) caution that we must be mindful of the conflict between being a therapist and a researcher; ‘...A therapist attempts to develop an individual, including his or her strengths, and realistic self-perceptions, whereas a researcher attempts to develop ideas, and indirectly people.’ (p. 102). During the interviews I was aware of the conflict of being a researcher and the ethical dilemma of feeling that my role as that of a therapist might have been helpful for the participants. In particular, I felt this when the women were being quite stoic, when the men were emotional, and throughout the interviews when I felt that an individual was not being heard. A therapeutic intervention would possibly have made a difference to the participant’s well-being, but it wasn’t appropriate to make a therapeutic intervention because the important ingredient for the interview was myself as the researcher not as a therapist. Moreover, while both therapy and research are concerned with a frank exploration of a person’s experience, it is ethically more problematic to probe into the darker elements of a person’s experience when you are in the role of the researcher. This may have had the effect of producing more stoic and positive accounts of the participants’ experiences, but they had agreed to the interview, they had not agreed to therapy.

This awareness further developed my sense of who I am as a researcher, a therapist and a human being. Chenail (1996) suggested that those in the role of being a therapist and a researcher should ask themselves 50 questions in

order to gain greater self-awareness. The questions I have found most helpful are where he asks us to consider our strengths and weaknesses as a researcher, a therapist and a person, and what we can do to improve our strengths and overcome our weaknesses. Through my experience as a reflexive researcher in the current project, I have become even more reflexive in my practice as a therapist and use these questions as prompts for my process notes.

The experience of being a researcher has also impacted on my practice in working with individuals and couples. As a result of the current research I have a deeper understanding of the complexity of relationships. When working with couples I find that I explore on a deeper level some of the pathways that establish relationship problems. I am more aware of the need to separate out the negative interactions, stress or conflict that impact negatively on the individuals within the coupledness. Sometimes when working with couples there are echoes of the participants' narratives, in particular when a couple is going through a stressful life event. A greater understanding of the gender differences has become an important element in identifying and interpreting patterns of communication, denial, distress and the power dynamics within the coupledness. The need for sessions of individual therapy alongside couple's therapy has become more apparent, and the need for couple's therapy when a client presents for individual therapy is sometimes indicated as a more constructive way forward.

## 7. REFLEXIVITY

In this final section on reflexivity I consider how my decision to have a baby and my personal connection to Marfan syndrome may have affected the research. My gender and the gender of the participants are also considered further from a reflexive stance. I explore how my personal experiences may have impacted on how I interviewed the participants and how I analysed the data. I consider my assumptions in the interview and reflect on what I possibly expected to hear based on my previous experiences, what I did actually hear and how my assumptions were challenged.

I decided to share with the participants that Marfan syndrome had been diagnosed in my family as I thought this might be helpful for the participants when talking with me about Marfan syndrome. However, I decided not to share the fact that I was a mother to ensure that the focus of the interview remained on the participants' experience of decision-making to have a baby. Before the interviews I talked with participants until I felt that we were all ready to begin recording. The participants did not ask whether I was a mother or in a relationship before or during the interviews. However, we did discuss Marfan in our respective families.

As previously mentioned, I believe that the participants might have assumed that I was a mother because I do look 'motherly'. Occasionally participants would make a point regarding becoming a mother or a father and then end it with '...you know what I mean?' or '...isn't it?' or '...don't you?' when talking

about the decision-making. I responded by acknowledging this and then focussing on the point they had made to find out more information or to clarify that I had understood what they were telling me. Following the interviews I mentioned aspects of my personal experience of making the decision to have a child and being a mother.

During the interviews, I could identify with the women's desire to conceive and have a child. In particular, when Belle was talking about 'the broody nerve' taking over and when Cathy was talking about feeling 'useless' if she couldn't have a child. I had a strong sense of the emotions they may be feeling. I often found myself aware that I wanted to probe more into their experiences of longing for a child, and I did when appropriate. I have to admit that at the same time I was tempted to share something of my own experience. At times, I think I was reluctant to probe more as I could sense a possible vulnerability or denial in the women behind the strengths and I didn't feel it would be ethical to break through this in the interview. But perhaps this emanated more from my own experience twenty-three years before and my naïve thinking about what it means to have that experience in today's society.

I took an optimistic view when we decided to have a baby and perhaps on reflection I carried this optimistic stance into the interviews. Therefore, I was possibly more ready to hear the stoicism and not so much of the distress of my participants. However, in recognising this potential blind spot I was more alert to understanding the perspectives of the participants' experience.

I was surprised to find that the couples spoke to others so openly about their decision-making. From my own experience I would have anticipated that it would have been a private matter between them. When I wanted to have a baby I didn't talk as openly as the women did in the interviews. Reflecting on this, I wondered if, had I discussed it openly with others, it might have made me feel vulnerable or disloyal to my husband.

During the analysis, I was surprised to find that one of the themes was 'Her decision is already made', and in the first draft of this thesis I included an exclamation mark following the theme. Reflecting on this, I think I initially saw the irony in this theme emerging, given the title of this research project on 'decision-making'. However, I think I also assumed that the men and the women in the couples would go through some sort of decision-making process. There was evidence that they did, but I was surprised that the women demonstrated what I initially considered to be strengths and admirable qualities in having made that decision from the outset. At the interview stage and during the analysis, I believe I may have been biased towards seeing more of the women's strengths in their narratives as opposed to their distress as I have seen this demonstrated in my own family, in my own experience of decision-making to have a baby, and in my previous research on breast cancer. However, from personal experience in my family, the underlying distress is apparent and openly discussed, but it is the strengths that shine through.

On reflection, I suppose I should not have been surprised that the women's decision to have a biological baby was already made. I had already made that decision too when my husband and I were discussing having a baby. In coming to understand the women's narratives, I recognised that I would probably have made the same decision as them, despite any consequences. I was slightly surprised to hear the men's narratives regarding their caution in making the decision to have a biological child. However, I did not expect to find that the men coped alone with their distress. Reflecting on my own experience and viewing our situation through a different lens informed by this research, I wondered if there may have been more going on in our relationship than I realised at the time.

The findings demonstrated consistent gender differences and I suspected that this would be the case. But I did not expect the extent to which there would be such consistencies in making the decision. Some of the differences were naturally apparent in the interviews, but the amount and type of differences that emerged during the analysis were at times surprising. In the two superordinate themes 'Her decision is already made' and 'Existing with the fallout of the decision' and again in the theme of 'Sharing responsibility for decision' under the superordinate theme 'Creating an informed decision', the differences are the most distinct.

I sometimes had the sense that the men would have liked to talk with me more about their side of the story; in particular, with Jack, Adrian and James, who became quite emotional at times during the interview. It is possible that

the men were sending out the cues for me to respond to their distress in order to collude with them to emphasise to their partners the emotional side of their story. When the men were talking about their emotional experience I sensed that this might have been the first time that they had had the opportunity to tell their side of the story and feel heard. Following the interviews, some of the men's words remained with me for a significant amount of time. I consider that the three themes 'Sharing responsibility for decision', 'He fears losing her' and 'He copes and cries alone' were among the most powerful themes for the men that emerged during the analysis of the data. On reflection, I wonder whether they would have shared their emotional feelings with me to such an extent if I had been a male researcher. I think it is important to acknowledge the limitations of my gender in understanding the men's perspectives.

Reflecting on how I heard the different perspectives of the men and the women during their decision-making and the decision to go ahead despite the risks, I had to examine my beliefs about the decision they had made. I certainly admired and understood the women's strengths, but at the same time I felt empathy and understanding for the men's concerns and hesitation. I was aware of trying to maintain neutrality and interviewing the couples provided me with what I considered to be a balanced view of what it was like to make the decision. Both the men and women appeared to share their side of the story quite freely and openly and often I could feel the tension between them, particularly when they were discussing making an informed decision and their differences in sharing the decision. Initially, I assumed that when the men were describing their distress, and in some cases tears, this would

be purely about their concerns in decision-making. On reflection, there may well have been other contributing factors outside the relationship that might have changed the language and meaning of their tears and distress. Nevertheless, I firmly believe that there is no right or wrong answer in making the decision.

## 8. CRITIQUE AND SUGGESTIONS FOR FURTHER RESEARCH

There are several constructive thoughts regarding the current study and these are now discussed in depth with a view to generating potential ideas for further research.

In line with the aims of IPA the sample was small in number, homogenous and purposively selected. Furthermore, the couples essentially were volunteers likely to belong to a similar social, educational and economic status. Therefore, it is possible that the sample represented a particular subsection of the population of couples making a decision to have a baby. Regarding generalisations, IPA acknowledges caution and places them in the particular (Smith et al, 2009). Future research should consider decision-making from the perspective of couples from a variety of backgrounds at different stages of decision-making, with a view to identifying significant themes highlighting similarities and differences to achieve a more universal approach.

It is acknowledged that the research focused on couples' experiences of desiring a baby, and the couples' accounts demonstrated the overwhelming longing for a child. It is possible that couples with a different experience were less likely to come forward to volunteer for the interview and discuss their decision-making with a stranger for the purposes of research. It would be interesting to investigate and compare decision-making from this alternative point of view.

Furthermore, the research represented couples that chose to go ahead and have a biologically related child and therefore the findings reflect the drive, identity, emotions, beliefs, actions, behaviours, responses, wants, needs and values of couples coping with the prospect of a high-risk pregnancy. Further exploration of the subjective meaning of a high-risk pregnancy could provide more insight into this experience. However, research with couples choosing not to have children or who are involuntarily childless would most probably generate different findings, reflecting a possible need for the provision of psychological support. It would be most interesting to carry out a study using such sampling to understand this particular group in more depth.

In addition to these considerations, the couples were interviewed together as a couple to reflect and emphasise the aims of the research title. It was considered that interviews with individual males and females would dilute the dynamics of the coupledness and thereby divert the focus of the research objectives. Interviewing participants as a couple was relevant from the perspective of counselling psychology in viewing the relationship in context (Street, 2006), as it may be more likely that they would present as a couple within a multidisciplinary team.

However, it could be argued that interviews with the couples was restrictive for the individuals within the relationship and therefore it is important to consider what may have been lost by not interviewing the participants individually. I was mindful of what may not be said in the couple interview. There is a strong possibility that individuals may hold back on providing

certain information in front of their partner for fear of upsetting them or causing concern. Similarly, they may appear more positive and stoic than they feel, reflecting a mismatch between verbally expressed and actual thoughts, emotional feelings, physical feelings and behaviours. Another aspect of interviewing couples is that the data may reflect power issues within the relationship, along with stereotypical gender norms. For example individual interviews may yield data that provides unique and individual responses and thereby a more in-depth representation of the experience highlighting the impact on health, identity, and well-being. An exploration of individual male and female experiences over the course of decision-making may open up some areas and yield some further insights into the process of decision-making. A greater emphasis on individual thought processes could be an interesting avenue for future research.

In considering the title of the present research, it may be revealing to interview men whom had Marfan partners who they lost during childbirth. Another area worthy of investigation may be interviewing couples where there is a male diagnosis of Marfan syndrome, to study the ways partners jointly impact one another. Also, in the light of the women's decision already being made in the current study, it would be interesting to interview young adults affected with Marfan syndrome to understand their reproductive thought processes at this stage of their life.

The distinct gender differences in decision-making in the current research suggest that the gender dynamics in decision-making to have a baby in

general should be considered as a valuable area for future research. In the same context, decision-making to have a child in the event of illness generally in either males or females would possibly generate some relevant findings. Research into couples with fertility concerns and the process of assisted conception would perhaps shed more light on the experience and the dynamics within the relationship. There may also be a need for further research into the gender dynamics of decision-making overall, as there is possibly an association with how couples make a decision in other areas of their relationship.

An interesting perspective of the researcher as a potential unwitting couples' counsellor has emerged from the current research. Although this has important implications for this and future research, it has highlighted that there is a need for further research into the effects of the transference and countertransference when studying close relationships. Specifically, it would be interesting to carry out a comparison of the experience of being a couples' counsellor researcher and the experience of being interviewed as a couple by that researcher.

Another consideration concerned the age of the children at the time of the interviews. There was some difficulty accessing couples that were actually going through the decision-making process and only one couple volunteered for interview. Broadening the search to couples that had already made the decision and had children proved to be more fruitful and five couples came forward. Although I was cautious regarding the retrospective data, it would be

interesting to interview couples going through the live experience of decision-making in order to compare the present findings.

Interestingly, in the current study the accounts of Belle and Adrian, who were actively going through the decision to have a baby, were strikingly similar to, and could be mapped onto the data from, the other five couples that had already made their decision and had children. I am aware that the experience of interviewing Belle and Adrian first and hearing their story may have had an impact on the subsequent interviews regarding the questions I asked and how I interviewed. However, the retrospective accounts demonstrated that the decision-making was an intense time for the couples because things aired at the time and the memories remained fresh for them. Comparison of the current and retrospective accounts seeking convergence and divergence revealed that the memories were likely to have remained loyal to concerns, and provided an indication or suggestion of what concerned couples at the time of decision-making. I would therefore argue that an advantage of the current study is the complex picture of the couples' experiences of decision-making that has emerged in the findings, which is reflected in the enduring and embedded nature of the experience in their lives. However, this warrants further investigation to see if that link is typical.

## **9. CONTRIBUTION**

This section focuses on the contribution of the current research and overall implications for the profession of counselling psychology. The meaning of the findings is considered in the light of how the impact of this new knowledge could potentially translate into a more effective environment for people affected by Marfan syndrome. However, it may also have implications for similar genetic conditions and perhaps other couples-related issues such as infertility. The limitations of the current service provision and resources are explored and suggestions of how they can be overcome are proposed. Specific areas where the contribution of this research could make a difference are discussed under the following headings relating to the real world of this particular population: service provision; Internet resources; the media; Marfan associations and groups; psychological counselling and psychotherapy.

### **9.1 Service provision**

The couples' accounts of their experience with medical professionals', such as doctors, consultants, cardiologists and genetic counsellors demonstrated a lack of knowledge or empathetic understanding regarding the emotional impact of coping with Marfan syndrome and decision-making to have a baby. This is consistent with literature that highlights that there is clearly much room for improvement in this area (Budysh et al, 2012). The personal experience of couples' reproductive decision-making with medical professionals has been previously neglected in the literature, due to the predominantly quantitative approach of using questionnaires, and the focus on patients as individuals. It

is therefore argued that the current research has significant clinical relevance and made a unique and valuable contribution to the understanding of this particular population. Additionally to Marfan syndrome there are further potential applications of the research findings. The care of patients with other genetically inherited conditions should also be considered. This research may have implications for healthcare professionals identifying the psychological needs of couples and individuals experiencing infertility, other fertility concerns or indeed any couple or individual making the life-changing decision to have a baby.

This study has highlighted that there is a need for patients to have the opportunity to voice their emotional concerns during consultations with professionals as this could open the pathways for a referral to an appropriate psychological service. It appears that a crucial aspect of this study concerns the many gaps in service provision. Since the 1980's, psychologists have been filtering into a number of medical settings (Taylor, 1990), but it seems that there remains a substantial lack of psychological components in the present system.

Another feature of the study revealed the complexity of negotiating a high-risk pregnancy and there was an emergent picture suggesting the poverty of emotional, educational and professional support at this time. I believe there might be echoes here of my discussions with professionals before starting this project, where I found that their knowledge of Marfan syndrome was either limited or non-existent. However, it is important to be mindful that some of the

couples were talking retrospectively, their children's ages differ and therefore what was available in terms of support may have changed over the years. The current research has provided a valuable insight into this crucial aspect of service provision and concludes that this has important implications for healthcare professionals, not only in caring for men and women coping with a high-risk pregnancy, but also during their decision-making to have a baby.

Additionally, the men's narratives emphasised the unique way in which they coped with decision-making and a high-risk pregnancy. Their vulnerability was an important aspect of the experience, but their emotions were tempered by the hegemonic masculine ideology associated with Western society. They bore their distress alone as they conformed to the cultural and social norms associated with masculinity. It is therefore argued that this study has made a unique and valuable contribution to the current understanding of men's individual needs at this time, highlighting the striking differences between a man's and a woman's world within a relationship. Moreover, it is possible that this research has implications for men's emotional well-being in a wider context of decision-making and pregnancy generally. However, further research would be necessary to explore and confirm this. It is considered that healthcare professionals need more awareness of men's problems at this time as it would be invaluable in identifying psychological distress in this seemingly forgotten majority, and have important implications for the role of counselling psychologists within a multidisciplinary team. As counselling psychologists, we must challenge the gender assumptions in therapy (Day, 1992), and

recognise that many males and females conform to culturally shaped ideas of masculinity or femininity, confusing our perception of their coping abilities.

Of note, The British Heart Foundation and Marfan Association UK emphasise the psychological burden of making a decision to have a baby when an individual has Marfan syndrome: *'It is very important that the unaffected partner is allowed to express his or her feelings about the advisability of pregnancy, since it would be their responsibility to care for an affected partner and possibly also an affected child'* (British Heart Foundation, 2002, p. 30).

Similarly, a recent clinical report updating key areas for care of Marfan patients clearly states: *'Health care professionals also must be cognisant of the patient's emotional issues, including body image concerns, activities, sexuality, reproductivity and mortality'* (Tinkle, 2013). Surprisingly, it appears from the findings in the current research that this advice is not heeded within the existing service provision. Seemingly, patients' needs are not being met.

It is emphasised in the NHS information for GPs, other medical professionals and patients, that genetic counselling is not a form of psychological counselling or therapy and is not to be confused as such (www.patient.co.uk, 2013; www.nhs.uk, 2013). But, as the findings have shown, the processes involved in couples' decision-making are complex and may require more than an explanation of the facts or information-giving on the options available, regarding having a baby with a genetic condition. Although the discipline of counselling psychology has the ability to introduce an element of evidence-based psychological thought into the training and practice of genetic counsellors, it is perhaps ambitious to expect genetic counselling to provide

'counselling' in the psychological sense. This is primarily due to a conflict of interest that may arise for genetic counsellors and other healthcare professionals, blurring the boundaries and purpose of service provision. It is suggested that genetic healthcare professionals should maintain a responsibility to recognise their limitations, identify the psychological needs of patients and thereby make an appropriate psychological referral.

Following these last points, it should be acknowledged that the research and findings focused on the decision to have a child, but the implications for couples or individuals who choose not to have a child, or are involuntarily biologically childless, should also be considered in the light of the shape of service provision and psychological therapy. However, there are important implications for the role of psychological therapists and service provision for men and women for individual or couple therapy. Health professionals should be suitably trained to recognise who is in need of psychological support in order to facilitate suitable referrals, help decide how that service is delivered and thereby provide appropriate psychological interventions. It is envisaged that an effective service provision will be mindful that psychological issues are invariably not voiced by patients and the challenge for service providers is to recognise this and enable an appropriate signpost to psychological services.

It is argued that due to the detailed picture that has been constructed of the couples' experience of decision-making, reflecting both its complexity and embedded nature, a pertinent role for counselling psychologists within the current service provision has emerged. Counselling psychologists could

provide valuable expertise in developing, writing and delivering educational training programmes for doctors, consultants, genetic counsellors and other healthcare professionals such as osteopaths, dentists, opticians and physiotherapists. These programmes would serve to educate and raise awareness of the psychological effects of Marfan syndrome at all levels of training, including continuing professional development.

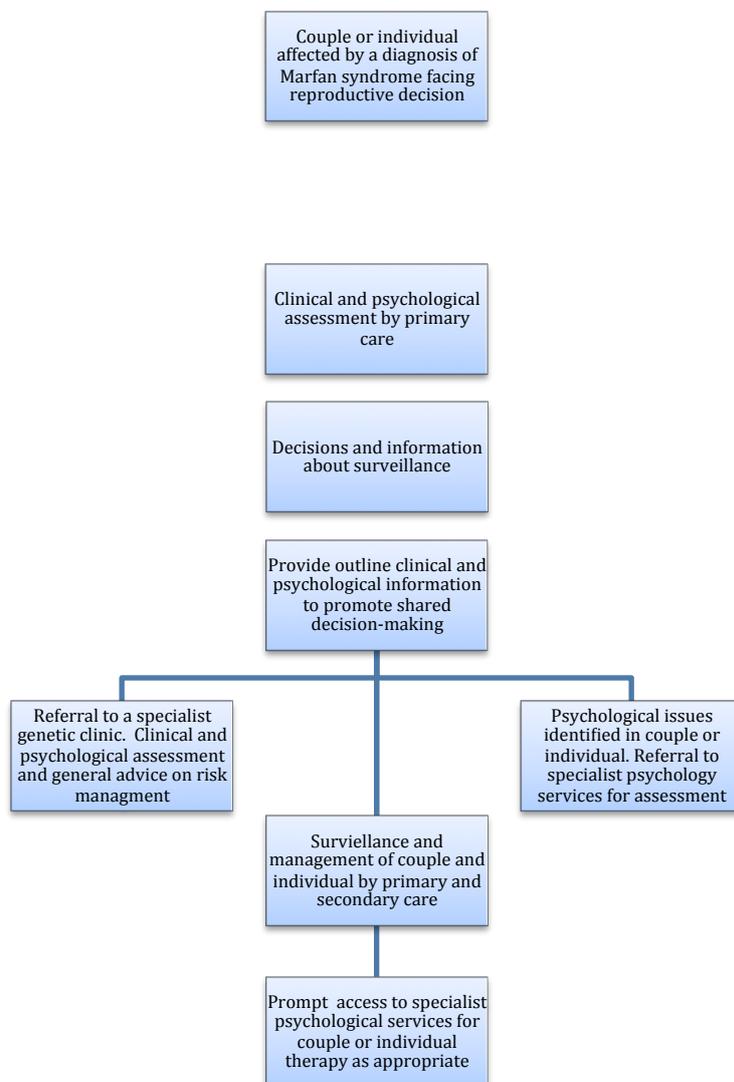
Counselling psychologists could have an intrinsic role in developing psychological services best suited to addressing the needs of patients. One possible way of delivering this psychological service would be to provide a 'one-stop shop' within a multidisciplinary team emphasising the importance of holistic care. The current services are highly centralised in London at St Georges Hospital and 17 other centres throughout the region. Considering that London represents only 19% of the Marfan population (Marfan Association Information, 2010), it is questionable whether this is an effective and economical use of resources. It would seem from the current research and discussions with service providers that referral guidelines and protocols for management do not include a means of coordinating specialist psychological expertise.

The National Institute for Clinical Excellence (NICE) guidelines (2013) for couples considering a pregnancy when a woman has a genetically inherited condition, recommend a service providing information that a woman and her partner can use in order to make informed choices about planning a pregnancy to ensure the best possible outcome for the couple and the baby.

The psychological component regarding the best possible outcome appears to be absent. The current research has highlighted that this is evidently an oversight.

The NICE guidance pathway (2013) for genetically inherited breast cancer may provide a useful template for the psychological care, assessment and management of Marfan patients. The pathway includes 'referral to a specialist genetic clinic'; 'providing information to promote shared decision-making'; 'assessment and management in primary and secondary care'; 'information and psychological support'. The pathway applied to the care of couples and the individuals within the coupledom has been adapted for the purposes of decision-making to have a baby when a woman has the potentially life-threatening condition of Marfan syndrome. The current research has emphasised that the needs of the individuals within the couple should be considered throughout the pathway. There are related complications to breast cancer and the pathway could include similar elements as suggested in Figure 1 below.

**Figure 1.** Suggested pathway overview for couple or individual with a diagnosis of Marfan syndrome facing reproductive decision.



It is thought that referrals to specialist psychological services should be made available to couples at a primary care level and from the outset of contact with secondary care. The findings in the current research also suggest that referrals should be considered throughout decision-making, pregnancy and beyond.

As the findings in the current study indicate, there is most probably much progress to be made in government-based policy for service provision to support couple relationship counselling. Ensuring that the necessary resources of public and private financial support are made available entails gathering evidence-based outcome data to provide stronger justification for couple therapy (Cowan & Cowan, 2012). Couple-based interventions can promote healthier coping in major health crises, not least because their behaviours are interdependent; research design should therefore assess couple-coping rather than just individual outcomes (La Guardia, 2012).

## **9.2 Internet resources**

An interesting insight that this study has provided is a greater understanding of the couples' experiences of Internet resources. Although the couples' accounts demonstrated a need to talk with a person for their emotional needs, the Internet proved to be an inefficient and potentially harmful resource. There is clearly a need for monitoring the quality of medical information on the Internet, as this is potentially a rich source of information. It can offer a wealth of health information, in particular for people with rare diseases, and it is usually available all the time (Lasker, Sogolow, & Sharim, 2005).

The present research is therefore of particular value and significance as it suggests that professional signposting to good quality Web-based medical, psychological and relevant social information sites, might be beneficial for those with a genetic condition in making a decision to have a baby. Counselling psychologists could provide a wealth of information to assist in composing helpful websites based on the current findings. They could advise the medical and other professionals who write the current programmes, which, according to the current research, are not fit-for-purpose. Counselling psychologists with appropriate knowledge of genetic conditions such as Marfan syndrome could offer Internet-based counselling services or provide training and supervision to other therapists offering online counselling.

### **9.3 Marfan associations and support groups**

The literature and information leaflets produced by Marfan associations and support groups for patients and professionals are written mainly from a medical perspective. More often than not they skim the surface of the psychological elements of coping with these important aspects of the condition. From childhood concerns through to adult issues many of the findings of the current study remain unrecognised.

As with web-based information, counselling psychologists informed by the theoretical insights and considerations of the current research could facilitate production of relevant literature by offering expertise, tailoring the content to a specific psychological context.

It is envisaged that an increased psychological perspective would essentially provide a more balanced picture of Marfan syndrome to complement and supplement the existing medical literature. The literature would be pertinent for patients and their friends and family, professionals, educational institutions and the workplace.

#### **9.4 The media**

Findings from the current research have the potential of heightening public awareness of the psychological effects of reproductive decision-making with a diagnosis of Marfan syndrome through media resources. Radio, television, newspapers and magazines are renowned powerful voices for imparting or generating new knowledge and sparking debate. These resources have been employed in the past by the author and it is intended that they will be explored to emphasise the importance of this research.

#### **9.5 The practice of counselling psychology and psychotherapy**

There are significant implications for counselling psychologists and other professionals such as counsellors, psychotherapists, and relationship therapists involved in couples counselling, family therapy and individual counselling for women, men and young people with Marfan syndrome and other related conditions.

It is perhaps important to consider what constitutes counselling psychology and what distinguishes it from clinical practice, in order to understand and identify the value of its contribution in this research and therapeutic practice in

general. Although counselling psychologists could have an important role in the education of practitioners such as genetic counsellors as described above, the clinical treatment approach for genetic conditions in the NICE guidelines (2013) is firmly set within the medical model and the positivist paradigm. As such, it could be argued that counselling psychology, with its firm foundation in a humanistic philosophy of the human sciences (Strawbridge & Woolfe, 2003), can provide a holistic approach that will not define clients as merely suffering from Marfan syndrome.

It could be said that one of the main epistemological differences between a clinical perspective and that of counselling psychology is that clinical practice often maintains a distance between the practitioner and the patient (Rawson, 2012). Historically, counselling and clinical psychology derive from the fundamentally different perspectives of relational versus clinical respectively. Although it appears that within the NHS and other organisations, the historical gap between counselling psychology and clinical psychology has narrowed over the years, with the two disciplines converging as essentially scientist-practitioners working with similar patients, it has been suggested that as the identities of counselling and clinical psychology become increasingly blurred, they will eventually merge together (Giddings, 2009; Turpin, 2009).

However, it remains that the focus of therapy from a traditional clinical stance may typically rely on finer and finer assessment, formulation and description of the presenting problems. Such an approach tends to reduce the patient to the sum of its parts. While I understand that it is necessary to acknowledge

certain symptomologies of Marfan syndrome, the current research has demonstrated that there is also a bigger picture of the complex and multifaceted aspects of making a decision to have a baby with this genetic condition.

Interestingly, the current research has had a significant impact on my work with clients, in particular with couples. Viewing relationships through a different lens informed by the research has enabled a richer understanding of how I would work with Marfan couples, couples where the wife has been diagnosed with a genetic condition, couples coping with infertility or couples who were considering pregnancy. Working with these couples involves entering into an intimate and private area of life and understanding the couple interaction. The current research has further emphasised the importance of a holistic therapeutic relationship with such couples, in particular when there is illness, and has stressed that the protection of clinical distance would be a hindrance. Furthermore, the relational dysfunctions and triumphs that keep the relationship going or destroy it under these circumstances have become clearer, highlighting that a more in-depth exploration of these significant issues during the sessions may be needed. Direct access to the client's world via the therapeutic relationship is not to be underestimated.

The research findings have highlighted that a couples-based approach to therapy may be appropriate when there are health concerns, as it is a couple problem due to the interdependent nature of the relationship. However, it is the men's distress that highlights a more pressing need for individual therapy

to support them through the difficult time of decision-making and perhaps beyond. The women's denial, discussed below, possibly indicates a cautious case for individual therapy for them too. This suggests that the possibility of individual therapy alongside couple's therapy should to be considered.

There are some related critical issues to be mindful of for all psychological practitioners. There is a need for heightened awareness of each partner's relationship with the external world and appropriate use of the therapeutic relationship as a direct way of accessing those worlds. For these clients it is considered even more important for therapists to understand each partner's relationship with themselves and each other, focusing on both individuals and the space between them (Berscheid, 1999). For example, the research findings have demonstrated that a female client with Marfan syndrome has a relationship with the female self who has the condition, but there is also their relationship with the female self that does not define them with that condition. In addition, there is the female client's relationship with her partner. The male partner without the condition has a relationship with the self as both an individual and as a male in relationship with a woman and with a woman who has the condition. There is also the relationship between therapist and clients. Therefore it is crucial to recognise that in working with couples and illness there are several relationships at play and the relationship issues become compounded as such.

There were some striking instances of denial where the women relentlessly downplay the risks and go on to have a biological baby, regardless of the risks

to themselves or their child. Constructively denying or regulating negative emotions has been associated with greater personal well-being (Vohs & Finkel, 2006). However, little is known as to how this individual emotional regulation affects the general functioning of the couple relationship (Tran & Simpson, 2012). It is arguable whether it would be morally correct for a therapist to break through the denial of risk for these women, as it could be seen as their way of coping; a defence mechanism in the Freudian sense. This is a complex issue, but therapeutically it would no doubt hang between the therapist and clients. Therefore, it is considered that in this context, simply recognising and acknowledging the denial to the client would be the most transparent and effective intervention. The aim should not be to reach a compromise or to weaken the defence of denial, but rather to enhance the client's ability to understand the implications and optimise the outcome of therapy (van Deurzen-Smith, 2002).

The medical component of Marfan syndrome or other similar potentially life-threatening conditions suggests that an understanding of the disorder may be necessary in order that therapists may be equipped to work with these clients. The current research has indicated that, for the purposes of psychological therapy, the topic is not about the illness; it is about how they are psychologically unsettled and how we as therapists navigate them through it. Therapy usually stirs up the bottom of the pond; with a potentially life-threatening condition there are the reactions of self and others to consider and thinking about death is very much in the frame. The aim of therapy is to re-pattern and make different connections in order to allow the sediment to

settle. Therefore, I would argue that some naivety might be beneficial for the therapist, as being too informed about the condition could make it clinical and cloud therapy.

An overarching contribution of the research to the practice of counselling psychology is the development of a broader conceptualisation of couples coping with Marfan syndrome relating it to psychological functioning and quality of life. As a result it is argued that this places Marfan syndrome in a wider arena rather than exclusively in a clinical or medical setting. It is hoped that in line with the discipline of counselling psychology, the findings of the current research will enlighten therapists and thereby help them to provide appropriate support specific to the context of reproductive decision-making in Marfan syndrome, other related genetic conditions, and reproductive decision-making generally in all couples and the individuals.

## 10. CONCLUSION

This research has explored an important aspect of coping with the genetically inherited condition of Marfan syndrome: the experience of couples' decision-making to have a baby when there is a female diagnosis of Marfan syndrome. This is of potential interest to counselling psychologists due to its possible significant implications for psychological well-being. Counselling psychologists have suitable training and skills and are well placed to offer the appropriate therapy for couples coping with health-related issues and they increasingly work in a wide variety of settings with a broad spectrum of clients (Ponterotto, Kuriakose & Granovskaya, 2008). Issues relating to health are now being placed in the mainstream arena and the unique skills of counselling psychologists are now valued in areas that were once dominated by specialist clinical teams (Corney, 2006).

The literature review highlights the poverty of research aimed at the psychological experience of reproductive decision-making for people affected with a genetically inherited condition. There is a need for more interest in understanding how individuals with a genetic condition make reproductive decisions in order to explore the psychological dynamics (Richards, 1999). The extant research is mainly quantitative, objective and presents a scant mixed picture. It is often simplistic, focusing on different aspects of decision-making, neglecting the complex and diverse nature and meaning of this experience. Furthermore, it seems that in reviewing the literature and exploring the current service provision for this group of patients it tends to

ignore the intricate psychological dimensions of how couples make sense of and give meaning to their experience.

The research explored the in-depth experience of decision-making for six couples when there is a female diagnosis of Marfan syndrome. The study adopted the qualitative approach of IPA. It was an ideal choice for the research aims, well suited to the health-related nature of the topic and considered the relational dimensions and complexities of the couples' experiences of decision-making. IPA has made a significant impact on health-related topics and is particularly valuable in its unique qualities that have contributed to qualitative investigations in applied psychology (Eatough and Smith, 2008). Furthermore, I would argue that IPA is commensurate with the philosophy of counselling psychology.

Much of the research into decision-making with genetic conditions assumes that individuals merely weigh up the pros and cons of the options (Broadstock & Michie, 2000), and there is a dearth of research studies aimed at how individuals in the real world make important decisions and explain the reasoning behind those decisions (Smith, Michie, Stephenson and Quarrell, 2002). Furthermore, there is a lack of research aimed at understanding the needs of people making a decision with a genetic condition and role of emotions in decision-making in particular in the context of gender differences. However the current research has demonstrated the complexity of this process and in line with the aims of IPA has demonstrated the variability and diversity of the couples' experiences in assessing the risks concerned. There

are commonalities in the female experience and in the male experience but as a couple, there are elements of the interdependent nature of their relationship that are critical when considering psychological research or psychological therapy. Hence the value of interviewing the participants as a couple, despite the potential methodological issues outlined above.

The rich insights and considerations generated by the current research have allowed an important contribution to a deeper understanding of what it is like for couples to go through the experience of decision-making to have a baby. The idiographic approach of IPA enabled me to give a detailed in-depth analysis that accessed and represented the actual nature of their experience. It is anticipated that the research will inform a wide range of professionals, individuals and organisations. Psychologists, psychotherapists, healthcare professionals, the media, the public and the Government could offer appropriate and proficient support for these patients through gaining new knowledge and a greater understanding of the impact of this experience.

It is hoped that through a deeper and more meaningful understanding of couples' decision-making to have a baby when there is a female diagnosis of Marfan syndrome, the direction and shape of clinical and therapeutic practice will change to reflect the link between the desire for children and its emotional relevance. As counselling psychologists, we are encouraged to engage empathically with the client's world (British Psychological Society, 2000) and it is hoped that this skill may to some extent be transferable to other healthcare professionals to facilitate referrals for appropriate psychological care.

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## APPENDICES

**APPENDIX A****HOW DO COUPLES MAKE A DECISION TO HAVE A BABY WHEN A WOMAN HAS MARFAN SYNDROME?**

My name is Annie Hinchliff and I have close family members affected by Marfan syndrome.

As a Chartered Psychologist my experience has moved from the personal to the professional as I have become increasingly interested in how other families cope with Marfan syndrome.

Currently, I am carrying out some research into couples' decision-making to have a baby when a woman has been diagnosed with Marfan syndrome. I want to share and understand what it is like for couples to make that decision.

The aim of the research is to advance the awareness and understanding of Marfan syndrome and provide a non-medicalised perspective on couple's decision-making to have a baby. I seek to generate new knowledge with a view to improving the present support system.

Would you like the opportunity to be one of six couples to talk with me about your experiences? As a woman you will have a diagnosis of Marfan syndrome. Equally important, as a man you will be sharing the experience of decision-making with your partner. In addition, as a couple you will have received genetic counselling.

We shall meet for one confidential interview, lasting about one and half hours, to talk about your experiences. You will be kept up-to-date with my work throughout the research and presented with the findings at the end. This is an opportunity to explore your thoughts and feelings, and to be at the forefront of research into how we can advance the level of care offered to couples now and in the future in their decision-making to have a baby when a woman has Marfan syndrome.

Interested? You can contact me in confidence using the links below and I have an information sheet giving comprehensive details about the research, which can be sent to you without obligation.

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## APPENDIX B

### WHO'S LEFT HOLDING THEBABY? EXPLORING COUPLES' DECISION-MAKING FOLLOWING A MATERNAL DIAGNOSIS OF MARFAN SYNDROME.

Preliminary list of emerging themes with page and line numbers

#### Interview 2 – Clare and James

- 1.8 sharing doubt re having children
- 1.10 sharing MFS with each other
- 1.13 she wants him to know the facts re MFS
- 1.14 he accepts MFS
- 1.15 she tests his love of her
- 1.18 seeks professional advice re having children
- 1.30 exploring the options
- 1.33 IVF as risky to mother
- 1.36 deciding on natural route
- 1.40 having opposing perspectives
- 1.41 deciding to let fate take its course
- 1.48 comparison of MFS to other medical conditions
- 2.3 leaving things to fate
- 2.9 the importance of timing
- 2.17 looking at options
- 2.19 seeking professional medical help
- 2.22 assessing the risks
- 2.25 making an informed decision
- 2.27 the importance of timing
- 2.31 medics approval as important in decision
- 2.38 natural route is her decision
- 2.41 he needs to explore options
- 2.44 she feels medical intervention in conceiving is chemical, stressful, unethical and 'playing God'
- 2.45 if her mother had genetically tested her she would not be here
- 3.1 her decision is personal
- 3.3 his decision is detached and pragmatic
- 3.7 they discuss dilemma of decision
- 3.11 she is prepared for being mother to MFS child
- 3.13 her experience of MFS as an asset
- 3.14 sharing the experience of MFS as a child with partner
- 3.18 the reality of MFS
- 3.20 comparison of MFS with other illnesses
- 3.22 rationalising MFS
- 3.31 his empathy for her
- 3.32 an emotional decision for her
- 3.33 the strength of the relationship
- 3.36 exploring all the options
- 3.39 evaluating the risks to her
- 3.44 exploring all the options
- 3.49 concerns re suitability for adoption
- 4.2 making the decision final
- 4.5 importance of medical approval

**APPENDIX B CONTINUED**

- 4.16 decision not 'if' but 'when and how'
- 4.18 she wants him to know facts re MFS
- 4.18 he accepts MFS
- 4.19 Importance of love and strength of relationship
- 4.35 genetic counselling and information and giving options
- 4.40 assessing the risks of genetic testing
- 4.46 genetic testing rejected as would not abort anyway
- 5.1 he is supportive role
- 5.7 the need to make decision pre-conception
- 5.10 she feels guilt re MFS in making decision
- 5.15 she owns her MFS
- 5.19 they make a joint decision
- 5.20 she feels responsible for passing on the MFS gene
- 5.22 understanding the meaning of having a child with MFS
- 5.25 he is optimistic re medical advancement
- 5.33 he is pragmatic
- 5.40 his internal v external locus of control over events
- 5.33 she is optimistic re medical advances
- 5.40 keeping informed re MFS
- 6.5 coping with other MFS couples' negative views on having children
- 6.9 the role of fate
- 6.40 the offer of counselling vague – opposing memories of availability
- 7.2 doing their own research
- 7.7 making the decision together
- 7.10 genetic testing NHS v private
- 7.19 importance of timing
- 7.34 timing and risk to mother
- 8.11 the importance of family support
- 8.16 the effects of her parents wishes on the decision
- 8.21 assessing the cost to the child
- 8.25 drawing on her mother's experience of caring for her as a child
- 8.27 highlighting her mother's guilt with her
- 8.34 comparison of self with her mother
- 8.37 the value of being a Mum with MFS and caring for a child with MFS
- 8.42 empathy re her mother's guilt and MFS
- 8.46 the value of past experience
- 9.5 she has insider knowledge in dealing with child's medical issues
- 9.11 her insight makes being a mother to MFS child less scary
- 9.20 she compares mother's situation with her own
- 9.25 optimism re medical advancement
- 9.31 he is pragmatic
- 9.32 she is emotion
- 9.33 the child as a person not an illness
- 9.43 coping with family responses to decision
- 9.45 assessing the role and expectations of their parents
- 10.1 coping with family reactions to decision
- 10.15 listening to others but owning the decision
- 10.24 exploring the options of having a child
- 10.28 child considered as they explore the options

**APPENDIX B CONTINUED**

- 10.32 exploring options as 'peace of mind'
- 10.36 she wants his to own his part of decision
- 10.39 she checks out how he feels about the decision
- 10.42 she needs to share guilt re MFS to prevent repercussions from him
- 11.3 considering their parents opinions in the decision
- 11.9 her mother as positive re decision
- 11.10 her emotions are up and down
- 11.13 he empathises with her
- 11.16 a massive decision
- 11.22 her mind is made up from an early age
- 11.25 adoption as a last resort
- 11.30 her drive to have child overrides everything
- 11.38 decision to have children made early in relationship
- 11.44 accepting limitations to number of pregnancies
- 12.9 he compares self with own father
- 12.35 importance of family support
- 12.38 exploring options with the family
- 12.49 coping with high-risk pregnancy
- 13.2 coping with the reactions of family
- 13.5 her fear of loss for self and baby
- 13.9 coping with reality of high-risk pregnancy
- 13.13 coping with risk of who's left holding the baby
- 13.18 coping with the reality of child with MFS
- 13.22 guilt of having a MFS child
- 13.27 MFS information day as helpful
- 13.30 the internet as research tool for MFS
- 13.35 comparison with others affected by MFS
- 13.40 coping with others negative perception of choice to have a baby
- 14.1 intensity of the process of decision-making
- 14.7 the romance of decision is diminished by clinical intervention
- 14.12 she feels selfish in making the decision
- 14.17 comparison with others with MFS
- 14.20 the strength of the relationship
- 14.23 she wants to be pregnant
- 14.26 acceptance of uncertainty
- 14.29 the gamble of pregnancy with MFS
- 14.32 he is influenced by others opinions
- 14.41 he feels emotion but mostly hides it
- 14.46 wants to protect wife and be strong and stoic for her
- 14.50 he bottles up emotion
- 15.1 she feels guilt re child and MFS
- 15.8 he is prepared for baby's diagnosis of MFS
- 15.12 unconditional love for child in decision-making
- 15.15 she is determined to have natural pregnancy
- 15.18 he goes along with the decision
- 15.24 he is fact driven, informed, practical
- 15.31 she wants partner to own decision
- 15.34 her urgency of time and age
- 15.37 deciding a cut off point in decision

**APPENDIX B CONTINUED**

- 15.39 she does not want to take full responsibility in decision
- 15.42 it's a female decision!
- 15.45 she wants him to own decision
- 15.48 he is compliant?
- 16.14 he feels talking through decision with a professional may have been helpful
- 16.18 in hindsight he recognises the emotion he felt
- 16.22 decision as informed v trusting instincts
- 16.25 she does not see counselling as helpful for her
- 16.28 the experience of other MFS couples as potentially helpful
- 16.35 she has made her decision
- 16.37 the options are explored for him
- 16.45 she wants him to make informed decision
- 16.48 she fears repercussions and blame
- 17.5 he is aware of her guilt
- 17.8 they consider situation of 'who's left holding the baby'
- 17.12 he needs to work through options in WLHTB situation
- 17.16 her concern is for baby
- 17.19 his concern is for her health
- 17.23 proving her ability to have children
- 17.25 she feels insecure in the relationship without children
- 17.38 survival of his wife as more important than baby
- 17.43 he shares the MFS experience with her
- 17.46 he has concerns re impact of pregnancy on her body
- 17.49 the strength of the relationship
- 18.3 he wants to share the experience of pregnancy
- 18.7 he copes with anxiety and concerns re wife's wellbeing
- 18.11 coping with uncertainty and the unknown
- 18.20 he is stoic and holds back his emotion
- 18.23 he has emotional build up until it overflows
- 18.26 he feels insecure re losing her
- 18.28 he has emotional on/off switch
- 18.34 he is more concerned about her than baby
- 18.36 she is more concerned re baby
- 18.43 he does not share his concerns with others
- 19.2 he does not share feelings with his family
- 19.17 he shares his concerns with her mother as they have common interest
- 19.30 generally he copes alone
- 19.33 she feels he would have benefitted from talking to a professional
- 19.37 he would like to have talked through his concerns with professional
- 19.41 coping in their own worlds
- 19.44 she does not share her fears with husband
- 20.1 he feels the need to open up with professional help
- 20.6 he copes alone to protect her
- 20.9 he needs sounding board but knowledge of MFS not essential
- 20.14 he sees value of talking through the emotional issues with professional
- 20.17 medics as providing information on MFS
- 20.21 she plans for others to be 'left holding the baby' if needed
- 20.26 maternal instincts for survival of child

## APPENDIX B CONTINUED

- 20.31 her concerns of WLHTB cause anxiety in husband and family
- 20.38 they protect each other from upset
- 20.40 coping with her mums concerns re her pregnancy
- 20.45 the altruistic nature of being a MFS mum
- 20.49 she plans for care of baby if she is dead
- 21.1 she wants baby to be safe if she dies
- 21.8 he has thoughts of being left holding the baby
- 21.10 he can't imagine working and caring for baby
- 21.15 losing his wife means coping with loss of her and responsibility of baby
- 21.20 she rationalises her dying by giving other causes of death
- 21.24 he draws on past experience of her health scares
- 21.29 he fears pregnancy as 'a step too far' for her
- 21.37 the value of her mother as a support for him
- 21.47 the strength of the relationship
- 21.48 she wants him to agree decision
- 21.49 she feels decision is asking a lot of him
- 22.1 she does not want sole responsibility of decision
- 22.5 she has to consider the facts as worst-case scenario
- 22.9 she tests his love and commitment to her and decision
- 22.18 he sees reality of her MFS due to health scares
- 22.23 she feels it is easier for her to cope than others around her
- 22.29 her mother helps him to cope with MFS
- 22.33 stoic coping of her mother and husband
- 22.35 her frustration at coping with MFS
- 22.37 her strive for independence
- 22.40 she feels emotional during the decision-making process
- 22.42 she feels calm after decision is made
- 22.46 making the decision creates certainty and feeling of control
- 23.1 ignorance is bliss
- 23.3 the need to normalise MFS in self and child
- 23.49 she does not share decision-making with friends
- 24.7 he shares some concerns with her friend's husband
- 24.25 friends as supportive in a listening role
- 24.41 she feels selfish and envious re friends' pregnancies
- 24.43 coping with emotions caused by other pregnancies
- 24.48 she checks out his feelings re decision
- 25.1 she has concerns she may deprive his of a child
- 25.7 he attempts to dismiss problems and cope in the here and now
- 25.10 he hides his concerns
- 25.10 balancing out the concerns in the relationship
- 25.17 she needs proof of his love and commitment
- 25.22 she does not want to carry responsibility of decision
- 25.28 the strength of the relationship
- 25.31 he understands that she wants him to make an informed decision
- 25.38 decision for him is a problem to be solved
- 25.41 problem solving - she is 'top down' and he is 'bottom up' processing
- 26.1 a massive decision
- 26.5 he is driven by competition in solving the problem of making a decision

**APPENDIX B CONTINUED**

26.9 she is driven by determination to prove her decision is OK

26.13 she wants to prove herself as a woman

26.36 the interview a highlighting emotional process

26.41 he finds interview as:

a. helpful exploring the experience of making the decision

b. validating the emotional aspect of the decision

27.7 she feels guilty and selfish re having baby with MFS

27.17 they make quicker decision to have second baby

27.32 the importance of age and timing

27.36 she assesses risks to self but highlights determination to go ahead anyway

28.3 accepting the role of fate

## APPENDIX C

### WHO'S LEFT HOLDING THEBABY? EXPLORING COUPLES' DECISION-MAKING FOLLOWING A MATERNAL DIAGNOSIS OF MARFAN SYNDROME.

Emerging themes arranged in **clusters** with page and line numbers

#### **Interview 2 – Clare and James**

##### ADAPTING

- ad 1.14 he accepts MFS
- ad 11.44 accepting limitations to number of pregnancies
- ad 14.26 acceptance of uncertainty
- ad 15.12 unconditional love for child in decision-making
- ad 15.8 he is prepared for baby's diagnosis of MFS
- ad 17.8 they consider situation of 'who's left holding the baby'
- ad 18.11 coping with uncertainty and the unknown
- ad 22.46 making the decision creates certainty and feeling of control
- ad 22.5 she has to consider the facts as worst-case scenario
- ad 23.3 the need to normalise MFS in self and child
- ad 25.7 he attempts to dismiss problems and cope in the here and now
- ad 26.1 a massive decision
- ad 3.11 she is prepared for being mother to MFS child
- ad 3.22 rationalising MFS
- ad 4.18 he accepts MFS
- ad 5.22 understanding the meaning of having a child with MFS
- ad28.3 accepting the role of fate

##### ASSESSMENT

- as 1.33 IVF as risky to mother
- as 1.36 deciding on natural route
- as 1.41 deciding to let fate take its course
- as 1.48 comparison of MFS to other medical conditions
- as 10.24 exploring the options of having a child
- as 10.28 child considered as they explore the options
- as 11.16 a massive decision
- as 11.25 adoption as a last resort
- as 12.49 coping with high-risk pregnancy
- as 13.13 coping with risk of who's left holding the baby
- as 13.18 coping with the reality of child with MFS
- as 13.9 coping with reality of high-risk pregnancy
- as 14.1 intensity of the process of decision-making
- as 14.29 the gamble of pregnancy with MFS
- as 15.34 her urgency of time and age
- as 15.37 deciding a cut off point in decision
- as 17.12 he needs to work through options in WLHTB situation
- as 17.16 her concern is for baby
- as 17.19 his concern is for her health
- as 18.36 she is more concerned re baby
- as 2.22 assessing the risks

## APPENDIX C CONTINUED

as 2.27 the importance of timing  
 as 2.3 leaving things to fate  
 as 2.38 natural route is her decision  
 as 2.44 she feels medical intervention in conceiving is chemical, stressful, unethical and 'playing God'  
 as 2.45 if her mother had genetically tested her she would not be here  
 as 2.9 the importance of timing  
 as 21.24 he draws on past experience of her health scares  
 as 21.29 he fears pregnancy as 'a step too far' for her  
 as 22.18 he sees reality of her MFS due to health scares  
 as 27.17 they make quicker decision to have second baby  
 as 27.32 the importance of age and timing  
 as 27.36 she assesses risks to self but highlights determination to go ahead anyway  
 as 3.39 evaluating the risks to her  
 as 3.49 concerns re suitability for adoption  
 as 4.16 decision not 'if' but 'when and how'  
 as 4.2 making the decision final  
 as 4.40 assessing the risks of genetic testing  
 as 4.46 genetic testing rejected as would not abort anyway  
 as 5.7 the need to make decision pre-conception  
 as 6.9 the role of fate  
 as 7.19 importance of timing  
 as 7.34 timing and risk to mother  
 as 8.21 assessing the cost to the child  
 as 9.20 she compares mother's situation with her own  
 as 9.45 assessing the role and expectations of their parents  
 as 9.47 helpful exploring the experience of making the decision

### INFORMATION

I 1.13 she wants him to know the facts re MFS  
 I 1.18 seeks professional advice re having children  
 I 1.30 exploring the options  
 I 10.32 exploring options as 'peace of mind'  
 I 13.27 MFS information day as helpful  
 I 13.30 the internet as research tool for MFS  
 I 16.22 decision as informed v trusting instincts  
 I 16.37 the options are explored for him  
 I 2.17 looking at options  
 I 2.19 seeking professional medical help  
 I 2.25 making an informed decision  
 I 2.31 medics approval as important in decision  
 I 2.41 he needs to explore options  
 I 20.17 medics as providing information on MFS  
 I 23.1 ignorance is bliss  
 I 25.31 he understands that she wants him to make an informed decision  
 I 25.38 decision for him is a problem to be solved  
 I 25.41 problem solving - she is 'top down' and he is 'bottom up' processing  
 I 3.13 her experience of MFS as an asset  
 I 3.14 sharing the experience of MFS as a child with partner

## APPENDIX C CONTINUED

I 3.18 the reality of MFS  
 I 3.20 comparison of MFS with other illnesses  
 I 3.36 exploring all the options  
 I 3.44 exploring all the options  
 I 4.18 she wants him to know facts re MFS  
 I 4.35 genetic counselling and information and giving options  
 I 4.5 importance of medical approval  
 I 5.25 he is optimistic re medical advancement  
 I 5.33 she is optimistic re medical advances  
 I 5.40 keeping informed re MFS  
 I 7.10 genetic testing NHS v private  
 I 7.2 doing their own research  
 I 9.25 optimism re medical advancement

### IDENTITY

id 11.10 her emotions are up and down  
 id 11.22 her mind is made up from an early age  
 id 11.30 her drive to have child overrides everything  
 id 12.9 he compares self with own father  
 id 13.22 guilt of having a MFS child  
 id 13.5 her fear of loss for self and baby  
 id 14.12 she feels selfish in making the decision  
 id 14.23 she wants to be pregnant  
 id 14.41 he feels emotion but mostly hides it  
 id 14.46 wants to protect wife and be strong and stoic for her  
 id 14.50 he bottles up emotion  
 id 15.1 she feels guilt re child and MFS  
 id 15.15 she is determined to have natural pregnancy  
 id 15.18 he goes along with the decision  
 id 15.24 he is fact driven, informed, practical  
 id 15.42 it's a female decision!  
 id 16.18 in hindsight he recognises the emotion he felt  
 id 16.35 she has made her decision  
 id 17.23 proving her ability to have children  
 id 18.20 he is stoic and holds back his emotion  
 id 18.23 he has emotional build up until it overflows  
 id 18.28 he has emotional on/off switch  
 id 20.26 maternal instincts for survival of child  
 id 20.45 the altruistic nature of being a MFS mum  
 id 20.6 he copes alone to protect her  
 id 21.10 he can't imagine working and caring for baby  
 id 21.8 he has thoughts of being left holding the baby  
 id 22.35 her frustration at coping with MFS  
 id 22.37 her strive for independence  
 id 22.40 she feels emotional during the decision-making process  
 id 22.42 she feels calm after decision is made  
 id 24.41 she feels selfish and envious re friends' pregnancies  
 id 24.43 coping with emotions caused by other pregnancies  
 id 25.1 she has concerns she may deprive him of a child

**APPENDIX C CONTINUED**

id 25.10 he hides his concerns  
 id 26.13 she wants to prove herself as a woman  
 id 26.5 he is driven by competition in solving the problem of making a decision  
 id 26.9 she is driven by determination to prove her decision is OK  
 id 27.7 she feels guilty and selfish re having baby with MFS  
 id 3.1 her decision is personal  
 id 3.3 his decision is detached and pragmatic  
 id 3.32 an emotional decision for her  
 id 5.1 he is supportive role  
 id 5.10 she feels guilt re MFS in making decision  
 id 5.20 she feels responsible for passing on the MFS gene  
 id 5.33 he is pragmatic  
 id 5.40 his internal v external locus of control over events  
 id 515 she owns her MFS  
 id 8.34 comparison of self with her mother  
 id 8.37 the value of being a Mum with MFS and caring for a child with MFS  
 id 8.46 the value of past experience  
 id 9.11her insight makes being a mother to MFS child less scary  
 id 9.31he is pragmatic  
 id 9.32 she is emotion  
 id 9.33 the child as a person not an illness  
 id 9.5 she has insider knowledge in dealing with child's medical issues

**RELATIONSHIP**

r 1.10 sharing MFS with each other  
 r 1.15 she tests his love of her  
 r 1.40 having opposing perspectives  
 r 1.8 sharing doubt re having children  
 r 10.36 she wants him to own his part of decision  
 r 10.39 she checks out how he feels about the decision  
 r 10.42 she needs to share guilt re MFS to prevent repercussions from him  
 r 11.13 he empathises with her  
 r 11.38 decision to have children made early in relationship  
 r 14.20 the strength of the relationship  
 r 14.7 the romance of decision is diminished by clinical intervention  
 r 15.31 she wants partner to own decision  
 r 15.39 she does not want to take full responsibility in decision  
 r 15.45 she wants him to own decision  
 r 15.48 he is compliant?  
 r 16.45 she wants him to make informed decision  
 r 16.48 she fears repercussions and blame  
 r 17.25 she feels insecure in the relationship without children  
 r 17.38 survival of his wife as more important than baby  
 r 17.43 he shares the MFS experience with her  
 r 17.46 he has concerns re impact of pregnancy on her body  
 r 17.49 the strength of the relationship  
 r 17.5 he is aware of her guilt  
 r 18.26 he feels insecure re losing her  
 r 18.3 he wants to share the experience of pregnancy

**APPENDIX C CONTINUED**

- r 18.34 he is more concerned about her than baby
- r 18.7 he copes with anxiety and concerns re wife's wellbeing
- r 19.41 coping in their own worlds
- r 19.44 she does not share her fears with husband
- r 20.38 they protect each other from upset
- r 21.15 losing his wife means coping with loss of her and responsibility of baby
- r 21.20 she rationalises her dying by giving other causes of death
- r 21.47 the strength of the relationship
- r 21.48 she wants him to agree decision
- r 21.49 she feels decision is asking a lot of him
- r 22.1 she does not want sole responsibility of decision
- r 22.9 she tests his love and commitment to her and decision
- r 24.48 she checks out his feelings re decision
- r 25.10 balancing out the concerns in the relationship
- r 25.17 she needs proof of his love and commitment
- r 25.22 she does not want to carry responsibility of decision
- r 25.28 the strength of the relationship
- r 3.31 his empathy for her
- r 3.33 the strength of the relationship
- r 3.7 they discuss dilemma of decision
- r 4.19 Importance of love and strength of relationship
- r 5.19 they make a joint decision
- r 7.7 making the decision together

**SUPPORT**

- s 10.1 coping with family reactions to decision
- s 10.15 listening to others but owning the decision
- s 11.3 considering their parents opinions in the decision
- s 11.9 her mother as positive re decision
- s 12.35 importance of family support
- s 12.38 exploring options with the family
- s 13.2 coping with the reactions of family
- s 13.35 comparison with others affected by MFS
- s 13.40 coping with others negative perception of choice to have a baby
- s 14.17 comparison with others with MFS
- s 14.32 he is influenced by others opinions
- s 16.14 he feels talking through decision with a professional may have been helpful
- s 16.25 she does not see counselling as helpful for her
- s 16.28 the experience of other MFS couples as potentially helpful
- s 18.43 he does not share his concerns with others
- s 19.17 he shares his concerns with her mother as they have common interest
- s 19.2 he does not share feelings with his family
- s 19.30 generally he copes alone
- s 19.33 she feels he would have benefitted from talking to a professional
- s 19.37 he would like to have talked through his concerns with professional
- s 20.1 he feels the need to open up with professional help
- s 20.14 he sees value of talking through the emotional issues with professional
- s 20.21 she plans for others to be 'left holding the baby' if needed
- s 20.31 her concerns of WLHTB cause anxiety in husband and family

**APPENDIX C CONTINUED**

- s 20.40 coping with her mums concerns re her pregnancy
- s 20.49 she plans for care of baby if she is dead
- s 20.9 he needs sounding board but knowledge of MFS not essential
- s 21.1 she wants baby to be safe if she dies
- s 21.37 the value of her mother as a support for him
- s 22.23 she feels it is easier for her to cope than others around her
- s 22.29 her mother helps him to cope with MFS
- s 22.33 stoic coping of her mother and husband
- s 23.49 she does not share decision-making with friends
- s 24.25 friends as supportive in a listening role
- s 24.7 he shares some concerns with her friend's husband
- s 26.36 the interview a highlighting emotional process
- s 26.41 he finds interview as:
  - s 6.40 the offer of counselling vague – opposing memories of availability
  - s 6.5 coping with other MFS couples' negative views on having children
  - s 8.11 the importance of family support
  - s 8.16 the effects of her parents wishes on the decision
  - s 8.25 drawing on her mother's experience of caring for her as a child
  - s 8.27 highlighting her mother's guilt with her
  - s 8.42 empathy re her mother's guilt and MFS
  - s 9.43 coping with family responses to decision
- S validating the emotional aspect of the decision

## APPENDIX D

### WHO'S LEFT HOLDING THE BABY? EXPLORING COUPLES' DECISION-MAKING FOLLOWING A WOMAN'S DIAGNOSIS OF MARFAN SYNDROME.

#### Interview 2 – Clare and James

Gender CLUSTERS with page and line numbers

m = male

f = female

b = both

#### ASSESSMENT

Both 2

- 2as/b 1.36 deciding on natural route
- 2as/b 1.41 deciding to let fate take its course
- 2as/b 1.48 comparison of MFS to other medical conditions
- 2as/b 10.24 exploring the options of having a child
- 2as/b 10.28 child considered as they explore the options
- 2as/b 11.16: 26.1 a massive decision
- 2as/b 11.25 adoption as a last resort
- 2as/b 12.49: 13.9 coping with high-risk pregnancy
- 2as/b 13.13 coping with risk of who's left holding the baby
- 2as/b 13.18 coping with the reality of child with MFS
- 2as/b 14.1 intensity of the process of decision-making
- 2as/b 14.26 acceptance of uncertainty
- 2as/b 14.29 the gamble of pregnancy with MFS
- 2as/b 15.37 deciding a cut off point in decision
- 2as/b 17.8 they consider situation of 'who's left holding the baby'
- 2as/b 18.11 coping with uncertainty and the unknown
- 2as/b 2.22 assessing the risks
- 2as/b 2.9/27: 7.19/34: 27.32 the importance of timing and risk to mother
- 2as/b 2.3: 6.9: 28.3 accepting the role of fate
- 2as/b 22.46 making the decision creates certainty and feeling of control
- 2as/b 27.17 they make quicker decision to have second baby
- 2as/b 3.22 rationalising MFS
- 2as/b 3.39 evaluating the risks to her
- 2as/b 3.49 concerns re suitability for adoption
- 2as/b 4.16 decision not 'if' but 'when and how'
- 2as/b 4.2 making the decision final
- 2as/b 4.40 assessing the risks of genetic testing
- 2as/b 4.46 genetic testing rejected as would not abort anyway
- 2as/b 5.22 understanding the meaning of having a child with MFS
- 2as/b 5.7 the need to make decision pre-conception
- 2as/b 8.21 assessing the cost to the child
- 2as/b 22.5 she has to consider the facts as worst-case scenario
- 2as/b 23.1 ignorance is bliss
- 2as/b 25.41 problem solving - she is 'top down' and he is 'bottom up' processing
- 2as/b 3.18 the reality of MFS
- 2as/b 3.20 comparison of MFS with other illnesses
- 2as/b 9.33 they view the child as a person not an illness
- 2as/b 9.45 assessing their role and expectations of their parents
- 2as/b 13.22 guilt of having a MFS child

## APPENDIX D CONTINUED

### Female 2

- 2as/f 1.33 IVF as risky to mother
- 2as/f 15.34 her urgency of time and age
- 2as/f 17.16:18.36 her concern is for baby
- 2as/f 2.38 natural route is her decision
- 2as/f 2.44 she feels medical intervention in conceiving is chemical, stressful, unethical and 'playing God'
- 2as/f 2.45 if her mother had genetically tested her she would not be here
- 2as/f 27.36 she assesses risks to self but highlights determination to go ahead anyway
- 2as/f 9.20 she compares mother's situation with her own
- 2as/f 3.13 her experience of MFS as an asset

### Male 2

- 2as/m 15.8 he is prepared for baby's diagnosis of MFS
- 2as/m 17.12 he needs to work through options in WLHTB situation
- 2as/m 17.19 his concern is for her health
- 2as/m 21.24 he draws on past experience of her health scares
- 2as/m 21.29 he fears pregnancy as 'a step too far' for her
- 2as/m 22.18 he sees reality of her MFS due to health scares
- 2as/m 4.18 he accepts MFS

### IDENTITY

#### Female

- id/f 11.10 her emotions are up and down
- id/f 11.22 her mind is made up from an early age
- id/f 11.30 her drive to have child overrides everything
- id/f 13.5 her fear of loss for self and baby
- id/f 14.12 she feels selfish in making the decision
- id/f 14.23 she wants to be pregnant
- id/f 15.1 she feels guilt re child and MFS
- id/f 15.15 she is determined to have natural pregnancy
- id/f 15.42 it's a female decision!
- id/f 16.35 she has made her decision
- id/f 17.23 proving her ability to have children
- id/f 20.26 maternal instincts for survival of child
- id/f 20.45 the altruistic nature of being a MFS mum
- id/f 22.35 her frustration at coping with MFS
- id/f 22.37 her strive for independence
- id/f 22.40 she feels emotional during the decision-making process
- id/f 22.42 she feels calm after decision is made
- id/f 23.3 the need to normalise MFS in self and child
- id/f 24.43 coping with emotions caused by other couples pregnancies
- id/f 25.1 she has concerns she may deprive him of a child
- id/f 26.13 she wants to prove herself as a woman
- id/f 26.9 she is driven by determination to prove her decision is OK
- id/f 27.7 she feels guilty and selfish re having baby with MFS
- id/f 3.1 her decision is personal
- id/f 3.32: 9.32 an emotional decision for her
- id/f 5.10 she feels guilt re MFS in making decision
- id/f 5.15 she owns her MFS
- id/f 5.20 she feels responsible for passing on the MFS gene

## APPENDIX D CONTINUED

id/f 8.34 comparison of self with her mother  
 id/f 3.11: 8.37/46: 9.5 the value of being a Mum with MFS and caring for a child with MFS  
 id/f 9.11 her insight makes being a mother to MFS child less scary

### Male

id/m 14.46 wants to protect wife and be strong and stoic for her  
 id/m 14.50/41: 18.20 he is stoic and bottles up emotion  
 id/m 15.18 he goes along with the decision  
 id/m 15.24 he is fact driven, informed, practical  
 id/m 16.18 in hindsight he recognises the emotion he felt  
 id/m 18.23 he has emotional build up until it overflows  
 id/m 18.28 he has emotional on/off switch  
 id/m 21.10 he can't imagine working and caring for baby  
 id/m 21.8 he has thoughts of being left holding the baby  
 id/m 25.10 he hides his concerns  
 id/m 25.7 he attempts to dismiss problems and cope in the here and now  
 id/m 26.5 he is driven by competition in solving the problem of making a decision  
 id/m 3.3: 5.33 his decision is detached and pragmatic  
 id/m 5.1 he is supportive role  
 id/m 5.40 his internal v external locus of control over events  
 id/m 20.6 he copes alone to protect her  
 id/m 12.9 he compares self with own father

### RELATIONSHIP

#### Both

r/b 1.8 sharing doubt re having children  
 r/b 1.10 sharing MFS with each other  
 r/b 1.40 having opposing perspectives  
 r/b 11.38 decision to have children made early in relationship  
 r/b 14.20: 21.47: 25.28: 3.33: 17.49: 4.19 the importance of love and strength of the relationship  
 r/b 14.7 the romance of decision is diminished by clinical intervention  
 r/b 15.12 unconditional love for child in decision-making  
 r/b 19.41 coping in their own worlds  
 r/b 20.38 they protect each other from upset  
 r/b 25.10 balancing out the concerns in the relationship  
 r/b 3.7 they discuss dilemma of decision  
 r/b 5.19: 7.7 they make a joint decision  
 r/b 3.14 sharing the experience of MFS as a child with partner

#### Female

r/f 1.13: 4.18 she wants him to know the facts re MFS  
 r/f 1.15: 22.9: 25.17 she tests his love and commitment to her and decision  
 r/f 10.36: 15.31 she wants him to own his part of decision  
 r/f 10.39: 24.48 she checks out how he feels about the decision  
 r/f 15.39: 22.1: 25.22 she does not want to take full responsibility in decision  
 r/f 16.45 she wants him to make informed decision  
 r/f 16.48 she fears repercussions and blame  
 r/f 17.25 she feels insecure in the relationship without children  
 r/f 19.44 she does not share her fears with husband

## APPENDIX D CONTINUED

r/f 21.20 she rationalises her dying by giving other causes of death

r/f 21.48 she wants him to agree decision

r/f 21.49 she feels decision is asking a lot of him

r/f10.42 she needs to share guilt re MFS to prevent repercussions from him

Male

r/m 1.14 he accepts MFS

r/m 11.13 he empathises with her

r/m 15.48 he is compliant?

r/m 17.38: 18.14 survival of his wife as more important than baby

r/m 17.43 he shares the MFS experience with her

r/m 17.46 he has concerns re impact of pregnancy on her body

r/m 17.5 he is aware of her guilt

r/m 18.26 he feels insecure re losing her

r/m 18.3 he wants to share the experience of pregnancy

r/m 18.7 he copes with anxiety and concerns re wife's wellbeing

r/m 21.15 losing his wife means coping with loss of her and responsibility of baby

r/m 3.31 his empathy for her

r/m 16.37 the options are explored for him

r/m 25.31 he understands that she wants him to make an informed decision

PROFESSIONAL SUPPORT

Both

ps/b 1.18: 2.19 seeks professional advice re having children

ps/b 11.44 accepting limitations to number of pregnancies

ps/b 13.27 MFS information day as helpful

ps/b 13.30 the internet as research tool for MFS

ps/b 2.25: 16.22 decision as informed v trusting instincts

Ps/b 16.28 the experience of other MFS couples as potentially helpful

Ps/b 19.33 she feels he would have benefitted emotionally from talking to a professional

ps/b 1.30: 2.17: 3.36/44 exploring all the options

ps/b 2.31: 4.5 medics approval as important in decision

ps/b 20.17 medics as providing information on MFS

ps/b 4.35 genetic counselling and information and giving options

ps/b 5.25: 9.25 they are optimistic re medical advancement

ps/b 5.40 keeping informed re MFS

ps/b 6.40 the offer of counselling vague – opposing memories of availability

ps/b 7.10 genetic testing NHS v private

ps/b 7.2 doing their own research

ps/b10.32 exploring options as 'peace of mind'

ps/b 26.42/36 helpful exploring the experience of making the decision in the interview

Female

ps/f 16.25 she does not see counselling as helpful for her

Male

ps/m 16.14: 19.37 he feels talking through decision with a professional may have been helpful

ps/m 2.41 he needs to explore options

ps/m 20.1/14 he sees value of talking through the emotional issues with professional

ps/m 20.9 he needs sounding board but knowledge of MFS not essential

ps/m 25.38 decision for him is a problem to be solved

ps/m 26.41 he finds interview as validating the emotional aspect of the decision

## APPENDIX D CONTINUED

## SOCIAL SUPPORT

## Both

- Ss/b 9.43: 10.1: 13.2 coping with family reactions to decision
- Ss/b 10.15 listening to others but owning the decision
- Ss/b 11.3 considering their parents opinions in the decision
- Ss/b 11.9 her mother as positive re decision
- Ss/b 8.11: 12.35 importance of family support
- Ss/b 12.38 exploring options with the family
- Ss/b 13.35: 14.17 comparison with others affected by MFS
- Ss/b 13.40 coping with others negative perception of choice to have a baby
- Ss/b 22.33 stoic coping of her mother and her husband
- ss/b 24.25 friends as supportive in a listening role
- ss/b 6.5 coping with other MFS couples' negative views on having children
- ss/b 8.16 the effects of her parents wishes on the decision

## Female

- ss/f 20.21/49: 22.1 she plans for others to be 'left holding the baby' if needed
- ss/f 20.31 her concerns of WLHTB cause anxiety in husband and family
- ss/f 20.40 coping with her mums concerns re her pregnancy
- ss/f 22.23 she feels it is easier for her to cope than others around her
- ss/f 23.49 she does not share decision-making with friends
- ss/f 8.25 drawing on her mother's experience of caring for her as a child
- ss/f 8.27 highlighting her mother's guilt with her
- ss/f 8.42 empathy re her mother's guilt and MFS
- ss/f 24.41 she feels selfish and envious re friends' pregnancies

## Male

- ss/m 14.32 he is influenced by others opinions
- ss/m 18.43 he does not share his concerns with others
- ss/m 19.17 he shares his concerns with her mother as they have common interest
- ss/m 19.2 he does not share feelings with his family
- ss/m 19.30 generally he copes alone
- ss/m 21.37: 22.29 the value of her mother as a support for him
- ss/m 24.7 he shares some concerns with her friend's husband

## APPENDIX E

### WHO'S LEFT HOLDING THE BABY? EXPLORING COUPLES' DECISION-MAKING FOLLOWING A WOMAN'S DIAGNOSIS OF MARFAN SYNDROME.

#### Interview 2 – Clare & James

#### TABLE OF SUPERORDINATE THEMES AND THEMES

Themes	Page/line	Key words
<b>A. HER DECISION IS ALREADY MADE!</b>		
A predetermined decision	11.22	I knew from early
age		
Female drive and identity	11.32	I wanted children
Urgency of timing and age	15.33	time's dragging on
MFS as strength and resilience	26.11	I have got to prove
myself		
Comparison with past self	3.11	I have been
through it		
Comparison with her parents	9.11	I'm not scared
<b>B. CREATING AN INFORMED DECISION</b>		
She wants him to know the facts	25.28	no skeletons or
surprises		him to
Sharing responsibility for decision	16.48	
blame me		
Coping with doctors	4.5	medical all clear
The meaning of genetic counselling	4.35	gave us our
options		
The role of the internet	13.30	on internet for
hours		
Exploring options	3.35	could have gone
IVF		
Assessing the risks	2.23	what's the risk
Optimism of medical advancements	5.22	progressively
improved		
<b>C. FINDING SUPPORT</b>		
The effects of her mother's experience	11.9	she focussed on
positives		
The strength of the relationship	4.15	run for the hills if
you want		
Temporary parallel lives	19.41	I was really scared
as well		
Dynamics in the family	19.1	don't really talk
Limitations of friends' support	24.28	they just listened
The role of support groups	16.27	spoken to a couple
Psychological counselling as helpful for him	16.14	a benefit for us
The interview as a therapeutic experience	26.36	a lot more
emotional		
The essence of helpful professional support	20.9	having a sounding
board		

## D. EXISTING WITH THE FALLOUT OF DECISION

Coping with high-risk pregnancy	17.46	impact on her body
Her concern is for the child	13.7	save the baby
Breaking the news to a child issues	8.35	teenager having
He fears losing her about you	18.33	more concerned
He copes and cries alone emotions	18.20	eruption of
Considering 'Who's left holding the baby?' myself	20.14	holding a baby
Feeling responsible, selfish, guilty	15.2	I am so sorry
Acceptance of gamble, uncertainty, fate	14.28	luckiest bloke

## APPENDIX F

## MASTER TABLE OF THEMES FOR THE GROUP

## A. HER DECISION IS ALREADY MADE! (pink)

*A predetermined decision (D)*

Belle and Adrian	always been set	14.47
Clare and James	I knew from an early age	11.22
Jack and Joan	from an early age	1.36
Theresa and Tom	in my future for sure	4.15
Carl and Cathy	different wants and needs	1.16
Lorraine and Bob	don't want kids!	5.40

*Female drive and identity (I)*

Belle and Adrian	very female desire	20.32
Clare and James	I wanted children	11.32
Jack and Joan	want to have this child	10.8
Theresa and Tom	it's the desire to have kids	10.20
Carl and Cathy	feel useless	17.18
Lorraine and Bob	nothing stopped us	1.17

*Urgency of timing and age (T)*

Belle and Adrian	a progressive disorder	1.23
Clare and James	times dragging on	15.33
Jack and Joan	perfect time	15.10
Theresa and Tom	down to the crunch	9.2
Carl and Cathy	had to push on	9.20
Lorraine and Bob	a huge factor	20.14

*MFS as strength and resilience (S)*

Belle and Adrian	I am not bothered	21.43
Clare and James	I have got to prove myself	26.11
Jack and Joan	want to do it	22.4
Theresa and Tom	if it's MFS we're Ok	4.28
Carl and Cathy	gift of life	6.13
Lorraine and Bob	I carried a gene	6.4

*Comparison with past self (CS)*

Belle and Adrian	A well childhood	3.13
Clare and James	I have been through it	3.11
Jack and Joan	you are not having children	15.32
Theresa and Tom	diagnosed and going to die	4.49
Carl and Cathy	never told it was MFS	11.35
Lorraine and Bob	never affected me	6.27

## APPENDIX F CONTINUED

**B. CREATING AN INFORMED DECISION (yellow)***She wants him to know the facts (F)*

Belle and Adrian	what I'm up against	7.18
Clare and James	no skeletons or surprises	25.28
Jack and Joan	100% honest	1.8
Theresa and Tom	implications genetic	1.19
Carl and Cathy	'we' didn't realize	1.24
Lorraine and Bob	he doesn't have a clue	7.14

*Sharing responsibility for decision (DEC)*

Belle and Adrian	adoption without doubt	20.20
Clare and James	him to blame me	16.48
Jack and Joan	'we' discussed	1.17
Theresa and Tom	in denial	11.18
Carl and Cathy	not me with marfans	1.38
Lorraine and Bob	she would tell me	22.25

*Coping with doctors (E)*

Belle and Adrian	don't see benefit	9.1
Clare and James	medical all clear	4.5
Jack and Joan	explained a little more	9.37
Theresa and Tom	lack of knowledge	6.17
Carl and Cathy	little he knew of marfan	8.27
Lorraine and Bob	not even doctors know	11.30

*The meaning of genetic counseling (G)*

Belle and Adrian	don't include me	6.48
Clare and James	gave us our options	4.35
Jack and Joan	all the information	4.30
Theresa and Tom	it's quite clinical	16.40
Carl and Cathy	never knew it existed	6.45
Lorraine and Bob	not offered	6.19

*The role of the internet (INT)*

Belle and Adrian	-	
Clare and James	on internet for hours	13.30
Jack and Joan	worst-case scenario	16.4
Theresa and Tom	get horrified	18.49
Carl and Cathy	start me worrying	18.4
Lorraine and Bob	panics people	16.39

## APPENDIX F CONTINUED

*Exploring options (OP)*

Belle and Adrian	rather adopt	3.39
Clare and James	could have gone IVF	3.35
Jack and Joan	may have to adopt	8.22
Theresa and Tom	not consider aborting	16.45
Carl and Cathy	certain genetic problems	8.47
Lorraine and Bob	-	

*Assessing the risks to child (R)*

Belle and Adrian	50/50 chance born healthy	7.28
Clare and James	what's the risk	2.23
Jack and Joan	chances 55% hesitant	7.14
Theresa and Tom	50/50 chance	1.28
Carl and Cathy	gamble with child's life	2.22
Lorraine and Bob	kids not worse than me	1.9

*Optimism of medical advancements (MED)*

Belle and Adrian	they can fix anything	23.16
Clare and James	progressively improved	5.22
Jack and Joan	not as ignorant	3.43
Theresa and Tom	good quality of life	5.7
Carl and Cathy	in another twenty years	2.29
Lorraine and Bob	years ago 'what's that?'	5.19

**C. FINDING SUPPORT (green)***The effects of her mother's experience (ME)*

Belle and Adrian	doesn't want us to try	10.21
Clare and James	she focused on positives	11.9
Jack and Joan	never have children	1.18
Theresa and Tom	my wife's mum pushed me	7.30
Carl and Cathy	keep things from her	11.22
Lorraine and Bob	I could speak to her	12.34

*The strength of the relationship (SR)*

Belle and Adrian	love's a powerful thing	1.40
Clare and James	run for the hills if you want	4.15
Jack and Joan	us against the world	22.45
Theresa and Tom	part/parcel relationship	3.45
Carl and Cathy	she is my soul-mate	12.17
Lorraine and Bob	I will tell him	18.16

**APPENDIX F CONTINUED***Temporary parallel lives (PL)*

Belle and Adrian	I feel alone	13.49
Clare and James	I was really scared as well	19.41
Jack and Joan	use your strength	12.26
Theresa and Tom	kept it to myself	11.8/25
Carl and Cathy	lull in communication	18.37
Lorraine and Bob	bloke's the strong one	14.40

*Limitations of friends support (FR)*

Belle and Adrian	don't think they realize	12.21
Clare and James	they just listened	24.28
Jack and Joan	what advice can they offer	17.8
Theresa and Tom	not chatted to friends	8.44
Carl and Cathy	they were philosophical	13.2
Lorraine and Bob	I have a close friend	17.29

*The role of support groups (SG)*

Belle and Adrian	other people not accessible	9.34
Clare and James	spoken to a couple	16.27
Jack and Joan	MFS patient who had child	18.7
Theresa and Tom	contact with a couple	17.20
Carl and Cathy	people in the same situation	19.23
Lorraine and Bob	not offered anything	22.2

*Psychological counseling as potentially helpful (PC)*

Belle and Adrian	a great help	9.28/8.11
Clare and James	a benefit for us	16.14
Jack and Joan	seems like a weakness	11.45
Theresa and Tom	no need for third party	17.12
Carl and Cathy	should be available	15.38
Lorraine and Bob	-	

*The interview as a therapeutic experience (INT)*

Belle and Adrian	nice to have intermediate	24.12
Clare and James	a lot more emotional	26.36
Jack and Joan	-	
Theresa and Tom	like we are talking to you	18.31
Carl and Cathy	OK to talk about stuff	24.25/41
Lorraine and Bob	nice to have somebody	23.18

## APPENDIX F CONTINUED

**D. EXISTING WITH THE FALLOUT OF DECISION (blue)***Coping with high-risk pregnancy (HR)*

Belle and Adrian	from day one a team	21.17
Clare and James	impact on her body	17.46
Jack and Joan	time scans testing	11.1
Theresa and Tom	things can go wrong	14.43
Carl and Cathy	wife at risk tough horrible	14.5
Lorraine and Bob	wow bloody hell	4.4

*Her concerns are for child (LC)*

Belle and Adrian	worse than me	3.3
Clare and James	save the baby	13.7/20.14
Jack and Joan	child not myself	23.40
Theresa and Tom	not play sport	10.48
Carl and Cathy	I could have given her MFS	4.12
Lorraine and Bob	really shocked	21.25

*He fears losing her (LH)*

Belle and Adrian	wheel you around	14.25/22.29
Clare and James	more concerned about you	18.33/20.14
Jack and Joan	never forgive myself	8.35
Theresa and Tom	her the priority	11.33
Carl and Cathy	scary fucking moron	16.20
Lorraine and Bob	stress on her heart	10.3

*He copes and cries alone (CA)*

Belle and Adrian	cry in the shower	19.46
Clare and James	eruption of emotions	18.20/14.40
Jack and Joan	on my own crying	23.20
Theresa and Tom	in a kind of bubble	15.20
Carl and Cathy	terrified to the core	14.34
Lorraine and Bob	bottle things up	4.22

*Feeling responsible, selfish, guilty (G)*

Belle and Adrian	seems selfish even	20.45
Clare and James	I am so sorry	15.2
Jack and Joan	I had let it happen	5.1
Theresa and Tom	-	
Carl and Cathy	selfish tough guilty	5.8
Lorraine and Bob	I'm all right Jack	12.18

*Acceptance of gambling, uncertainty, fate (AC)*

Belle and Adrian	big step into the unknown	11.26
Clare and James	luckiest bloke	14.28
Jack and Joan	accept those children	4.23
Theresa and Tom	the odds increase	10.1
Carl and Cathy	out of my control	18.1
Lorraine and Bob	you're just guessing	16.

**APPENDIX G**

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**WHO'S LEFT HOLDING THE BABY? EXPLORING COUPLES' DECISION-MAKING  
 FOLLOWING A WOMAN'S DIAGNOSIS OF MARFAN SYNDROME.**

**SUMMARY OF FINDINGS**

The analysis of the data, verbatim transcripts derived from single eighty-minute interviews with six couples, identified four main themes, each of which is supported by a number of super-ordinate themes. A theme or super-ordinate theme was only included if it applied to at least five of the couples.

**A. Her decision is already made!**

*A predetermined decision*  
*Female drive and identity*  
*Urgency of timing and age*  
*MFS as strength and resilience*  
*Comparison with past self*

**B. Creating an informed decision**

*She wants him to know the facts*  
*Sharing responsibility for decision*  
*Coping with doctors*  
*The meaning of genetic counselling*  
*The role of the Internet*  
*Exploring options*  
*Assessing the risks to child*  
*Optimism of medical advancements*

**C. Finding support**

*The effects of her mother's experience*  
*The strength of the relationship*  
*Temporary parallel lives*  
*Limitations of friends' support*  
*The role of support groups*  
*Psychological counselling as potentially helpful*  
*The interview as a therapeutic experience*

**D. Existing with the fallout of the decision**

*Coping with high-risk pregnancy*  
*Her concerns for child*  
*He fears losing her*  
*He copes and cries alone*  
*Feeling responsible, selfish, guilty*  
*Acceptance of gamble, uncertainty, fate*

### **A. HER DECISION IS ALREADY MADE!**

The first theme, 'her decision is already made!' shows how five of the female participants had given some considerable thought at an early age to the prospect of having children later in life. There was some considerable focus during the interviews on the notion that the women believe so strongly that they must have a baby, there was not really a decision to be made for them. From the women's narratives, it appears that their decisions were largely based on their personal longing to have their own baby and fulfil what they considered to be their role as a woman. Over time their age and state of health was 'a huge factor'. Their personal experience of adjusting and adapting to Marfan syndrome gave them strength and resilience to cope with the future. In making their decision they made comparisons with themselves as a child. These were physical and psychological comparisons and provided a measure for them when evaluating the risks for their own child.

#### ***A predetermined decision***

Five out of the six women described how they had given some considerable thought to having children later in life. The interviews captured the determination with which the women achieved their ultimate goal of having a child through natural childbirth. Their determination to have children shone through and, although they acknowledged that they do have some worries, they would not let them stand in their way of having a baby.

#### ***Female drive and identity***

The ability to conceive, carry a child and give birth for most women are essentially what it means to be a female. Many women will identify with the longing that the participants portrayed in their accounts of the passion with which they fought their way through the minefield of potential threats to their goal of having a child. Their descriptions suggested that the desire to have a child was out of their control as a physically-driven phenomenon. When the powerful female drive or desire was fulfilled by having a child, the perception of the risks associated with Marfan syndrome in considering further children was increased. The value of the self as a woman would be diminished considerably if they were unable to produce the child they had decided they wanted. Having a child placed a practical value on them and made them effective as a female. Not becoming pregnant and giving birth to a child would mean a life unfulfilled. Their sense of self and femininity would be threatened.

#### ***Urgency of timing and age***

Having made their decision, the women wanted to become pregnant sooner rather than later. The decision was concrete for these women who had lived through the experience of coping with Marfan syndrome and they resented having to wait. There was a striking difference between the females' determination and as we shall see later, their partners' caution. The women had made their decision and there was a sense of anxiety and frustration as they recounted their readiness to go ahead and cope with the practicalities of their situation. There was often a narrow margin of opportunity for them to become pregnant. The urgency was particularly evident when they considered age and the lengthy process of following the various options to have a baby. Although their husbands were mostly included in the decision-making process, many of the women illustrated their need to eliminate the other options as quickly as possible so that they could proceed with a natural pregnancy.

#### ***MFS as strength and resilience***

The women's decisions were partly based on the strength and resilience they had acquired over the years as they coped with MFS. This suggests that they have already thought through the issues when considering having a baby. This was an admirable quality that pulled them through the decision-making with their husbands. The women often had to convince their husbands, who sometimes expressed quite extreme concerns for their wife's safety that they were able to cope with the risks or any potential obstacles that might stand in their way of having a child. Generally their strength and resilience emanated from their direct and personal experience of Marfan's syndrome as a life-threatening condition.

### *Comparison with past self*

The women drew on their own experience of being a child with Marfan as a benchmark to prove that in making their decision they had thought through the issues that may arise for their own children. They explained that they thought quite positively about their decision and the risk of having a child with MFS. Therefore, one was left with the strong sense that they feel their own child would have a similar experience. They had a realistic personal view of the implications of being a child with Marfan, but having made the decision to have a child they checked out how prepared they would be if their children were to have Marfan. At the same time they tried to objectify their thinking by cross-checking their husband's understanding of the implications of having a child with Marfan.

## **B. CREATING AN INFORMED DECISION**

In the second theme the participants described how they used different sources of information to help them in an attempt to create some certainty as they weighed up the pros and cons in considering childbirth. The women expressed the need to share the facts of Marfan's syndrome with their partners from early on in their relationship and wanted them to understand the risks and implications of having a baby. Having made every effort to ensure that their partners knew the facts, they then encouraged them to share the decision-making, as they didn't want to take sole responsibility for the outcome. They sought advice from various doctors and although there was some reliance on the doctor's 'all clear' to go ahead, there was evidence of a poverty of understanding of Marfan syndrome among the doctors and medical professionals in general. The meaning of genetic counselling was considered and four out of the five couples described their experience. The Internet was a source of information on Marfan syndrome, but the descriptions and implications of the condition also provided a cause for concern. Five out of the six couples described how they explored the different options of having a baby, including adoption and IVF, however this was more for the men's benefit than the women's. They generally assessed the 50/50 risk of having a child affected by Marfan's and were comforted by their optimism that medical knowledge of Marfan's was advancing at a steady and encouraging rate.

### *She wants him to know the facts*

The women wanted their husbands to be aware of the implications of Marfan syndrome. They encouraged them to understand the meaning of coping with a life-threatening condition and what that may mean in pregnancy and childbirth. They had educated themselves to a point where they had a fairly good understanding of Marfan syndrome and what to expect in pregnancy and childbirth, but they also wanted to ensure that their husbands were fully aware of the implications of Marfan syndrome. The women were usually 100% honest with their husbands at the beginning of the relationship, wanting them to know the implications of living with a genetic condition.

### *Sharing responsibility for decision*

Sharing responsibility for the decision was a significant consideration. The women either didn't want to take sole responsibility, as they didn't want to take the blame for any consequences, or conversely, one couple felt that it was acceptable for the woman to take control of the situation and to keep her husband informed. They talked about exploring and researching every option and aspect of having a baby, but the women felt that this was more for their husbands' benefit than theirs, as they didn't want to take the blame for anything.

### ***Coping with doctors***

The couples had positive and negative experiences in visiting doctors. However, they all recounted the various difficulties they were faced with from time to time as they sought medical advice and information to help them with making their decision. There were various reports regarding visits to doctors at the surgery or hospital to seek advice, to have a check-up, to discuss what to expect from a pregnancy, the risks, giving birth, how they will be afterwards. One couple felt that they made their decision without thoroughly considering the implications for giving birth when a woman has Marfan, and told me that they would have found it helpful to have more information about what to expect. Some couples had frustrating experiences with doctors. They mentioned the 'lack of knowledge' of

MFS generally among the doctors. One couple told me that over the years they had learnt to try and work differently with the doctors when the system wasn't working for them. Another couple told me that they had learnt that if you get frustrated with the doctors you 'get nowhere', so they had learnt to 'pull back and come at it from a different angle'. However they could see the lack of knowledge from both their perspective and the doctors and understood that the doctors 'don't come across Marfans that often, so they are not going to know about it'. Another couple were told by a local cardiologist and a gynaecologist that under no circumstances should they have children. They sought specialist Marfan advice, were told to the contrary, and eventually had their children.

### ***The meaning of genetic counselling***

The role of genetic counselling was considered as the participants described how they experienced this service. Four out of the six couples attended an appointment for genetic counselling, it wasn't offered to one couple and another couple were unaware that it existed. They usually had to ask their GP for a referral to a genetic counsellor to help them with their decision. The various reports described the meetings as 'fairly helpful', 'more medical, matter of fact than emotional', 'impersonal', 'I didn't feel included' (husband), 'I didn't understand', 'just giving us our options and what is available', 'medical tests and all men and women thing with circles and squares', 'it's quite clinical and these are the facts'.

### ***The role of the Internet***

The participants described how they searched the Internet for information but generally they did not find it helpful in making their decision. One couple did not have access to the Internet but other participants told me their reactions to the various websites, including NHS Direct, they visited. They felt that the information can 'be quite scary' and 'very upsetting reading that, you know the worst case scenario', 'panics people', 'start me worrying because it said about all the complications'

### ***Exploring options***

Five out of the six couples explained how they explored all the options available to have a baby before they decided on the natural route. They considered adoption, IVF, prenatal genetic diagnosis (PGD) and pre-implantation genetic diagnosis (PIGD). Aborting a foetus was ruled out. This could be quite a frustrating time for some of the couples as they sought to understand the decision from each other's perspectives. It was an emotional time as they considered the options available.

### ***Assessing the risks to child***

The participants considered the risks involved as they tried to make their decision. The dominant risk they considered was the 50/50 risk of their child inheriting the gene. The 50/50 risk was generally acceptable, but it was the males who could be more cautious and consider the risks as more risky than the females. This is further confirmation that the females had already decided that they were going ahead with the pregnancy regardless of risk. This was also an emotional time as some couples considered how a higher risk than 50% may have

made a difference to their decision. Generally though the assessment of risks was more based on the severity of the woman's condition.

#### ***Optimism of medical advancements***

Medical advancements were a major consideration while the participants were considering the outcome of their proposed decision. They expressed how they felt optimistic that the medical knowledge of MFS would continue to improve as it had done since the women were younger. There were reports of '...they can fix things anything these days', 'we are not as ignorant as we were twenty years ago', 'it has progressively improved over the last thirty years',

Generally, the men were concerned about how Marfan's would affect their wife's health in pregnancy and childbirth. The women considered Marfan syndrome as a non life-threatening condition in today's advanced medical conditions, raising the odds in favour of making a decision to have a child. They had a realistic sense of the risk being lower in the present medical environment than it was when they were children. Advances in heart surgery, quality of life and life-expectancy as marginally below that of others without the condition were all discussed in a favourable light and helped to substantiate their decision.

### **C. FINDING SUPPORT**

The participants described the different sources of support they experienced during the decision-making. The women's mothers had an influence on the situation for both the women and their partners. Love and the strength of the couples' own relationship was an overwhelming factor in their support structure. However, for a while, they psychologically lead parallel lives as they employed their individual coping strategies. There was a limitation to the extent to which friends provided support. A support group with other couples who had successfully been through the experience of decision-making was considered as potentially helpful. The role of psychological counselling was explored and viewed as potentially helpful. Interestingly, they described the interview as a therapeutic experience. This section concludes with the couples giving positive and negative evaluations of the health professionals and what they consider to be the essence of helpful support.

#### ***The effects of her mother's experience***

I asked the participants about their relationship with their family and how it was when they were going through the decision to have a baby. They generally did not share the decision with his parents or her father, but the women's mothers all had some effect on the decision-making process. The women's mothers could be supportive, knowledgeable, positive, encouraging and respectful of their daughter's and son-in-laws' situation. However, two of the couples did not share their experience of decision-making with the woman's mother and they felt they had to be stoic, as they had to consider their mother's feelings, fears and concerns regarding Marfan syndrome and pregnancy.

#### ***The strength of the relationship***

On a number of occasions in the interviews the couples demonstrated the extent to which they loved, respected and wanted to be together. Marfan syndrome was part of the relationship and this provided a secure base from which they could make a decision to have a baby. The strength of their relationship was intensively captured in the interviews and the following extracts: 'Love's a powerful thing I guess'; 'I told him he could run for the hills if he wanted'; 'you make a decision using heart and head'; 'it was us against the world'; 'it (Marfans) would have been part and parcel of our relationship for a long time'; 'I don't see it as Marfans I just see my wife.

### ***Temporary parallel lives***

There was evidence that the participants coped in their own individual world at times and did not share their thoughts or concerns with their partners. This was because they did not want to show any vulnerability, become another problem for their partner to deal with or they needed to conserve their strength to cope with their situation. It could be difficult for them to talk to each other about certain things and temporarily withdrawing could give them the opportunity to process any problems and work out the best coping strategy. Honesty was not always the best policy as they temporarily felt that the relationship would benefit from a modicum of independence. This was sometimes because they thought it would end up in a row or because it would cause hurt, or because they felt their partner had their own problems and didn't want to hear or have to cope with their problems.

### ***Limitations of friends' support***

I wondered if the couples talked to their friends when they were making the decision. And if they did what was it like. Although four out of the six couples talked to friends, including close friends, they found that the amount of support they could offer at that time was limited. Friends found it difficult to fully understand Marfan syndrome and the implications of what it was like to make the decision to have a child. Many of their friends already had children, which could leave them coping with the social expectations of others or feeling the odd ones out in their group of friends.

### ***The role of support groups***

Talking to other people in the same situation as them was something they felt would be helpful. The women, in particular, considered that sharing other couples' experiences of making the decision and going through pregnancy and childbirth would be a significant source of support. The Marfan support groups were mostly helpful for those who contacted them. There was an overwhelming need for a personal touch within the support group structure.

### ***Psychological counselling as potentially helpful***

Having established that none of the couples were offered or attended psychological counselling, I asked them if they would have found it helpful. There was a distinct difference between the male and female responses. Five out of the six females felt it would not have been of benefit to them, but they felt it could have been helpful for their husbands to have had some emotional support from a third party at the time they were going through the decision-making process. Five out of the six men initially denied that talking to a third party would have been helpful for them, but on remembering that it was quite an emotional time for them, they then confessed that 'I could talk things through without having to worry', 'a counsellor maybe', 'I think I could certainly have done with counselling', 'could have been of benefit' and 'it should be available'.

### ***The interview as a therapeutic experience***

At the end of each interview I gave the couples the opportunity to share what it was like for them talking about their experiences with me in the interview. They found the interview to be a positive experience. It had been a space to hear what each other had to say and to try and make sense of their thoughts. In talking about their experiences, the memories or the processes were often much more emotional than they had realised at the time.

## **D. EXISTING WITH THE FALLOUT OF DECISION**

The final theme is a powerful exploration of the final stages in the decision-making process. The participants described their thoughts as the focus of their attention shifted from the hypothetical to the practical. In the first sub-theme, living with the physical and psychological consequences of coping with a high-risk pregnancy are explored. The women expressed their concerns for their child's well-being, which overrode the risks to themselves.

The men had an overwhelming fear of losing the woman they love in childbirth and talked about how they coped and often cried alone. The men considered the reality of 'Who's left holding the baby?' as they struggled in coping with the ongoing uncertainty of the risks to their wives in pregnancy and childbirth. Five out of the six couples reported how they sometimes felt responsible, selfish or guilty for putting a child at risk of inheriting Marfan syndrome. However, there was also an acceptance of the 50/50 gamble, uncertainty and fate.

#### ***Coping with high-risk pregnancy***

The participants described the impact of coping with a high-risk pregnancy. It was the men who defined the tension between wanting a child and coping with the risk of their wives being pregnant and giving birth. Often there was much waiting for results and the go-ahead to become pregnant from the medical professionals. Pregnancy had to be 'more cautious and considered'. The women coped stoically with the high risks involved. It was more difficult for the husbands to witness their wives going through a high-risk pregnancy and childbirth.

#### ***Her concerns are for child***

The women were more concerned about their babies than the risks to themselves. There were concerns that they could give the child Marfan syndrome and they expressed their wishes to put the baby first if they were faced with a life-threatening situation during the birth. There were concerns regarding their fears of who would care for the child if the women were unable to do so, and it was important to ensure a safe and secure future for their unborn child.

#### ***He fears losing her***

While the women were altruistically concerned with the risks to their child, all the men expressed a fear of losing their wives. The immense risks to a woman with Marfan syndrome in pregnancy and childbirth were not to be underestimated as the men considered the responsibility of making the decision and the very real possibility of 'worst case scenario'. They imagined what the possible outcome of their decision to have a baby could be, and they were apprehensive about how they would cope if they were left to care for their child.

#### ***He copes and cries alone***

Generally the men 'bottled up' their emotions as their wives coped with the practicalities of pregnancy and childbirth. The purpose of not sharing their feelings with their wives was to protect her from having any additional problems to cope with. This sub-theme contained some of the most heartfelt comments from the men as they struggled to cope with the decision. They explained how they felt at the time: 'preparing for the worst', 'it makes you cry at times', 'just had to bottle it up inside me', 'plenty of tears, floods of tears, 'I was petrified. I was absolutely terrified to the core and the only thing I knew to do was to keep that inside of me'.

#### ***Feeling responsible, selfish and guilty***

The participants all experienced feeling responsible, selfish and guilty. They were keen to have a family and would have found it hard not to have children. However, they also felt that the decision could be somewhat selfish. They wanted to share the experience of life as this gave meaning to their existence as a couple. They would feel responsible and guilty if their child was diagnosed with Marfan's, but they would love and nurture that child whatever the circumstances.

#### ***Acceptance of gambling, uncertainty and fate***

Having decided to go ahead and have a baby, the couples recognised the limitations of making a fully informed and accurate decision. It made sense to define their situation with a generally positive view of the prospective outcome. They acknowledged that the future may not be perfect, but their choice was partly based on what they thought they could cope with at the time. They had assessed the family history and considered the personal, social and emotional consequences of the risks. They demonstrated that the burden of making the

decision was not to be underestimated. Although the women were the leaders in the decision-making, they were both intent on sharing their life with a child. The couples accepted the uncertainty and recognised the impossibility of making a fully informed decision. They embraced the opportunity to make the decision regardless of the risks. The following extract from one of the women summarises the acceptance of gambling, uncertainty and fate that was evident in the other couples' reports of decision-making: 'If you're going to have a child, you're going to have a child. Women all over the world have children and they will accept those children no matter what's wrong with them'.

## APPENDIX H



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Annie Hinchliffe  
DCPsych Doctoral Conversion Programme  
Metanoia Institute

Our ref: 4/08-09

6<sup>th</sup> April 2009

Dear Annie

RE: *'Who's left holding the baby?' Exploring couples' experiences of decision-making following a female diagnosis of Marfan syndrome*

I am pleased to let you know that the above project has been granted ethical approval by Metanoia Research Ethics Committee. If in the course of carrying out the project there are any new developments that may have ethical implications, please inform me as Integrative department representative for the Research Ethics Committee.

Yours sincerely,

A handwritten signature in black ink, appearing to read 'Vanja Orlans', is written over a horizontal line.

Professor Vanja Orlans  
On behalf of: Metanoia Research Ethics Committee