

A Qualitative Study Exploring
the Psychosocial Issues in Patients with
Vascular Ehlers Danlos Syndrome.

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Declaration

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Abstract

Background

Vascular Ehlers Danlos Syndrome (vEDS) is a subtype of Ehlers Danlos Syndrome characterised by thin translucent skin, easy bruising, and an increased risk of sudden death due to vascular rupture. There is minimal research addressing the psychosocial implications of living with vEDS. Therefore, this exploratory qualitative study aimed to investigate the psychosocial impact of living with vEDS, to establish the effects of the condition in everyday life.

Methods

Participants were recruited through the UK vEDS support group Annabelle's Challenge. In-depth semi structured telephone interviews were conducted and analysed using thematic analysis. Participants also received a debrief phone call one month after their interview.

Results

In total, 21 participants were recruited, producing a response rate of 61%. The experiences reported by interviewees generated six dominant themes. Physical limitations to daily life surfaced as an area that significantly impacted individuals. The following five themes of risk perception and existential thoughts, communication and support, responsibility, loss and finally resilience and coping mechanisms also emerged from data analysis. Increased mental health issues were seen amongst the group and the vast majority commented on frustration and lack of knowledge amongst healthcare professionals, along with concern for future generations.

Conclusions

The results suggest that vEDS patients are faced with an abundance of psychosocial issues and permit the need for change within clinical practice in order to support these individuals effectively. Current care for patients could be improved by upskilling genetic counsellors to address the psychosocial side of vEDS via psychotherapeutic approaches. There is scope for various future research projects for vEDS, including the significance of age at diagnosis, quality of life, coping mechanisms, and relationships.

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List of abbreviations

vEDS: Vascular Ehlers Danlos Syndrome

EDS: Ehlers Danlos Syndrome

MFS: Marfan Syndrome

HCM: Hypertrophic Cardiomyopathy

LDS: Loeys-Dietz Syndrome

DASS: Depression Anxiety Stress Scale

TAA: Thoracic Aortic Aneurysm

TGF- β : Transforming growth factor beta receptor

LQTS: Long QT Syndrome

SF-36: Short Form 36 Health Survey

ICD: Implantable defibrillator

hEDS: Hypermobility EDS

AC: Annabelle's Challenge

SMREC: School of Medicine Research Ethics Committee

GDPR: General Data Protection Regulation

JCA: Juvenile Chronic Arthritis

WGS: Whole Genome Sequencing

MDT: Multidisciplinary Team

HCP: Healthcare Professional

A&E: Accident and Emergency

GC: Genetic Counsellor

QoL: Quality of Life

HRQoL: Health Related Quality of Life

PROMS: Patient Reported Outcome Measures

Chapter one: Introduction

I originally discovered this research project during my first year of study, in the list of dissertation topics released by the MSc in Genetic and Genomic Counselling. The project was described as a qualitative study exploring the psychosocial impact of living with Vascular Ehlers Danlos Syndrome (vEDS) and was presented by clinical supervisor Claire Green.

As an individual with a background in human biology, I recognised the physical limitations presented to patients with life limiting conditions and appreciated the importance of understanding the psychological implications from the patient's perspective. Throughout the MSc, I developed an interest in learning about the psychosocial impact of living with a genetic condition and felt challenged to apply this knowledge in a practical setting, such as research for an MSc.

I favoured this research study for three reasons, all of which motivated me to pursue this topic. Firstly, investigating the psychosocial impact of a genetic condition was my initial and most profound motivation for embarking on this research journey. Secondly, the research had not been investigated before and so results from the study would be ground-breaking within the field. Lastly, the project presented the opportunity to conduct a qualitative, patient-centred research project, using data collection methods such as qualitative interviews to acquire information. I wanted to develop my skills and personally interview participants to gain rich data about their lives, as I preferred this technique in comparison to questionnaires and surveys.

From the conversations with Claire Green and my additional research into the condition, I became aware of the life-limiting nature of vEDS, which highlighted the importance of conducting research into this area. Previous research into this field focused on providing an insight into a vEDS patient experience, via comparison with similar conditions such as Marfan Syndrome and Loeys-Dietz. Focusing my dissertation on a specific patient group, motivated me further, as not only would I conduct innovative research, but also offer a platform for the individuals involved to be heard.

Ultimately, identifying the challenges faced by individuals with vEDS, whilst providing insight into their day to day lives has great potential. The outcomes of this study could significantly influence the care and support for this patient group now and in the future. I value the importance of this research project in providing a basis for additional, focused studies to take place in similar areas.

Chapter two: Literature Review

2.1. Background

Ehlers Danlos Syndrome (EDS) is a group of heterogeneous connective tissue disorders that arise due to a defect in collagen (Steinman and Royce, 2003). This literature review focuses specifically on a subtype of EDS known as Vascular Ehlers Danlos Syndrome (vEDS). Those affected by vEDS experience bruising, skin translucency and an increased risk of sudden death due to vascular rupture, resulting in an average lifespan of 49 years old for men and 53 years old for women (Byers *et al.* 2017). This literature review aims to identify and critically evaluate the published evidence regarding the psychological and social impact of living with vEDS and how it affects individual's daily lives. The gaps identified from this review will help formulate a research question which will be addressed later on.

2.2 Literature search methodology

The lack of published literature discussing the psychosocial aspects of living with vEDS was demonstrated in the initial search and produced the results seen in table one below.

Table 1 - Search results from literature review using the terms 'Vascular Ehlers Danlos Syndrome,' 'psychosocial,' 'social,' 'family implications,' 'wellbeing,' 'well-being,' 'occupation' and 'work.'

	Web of Science		PUBMED		Cardiff Library		Google Scholar	
	Overall	Useful	Overall	Useful	Overall	Useful	Overall	Useful
Number of results produced	33	10	2	1	2	1	5	1

The Web of Science database was selected first, which produced the highest number of results related to this topic area. There was an overlap in the literature displayed across additional databases, including PubMed, Cardiff University online library and Google Scholar. Therefore, the table displays the new articles identified across subsequent databases. Citation lists from relevant works were used to detect additional articles and founded the most relevant paper from a symposium. Search results also discovered relevant published research for other forms of EDS.

A thorough review of the EDS literature has not been included, as focusing on other conditions with similar clinical features to vEDS was more relevant. Published literature exploring these conditions was also collated and is discussed within this review.

2.3. Vascular Ehlers Danlos Syndrome

Vascular EDS (vEDS) or EDS IV, is an autosomal dominant disorder caused by mutations in the COL3A1 gene (Zilocchi *et al.* 2007). COL3A1 mutations present several clinical implications including excessive bruising and tissue fragility, predisposing patients to premature arterial rupture, bowel ruptures, aneurysms, and sudden death (Beighton *et al.* 1998). Full body examinations, biochemical testing, and skin biopsies, if required, are used to diagnose what researchers believe to be the most severe subtype of EDS (Paepe *et al.* 2004).

Although a cure for vEDS has not been established, Celiprolol, a cardio selective adrenergic blocking agent can be administered to prevent major complications of the condition. A randomised, open blinded end-point trial investigated the effects of Celiprolol on 53 vEDS patients situated in France and Belgium and concluded that the treatment was effective when compared to control groups (Ong *et al.* 2010). Despite the advantages of a randomised open blind trial in reducing bias, only 62% of participants had a confirmed diagnosis of vEDS. Nevertheless, recent evidence has shown that survival rates amongst 144 vEDS participants were 80.7% in those treated with Celiprolol, compared to 48.5% in those that did not receive treatment (Frank *et al.* 2019).

2.4. Psychosocial impact of Vascular Ehlers Danlos Syndrome

The psychosocial impact of vEDS is too often neglected (Byers *et al.* 2017) and published literature focuses on the psychosocial impact of living with other forms of EDS which have minimal clinical similarities with vEDS.

2.4.1 EDS studies

An early American study investigated the psychosocial impact of living with EDS via questionnaires and interviews with 41 patients. Analysis uncovered three major difficulties individuals faced including pain, complications during pregnancy and restrictions in everyday life due to the fear of injury/bruising (Lumley *et al.* 1994). But the study was conducted over 20 years ago, when the recognition, support and healthcare management for EDS was not as profound as it is today.

Furthermore, semi structured interviews with 17 hypermobile EDS participants (hEDS) were analysed using thematic analysis which generated five main themes displayed in table two below, encapsulating the psychosocial impact of the condition for these individuals (Bennett *et al.* 2018).

Table 2 - Five main themes identified when interviewing hEDS patients about the psychosocial impact of their condition (Bennett *et al.* 2018).

Theme	Description
Healthcare limitations	Lack of awareness from healthcare professionals
Restricted life	Pain, fatigue, instability led to difficulty completing daily activities
Social stigma	Invisible condition, not being able to keep up with family and friends
Fear of the unknown	Not knowing when the next injury was going to occur, no reliable information about their condition
Ways of coping	Accepting condition, building social networks, finding out more about EDS and adapting their lifestyle

Evidently, restrictions to everyday life, and pain are dominant psychosocial issues faced by EDS patients. However, there are distinct differences between EDS and vEDS such as the increased risk of sudden death, possibly eliciting additional psychosocial issues for vEDS patients.

2.4.2 vEDS studies

Published literature addressing the psychosocial challenges specific to vEDS patients is minimal. Of the few studies reported, vEDS is investigated with other patient groups, commonly Marfan Syndrome (MFS) and Loeys Dietz Syndrome (LDS). Connors *et al.* (2012) explored the types of psychosocial adjustment present in patients with LDS, MFS and vEDS. 21 Australian participants were interviewed and completed the Depression Anxiety and Stress Scales (DASS) and COPE questionnaires. The central themes expressed by patients included grief and loss, reaching a resolution, support, and future worries about children. Additionally, patients felt that a lack of connection with their peers was the biggest unmet need that they struggled with. Although the DASS questionnaire has been praised for its validity and is described as a true measure for patient experiences (Buck, 2014), supporting its use within this study, the Australian cohort limits the generalisability of these results to patients in the UK.

Velvin and Johansen (2018) presented innovative research at the Marfan Syndrome and related disorders research symposium in Amsterdam 2018. 22 Norwegian patients, 11 of which had a confirmed diagnosis of vEDS undertook focus group interviews. The findings demonstrated the impact of psychological distress, fatigue and chronic pain in family life and daily function.

Notably, contradictory advice from healthcare professionals and the lack of rehabilitation programmes available for patients, significantly impacted patient's quality of life and involvement in day to day activities. vEDS affected the personal, social and medical aspects of individuals lives, summarised below.

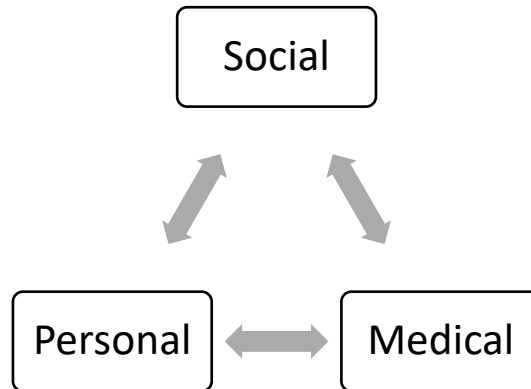


Figure 1 - Cycle displaying the dominant themes that impacted psychosocial wellbeing in patients with MFS, vEDS and LDS, adapted from the findings presented by Velvin and Johansen (2018).

The outcomes of this study are revolutionary in providing an insight into vEDS patients' lives from a psychosocial perspective. Velvin and Johansen (2018) expand on the findings presented by Connors *et al.* (2012), whilst emphasising the differences between vEDS and EDS patients in regard to psychosocial wellbeing, as reported by Bennett *et al.* (2018) and Lumley *et al.* (1994). However, this study has its shortcomings, as the results depict the experiences of LDS, MFS and vEDS patients collaboratively, not differentiating the findings specific to vEDS patients. Additionally, the location in which the study sample is based reduces the generalisability of the results to patients in the UK.

Whilst qualitative research is useful in exploring areas that are not well-understood, findings from such studies are not reliable and require confirmation in a larger, more representative sample. It is therefore unclear whether the results are “statistically significant or due to chance” (Atieno, 2019, pp. 18). Nonetheless, Connors *et al.* (2012) and Velvin and Johansen (2018) have highlighted the range of psychosocial issues patients are met with, whilst bringing light to the importance of consistent support and guidance from healthcare professionals.

2.5. Psychosocial impact of living with Aortopathies

vEDS is part of a group of genetic conditions known as Aortopathies which are characterised by Thoracic Aortic Aneurysms (TAA). vEDS has similarities and strong genetic origins with MFS and LDS (Goyal *et al.* 2017).

Wider research into published literature on associated conditions such as LDS and MFS, is valuable in indicating the possible psychosocial implications that may present in vEDS patients. Similar search terms were used across the databases listed in table one. The addition of ‘Thoracic Aortic Aneurysms’ ‘Loeys Dietz’ and ‘Marfan Syndrome’ were used to formulate relevant literature.

2.5.1 Loeys-Dietz syndrome

Loeys-Dietz syndrome is a connective tissue disorder caused by mutations in the transforming growth factor beta receptor (TGF- β) (Lindsay *et al.* 2012) and is largely characterised by aortic aneurysms and cardiovascular complications like those displayed in vEDS patients.

Literature analysing the psychosocial impact of living with LDS is minimal, with most studies investigating the Aortopathies collectively. MacCarrick *et al.* (2014) explained the potential psychosocial issues that can arise when living with a diagnosis of LDS, including relationships with family and friends, the need for medical assistance and understanding of the condition and feelings of isolation, whilst fearing for the future. Clearly, there are psychosocial similarities between LDS and vEDS as these results mirror those presented by Connors *et al.* (2012) and Velvin and Johansen (2018).

2.5.2 Marfan Syndrome

Marfan syndrome is an inherited connective tissue disorder. The clinical presentation of the condition affects the cardiovascular and skeletal systems, the ocular, dura and lungs (Tinkle and Saal, 2013; Aubart *et al.* 2015). Like vEDS, patients suffering from MFS are at constant risk of aortic rupture that can ultimately lead to sudden death.

Van-Tongerloo and Paepe (1998) conducted semi-structured interviews with 17 MFS patients aged between 17 and 35 years old. They discovered that the condition placed a significant burden on participants daily physical activities, social life, school journey and career. Coping with the diagnosis alongside the importance of accurate information from healthcare professionals, also surfaced as areas of significance.

2080 MFS participants from support groups in Denmark, Belgium, Germany, and France, completed a study-specific questionnaire. 90% of participants felt the condition had a negative influence on their sexual relationships, leading to a negative perception of body image. Furthermore, MFS limited individuals educational and work-related opportunities presenting the possibility that such issues also extend to vEDS patients (De Bie *et al.* 2004). Although, developing a study-specific questionnaire

reduces validity in research, as unlike validated instruments they have not been tested extensively to ensure they are fit for purpose (Jones *et al.* 2013).

Analysis of 13 quantitative and two qualitative articles addressing the psychosocial impact of living with MFS discovered the following common themes: decreased quality of life and challenges in family life, sexual relationships, physical restrictions and education (Velvin *et al.* 2015). Body image continues to be significantly impaired amongst individuals, with recent evidence demonstrating negative body evaluations in both men and women with MFS (Hansen *et al.* 2020). Evidently, the psychosocial impact of living with MFS has not drastically changed over time, supporting previous findings.

Velvin *et al.* (2016) designed a study-specific questionnaire adapted from published and validated questionnaires (table three) to address the specific challenges of MFS. The overall findings indicated that MFS patients have a low quality of life and experience physical limitations due to chronic pain, preventing patients from carrying out day to day activities including work and study. In addition to this, a significant connection between increased levels of fatigue and decreased work participation and quality of life, was identified suggesting that fatigue may be “the most disabling symptom of MFS” (pp. &74).

Table 3 - Study specific questionnaire design, showing the validated questionnaires that were used in order to create the questionnaire for the study.

Topic being assessed in patients	Previous questionnaires the questions were adapted from	Critical appraisal/comments
Work participation	Norwegian Labour Force study	Has not been tested for reliability and validity
Satisfaction with life	Satisfaction with Life Scale	Only measures the cognitive component of satisfaction with life, not the affective component too (Pavot <i>et al.</i> 1991)
Chronic pain	Standardised Nordic Questionnaire	Chronic pain is difficult to measure as its subjective and multifaceted
Fatigue	Fatigue Severity Scale	Has good psychometric properties and is valid and reliable (Whitehead, 2009)

A final conclusive review of 41 published articles exploring the psychosocial aspects of living with MFS was conducted by Nielsen *et al.* (2019) and revealed that irrefutably, MFS negatively impacted patients' quality of life, work participation and reproductive decision-making, supporting the evidence presented in the literature discussed.

The collection of findings reported are valuable in providing insight into the several psychosocial issues patients with aortic diseases face and further expand on the possible psychosocial challenges vEDS patients may be confronted with.

2.6. Psychosocial impact of living with Sudden death disorders

Long QT syndrome (LQTS) and Hypertrophic Cardiomyopathy (HCM) also place patients at risk of sudden death due to a cardiac complication. Appreciating the psychosocial experiences of these patient groups can enhance the knowledge available on the psychosocial issues vEDS patient's experience. The terms 'psychosocial' 'social' 'psychological' 'wellbeing' and 'well-being' alongside 'Long QT Syndrome' and 'Hypertrophic Cardiomyopathy' were used across scientific databases to formulate results.

2.6.1 Long QT syndrome

Long QT syndrome is a congenital condition affecting the repolarisation of the heart, placing patients at risk of life-threatening arrhythmias (Modell and Lehmann, 2006). Those living with the condition are faced with a variety of psychosocial issues that significantly impact their lives. A qualitative study with seven Norwegian adults with LQTS uncovered four main themes displayed in table 4 below. Particularly, participant's main concern was for their children, grandchildren, and future generations (Andersen *et al.* 2008).

Table 4 - Psychosocial issues faced by patients with Long QT syndrome, adapted from Andersen et al. (2008).

Main theme	Subtheme	Subtheme	Subtheme
Positive responses to the diagnosis	Initial relief	Early and gradually acquired information	
Anxiety and worry	Uncertainty and loss of control	Unrelated emotions and dramatic events	Concern for others
Limitations and loneliness	Limitations in daily life	A need for someone with whom to talk	Difficult choices when considering pregnancy and childbirth
Risk and existentiality	A normal and fulfilled life	Death as an acceptable aspect of life	Trust and incompetence in healthcare

Clearly, individuals with LQTS face an abundance of psychosocial issues. However, the inclusion criteria used to recruit participants in this study was not consistent and lacked uniformity resulting in a considerably diverse group of LQTS patients. Identifying the study sample in a reliable and consistent manner is crucial in ensuring that the target population is represented effectively (Garg, 2016) questioning the themes generated, as the individual experiences of the condition differed greatly. Despite this, such findings are invaluable in understanding the possible experiences of vEDS patients.

An additional insight into the experience of living with LQTS was identified in children, who expressed that they felt like outcasts and used images (figure 2) to explain how the condition had affected their lives (Chattha and Zelenietz, 2011). Such findings encourage future research into children's experiences of vEDS, as they may encounter similar troubles.



Figure 2 - Drawing by a young male with LQTS expressing how he feels about having the condition. Adapted from Chatta and Zelenietz (2011).

2.6.2 Hypertrophic Cardiomyopathy

A common cardiac disease with varying age of onset, also known to cause sudden death is Hypertrophic Cardiomyopathy (HCM) where an individual has hypertrophy of their ventricular wall (Christiaans *et al.* 2008).

Health related quality of life was assessed in 137 patients with HCM via the Short Form 36 Health Survey (SF-36) and Hospital Anxiety and Depression Scale. Patients showed to have lowered scores in all dimensions of the SF-36, highlighting the effects of the condition in their physical functioning, emotional problems, mental health, body pain, social functioning, and general health perceptions. Patients also showed increased levels of anxiety and depression compared to the norm (Cox *et al.* 1997). Despite the use of these findings in understanding the experiences of patients with HCM, they were reported over two decades ago, and the progressive acceleration of published scientific literature can lead to the questioning of results from previous papers (Pautasso, 2013).

But the SF-36 questionnaire was employed in a Swedish population of HCM patients with implantable defibrillators (ICD) who scored lower than normal on all areas of the SF-36, apart from bodily pain (Magnusson *et al.* 2016). Although Cox *et al.* (1997) did not specify whether participants had an ICD, the findings here support the notion that HCM patients have low quality of life and face considerable challenges. Additional questionnaires have detected increased levels of anxiety and depression amongst patients suffering from HCM as well as alcohol abuse (Stephoe *et al.*

2000; Ingles *et al.* 2008; Teo *et al.* 2014). The literature indicates that HCM results in increased depression and anxiety, and reduced quality of life which may also be the case for vEDS patients.

Spanaki *et al.* (2016) analysed 152 children with HCM (3-18 years old) and their parents from the UK, using the Paediatric Quality of Life Inventory and the strengths and difficulties questionnaire. Both the children and their parents reported a significantly reduced quality of life when compared to controls. Furthermore, parents informed that the diagnosis of the condition led to the reconsideration of lifestyle choices for their child and family due to the risk of sudden death, showing the significance of understanding how life-threatening conditions affect children and their families. Spanaki *et al.* (2016) reinforce the importance of a multidisciplinary team that includes psychological support, in improving a patients overall understanding of their condition and as a mechanism to prevent further psychosocial maladjustment in the future. Doubtlessly, premature understanding of psychological and social wellbeing is invaluable and could be of benefit to vEDS patients as well.

2.7. Conclusion and importance of my research

The research reviewed here has addressed the psychosocial impact of living with vEDS, through related conditions. Table five summarises the frequent psychosocial issues that surfaced throughout the literature, demonstrating the potential challenges vEDS patients may also face.

Table 5 - Summary of the psychosocial issues and concerns that surfaced throughout this literature review on vEDS and related conditions.

Psychosocial issue/concern	Condition and Reference
Pregnancy/worries about future children	EDS: Lumley <i>et al.</i> (1994) TAA disorders: Connors <i>et al.</i> (2012)
Reproductive decision making	MFS: Nielsen <i>et al.</i> (2019) LQTS: Andersen <i>et al.</i> (2008)
Physical functioning and restrictions to everyday life	EDS: Lumley <i>et al.</i> (1994) LQTS: Andersen <i>et al.</i> (2008) MFS: van-Tongerloo and Paepe (1998) HCM: Cox <i>et al.</i> (1997) MFS: Velvin (2015)
Chronic pain and fatigue	EDS: Lumley <i>et al.</i> (1994) vEDS: Velvin and Johansen (2018) MFS: Velvin (2016) MFS: Hansen <i>et al.</i> (2020) HCM: Cox <i>et al.</i> (1997)
Limitations to social life	MFS: Tongerloo and Paepe (1998) HCM: Cox <i>et al.</i> (1997)
Family life and lifestyle choices	vEDS: Velvin and Johansen (2018) HCM: Spanaki <i>et al.</i> (2015) MFS: Velvin (2015)
Finance	vEDS: Velvin and Johansen (2018)
Sexuality and partnership	vEDS: Velvin and Johansen (2018) MFS: Velvin (2015)
Grief and loss	TAA disorders: Connors <i>et al.</i> (2012) LQTS: (children) Chatta and Zelenietz (2011)
Limitations in working life and study	MFS: Velvin (2016) MFS: De Bie <i>et al.</i> (2004)

	MFS: Tongerloo and Paepe (1998) MFS: Hansen <i>et al.</i> (2020) MFS: Nielsen <i>et al.</i> (2019) MFS: Velvin (2015)
Loneliness/isolation	LQTS: Andersen <i>et al.</i> (2008) LDS: MacCarrick <i>et al.</i> (2014)
Substance abuse	HCM: Teo <i>et al.</i> (2014)
Frustration with healthcare professionals	EDS: Bennett <i>et al.</i> (2018) vEDS: Velvin and Johansen (2018) LQTS: Andersen <i>et al.</i> (2008)
Anxiety and worry	LQTS: Andersen <i>et al.</i> (2008) HCM: Cox <i>et al.</i> (1997) HCM: Steptoe <i>et al.</i> (2000) HCM: Ingles <i>et al.</i> (2008) HCM: Teo <i>et al.</i> (2014)
Fear of the unknown/uncertainty	EDS: Bennett <i>et al.</i> (2018) LDS: MacCarrick <i>et al.</i> (2014) LQTS: Andersen <i>et al.</i> (2008)
Quality of life	MFS: Nielsen <i>et al.</i> (2019) HCM: Spanaki <i>et al.</i> (2016) MFS: Velvin (2015)
Communication (need for support)	LQTS: Andersen <i>et al.</i> (2008)
EDS: Ehlers Danlos Syndrome	
LDS: Loeys Dietz Syndrome	
LQTS: Long QT Syndrome	
HCM: Hypertrophic Cardiomyopathy	
MFS: Marfan Syndrome	
TAA disorders: Thoracic Aortic Disorders	

Noticeably, there is a substantial need for research focused on investigating the psychosocial impact of living with vEDS, specifically. In order to address this gap in the literature, this research project will employ a qualitative approach to explore the question “What psychosocial issues are faced by patients with Vascular Ehlers-Danlos Syndrome?”. This will be the first project to gain an in-depth understanding of the experiences, challenges and difficulties that lead to impaired psychosocial wellbeing in vEDS patients. The outcomes of the project have the potential to influence patient care and support.

2.8. Research question

What psychosocial issues are faced by patients with Vascular Ehlers Danlos Syndrome?

2.9. Study aims and objectives

To effectively answer this research question, this study aims to:

- Identify the psychosocial issues of living with vEDS.
- Establish the day-to-day experience of living with the condition, including its affects in regard to work and study.
- Explore how patients feel in regard to living with a life limiting condition.
- Identify the impact of the condition on patient's mental health.
- Establish the impact of the condition on patients' social lives and desire to socialise.
- Understand how the condition affects relationships with family and friends.
- Address the concerns of patients with affected children.
- Explore individuals' perceptions of having a family.

Chapter three: Methods

3.1 Study design

This research study explored the day to day experiences and challenges of individuals with vEDS, showcasing their viewpoints and opinions, to inform current literature. As the nature of the research question takes an exploratory approach, qualitative methods deemed the most suitable, along with thematic analysis to analyse the data. Qualitative research is praised for “providing valuable insights into the ways we conceptualise health, illness and patients’ experiences” (Chafe, 2017, pp. 13). Despite being criticised due to the quality of information produced, relying on the skill of the researcher and personal bias (Anderson, 2010), qualitative approaches produce thick, detailed descriptions of participants feelings, opinions and experiences (Rahman, 2016).

Conducting semi-structured interviews as opposed to surveys and questionnaires enabled a dynamic, in-depth data collection process. DeJonckheere and Vaughn (2019) support the use of semi-structured interviews and describe the method as a gateway to “delving deeply into personal and sometimes sensitive issues” (pp. 3), highlighting the appropriateness of this technique for an exploratory research project.

Individual telephone interviews provided participants with the flexibility to schedule their interviews around their personal commitments. Although face-to-face interviews were presented as a possibility, telephone interviews were preferred by both parties, due to the additional time and effort a face-to-face interview may require. vEDS is a very rare condition and patients are based throughout the UK, some of which have physical limitations making travel difficult. It was not feasible or financially viable for the researcher to travel to patients to conduct interviews within the time constraints of the project.

3.2 Target sample

This study targeted individuals with a confirmed diagnosis of vEDS that live in the UK. A recruitment advert was shared amongst members of UK vEDS charity support group, Annabelle’s Challenge (AC). At AC, diagnosis is confirmed in members by a screening process that includes an application form, phone call and request for the vEDS diagnosis letter, if they are not known and/or diagnosed by the EDS National Diagnostic Service. The researcher did not have access to participants medical records.

The study aimed to recruit a representative sample of vEDS patients, to ensure participants were of various ages and from a wide geographical demographic in the UK.

Purposive sampling was the chosen method because it enables researchers to identify and select groups of individuals that will provide information-rich data from a limited source or population (Palinkas *et al.* 2013).

Defined inclusion and exclusion criteria (table 6) were used to select the target sample of participants.

Table 6 - Inclusion and exclusion criteria used in recruitment

Inclusion criteria	Exclusion criteria
Confirmed medical diagnosis of vEDS	Children (due to the novelty of the study in investigating adult experiences)
Over the age of 18 years old	Vulnerable adults i.e. those that require additional psychological support or daily care
Capacity to consent	Based outside of the UK

An achievable target sample of 10-15 participants was set, considering the rarity of the condition and the time frame in which the project was taking place. This number of participants was considered large enough to achieve data saturation, in which no new themes or patterns would emerge if further data was collected (O'Reilly and Parker, 2013).

3.3. Ethical procedures

An initial ethical approval application was completed and submitted to Cardiff University School of Medicine Research Ethics Committee (SMREC) for review on 4th May 2019. The application included the research protocol (Appendix A) along with the proposed recruitment materials. A letter was received on 28th May 2019 (Appendix B) requesting minor amendments, including clarification of information in recruitment documents and the inclusion of Cardiff University's General Data Protection Regulations (GDPR) guidance. The study received ethical approval from SMREC on 29th July 2019 (Appendix C). An application to the NHS Research Ethics Committee was not necessary, as this study did not recruit NHS patients, or their relatives/carers.

Cardiff University SMREC was contacted again, to confirm whether the original consent provided by participants covered the use of information from the debrief phone call in the project write up. On 16/04/2020 the committee reported that the consent form covered the use of information from the debrief phone call.

3.4 Recruitment

3.4.1 Recruitment documents

After ethical approval was received recruitment documents were shared with the founder and CEO of AC. A recruitment advert (Appendix D) detailing the research protocol was displayed on the charity website and emailed to potential participants who met the inclusion criteria. Individuals that expressed an interest received a recruitment pack containing an invitation letter (Appendix E), participant information sheet (Appendix F), and consent form with reply slip (Appendix G).

Participants were asked to return consent forms via email to the CEO at AC. Participant information including names and contact details were collated and stored on a password protected excel spreadsheet. The spreadsheet and consent forms in a PDF, were forwarded to the researcher.

3.4.2 Recruitment procedures

Participants that displayed an interest were sent an initial email on 13/09/2019, introducing the project, requesting a convenient date and time for the initial phone call. Potential participants that did not respond were emailed a final time on 07/10/2019. No further emails were sent to those who remained unresponsive, to ensure they did not feel overwhelmed or pressured to take part.

The initial phone call lasted around 5-10 minutes and presented the opportunity to address any questions about the study. A date and time for the interview was also organised and confirmed via email. After each interview, participants were contacted to thank them for taking part in the study and verify the date and time for the debrief phone call. If, at the time of their proposed interview participants did not answer the phone, an email was sent offering the option to rearrange if they wanted to do so.

Around one month after each interview, participants received a debrief phone call to tackle any distressing aspects of the experience, offering individuals the chance to reflect on the process. In published literature, the addition of a debrief phone call in qualitative research methodology, is a strategy used to minimise any emotional distress participants encounter from their interview experience, providing benefit to both participant and researcher (Draucker *et al.* 2009). In this instance, the debrief phone call was extremely beneficial as the reflective process highlighted the positive and negative outcomes of the interview for participants.

3.5 Data collection

Data was collected using semi-structured telephone interviews, conducted by the author.

Telephone interviews have been criticised for the absence of visual cues that would be present in a face-to-face interview, resulting in a loss of contextual and nonverbal data, along with the potential for participants to get distracted in their own environment (Novick, 2008). In spite of this, telephone interviews/counselling have proved to be effective in genetic counselling settings. A large randomised control trial investigated the experiences of genetic counsellors (GC) when delivering telephone or face to face counselling. After conversations with 479 high risk women interested in BRCA1/2 testing, GC's believed the key elements of genetic counselling practice could be delivered effectively regardless of the method (Jacobs *et al.* 2016).

From a patient perspective, women that were eligible for BRCA1/2 testing reported high satisfaction with the telephone interactions they had with a GC (Tutty *et al.* 2019). The evidence presented here, supports the use of telephone interviews within research, as the method has proved to be effective in clinical settings and for research purposes. The method also provides a platform when gathering sensitive information from participants (Lechuga, 2012).

Interviews ranged from 45 minutes to one and a half hours, the majority of which lasted one hour. All interviews were audio recorded and anonymised.

3.6 Data treatment

Participant confidentiality was maintained throughout the data collection process. Numbers were used to classify interviews and any identifiable information was removed from transcripts and shared materials. Interview audio recordings were destroyed after each interview was transcribed. All physical documentation such as consent forms, were sent to Marion McAllister at Cardiff University to be stored in a secure locked cabinet.

The research project conformed to Cardiff University's GDPR throughout, as the recommended key privacy information (table 7) was provided to participants. Participants were signposted to the Cardiff University Data Protection Officer if they had any queries (Appendix E). Data was stored according to the university's data retention scheme <https://www.cardiff.ac.uk/public-information/policies-and-procedures/record-management-policy-and-retention-schedules>

Table 7 - Key privacy information advice from Cardiff University GDPR

	Key privacy information
1	Purpose of the research
2	What is involved when taking part in the research
3	Benefits and risks in participating in the research
4	Details of the research
5	Procedures for withdrawing from the project
6	Planned usage, dissemination, storage, and publishing of the data
7	Strategies for ensuring ethical use of the data
8	Procedures for safeguarding personal data, maintaining confidentiality, and anonymising data (data archiving, sharing and reuse)

3.7 Measures

In order to answer the research question and engage with the exploratory nature of the project, an interview guide was developed and submitted to SMREC for approval (Appendix H). The interview guide included several open questions designed specifically to address the objectives and academic rationale of the project as stated in chapter two, section nine.

The interview guide directed each interview, navigating the researcher and participant through the fundamental questions required. The exploration of certain topics that were not included in the guide, but were relevant in addressing the research question, were also explored.

Interview guides set a standard and maintain focus ensuring comparison can be made from the data produced. Additionally, semi-structured interviews provide an element of flexibility, where further questions can be asked, to elaborate on developing themes (Young *et al.* 2018). However, the supportive evidence presented here is based on the use of interview guides in conservation science research, limiting its application to patient-centred healthcare related projects. Nonetheless, semi-structured interviews achieve an optimum use of time and “keep the interview focused on the desired line of action” (Jamshed, 2014 pp. 87).

3.8 Data analysis

Thematic analysis was employed to analyse the data, using the step-by-step guide (figure 3) designed by Braun and Clarke (2006). Analysis began during the data collection process, which involved familiarising with the data produced from interviews, noting down potential themes and points of

interest. Interview transcription permitted a further in-depth exploration of the data, before NVivo software was used to unpick the data further.

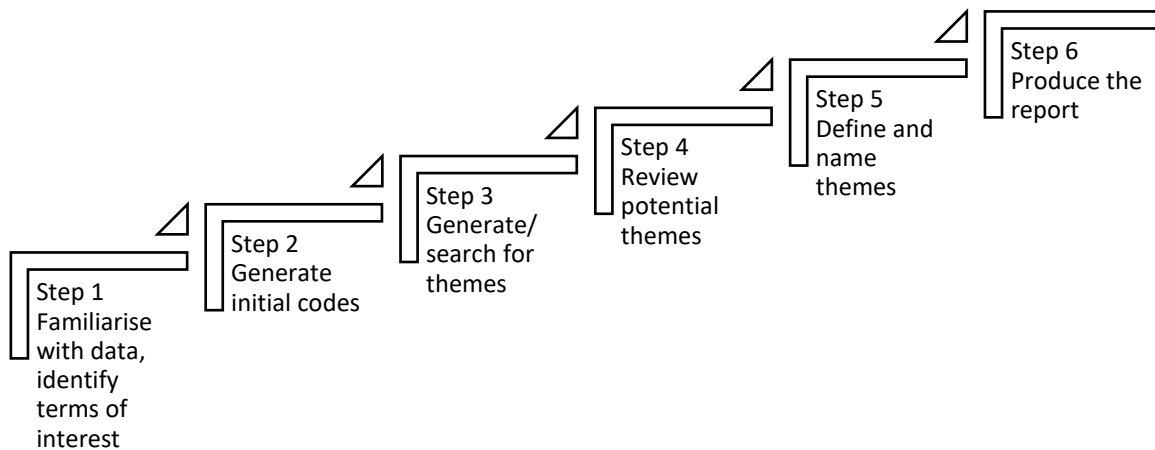


Figure 3 - Adapted from Braun and Clarke (2006), the step by step guide to thematic analysis used in this project.

Each transcript was carefully annotated and coded using short phrases that depicted the main features present within the text. Familiar codes and short phrases were joined, whilst others that described more than one aspect of the data were split. The initial themes produced from data analysis took a categorical approach, dividing the data into groups and subgroups rather than overarching themes. Castleberry and Nolen (2018) suggest an effective way of identifying themes when using thematic analysis, by looking at patterns within the codes and using related codes to “see a bigger picture of what is being portrayed” (pp. 809). But their methodological literature review explores the application of thematic analysis in pharmaceutical education and so cannot account for the use of analysis techniques in healthcare research. In addition to this, it is difficult to draw meaningful conclusions from methodological approaches (Mallet *et al.* 2012). Regardless, thematic analysis is a useful method in novel research, as themes and codes are developed directly from the data within the text, creating representative themes and subthemes (Vaismoradi *et al.* 2013).

Braun and Clarke’s (2006) step-by-step guide was followed throughout, taking into consideration the comments made by Castleberry and Nolen (2018). Additional review of the themes, focused on detailing the topics that ran all through the data, illustrating the key aspects that represent the participants experiences, in answer to the research question. A thematic map was produced and is displayed in figure 5 which helped establish the structure of the results.

Chapter four: Results

4.1 Introduction

The results from recruitment and data analysis will be discussed in this chapter. A description of the study response rate (table 8) and demographics of the sample group have been reported (table 9). The outcomes of thematic analysis conducted on the interview data are also presented. Despite the varying factors amongst participants including severity of symptoms and age at diagnosis, distinct similarities were identified throughout the data analysis process and six major themes were classified: Physical limitations to daily life, risk perception and existential thoughts, communication and support, responsibility, loss and resilience and coping mechanisms. An overview of the themes and subthemes have been illustrated in a thematic map displayed in figure 5.

4.2 Recruitment and response rate

The recruitment process is described below in figure 4. 34 participants returned their consent forms and a final total of 21 participants were recruited and completed interviews, producing a response rate of 61%.

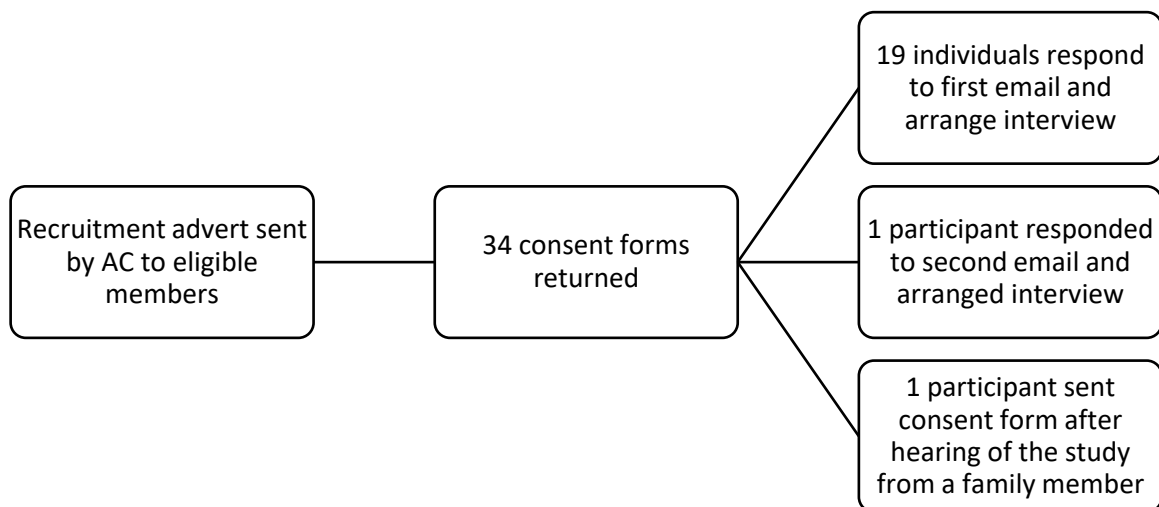


Figure 4 - Flowchart describing the recruitment process.

Table 8 - Response rate from recruitment

Participants	Reply slips returned	Interviewed	Response rate
AC vEDS member	34	21	61%

4.3 Sample characteristics

The demographic characteristics of the 21 participants are displayed below in table 9. All participants are identified using 'PN' followed by a number to ensure anonymity. The mean age was 41 years old (range 19-61) including 18 women and three men. Partner status, whether they had children and whether they had experience of a serious vEDS event was also included.

Table 9 - Participant demographics

Interview	Age	Partner Y or N	Male or female	Children Y or N	vEDS dramatic event Y or N
PN001	32	Y	F	N	N
PN002	37	N	F	Y	Y
PN003	61	N	F	Y	Y
PN004	23	Y	F	N	N
PN005	27	Y	F	N	N
PN006	44	Y	F	Y	Y
PN007	44	N	F	Y	Y
PN008	39	Y	F	Y	N
PN009	54	N	F	Y	Y
PN0010	46	N	F	N	Y
PN0011	20	N	M	N	N
PN0012	19	N	M	N	N
PN0013	39	Y	F	N	N
PN0014	54	Y	F	Y	Y
PN0015	34	N	F	Y	N
PN0016	57	N	F	Y	N
PN0017	52	Y	F	Y	N
PN0018	46	Y	F	N	N
PN0019	42	N	M	N	Y
PN0020	43	Y	F	Y	Y
PN0021	57	Y	F	Y	N

4.4 Results from thematic analysis

Thematic analysis of all 21 interviews discovered six dominant themes that address the research question when exploring the psychosocial impact of living with vEDS. Each overarching theme contains subthemes, summarised in the thematic map in figure 5.

All interviews were analysed to reflect the patient experience and achieve data saturation. Each theme and its associated subthemes are discussed below, along with illustrative quotes from the interviews.

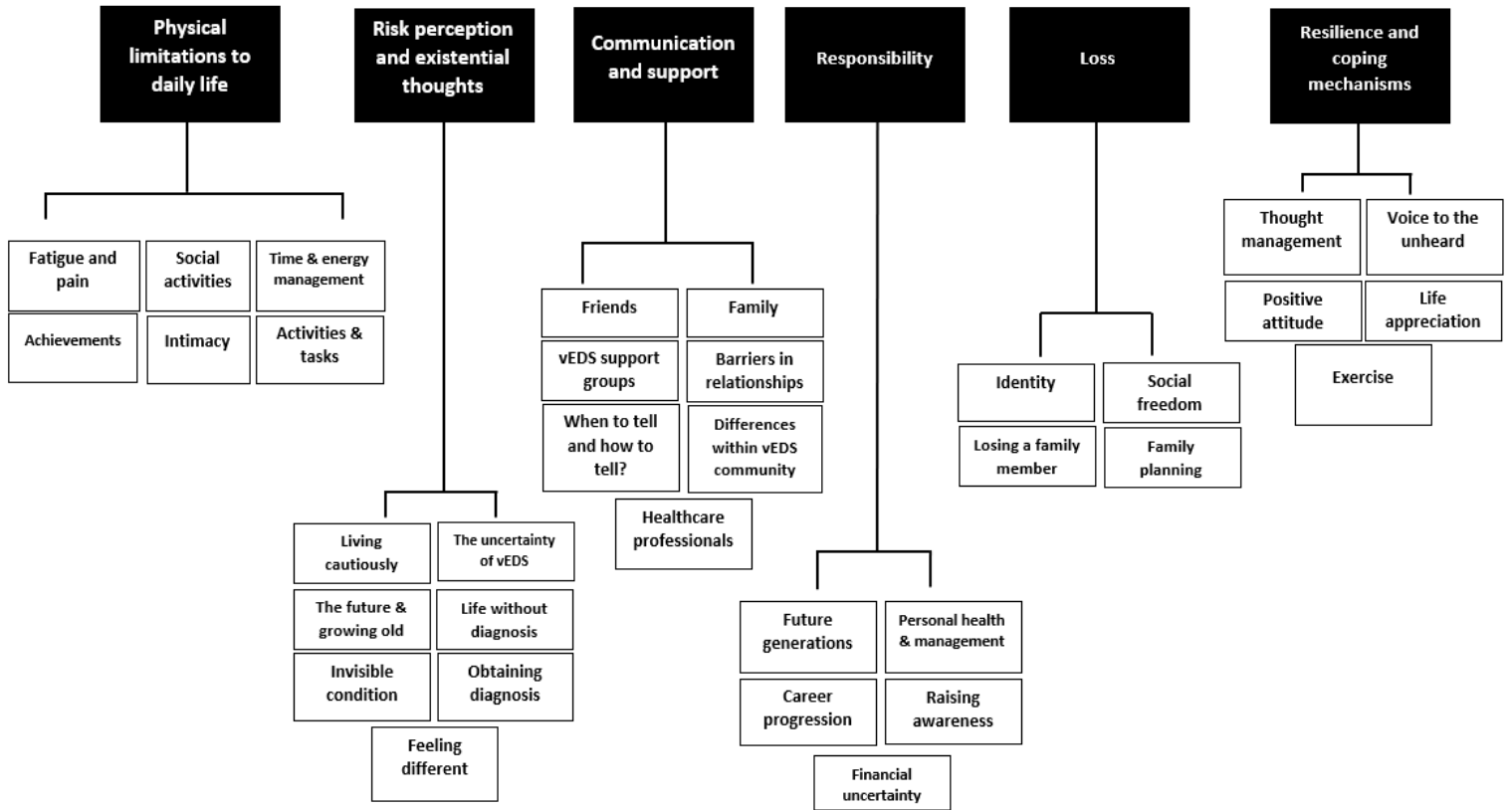


Figure 5 - Thematic map displaying the results from thematic analysis. Six dominant themes were established. Associated subthemes are included and have been shortened due to spacing.

4.5 Physical limitations to daily life

Participants expressed how the physical part of the condition directly or indirectly affected the psychosocial aspects of their lives and that the relationship between physical health and its impact on psychosocial health was often dismissed and unnoticed.

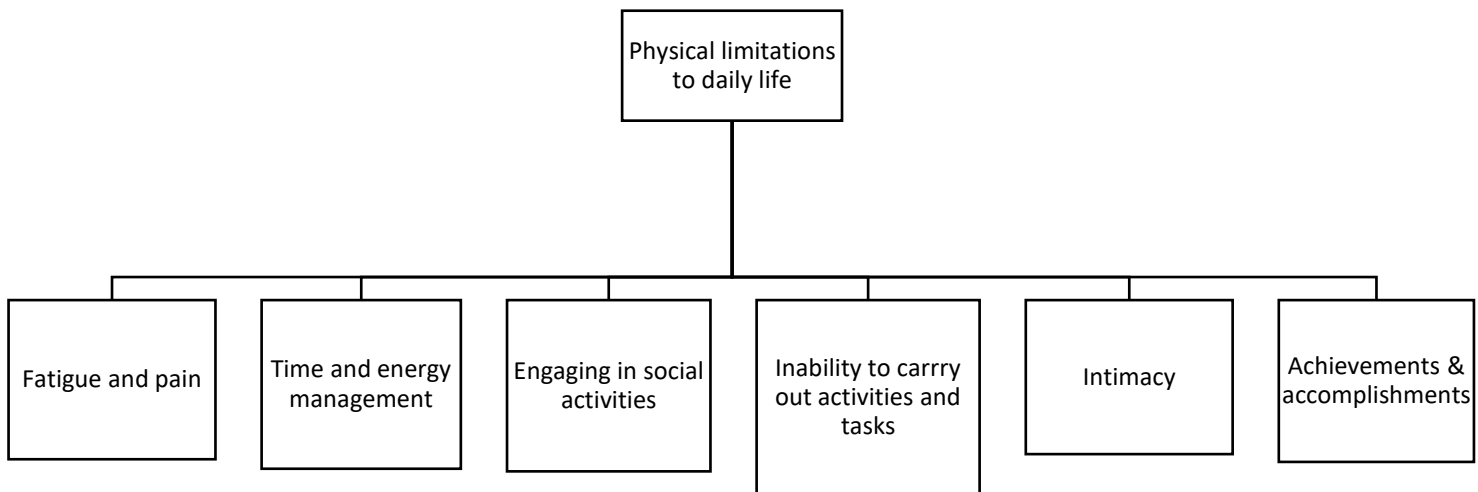


Figure 6 - Thematic map specific to the theme of 'Physical limitations to daily life'. The arrows navigate to the subthemes and additional layers of what analysis uncovered.

4.5.1 Impact of fatigue and pain on everyday life

Fatigue and pain were reported as significant barriers in participants lives, impacting their day to day functioning.

4.5.1.1 Fatigue

When prompted to discuss the daily challenges faced due to vEDS, fatigue was mentioned repeatedly, provoking feelings of frustration:

"It's the fatigue, I've just not got the energy...I have to be wary of pushing myself"

"the tiredness, the pain, the frustration of not being able to open a milk bottle"

Living with fatigue was a continuous challenge for participants, as it impacted every aspect of their lives, to the extent that some individuals felt finding a cure for their fatigue would solve almost all of their problems.

4.5.1.2 Pain and pain management

Pain was discussed in relation to chronic pain, pain from bruising or skin tearing and sudden unknown pain eliciting anxiety and worry. Managing pain and injuries consumed a sizeable amount of participants time, resulting in frustration and perceived reduction in quality of life:

"it's just pain and managing injuries. You go to open a can of beans and your hand bruises"

“Because I was just in so much pain...I just had no quality of life at all.”

Consequently, individuals explored the concept of discovering their own pain thresholds in relation to the condition, to help manage and rationalise their pain and worry:

“it took quite a few years for me to find what my baseline was, what my normal was”

“just trying not to be constantly on edge of any sort of sudden pain, whether that's going to be the one that takes you”

Understandably, attempting to manage and adjust personal pain thresholds, remains a basis of concern for individuals.

4.5.2 Time and energy management

Participants felt the condition determined their daily tolerance, reducing their daily activities and limiting them to a certain number of tasks each day:

“I can only do one thing a day...and that's the challenges I face really”

“if I've been really good one day, I might not be able to...do what I want the day after”

Therefore, patients explained that forward thinking and planning became a crucial part of their day to day lives to ensure their energy was managed efficiently as seen below.

“managing my energy, making sure I have enough time and energy to do these things.”

“If I know I need to clean the house one day, then I can't wash my hair on that same day because that will use up all my energy”

“I do everything in phases”

Lastly although thankful that they are being monitored, hospital appointments were often challenging because of the significant effort required for attendance.

4.5.3 Engaging in social activities

Physical health appeared to directly affect participants' ability to engage in social activities. Despite their frustration, recognising their own limits was important when deciding whether to attend a social event:

“it limits it a bit in that I have to say oh I can't come to this. And there's actually no tangible reason that they would understand why I can't get to something...because I'll be too tired...I know it'll be too much”

Furthermore, participants stressed that in some cases, engaging in social activities presented more strain than enjoyment, due to the need to risk assess each situation, playing devil's advocate as to what the worst-case scenario would result in:

“if it's going to be really busy and you're going to get knocked and shoved and there's not anywhere to sit...is it an upstairs event? Am I going to be able to get upstairs?”

As well as the anxiety surrounding physical engagement in social activities, judgement from others was also an anxiety inducing factor for patients. It is clear that participants face an abundance of concerns regarding their social lives, that result from the physical limitations of the condition.

4.5.4 Inability to carry out activities and tasks

A handful of participants reporting difficulties carrying out simple tasks. Seemingly minimal, easy tasks are now obstacles participants face each day.

“I mean, at night I look at the stairs and it might as well be a mountain to climb you know.”

“I can't walk very far now, and I can't lift certain things, you know, do things that I would have been able to before.”

This feeling of helplessness substantially impacted individuals, who felt they were continuously questioning whether they should or should not carry out certain tasks, as captured in the quote below:

“Ummm constantly...like assessing can I do this. Should I be doing this?”

Participants also stressed the importance of information, knowledge, and parameters from healthcare professionals to help manage their physical health.

4.5.5 Achievements and accomplishments

The practical limitations of vEDS became apparent as individuals voiced that over time, they became increasingly aware of how the condition restricted them from achieving certain milestones and accomplishing goals in life:

“I'm looking at peers my own age and they're still working full time...They're joining the gym. You know, they're going on holidays.”

“everyone has these life milestones and umm mine are just yeah I just don't know how to adjust mine, it's difficult.”

Adjusting to the physical and psychological restrictions of the condition proved difficult for participants, as the uncertainty of time the condition presents, added pressure for them to achieve their goals.

4.5.6 Intimacy

As the interviews permitted the exploration of sensitive and private information, the topic of intimacy arose amongst participants with a partner. The physical aspects of the condition raised concern for participants, fearing they may provoke a vascular episode during intimacy, the experiences of which are depicted in the quotes below:

“just being affectionate, he can hurt me without meaning to.”

“so now, he doesn't want to hurt me, so that's affected our relationship”

“when it comes to sex, you worry about your blood pressure and about having/causing a vascular episode and things like that”

Despite only a handful of female participants discussing intimacy across the interviews, it is evidently a topic that others may want to explore if prompted and given the opportunity to do so.

4.6 Risk perception and existential thoughts

Participants were exceptionally aware of the uncertain life expectancy associated with the condition, which influenced varied attitudes towards the future amongst the group.

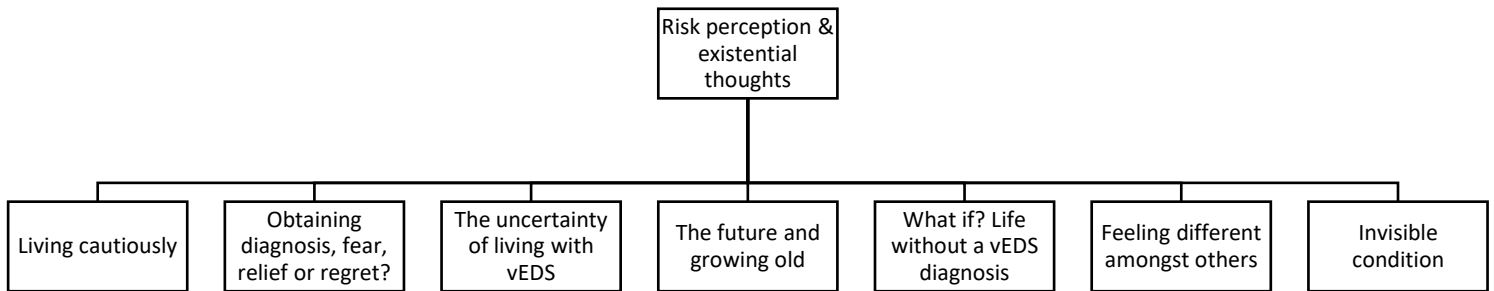


Figure 7 - Thematic map displaying the theme of 'Risk perception and existential thought's'.

Individuals were asked how often they think about their diagnosis (figure 8) providing insight into their perceptions of how much the condition affects them on a daily basis.

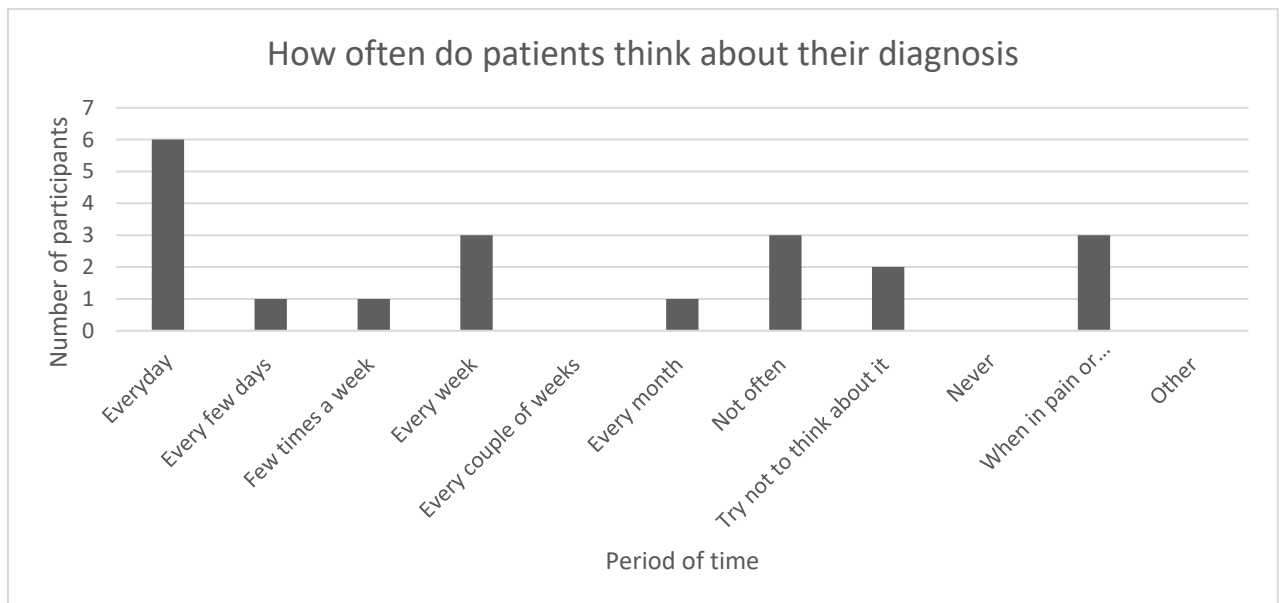


Figure 8 - Graphical representation of how often participants think about their diagnosis.

4.6.1 Living cautiously

Almost half of the participants had experienced a serious vEDS event; an episode resulting in surgery or prolonged hospital admission. Traumatizing experiences subsequently led to caution, anxiety, and fear:

“you’re like a ticking time bomb and don’t know when the next rupture is going to happen”

“after having such a big leg rupture, I was just terrified that no matter what I did I was going to have another major rupture”

In addition, individuals were fearful of tasks they believe caused their event, adding another depth of anxiety and wariness:

“because it happened to me on the stairs...I was just scared that every time I used the stairs...that was going to cause another rupture”

It was clear that a serious vEDS event had a significant impact on participants lives as they lived in constant fear of subsequent recurrence.

4.6.2 Obtaining diagnosis, fear, relief, or regret?

As expected, due to the variability in presentation of vEDS, receiving a diagnosis surfaced an array of responses summarised in table 10 below. For some, receiving their diagnosis was a relief following months/years of symptoms. For others, diagnosis elicited fear and emphasised the uncertainty associated with the condition.

Further to receiving and adjusting to their own diagnosis, participants with children and grandchildren were faced with an additional layer of grief. Although regret was not a term used to discuss the possible setbacks, several participants deliberated whether receiving a diagnosis was beneficial, or whether continuing life without knowing would have been better.

Table 10 - Responses to receiving diagnosis, with illustrative quotes

Response to receiving diagnosis	Quotes
Relief	<p><i>“HA! [laughs] I have got something wrong with me... [laughs] I was right, it was definitely a relief.”</i></p> <p><i>“The lead up to it was quite stressful...But when I was actually diagnosed, it was almost quite a relief.”</i></p>
Fear, devastation, and uncertainty	<p><i>“that period I felt my whole world had fallen apart, with that diagnosis. Everything that I knew and everything that I had...ever dreamed of [pauses] just was shattered to pieces”</i></p> <p><i>“So you do start thinking oh my god... You know, you're virtually in a coffin already and you can't do this.... You can't do that... You can't...”</i></p>
Grief	<p><i>“Receiving my diagnosis, was on one level. [INT: hmm] Then the consequent knock on effect of my children and my grandchildren was a very different level, and I found that even more difficult”</i></p>
Deliberating decision to receive diagnosis	<p><i>“yeah it was much easier not knowing [laughs]”</i></p> <p><i>“a bit of me is like why did I push for this diagnosis. You know, why did I like push for them to test me? Because now I've got the results...I'm like, I'm not sure, I want to know [laughs]”.</i></p>

It is obvious that for some participants the end of their diagnostic odyssey brings relief, whilst for others receiving a diagnosis triggered devastation and panic.

4.6.3 Facing mortality the uncertainty of living with vEDS

The conscious awareness that vEDS is a life limiting condition meant participants faced the difficult concept of confronting their mortality, whilst living with the uncertainty of life and life expectancy.

“it's like we were given a life sentence...and you realise how fragile your life is”

“So just that the general difficulty of...facing your mortality”

“But it's different from being given a diagnosis of cancer. Someone can say unfortunately, you've got six months, with us, we don't know whether got six months or six years or 20 years.”

Participants likened vEDS to cancer, in an attempt to contextualise their uncertainty and concerns against a well-known supported condition. Living without a definitive time scale exacerbates the uncertainty surrounding life expectancy and some individuals commented on their fears of dying and living everyday as if it was their last:

“in the shower, I'm almost like preparing the body, I think to myself is this the last time I'm going to paint my nails...is this the last time that I'll put that colour on...will somebody else be tidying me up when the time comes”

Over time, some individuals, disclosed how they had learned to accept death and come to terms with their own mortality. A final noticeable concept that surfaced was how participants normalised the

uncertainty of life that vEDS presents. Ten participants used a version of the following examples to normalise the ambiguity of their life expectancy:

“Everybody lives with the big unknown...you could get run over tomorrow.”

“it's the same for all of us...we could get run over by a bus tomorrow...”

Linking the uncertainty of vEDS, to the ambiguity of life of someone in the general population, was used to highlight that no one is in control of their mortality.

4.6.4 The future and growing old

There was a varied response from participants when discussing how they view their future.

Questioning whether to make future plans and perceptions of growing old highlighted the differing opinions amongst the study sample:

“do I need to plan that far ahead because will I still be here?”

“I'm always thinking to myself; I won't have a retirement because I'm going to die young [laughs]”

“I was just so frightened that booking something in advance...was going to tempt fate in a way that I wouldn't make it.”

The idea of not living long enough to see pre-organised plans through, stresses how life expectancy is a substantial existential thought in individuals' lives. On the other hand, certain participants showcased their conscious efforts to live life in the moment, and make decisions for now, as they recognise the time sensitive nature of their condition:

“because I know I may not be around forever, I haven't got that ummm...the outlook of oh we can do it next year...if I want to go and see a gig or a play or something...I'm going to do it.”

Whilst certain individuals had decided their approach to the future, others felt continually conflicted, hoping to establish a balance:

“Trying to get that balance of trying to plan for the future, but also live in the moment”

Discussions around the future helped individuals gain perspective on living life with vEDS, something which, several participants disclosed they had not articulated before the interview.

4.6.5 What if? Life without a vEDS diagnosis

Although participants were learning to accept their vEDS diagnosis, upon reflection, ‘what if?’ comparisons emerged, questioning life and decision making:

“A lot of my decisions have been made because of my condition... [continues crying]”

“I think it's very hard to maintain a healthy perspective and objectivity about living...When you're living with a condition like this and it's affected, probably most of the decisions I've made in my life”

Practising reflexivity in this area triggered sadness as participants explored where their lives would be if they did not have their condition.

The age in which participants were diagnosed with the condition varies and this is visually presented in the graph below (figure 9). Noticeably, most individuals were diagnosed later in life and this, for many, was considered beneficial:

“it's like we were given a life sentence...I feel...I've had to like slow burn on it, and learn things and adapted along the way”

“You know, we've lived our life in complete ignorance. Most importantly, lived a childhood with no restrictions as a result of this diagnosis.”

Participants felt that if they had been diagnosed at a younger age, it would have restricted their childhood and adolescent years, therefore proving that age at diagnosis is a factor to consider when scaling how vEDS impacts individuals on a psychosocial level.

However, certain participants that received their diagnosis during their teenage years were grateful, as it helped guide their management of the condition through adolescence:

“but luckily, like I guess I was lucky I was diagnosed then actually”

“by the time I got my diagnosis, I was just ready to know what it was...it was a relief”

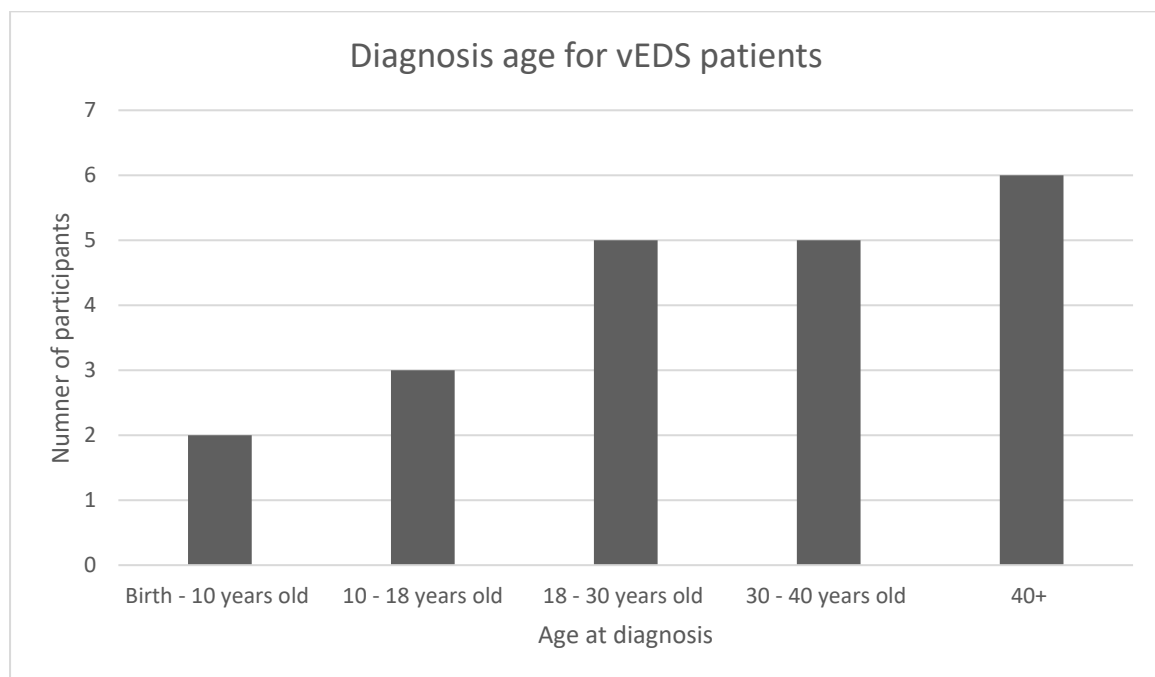


Figure 9 - Graph displaying the age at diagnosis for participants in the study. This data was collected during interviews.

4.6.6 Feeling different amongst others

Across the study sample, it became evident that participants were aware of how vEDS sets them apart from others and the concept of not being normal emerged as shown in the quotes below:

“I guess I just think of myself as not as good you know, like not good enough. I'm not as good as them and I'm not normal.”

“Yeah it's kind of instilled in me that I'm not normal and that there's something not right with me.

Feeling different and not fitting in proved to be a major psychological challenge for participants as they expressed feelings of isolation, loneliness, and self-doubt, realising that they are on their own, altering their perceptions of themselves. One participant highlighted that although being normal is deemed boring by society, after living with vEDS, a normal life was all she desired:

“It's so funny because normal is just something that society goes urgh normal who wants that...but I want normal [laughs]”

4.6.7 Understanding vEDS as an invisible condition

A particular frustration that appeared across the study sample was the notion of living with a hidden condition. For many, vEDS affected their physical health but this was not visible to others.

“It's a silent illness because there isn't anything on the outside that shows that you have it”

“you can't see that anything's wrong with me. I think that's the biggest thing”

“It's very difficult to understand something you can't see.”

Appearing normal on the outside, for some, was a relief as they were able to mask their condition from others. However, the invisible nature of vEDS created more complications, specifically in the form of judgment from others:

“People look at me and they say, you know, you look so well”

“umm it's still hard because I don't look ill and that's the thing...I have a blue badge and I see people looking at me. And I think they're thinking why has she got a blue badge it doesn't look like there's anything wrong with her”

Living with a hidden condition presents its own challenges, as the reactions participants received from others affected how they coped with their diagnosis leading to further feelings of isolation.

4.7 Communication and support

Communication surfaced across the data as a vital component when living with and managing life with vEDS.

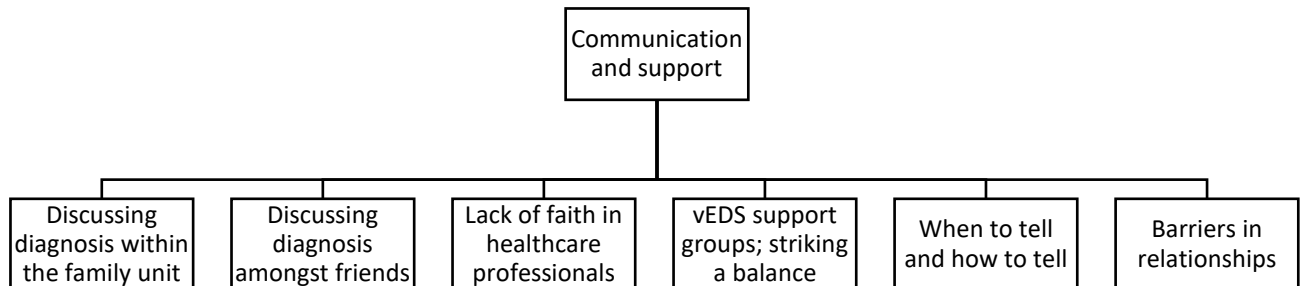


Figure 10 - Thematic map specific to the theme of 'Communication and support'. The arrows navigate to the subthemes.

4.7.1 Discussing diagnosis within the family unit

It became evident that communication within the family was important for individuals, as disclosing information about the condition provided support within the family unit:

"I think it's brought us closer together...And they've obviously been very supportive."

Communicating within the family about vEDS proved to carry responsibility, as participants held potentially life-changing information regarding other family members:

"It's a bit like, well, until I told everybody, I guess I've got that burden of responsibility."

On the other hand, patients discussed how not communicating protects family members from the condition, as they acknowledge the possible anxiety, discussion can cause.

"so, with my family I do...I find myself not bringing it up because I don't want to upset them."

"so probably I do steer away from it...maybe to protect them from how horrid it is..."

It is important to recognise that decisions around vEDS conversations are not only based on personal desires to share information, but also the protection of themselves and family members.

4.7.2 Discussing diagnosis amongst friends

The role of communication is just as powerful with friends, however, unfortunately, for some, communicating with friends about the diagnosis, led to the breakdown of friendships, due to trust, support and understanding:

“I've lost friendships because people hadn't believed me.”

“you've got to be there for them as well...it did impact some of the friendships that I have”

Furthermore, reasons for not discussing vEDS with friends ranged from potentially burdening friends, to the uncertainty of the condition, both of which showed to place strain on the friendship:

“I did almost die twice [laughs] its quite a burden to put on people...so it did impact my friendship groups then...”

“I didn't used to tell them about my condition...It wasn't something I wanted to burden anyone with.”

The quotes above create additional evidence on the impact vEDS has on the psychosocial wellbeing of participants. It is clear that the social aspects of the condition are significantly affected.

4.7.3 vEDS support groups; striking a balance

Participants have been part of several support groups throughout their diagnosis. Companionship and having someone who understands entirely what vEDS is and how it can affect life, was a leading advantage of attending a support group:

“I think it's just nice knowing you're not on your own.”

“people around you can be supportive, but to an extent, whereas I suppose if you meet someone who also has it...they know exactly what's going on.”

Building relationships with individuals from a support group had a positive effect for participants. However, due to the nature of the condition, participants were deeply affected when a member of the vEDS community died. In several instances this topic triggered an array of sadness and emotion.

“So, when somebody in the vEDS community died...I was absolutely besides myself, it was awful”

The death of members within the support group had a significant impact on participants wellbeing as it was a stark reminder of the fragility of the condition. The constant updates received on death within the community proved difficult for participants to handle:

“it just highlights the...It's a real double-edged sword of just getting that balance right”

It is clear that decisions around actively taking part in a support group are based on friendship, loss, and wellbeing, all of which participants must weigh up, to minimize the effects on their psychosocial health.

4.7.4 Differences within the vEDS community

Individuals appreciated the sense of community and support gained from being part of a vEDS support group. However, participants also brought light to the negative attitude some patients take when approaching the condition:

“they weren't able to see past their own pain and suffering; it seems to just be a place to vent rather than to try and support”

Participants recognised that the less favoured opinions and comments were few and far between but felt that it deterred from the purpose of a community group. In addition to this, some reported an element of competition between patients comparing their symptoms:

“like it's almost a competition, who's got the worst signs and symptoms of their condition”

Participants have shown that by comparing themselves to other vEDS patients they are able to recognise fewer positive behaviours and opinions from those within the community.

4.7.5 When to tell and how to tell

Discussing the diagnosis for the first time proved to be a substantial obstacle for some participants. Individuals spoke of different methods they had attempted when communicating about the condition for the first time:

“I tried different tactics of either not telling them about the condition or telling them straight away”

“so new friendships, new people I meet I tend not to tell [laughs]”

The above quotes emphasise the conscious decision participants make when meeting new people. Some found it easier to keep the condition a secret, but this presented its own difficulties:

“not telling some people, it created a bit of a barrier...because you're always very conscious of not saying anything that might lead into a conversation that you don't want to have”

Therefore, the majority felt it was easier to tell than not tell when meeting new people. However, affected parents, with affected children also faced difficulty in deciding when and how to tell their children about the condition:

“And it's one of the hardest things ever. Thinking should I tell them or should I not and if I should...When should I tell them?”

4.7.6 Barriers in relationships

Each interviewee was questioned about the impact vEDS had on their relationships with their partners, the responses of which are summarised in table 11 below. Participants commented on the shift in dynamic and expectations with their partners, as priorities and needs changed over time. The positives of discussion centred around the benefits of working together and managing the effects of the condition as one. Non-communication within a relationship focused on protecting partners from the condition, despite placing additional stress on participants lives.

Acknowledging the impact, the condition had on individuals and their partners, led quite a few participants to retrospectively examine their decisions. Witnessing participants talk about potentially never entering a relationship if they knew the consequences of the condition, despite the support from their partners being fundamental to their management of vEDS, highlights the detrimental effects of vEDS within relationships.

Articulating the uncertain nature of the condition to potential partners, was undoubtedly challenging for participants as they experienced feelings of guilt and recognised the fragility of their own life and how this would unfairly influence a new relationship.

Table 11 - Impact of vEDS in relationships

Impact of vEDS on relationships	Quotes
Shift in dynamic	<p><i>“He sees me just as an ill person... we’ve gone from husband to wife. Partners who love you, to kind of...Patient care and protector”</i></p> <p><i>“my husband feels he’s got to...protect me...”</i></p> <p><i>“you have that change of dynamic that I’m not the wife that I wanted to be and I’m not the wife that he expected when he married me. He didn’t, he looked at me, as, you know, this bright slim young thing that was fun and was doing all the things. And that’s just gone to hell after”</i></p>
Non-communication/protection	<p><i>“I think with relationships with vEDS, you spend so much energy, emotional energy, protecting yourself and protecting everybody else, you know...”</i></p>
Retrospective thoughts	<p><i>“I wouldn’t have...I wouldn’t have sought a partner because of the consequences of this”</i></p>
Entering new relationships	<p><i>“it does seem to me that the condition scares people away from things because obviously people are looking long term”</i></p> <p><i>“I found even just entering into a relationship, I found quite difficult...because you know what if you’re not there”</i></p> <p><i>“So, when you go into a relationship, you think, is it fair to form that attachment?”</i></p>

4.7.7 Lack of faith in healthcare professionals

Throughout data analysis it was noted that participants had contact with a range of healthcare professionals (HCP) to assist in the management of the condition (figure 11).

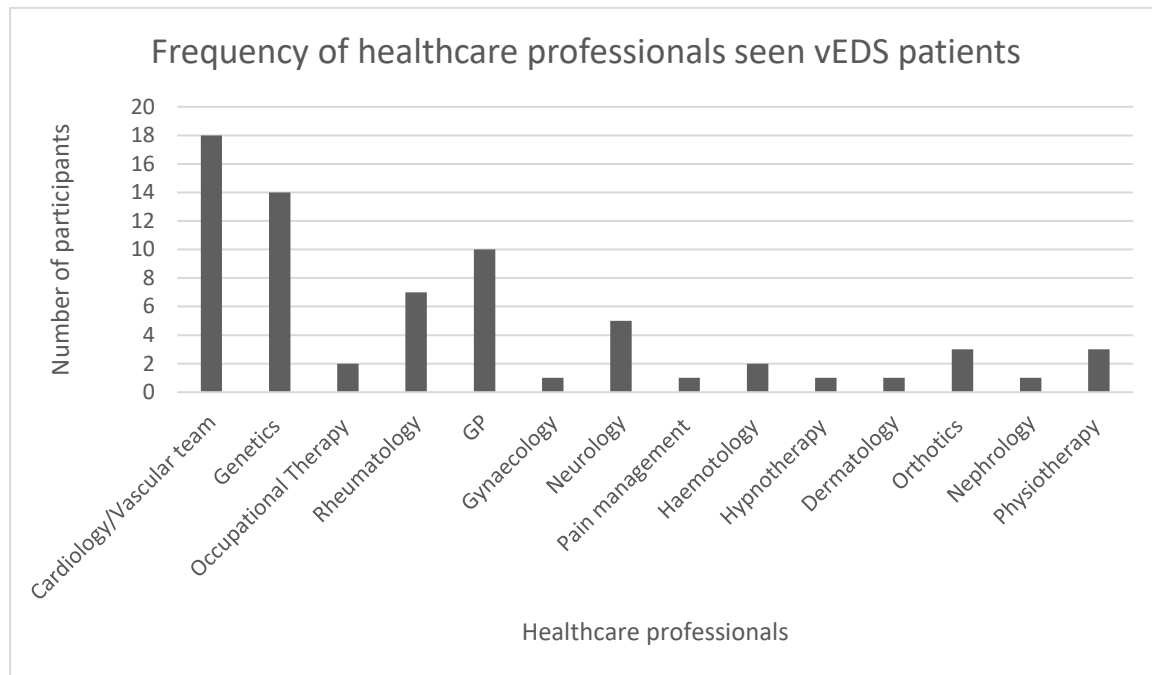


Figure 11 - Graph displaying the healthcare professionals' participants regularly come into contact with.

Although some participants praised the support they had received from HCP's, the majority felt they were not supported enough, as knowledge and awareness of the condition was minimal:

"I find that very frustrating because I don't think in this day and age, we as patients... We shouldn't have to go and explain to the medics"

Unsurprisingly, participants yearned for the relevant support and guidance needed to help manage their condition. When these basic expectations were not met, participants felt isolated and fearful for their health:

"I see all these different consultants for different things and then at the end of it they decide there is not really anything they can do...so I'm like great so I'm just left with this body that's falling apart...just getting worse if anything and I think that affects me"

Participants expressed the need for HCP's to provide guidance on the foundations of vEDS to aid overall management:

"having somebody that could help and be like a jack of all trades that would know precisely...what information to give to employers...to use in schools...to give to health professionals and paramedics."

The lack of support from those that care for patients, affected both the physical and mental wellbeing of participants. Noticeably, in figure 12 increased awareness from HCP's was the support patients

desired most, to improve their overall experience with the condition. Specifically, knowledge and understanding from general practitioners and doctors in emergency care would make a significant difference to the management of vEDS.

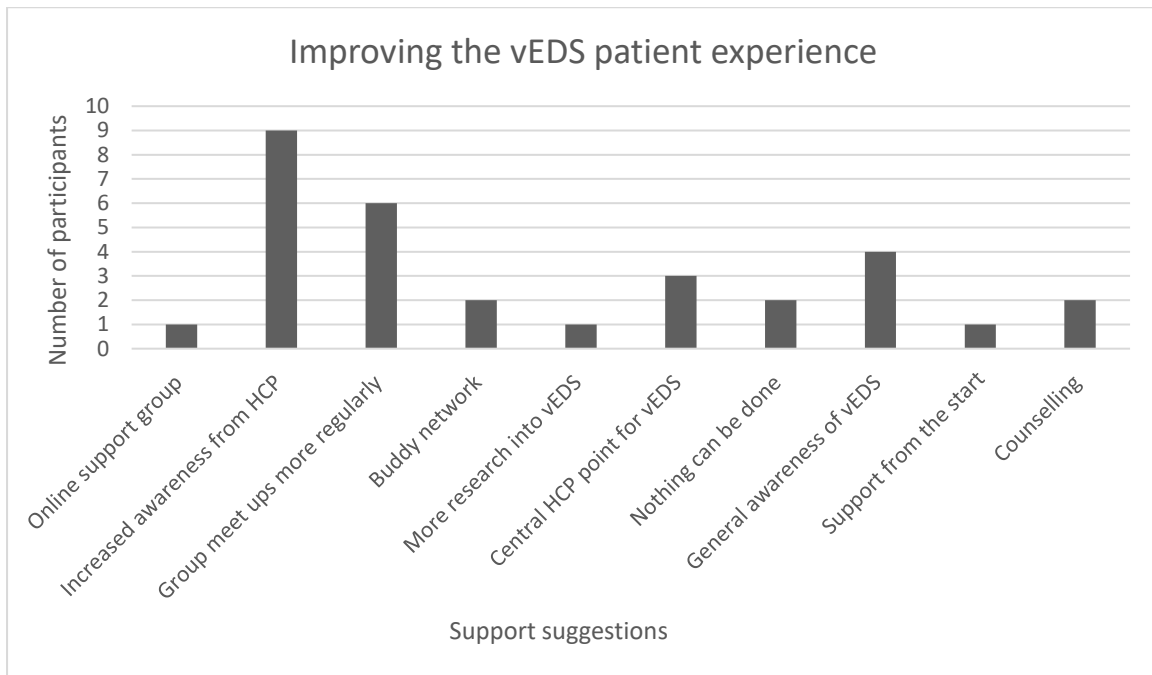


Figure 12 – Support participants desired to aid the management of their condition.

4.8 A world of responsibility

A profound feeling of responsibility surfaced throughout the interviews in different forms, which identified responsibility as an overarching theme.

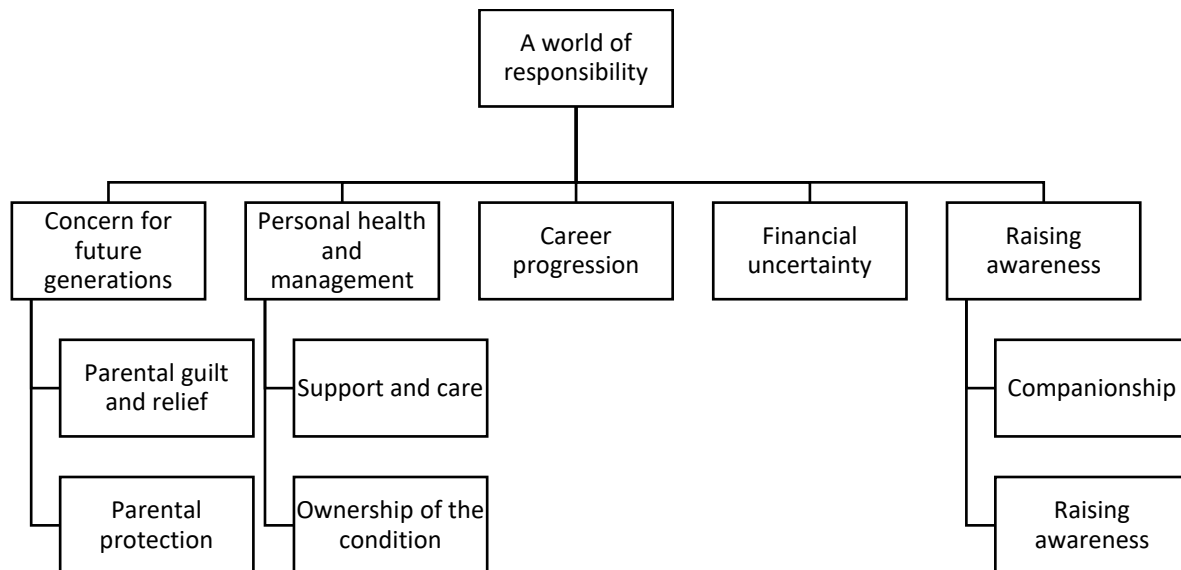


Figure 13 - Thematic map specific to theme of 'A world of responsibility'. The arrows navigate to the subthemes and additional layers of what analysis uncovered.

4.8.1 Concern for future generations

As displayed in table 2, 12 participants had children, a handful of which had a vEDS diagnosis.

4.8.1.1 Parental guilt and grief

Those who had passed the condition onto their children spoke of suffering from continual guilt, feeling entirely responsible for their child's health and future.

"because you're almost thinking well how long has this child has got? I've basically given her a death sentence straight away"

"I do feel guilty that I've passed this thing on...and they're suffering they are not leading the normal lives that they should be leading"

Caring for a child with vEDS, whilst knowing the condition originated from them, escalated participants grief and guilt for their child. Individuals voiced the difficulty they are confronted with when attempting to come to terms with the fragility of their child's life and future:

"sort of psychological impact cos you've got to, you almost start grieving, umm for a child that is actually alive"

The weight of responsibility and remorse faced by participants was an obvious burden to their everyday lives, adding another dynamic to the psychosocial impact of the condition.

4.8.1.2 Parental protection

It became obvious that a significant fear for participants was the possibility that they may die before their children. One participant shared the vivid reality of her views towards death in relation to her children:

“if it happens to me first, I’m not there to protect them and to look after them and so a part of me thinks I want them to go first and then me...[pauses] that sounds really morbid sorry...”

Not only does this showcase the realism of living with vEDS and caring for children with the same condition, but it also highlights the importance of protection for parents, that face the responsibility of caring for their children whilst suffering with the condition.

4.8.2 Personal health and management

Individuals addressed the dynamics in responsibility for their health and personal management throughout their lives.

4.8.2.1 Support and care

The varied progression of vEDS amongst patients, drew attention to the possible need for additional support and care, resulting in a shift in parenting roles as seen here:

“She in effect, became mum and she looked after me and she ran the house while I was incapacitated...And I think the responsibility shifted at that time.”

Participants sought to prevent changes in the family dynamic, however, found themselves in a position where this was no longer possible, as they required additional support.

4.8.2.2 Ownership of the condition

In Section 4.7.7, the lack of faith and support from healthcare professionals was presented as a significant challenge for participants. Consequently, individuals expressed the need to take control of the condition, in order to ensure they receive the correct care:

“Do not presume that other healthcare professionals will know much about it because it's so unusual...You must become the master.”

“I don’t want you to let vEDS own you...you need to own it”

The concept of owning the condition provides individuals with a sense of authority over their health and management. Additionally, the responsibility to keep themselves healthy, to prevent a serious vEDS event and gain control of the condition was also important:

“I’ve done everything that I can to make myself a healthier person since the diagnosis...even though I was quite healthy before, now I’m like, extra healthy than what I was”

4.8.3 Career progression

Throughout the data it was evident that vEDS placed excessive strain on an individual's ability to work and study. Participants showed pervasive opinions on their career progression in relation to the condition, noting feelings of frustration and defeat when vEDS intervened with their ability to work:

"I feel guilty for not working...because I feel as if I've umm [pauses] let myself be defeated by vEDS"

"I wouldn't particularly give much more crap if it didn't have complications in the way it does for my degree."

Patients were left with the additional pressure that went with working and managing their health.

Judgment from others within the working environment added an additional layer of concern:

"I felt disabled when I was at work because of the way I was treated"

"work made me feel like [pauses] I was less of a person because of the condition"

In the same way that participants are responsible for their own and their children's health, the growth of their career was also a life factor affected by vEDS.

4.8.4. Financial uncertainty

The uncertainty that vEDS places on individuals' lives not only affects their current financial situation, but also increases anxiety around savings and financial stability, producing hesitant views on saving for pensions and the future:

"should I be paying into a pension? Chances are I won't be here to draw a pension. So, should I just keep that money now?"

"when you're talking about like pension plans and stuff, I'm like, do I need a pension plan. Should I have a pension plan? Or should I just save my money? [laughs]"

Participants acknowledged responsibility for financial management and future planning, however, the ambiguity regarding life expectancy in vEDS provoked deep thought and deliberation.

4.8.5 Raising awareness

Certain patients believed that having the condition presented them with the responsibility to share their story and raise awareness of vEDS.

4.8.5.1 Companionship

Mature participants and those that had lived with the condition for a long time, noticed the value of sharing their experiences with other vEDS patients to provide a positive perspective on their future:

"I wouldn't mind sharing that experience with people to give some hope"

"I know they get the positivity from me of being 52 and overcoming different things and still surviving"

Participants recognised the benefits that can arise from their stories and felt responsible to communicate these with others.

4.8.5.2 Raising awareness amongst others

Participants also acknowledged the need to increase awareness of vEDS amongst healthcare professionals and their community:

“I think understanding, helping people understand, helping doctors understand, some ambulance people don’t even know what it is either”

“I thought [laughs] I should go round telling people how to kind of like...spot vEDS”

Through education, fundraising, increasing understanding and taking part in research, individuals felt productive as they worked collaboratively to raise the awareness of the condition in an attempt to improve overall support and care.

4.9 Loss

Loss as a result of vEDS was experienced in some form by each participant, throughout their diagnosis. Influencing factors affected the type of loss individuals endured.

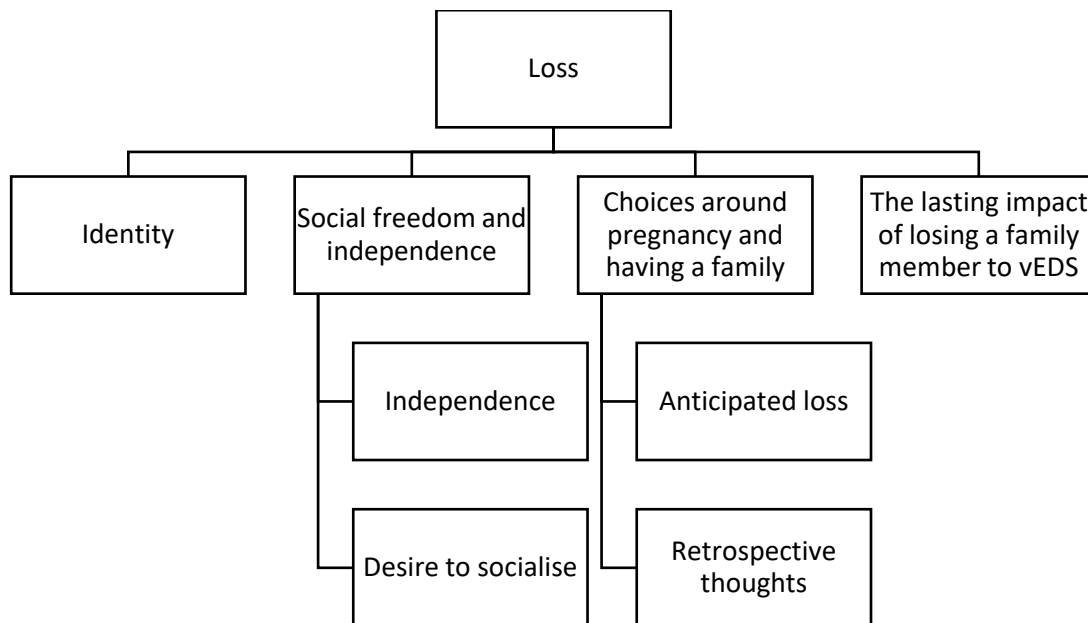


Figure 14 - Thematic map specific to theme of 'Loss'. The arrows navigate to the subthemes and additional layers of what analysis uncovered.

4.9.1 Identity

Adjusting to life with vEDS led to the acceptance that the condition was now part of their identity, stirring several emotions as participants consistently wondered who they would be, if they did not have vEDS:

“You wonder who you would be if you didn't have it [starts crying]”

Certain individuals spoke of how they struggled to adapt their lives, feeling that as the condition progressed, they were losing a part of themselves. Specifically, for certain female participants, they felt the condition had stolen their identity in regard to having children:

“Difficult in the sense that a lot of people's identity is being a mum like at 30”

The inability to have children shifted participants identity as they became increasingly aware of the loss vEDS presented them with, especially in comparison with friends.

4.9.2 Social freedom and independence

For numerous participants, the condition directly affected their independence and confidence which in turn, reduced their desire to socialise, captured through the quotes in table 12.

4.9.2.1 Independence

Lowered independence surfaced as a key factor for individuals when considering how the condition affected their social freedom. Relying on others, led to increased self-doubt and a loss of confidence, in their own abilities on a personal and social level.

4.9.2.2 Desire to socialise

It seemed that there was a difference in social confidence between patients that were diagnosed at a young age, compared to their older associates. Younger individuals suffered more intensely with social situations, as they doubted themselves and their interactions with others. In comparison, those that were diagnosed later in life, appeared more confident, as they felt having lived most of their lives without knowing about the condition played to their advantage.

Table 12 – The impact of vEDS on social freedom and independence.

Effects of vEDS on independence and desire to socialise	Quotes
Independence	<p><i>“I do struggle to ask people for things...because I’ve always been very independent and that’s really... caused me the most grief”</i></p> <p><i>“I seriously could have just taken on the world myself. And I find that difficult, actually relying on other people”</i></p>
Desire to socialise in younger participants	<p><i>“I know it’s probably affected my confidence; I think I’m a bit socially retarded anyway and I’m not very good in social situations”</i></p> <p><i>“the social aspect of it has been the biggest impact and the hardest to deal with [pauses] for me”.</i></p>
Desire to socialise in mature participants	<p><i>“because I’ve got what I’ve got, that shouldn’t stop me doing any of these things?”</i></p> <p><i>“it’s not made me think, oh I don’t want to go out I want to say in... it’s made me more okay right I’m going to make sure I see this group of friends this week and that group of friends next week”</i></p>

The differences displayed between age groups could be explained by several contributing factors. Nonetheless, the loss of social freedom had a substantial impact on the psychosocial wellbeing of vEDS patients, especially within specific age groups.

4.9.3 Choices around pregnancy and having a family

The perceptions of having a family differed amongst the patient group, as male and female participants expressed the difficulty faced when managing their reproductive options.

4.9.3.1 Anticipated loss

For many, processing the possibility that they may not be able to have children was extremely difficult. The quotes below encapsulate the mentality a few participants had adjusted to:

“I always thought I’d have a family and now I don’t...[pauses] I just have to deal with that”

“I just have to readjust my expectations I guess.”

“I want children one day...but I might not give birth to them”

The decision-making process was a real challenge for participants and one individual discussed how deciding not to have children was very distressing:

“I’ve chosen not to have children.... which is quite hard actually [starts crying]”

Despite recognising that the decision not to have children protects their health and the health of a potentially affected child, this aspect of vEDS significantly impacted patient's lives.

4.9.3.2 Retrospective thoughts

Those diagnosed with the condition after having children, explored their thoughts on having a family retrospectively. The pregnancy and birth of their children, as well as knowledge about the risk the condition poses during childbirth, surfaced as the main factors when discussing perceptions of having children now:

“but then if I knew I'd had the condition, then maybe I wouldn't of had my daughter,”

“after having my daughter and the trouble that I'd gone through, there is no way on this earth I would ever put myself through it again [laughs]”

Again, age at diagnosis influences the outlook participants have on certain aspects of living with vEDS. Opinions on alternative options to having a family varied significantly amongst the patient group. A few individuals felt uncomfortable with the concept of adopting a child. Others had embraced the idea and one participant adopted on the basis of her diagnosis:

“I have adopted a child...because of the condition”

It is important to recognise that although alternative options are available, participants may struggle with the loss of not having their own biological child.

4.9.4 The lasting impact of losing a family member to vEDS

The physical loss of a family member to vEDS influenced the approach a proportion of participants took to living their lives. Experiencing a loss, created a basis when considering how their own death would impact their families, especially if they were not the first to be diagnosed.

“I think when you see the impact on your family, on your loved ones, you're very aware of how that could be if something happened to you”

Participants voiced that as the condition progressed, they became increasingly aware of how the condition affected their lives and the lives of those around them. Losing a family member adjusted participants perceptions of their life expectancy, to those they had lost as a result of the condition.

“even the fact that my brother died when he was 29, when I hit my 30th birthday that really sort of hit me, because I thought you know I'm older now than he was”

“When the levels above you have gone, you know that you're the next in line kind of thing”

Individuals that lost a close family member with the condition were left feeling alone and isolated:

“what I found most difficult about losing my brother was that he was the person that understood what I was going through...now I'm on my own and no one understands it.”

4.10 Resilience and coping mechanisms

Participants acknowledged that vEDS had limited their lives in several ways, yet throughout each interview, individual's strength and determination to carry on despite their circumstances remained absolute. Table 13 showcases the direct quotes from participants, that correlate with the coping mechanisms they found most effective.

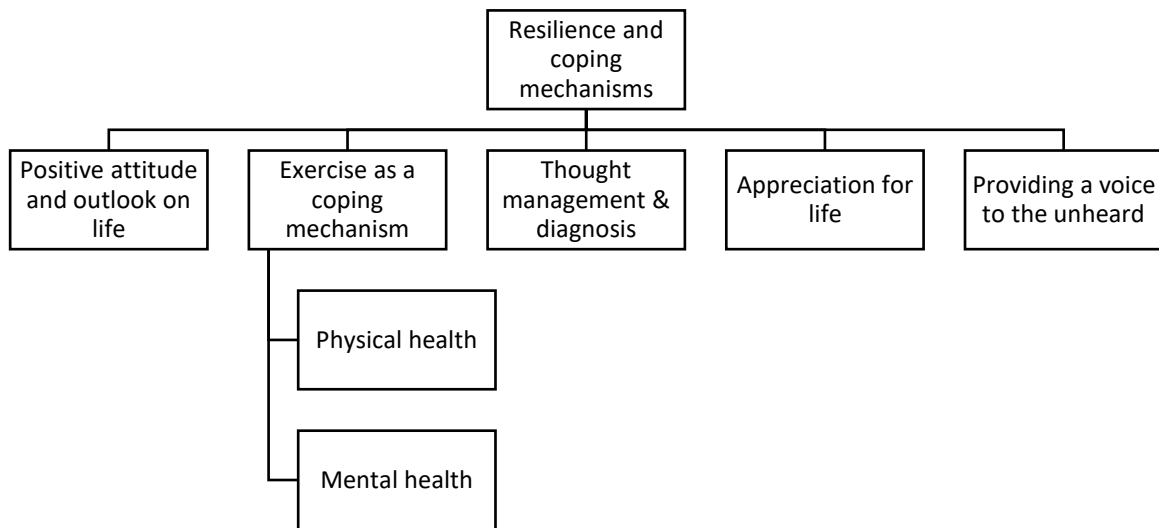


Figure 15 - Thematic map specific to theme of 'Resilience and coping mechanisms. The arrows navigate to the subthemes and additional layers of what analysis uncovered.

4.10.1 Positive attitude and outlook on life

Maintaining a positive attitude appeared to be of significant importance for participants as they demonstrated that changing their perspective enabled them to embrace the condition, ensuring that they remained determined despite setbacks. The resilient nature of individuals within the study group was inspiring, as they recognised that maintaining a positive outlook towards life was an empowering coping mechanism.

4.10.2 Exercise as a coping mechanism

The physical ability amongst participants varied, restricting some more than others.

4.10.2.1 Physical health

Managing health and fitness was greatly important in providing participants with control over their bodies. It was reported that engaging in physical exercise, directly led to improvements in personal health and increased stamina to help individuals get through the day. Although not all participants

were able to exercise regularly, those that were able experienced considerable health benefits, which establishes exercise as a valuable coping mechanism.

4.10.2.2 Mental health

Several participants experienced mental health issues as a result of the condition (figure 16) some of whom felt exercise helped with their mental wellbeing. Counselling and CBT were the most frequently accessed mental health support networks.

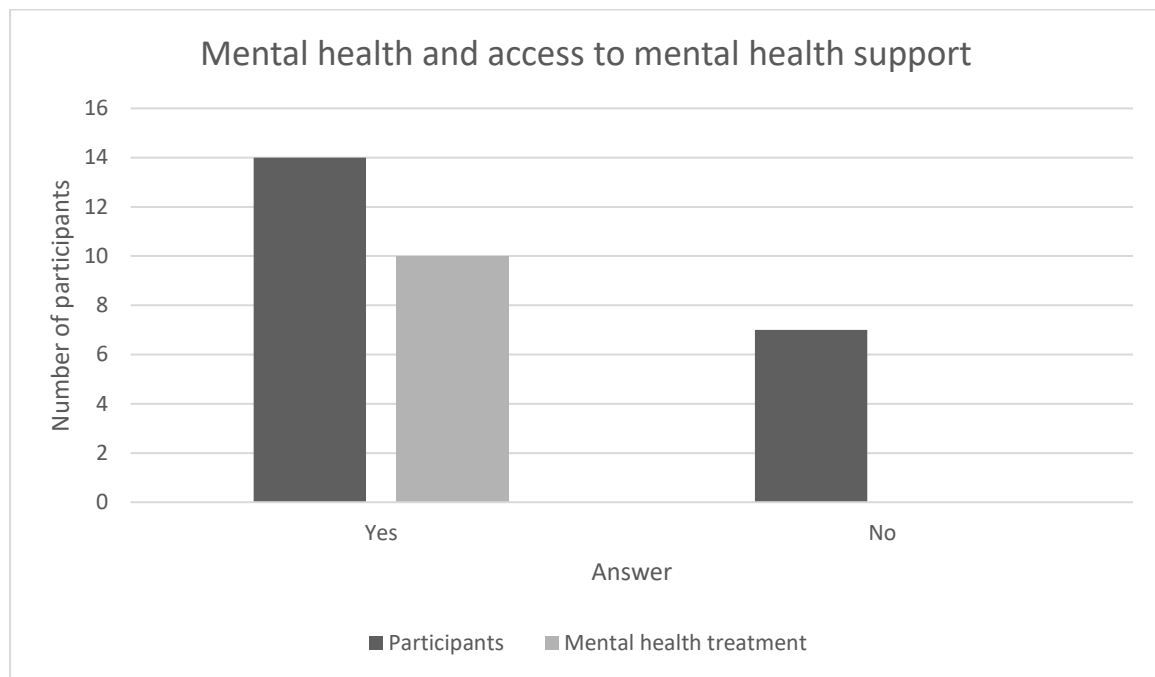


Figure 16 - Number of participants that suffered from mental health issues as a result of vEDS, including the number of those that sought support including counselling, CBT and medication.

As well as the physical advantages, exercise was noted to aid participants' mental health, as having a healthier body, led to a more optimistic outlook on life. Recognising the relationship between physical and mental wellbeing for vEDS brings a new dimension to exploring effective coping methods.

4.10.3 Thought management and diagnosis

Speaking with a diverse cohort of vEDS patients unveiled the different approaches individuals took to live with their diagnosis. For some, ignorance was bliss as they explained the benefits of not acknowledging the condition in their lives and blocking it out. Although initially interpreted as denial, these participants spoke of how they had accepted their diagnosis but chose to compartmentalise vEDS, separating it from other aspects of their lives.

4.10.4 Appreciation for life

Participants held a profound appreciation for life and a strong sense of gratitude, recognising the importance of valuing their lives. Adopting this approach enabled individuals to face their struggles from a position of gratefulness, being able to acknowledge their personal journeys in a positive light.

4.10.5 Providing a voice to the unheard

In several examples, participants acknowledged and compared the provisions available to well-known conditions such as cancer. Participants expressed their frustration at being overlooked in regard to research studies and for many this was the driving factor behind their participation in this research project as it provided a platform for their voices.

A mixture of opinions arose in regard to how individuals found the interview experience. Although distressing for some, the overriding consensus from participants was focused on the advantages of the interview. Allowing patients to articulate their feelings and emotions and having someone listen to their story, gave them a sense of power, especially as they realised their input would raise awareness and influence future care.

Table 13 - Illustrative quotes reflecting the coping mechanisms used by vEDS patients.

Coping mechanisms	Quotes
4.10.1 Positive attitude and outlook on life	<p><i>“we accept we’ve got this condition and we can make adaptations, and by making adaptations means you can still do these things, but we just have to be slightly creative in how we do them”</i></p> <p><i>“you’ve got it, there’s no point in wasting your time thinking about it in a negative way”</i></p>
4.10.2.1 Exercise as a coping mechanism – physical health	<p><i>“so, going to the gym and things makes me feel better within myself... It makes me feel sort of healthier and things”</i></p> <p><i>“I done quite a lot in the gym.... Just trying to keep my physical fitness up, because if my physical fitness is better, then I can do a bit more without getting so knackered”</i></p>
4.10.2.2 Exercise as a coping mechanism – mental health	<p><i>“It has helped my body; I can notice a change in my body...it has made me stronger... And that makes me feel better psychologically”</i></p> <p><i>“so, it is good, I need exercise, if I didn’t exercise, I know psychologically I’d be worse and also my body would be so weak and then that would make me feel worse psychologically as well”</i></p>
4.10.3 Thought management and diagnosis	<p><i>“I absolutely blank out my condition, and completely pretend it doesn’t exist”</i></p> <p><i>“I’m quite happy to just put it in a box and not worry and not think about it. And that is my coping mechanism”</i></p>
4.10.4 Appreciation for life	<p><i>“I am very, very, very lucky. And that does.... It makes me probably worry less subconsciously.”</i></p> <p><i>“I’m just grateful for every year that I’m here”</i></p>

	<p><i>“That just makes you on the one hand feel very grateful...that you're not going through some of those difficulties, umm but it also is a stark reminder that everything isn't quite as rosy as it might appear”</i></p>
4.10.5 Providing a voice to the unheard	<p><u>Condition comparison</u> <i>“If I was to say I had cancer people give you sympathy, a treatment plan, people would know what you're talking about, especially medical professionals”</i></p> <p><i>“I get slightly cross when I see all the funding for cancer research because I think... They had all this support and funding for years. And there's nothing for what I have”</i></p> <p><u>Interview empowerment</u> <i>“It's nice to talk to somebody... and they listen”</i></p> <p><i>“I signed this research thing because I wanted.... I thought, well, it'd be a bit hypocritical to complain about the condition and not at least attempt to advance signs and the knowledge of it.”</i></p> <p><i>“I think it is good to recognise how you feel...I couldn't have this conversation with somebody I loved.”</i></p>

4.11 Summary of key findings

The impact Vascular Ehlers Danlos Syndrome has on an individual’s psychosocial wellbeing has been revealed through the themes presented. Whilst each individual experience of the condition differed, the overarching themes present the patient experience.

The themes were dynamic and interlinking, encompassing all the factors of patients’ lives most affected by the condition. The data indicates that the psychosocial consequences of living with vEDS comprises of several factors, although one aspect may affect an individual considerably more than another, but when combined, said factors significantly impact individuals’ lives. Physical limitations to daily life, showed to have the greatest overall impact, whilst age at diagnosis, grief and loss and the uncertainty of the condition were factors that affected individuals lives in different ways. The themes presented will be discussed in context with the literature in the next chapter.

Chapter five: Discussion

5.1 Overview

This study aimed to investigate the psychosocial impact of vEDS. For the first time, this research study focused specifically on the experiences of patients living with vEDS in the UK, exploring how the condition affected particular aspects of individuals' lives, as well as day to day living. The findings will be discussed in context to prior research discoveries including Velvin and Johansen (2018) which is referred to throughout this discussion. Their findings highlighted the impact of psychological distress, fatigue and pain in family life and daily function, as well as inconsistent support from healthcare professionals. The strengths and limitations of this research study will be assessed, along with implications for clinical practice and future research.

5.2 Day to day living

5.2.1 Fatigue

This current research study is the first to provide an in-depth analysis into the relationship of fatigue and psychosocial wellbeing in patients with vEDS. Hardy and Studenski (2010), defined fatigue as a range of characteristics that have several outcomes for patients, divided into mental and physical categories (figure 17).

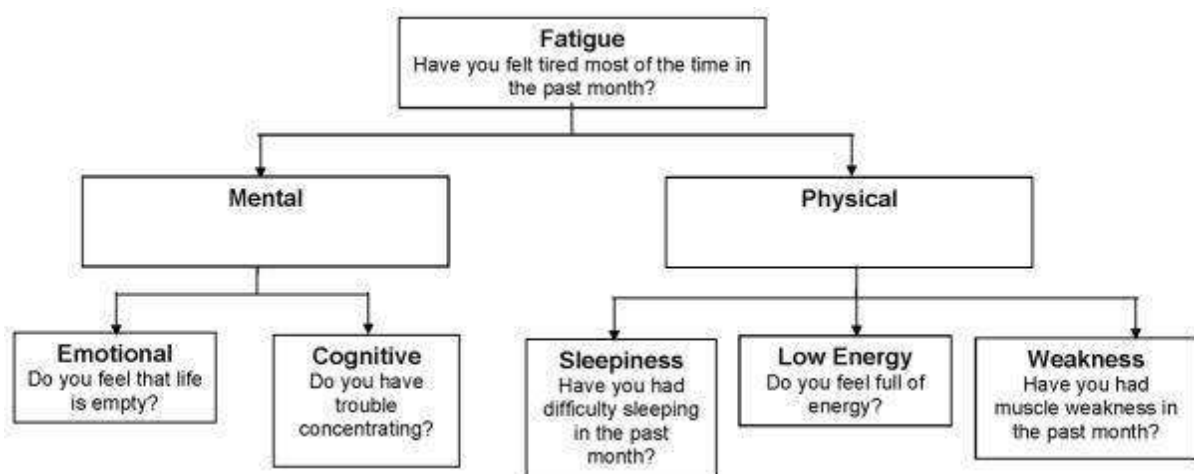


Figure 17 - Model of fatigue displayed by Hardy and Studenski (2010).

Their work compartmentalised fatigue using data from primary care patients aged 65+ years. The experiences of fatigue in the elderly population may differ greatly to those in vEDS patients and so are not representative. Nonetheless, Giroux *et al.* (2016) support the classification of fatigue into mental and physical parts, as the physical aspects include limited endurance, fluctuating capacity to carry out daily activities and difficulties with mobility. The mental aspects focus on cognitive functioning and daily productivity, as well as difficulty concentrating.

Dissimilarly, fatigue for vEDS patients in this study, is not a split entity between mental and physical outcomes, but instead presents as a direct relationship, where the physical limitations impact mental and psychosocial wellbeing.

Fatigue has shown to have widespread effects in EDS patients, impacting emotional, social, and physical wellbeing (Hakim *et al.* 2017). Furthermore, fatigue significantly restricts the lives of EDS patients, leading to difficulty completing day to day tasks, as reported by Bennett *et al.* (2018) discussed in table 2 in Section 2.4.1. Undoubtedly, fatigue has an extensive impact in vEDS and EDS patients' lives and the findings from this project support and expand on those displayed by Connors *et al.* (2012), and Velvin and Johansen (2018). Recognising the cause and effect relationship between fatigue and impaired psychosocial wellbeing is therefore crucial when supporting patients with this symptom.

5.2.2 The cycle of psychosocial challenges

Velvin and Johansen (2018) summarised that psychosocial stress was mainly observed as a cycle that included social, medical, and personal factors for patients. The findings from this current research study, support and develop this cycle further, as seen in figure 18 below.

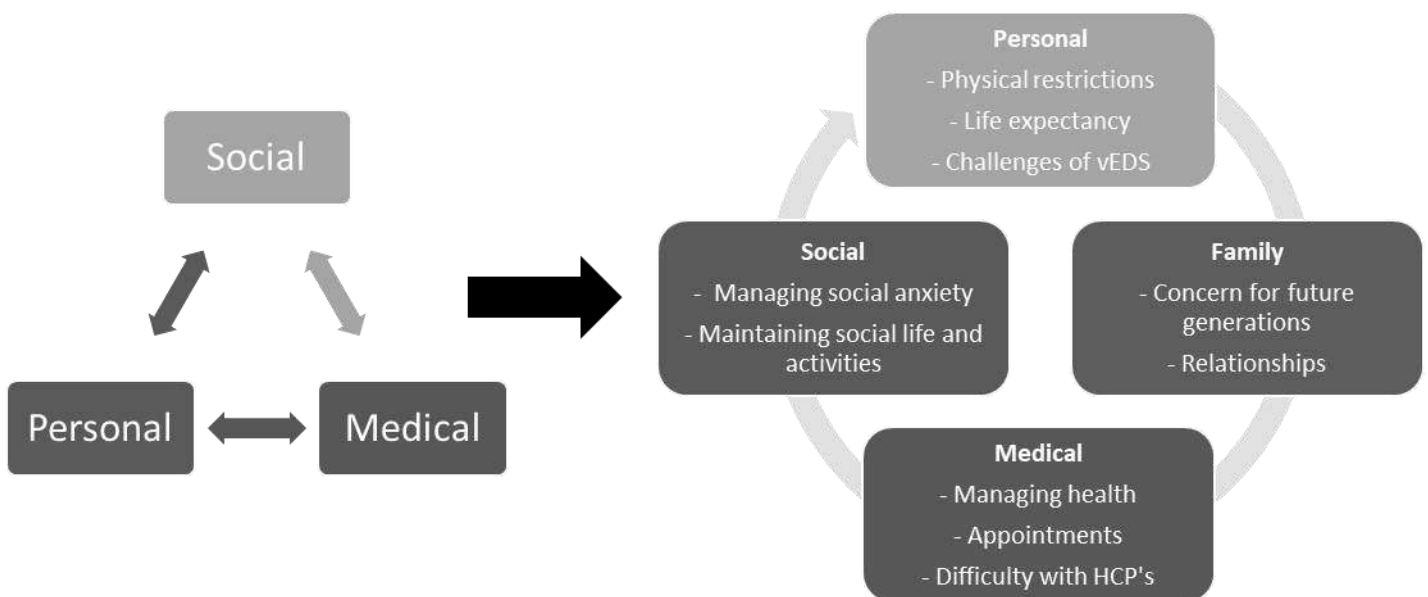


Figure 18 - On the left is the original cycle proposed by Velvin and Johansen (2018), which has been adapted to include the findings from this research study, displayed on right with the addition of family.

The introduction of 'family' into the cycle, captures the experiences of the 21 vEDS participants that took part in this research study. According to the family systems theory, the relationships between the immediate nuclear family and extended family influence one another (Papero *et al.* 2018).

Therefore, any anxiety or challenges patients face, affect their personal lives, the lives of their immediate and extended family and consequently have a ripple effect across wider social systems including work, education, and healthcare.

5.3 Ambiguity of vEDS

The uncertainty of vEDS and the sudden risk of a serious event, potentially leading to death, applied additional worry and distress for individuals, directly impacting their psychosocial wellbeing.

The paper titled 'Am I going to die today' interviewed 14 LQTS patients from New Zealand and displayed the detrimental impact sudden death had on patients quality of life, as they tried to cope with the concept of an uncertain ending, which significantly contributed to low psychosocial wellbeing (Watson, 2011). Although the study sample reflects the views of patients in New Zealand, reducing the generalisability of the results, Watson (2011) recognised the impact of uncertainty on psychosocial wellbeing, supporting the concerns voiced from vEDS patients in this study and those in previously discussed in the literature (Bennett *et al.* 2018; MacCarrick *et al.* 2014; Andersen *et al.* 2008).

Economy and finances surfaced as a concern for MFS, vEDS and LDS patients interviewed by Velvin and Johansen (2018). In this study, participants encountered conflicting views in regard to managing their finances as they constantly toyed with the reality that they may not reach retirement age and so saving for a pension was somewhat meaningless. But, on the other hand individuals admitted that paying into a pension ensured financial stability for their future and presented the option for early ill health retirement, with recent research showing that 31% of 52 vEDS and LDS patients had left work before retirement age (Johansen *et al.* 2019). This current research study is the first to expand on the financial burden vEDS places on patients' lives emphasising the need for financial support and advice to assist patients.

5.4 Decision making

Entering into a relationship when living with a progressive genetic condition involved a considerable amount of thought and concern for participants, as they recognised the long-term commitment involved. Similarly, patients with Huntington's Disease and breast cancer felt that deciding whether

to date along with finding the right time to inform partners of their condition, were the main anxieties faced in regard to entering a relationship (Klitzman and Sweeney, 2010).

Those that had not started a family and were aware of the condition, faced the difficulties surrounding reproductive decision making. Patients were aware of the significant risk of pregnancy and childbirth, as interviews with 39 female vEDS patients revealed that 14.5% encountered life threatening complications (Murray *et al.* 2014). Therefore, when making decisions about starting a family, patients are not only considering their child's life, but their own lives too.

Reproductive decision making is challenging for individuals with a genetic condition as there is the constant fear that they will pass the condition onto their children. Barlevy *et al.* (2012) conducted focus groups and interviews with families that had a history of LQTS and sudden death. For some, the inability to monitor their child's health and the fear of their child's future health, were deciding factors in reproductive decision making. One participant explained "I can have them x-rayed...but I have no control over their hearts and that scares the living shit out of me" (pp. 6). As LQTS is very similar to vEDS, these findings highlight the multitude of thoughts and factors to consider in reproductive decision making for patients.

5.5 Family life

Concern for children, partners and future generations surfaced throughout the interviews as the impact of diagnosis was pertinent in the family unit.

According to the family systems theory, a family is considered to be an emotional unit, where each individual has a specific role, in which their behaviours impact other members and potentially lead to dysfunction or imbalance in the system (Bowen, 1966). A genetic diagnosis can impact the family unit in several ways, as families adjust to coping with; the possible genetic risk, possible decision-making for additional family members and attempting to incorporate new information about a genetic condition into the family circle (Mendes *et al.* 2017). Rolland and Williams (2005) developed the Family Systems Genomic Illness Model to highlight the impact of a genetic condition within a family unit, acknowledging the ongoing processes that needs to be considered. Medical information, communicating risk, living with uncertainty and anticipatory loss such as inevitable future losses, showed to elicit significant psychosocial challenges within a family and are similar to the findings presented in this research project.

5.5.1 Worry on behalf of family members

Worry on behalf of family members and concerns for future generations have surfaced as complex challenges for vEDS and associated conditions (Lumley et al 1994; Andersen *et al.* 2008; Connors *et al.* 2012; Velvin and Johansen, 2018). Smith *et al.* (2013), reviewed 34 studies relating to the experiences of living with a child with a long-term condition, which identified similar concerns to those discovered in this study (table 14).

Table 14 - Concerns displayed in parents with a child that has a long-term condition (Smith *et al.* 2013). Results are displayed in comparison to those discovered in this study.

Concerns from vEDS parents in this research study	Patients' lives (this research study)	Their child's life (this research study)	Themes identified in research by Smith <i>et al.</i> 2013
Life expectancy	If they die, will their children receive adequate care	Limiting life expectancy for their child	Grief and loss, physical and emotional overburden
Understanding	Continually learning about how the condition affects their lives	Recognising the challenges, the condition presents for their child	Learning about the condition, working with healthcare professionals
Health management	Ensuring they manage their health	Managing the care and support for their child	Monitoring symptoms and responding to the changes in their child's condition
Social engagement	Managing their own social lives, despite their own health and childcare	Encouraging their child to engage in social activities despite their condition	Maintaining normality
Relationships	Ensuring there are people around to support with their care	Ensuring their child is also surrounded by family/friends	Seeking support systems, maintaining relationships

Although Smith *et al.* (2013) did not elaborate on the specific chronic conditions analysed, comparing the findings from this research project against literature focusing on the impact of caring for a child with a chronic condition, highlights the similarities in patient experiences.

5.5.2 Relationships

The role partners play in helping patients manage their condition is displayed in the Venn diagram below.

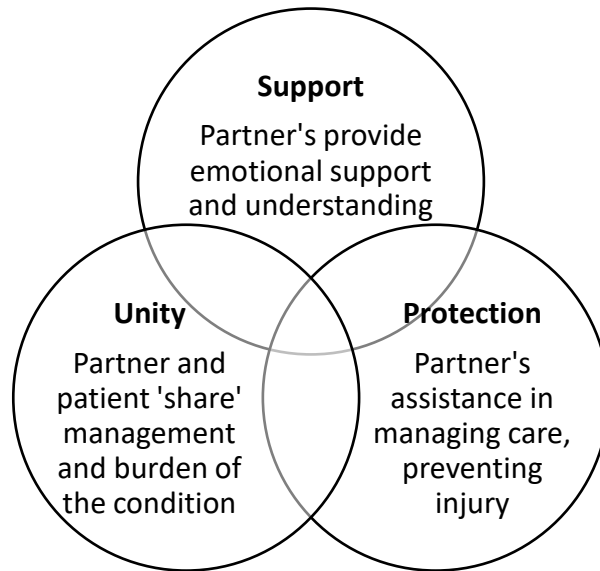


Figure 19 - Support received from partners, as reported by patients in this research study. The Venn Diagram highlights that there is a continual overlap.

Martire and Helgeson (2017) investigated how chronic illness was managed within couples and discovered various relationship constructs, several of which align with the comments made by participants in this study (table 15).

Table 15 - Relationships constructs identified by Matire an Helgeson (2017), which can be applied to the findings from this study.

Relationship construct	Description
Supportive	Emotional assistance with partners illness
Overinvolved, overprotective	Excessive assistance that undermines patient independence
Shared responsibility and coping	Patient and partner work together to manage and cope with the illness

Participants were extremely grateful for the support they received but, in some cases, overprotection became a frequent frustration, as they felt their partners doubted their ability to do certain things. Nevertheless, the supportive, shared responsibility and coping constructs accurately depict the experience of most vEDS patients with their partners, validating the findings from this research study.

Several participants felt it was worth noting the need for psychological support for their partners, regardless of whether they discussed the condition openly with them.

Recent investigation into the challenges of living with a spouse with a chronic illness, brought light to some of the obstacle's partners face including adapting their social lives, seeking support, and worrying about the future (Eriksson *et al.* 2019). However, the paper fails to specify the chronic illnesses partners suffered, restricting the application of these findings to vEDS patients. Regardless, Eriksson *et al.* (2019) emphasise the need for support for partners of those with a chronic condition, justifying assistance for partners of vEDS patients, where fragility of life is intensified.

5.6 Grief and loss

The evidence from this study highlighted the abundance of loss individuals experienced due to their vEDS diagnosis.

5.6.1 Anticipated loss

Throughout the interviews anticipated loss surfaced in relation to:

1. The loss of future health and identifying as a healthy person.
2. Possibly losing a family member to vEDS.
3. Not being able to have children and identify as a mother/father.
4. Anticipating the loss of an affected child's health.

The extent to which anticipated loss affects individuals' lives has been discussed in-depth throughout this chapter of the discussion.

5.6.2 Grieving for a child with vEDS

Parents of a child with vEDS anticipated the loss of their child's health and future as they adapted to the inevitable restrictions of the condition, subsequently leading participants to feel that the future plans set out for their children may not play out as expected.

Parents of children with Juvenile Chronic Arthritis (JCA) expressed their experiences of caring for their child as a "a grief you have inside you" (Sallfors and Hallberg, 2003, pp. 199). Although JCA has minimal similarities with vEDS, supportive evidence can be found in related disorders such as MFS. Child and Rowntree (2016) found that grieving for the loss of perfect health in a child and worry for their child's future, were commonly reported by parents. The authors suggest that psychological support and contact with another family who have an older child with MFS would be

beneficial to parents in managing and rationalising the continual grief and anticipated loss they experienced for their child. Such interventions may also be beneficial for vEDS parents.

A paper titled ‘When I go into wake them, I wonder’, depicted the reality of caring for a child with LQTS, as 19 out of 31 parents lived with the uncertainty of losing their child prematurely due to the condition. Farnsworth *et al.* (2006) acknowledged that the sample of parents recruited had older or adult children at the time of their interview, so these individuals provided retrospective answers. The difficulties in establishing a cause and effect relationship in retrospective accounts due to possible missing data, limits the application of these results (Toftthagen, 2012). Nevertheless, this study emphasises the impact of caring for a child with a sudden death disorder, validating the findings from this current research project and supporting those discussed by Spanaki *et al.* (2016) as parents reconsidered their child’s life choices.

5.6.3 Loss of a family member

Losing a family member to vEDS shaped participants’ perceptions of their own health and life expectancy. A case study revealed that a woman living with vEDS, felt the loss of her mother to the condition, triggered her artery dissection due to psychological distress (Shimoyama *et al.* 2017). The main weakness of this study is the use of a case study approach, as it focuses on an individual data account producing ungeneralisable results. However, case studies do permit the in-depth exploration of complex issues (Crowe *et al.* 2011) and in this case, present an area where additional psychological support may be needed.

5.6.4 Disenfranchised grief

Disenfranchised grief has been described as the grief “that persons experience when they incur a loss that is not or cannot be openly acknowledged, publicly mourned or socially supported” (Doka, 1989, pg. 4). There are several reasons as to why disenfranchised grief occurs, some of which include lack of acknowledgement for the loss and the nature of the loss involved (Doka, 2009). The root of disenfranchised grief for vEDS patients may stem from the invisible nature of their condition, which potentially results in individuals feeling alienated and unsupported by society, when they experience grief surrounding their diagnosis (Thompson *et al.* 2016). Examples from participants touched on the socially accepted grieving ritual for cancer patients, as they continually receive sympathy and support. Doka’s original theory of disenfranchised grief is based upon the concept that grief is separate from a person’s identity, proposing it is linear and a temporary state of being (Barney and Yoshimura, 2020). This is not the case for the vEDS participants within this study, as the diagnosis lead to continual grief and loss throughout their lives.

In addition to this, the invisible nature of hEDS has shown to leave patients feeling judged and outcast as discussion of the condition confuses others. “Other people’s judgement...if you don’t fit that notion of what disabled is in their head, they get confused”, Bennet *et al.* (2019, pp. 4) encapsulates the effects of living with a condition that others cannot see or understand and therefore cannot sympathise with.

5.6.5 Compounded grief

Compounded or complicated grief can be described as a form of grief with heightened intensity and a prolonged duration (Shear *et al.* 2011). Participants spoke of a never-ending grieving process in relation to receiving their diagnosis, and the subsequent testing of family members. There are several factors that can exacerbate grief including: the circumstances surrounding loss and historical, social and personality factors (Worden, 2010). The findings from this current research study present compounded grief as an additional dimension of loss that participants face.

5.7 Age at diagnosis

Age at diagnosis influenced participants’ experiences, decisions, and attitudes towards their health. The viewpoints displayed by individuals diagnosed at an older age showcased the advantages of receiving a diagnosis later in life.

A study with 18 vEDS patients revealed the median age at diagnosis was high, at around 30 years old. Johansen *et al.* (2020) speculate that presumably by this age, individuals would have chosen their career and educational paths before receiving their diagnosis. They hypothesise that as the age at diagnosis reduces for vEDS patients in the future, it will influence individuals’ choices on their education and work. The study methods used to collect this data involved self-reported postal questionnaires, which have been criticised due to potential bias and the lack of detail in responses provided by participants (Demetriou *et al.* 2015). Despite this, it is clear that the findings from Johansen *et al.* (2020) validate those discovered in this research study, as a diagnosis later in life did not impact participants’ occupation or education, in the same way that it did for those diagnosed at a younger age.

A diagnosis later in life appeared to reduce anxiety around social activities and decisions around having a family. As many explained their guilt and grief in passing the condition onto their children, or the traumatic experiences they encountered during their pregnancy and childbirth, they were able to recognise that a later diagnosis did not present a decision making process in regard to having children.

Although participants reported personal advantages to receiving a diagnosis later in life, it is worth considering the medical advantages of being diagnosed at a younger age. In the UK, if a child is at 50% risk of inheriting vEDS, they are treated as if they had a confirmed diagnosis. Therefore, advice on the condition is provided early on, increasing understanding, whilst ensuring individuals are equipped with tools to manage their health effectively. In addition to this, an early diagnosis ensures that protective lifestyle factors are instilled from a young age, guiding patients on appropriate low impact hobbies.

Exposure to a genetic diagnosis at a young age empowers children as they receive greater autonomy over their own health needs and behaviours. Furthermore, learning of the condition early on prepares children for the future obstacles they may face, such as reproductive choices and planning their care. Finally, normalising conversations surrounding the topic, help children adapt to the condition becoming a part of their identity (Rowland and Metcalfe, 2013).

Metcalfe *et al.* (2011) conducted semi-structured interviews with children (8-11 years) and young people aged 12+ years, from families affected by a genetic condition. Table 16 compares the thoughts expressed by those that received their diagnosis at a young age, against individuals that learned about it as they got older i.e. in their teenage years.

Table 16 - Participants describing their experiences of learning about their diagnosis. Participants reflected retrospectively and spoke about how it felt to receive their diagnosis during their teenage years or at a younger age.

Thoughts on learning of the diagnosis at a younger age	Thoughts on learning of the diagnosis in teenage years
Learning about the condition with gradual information from a young age increased understanding over time	Difficulty adjusting to the condition
Provided insight into the condition along the way	Difficulty understanding the risks of the condition to themselves and family members
No shock, as they did not learn about all the information at once, less overwhelming	Shock of learning about it all at once

Receiving a diagnosis during teenage years, when individuals are starting to cement their own identity, whilst pulling away from their family unit, can be a really difficult time to learn and come to grips with a life changing condition such as vEDS.

Hence, it is important to drip feed information to children to aid understanding, acceptance, and adaptation to a genetic diagnosis, thus supporting the benefits of receiving a diagnosis at a young age.

Sequencing the exomes or genomes of new-born babies for rare disease could permit a lifetime of tailored medical care and identify rare disease that may affect a child's life from birth or as they get older (Kingsmore, 2015, Johnston *et al.* 2018). However, expanding new-born screening to a Whole Genome Sequencing (WGS) approach raises ethical issues surrounding societal questions about nonmaleficence, beneficence, autonomy, and the preservation of each child's open future (Berg *et al.* 2017; Lantos, 2019). The conflicting attitudes towards WGS in new-born babies, raises considerable healthcare and ethical challenges, but it cannot be ignored that such technologies may provide long-term benefits in managing rare diseases such as vEDS.

For certain participants, the uncertainty of their diagnosis lead to prolonged symptoms and serious events that were unexplained throughout their lives. For others receiving a diagnosis at a younger age helped with the overall management of the condition. Evidently, understanding how age at diagnosis may influence psychosocial wellbeing can aid the management of the condition further.

5.8 Inner strength and mental health

Mental health issues arose as a result of the condition, presenting additional challenges for certain patients, resulting in the need for counselling and medicinal support. In the face of all these challenges, this cohort of participants were admirable, as they courageously placed themselves in a vulnerable position and spoke openly about their experiences to benefit the wider community. Participants' motivations to take part in this current project are similar to those seen across other patient groups. Cancer and cardiac patients reported that their motivations to take part in research were based on altruistic and intellectual reasons, including the benefit to society, and helping future patients. Having an interest in the research question at hand was also a driving factor (Goodman *et al.* 2019; Soule *et al.* 2016).

5.9 Importance of reflection

Individuals were given the opportunity to reflect on their experience of taking part in the project during the debrief phone call and addressed any emotions or thoughts that arose as a result of the process. The coping strategy of positive reappraisal can explain certain participants' responses.

As participants reflected on their interview experience, they acknowledged the value of their discussion and were sincerely thankful that they were given the opportunity to speak on behalf of the

vEDS community. Positive reappraisal is adapted by individuals as they find “personally-relevant positive meaning from an experience in the face of its negative reality” (Nowlan *et al.* 2015, pp. 475). The systematic literature review presented here, is focused on positive reappraisal in older adults with declining health issues, reducing the generalisability of the results to this research project, as vEDS occurs in all ages. However, positive reappraisal has shown to play a significant role in helping individuals adjust to and manage negative situations, discovering meaning in their actions (Garland *et al.* 2009; Finklestein-Fox *et al.* 2018), and can be seen as a coping mechanism used by individuals within this study.

The vulnerability of the interview made some individuals aware of how they dismiss their thoughts and feelings in relation to the condition and place them in the back of their mind to help them cope. No one found themselves thinking about the interview negatively, but the debrief phone call revealed that all participants were aware of the power of the interview in addressing their own concerns and providing a platform.

5.10 Limitations and strengths

The limitations and strengths of the research presented have been summarised in table 17 below.

A key strength of this study was that the sample of participants represented the target population as 21 vEDS patients, both male and female, within the UK participated. It is worth noting the novelty of this research project, as it includes the largest set of psychosocial interview data, from solely vEDS patients and has supported and expanded on previous research such as Velvin and Johansen (2018) and Connors *et al.* (2012).

Table 17 - Summarising the strengths and limitations of the research study

Strength	Limitation
Generalisable: representative sample and ‘large’ sample size for an exploratory study – results can be applied to wider vEDS community	Sample bias: potential that individuals more engaged in the vEDS charity, decided to take part in the research study, reducing external validity of the results
Valid: data saturation achieved; no new themes emerged after 15 th interview	Medical records: no access to patients records to confirm diagnosis, diagnosis confirmed internally at AC using the screening process described in chapter three, section 3.2. But 95% of participants are known to the EDS Service, so would therefore have a confirmed diagnosis
Reliable: results are consistent with previous published literature on vEDS and associated conditions	Small number of men: disproportionate number of male participants, results do not accurately represent the male experience
Rich, detailed data: semi-structured interviews permitted guided discussion and ensured a uniform approach in data collection. Also enabled participants to explore the impact of the condition surfacing new topics and potential themes such as intimacy	Interviewer bias: novice qualitative researchers are more likely to demonstrate interview bias due to lack of scientific rigour and subjective opinions (Noble and Smith (2015))

Vasileiou *et al.* (2018) analysed sample size sufficiency over a 15-year period in qualitative research. They explained that results can be viewed as generalisable if the study sample was large enough to unfold new levels of understanding, but small enough to ensure that the deep, rich informative nature of qualitative methods is not overlooked. With this in mind, the outcomes of this study can be considered as generalisable to the wider vEDS community as the study methodology aligned with the guidance above.

Another strength of this study is that the validity of the results is supported through achieving data saturation by the 15th interview when no new themes emerged. There is no standard guidance on the number of interviews needed to achieve data saturation and some report that 20 to 30 participants would suffice (Marshall *et al.* (2013). However, the framework used to reach data saturation estimates is still widely unknown (van-Rijnsoever, 2017). In qualitative research it is at the discretion of the researcher to decide whether to continue interviewing and collecting data, if the goals of the research have been achieved (Fugard and Potts, 2014). Therefore, the decision to conduct 21 interviews, was based on two factors. The first being the novelty of the research, as it was an innovative exploratory study, so including more participants ensured that data saturation was met. Secondly, on reflection, the author was a novice researcher with no experience conducting qualitative research, so aimed to guarantee data saturation.

The reliability of the study is supported by comparing the findings with those from previous research studies, on vEDS and similar conditions. The consistency of the results presented in the data, align with the common themes and psychosocial challenges reported in the literature review. Table 18 compares the findings from this research study against the published literature exhibited in table 5 in the literature review.

Table 18 - Comparison of the findings from this research study against those reported in previous literature.

Psychosocial issue/concern	Condition and Reference	Finding from this research study
Pregnancy/worries about future children	EDS: Lumley <i>et al.</i> (1994) TAA disorders: Connors <i>et al.</i> (2012)	Decisions around having a family, concern for future generations
Reproductive decision making	MFS: Nielsen <i>et al.</i> (2019) LQTS: Andersen <i>et al.</i> (2008)	Loss of having a family, reproductive decision making
Physical functioning and restrictions to everyday life	EDS: Lumley <i>et al.</i> (1994) LQTS: Andersen <i>et al.</i> (2008) MFS: van-Tongerloo and Paepe (1998) HCM: Cox <i>et al.</i> (1997) MFS: Velvin (2015)	Covered throughout, physical limitations to daily everyday life
Chronic pain and fatigue	EDS: Lumley <i>et al.</i> (1994) vEDS: Velvin and Johansen	Physical restrictions to everyday life

	(2018) MFS: Velvin (2016) MFS: Hansen <i>et al.</i> (2020) HCM: Cox <i>et al.</i> (1997)	
Limitations to social life	MFS: Tongerloo and Paepe (1998) HCM: Cox <i>et al.</i> (1997)	Anxiety in social situations, reduced desire to socialise, support and communication with friends
Family life and lifestyle choices	vEDS: Velvin and Johansen (2018) HCM: Spanaki <i>et al.</i> (2015) MFS: Velvin (2015)	Family life, communication within the family, decision making, responsibility
Finance	vEDS: Velvin and Johansen (2018)	Responsibility of personal finances and savings including pensions
Sexuality and partnership	vEDS: Velvin and Johansen (2018) MFS: Velvin (2015)	Intimacy and relationships with partners, barriers to relationships, communication within relationships
Grief and loss	TAA disorders: Connors <i>et al.</i> (2012) LQTS: (children) Chatta and Zelenietz (2011)	Loss in patient's lives and the grieving responses to this, loss of child's health, anticipated loss, disenfranchised grief, compounded grief
Limitations in working life and study	MFS: Velvin (2016) MFS: De Bie <i>et al.</i> (2004) MFS: Tongerloo and Paepe (1998) MFS: Hansen <i>et al.</i> (2020) MFS: Nielsen <i>et al.</i> (2019) MFS: Velvin (2015)	Career progression and educational limitations, achievements, and accomplishments
Loneliness/isolation	LQTS: Andersen <i>et al.</i> (2008) LDS: MacCarrick <i>et al.</i> (2014)	Isolation, comparison, reduced confidence, and independence
Substance abuse	HCM: Teo <i>et al.</i> (2014)	N/A
Frustration with healthcare professionals	EDS: Bennett <i>et al.</i> (2018) vEDS: Velvin and Johansen (2018) LQTS: Andersen <i>et al.</i> (2008)	Lack of support from healthcare professionals influencing care

Anxiety and worry	LQTS: Andersen <i>et al.</i> (2008) HCM: Cox <i>et al.</i> (1997) HCM: Steptoe <i>et al.</i> (2000) HCM: Ingles <i>et al.</i> (2008) HCM: Teo <i>et al.</i> (2014)	Quantitative data on the prevalence of mental health across the study cohort, anxiety around health and health management
Fear of the unknown/uncertainty	EDS: Bennett <i>et al.</i> (2018) LDS: MacCarrick <i>et al.</i> (2014) LQTS: Andersen <i>et al.</i> (2008)	Risk perception and existential thoughts, living cautiously, facing mortality, the future and growing old
Quality of life	MFS: Nielsen <i>et al.</i> (2019) HCM: Spanaki <i>et al.</i> (2016) MFS: Velvin (2015)	N/A
Communication (need for support)	LQTS: Andersen <i>et al.</i> (2008)	Communication and support; family, friends, support groups
EDS: Ehlers Danlos Syndrome		
LDS: Loeys Dietz Syndrome		
LQTS: Long QT Syndrome		
HCM: Hypertrophic Cardiomyopathy		
MFS: Marfan Syndrome		
TAA disorders: Thoracic Aortic Disorders		

5.11 Implications for clinical practice

Recognising the variability of vEDS across the study sample emphasised the need for individual patient care recommendations and not a blanket ‘one size fits all’ approach. For many participants understanding and awareness from HCP’s was imperative to their experience with the condition. Similar concerns were reported in patients with MFS, LDS and vEDS by Bennet *et al.* (2018), Velvin and Johansen (2018) and Andersen *et al.* (2008).

Sulli *et al.* (2018) investigated the volume of published literature and guidelines available to healthcare professionals regarding EDS. In most scenarios, vEDS was diagnosed after a near death event and there was little/no educational literature and clinical practice guidelines for vEDS available. Although the systematic review was conducted by a European Network which excludes the UK, Sulli *et al.* (2018) showcase the unmet needs of vEDS patients as there is minimal literature provided for healthcare professionals.

The UK EDS National Diagnostic Service was founded in 2009 and supports vEDS patients across the country.

The service is commissioned to see 200 patients a year but follow up appointments are offered in some cases. Since 2011 patients have attended a multidisciplinary team (MDT) clinic with cardiology and genetics consultants, a cardiology clinical nurse specialist and a GC. Patients are seen annually for clinical follow up with appropriate imaging and treatment at one of the EDS services in Sheffield Children’s Hospital. Some patients’ care is managed by local clinicians, who contact the EDS service for specialist advice. Sheffield have recently advanced the management for the psychosocial aspects of the condition, as patients now meet with a GC and a staff member from AC.

Care plans are not efficient for vEDS patients as they see the HCP available at the time they present to Accident and Emergency (A&E). Each patient receives an A4 medical information form and emergency wallet card from the EDS service and are encouraged to pass these onto the medical alert team, to ensure that the HCP managing patients care is educated and aware of the condition.

There are approximately 300 patients in the UK with vEDS, so it is difficult to educate all local HCP’s about the condition. Overall, at service evaluation, patients report a high level of satisfaction with the EDS service. Reassuringly, there is an MDT approach to managing vEDS patient care in the UK, however, there is room for improvement within the service, specifically in terms of the role of GC’s in supporting the psychosocial and coping aspects of the condition (figure 20).

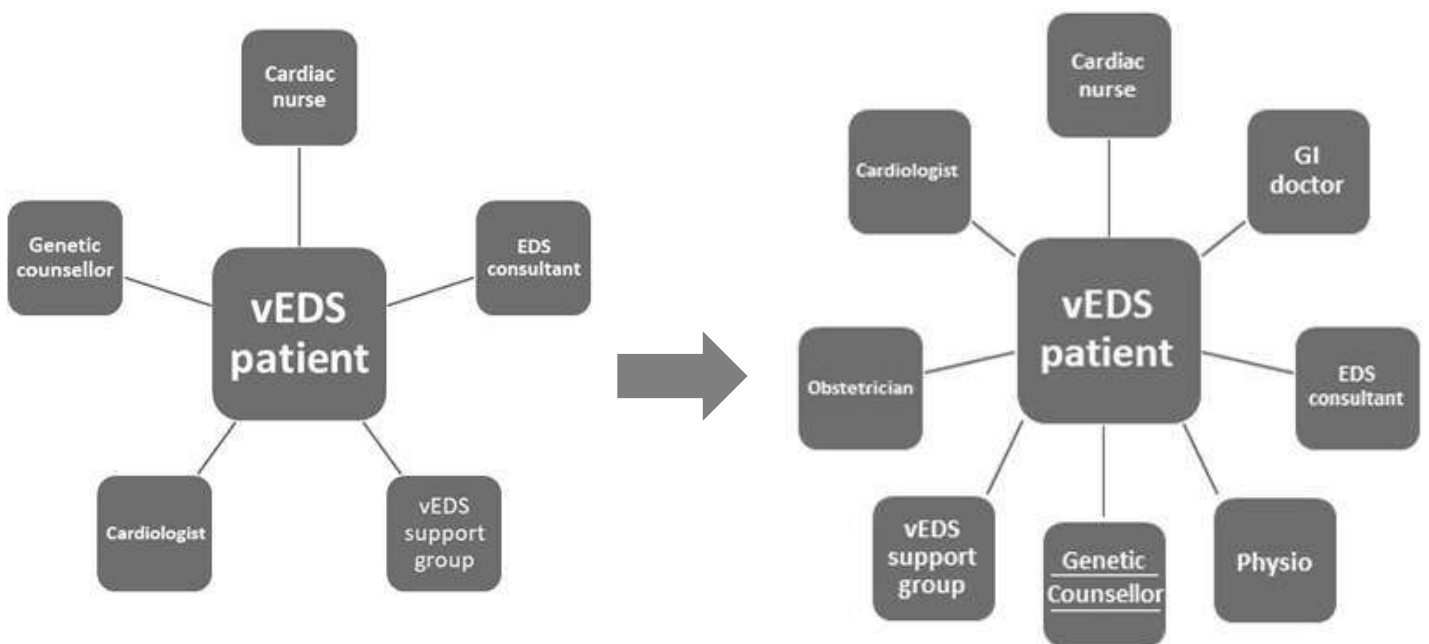


Figure 20 – Left: the current MDT team for vEDS patients in the EDS National Diagnostic Service. Right: Potential improvements of the service, focusing on all clinical aspects of vEDS, including Gastrointestinal (GI) care, obstetricians and physiotherapists.

The reciprocal engagement model of genetic counselling is built on the foundations of “patient-centred education and counselling, understanding and appreciation of the patients unique situation, support and guidance to patients to build rapport and trust and facilitative decision making” (Schmidlen *et al.* 2018, pp. 2). Although the model emphasises the use of counselling and therapeutic engagement with patients, this is not necessarily the case in the UK, due to limited time and resources for follow up appointments, along with additional difficulties displayed in table 19 below. In most settings genetic counselling takes a psychoeducational approach, with a focus on providing information, rather than fulfilling the reciprocal engagement model of genetic counselling. Biesecker (2020) highlights that current genetic counselling is primarily centred on “counsellor speak” (pp. 4) focusing on genetic information provision, with minimal attention to patients’ psychological needs.

Table 19 - Adapted from Biesecker (2020) displaying the potential difficulties genetic counsellor’s face when attempting to deliver a psychotherapeutic and psychoeducational approach.

Obstacle	Explanation
Development of genomics	Focuses GC attention on science
Ability to explain genetic concepts	Individual capacity to explain how genetics works
Complex psychological responses	GC’s may feel intimidated to address these
Graduate education	Less prepared to provide psychosocial support

Addressing issues such as grief and loss, relationships and the emotional elements that arise when living with a genetic condition, facilitated decision making and knowledge retainment in patients with genetic conditions (Edwards *et al.* 2008). Additionally, focusing on the emotional and psychosocial needs of patients elicited better psychological results and in turn supported knowledge-based outcomes (Austin *et al.* 2014).

Psychologists place greatly within a vEDS patients’ MDT, as their skills can effectively address psychosocial distress and wellbeing and improve the communication of genetic risk amongst relatives (Mazella *et al.* 2020; Caleshu *et al.* 2016). Equally, GC’s play a vital role in supporting all patients, especially when promoting a psychotherapeutic approach to patient care. Additional training in counselling and psychological support, such as CBT and psychotherapy methods which patients in this study frequently accessed, can further promote the psychotherapeutic approach, and showcase the importance of GC’s in facilitating patient’s psychosocial wellbeing effectively.

5.12 Future research prospects

As literature surrounding the psychosocial aspects of vEDS is still in its infancy, there are several opportunities for additional studies (figure 21).

It appears that vEDS patients are willing to take part in research, as 82.9% of 300 vEDS patients from a support group wanted to share their experiences to help researchers, physicians and others with the condition, reported via questionnaires (Shalhub *et al.* 2020). The dissemination of this survey into a support group via social media platforms such as Facebook, questions the validity of the sample group as patient diagnoses were not confirmed. Furthermore, some of these groups do not have identifiable moderator membership admission procedures, so anyone could complete the survey (Gelinas *et al.* 2017). Nonetheless, the positive response to participation from vEDS patients supports future studies.

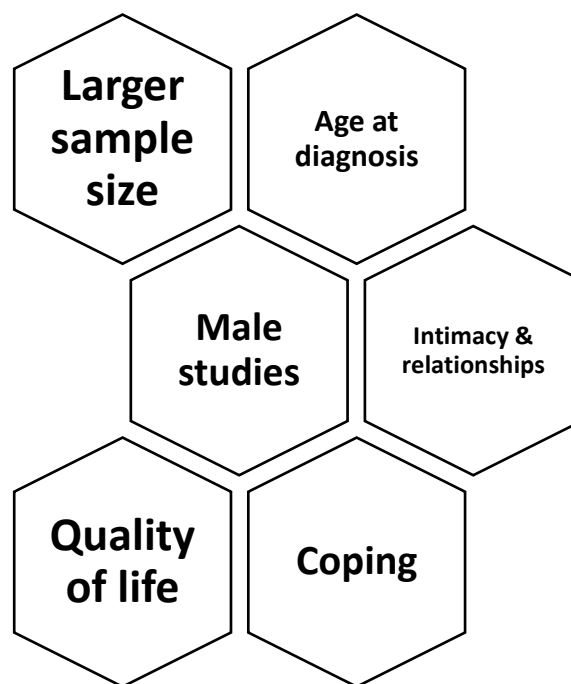


Figure 21 - Future research opportunities to consider investigating in vEDS patients.

5.12.1 Sample size

The recruited sample size is relatively large considering there are only 300 vEDS patients in the country, creating difficulties when attempting to repeat this study amongst a larger group. Therefore, additional psychosocial studies could focus on the different aspects of vEDS such as the impact on patients under the age of 18 using quantitative or qualitative methods, or the effects of the condition on affected and unaffected family members.

5.12.2 Age at diagnosis

Research into how age at diagnosis affects vEDS patients and their management of the condition, could impart further knowledge on the differences in experiences between ages. Interviews with

adolescent MFS patients discovered that difficulty keeping up with peers, feeling different to others and self-esteem/image were significant challenges for individuals (Warnink-Kavelaars *et al.* 2019).

Moreover, the review of 630 clinical records of patients with vEDS revealed that arterial rupture was responsible for all deaths in young males. Additionally, 18% of male deaths were caused by vascular dissection or rupture which occurred before the age of 20 (Pepin *et al.* 2014). Exploring the impact of vEDS in teenagers and young people could expose the difficulties these individuals face on a daily basis. Such studies warrant the need for additional support for younger patients with the condition, such as a buddy network with older individuals affected by vEDS, or psychological support to help boost confidence and address anxiety.

5.12.3 Male studies

As mentioned, the study sample included three male participants, two of which were under the age of 25. Therefore, experiences from a larger cohort of male participants, as well as insight into the condition for men over 25 affected by the condition, is required as this group was significantly underrepresented in this study.

5.12.4 Intimacy and relationships

Participants spoke of the pain involved in sexual intimacy and the fear of provoking a vascular episode. Women who had experienced perineal trauma commented on the impact this had on intimacy. Painful and difficult sexual intercourse alongside dissatisfaction with sex life were expressed by participants (Rathfisch *et al.* 2010; Gommesen *et al.* 2019). Additionally, 60% of 174 patients with MFS reported a lack of sex drive as a result of symptoms such as pain (Peters *et al.* 2002). Psychosocial investigation into living with joint hypermobility EDS also revealed difficulties with sex and intimate relationships, as a result of the fatigue and pain that the condition presented (Bennett *et al.* 2019).

Evidently, pain associated with intimacy can impact individuals' lives significantly. Only a handful of participants explored this topic in detail and explained that it is often overlooked when considering how the diagnosis affects their lives, affirming the opportunity for further research into the impact vEDS has on relationships and the experience of partners.

5.12.5 Quality of life studies

There is minimal published literature investigating the Quality of life (QoL) in vEDS patients specifically, as most studies investigate this collaboratively with other conditions (Velvin *et al.* 2019a). Berglund *et al.* (2015) distributed a postal survey measuring health related QoL (HRQoL)

and anxiety and depression. Of the 250 responses, 10 of which had vEDS, a lower HRQoL was reported amongst the Swedish participants, as well as higher levels of anxiety and depression. As the results are presented for the sample collectively, it is difficult to gauge the HRQoL in vEDS patients exclusively.

When developing a QoL study for a rare disease there are certain criteria and factors to consider. One method used to assess QoL is via Patient Reported Outcome Measures (PROMS) as they provide direct reports from patients regarding their health, their condition and their QoL. When designing PROMs for rare conditions, the development of content validity is important and input from patients with the lived experience of the condition should be included. PROMS can benefit clinicians and inform consultations with patients, promoting patient-centred care (Slade *et al.* 2018; Rottenstein *et al.* 2017). However, developing a condition specific QoL scale is a significantly time-consuming research process, where funding is rarely provided. But, utilising the findings from this project and additional input from vEDS patients could help develop an effective QoL study.

5.12.6 Role of exercise and coping mechanisms

Generally, studies investigating QoL in genetic conditions focus on psychological wellbeing, physical manifestations, and illness perceptions, with minimal attention on effective interventions for patients. Cohen and Biesecker (2010) emphasise the need for QoL research that addresses the coping strategies used by patients, in attempt to enhance and improve their quality of life.

This research study highlighted exercise and positive mental attitude as pivotal coping mechanisms in individuals' lives. Alternative help, a conscious way of living and positive attitudes showed to be the most commonly reported management techniques in 579 patients with chronic pain on a study questionnaire (Bussing *et al.* 2010). However, this study makes no attempt to address the potential social desirability bias in participants responses, as individuals may have wanted to look like they are actively using coping mechanisms to manage their condition (Rosenman *et al.* 2011).

Physical exercise as a coping mechanism has been explored in individuals with TAA disorders including vEDS. 36 MFS, LDS and vEDS patients expressed the struggle to strike a balance between over and under exercising emphasising the need for additional research in this area to establish a safe balance (Velvin *et al.* 2019b). Research into interventions and effective coping mechanisms has been explored in other EDS types. 17 participants with hEDS expressed the coping mechanisms they used to address the psychosocial impact of their condition and these are summarised in figure 22 below (Bennett *et al.* 2019).

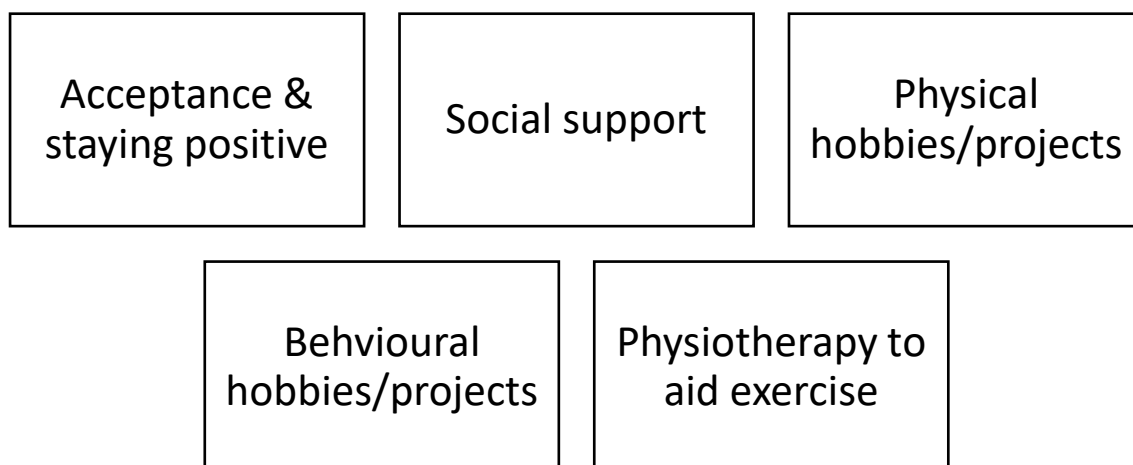


Figure 22 - Coping mechanisms used by hypermobile EDS patients.

For vEDS patients specifically, research is needed to further identify the possible coping mechanisms patients use, as they may correlate with those displayed in EDS patients also.

5.13 Conclusion

This study is the first to reveal the psychosocial issues faced by vEDS patients, addressing the original research question throughout this qualitative research analysis. The initial aims of this study have been met through the in-depth exploration of patient experiences, providing rich information to meet the research objective. Patients discussed their daily lives in great detail which revealed the relationship between the physical limitations of the condition, and the impact on psychosocial wellbeing. It is evident that the condition presents many diverse challenges, ranging from social engagement and relationships, to decision making and future plans.

This study emphasises the combination of factors that contribute to an individual's overall experience with vEDS. Understanding the role of factors, such as communication and the responsibility patients carry alongside their diagnosis, highlight the widespread impact of the condition amongst family members, friends, and partners.

Gaining insight into the influence of loss through various participant perspectives, enabled a greater understanding of internalised thoughts and troubles. This study consolidates the widespread psychosocial impact of vEDS identified in previous research, whilst also exploring new areas such as intimacy.

The participant's resolve to adapt to their diagnosis and actively use coping mechanisms encapsulated their overriding strength and perseverance. A greater understanding of; the impact of age at diagnosis, the experience of male patients and also partners will require further research. This study supports the introduction of professional educational programmes to boost awareness and understanding of vEDS within clinical practice. Clinicians should aim to review and improve the MDT to further address the psychosocial issues identified in this research, whilst considering upskilling genetic counsellors to also support patients.

Chapter six: Reflective discussion of the methods used

6.1 Introduction

This dissertation process proved to be an invaluable learning experience, involving a personal transformation increasing my confidence as I completed what initially seemed to be an impossible task. As I entered each stage of the project I was confronted with new challenges, all of which became valuable lessons that contributed to my growth as a researcher.

Figure 23 below utilises John's model of reflection, to provide a visual representation of my experience throughout this process, which is explored in detail during this chapter. I have detailed the numerous setbacks I encountered along with valuable advice for future students conducting their first research projects.

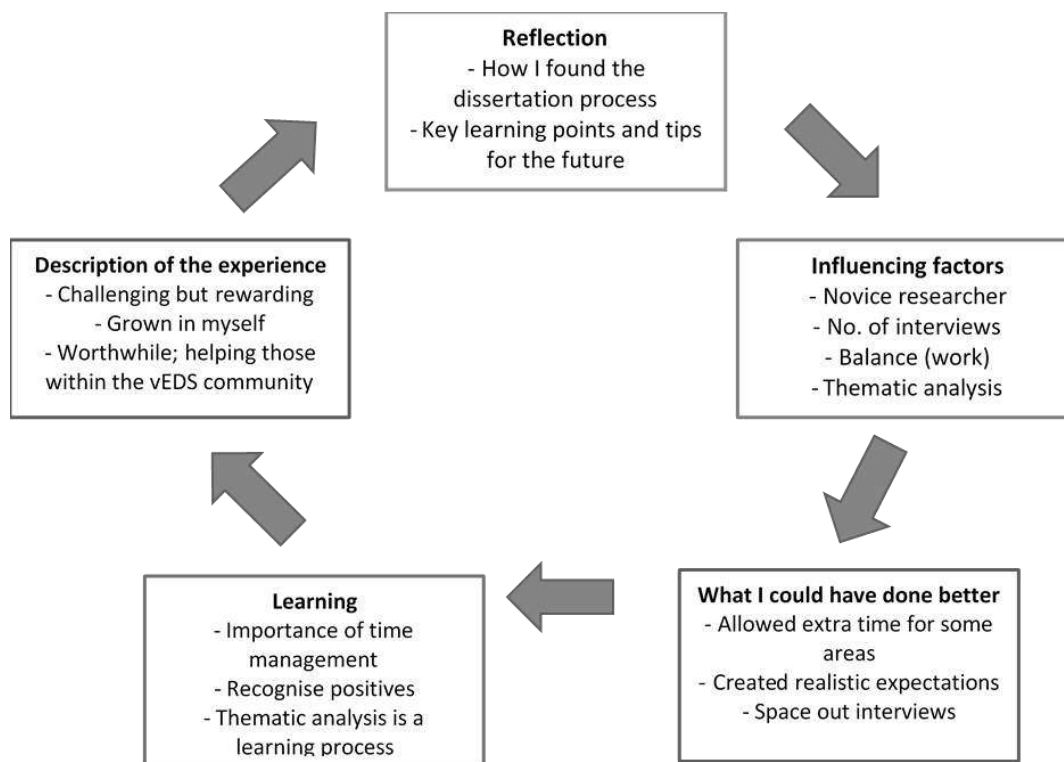


Figure 23 - John's model of reflection used to explore the experience and learning points.

6.2 The perfect match

On reflection, I am happy with my choice of research project. Upon meeting the project supervisor, I was struck by her passion for the topic and gained an understanding of the study's objectives and the support I would receive in reaching those aims.

Although I was anxious about conducting my first ever interviews, data collection turned out to be my favourite part of the dissertation process.

I built a good rapport with my study sample and strived to interview all participants that came forward (21) despite my initial aim being 15. Enjoying and embracing my dissertation topic was of great advantage in my methods and throughout the process.

I advise that future researchers ensure they choose a study that they are interested in and can grow to be passionate about, as this made a significant difference to my dissertation experience. After completing the project, I can honestly say that this research topic, was the perfect match for me. I was not only driven by my personal motivations, but I understood the importance of the project for my participants and wanted to voice their experiences.

6.3 Organisation and timing are key

“Do as much as you can as early as you can” the advice written by Hodgson and Rollick (1989) in their handbook to surviving research. I pride myself in being an organised individual and this has been of significant benefit throughout this process. Creating daily, weekly, and monthly plans, with realistic deadlines and arranging meetings with my supervisor in advance, helped me juggle my job, lectures, assignments and my dissertation. After hearing of the time-consuming nature of the ethical approval process, I was keen to begin my application as early as possible. I also recorded all the information regarding my participants in an excel spreadsheet which acted as a guide throughout the process (table 20).

Table 20 - Participant information spreadsheet used throughout the recruitment, data analysis and data collection process. Participants are identified by their unique reference number.

Study/ interview reference	Consent received	Date initial contact email sent	Initial telephone conversation date	Interview date	Debrief phone call date	Interview recorded	Interview transcribed	Interview analysed and coded
PN002	YES	Date	Date	Date	Date	Y	Y	Y

Submitting my ethics application early, allowed me to begin recruiting at the start of my third year of the MSc and complete data collection by December 2019, for which I am thankful as I witnessed the impact of the Covid-19 pandemic on my colleagues recruitment strategies.

Timing and organisation were therefore key to my methodological process and had I not started as early as I did; I would not have accomplished what I have today.

Interviewing, transcribing, coding, and analysing 21 interviews, as well as working to deadlines to submit draft chapters was a huge task. I strongly recommend that future researchers develop a reasonable plan with realistic deadlines to ease the pressure of time constraints. Lastly, I advise submitting the ethics application early to aid time management and aim to keep a consistent record of all participant correspondence, activity, and research data.

6.4 A time to debrief

Including a debrief phone call in my methods, provided myself and participants with the opportunity to reflect on the overall experience. Some participants explained that the interview process emphasised the fragility of the condition and brought light to uncomfortable areas such as mental health. The debrief phone call highlighted that the interviews surfaced some unwanted emotions which individuals found hard to contend with. Notwithstanding, some participants stated that the interview process had inspired them to take greater control over the management of their condition and felt motivated to set higher personal aims.

A handful of participants likened the interview to speaking with a friend, which helped them provide open and honest answers which was comforting to hear as a novice researcher and demonstrated that debrief phone calls offer value in qualitative research. This was a great advantage to my methodology, and I would advise that future qualitative researchers consider including a debrief phone call in their approach. As a novice researcher, listening to how individuals found the interviews and their overall reflection of the process restored my confidence in conducting research and successfully navigating sensitive conversations.

6.5 Establishing a balance

Naturally, completing an MSc dissertation project from start to finish would elicit increased stress, especially due to the heightened workload, my perfectionism and the responsibility associated with the task. I initially misjudged the intense amount of work associated with a relatively large data set. This resulted in elevated stress levels as my work capacity was being constantly stretched. I struggled with the irrational guilt when engaging in non-dissertation activities and whilst being further along in the process than my colleagues did provide some reassurance, it also left me feeling somewhat alone. Overtime I accepted that stress was an inevitable aspect of the process and adopted a positive understanding of stress as a driving factor.

Once the UK entered lockdown in response to Covid-19 my daily routine was severely disrupted, as I could no longer attend the library or follow my structured timetable. Initially, the uncertainty and fear surrounding the virus made it difficult to maintain the balance I had achieved.

Adapting to both working and completing my dissertation from home during such unprecedented times was challenging.

This experience has enabled me to further develop my stress management skills and recognise an unhealthy level of stress. I advise future researchers to recognise their own stress thresholds and practice self-care, as this will contribute to positive mental wellbeing. Maintaining a work/study/life balance is difficult but is a key skill that will benefit future challenges.

6.6 Learning as I went along

6.6.1 A novice researcher

This process presented a steep learning curve. Despite being warned that including 21 participants would result in a mountainous workload, I did not quite realise how many hurdles such a large amount of data would incur, many of which were time-related barriers such as transcription and adhering to prescribed word counts. However, I am proud that I included all patients that wanted to take part. In hindsight, conducting my interviews in quick succession prolonged the process of transcription and analysis of the data, but it also enabled me to maintain momentum and retain a uniform interview technique.

6.6.2 Thematic analysis

“You’ve got to be prepared to let go of your data”, a statement by Braun and Clarke cited in their successful guide for qualitative research (Braun and Clarke, 2013 pp. 234). Regardless of my initial efforts to utilise thematic analysis effectively, I was left feeling deflated as my inexperience with the technique resulted in a categorised analysis of the data set. Acknowledging the faults in my approach to thematic analysis highlighted my errors:

- 1) My initial categorical approach was surface level and required review and refinement, which is common when using the technique.
- 2) I had become attached to my data, creating difficulty in establishing which parts were not relevant to my study.

6.6.3 Interview technique

I made a conscious effort to uphold my position as a researcher when interviewing participants, but admittedly struggled to prevent myself from shifting between a researcher, and a training genetic counselling student equipped to provide psychosocial support. Despite restricting myself, my counselling skills occasionally surfaced in reaction to participants distress, along with empathetic responses. I have always considered myself to be a resilient individual however, some interviews

were very emotive and overwhelming. Initially, I felt awkward using silence as a technique during the interview process, but I soon understood that it allowed time for the participants to articulate their thoughts which resulted in in-depth answers.

I strongly advise that those embarking on qualitative, emotive research projects consider their experience and knowledge at each stage. It is worth noting that as a novice researcher you are continually learning along the way and support and guidance from your supervisor and colleagues is invaluable.

6.7 Lessons learned

Reflecting on the development of this project has highlighted the personal growth I have made as a result of the lessons I have learned. I would describe the entire process as a learning curve and figure 24 displays the key learning points I have taken from this experience. Figure 25 summarises my advice to future researchers.

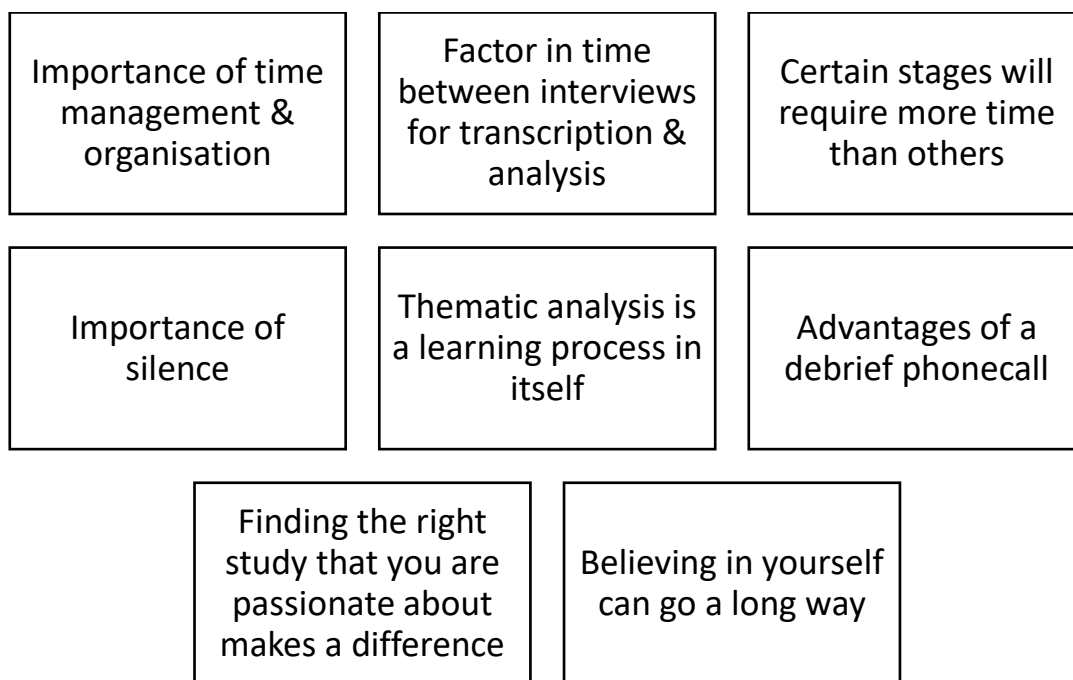


Figure 24 - Summary of the key learning points I have gained from conducting this research project.

6.7.1 Theoretical planning cannot account for obstacles along the way

I learned that planning is crucial to the process, but no matter how organised you are, some areas require more time and effort than others. My time estimation for transcription was unrealistic and I did not account for the additional time needed in data analysis, both of which affected my overall schedule. Lesson one therefore emphasises that this process is dynamic, and it is likely that timescales will change.

6.7.2 Thematic analysis is a process in itself

I had briefly touched on thematic analysis in lectures and tutorials during the MSc. It is no surprise therefore, that this stage of the dissertation evolved into an entire lesson in itself, as I attempted to understand how to utilise the technique efficiently, to draw out conceptual themes from my data. I advise that once future researchers have established their data analysis method, they familiarise themselves with the technique, especially with practical examples.

6.7.3 A dissertation project results in personal growth and development

I have learned that conducting research highlights the importance of development and personal growth, as acquiring knowledge and self-assurance at each stage was important. Building rapport with my participants, and noticing that they felt comfortable speaking to me, increased my confidence which organically contributed to the development of my interview technique.

6.7.4 Inexperience does not imply incapability

The greatest lesson I have learned is that experience does not determine capability. Initially, I believed that because I had not carried out qualitative research, or interviewed participants, I did not have the capacity to complete the project. Recognising my own strengths and limitations throughout the process, along with accessing practical and emotional support from my family, friends, and supervisor enabled me to learn, grow and progress along the way. Believing in yourself is valuable and completing this project has proved that I can do anything I put my mind to!



Figure 25 - Summary of the advice to future researchers conducting an MSc dissertation.

6.8 Final reflections

This study has successfully addressed the research question and exceeded my expectations, by providing rich informative results that showcase the vast implications of living with vEDS. I overcame several hurdles but managing the amount of data remained a constant challenge throughout the process.

I have been immersed in the realism of living with vEDS and gained insight into the challenges vEDS patients face. This invaluable experience has provided me with an abundance of transferable skills and knowledge which can be applied to all aspects of my life including my future career in genetic counselling. I am confident that this study has the potential to influence clinical practice and has motivated me to embrace future research prospects. I feel I have successfully used this opportunity to voice the opinions, thoughts, and experiences of those within the vEDS community.

The reassuring and attentive nature of my supervisor, along with the endless support from family and friends were vital to getting through this process. Although I am now confident in conducting qualitative research, I acknowledge that there are several areas for further development. Incorporating the lessons I have learned from this process, into future research opportunities will contribute to my growth as a researcher.

Embarking on this journey and completing an MSc dissertation is my greatest academic achievement. I am immensely proud that I continued to push myself further than ever before, despite several personal and academic challenges that arose. Continuing the data analysis and write up of my project during a global pandemic was an additional hurdle that had a profound impact on my journey towards the end of the process. As I write this, I am very proud of myself for completing this project to what I believe is an influential standard. This process has highlighted my key strengths including my confidence, resilience, and determination, all of which were vital foundations to this project. My lasting message to future researchers is to have faith in yourself and get going, as the first step is always the hardest to take.

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Appendices

Appendix A: Research protocol

Research protocol

Project title

What psychosocial issues are faced by patients with Vascular Ehlers Danlos Syndrome?

Purpose of the project and its academic rationale

The purpose of this project is to explore the psychosocial issues patients with Vascular Ehlers Danlos Syndrome (vEDS) are faced with and the overall patient experience.

This study will investigate and aim to gain a deeper level of understanding of:

- The psychosocial issues displayed in patients with vEDS e.g. 'how do patients cope day to day' and how patients feel about their diagnosis.
- The challenges faced by an affected individual's family
- The overall patient experience and identifying
- The obstacles faced by vEDS patients,

Achieving information surrounding these topics can benefit families, friends and healthcare professionals in accommodating the needs of patients with vEDS, ensuring that maximum support is available to affected individuals. Additional information regarding the use of this research topic and its academic rationale can be found below.

Background

Ehlers Danlos Syndrome or EDS, refers to a group of heterogeneous connective tissue disorders, which can result from a defect in collagen (Royce and Steinman. 2003). Initially, EDS was categorised into six main subtypes but, the evolution of genetics and the rise of next generation sequencing lead to the discovery of several mutations in novel genes associated with collagen and developments in the diagnostic criteria of EDS and so, there are 13 subtypes of the condition today (Beighton *et al.* 1998, Malfait *et al.* 2017). Vascular EDS (vEDS) is an autosomal dominant disorder that places patients at risk of spontaneous rupturing of their hollow organs and arteries, which evidently can result in premature death (Milewicz *et al.* 2014). The burden of living with EDS, that has little treatment and uncertainty of what each day will hold, would expectedly have several physical and psychological consequences to an individual's health and lifestyle, however, there is little research which addresses this.

Lumley *et al.* (1994) analysed a dominantly female sample, with a small number of male participants and seven children, using interviews and questionnaires to gain an insight into the psychological

Version: 1

Date: 04/05/2019

wellbeing of an EDS patient. All 41 participants had a clinical diagnosis of Classical EDS, which is associated with fragile, elasticated skin, atrophic scars and/or hematomas or cysts on the extremities of the body. The results from testing identified 3 main concerns that were expressed by patients:

1. Problems in day to day living associated with pain
2. Pregnancy and reproduction
3. Cautiousness to engage in certain activities due to a fear of injury

Significant advancements in medicine and the increasing knowledge available about EDS, makes it difficult to assume that these are the concerns present in EDS patients today, as the study was conducted over two decades ago. Also analysing a dominantly female population, limits the generalisability of the results, as we are unaware of what males with EDS experience, as it could be different.

The association between hypermobile and classical EDS, and psychiatric disorders has surfaced in recent research. Berglund *et al.* (2015) received 250 responses from members of the Swedish National EDS Association on The Short form Health Survey (SF-36) and The Hospital Anxiety and Depression Scale (HADS). Significant differences were displayed between men and women in the physical and mental components of the SF-36, reinforcing the importance of having results from both genders. Notably, EDS patients recorded a lower quality of life when compared to a control group and the highest rated anxiety measures included "sudden feelings of panic" and "feeling as if something bad will happen" (Berglund *et al.* 2015, pg. 3).

A similar study focused on the presence of psychiatric disorders in EDS patients in Canada. A retrospective chart review took place for hypermobile EDS patients, using medical and psychiatric data that had been reported by patients during genetic assessment. Of the 106 patients within the cohort, 42.5% had one psychiatric disorder, whilst 22.7% had 2 or more (Hershenfeld *et al.* 2016). Table 2 displays the wide range of psychiatric disorders identified in hypermobile EDS patients.

Table 1: Types and frequencies of psychiatric disorders adapted from results by Hershenfeld *et al.* (2016).

Psychiatric disorder	N (%)
Any psychiatric disorder	45 (42.5)
Depression	27 (25.5)
Anxiety disorders	25 (23.6)
ADHD	7 (6.6)
PTSD	5 (4.7)

Borderline personality disorder	4 (3.8)
Bipolar disorder	2 (1.9)
Anorexia nervosa	2 (1.9)
Learning difficulties	2 (1.9)
Conduct disorder	2 (1.9)
Asperger disorder	1 (0.9)
Schizoaffective disorder	1 (0.9)
Schizotypal disorder	1 (0.9)

However, retrospective chart reviews have shown to be vulnerable to methodological error and can lead to the underestimation of disease when used in research (Matt and Matthew, 2013).

Additionally, the use of self-reported methods, have been criticised due to the potential of “socially desirable and extreme responding” from patients (McDonald, 2008, pg. 94). Collectively, these limitations can widely affect the results produced. Regardless, Berglund and Hershenfeld have publicised the association between EDS and the development of psychiatric disorders.

Educational needs are an aspect to be considered in individuals with chronic conditions. Giroux *et al.* (2016) believe that appreciating educational factors as well as clinical considerations in all subtypes of EDS could “help students with their academic and psychosocial development” (pg. 408). They stressed that a vast majority of EDS research concentrates on clinical issues which aids medical professionals, but there is no attention paid to students, support workers and educators. Giroux have shown the importance of understanding not just the clinical barriers, but also the psychosocial barriers faced by patients with EDS.

The relevant research currently available in this area largely focuses on psychological wellbeing in all patients with EDS, regardless of the type. There is no attention paid to the psychological impact vEDS can have on patients, emphasising the need for research in this area. Conditions such as Loeys-Dietz syndrome (LDS) and Marfan syndrome display similar features to vEDS than other subtypes of EDS themselves. LDS is largely characterised by aortic aneurysms and cardiovascular complications like those displayed in vEDS patients. Marfan syndrome (MFS) is an inherited connective tissue disorder which has varying clinical presentation and like vEDS, patients are at constant risk of aortic rupture that can ultimately lead to sudden death.

This present study aims to explore the psychosocial issues faced by patients suffering from vEDS, due to the severity of this subtype. By doing so, a greater understanding of the vEDS patient experience

can be achieved, which can improve the support available to patients provided by healthcare professionals, families and friends, as well as set a base for additional research to take place.

Description of methods and measurements

Overall approach

An exploratory style will be used for this project. Qualitative methods will be adopted, to establish the importance of any identified psychosocial issues within vEDS patients. Qualitative methods are appropriate for research areas that are not well-understood and where exploring people's views and perceptions are important. This project will employ the use of semi-structured interviews to gain information from participants and this flexible research style will identify key subject areas expressed by patients.

Sample and recruitment

Participants will be recruited through the Annabelle's Challenge, a UK charity for vEDS that raises awareness campaigns and offers support and advice for people with and families of individuals living with the condition. Jared, the founder and CEO of the charity will use Annabelle's challenge social media platform, newsletter and website to publicise this research project along with mine and Jared's contact details, for potential participants to express their interest. Participants with a confirmed diagnosis of vEDS, over the age of 18, which can provide written informed consent will be suitable for the project. Jared will then circulate the recruitment documents (participant information sheet, invitation letter and consent form) to any participants that are interested in joining the study. vEDS is a rare form of EDS within the UK and is estimated to affect at least 1 in 50,000 Steinmann et al. 2002). Recruiting 10-15 participants will be sufficient and manageable to investigate and help me achieve data saturation. Once consented into the study, I will contact the participants for an initial telephone call to discuss the study in more detail and address any questions they may have. This initial phone call will also be used to arrange the date and time for their interview for questioning.

Data collection

Interviews will take place via telephone call or home visits, as the members and families that are part of Annabelle's challenge are located around the UK. If home visits are required for some participants, these will take place on a one-to-one basis, allowing the researcher to interpret non-verbal cues through "body language, facial expression and eye contact" providing a deep understanding of how patients are feeling (Ryan et al. 2009, pg. 310).

All phone calls and interviews will be recorded using a Dictaphone. These recordings will be uploaded onto the password protected computer and will then be deleted from the Dictaphone device immediately. Interview transcripts will be anonymised and pseudonyms will be used.

A checklist will be used to briefly guide the interview, which provides an opportunity for 'in-depth probing' whilst ensuring the interview stays within "the parameters traced out by the aim of the study" (Berg 2007, pg. 39). I will take brief notes and also summarise each interview in a short audio recording for my own reference.

Data treatment

My Cardiff University email account will be used to communicate with participants and store the data. Data will be saved in individual participant folders on a password protected computer. Each interview (both phone call and face-to-face) will be transcribed and numbered to eliminate any identifying characteristics. Each folder will contain the participant's interview recording, the summary recording I create after the session, the transcription of the interview and any other associated documents. Confidentiality will be addressed throughout the entire research study, but particular attention will be paid at 4 main points; during research planning, data collection, data analysis and the distribution of research results, similar to the dominant approach of confidentiality (Kaiser 2009).

Any reply slip or consent form that has been returned from potential participants indicating willingness to participate in the study will be sent to myself, for the care of Marion McAllister at Cardiff University. This will ensure that all reply slips/consent forms that contain participant's personal information (name, phone number, email address) are appropriately stored in a cabinet in a locked office at Cardiff University to protect participant confidentiality.

Data analysis

Audio recordings will be transcribed and thematic analysis will be used. This form of analysis is appropriate for my research study as it emphasises participant's perceptions, feelings and experiences as the paramount object of the study. Thematic analysis is beneficial in exploratory research and will allow me to identify the key themes present amongst the responses I receive from participants. Using thematic analysis will allow me to present a rich description of my data in relation to my research question and tie together the various strands and themes I identify during my analysis.

Version: 1

Date: 04/05/2019

Outputs

I am to present my research at the Annabelle's Challenge UK conference targeted at individuals with vEDS. Additionally, I am to publish the project in the EDS UK support group as well as in related peer-reviewed journals such as the Journal of Genetic counselling. The project will be written up as an MSc Dissertation submission to Cardiff University.

Participants, recruitment methods, number of participants, age, gender, exclusion and inclusion criteria

- Participants will be recruited through the Annabelle's Challenge, a UK charity for vEDS
- The study will be publicised by the founder and CEO of the charity via social media platforms, newsletters and their website.
- I will aim to recruit a minimum of 10-15 participants, I may be able to recruit more participants due to the flexible nature of the data collection process (telephone calls and home visits)
- Individuals taking part in the study must be 18 years old and over
- Gender: there is no gender specificity in this study
- Exclusion criteria: will include any adults that are considered to be in a vulnerable state e.g. require additional psychological support or daily care
- Inclusion criteria: Any individual with a confirmed medical diagnosis of vEDS, is over the age of 18 and has capacity to consent to this study.

Consent and participant information agreements, debriefing

- The consent form is on a separate document
- The participant information sheet is on a separate document
- The invitation letter is on a separate document
- The interview guide/checklist is on a separate document

After their interview, there will be a short debrief element, where participants will be encouraged to ask any questions and speak about how they found the interview process. One month after their interview, participants will be contacted for a final debrief of the whole project, here they will be notified about what happens next in terms of their data, how the research will be used and how to access any relevant support services that they may require.

Clear statement of ethical considerations raised by the project – how to deal with them

The investigation requires participants to speak of their life experiences whilst living with a potentially fatal condition and so patients may find the experience unsettling. The initial phone call with participants cover the potential distress they may experience, giving them an opportunity to ask me questions related to this element of the project. Any participants that require professional help during or after the study will be signposted to relevant services such as their GP, counselling services and support group networks such as Annabelle's challenge, the leading vEDS patient support group in the UK (Annabelle's challenge, 2018). Patients can also seek advice from the genetic counsellors at the EDS National Diagnostic Service, who I will signpost patients to if they need. Throughout the process of the study, patients will be reminded that they have the right to withdraw themselves and any data from the study, if they feel overwhelmed by the process.

Any distress experienced by the researcher will be overcome by discussions with course leaders, supervisors and fellow students if necessary. Any interviews taking place within participant's homes will abide by the Cardiff and Vale University Health Boards Lone Worker Policy (Cardiff and Vale University Health Board, 2017 <https://intranet.cardiff.ac.uk/intranet/staff/documents/health-safety-and-wellbeing/core/lonewkg.pdf>). This ensures the researcher is safe when interviewing participants in their home environment. Relevant contact information such as the both mine and my supervisors email address and my phone number will be provided for participants to use if needed.

Estimated start date and duration of project

Once we have received ethical approval from the Cardiff University School of Medicine board to go ahead with the project, the following timeline will be used as guidance. A table version of this information can be found in appendix six.

1. Sample recruitment will take place from 30/08/2019 until 01/10/2019.
2. Data collection (phone-call interviews/home visits) will take place from 10/10/2019 until 13/12/2019.
3. Data analysis will take place during the data collection phase and will finish by 30/01/2020.
4. My initial draft of my dissertation chapters will be written up and completed by 30/02/2020 and this will be reviewed by my academic supervisor, we will discuss the feedback together.
5. The first complete draft of the dissertation project will be completed by 29/04/2020, in which I will receive final feedback from my academic supervisor and we will discuss the final changes that need to be made.
6. Final amendments will be made before submission of the project on 20/05/2020.

Appendix one – Project timeline

	Ethics	Recruitment	Collect data	Analyse data	Initial dissertation chapters	Complete first draft	Submit final dissertation project
Start date	05/01/2019	30/08/2019	10/10/2019	15/10/2019	15/10/2019	20/03/2020	30/09/2020
Finish date	30/04/2019	01/10/2019	13/12/2019	30/01/2020	30/02/2020	29/04/2020	20/05/2020
Duration (days)	115	32	64	107	152	10	51

Appendix B: Cardiff University School of Medicine Research Ethics Committee, initial letter.



School of Medicine
Yr Ysgol Meddygaeth

Cardiff University
Main Building
Heath Park
Cardiff CF14 4XN
Wales, UK
Prifysgol Caerdydd
Prif Adeilad
Parc y Mynydd Bychan
Caerdydd CF14 4XN
Cymru, Y Deyrnas Unedig

Tuesday 28th May 2019

Leanne Barrett,
MSc Genetic & Genomic Counselling,
Postgraduate Taught Courses
School of Medicine
Cardiff University

Dear Leanne,

Re: What psychosocial issues are faced by patients with Vascular Ehlers Danlos Syndrome?

SMREC Reference Number: 19/50

This application was reviewed by the Committee on Tuesday 21st May 2019.

Ethical Opinion

On review the Committee have asked for the following issues to be addressed:

1. Provide clarification to the Committee and participants as to Marion McAllister's role in the study.
2. Ensure that you provide clarification to participants in the Participant Information Sheet that they will be audio recorded.
3. Provide clarification as to how you intend to notify participants of the outcome of the study.
4. Please amend the Participant Information Sheet to include the University's guidance on GDPR. Please see Appendix A of the attached for suggested wording to include.

Please send all documents addressing the points above to the Committee Secretary, Mrs Claire Evans, via email.

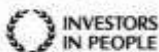
Documents Considered

Document Type:	Version:	Date Considered:
Application	V1 04/05/2019	21/05/2019
Research Protocol	V1 04/05/2019	21/05/2019
Letter to Participants	No Date or Version	21/05/2019
Participant Information Sheet	V1 04/05/2019	21/05/2019
Participant Consent Form	V1 05/04/2019	21/05/2019
Interview Guide	V1 04/05/2019	21/05/2019

Yours sincerely,

Dr Jonathan Hewitt
Chair, School of Medicine Research Ethics Committee

CC: Dr Claire Green



Registered Charity, no. 1136855
Eusen Gofhrestedig, rhif. 1136855

Appendix C: Cardiff University School of Medicine Research Ethics Committee letter of approval



School of Medicine
Yr Ysgol Meddygaeth

Cardiff University
Main Building
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Wales, UK
Prifysgol Caerdydd
Prif Adeilad
Parc y Mynydd Bychan
Caerdydd CF14 4XN
Cymru, Y Deyrnas Unedig

Monday 29th July 2019

Leanne Barrett,
MSc Genetic & Genomic Counselling,
Postgraduate Taught Courses
School of Medicine
Cardiff University

Dear Leanne,

Re: What psychosocial issues are faced by patients with Vascular Ehlers Danlos Syndrome?

SMREC Reference Number: 19/50

This application was reviewed by the Committee on Tuesday 21st May 2019. Further revised documents were reviewed on Thursday 25th July 2019.

Ethical Opinion

On review, I can confirm ethical approval for this study.

Conditions of Approval

The Committee must be notified of any proposed amendments to the methodology and protocols outlined in your submission. Also, any serious or unexpected adverse reactions that may arise during the course of the study must be reported to the Committee. As a condition of this approval, the Committee retains the right to audit and review the study for our own records.

Documents Considered

Document Type:	Version:	Date Considered:
Application	V1 04/05/2019	21/05/2019
Research Protocol	V1 04/05/2019	21/05/2019
Letter to Participants	No Date or Version	21/05/2019
Participant Information Sheet	V1 04/05/2019	21/05/2019
Participant Consent Form	V1 05/04/2019	21/05/2019
Interview Guide	V1 04/05/2019	21/05/2019
Research Protocol	V2 19/07/2019	25/07/2019
Participant Information Sheet	V2 19/07/2019	25/07/2019

With best wishes for the study of your study.

Yours sincerely,

Dr Jonathan Hewitt
Chair, School of Medicine Research Ethics Committee



CC: Dr Claire Green



Registered Charity, No. 1139955
Eiddo Gŵmestudig, nif 1139955

Appendix D: Recruitment advert on Annabelle's Challenge website

Freephone helpline 0800 917 8495

[REDS4VEDS DAY](#)



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Psychosocial Research Study

What psychosocial issues are faced by patients with Vascular Ehlers-Danlos Syndrome?



Lead researcher: Leanne Barrett, MSc Genetic and Genomic Counselling Student.

We are delighted to announce our first research study which will commence in September 2019 with recruitment taking place during August 2019. Our researcher is Leanne Barrett MSc, a Genetic and Genomic Counselling Student assigned to Annabelle's Challenge by Cardiff University working closely with Claire Green at the NHS EDS National Diagnostic Service in Sheffield.

Purpose of the study
This study aims to explore the psychosocial issues faced by patients suffering from Vascular Ehlers Danlos Syndrome (vEDS). Most of the research studies done in this area focus on other subtypes of EDS, with none paying specific attention to vEDS.

Psychosocial issues relate to factors that impact an individual's psychological and social functioning. By interviewing patients with vEDS, we hope to gain a deeper understanding of the vEDS patient experience, which may in turn, improve the support available to patients provided by healthcare professionals, families and friends, as well as set a base for additional research to take place.

Recruitment Criteria

You can take part in this study if you have a confirmed clinical diagnosis of vEDS, are over the age of 18 and have the capacity to consent to take part in this research based in the UK.

Project timeline

1. Consent process: August 2019
2. Initial phone call with Leanne: September 2019
3. Interview phone call or home visit with Leanne: Anytime from October 2019-December 2019, this interview will be audio recorded.
4. Debrief phone call with Leanne: One month after your interview (latest January 2020)
5. Results of the study expected from June 2020.

Confidentially

All of your personal data and information will be stored on a password protected computer. All information will be anonymised and pseudonyms (false names) will be used to identify the documents.

What do I have to do?

If you decide to take part in this study, send us a participation request (see below) and you will be sent a consent form which will be acknowledged and stored safely at Cardiff University. The lead researcher, Leanne Barrett, will then contact you for an initial phone call to introduce herself and address any questions you have about the research.

During this initial phone call, you will arrange with Leanne whether a telephone interview or home visit (face to face) interview suits you best. You will then arrange a time and date for your interview to take place.

You will only have one interview with Leanne, whether it be phone call or face to face and this will last from anywhere between 30 minutes to an hour. All interviews (both phone call and face to face) will be audio recorded. At the end of the phone call you will have the chance to express how you found the interview and ask any additional questions if needed. All audio recordings will be stored on a password protected computer.

You will also arrange a final debriefing telephone call with Leanne one month after your interview. This will provide you with the opportunity to go through any additional questions or concerns you have regarding your part in the research study. You can also express your experience of being in the study and explain what went well and what could have been improved. Leanne will inform you of the next steps of the study and how your interview will be used in her dissertation project. You will not be required to provide any additional information or time after this stage.

What will happen to the results of the study?

The results from this study will be published as a student dissertation project for Cardiff University. A short, summative article will be created to highlight the signature findings of the project. This summary document will be sent to you, via your preferred method of contact (either by post or via email) so that you are aware of the outcome of the study.

The results may also be published in related peer-reviewed scientific journals such as the Journal of Genetic Counselling. The results may also be shared on the EDS UK support group website and on Annabelle's Challenge website and social media platforms. Again, none of your personal information will be included in the publication.

Results of the study expected from June 2020.

Appendix E: Invitation letter



Dear participant,

You are being invited to take part in a research study looking at the psychosocial impact of living with Vascular Ehlers Danlos Syndrome (vEDS). Before you decide whether or not you want to take part, it is important for you to understand why the research is being done and what it will involve. Please take time to read the attached participant information sheet carefully and discuss it with others if you wish.

Please do not hesitate to contact me if there is anything that is not clear, or if you would like more information. Please take the time to decide whether you would like to take part in this project, thank you.

Sincerely,

*Leanne Barrett
MSc Genetic and Genomic Counselling Student
Cardiff University
BarrettLV@cardiff.ac.uk*

Version: 2

Date: 07/08/2019

Appendix F: Participant information sheet



Participant information sheet



Project title

What psychosocial issues are faced by patients with Vascular Ehlers Danlos Syndrome?

Lead researcher

Leanne Barrett, MSc Genetic and Genomic Counselling Student

Purpose of the study

This study aims to explore the psychosocial issues faced by patients suffering from Vascular Ehlers Danlos Syndrome (vEDS). Most of the research studies done in this area focus on other subtypes of EDS, with none paying specific attention to vEDS. Psychosocial issues relate to factors that impact an individual's psychological and social functioning. By interviewing patients with vEDS, we hope to gain a deeper understanding of the vEDS patient experience, which may in turn, improve the support available to patients provided by healthcare professionals, families and friends, as well as set a base for additional research to take place.

Why have you been chosen?

You have been chosen to take part in this study because you have a confirmed clinical diagnosis of vEDS, are over the age of 18 and have the capacity to consent to take part in this research.

Do I have to take part?

It is entirely up to you to decide whether or not you want to take part in this project. If you do decide to take part you will be given this information sheet to keep and be asked to sign a consent form. If you decide to take part, you are still free to withdraw at any time, without giving a reason.

What will happen to me if I take part?

Your involvement in the project will include:

1. Consent process: August 2019
2. Initial phone call with Leanne: September 2019
3. Interview phone call or home visit with Leanne: Anytime from October 2019-December 2019, this interview will be audio recorded.
4. Debrief phone call with Leanne: One month after your interview (latest January 2020)

Confidentiality

All of your personal data and information will be stored on a password protected computer. All information will be anonymised and pseudonyms (false names) will be used to identify the documents.

Any reply slip or consent form that you return will be sent to Leanne, for the care of Marion McAllister a senior lecturer within the School of Medicine at Cardiff University. Marion's role within this research project is solely based on collating and storing confidential information. This will ensure that all documents containing your confidential information (name, phone number, email address) are appropriately stored in a cabinet in a locked office at Cardiff University and comply with the data protection policy mentioned below.

What do I have to do?

If you decide to take part in this study, you will sign the consent form which will be acknowledged and stored safely at Cardiff University. The lead researcher, Leanne Barrett, will then contact you for an initial phone call to introduce herself and address any questions you have about the research. During this initial phone call, you will arrange with Leanne whether a telephone interview or home visit (face to face) interview suits you best. You will then arrange a time and date for your interview to take place.

You will only have one interview with Leanne, whether it be phone call or face to face and this will last from anywhere between 30 minutes to an hour. All interviews (both phone call and face to face) will be audio recorded. At the end of the phone call you will have the chance to express how you found the interview and ask any additional questions if needed. All audio recordings will be stored on a password protected computer.

You will also arrange a final debriefing telephone call with Leanne one month after your interview. This will provide you with the opportunity to go through any additional questions or concerns you have regarding your part in the research study. You can also express your experience of being in the study and explain what went well and what could have been improved. Leanne will inform you of the next steps of the study and how your interview will be used in her dissertation project. You will not be required to provide any additional information or time after this stage.

Are there any risks?

If you decide to take part in this project there are no physical risks to you. There is the possibility that you may find talking about your life experience living with vEDS, emotionally challenging and unsettling. If you do feel distressed at any point within the study you can contact Leanne who will signpost you to additional support services such as your GP, genetic counsellors at the National EDS diagnostic service and other relevant organisations.

The final phone call with Leanne will allow you to express any concerns you may have after the interview as well. Again, she can signpost you to the relevant services if needed.

What will happen to the results of the study?

The results from this study will be published as a student dissertation project for Cardiff University. A short, summative article will be created to highlight the signature findings of the project. This summary document will be sent to you, via your preferred method of contact (either by post or via email) so that you are aware of the outcome of the study. The results may also be published in

related peer-reviewed scientific journals such as the Journal of Genetic Counselling. The results may also be shared on the EDS UK support group website and on Annabelle's challenge website and social media platforms. Again, none of your personal information will be included in the publication.

Organisation and funding

This research project has been organised by Cardiff University for their post graduate student dissertation projects, on behalf of Claire Green. There is no funding for this project. Lead researcher Leanne Barrett will be supervised by Claire Green, a genetic counsellor in Sheffield who has created the basis for this project to take place.

Data protection

Cardiff University is the Data Controller of this research project and is committed to respecting and protecting your personal data in accordance with your expectations and Data Protection legislation. The University has a Data Protection Officer who can be contacted at inforequest@cardiff.ac.uk. Further information about Data Protection, including your rights and details about how to contact the Information Commissioner's Office should you wish to complain, can be found at the following: <https://www.cardiff.ac.uk/publicinformation/policies-and-procedures/data-protection>

Your data will be stored according to the university's data retention scheme <https://www.cardiff.ac.uk/public-information/policies-and-procedures/record-management-policy-and-retention-schedules>

Contact for further information

I hope you have found this information sheet useful. I understand that there is a lot of information to think about. So if you have any questions, please do not hesitate to contact me via my email address: BarrettLV@cardiff.ac.uk

Thank you,

Leanne Barrett
MSc Genetic and Genomic Counselling Student

Appendix G: Consent form

Participant consent form



Project title: What psychosocial issues are faced by patients with Vascular Ehlers Danlos Syndrome?

Name of Researcher: Leanne Barrett, MSc Genetic and Genomic Counselling student

Please initial each box:

1. I confirm that I have read and understand the information sheet dated 19/07/2019 (version 2) for the above study and have had the opportunity to ask questions.
2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason.
3. I understand that the telephone or face to face interview that I take part in, will be recorded and any identifying information will be anonymised.
4. I understand that should I need it, additional psychological support is available and I can ask about this at any time during the study.
5. I understand that the results from this study will be published as a student dissertation for Cardiff University.
6. I understand that any information I provide will be anonymised.
7. I understand that this research may be published in scientific journals.
8. I agree to take part in the above study.

Name of Participant Date Signature

Name of Person taking consent Date Signature
(if different from researcher)

Researcher Date Signature

Version: 2

Date: 07/08/2019

Appendix H: Interview guide

Interview guide

1. Introduction to the study

- Explain how the interview will take place
- Remind the participant that they are free to pause or stop the interview at any point
- Provide the participant with a brief overview of what the interview will involve

2. Background questions

- Could you tell me about how you came to be diagnosed with the condition?
- How does the condition affect you?
- What healthcare professionals did you come into contact with during the process of you becoming diagnosed?

3. General experience

- Day to day experience living with vEDS – any immediate comments?
- Occupation – does the participant have a job? If so how does vEDS affect this?
- What are the challenges participants face on a day to day basis?
- How do participants feel compared to others they come in contact with on a daily basis?
- How often do you think about your diagnosis?

4. Psychological impact

- How do patients feel knowing that they have a potentially fatal condition?
- How do participants feel about their future considering the nature of the condition?
- Has the participant experienced any mental health issues e.g. depression, anxiety?
- How has vEDS affected the participant's independence?

5. Social impact

- How has the condition affected the participant's social life?
- Do they feel limited in certain areas of their lives due to the condition?
- Has the condition affected their desire to socialise?
- Do participants feel that having a support group is useful in living with their condition?

6. Relationships with family and friends

- How do they feel vEDS has affected their relationships with others?
- Do participants talk about the condition with their family and friends?
- If the participant has children, how do they discuss the diagnosis with them?
- Has the condition affected participant's perceptions of having a family?