

**Women's experiences of living with a rare disease,
lymphangiomyomatosis (LAM):
A life history study**

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A thesis submitted in fulfilment of the requirements for the degree of
Doctor of Philosophy

Sydney Nursing School
The University of Sydney

2016

ABSTRACT

This study explored the experience of women living with lymphangiomyomatosis (LAM) over their life course. LAM is a rare, chronic, potentially life limiting, multisystem condition. It affects almost exclusively women and is characterised by progressive cystic lung disease. This study was undertaken to understand the meaning of women's experiences of living with LAM over time and how these experiences were affected by the rarity of LAM.

Life history methodology was used to explore illness experience across their life course. Gadamer's hermeneutic philosophy and Rosenthal's (1993) biographic interpretive method of narrative analysis provided the theoretical framework for the study. Semi-structured life history interviews were conducted with 19 women living with LAM. Factual data was extracted from the medical record of each participant. Data analysis involved constructing a biographical account of the objective facts of each woman's illness and analysing each life story for turning points and themes. Following cross case analysis, a collective life history which focused on the participants' illness experiences, was constructed.

An understanding of the complexity of their experiences from the onset of symptoms to the late stage of their illness was uncovered. The participants showed that they lived with LAM in a context of constant change, from the day to day fluctuations of everyday life to turning points of significant change related to their illness or social world. Diagnosis, a long hospital admission, commencing oxygen therapy, experiencing respiratory failure, and receiving a transplant were turning points when they experienced significant life disruption. The rarity of LAM created feelings of isolation and uncertainty at diagnosis and a need for self-reliance and self-advocacy to access appropriate information, care and treatment.

This study illuminates the process of how the participants developed resilience as they adapted to their illness in a period of transition. Resilience was a dynamic learning process and outcome by which they gained knowledge and competence in illness self-management, and found meaning through their experiences of autonomy, agency, social connectedness, spirituality and personal growth. It accumulated throughout life through everyday experiences as well as adversity. Resilience was associated with positive self-beliefs and enabled them to experience wellness and constructively manage illness-related and social changes over the course of their illness.

DEDICATION

This thesis is dedicated with appreciation to the nineteen women living with
LAM who participated in this study

ACKNOWLEDGEMENTS

I wish to acknowledge and sincerely thank the women who participated in this study and so generously shared their time and life stories with me. It was an honour for me to meet each one. The insights they have provided have given me a deeper understanding of the experience of living with LAM. I hope this thesis will inform healthcare professionals and help improve support for women living with LAM in the future.

Thank you to my supervisors, Associate Professor Murray Fisher and Dr Jennifer Green, for their wise guidance, patience, support, and encouragement throughout this PhD journey. I particularly thank them for taking the time to read and provide valuable feedback on numerous drafts of my chapters during the final stages of my candidature. Thank you especially to Murray for having inspired my interest in life history during my Bachelor of Nursing studies and, then on, for having been my mentor through the extended period of supervising both my Honours and PhD theses.

I acknowledge and thank Professor Allan Glanville, Director of Thoracic Medicine, St Vincent's Hospital, Sydney, my hospital supervisor, for his support of this research project and his assistance in the early stages of recruitment of the participants.

I acknowledge and sincerely thank Dr Jennifer Blundell, Honorary Associate Senior Lecturer at the University of Sydney, for final editing and formatting of this thesis.

My very special thanks go to my mother Nola Clare and dear friend, Sharon Hodgins, for their unwavering support, encouragement, and belief in me that I could see this thesis through to completion. They have been a much valued source of love and connectedness during the lonely times of writing. In particular, I thank Nola for her sustaining daily messages and inspiring poetry, and for reading and providing helpful editing suggestions for many of my chapters. Thank you to friends, Frances Nethery, Andrew Poidevin, and John and Olga Aitchison for their support and friendship.

My love and deep thanks go to my husband Bernie and children, Tim, Dominic, Andrew, and Laura, for their love, support and patience over the long course of my thesis. They witnessed the highs and lows of the journey and I am very grateful to them for 'sticking by me' throughout.

DECLARATION

I certify that the work presented in this thesis has not previously been submitted for a degree or as part of the requirements for a degree except as fully acknowledged within the text.

I certify that this thesis has been written by me and any assistance that I have received while undertaking this research and in the preparation of the thesis itself is acknowledged.

I certify that, to the best of my knowledge and belief, this thesis does not contain any material previously published or written by another person except where due reference is made in the text.



Denise Haylen

December 2015

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Chapter 1

INTRODUCTION

1.1 Introduction

This life history study explores women's experiences of living with lymphangiomyomatosis (LAM) over their life course. It illuminates the meaning of their experiences through the progressive stages of LAM as they lived their everyday lives within their individual social contexts, and interacted with health professionals in healthcare settings. This research highlights the complex issues related to women's experiences of living with LAM, including how their experiences were affected by the rarity of LAM.

This chapter is an introduction to the thesis. It begins with a background to the study, including an account of how this research journey began. The literature review which was conducted to locate and analyse the available literature related to the lived experience of women with LAM to determine the need for the study is presented. The literature review is followed by a statement of the aims of the study, the research question to be answered, and significance of the research. The chapter concludes with an overview of the structure of the thesis.

1.2 Background

LAM is a rare multisystem disease affecting almost exclusively women and characterised by progressive cystic lung disease (Ryu et al. 2006). LAM lung disease occurs sporadically, or in 26-38% patients with tuberous sclerosis complex (TSC), an inherited genetic disease characterised by mental retardation, autism, and lesions in various organs, and which can also affect men (Johnson et al. 2010; Taveira-DaSilva & Moss 2015). Sporadic LAM is not inherited and predominantly affects women (Taveira-DaSilva & Moss 2015). There has been only one case documented of a man without TSC diagnosed with LAM (Schiavina et al. 2007). While targeted therapies have recently been developed to treat symptoms of LAM, there is no cure. Women with advanced disease may be prescribed supplementary oxygen therapy and those with severe disease may be recommended for lung transplantation (Johnson et al. 2010).

The first case of LAM in a TSC patient was documented in 1918, and in a patient without TSC by Van Stossel in 1937 (Glassberg 2004; LAM Foundation 2015). The medical literature on LAM was limited to a few reviews and case reports until the establishment of the LAM Foundation in the US in 1995 by Sue Byrnes whose daughter had LAM, and the US National Registry for LAM in 1997 which provided a database for research. Since 1997 the LAM Foundation has raised over \$15 million dollars to fund research which led to the discovery of a genetic link between LAM and TSC (LAM Foundation 2015; Tattersfield & Glassberg 2006).

1.2.1 Epidemiology

LAM was previously considered to be a fatal condition that affected women of childbearing years. It is now regarded as a chronic disease of both pre- and post-menopausal women (Taveira-DaSilva & Moss 2015), with a median transplant-free survival time of 29 years from the onset of symptoms (Oprescu et al. 2013). The prevalence of sporadic LAM worldwide has been estimated at 3.3-7.7 per million women, and in Australia 5.17 per million women (Harknett et al. 2011). While the number of women diagnosed with LAM who registered with the US LAM Foundation and UK LAM Action databases increased over the years 1995-2005, Cohen et al. (2005) could not state that the incidence of LAM was increasing. Rather, they attribute these increases to improved recognition of milder LAM symptoms and subsequent diagnosis in women over 40 years. Harknett et al. (2011) estimated there are between 15,000 and 23,000 patients with LAM worldwide, while in Australia 104 women are known to be living with LAM (LAM Australia Research Alliance 2015). According to the LAM Foundation (2015) there may be 250,000 women with LAM worldwide who are undiagnosed or have been misdiagnosed with asthma, emphysema or bronchitis. LAM is not specific to any race or ethnic group (Taveira-DaSilva et al. 2010).

1.2.2 LAM as a disease

1.2.2.1 Pathology

LAM is characterised by a proliferation of abnormal smooth muscle cells called LAM cells which surround, infiltrate and obstruct airways, blood and lymph vessels (Glassberg 2004). Thin-walled cysts in the lung, ranging in size from a few millimetres to a few centimetres in diameter, are formed by the degradation of the collagen and elastin fibres of the lung and obstruction of the terminal airways by LAM cells and nodules (Taveira-DaSilva et al. 2006). The cysts can rupture into the pleural space causing spontaneous lung collapse

(pneumothorax) (Almoosa et al. 2006). Changes in the lung can cause abnormalities in lung function, including airflow obstruction, reduced lung diffusing capacity, and gas exchange abnormalities (Taveira-DaSilva & Moss 2015). Receptors for oestrogen, progesterone and growth factors have been identified on LAM cells (Taveira-DaSilva et al. 2006).

Blockages of the lymph channels cause the formation of chyle-filled lymphatic masses called lymphangioliomyomas most frequently in the peritoneum, pelvis and mediastinum (Taveira-DaSilva & Moss 2015). LAM cells also cause thickening of the thoracic duct, dilation of lymph channels, and the formation of multiple narrow channels causing chylous pleural effusions and chyloptysis (Taveira-DaSilva et al. 2006). Angiomyolipomas, tumours in the kidney, are composed of LAM cells, fat cells, and poorly differentiated vascular structures (Taveira-DaSilva et al. 2006). Blockages in the blood vessels in the lung cause haemoptysis (Johnson 1999).

1.2.2.2 Pathogenesis

Research conducted over the last 15 years has found that LAM is caused by mutations in the tuberous sclerosis tumour suppressor genes, TSC1 and TSC2 (LAM Foundation 2015; Taveira-DaSilva et al. 2006). These mutations cause the proliferation of LAM cells by dysregulating the mTOR (mammalian target of rapamycin) signalling pathway, a protein complex which regulates cell growth, metabolism, proliferation and survival (LaPlante & Sabatini 2009; Taveira-DaSilva & Moss 2015). LAM cells have metastatic properties, shown by the detection of LAM cells in the donor lungs of patients who had received lung transplants (McCormack 2008), and, while there is not general agreement, it has been suggested that LAM be reclassified as a “low-grade, destructive, metastasizing neoplasm” (McCormack et al. 2012, p.1210).

1.2.2.3 Clinical Features of LAM

There is a high level of clinical variability with LAM (Cohen et al. 2005). Patients frequently present with recurrent pneumothoraces (57%) or progressive breathlessness (73%), and may also experience a chylous effusion (12%), abdominal masses (29%), bleeding from a ruptured angiomyolipoma in the kidney or abdomen (32%), or haemoptysis (32%) as their first sign of LAM (Ryu et al. 2006; Taveira-DaSilva & Moss 2015). Cough (6%), chest pain (5%), chyloptysis, and wheeze are much less common symptoms (LAM Foundation 2015). The average number of recurrent pneumothoraces for an individual woman is 4.4 (McCormack 2008). Angiomyolipomas occur more frequently in patients with TSC-LAM (93%) than those with sporadic LAM (32%) while lymphangioliomyomas and chylous

effusions are more common in sporadic LAM (Ryu et al. 2006; Taveira-DaSilva & Moss 2015). There is also a high level of variability in the rate of progression of LAM. For some women, LAM progresses slowly over many years while for others, usually younger women, it can advance rapidly (Taveira-DaSilva & Moss 2014). Johnson et al. (2004), in an analysis of 57 patients in the UK LAM database, reported that 23% of those patients required home oxygen after ten years.

1.2.2.4 Diagnosis

There is generally a delay from onset of symptoms to diagnosis due to the more general nature of early clinical findings and many physicians' lack of knowledge of LAM which can lead to misdiagnosis of asthma, emphysema or chronic bronchitis (Glassberg 2004). Johnson (1999) reported the mean interval between onset of symptoms and diagnosis was 4 years but ranged from 0 to 25 years. LAM is usually diagnosed by high resolution CT scanning (HRCT) and/or lung biopsy (Johnson et al. 2010). Recent research has discovered that VEGF-D (a lymphatic vascular growth factor), at a serum level of >800pg/ml, is a valuable biomarker in diagnosing LAM (Young et al. 2013).

1.2.2.5 Medical management of LAM

Until recently there was no effective therapy for LAM and lung transplantation was the only treatment option (Taveira-DaSilva & Moss 2014). The discovery of the role of the mTOR pathway in the pathogenesis of LAM has led to the development of therapies which target this pathway to treat LAM. Two mTOR inhibitors, sirolimus and everolimus, have been found to be “effective in stabilising lung function, and reducing the size of chylous effusions, lymphangiomyomas, and angiomyolipomas” (Taveira-DaSilva & Moss 2014, p.116). Like cancer, it is believed that LAM may be best treated with therapies targeted at pathways involved in the pathogenesis of the disease, and research is continuing into other treatments (Taveira-DaSilva & Moss 2014).

A pneumothorax is treated initially by chest tube drainage but, as the risk of recurrence is greater than 70%, a woman may undergo a chemical or surgical pleurodesis (Taveira-DaSilva & Moss 2014). Chylothorax is treated by chest tube drainage and possibly a fat free diet (Johnson et al. 2010). Renal angiomyolipomas are monitored and those more than 4cm in diameter, which are at greater risk of bleeding, may be embolised (Johnson et al. 2010). Lung transplantation is considered when a woman's lung function is less than 30% of the

predicted value for her age, sex, and size, and she is unable to carry out activities of daily living (Taveira-DaSilva & Moss 2014).

Pregnancy has been associated with an increased risk of pneumothorax and chylothorax (Johnson et al. 2010). The European Respiratory Society, in its Guidelines for the Diagnosis and Management of LAM, while stating that becoming pregnant is the patient's decision and advice should be given on an individual basis, recommends that women with moderate to severe disease or rapidly declining lung function pregnancy avoid pregnancy (Johnson et al. 2010). Women with mild disease are warned of the potential risks of pregnancy (Taveira-DaSilva & Moss 2014). Women with well-preserved lung function may travel by air, those with advanced disease need to be evaluated for the need for oxygen during the flight, and women with a recent or known untreated pneumothorax are advised not to fly at all (Johnson et al. 2010).

1.2.3 LAM as a rare disease

The US definition of a rare disease is one that affects less than 200,000 individuals. This corresponds to 2000 in Australia and a prevalence of 1-8 in 10,000. The European Community definition is less than 5 in 10,000 (Aronson 2006). LAM is readily classified as a rare disease by these criteria. There are more than 6,000 known rare diseases (Rare Disease UK 2016). While the prevalence of each rare disease is low, rare diseases in total affect a significant number of people. It has been estimated that there are 29 million people living with rare disease across the European member states (EURORDIS 2009). In Australia it is estimated that there are 1.2 million people living with a rare disease (Department of Health Western Australia 2013), representing six to eight per cent of the Australian population (Rare Voices Australia 2013).

A rare disease such as LAM may also be referred to as an orphan disease or "neglected disease" (Aronson 2006, p.127). Because of their rarity orphan diseases have difficulty attracting funding for research as it is not profitable for drug companies to research and manufacture drugs that would benefit people suffering from these diseases (Gericke et al. 2005). Gericke et al. (2005) argued that rare diseases merit scientific study not only for moral reasons but because research into rare diseases often provides valuable insights into more common conditions as many rare diseases result from a single genetic alteration. Research into LAM has already benefited other rare diseases such as Cowden disease, Peutz-Jeghers syndrome and Proteus syndrome and it is hoped will give insights into other proliferative disorders such a cancer and asthma (Juvet et al. 2007; McCormack 2008).

Since 2003 LAM has come under the umbrella of the Rare Lung Diseases Consortium, part of the US Rare Diseases Clinical Research Network. This consortium collects clinical data from around the world into a centralised database and conducts clinical trials testing treatments for rare lung diseases (Rare Lung Diseases Consortium 2015). This allows areas with much lower numbers of individuals with LAM to contribute data for research and benefit from such research.

In Europe and North America patient-driven alliances have formed to represent and advocate for people living with rare diseases. The National Organisation for Rare Disorders (NORD), formed in the US in 1983, has more than 230 patient organisation members and runs education, advocacy and research programs (NORD 2015). The European Organisation for Rare Diseases (EURORDIS) was founded in 1997 and represents 30 million people, 692 rare disease patient organisations in 63 countries, and more than 4000 rare diseases (EURORDIS 2009). Rare Disease UK was established in 2008 to represent over 160 organisations of people with rare conditions (Rare Disease UK 2016). Rare Voices Australia formed in 2012 to advocate and promote health policy to benefit people with rare diseases (Rare Voices Australia 2013). Australia has yet to put in place a National Rare Disease Plan even though a 2013 scoping paper recognised the need for one (Department of Health Western Australia 2013). Patients with LAM are represented by the US LAM Foundation and LAM patient organisations in individual countries worldwide. The LAM Australia Research Alliance (LARA) has represented women living with LAM in Australia since 2006 and is a member of the Worldwide LAM Patient Coalition, formed by the LAM Foundation in 2007 to facilitate clinical research into LAM (LARA 2015; LAM Foundation 2015).

1.2.4 Beginning the research journey

My personal interest in LAM began nine years ago when I viewed a television documentary about two women who were living with LAM. I became aware that the hospital where I was working as a cardiothoracic nurse was a centre of LAM medical expertise where a number of women with LAM were treated. At that time, I had never heard of this rare disease although I recalled in the past a young female patient on the ward who had written the very long name of her illness on a piece of paper so that staff would know how to spell and pronounce it. On reflection, it is possible the word was ‘lymphangiomyomatosis’.

Subsequently I informally met a woman with LAM in an outpatient clinic. This had a deep impact on me as a brief meeting stretched into more than two hours during which time the woman told me her story with great feeling and, at the end of our meeting, handed me a

typed version she had been carrying with her. Listening to her story and being entrusted with it convinced me that it was important to hear what these women had to say about their lives with LAM. I wanted to understand more about how women experienced living with this rare disease. I embarked on an Honours thesis as the first stage of pursuing my nursing inquiry. The study involved conducting two life history interviews with one woman with LAM as a single case study. This doctoral thesis now forms the second stage of the inquiry.

1.3 The Literature Review

This literature review aimed to locate and review the literature on the lived experience of women with LAM in order to identify gaps in the research literature. The systematic search process is illustrated in Figure 1.1 and the results of the individual database searches are shown in Table 1.1. A paucity of literature was found on women's experiences of living with LAM. As LAM is both a progressive disease with significant effects on lung function and a rare disease, the literature search was expanded to include literature on the experience of living with other rare diseases, including rare lung diseases. The LAM literature was examined in relation to this wider body of rare disease research. The literature reviewed which related to the experiences of women living with LAM is summarised in Table 1.2, located in this chapter. Two additional tables located in Appendix 2 summarise the literature related to the experiences/perspectives of people living with a rare lung disease – pulmonary hypertension and idiopathic pulmonary fibrosis, and the literature related to the experiences/perspectives of people living with a rare disease. The literature search and review is discussed below, firstly in relation to the lived experience of LAM and, secondly, in relation to the experience of living with a rare disease.

1.3.1 Living with LAM

The literature search for the available literature on the experience of living with LAM was conducted using CINAHL, MEDLINE, PsycINFO, Sociological Abstracts, Expanded Academic ASAP, and ProQuest Dissertation and Theses databases (see Table 1.1). A search of each database was conducted using the keyword 'lymphangiomyomatosis'. The MEDLINE search (with limits "English" and "humans") revealed 1033 citations related to LAM; CINAHL, 113; Expanded Academic ASAP, 168; PsycINFO, 6; and ProQuest Dissertation and Theses, 164 theses. Sociological abstracts yielded no literature on LAM. The majority of citations located were biomedical and documented the epidemiology, physiology, pathology, pathogenesis, radiological investigation, diagnosis and management

of the disease, and included scientific research, clinical trials, case reports, observational studies, and cross-sectional surveys.

Additional keywords of qualitative, interview*, narrative*, experience*, quality of life, perspective*, subjective, survey*/surveys and questionnaires, and focus group* were applied individually to the MEDLINE search as separate searches in order to capture literature related to the lived experience of women with LAM. These searches yielded a total of 129 citations that were screened. The limited number of citations retrieved from CINAHL, Expanded Academic ASAP, PsycINFO, and ProQuest Dissertation and Theses databases were screened for relevance without applying additional keywords. Figure 1.1 is a PRISMA diagram which illustrates the systematic processes of the literature search (Moher et al. 2009). A total of 580 citations were identified from the databases and screened. After screening, 560 citations were excluded, including reports of scientific and medical research, clinical trials, case reports, an article which was not in English, and a book chapter on qualitative methodology. Twenty citations remained after exclusion. When duplicates were removed from these, 12 full-text articles and two doctoral theses were assessed for eligibility for inclusion in the literature review. Two articles (a book review and an evaluation of clinical data) were excluded. An additional citation was identified in the reference list of one article (Carel et al. 2010). This was Carel's (2008) longer personal narrative and philosophical phenomenological reflection on her LAM illness experience published as a book entitled *Illness*. A second edition of this book was published in 2013 (Carel 2103b) and this more recent edition was included in the review. A total, therefore, of 13 pieces of literature (nine full-text articles, one book, and two doctoral theses) were related to women's experiences and perspectives on living with LAM and included in the literature review. Table 1.1 displays the database search for literature. It contains six duplicates as it shows the search and retrieval of relevant literature from each database. Five references were present in more than one database. Table 2.1, Appendix 2, summarises the literature included in the review following the removal of the duplicates.

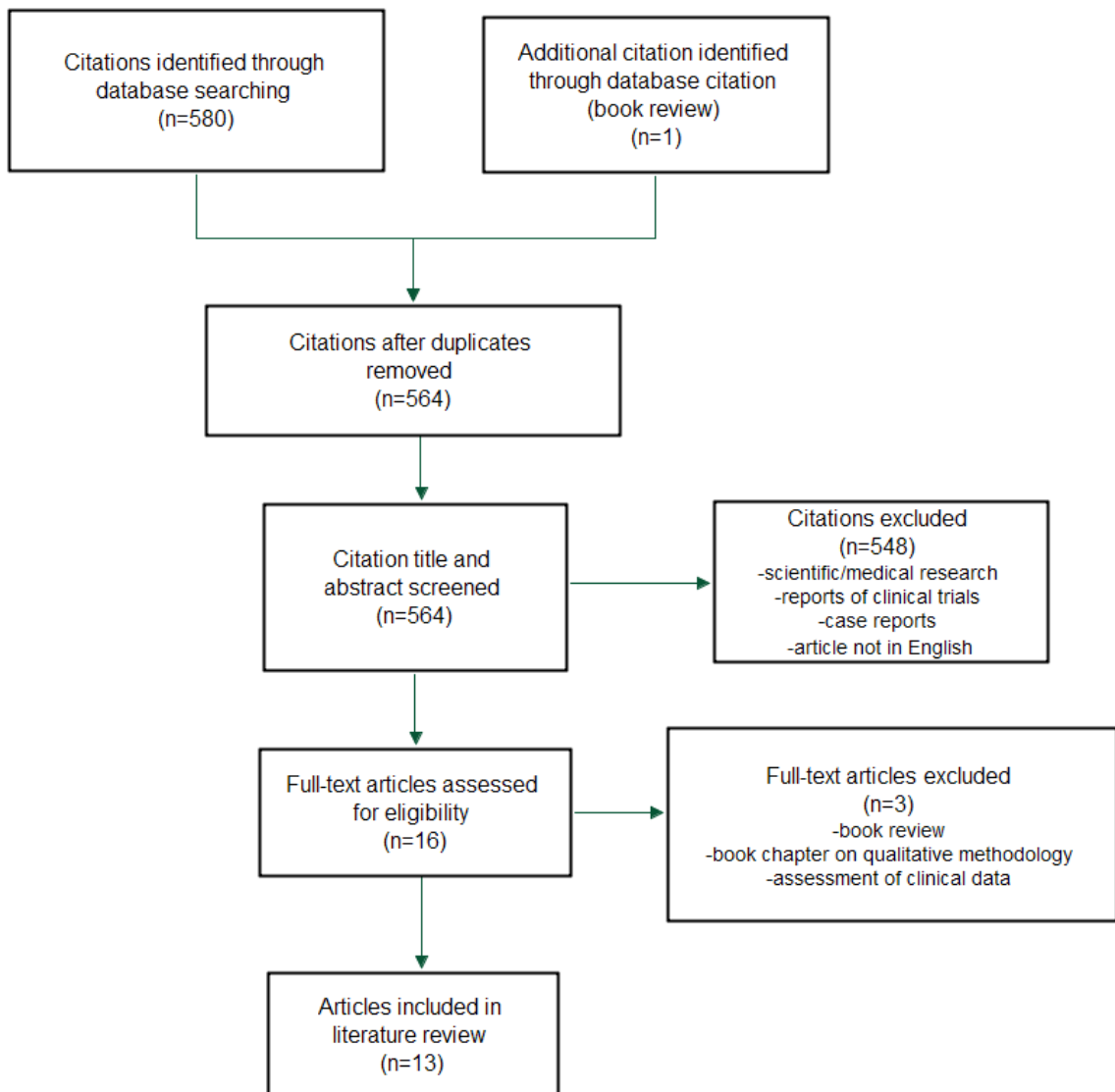


Figure 1.1 PRISMA diagram illustrating the systematic literature search on the experience of living with LAM (from Moher et al. 2009)

Table 1.1: Database search for literature on the experience of living with LAM

Database	Keywords	Available Literature on LAM	Literature on Experiences/ perspectives of women with LAM	References
MEDLINE	lymphangi leiomyomatosis (limits: English, human)	1033		
MEDLINE	lymphangi leiomyomatosis qualitative, interview*, narrative*, experience*, quality of life, perspective*, subjective, survey*/surveys and questionnaires, focus group*	129	9	Hoy 2016; Walker et al. 2015; Belkin et al. 2014; Carel et al. 2010; Cohen et al. 2009; Young et al. 2006; Ryu et al. 2006; Pollock-BarZiv et al. 2005; Cohen et al. 2005
CINAHL	lymphangi leiomyomatosis	113	5	Hoy 2016; Carel et al. 2010; Haylen et al. 2009; Ryu et al. 2006; Young et al. 2006
Expanded Academic ASAP	lymphangi leiomyomatosis	168	1	Young et al. 2006
PsycINFO	lymphangi leiomyomatosis	6	1	Vafamand 2014
Sociological Abstracts	lymphangi leiomyomatosis	0		
ProQuest Dissertation and Theses	lymphangi leiomyomatosis	164	2	Vafamand 2014; Pollock-BarZiv 2005
Additional citation			1	Carel (2013b)

The literature search yielded only two qualitative studies on women's experiences of living with LAM. Belkin et al.'s (2014) focus group study involved 37 participants, aged 34-68 years and diagnosed between 4.8 months and 39 years (mean 7.0 years). This study focused on symptoms and psychosocial experiences at the time the focus groups were conducted. Breathlessness and fatigue were the dominant symptoms experienced by the participants. Fatigue significantly affected their activities of daily living. The study did not examine the participants' experiences of diagnosis and, although sixteen women were using supplementary oxygen, their perspectives on their experiences of using oxygen therapy were not elicited. Belkin et al. (2014) reported that the primary psychological experience of LAM was one of "getting stuck with LAM". Fewer participants had adapted to living with LAM but it was not revealed how this was achieved.

Pollock-BarZiv's (2005) doctoral thesis employed mixed methods to elucidate the quality of life and psychosocial impact of LAM on lung transplant candidates and recipients. The exploratory, descriptive, qualitative arm of the study, using a narrative approach, involved 11 transplant recipients aged 33-55 years, two to 13 years post-transplant, and described the women's experiences from diagnosis to life after transplant. The study focused on participants' experiences while waiting for transplant and post-transplant. Many participants received little information and support when they were diagnosed. While one participant reported adapting to her illness, most, who were living with severe disease prior to transplant, felt unable to adjust to living with LAM and their subsequent loss of function. Most experienced anxiety and depression while waiting for a transplant, and stigma in relation to their use of supplementary oxygen. The participants' experiences of transplant varied but commonly they experienced both physical and emotional difficulties adjusting to their drug therapy, particularly steroid therapy. The quantitative arm of the study surveyed 31 women waiting for a transplant and 43 women who had received a lung transplant. Results reflected the qualitative findings. Pre-transplant quality of life was impaired by limited function, fatigue, anxiety and isolation, and improved after transplant.

Three personal narratives were located (Carel et al. 2010; Carel 2013b; Hoy 2016). Carel's narratives concerned her experiences of living with LAM from the onset of her illness when she was 35 to the time of writing the second edition of her book, *Illness*, seven years later. Carel et al. (2010) was a brief narrative written for a collection titled "A Patient's Journey", published in the British Medical Journal between 2007 and 2013 (Jessop 2014). Her book (Carel 2013b) is a more detailed narrative and reflection on her illness experience. Carel

was initially misdiagnosed as asthma and was given a prognosis of ten years when she was diagnosed in 2006. Hoy (2016), a physician with access to expert medical facilities, was diagnosed in a timely manner within six days in 2014. Carel used supplementary oxygen. She reported stigma in relation to her oxygen use, and miscommunication and being made to feel alienated by healthcare professionals. In contrast to the findings of Belkin et al. (2014) and Pollock-BarZiv (2005), Carel's (2013) narrative focused on her ability to adapt to living with her illness and experience health and wellbeing. Similarly, in her short personal narrative, Hoy (2016) reported accepting and adjusting to living with LAM and feeling comfortable with her new normality.

Seven quantitative studies were reviewed in relation to women's experiences and perspectives. Vafamand's (2014) doctoral thesis, through quantitative methodology employing predetermined questionnaires, examined statistically the effect of knowledge, attitude and lifestyle practices on the quality of life of 143 LAM patients. It concerned health-related attitudes and behaviours related to physical activity, diet and smoking. This study reported that the majority of respondents had positive attitudes towards being physically active, eating healthily and stopping smoking. More positive attitudes towards physical activity were associated with higher quality of life. There was no significant relationship between quality of life and attitudes and intent to change diet or smoking behaviours. Although Vafamand (2014) reported that confidence in handling the disease was associated with higher quality of life, the study found that greater knowledge of the disease was associated with lower quality of life. Lower quality of life was also associated with more severe symptoms and more restricted activity. It was observed that this finding contradicted earlier studies which found that increased knowledge was associated with better quality of life. Vafamand concluded there was a significant positive relationship between attitude, lifestyle practices and quality of life, and a negative relationship between knowledge and quality of life. This study did not elicit the participants' personal responses, and, confined to health-related attitudes and behaviours, did not explore other attitudes, practices or factors which might influence women's quality of life.

Ryu et al. (2006) outlined the baseline characteristics of 243 patients with LAM at the time of enrollment in the LAM Registry between 1998 and 2001. Using the SF-36 health survey and the St George's Respiratory Questionnaire (SGRQ), they found that LAM patients experienced impaired quality of life as lung function decreased. Results of both tests were compared with those of patients with chronic obstructive pulmonary disease (COPD). The

SF-36 physical and mental component scores were better for LAM patients than COPD patients. The lack of correlation between physical and mental components suggested both groups develop mechanisms to cope with the impact of their diseases. However, the results could not easily be interpreted due to a marked difference in age, sex and lung function between the two cohorts (Tattersfield & Glassberg 2006, p.3). The nature of the coping mechanisms was not investigated.

Walker et al. (2015) developed a Tool to Assess Quality of life in LAM (ATAQ-LAM) and surveyed a convenience sample of 69 LAM patients attending a LAM conference to test the tool. They reported that participants using supplementary oxygen and with reduced lung function had more impaired health-related quality of life than those not requiring supplementary oxygen. The main focus of the article was the development and validation of the tool and its ability to discriminate between patients with differing levels of LAM severity and health-related quality of life. How the women experienced life with supplementary oxygen was not explored in this quantitative study.

Cohen et al. (2009), in a survey of 328 women with LAM, reported that 55% of respondents avoided pregnancy due to concerns about the effect it might have on their condition. Women with mild disease were more likely to have favourable pregnancy outcomes. Women diagnosed before (n=12) or during pregnancy (n=15) had poorer lung function than women diagnosed after pregnancy (n=178). In addition, those diagnosed during pregnancy had increased premature births and miscarriages, and more breathlessness, pneumothorax and chylothorax. However, they found that there was no difference in quality of life, general wellbeing, depression and anxiety between women who had children and those who didn't. These findings demonstrate the complex issues involved in pregnancy for women with LAM and support the European Respiratory Society guidelines for the management of LAM which recommend women are offered individualised counselling regarding pregnancy (Johnson et al. 2010). The study did not reveal women's personal experiences or decision-making regarding pregnancy.

Three studies emphasised the need to take into account patients' subjective views of their disease, supporting the need for this current study. Pollock-BarZiv et al. (2005), in a cross-sectional study evaluating the functional status of 74 patients with LAM, reported that certain objective measurements of function (spirometry and six minute walk test) did not correlate well with patients' perceptions. They recommended that clinicians consider LAM patients' subjective assessment of their function to focus interventions and improve quality

of life. Similarly, Young et al. (2006), evaluating the opinions of 314 LAM respondents to a questionnaire on the management of pneumothorax, found a difference in opinion between LAM patients and their physicians, with most patients favouring more conservative management initially. One third of these respondents believed their physicians did not consider their preferences in this regard. Young et al. (2006) stated that understanding patients' perspectives enables cooperative decision-making and improves clinical outcomes related to the management of pneumothorax.

Cohen et al. (2005), in a cross-sectional survey of 328 women listed with the US LAM Foundation database and the UK LAM Action group, reported fatigue as a common symptom of LAM which negatively affected quality of life for 71.9% of their respondents. It occurred independent of lung function across all age groups and was not addressed by healthcare professionals. They concluded that further research was needed to explain and reduce fatigue experienced by women with LAM. Pollock-BarZiv (2005) and Belkin et al. (2014) similarly reported that fatigue was a prominent symptom causing significant impairment for their participants. This corresponds with other studies that have found that fatigue is a common aspect of many chronic illnesses and negatively affects quality of life for these people (Kralik et al. 2005; Small & Lamb 1999).

In summary, four quantitative studies and one mixed methods study examined quality of life in women with LAM and found that it was impaired. Only Vafamand (2014) measured the relationship of attitudes and lifestyle practices to quality of life. In the studies reviewed, lower quality of life was associated with reduced lung function and the use of supplementary oxygen (Ryu et al. 2006; Walker et al. 2015); severe symptoms, restricted activity, and knowledge of the disease (Vafamand 2014); fatigue (Cohen et al. 2005); and function and depression (Pollock-BarZiv 2005). Higher quality of life was associated with confidence in handling the disease and a positive attitude to physical activity (Vafamand 2014). Post-transplant respondents scored high levels of self-rated quality of life associated with lower anxiety and depression scores and higher energy/vitality levels and functional status (Pollock-BarZiv 2005). Two quantitative studies found that patients' perspectives of their function (Pollock-BarZiv et al. 2005) and medical management (Young et al. 2006) differed from objective measurements and physician perspectives and recommended these should be taken into account in clinical decision-making. The personal narratives of Carel (2010; 2013b) and Hoy (2016) revealed their ability to adapt to their limitations and experience health and wellbeing. These experiences contradict the findings of Belkin et al. (2014) of a

primary negative psychological experience of feeling “stuck” and frustrated associated with physical limitations and fatigue.

The review of the LAM literature revealed a limited understanding of women’s experiences of living with this condition. The quantitative studies demonstrated the association between quality of life and the physical symptoms of LAM, confidence and knowledge of the disease and attitude to physical activity; and the discrepancy between patient perspectives and medical assessments. They did not, however, uncover women’s personal experiences of living with their illness or how the rarity of their condition might affect their experience. Only two personal narratives and one qualitative focus group study revealed differing aspects of the experience of living with LAM. This review concluded there was a paucity of literature on women’s experiences of living with LAM.

1.3.2 Living with a rare disease

As such limited literature was found on the experience of living with LAM, it was decided to extend the literature search to include people’s experiences of living with other rare diseases, including rare lung diseases, and review the LAM literature in relation to this body of literature. Literature was sought which focused on the influence of the rarity of the condition on people’s illness experiences. The search for rare lung diseases also encompassed people’s experiences of progressive respiratory illness. There are more than 6000 rare diseases and many of these are genetic and affect individuals from birth (Rare Disease UK 2016). For the purposes of this study and because LAM is a disease of adult onset, the search was limited to adults’ experiences of living with rare disease, except for studies which were inclusive of a range of rare diseases affecting both children and adults. Literature on the experience of living with other respiratory diseases, chronic obstructive pulmonary disease (COPD), asthma, and cystic fibrosis, was not specifically reviewed. COPD is a disease commonly caused by smoking and primarily affects an older population. Asthma is common and treatable. Cystic fibrosis affects people from birth.

The search for literature on living with a rare disease was conducted using databases (MEDLINE, CINAHL, Expanded Academic ASAP, PsycINFO, ProQuest Dissertation and Theses, and Google Scholar), journal reference lists, and a journal specialising in rare disease, the Orphanet Journal of Rare Diseases. The database search terms used were rare lung disease* and rare disease*, with additional terms of qualitative or narrative* or interview* or experience*. This search aimed to extract a range of literature which focused

on the experience of rarity in a variety of diseases, including rare lung diseases, in order to provide a background of literature against which the literature on LAM could be further examined.

The search yielded 15 articles on the experience of living with two rare lung diseases, pulmonary hypertension (n=8) and idiopathic pulmonary fibrosis (n=7). See Appendix 2, Table 2.1. There was scant literature on the experience of living with other rare lung diseases. The search was limited to pulmonary hypertension and idiopathic pulmonary fibrosis as they were similar in character to LAM. Like LAM, pulmonary hypertension and idiopathic pulmonary fibrosis are rare, incurable, progressive lung diseases of adult onset, unrelated to smoking, and with similar common symptoms of breathlessness and fatigue, and may also affect younger people. See Appendix 2, Table 2.2, for a comparison of the characteristics of LAM, idiopathic pulmonary fibrosis and pulmonary hypertension. While pulmonary hypertension and idiopathic pulmonary fibrosis have significantly worse median survival times, these conditions can be compared with women's experiences of living with LAM. Idiopathic pulmonary fibrosis will be referred to as pulmonary fibrosis in this thesis for ease of reading.

Of the 15 articles reviewed on pulmonary hypertension (n=8) and pulmonary fibrosis (n=7), there were ten qualitative studies (Overgaard et al. 2016; Duck et al. 2015; Yorke et al. 2014; Kingman et al. 2014; Armstrong et al. 2012; Matura et al. 2012; Schoenheit et al. 2011; McDonough et al. 2011; Flattery et al. 2005; Swigris et al. 2005), two quantitative studies (Taichman et al. 2005; De Vries et al. 2001), one survey (Collard et al. 2007), one literature review (Belkin & Swigris 2014), and one mixed methods study (Pulmonary Hypertension Association Europe 2016). See Appendix 2, Table 2.1.

Twenty pieces of literature were retrieved which related to the experiences and perspectives of people living with other rare diseases (see Appendix 2, Table 2.3). Twelve studies were concerned with multiple rare diseases. These included three surveys conducted by rare disease associations (EURORDIS 2009; Rare Disease UK 2016; Rare Voices Australia 2013), one mixed methods study (Caputo 2014), five qualitative studies (Budysh et al. 2012; Garrino et al. 2015; Huyard 2009; Jaeger et al. 2015; Jessop 2014), two quantitative studies (Molster et al. 2016; Wallenius et al. 2009), and one literature review (Cohen & Biesecker 2010). Six studies related to rare cancers: neuro-endocrine tumour (Feinberg et al. 2013), vulval cancer (Jefferies & Clifford 2009/2011), penile cancer (Gordon 2013), mycosis fungoides – a rare skin lymphoma (Vitale 2005), leiomyosarcoma (Garau 2016 – a personal

narrative), and gallbladder cancer (Dockser Marcus 2010 – a personal narrative). Joachim and Acorn's (2003) study on scleroderma was included as it focused on the experience of living with a rare disease. Oksel & Gunduzoglu's (2014) article on women's experiences of living with scleroderma provided a further comparison of this condition.

The literature on the lived experience of LAM is now discussed in relation to the rare disease literature that was retrieved. Discussion concerns the experience of living with an incurable lung condition and how the rarity of a disease affects the illness experience. Four significant aspects of experience emerged from the rare disease literature: searching for a diagnosis, receiving the diagnosis, daily struggles, and searching for expert care and treatment in the healthcare system. While the issue of the effect of rarity on the illness experience was not specifically discussed in the literature on the lived experience of pulmonary hypertension and pulmonary fibrosis, in common with the rare disease literature, these studies revealed these aspects of experience to be important for participants.

1.3.2.1 Searching for a diagnosis

A period of misdiagnosis or delayed diagnosis was reported as a common experience for people with rare diseases in the majority of studies examined in this literature review. The participants of Pollock-BarZiv's (2005) study experienced variable onset of symptoms from sudden onset with lung collapse to gradual development of breathlessness over a period years which was difficult to diagnose. Most experienced multiple medical consultations and investigations with delays in diagnosis and being misdiagnosed before they received their diagnosis. Carel (2013b) was originally misdiagnosed with asthma.

Pollock-BarZiv's (2005) findings and Carel's (2013b) experience are reflected in two surveys conducted between 2003 and 2008 by the European Organisation for Rare Diseases, EURORDIS, on the experiences and expectations of 12,000 patients with 16 rare diseases on diagnosis and care in Europe (EURORDIS 2009). Patients with LAM were not included in these surveys. The results revealed 40% of patients were initially misdiagnosed and received inappropriate medical and psychological treatment, including psychiatric medication. Diagnosis was delayed between five and 30 years for 25% of patients. Similar findings are reported in surveys conducted by Rare Diseases UK (2016) and Rare Voices Australia (2013).

Misdiagnosis and delays in diagnosis were also found in studies of other rare diseases in various countries (Feinberg et al. 2013; Garau 2016; Garrino et al. 2015; Gordon 2013;

Molster 2016; Vitale 2005), pulmonary hypertension (Armstrong et al. 2012; Kingman et al. 2014), and pulmonary fibrosis (Belkin & Swigris 2014; Duck et al. 2015; Overgaard et al. 2016; Schoenheit et al. 2011), as were symptoms being attributed to psychosomatic causes (Armstrong et al. 2012; Duck et al. 2015). Jessop (2014, p. 108), in a narrative typology exploring how people with rare diseases structured their narrative accounts, referred to searching for a diagnosis in the context of a rare disease as “the quest for diagnosis”.

1.3.2.2 Receiving the diagnosis

Receiving their diagnosis had a significant impact on women with LAM and people with pulmonary hypertension and pulmonary fibrosis. They experienced shock and uncertainty (Carel 2013b; Flattery et al. 2005; Kingman et al. 2014; Overgaard et al. 2016; Pollock-BarZiv 2005); fear (Armstrong et al. 2012; Hoy 2016; McDonough et al. 2011; Swigris et al. 2005); anger and grief (Carel 2013b; Hoy 2016; Pollock-BarZiv 2005); anxiety (McDonough et al. 2011; Swigris et al. 2005); and isolation (Belkin & Swigris 2014; Kingman et al. 2014; Pollock-BarZiv 2005). People with other rare diseases also reported feeling isolated because of their diagnosis (Garrino et al. 2015; Huyard 2009; Jefferies & Clifford 2011).

A significant aspect of being diagnosed with a rare disease was the manner in which the diagnosis was delivered. Women with LAM and people with other rare diseases reported being given their diagnosis in an insensitive manner and in insufficient time (Carel 2013b; Schoenheit et al. 2011), and in inadequate or inappropriate conditions (EURODIS 2009). Multiple studies reported that participants were provided with little information (Carel 2013b; Collard et al. 2007; Duck et al. 2015; EURORDIS 2009; Garau 2016; Garrino et al. 2015; Gordon 2013; Jefferies & Clifford 2009; Pollock-BarZiv 2005; Rare Diseases UK 2016; Rare Voices Australia 2013) or emotional support (Armstrong et al. 2012; Belkin & Swigris 2014; Carel 2013b; EURORDIS 2009; Garau 2016; Garrino et al. 2015; Gordon 2013; Pollock-BarZiv 2005). According to EURORDIS (2009), problems with diagnosis arise from healthcare professionals’ general lack of awareness and knowledge of rare diseases.

When their information and support needs were not met by healthcare professionals many people with rare diseases and their families embarked on an independent search for information and support. This was reported by women with LAM (Carel 2013b; Hoy 2016)

and vulval cancer (Jefferies & Clifford 2009), people with pulmonary hypertension (Armstrong et al. 2012), mycosis fungoides (Vitale 2005), and other rare diseases (Rare Diseases UK). The internet became a significant but not always reliable source of information for these people. Although a physician herself, Hoy (2016) referred to Wikipedia and read outdated information of a ten year survival period for LAM, rather than consulting medical publications available at that time which indicated a median survival time of 29 years from diagnosis (Oprescu et al. 2013). While peer support organisations are a valuable source of information and support, a survey conducted by Rare Diseases UK (2016) found that four out of five respondents were not referred to these groups by their physicians when they were diagnosed. Similarly, women with vulval cancer (Jefferies & Clifford 2009) and people with neuro endocrine tumour (Feinberg et al. 2013), mycosis fungoides (Vitale 2005), and pulmonary fibrosis (Schoenheit et al. 2011) reported having difficulty finding others with their condition and disease specific support.

When they were diagnosed, women with LAM (Carel 2013b; Pollock-BarZiv 2005), and people with pulmonary hypertension (Kingman et al. 2014), pulmonary fibrosis (Duck et al. 2015; Schoenheit et al. 2011; Swigris et al. 2005) and scleroderma (Oksel & Gunduzoglu 2014) experienced disruption of their sense of self, and social roles, relationships, and interactions. Disruption was accompanied by feelings of loss of their previous life (Carel 2013b; Duck et al. 2015), independence (Duck et al. 2015; Overgaard et al. 2016; Pollock-BarZiv 2005; Schoenheit et al. 2011; Swigris et al. 2005), friends and social participation (Carel 2013b; Swigris et al. 2005), spontaneity (Carel 2013b; Duck et al. 2015; McDonough et al. 2011), and financial status (Carel 2013b; Schoenheit et al. 2011). Loss of independence and needing help from others was accompanied by feelings of guilt (Belkin et al. 2014; Duck et al. 2015; Pulmonary Hypertension Association Europe 2016), being a burden (Schoenheit et al. 2011), and loss of control (Pollock-BarZiv 2005; Duck et al. 2015).

1.3.2.3 Daily struggles

Four studies examined quality of life for people living with pulmonary hypertension and pulmonary fibrosis and found that it was impaired (Belkin & Swigris 2014; De Vries et al. 2001; Swigris et al. 2005; Taichman et al. 2005). The findings were associated with the effects of the disease rather than its rarity and reflected the quality of life findings in the LAM literature. For people with pulmonary fibrosis, reduced quality of life was associated with physical symptoms, fatigue and using supplementary oxygen (Swigris et al. 2005); and depression, anxiety and isolation (Belkin & Swigris 2014). De Vries et al. (2001), in a

quantitative study examining the relationship between quality of life, depressive symptoms and breathlessness inpatients with pulmonary fibrosis, reported that impaired quality of life for these people was associated with physical symptoms, fatigue, reduced mobility, limited activities of daily living and working capacity, and depression. Taichman et al. (2005), in a cross-sectional survey of health-related quality of life in patients with pulmonary hypertension, reported that respondents had severe impairments in both physical and emotional domains of health-related quality of life associated with measurements of reduced functional status.

Seven studies found that women with LAM and people with pulmonary hypertension managed symptoms of breathlessness and fatigue and the limitations these imposed on their activities and lifestyle (Armstrong et al. 2012; Belkin et al. 2014; Carel 2013b; Kingman et al. 2014; McDonough et al. 2011; Pollock-BarZiv 2005). They did this by pacing and adjusting their activity levels (Carel 2013b; McDonough et al. 2011), developing other hobbies (Carel 2013b; Kingman et al. 2014; McDonough et al. 2011), and redefining life (Carel 2013b; Flattery et al. 2005; McDonough et al 2011).

The sense of disruption experienced by Carel (2013b) and other people with rare disease is common to chronic illness. Bury's (1982, p.169) seminal study of rheumatoid arthritis recognised that chronic illness was a form of "biographical disruption", in that its disruptive effects encompassed a person's identity, relationships and everyday life within their social, cultural and familial contexts which may change over time. He found that a person's response to chronic illness involved searching for meaning, and an attempt to adjust and mobilise physical, cognitive, spiritual, social, material or financial resources. Further, Williams (1984) claimed people attempt to repair the disruption of chronic illness by reinterpreting their biography in a process of narrative reconstruction. Williams (2000) extended the concept of biographical disruption to include the concept of biographical continuity where people might apply a range of meanings to their illness depending on the timing, context and circumstances in which the illness occurs.

Reflective of these concepts, chronic illness has been described as a complex, shifting, multidimensional and disruptive experience for women living with various illnesses and conditions, including breast cancer, diabetes and fibrocystic breast disease (Packard et al 1991), rheumatoid arthritis (Plach et al. 2004), chronic fatigue syndrome and fibromyalgia (Asbring 2001), premature menopause (Boughton 2002) and ovarian cancer (Ryan 2005). Similarly, Levealahti et al. (2007), in a qualitative study exploring how people with

inoperable lung cancer conceptualize the onset of their illness, found that participants viewed their symptoms as having different meanings ranging from disruption to continuity of various aspects of their lives depending on their lifestyles and history. Kralik (2002, p.146), in a narrative inquiry into the meaning of 81 midlife women's experiences of living with chronic illness, extended the concept further, finding that women reconstructed their lives disrupted by illness in a fluctuating process of moving from a state of "extraordinariness" to "ordinariness".

In line with Kralik's (2002) findings, Carel (2013b) was able to experience health and wellbeing while living with LAM when her lung function stabilised due to treatment with sirolimus medication, and by focusing on being positive and living in the present. Hoy (2016, p. 238) described sirolimus as a source of hope in its ability to stabilise her LAM and improve her health. Like Carel (2013b), Cohen and Biesecker (2010), in a systematic review of quality of life in rare genetic conditions, and Caputo (2014), in a content analysis of written illness stories of 32 people with four rare diseases, found that people with rare diseases were able to experience good quality of life in spite of serious disability. According to their personal narratives, sirolimus was significant for Carel (2013b) and Hoy (2016) but no other studies have sought women's perspectives on this new treatment.

In contrast with the above studies which found that people with chronic illness can move from feelings of disruption to continuity, Belkin et al. (2014, p.3) reported that, while several participants adapted to their illness and showed defiance, most experienced "getting stuck with LAM" and felt frustration, resentment, worry and fear. Forty three percent of Belkin et al.'s (2014) participants were using supplementary oxygen and it is not clear if these feelings were related to their stage of illness or oxygen therapy. According to Duck et al. (2015), in a qualitative study of the experiences of people with pulmonary fibrosis, people with pulmonary fibrosis, with a low mean life expectancy of two to four years, felt a sense of loss and reported struggling with living with their illness, and being dependent on their oxygen to manage their breathlessness. Similarly, Pollock-BarZiv (2005) found that most of her participants were unable to adjust to their advancing disease before transplant as they experienced uncertainty, fear, anxiety, struggling to stay alive, and being completely dependent on others. It is not known how women with LAM who do not receive a transplant and progress to respiratory failure experience this stage of their illness.

The rarity of a disease can negatively affect people's social lives and create feelings of being stigmatised and isolated due to a lack of knowledge and understanding of rare disease in the

community. Pollock-BarZiv (2005) reported that when participants started oxygen therapy they felt an altered sense of self, took on a 'sick' role, and experienced stigma using oxygen in public. Carel (2013b) also felt stigmatised but she looked healthy when not using oxygen and subsequently also felt a lack of empathy from others. Yorke et al. (2014), in a qualitative study of the experience of living with pulmonary hypertension, found this effect was compounded by the lack of knowledge of their disease for people with pulmonary hypertension. Tattersfield and Glassberg (2006), in an editorial article on a national registry for LAM, noted that women with LAM can find that their local doctors and friends have not heard of the condition and their specialist physician may not previously have treated a patient with LAM. Joachim and Acorn (2003), in a focus group study of women living with scleroderma, found that this created feelings of social isolation and fear of rejection. Similarly, Jaeger et al. (2015), in a focus group study of the experiences of adults living with different rare diseases, reported that lack of understanding in the community affected the ability of their participants to participate in activities. Further, according to a survey conducted by Rare Voices UK (2016), social, educational and employment opportunities are restricted for people with rare disease. Vitale (2005), in a phenomenological study of the experience of living with mycosis fungoides as a rare disease, found that social isolation was compounded when people with mycosis fungoides were geographically isolated. In this context, patient organisations (Belkin et al. 2014; Carel 2013b; Duck et al. 2015) and online communities and forums (Kingman et al. 2014; Matura et al. 2012; Vitale 2005) have been found to be a valuable source of support for people with rare diseases.

Three studies revealed that participants' sexual lives were altered by the effects of their illness and that it was an important aspect of their experience. Oksel & Gunduzoglu (2014), in a phenomenological study of the experiences of 20 women living with scleroderma, reported that women with scleroderma experienced sexual dysfunction when their sexual lives were disrupted by their physical symptoms and altered body image. Similar findings were reported in a mixed methods study conducted by the Pulmonary Hypertension Association Europe (2016) for people living with pulmonary hypertension (2016) and in a qualitative study on quality of life for people with pulmonary fibrosis (Swigris et al. 2005). These studies found that their participants' illness caused a decrease in sexual activity and loss of libido which had a significant effect on their daily life. No studies have reported how LAM affects women's sexual lives.

Women with LAM face decision-making regarding pregnancy and options for having children. Carel (2013b) accepted the recommendation of her physicians to avoid pregnancy, prioritising staying alive over having children. Hoy (2016) and her partner harvested eggs and froze embryos but did not discuss their decision-making on this issue. In contrast to Carel (2013b), the participants of Boughton's (2002) phenomenological study of premature menopause, a non-life-threatening condition, felt the disruption of their expected plans for procreation as a sense of alienation and loss of their female identity. Pollock-BarZiv (2005) and Belkin et al.'s (2014) participants did not reveal their perspectives on this issue.

Women with LAM may be working and raising children born prior to diagnosis. Pollock-BarZiv (2005) reported that their participants perceived their identities changed as they had to relinquish or alter work and family roles as their illness progressed. They found it eventually impossible to carry out their mothering duties. Belkin et al. (2014), Carel (2013b) and Pollock-BarZiv (2005) did not discuss how women with LAM managed work and parenting while living with their illness.

1.3.2.4 Searching for expert care and treatment in the healthcare system

According to surveys conducted by EURODIS (2009), healthcare professionals' lack of awareness and knowledge is a significant barrier to people with rare disease being able to access quality health care and improve their quality of life. Similarly, Joachim and Acorn (2003, p.605), in a focus group study of the experience of living with scleroderma, stated that the "knowledge, attitudes and behaviour" of healthcare professionals play an important role in assisting people with rare conditions adjust to their illness. They asserted that nurses' ignorance of rare diseases produces stigmatising responses to patients, compounding patients' complex experience and creating misunderstanding. Joachim and Acorn (2003, p.605) summarised the experience of living with scleroderma as "complex" and one made "more complex" by the rarity of the condition.

Patients with rare diseases and their families feel marginalised in the health care system and frequently have to search for expert care and advocate for appropriate treatment (EURORDIS 2009). EURODIS (2009) reported that more than 25% of the respondents to their surveys struggled to access services and preferred to be managed in centres of expertise. Many relocated for this purpose. According to Vitale (2005), people with mycosis fungoides felt it was a burden to travel considerable distances to receive specialised care. In addition, people with rare diseases experience a lack of holistic, multidisciplinary care

(Jaeger et al. 2015; Rare Diseases UK 2016), difficulty in accessing social services (Wallenius et al. 2009; Yorke et al. 2014), and poor communication between medical specialists (Jaeger et al. 2015; Molster et al. 2016). Duck et al. (2015), in a qualitative study on the experience of living with pulmonary fibrosis, found that their participants similarly were frequently faced with poor service provision. Inequities in accessing effective therapy were also reported in the Australian context. According to Rare Voices Australia (2013), less than 1% of the Australian rare disease population is able to access effective treatment. It is not known how this applies to women living with LAM in Australia.

Huyard (2009), in a qualitative study examining to what extent people with rare disease think the rarity of their disease contributes to their difficult experiences, argued that difficult rare disease experience was a result of healthcare professionals failing to meet the moral needs of their patients rather than the rarity of the disease itself. This view recognises the influence of healthcare professionals on the experience of people with rare disease but does not take account of the impact, reported in the studies above, of the rarity of the condition on people's social lives in the broader community due to the lack of understanding they experience.

People with rare diseases, including women with LAM, advocate collectively, through patient organisations, to raise funds for research into their condition (LAM Foundation 2015; Joachim & Acorn 2003). In this way, Ayme et al. (2008), in an essay on patient empowerment for people with rare diseases, claimed people with rare diseases are empowered by their active participation in research and contribute to the development of scientific knowledge. Finn (2008), in an ethnographic study of empowerment in the alpha-1 antitrypsin community, found that advocacy and networking with other patients and care providers contributed to patients' empowerment. Carel (2013b) reflected on the stigma and lack of empathy she experienced in her interactions with healthcare professionals and stated that she had become her "own advocate" and "a patient activist" through her involvement with the UK LAM peer support organisation (p.156). Belkin et al. (2014) and Pollock-BarZiv (2005), however, did not explore women's experiences of advocacy or their encounters with healthcare professionals.

1.3.3 Summary

This review of the available literature on the experience of women living with LAM revealed a paucity of qualitative literature concerned with women's subjective illness experiences. Four quantitative studies and one mixed methods study reported quality of life

was impaired for women with pulmonary LAM (Cohen et al. 2005; Pollock-BarZiv 2005; Ryu et al. 2006; Vafamand 2014; Walker et al. 2015) Three quantitative studies sought women's perspectives on fatigue (Cohen et al. 2005), functional status (Pollock-BarZiv et al. 2005), and management of pneumothorax (Young et al. 2006), and supported the view that taking patients' subjective perspectives into account can improve clinical management, outcomes, and quality of life for women with LAM.

Only two qualitative studies (Belkin et al. 2014; Pollock-BarZiv 2005) and two women's personal narratives (Carel 2013b; Carel et al. 2010; Hoy 2016) were located. These described aspects of women's LAM illness experiences and were reviewed in relation to literature on other rare diseases. Carel (2013b) and Pollock-BarZiv (2005) demonstrated that receiving a diagnosis of LAM created a sense of disruption and feelings of isolation. Carel's (2013b) narrative focused on her ability to adapt to her illness and experience health and wellbeing. Similarly, Hoy (2016, p.238) was able to accept and adjust to her "new normalcy". In contrast, Pollock-BarZiv (2005) and Belkin et al. (2014) found that most of their participants had difficulty adjusting. It is not clear if this was related to the stage of illness of the participants.

Many areas remain unexplored. While Pollock-BarZiv (2005) explored the experiences of women from onset of symptoms to following lung transplantation, the study was focused on experiences of transplant and did not include women who reached the late stage of LAM and did not receive a transplant. It is not known whether most women with LAM adapt to their illness, as was the case for Carel (2013b) and Hoy (2016), or how the stage of their illness affects their ability to adapt. Many aspects of living with LAM have not been explored, for example, managing oxygen therapy, personal relationships, sexuality, the need to avoid pregnancy, parenting and working. While Carel (2013b) reflected on her experience of stigma in her relations with healthcare professionals, women's experiences of advocacy and interactions with healthcare professionals, identified as significant aspects of living with a rare disease in other studies, have not been explored in detail in relation to LAM. Carel (2013b) and Hoy (2016) benefited from stabilised lung function due to sirolimus therapy but no studies sought women's perspectives on this significant new treatment for women with LAM. Women's experiences in the Australian context, where the numbers of women with LAM are so limited, have not been investigated.

This literature review found a significant gap in the available literature on women's experiences of living with LAM and identified a clear need for further qualitative research

to address the lack of understanding in this area. Considering LAM is a chronic illness with marked clinical variability and rate of progress, a broad range of women's personal views of their illness is needed in order to understand the meaning of their experiences in their individual social contexts and healthcare settings at different stages of their illness across time.

1.4 Aims of the study and research question

The purpose of this nursing research is to explore and capture a comprehensive view of the experiences of women living with LAM over their life course. Specifically, the research aims to:

- increase understanding of the meaning of women's experiences of living with LAM over time across the illness trajectory;
- understand how the rarity of LAM affects the women's experiences;
- identify issues of concern for women living with LAM;
- enhance the existing body of nursing knowledge to inform the practice of nurses and other healthcare professionals and improve the care and support provided to women with this disease.

To address these aims, the study seeks to answer the following research question:

What are women's experiences of living with LAM over their life course?

1.5 Significance of the research

Although LAM is rare, increasing numbers of women are being diagnosed with this condition. Nurses may encounter LAM patients in community and hospital inpatient and outpatient settings as they are treated for LAM, undergo transplant workup or receive a transplant. As a rare disease it is not uncommon for nurses caring for these patients to have not heard of LAM or know very little about the disease and its impact on these women.

This research will provide accessible information for nurses, physicians and other healthcare professionals to increase the current limited understanding of women's experiences of living with LAM across their illness trajectory. The addition of the experiential dimension of LAM to the body of biomedical knowledge may improve clinical management, outcomes and quality of life for these women. Furthermore, informing healthcare professionals of the problems women encounter as they live with LAM may increase empathy and support for

them and reduce their sense of isolation in living with a rare disease. This study enables women living with LAM to have their voices heard and their needs addressed. Finally, the insights gained from this research may illuminate the experience of living with other rare diseases.

1.6 Overview of the thesis

The thesis is presented eight chapters.

Chapter 1 has introduced the thesis and provided a background to the study and an account of its origins. It has presented the literature review conducted to locate the available literature on women's experiences of living with LAM, demonstrated a paucity of literature on this subject, and established a need for further research. It has stated the aims of the study, the research question, and significance of this research.

Chapter 2 discusses the life history methodology used in this thesis in relation to its theoretical framework, recruitment of participants, data collection and analysis, rigor, and ethical considerations. The participants are introduced in brief biographies at the end of the chapter.

Chapters 3 to 6 present the findings of the study as a constructed, collective life history focused on women's experiences of living with LAM at significant turning points and periods of their illness. Chapter 3 presents women's experiences of being diagnosed with LAM. Chapter 4 concerns their life after diagnosis as they learned to live with their illness. Chapter 5 investigates their experiences of interacting with healthcare professionals. Chapter 6 explores their experiences of living with advanced disease, including experiences of oxygen therapy, respiratory failure and lung transplantation.

Chapter 7 discusses the findings in relation to existing theory and illness literature in order to link this research to broader issues and knowledge.

Chapter 8 discusses the implications of this research for nursing practice and directions for further research. The life history method is reviewed and limitations of the study considered. The thesis concludes with a personal reflection and concluding remarks.

1.7 Conclusion

This chapter has introduced the thesis, provided a background to the study and presented the literature review which was conducted to locate and analyse the available literature on the experiences of women living with LAM. Due to the paucity of literature identified, the

literature review was expanded to include the lived experience of other rare diseases. The LAM literature reviewed was then examined within this broader body of research. Significant gaps were identified in the literature on LAM and a need was established for this study on women's experiences of living with LAM. The aim of the study, research question and significance of the research were presented. Finally, an overview of the thesis was provided. The life history methodology used in the thesis is highlighted in the next chapter.

Chapter 2

METHODOLOGY

2.1 Introduction

Life history methodology was used in this study to answer the research question:

What are women's experiences of living with LAM over their life course?

The literature review undertaken in the previous chapter revealed a paucity of qualitative literature on this topic and suggested that understanding women's perspectives of their illness may improve support, clinical management and quality of life for women with LAM. This chapter presents the life history methodology used in the study. It is discussed in relation to the theoretical framework provided by Gadamer's (2004) hermeneutic philosophy and Rosenthal's (1993) biographical interpretive method of narrative analysis, followed by discussion of the research process, including participant recruitment, data collection and analysis, rigor, and ethical considerations. Finally, the participants are introduced in alphabetical order with a brief biography and factual account of their illness.

2.2 Theoretical framework

The studies of LAM and rare disease examined in the literature review reflected the concept of chronic illness experience as biographical, involving a person's identity, interactions, and everyday life in their individual social contexts over time (Bury 1982). To understand women's experiences of living with LAM from their perspective, this study is therefore concerned with personal meaning and subjective biographical knowledge situated in context and across time. Steger (2012, p.165) defined meaning as "the web of connections, understandings, and interpretations that help us comprehend our experience and formulate plans directing our energies to the achievement of our desired future". This definition of meaning alludes to the complex, interpretive nature of experience and provided direction in selecting the methodology of the study. To address the complex, meaning-focused, biographical nature of the research question, the methodology of this study adopted a life history approach and hermeneutic interpretive theoretical framework. The reasons for this are explained below.

Life history methodology explores human experience as part of a person's whole life. Denzin (2009) asserted that central features of life history are its focus on the individual and their interpretations of their life, and its capturing of events over time. Furthermore, according to Cole and Knowles (2001), it provides insights into the meaning of experience by examining how it is shaped by the complex interaction between personal, social, contextual and historical influences. In this way, according to Josselson (1995), life history increases understanding by revealing the multidimensional character of human meaning. Smulyan, (as cited in a personal opinion in Hatch & Wisniewski 1995, p.120), claimed that it can "demonstrate forces and powers that shape experience", and, as Denzin (1989) highlighted, give voice to marginalised people. A life history approach was therefore determined appropriate for this study as it aimed to give voice to women living with LAM who, as the literature review demonstrated, can feel isolated in communities and health care settings.

Denzin (1989, p.70) argued that life history reveals turning points, epiphanies and transformations by examining events and how they impact on an individual and their life trajectory. He regarded epiphanies as "moments of crisis altering the fundamental meaning structures in an individual's life". Similarly, McAdams et al. (2001) viewed turning points as life transitions involving significant change. Wethington (2003) claimed that identifying these points provides insights into how a person makes meaning in challenging life situations, and, according to Sparkes (as cited in Hatch & Wisniewski 1995), how they reconstruct identity and self over time. Life history, therefore, is a source of rich data to understand women's experiences of living with LAM at different stages of their illness over time.

According to Denzin (1989), life history constructs an account of a person's life based on their life story narrated in interviews. Widdershoven (1993, p.2) argued life history is hermeneutic because a person interprets their life and creates meaning as they tell their life story. As such, he claimed, human life is a "process of narrative interpretation". In light of the hermeneutic nature of life history, Gadamer's hermeneutic philosophy and theory of interpretation was adopted as the theoretical framework for this study.

2.2.1 Gadamer's hermeneutic philosophy

Gadamer (2004, p.390) claimed that "understanding occurs in interpreting" and "language is the universal medium in which understanding occurs". A central tenet of Gadamer's philosophy was the concept of prejudice. For Gadamer (2004), the prejudices of an

individual are their background understandings – that of the family, society, and state in which one lives. He argued that these form the historical reality of a person's being and always influence any experience. In this way, according to Koch (1996), a person's prejudices affect their interpretation of an interaction or text. Gadamer's philosophy can be applied to personal experience and the life history research process. A person understands their life as they tell their life story and a researcher gains understanding in interpreting the life story. Further, a person interprets their experiences through the lens of their prejudices as they tell their life story. Similarly, a researcher brings their prejudices to the interaction and interpretation of the life story.

When interpreting and understanding a text, Gadamer (2004, p.271) emphasised that one must "remain open to the meaning of the other person or text...this openness always includes our situating the other meaning in relation to the whole of our own meanings or ourselves in relation to it". He argued we do not exclude ourselves from the interpretation of a text but rather acknowledge our prejudices so that the text can present its own truth and otherness against our prior understandings.

In this study, as researcher, my own prejudices included my background as a practising clinical nurse; my experience of being in close contact with my father as he lived with late-stage Parkinson's disease; and my professional and personal values of respect for the dignity, values and beliefs of each participant, honesty, ethical awareness, reflexivity and empathy. In having met only one woman with LAM previously in the hospital clinic, I did not have preconceived ideas of the participants' illness experiences. I was, however, motivated by my experience of listening to the woman's story to hear the participants' stories and understand their experiences and concerns. I was also aware from the literature I had reviewed that my own lack of knowledge could contribute to the sense of isolation which women with LAM and other people with rare diseases can experience in healthcare settings. Therefore, committed to contributing a deeper understanding of their experiences to nursing knowledge, I brought a sense of curiosity and openness to uncovering meaning in this study.

The concept of prejudice and acknowledging one's background understandings is reflected in Frank's (2000, p.356) term "standpoint". According to Frank, a standpoint represents a person's perspectives and position in the world. These evolve through the person's individual and shared life experiences and the choices they make. Frank (2000) argued that taking a standpoint in narrative research involves an ethical act of self-reflecting on the

experiences and choices which have shaped one's perspectives and choosing to give precedence to some to inform the approach one takes to the research. Frank's (2000) standpoint, for example, of privileging a person's illness story over academic analysis as a means of informing clinical understanding reflected his own lived experience of serious illness and work as a therapist. Similarly, Thomas (2010) examined how her own life experiences of living with personal and family illness and disability and working as a sociologist shaped her approach to narrative research in her aim to improve cancer services by giving precedence to people's narratives of their illness experience and broader social contexts. Likewise, my work as a nurse, position as a PhD student, and experience of meeting a woman with LAM in the hospital clinic shaped my standpoint of prioritising the voices of my participants within the academic conventions of a doctoral thesis as a means of informing nursing practice to improve support for women with LAM.

Gadamer (2004, p.301) referred to the concept of "horizon" which he defined as "the range of vision that includes everything that can be seen from a particular vantage point". A person's horizon is their perspective and this is formed with the past as they continually test their prejudices. Gadamer (2004, p.305) claimed that understanding is always the fusing of past and present horizons, a "fusion of horizons". Applying this concept, Widdershoven (1993), held that, in telling a story of an experience, a person fuses their present view with their past perspective to change and enrich the meaning of the story. Gadamer (2004, pp.307, 309) asserted that "the meaning of a text goes beyond its author" so that "understanding is...always a productive activity". Further, he argued, "the discovery of the true meaning of a text...is an infinite process...new sources of understanding are continually emerging that reveal unsuspected elements of meaning". This can be seen when, in reading and interpreting a text, the perspectives or horizons of text and reader are fused to form a new, more encompassing meaning (Widdershoven 1993). Understanding, therefore, is never complete as it continually evolves with new experiences and interpretations.

The hermeneutic circle, a key concept of hermeneutics, describes "the experience of moving dialectically between the part and the whole" (Koch 1996, p.176). Gadamer (2004, p.302) argued "we must understand the whole in terms of the detail and the detail in terms of the whole". Narrative is interactive, and narrator, researcher and reader all take part in what Sandelowski (1991, p.162) referred to as the "hermeneutic circle of interpretation". Each brings their particular background/prejudices and prior perspectives/horizons to the interaction, influencing interpretation of the story. Rosenthal (1993) argued this is not a

problem but part of the interaction of the narrator with their social world. The hermeneutic circle can be seen in the interaction between participant and researcher during the interview, in the process of interpretation which involved moving between narrative segments and the whole of each participant's life story, and between each life story and the constructed life history.

According to Gadamer's philosophy, each participant's life story in this study reflected their prejudices and a fusion of their past and present perspectives. In turn, through the process of analysing and interpreting the life stories, my perspective as researcher was fused with the perspective of each participant. Further, in constructing the life history, meaning was created in fusing the multiple interpretations, perspectives and realities of the participants with my interpretations. Readers of the research will further enrich its meaning as they fuse their own perspectives with the interpretations presented in the thesis.

In deciding which approach to adopt for analysis the level of interpretation to be undertaken was considered. Plummer (2001, p. 179) suggested "a continuum of 'construction'" ranging from the unedited life history in which the participant's life story is not interpreted and speaks for itself, to the researcher's theoretical construction made independently of the participant's account. In between, the construction involves the participants speaking for themselves but their accounts are organised around themes and linked to theory. It was decided this level of interpretation was appropriate in meeting the aim of the study to give voice to the participants but also interpret their stories to uncover deeper levels of understanding.

2.2.2 The biographical interpretive method, Rosenthal (1993)

In accordance with the study aim and the hermeneutic interpretive framework, the biographical interpretive method of analysis, using Rosenthal's (1993) approach, was selected as the framework to guide the process of data collection and analysis. Chamberlayne et al. (2000) argued that the strength of this method is its ability to uncover deeper levels of personal meaning. The unit of analysis is the life story. Rosenthal (1993, p.61) believed that life history and life story are "continuously and dialectically linked and produce each other", but distinguished between life story and life history. According to Rosenthal (1993, p.89) the life story is narrated and constructed in the present moment at the time of the interview, while life history is the experienced "lived through life".

In line with Gadamer's (2004) concepts of prejudice and horizon, Rosenthal (1993, p.62) regarded a life story as a process influenced by a person's "biographical construct" or structure of meaning, comprising their past experiences, present perspective and anticipated future, within the context of their interaction with the researcher. According to Rosenthal, in telling their life story, a person is orientated by their "biographical construct" and selects what they interpret are biographically and thematically relevant experiences. Before analysing the thematic aspects of the life story, Rosenthal's method involved firstly extracting objective biographical data from the interview to determine which of these, and in what order, were relevant to the narrator and consequently selected for narration. In this study, objective facts would also be obtained from each participant's medical record.

According to Rosenthal (1993, p.60), the life story forms the "data base" from which the life history can be reconstructed. In this process experiences are reconstructed in the chronological sequence in which they occurred. The reconstructed life history represents the past perspective and "the biographical meaning that the experiences had at the time they happened", and reveals "transformation processes" in a person's life history (Rosenthal 1993, p. 67-69). Rosenthal viewed the reconstruction of the life story and the life history as hermeneutic as both are interpretations.

The layers of analysis involved in Rosenthal's method offered a number of benefits for answering the research question of this study. Firstly, it offered a clear process to uncover present and past meaning and therefore changes in meaning over time, as well as each participant's individual background/prejudices and how they influenced their interpretation of their experiences. It was additionally a way to examine the relationship between the objective disease process and participants' subjective experience of their illness. Finally, it allowed the participants' perspectives/horizons, revealed in the interviews, to be compared with the objective facts contained in their medical records.

Examining the participants' medical records was a means of gathering data related to their medical assessments and the chronological progress of their LAM. This data could then be compared with the participants' subjective views as narrated to reveal how the participants prioritised their illness experiences and any differences between their perspectives and the objective assessments recorded in their medical notes. The collection of the participants' medical data is consistent with life history research. According to Denzin (2009), life history materials consist of any documents that are related to a person's experiences and may include both public and private archival material, including secondary sources written by a

third person. The participants' life stories were the primary source of data while their medical records were secondary sources of data. Denzin (2009, p.224) argued that "a complete life history will combine as many primary and secondary sources as possible", while focusing on a person's personal account.

2.3 Recruitment of the participants

Originally it was proposed to use purposive sampling for this study. Participants of varying ages, social and cultural backgrounds, and at different stages of the disease were sought to give as broad a range as possible of experiences of living with LAM. It was intended to include participants with advanced disease who were using supplementary oxygen therapy, and who had received a lung transplant.

In accordance with HREC (Human Research Ethics Committee) approval, a physician at a hospital which provided a clinic for LAM patients agreed to send out invitations to potential participants to join the study. The aim of the study was explained to the physician and it was requested that potential participants were purposively selected. The physician, however, randomly invited women on the clinic register to participate in the study. Over a period of three months only five participants consented to join the study.

It was decided that it was necessary to find alternative means of recruitment. Following approval for an amendment to the research protocol by the HREC, the peer support organisation in Australia, the LAM Australia Research Alliance (LARA), was contacted for their assistance. The women welcomed someone taking an interest in their rare condition. The aim and design of the study was presented to a meeting of members and they indicated their support. Recruitment then became a process of network sampling as members of LARA networked within the organisation to inform other members of the study. Through this process a further fourteen women consented to participate in the study. Nineteen participants in total were recruited to join the study over a period of eight months.

Although purposive sampling had not been possible, network sampling achieved the aim of recruiting participants with a broad range of experience of living with LAM. The nineteen women participating in the study ranged in age from 34 to 68 years and had been diagnosed with LAM from between 18 months and 16 years. They came from a range of cultural backgrounds including Australian [n=11], European [n=5], Asian [n=1], South Asian [n=1] and South American [n=1], and were at different stages of the illness. Five women were living with advanced disease and using oxygen therapy, two had received lung transplants,

and one had experienced respiratory failure. Nineteen participants were recruited as the recruitment aim of a diverse range of participants with varying and key aspects of illness experience had been achieved. This is consistent with life history research which provides “intensity sampling” whereby certain participants provide detailed information and insight into particular aspects of experience (Plummer 2001, p.133). According to Plummer (2001, p. 21), there have been many life history studies with a sample size as low as one, for example, *Wladek Wisniewski* (Thomas and Znaniecki 1958), *Stanley* (Shaw 1966), *Jane Fry* (Bogdan 1974) and *Mrs Abel* (Strauss and Glaser 1977). A sample size of 19 is considered large for life history research. In the context of this study the sample was robust as nineteen participants provided a full range of experience of living with LAM across the illness trajectory.

2.4 Data collection

Data collection took place over a period of 18 months between November 2010 and May 2012. It involved conducting two interviews with each participant as well as accessing their hospital medical records.

2.4.1 Medical record data

Biomedical data was extracted from the participants’ medical records to provide a factual background to their illness experiences. It tracked the biomedical progress of their illness and included physician assessments, objective measurements of their lung function, records of investigations such as scans and six-minute walk tests, blood tests, treatments, procedures and hospital admissions. Notes were taken by hand from the medical records and de-identified as they were written. As per Rosenthal’s (1993) method, this factual data allowed identification of which aspects of their illness the participants regarded as significant and chose to narrate in their life story. Ideally, medical records were accessed between the first and second interview. However, logistically, this was not always possible due to processes of gaining ethical approval from three additional hospitals and because of the long distance travel required in some cases. The medical records of some participants, of necessity, were accessed during the same trip in which their second interview was conducted.

2.4.2 The Interviews

The first interview, as per Rosenthal’s (1993) method, was an open style of interview. After informal, friendly, introductory conversation, the participant was invited to tell the story of her life in whatever way she chose. Looking interested and listening without interrupting,

except to give verbal signs of attentiveness and acknowledgement, allowed the woman to choose to narrate what she regarded were the significant aspects of her life story in her own time and manner. The participants were all articulate and spoke freely about their experiences.

In keeping with Gadamer's (2004) hermeneutic philosophy, this style of interviewing was totally focused on the participant and her perspective to maintain an openness to her truth. In Josselson's (1995, p.30) words, an "empathic stance" was adopted. According to Josselson, an attitude of empathy involves continuity and receptivity, allowing aspects of the other to "permeate" one's self to gain a clearer perception and deeper understanding of them. This enabled a rapport to be established with each of the participants. Josselson (1995) argued that the "empathic stance" is hermeneutic because, in the process of interpreting another, an empathic researcher is aware of the efforts of the person to make meaning as they interpret themselves in telling their story.

The intended areas of focus for the first interview were life leading up to diagnosis, the process of diagnosis and how life changed with diagnosis, and for the second interview, life now and the issues living with a rare disease (see Interview Schedule, Appendix 1.4). The narratives of participants, however, did not necessarily follow linear time or the planned interview schedule. As Rosenthal (1993) observed, characteristic of life history interviews, narratives often flowed in subjective time, moving back and forth between the past and present in the first interview. Each interview was reflected on immediately after and notes were written of initial impressions of its content, context and participant/researcher interaction.

The second interview, conducted later after further reflection on the first interview, allowed issues to be followed up and topics not covered in the first interview to be raised. It was also an opportunity to clarify initial interpretations of meaning derived from the first interview. The second interview, therefore, was semi-structured and conversational. Verbal prompts such as, "how did you feel about...?" and "can you tell me more about...?" were used to explore topics in greater depth. Sensitive issues, for example, sexuality, were often discussed in the second interview after rapport had been established and the participant could feel a sense of trust and comfort with me. Interviews lasted between 49 and 240 minutes, on average between 120 and 150 minutes each. All interviews were recorded and conducted without any issues. A minority of participants expressed emotions during their interviews. When this occurred, I offered to pause or stop the interview. All wished to

continue so I allowed them to freely express their emotions and take their time in resuming their narrative. A social worker was available to be contacted by phone for further support if needed but this service was not required by any of the participants. None became distressed and all were settled at the end of the interview. Two participants specifically expressed that having the opportunity to reflect on, and talk about, their experiences was therapeutic.

Two interviews were conducted with each of 16 participants and three participants were interviewed once. Carol, who was very mildly affected by LAM, felt she had covered her story in the first interview and declined a second interview. Deb lived interstate in an area where there had been extensive flooding and it was not possible to schedule a second interview in the time available. Anna was interviewed once as she had been interviewed two years previously for an earlier pilot study. When randomly sent an invitation by the hospital physician, she made contact and indicated she wished to meet again for this current study.

Interviews were conducted at a time and place convenient to each participant and where they felt comfortable. Twenty seven interviews were conducted in the homes of the participants. Two interviews took place in hotel rooms, two in a private office of a workplace, one in a closed office of a university, and one in a private room of a hospital clinic. Each of these settings was quiet and private. One interview was conducted in a shopping mall and one in a cafe. These were less satisfactory due to the less private nature of the settings and the level of background noise. Each interview was transcribed verbatim and de-identified.

2.4.3 Data collection and the challenge of a rare disease

The rarity of LAM provided challenges for data collection. In the context of a rare disease, five participants lived in NSW and fourteen participants were located throughout Australia in four other states. Interviews had to be arranged for a time that was convenient to the participants and when I was able to travel to the relevant locations. Many of the participants were working or had other responsibilities or commitments that influenced scheduling of the second interview. For these reasons, the interval between the first and second interviews varied from two to 14 months. The mean interval was 7.5 months.

Travelling long distances to five states of Australia to conduct 35 interviews was time consuming and costly. The medical record departments of four hospitals located in four different states were attended to access participants' medical records. While long distance

interviews could alternatively have been conducted by phone or Skype it was felt to be important to be personally present with each woman to gain a deeper understanding of her experiences. This facilitated the building of excellent rapport with each participant and rich data collection.

2.5 Data analysis

Data analysis was carried out in three stages. For each participant, the data was analysed in two steps, by constructing a biographical account of their life, and analysing their life story. Comparing and contrasting the individual life stories in the third stage of analysis revealed common themes and points of difference. Following cross case analysis, the collective life history was reconstructed in chronological order.

Firstly, for each participant, the biographical account was constructed to represent the objective facts of her life and illness trajectory. These were the facts documented in her medical record, including background and biomedical information, and those facts that were free of interpretation narrated in her life story. They were extracted from notes taken from the medical record and from her life story as told at the time of the interviews. This information was then constructed in chronological sequence to show the objective process of her illness trajectory (See Appendix 3 for an example of a biographical account).

The second stage of analysis, using Rosenthal's (1993, p.67) method, is the "thematic field" analysis. According to Rosenthal (1993, p.64), the narrated life story represents "a sequence of mutually interrelated themes" and the thematic field is defined as "the sum of events or situations presented in connection with the theme that form the background or horizon against which the theme stands out as the central focus". Rosenthal's method of analysis involves a complex process of text sequentialisation whereby small sequences of text are interpreted in order through the generation and testing of hypotheses.

Rosenthal's process of thematic field analysis was modified at this stage of the analysis for the following reasons. Firstly, Rosenthal's (1993, p.70) aim of the thematic field analysis was to "interpret the nature and function of the presentation in the interview and not the biographical experiences themselves". This was not the primary aim of this study which sought to understand the meaning of illness experience. Rosenthal's method was directed at understanding the meaning of wartime experiences during the National Socialist era in Germany. It was developed as a way to uncover and interpret meanings that may be hidden from a narrator's consciousness as a coping mechanism or because they wished to present

themselves differently from their past. In contrast, this study was focused on developing deeper levels of meaning rather than uncovering hidden agendas.

Secondly, Rosenthal's method of sequential analysis, concerned mainly with structure and representation, was very prescribed, rigid and difficult to undertake with a number of cases in the time allotted to a doctoral thesis conducted by a single researcher. It lacked flexibility and room for creativity. Similar concerns were expressed by Jones (2004, p.49) who modified the biographical interpretive approach in a study of identity and the informal care role by reducing strict adherence to text structure sequentialisation to allow a more creative approach to data analysis. King (2000), in a study of home-based caring in West Germany, also modified the method, noting that qualitative methods evolve and that the need to answer an individual research question may require modification of the research tools used. In line with these adaptations of the method, Rosenthal (1993, p.59) noted that "methodology and methods of reconstructing life histories...are being continuously developed". It was decided that the process of sequential analysis would not provide meaningful interpretations in the context of this current study. Rosenthal's method was therefore modified at this stage of the analysis.

The data, instead, was explored for themes to uncover participant meanings. Analysis focused on identifying stories within the whole transcript. Rosenthal (1993) emphasised that analysing stories in the context of the whole text and the whole life preserves the gestalt of the biography in the sense of the underlying structure of personal meaning which guided the selection of stories as narrated in each interview. This approach was holistic and interpretive and congruent with Rosenthal's method and the underlying hermeneutic philosophy of the study. It was inductive in that themes came directly from the data.

The data consisted of 35 transcripts of interviews (two each with 16 participants and one each with three participants) in hard copy and word document, and deidentified notes made from each participant's medical record in a hard copy to record objective facts such as symptoms, investigations, diagnosis and progress of the disease. Hard copies of the transcripts and handwritten notes were kept in an individual folder for each participant. The word document copies of the transcripts were imported into Scrivener writing software into an individual project file created for each participant. Folders were created within each participant's project file for the transcripts, codes, turning points, and developing themes. Individual codes (e.g. breathlessness, fatigue, relationships, family, work), turning points (e.g. being diagnosed, starting oxygen therapy, finishing work, birth/death in family, being

listed for transplant) and emerging themes (e.g. loss, uncertainty, isolation, acceptance) were entered as a separate document within the folder to record relevant quotes, notes and reflections. Projects were also created for turning points and themes analysed across the participants. Blank-paged scrap books were used for drawing mind maps free hand. These were used to plot and develop my interpretation of themes. Electronic mind maps were created in iMindmap software to present the final themes (see Figures 3.1, 4.1, 5.1 and 6.1).

The hard copy transcript was firstly read, reread and reflected upon. Words and phrases that seemed significant and provided insights were highlighted in the text. The structure of the text was examined to identify which segments of text were narrations or stories. As per Rosenthal (1993), these were identified as those segments of text, generally longer sequences, where the participant told a story and elaborated upon an experience or event. These were also highlighted in the transcript.

Secondly, the narrative sequences were analysed intact, paying attention to their context in relation to the whole text, their temporal ordering within the text and their relationship to the initially highlighted words and phrases. The sequences were read more deeply to search for emerging patterns of meaning or themes. This method kept me immersed in the individual life and the data within the transcript, avoided fracturing the data, and kept a sense of the whole narrative and the context of individual stories within it. It also enhanced intuitive and creative responses which, according to Plummer (2001) and Cole and Knowles (2001), are a feature of life history research.

In particular, turning points, critical events or epiphanies were identified. Cole and Knowles (2001) and Denzin (1989) described these as organising constructs by which fundamental changes in meaning are conveyed within a life story. Following Rosenthal's (1993) method, the biographical account was compared to the life story to correlate the objective medical perspective with each woman's personal views and to see which events she had chosen to present, in what sequence, and their relative significance.

The descriptive identification of themes was followed by a more interpretive analysis. This process was often iterative with deeper levels of interpretation emerging with reflection and writing over time. Interpretations remained grounded in the text and relevant quotes representing the themes were selected. Developing interpretations were checked and agreed upon within the supervisory team. Analysis of the life story was complete when no new themes emerged.

The third stage of analysis involved conducting analysis across cases after each life story had been analysed. Mind mapping was used to plot themes visually during analysis of individual life stories and across cases. This allowed relationships between themes to be identified and more abstract concepts and overarching themes developed. Areas of difference between cases were also analysed. In this way shared qualities of women's experiences of living with LAM as well as unique features could be presented. Mind maps also provided a visual representation of the interpreted themes of the collective life history following analysis (see Figures 3.1, 4.1, 5.1 and 6.1).

Analysis of the participants' life stories revealed common turning points representing times of significant life change and adjustment, themes conveying the meaning of each turning point, and how the rarity of LAM affected their experiences. The turning points marked the progression of their illness over time from onset to late stage, and included being diagnosed, commencing oxygen therapy, receiving a transplant and experiencing respiratory failure. Analysis also revealed individual turning points in the participants' lives, for one a lengthy hospital admission, and for others significant changes in their personal social circumstances, as well as the importance of the women's interactions with healthcare professionals in influencing their illness experience.

Following cross case analysis, the collective life history was constructed to provide an understanding of the participants' experiences of living with LAM over their life course. In this process the interpreted life stories were merged to present the women's lives and illness experiences in chronological order, showing common turning points and experiences, their themes, areas of difference, and how meaning changed over time.

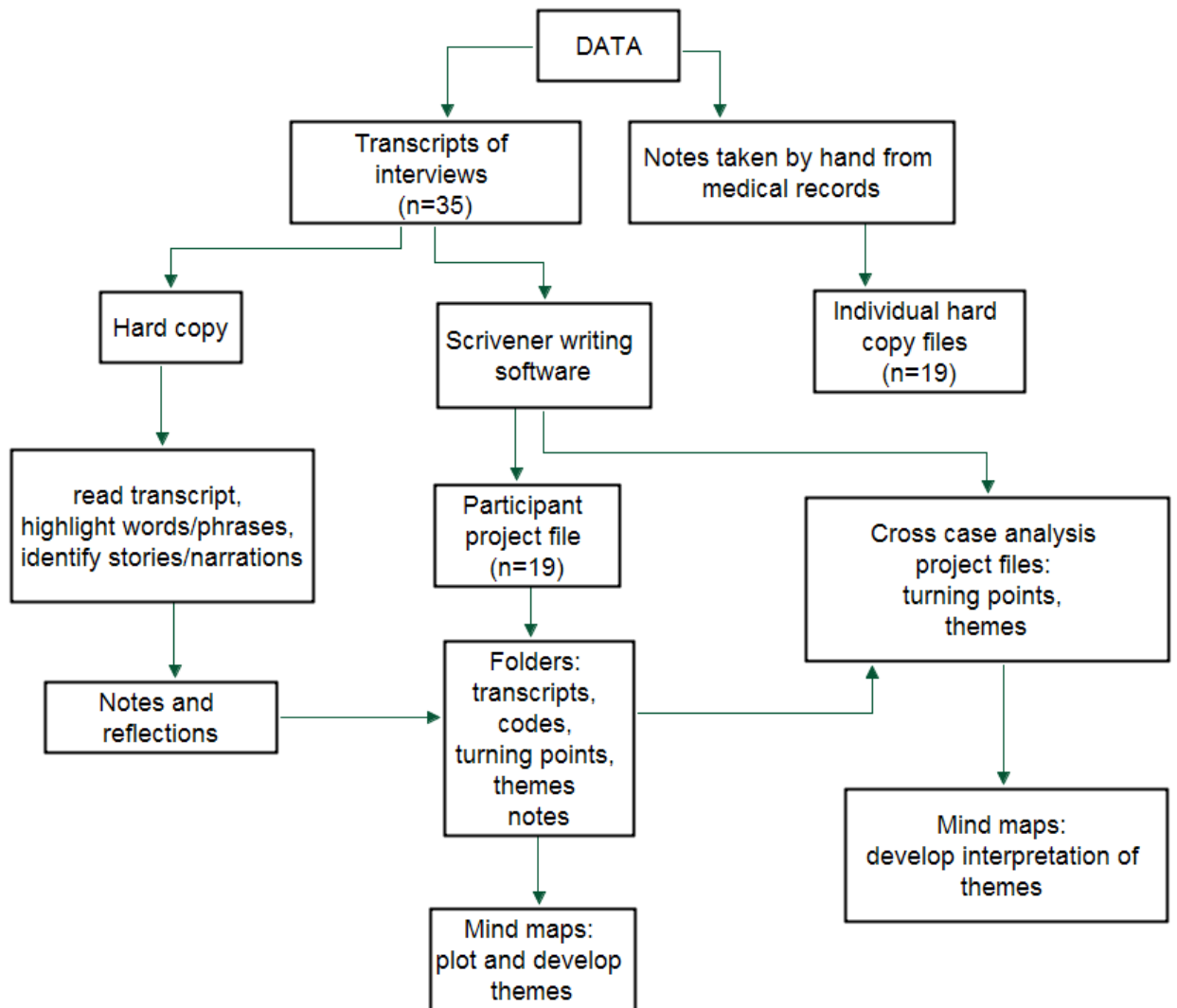


Figure 2.1 Data Analysis

2.6 Rigor

The trustworthiness of the study was ensured by applying a transparent framework and systematic approach to the research process, as described above. According to Meyrick (2006), this is a core principle of qualitative research quality. Variations were made to Rosenthal's method to best allow the research question to be answered. This accords with Popay et al.'s (1998) claim that flexibility in the design of the research with evidence of adapting it to the context of the study is a mark of quality research. The procedures, decision-making and interpretations were reflected upon, and discussed with the research supervisors. The use of mind mapping documented and demonstrated how interpretations were made.

Transcripts and interpretations were not returned to the participants. This decision was made in keeping with the underlying assumption of life history research that the life story told at the time of the interview is true for the narrator at that time and that later, meaning may change in the light of new experiences. Sandelowski (1993), Plummer (2001), and De Chesnay & Fisher (2015) similarly expressed this view. It is also consistent with Gadamer's (2004) hermeneutic view that meaning changes as a story is told and the horizon of the past is fused with the horizon of the present.

A key practice throughout the study was reflection and reflexivity. Reflections were recorded in field notes after each interview. Reflexivity, as described by Parahoo (2014), involved critically reflecting on my role as researcher, my background as a clinical nurse, my own values, beliefs, behaviours and presence, and how these might influence my interactions with each participant and interpretation of their stories. Reflexivity also included maintaining ethical awareness. A reflexive journal recorded reflections I made during the research.

Holloway (2005, p.276) stressed that the credibility of the research should be evident in its "truth-value" through interpretations that accurately reflect the participants' experiences. This research attempted to capture the participants' truth through the style of the interviews, the process of analysing the data, and presentation of the research. In the interviews, effort was made to be present in an open, empathic and friendly manner that privileged the participants' accounts of their lives. In the interviews with the participants I presented myself as a researcher. The participants were aware of my background as a practising clinical nurse but my role in interviewing them was as a researcher. While they did not

comment on this specifically, on reflection, I believe it may have created a level of comfort, openness and rapport as they narrated their stories to know that, as a nurse, I understood the biomedical aspects of their rare illness and they did not have to explain these to me, but also that, as a researcher, I was interested in knowing how their illness affected their lives beyond their physical symptoms. These factors and a prolonged period of engagement with the participants through two interviews and, within each interview, allowing them as much time as they needed to narrate their stories, enhanced the credibility of the study by enabling each woman time to reflect on the meaning of her experiences and express herself fully. Credibility was further strengthened by the process of analysis which involved long periods of immersion with the data. This allowed the time to take all aspects of the participants' lives into account, including the dynamic and changing nature of their subjective experience and meaning, and finding meaning in different as well as common experiences and so reach a deeper level of understanding.

The life history was constructed to allow each woman's voice to be heard and interpretations presented are supported by the participants' own words. Some participants are, of necessity, represented more often in the thesis due to their having experienced a wider range of aspects of living with LAM, for example, using oxygen therapy, experiencing respiratory failure, receiving a transplant, and undergoing an extended stay in hospital. This is in keeping with the aim of the study to explore a broad range of women's experiences of living with LAM and is consistent with the "intensity sampling" approach of life history research to provide comprehensive insight into certain aspects of experience (Plummer 2001, p. 133). It does not diminish the experiences of those with few symptoms or limitations and particular care was taken to ensure each participant's experience was included in the life history with at least one direct quote.

According to Polkinghorne (1995), Dollard's (1935) criteria for judging life history are relevant. As per Dollard (1935), the life history of this study encompassed the participants' social and cultural context, their embodied nature, their relationships, their choices and actions, and their history and past experiences. As claimed by Popay et al. (1998) and Streubert Speziale (2003), the potential for the research to be transferred to other situations was strengthened by articulating the findings of the study in relation to theory and previous knowledge and the implications for policy and practice.

2.7 Ethical considerations

Ethics approval for the study was granted by the HREC of St Vincent's Hospital in Sydney and was ratified by the ethics committee of the University of Sydney at which I am a doctoral candidate. All participants were over 18 years of age and spoke English. They received a Participant Information and Consent Form which informed them in a clear and easily understood manner of the nature, aims and process of the research project, and that they were able to withdraw consent at any time without any penalty and without their care being affected in any way. Participants freely gave their consent to be interviewed, for their interviews to be audiotaped and transcribed, and to have their medical records accessed. (See Appendix 1 for ethics documents)

During the recruitment process, a complaint was made to the HREC by a physician who was concerned that patients attending the public hospital clinic, who they considered to be under their care, had been recruited to the study without their prior approval. A second concern was that participation in the life history study would interfere with a concurrent clinical trial in which some patients were also participating. The matter was resolved following HREC review by a change to the research protocol whereby, within the hospital, individual physicians would be notified of the study and, at their own discretion, would choose to invite potential participants to join the study. Women who were willing to participate in the study were asked to contact the researcher to arrange mutually convenient times and places to obtain informed consent and conduct the interviews. An independent third party was preferred to sign the consent form as a witness to the signature of the participant. The majority of participants met me alone so a third party was not available to witness the consent. In this case the HREC approved that a file note could be made to that effect.

When participant consents had been received, ethical approval to access medical records also had to be sought from three other hospitals attended by some participants in Victoria, Western Australia, and Queensland. All provided access to the relevant medical records on the basis of the consent given by the participants and the ethics approval granted by the HREC of St. Vincent's.

Audiotapes and transcripts were available only to myself and my two university supervisors. During the research process data was stored on a password protected computer. Hard copies of the transcripts were stored in a locked desk in a locked room at the university. Data from the medical records was stored with the transcripts. At the completion of the study the research data will be deleted from the computer and audio recordings and transcripts will be

then located to a locked cupboard in a university office where they will be stored for seven years and then destroyed.

An ethical consideration in conducting research with a small group in the context of a rare disease is that some participants were known to each other and particular care had to be taken with de-identification of the data while at the same time preserving the meaning of each participant's narrative. This was achieved through the use of pseudonyms and changing or generalising identifying details such as nationality (e.g. Asian rather than specific nationality), and location (e.g. urban or regional rather than a specific state or city). The range of the participants' occupations are revealed in Chapter 4 but are not attributed to specific women. Particular caution was taken when discussing sensitive topics such as sexual relations and contraception. In this case the participants' quotes were not identified by their pseudonyms to further ensure their privacy.

These steps to preserve the participants' privacy were taken throughout the thesis, including the brief introductory biographies of each participant given below. As this is a life history study concerning how women experience living with LAM in the setting of their whole life, the biographies were needed to give context to the participants' experiences and the concerns they expressed and assist understanding of the interpreted themes within the collective life history. Further, the interpretations themselves will be presented collectively for future publication rather than as complete individual life histories so that individual lives are not identifiable. The privacy of the participants' physicians was also considered. Both male and female physicians are referred to as 'he' to avoid identifying particular physicians.

2.8 Introducing the participants

The participants are introduced in the following brief biographies which provide a factual account of their illness. These are summaries of the biographical accounts constructed from the participants' medical records and their narratives. Their subjective interpretations of their experiences are presented in the next four chapters. Summaries of the participants' social and medical backgrounds are provided in Appendix 3, Tables 3.1-3.4. Some participants were prescribed sirolimus and others everolimus for treatment of LAM. Both medications are mTOR inhibitors and have similar effects. For ease of reading sirolimus is the term used in this thesis for this class of medication.

Aiko is a 64 year old Asian woman. She is an only child. Her parents died within weeks of each other shortly before she was diagnosed. Aiko is married to an 88 year old Caucasian

man with multiple health issues including early cognitive impairment and limited mobility. She lived with her husband and cared for him until, after organising home nursing care for him, she moved to an interstate city to support her daughter and grandchild after her son-in-law's sudden death between our first and second interviews. Aiko was university educated and worked fulltime until she was diagnosed. Her interests include walking, yoga, and painting. She was fit and healthy and experienced only minor breathlessness for a number of years prior to being diagnosed. She was diagnosed when she was 52 following an open lung biopsy after an episode of pneumonia. She was found to have renal angiomyolipomas. Ten years after her diagnosis she was prescribed home oxygen to be used at night and when needed. She is now retired and was well at the time of interview.

Anna is a 37 year old Australian woman. Her parents had migrated from Eastern Europe. She has two sisters. She was married two years before she was diagnosed and divorced three years after her diagnosis. She currently lives in an urban area with her partner of six years. She is university educated and works fulltime. She enjoys walking, fashion, and sewing. She was fit and healthy and played various sports before she experienced her first pneumothorax when she was 23, the year she was married. Over the next two years she had further lung collapses. She was diagnosed at 25 following a lung biopsy taken during pleurodesis surgery. Six years after being diagnosed she was coughing up blood every day and had developed an abdominal lymphangiomyoma. This resolved after treatment with sirolimus. She experienced a major episode of depression and anxiety eight years after her diagnosis. This resolved with cognitive behaviour therapy and anti-depressants. At the time of interview she had been well and stable for three years.

Carol is a 66 year old Australian woman. She is a widow and lives alone in an urban area. She has two adult children and two grandchildren. She worked four days a week until she retired when she was 64. She currently does volunteer work. Her interests include walking, yoga and singing. She has osteoarthritis of the knee but is otherwise fit and healthy. She was diagnosed when she was 57 following a chest CT to investigate minor breathlessness (initially misdiagnosed as asthma) and a small pneumothorax. Carol has mild LAM and has been well and stable in the ten years since she was diagnosed.

Clare is a 34 year old Australian woman. She grew up on a rural property with her parents and brother. She lived with her partner in an urban area at the time of the first interview. In between the interviews she separated from her partner and was living with her parents at the time of the second interview. She has diploma level education. Her interests include walking

and painting. She had travelled extensively and worked overseas in a variety of jobs, and was fit and healthy and played various sports before the onset of LAM. She started to experience exhaustion and breathlessness when she was 31 and reduced her work to part time. She was misdiagnosed as having a chest infection. Her symptoms did not resolve and she was diagnosed a year later when she was 32 following a chest CT, and advised she would need a transplant in one to two years. Two months after her diagnosis she was found to have a chylous effusion and spent the following five months in hospital when the effusions persisted. The effusions finally resolved after she was treated with sirolimus. She only used oxygen at home for walking in the initial period after her discharge. At the time of the second interview she was well, working one day a week and investigating other possibilities for work.

Deb is a 52 year old Australian woman. Her parents are deceased and she has five siblings. She is divorced, has three adult children and lives alone in a regional town. She has professional training and works full time. She was fit and healthy before the onset of LAM. She enjoys being fit and outdoor activities such as camping, hiking, biking and gardening. Deb experienced breathless and chest pain walking uphill when she was 48. Her symptoms were thought to be cardiac in origin. She was diagnosed one year later following a lung biopsy. She is treated with doxycycline. She has experienced a gradual decrease in her exercise tolerance since her diagnosis but has otherwise been well and stable.

Eva is a 38 year old Eastern European woman. She came to Australia as a refugee fleeing civil war in her own country when she was 19. She lives with her husband in an urban area. Her mother and brother also live in Australia. She has Masters level university education and works fulltime. Her interests include meditation, yoga, and walking. She was fit and healthy and enjoyed dancing before the onset of LAM. She began to experience occasional chest pain and breathlessness, and a reduction in her exercise tolerance when she was 31. It was felt to be nerve pain. One year later she experienced acute breathlessness and panic during a plane flight. A chest X-ray showed pneumothoraces in both lungs. Bilateral pleurodeses were performed. A lung biopsy provided a diagnosis of LAM. Her lung function and exercise tolerance deteriorated significantly over the next two years. She was also found to have extensive abdominal lymphangioliomyomas. After she commenced sirolimus her symptoms and lung function markedly improved. She has been well and stable in the five years since then.

Helen is a 40 year old woman, born in Australia to European parents. She lived most of her life overseas. Her father died when she was 25. Her mother and brother still live overseas. Helen lives with her partner and two preschool aged children in a city suburb. She is university educated and has been working part time, two days per week, since her diagnosis. She was fit and healthy and enjoyed running, gym, dancing, swimming, yoga, and horse riding before the onset of LAM. She experienced breathlessness during both of her pregnancies. It was attributed to anxiety and depression. Her breathlessness continued after the birth of her second baby when she was 39. Three months later she was found to have a low oxygen saturation when she was taken to hospital with severe back pain. She was diagnosed with advanced LAM following a CT scan and commenced on continuous oxygen therapy and sirolimus medication. She was advised she would need a lung transplant soon and has had a pre-transplant assessment. She continues to work part time and enjoys going to the gym and bike riding.

Irena is a 47 year old woman, born in Australia to European parents. She has one older sister. She lives in a city suburb with her husband and adult stepchild. She is university educated and worked full time professionally until she retired when she was 45 due to her lung disease. Before the onset of LAM, she had been treated for some specific medical issues but was fit and healthy and enjoyed dancing and walking. She was diagnosed when she was 37 following a chest CT to investigate the haemoptysis she had had for six months. She experienced ongoing haemoptysis and progressive breathlessness walking uphill over the following five years and was prescribed home oxygen for walking. One year later her exercise tolerance had deteriorated further and she was found to have an abdominal lymphangiomyoma. It reduced in size when she started sirolimus. When she independently stopped her sirolimus after one year her lung function deteriorated and her abdominal mass increased in size. Two years later, when she restarted her sirolimus, her exercise tolerance improved, her haemoptysis resolved, and her lung function stabilised.

Jess is a 41 year old Australian woman. Her biological father (now deceased) and mother separated when she was four years old. Her mother remarried and later separated from Jess's stepfather when she was 16. Jess has been estranged from her mother for two years. She lives with her partner in an urban area. Jess has an Honours level university education. She works full time. She was fit and healthy before the onset of LAM and enjoyed gym, bike riding and walking. Jess experienced a sudden onset of abdominal bloating when she was 31. She was treated for a gut infection. Her symptoms did not improve and were attributed

to psychosomatic causes. Four years later, she again experienced abdominal bloating, accompanied by pain and fatigue, with her period. It was attributed to her appendix and later endometriosis. Twelve months later she was taken to hospital for severe abdominal pain. She was diagnosed with LAM following an abdominal ultrasound and lung biopsy. Her abdominal mass did not respond to treatment with radiotherapy and doxycycline over the next two years. Eight years after her initial onset of symptoms she experienced sudden breathlessness, chest pain and cough and was found to have a chylous effusion. A pleurodesis was performed. Some months later she commenced sirolimus after consulting a second physician. Within twelve months her abdominal mass had resolved, her lung function had improved and she was well and stable.

Julie is a 46 year old Australian woman. Her parents and sister live interstate. She lives with her husband and two adolescent children in a small country town. Julie has a college education and works part time. She smoked from when she was 18 until 41. She was otherwise fit and healthy and enjoyed a wide variety of sports before she was diagnosed. Julie first experienced chest and back pain due to a pneumothorax when she was 25. Over the following twelve months she experienced multiple pneumothoraces and ongoing shoulder pain, and underwent a pleurodesis. A CT scan showed small cysts but her pain was attributed to psychological causes. Six years later when she was 32, during her second pregnancy, a second pleurodesis and lung biopsy were performed after another pneumothorax. She experienced ongoing pain for another four years and then breathlessness on steps and hills. Three years later after an attack of severe bronchitis, she was found to have reduced lung capacity and was misdiagnosed as emphysema. She was finally diagnosed with LAM when she was 41 after a CT scan. A partial nephrectomy was performed for a renal angiomyolipoma. It was discovered that the lung biopsy taken nine years earlier had indicated LAM but the results had not been communicated. She commenced sirolimus when she was 45 after her breathlessness increased and her lung function deteriorated significantly. Although she experienced side effects from the sirolimus, within four months her exercise tolerance and lung function had improved. She was well and stable at the time of interview.

Louise is a 57 year old Australian woman. Her parents are deceased and she has two older siblings. She is university educated and had worked fulltime. At the time of her second interview she was not working after a change in the management of the organisation she was working for. She lives with her partner in an urban area and has two adult stepchildren.

She was fit and healthy before the onset of LAM. Her interests include art, reading, pets, gardening and walking. Louise first experienced intermittent haemoptysis, back pain and breathlessness while swimming when she was 38. She was diagnosed with LAM five years later when she was 43 following a CT scan. Since diagnosis she has been well and stable and felt a general improvement in wellbeing, exercise tolerance and lung function when she lost 10kg in weight as a result of diet and exercise.

Margaret is a 45 year old woman, born in Europe. Her family moved to Australia when she was seven years old. She has one older sister. Her parents separated when she was 16. Her mother since remarried. Margaret has a high school education and worked fulltime until she retired from work when she was 38 due to the progression of her illness four years after she was diagnosed. After finishing work, she took on the voluntary role of caring for dog rescue puppies for six years. She is single and lives alone with her two pet dogs in public housing accommodation. She has lived with asthma since she was three years old. She smoked for twenty years until she was 37. She enjoys reading, art, crafts, music, meditation and caring for her dogs. Margaret's symptoms started when she was 27 and developed gradually. She experienced breathlessness, abdominal pain, wheeze, haemoptysis, chest and renal pain over seven years. She was diagnosed with LAM when she was 34 following a CT of her chest and abdomen which showed lung cysts and multiple renal angiomyolipomas. Margaret's LAM progressed slowly over the next seven years until she required continuous oxygen therapy when she was 41. According to her medical record, when she was 43 she was noted to be unsuitable for a lung transplant due to "multi organ system problems". Two years later she was admitted to hospital for respiratory failure. She had decided she did not wish to be resuscitated if a life threatening situation arose. Multiple medical emergency calls were made for her respiratory distress and anxiety. At home she uses a motorised scooter to move about the house.

Mia is a 38 year old South American woman who migrated to Australia with her husband when she was 31. Her parents are separated. Her mother lives in Australia and her father overseas. She lives with her husband and adopted child in an urban area. She is university educated and worked full time until after she adopted her son when she changed to part time after a period of maternity leave. She enjoys gym, travel, art and photography. Mia has a long history of asthma but no other significant medical history. She sought medical advice when she was 34 for breathlessness on stairs and a persistent cough. She was diagnosed with LAM following a CT scan. Mia and her husband decided to apply for adoption when she

was diagnosed. After a long process over four years they were advised they could collect their baby from overseas. Mia needed to be accompanied by a doctor and use oxygen on the plane flight. Six years after she was diagnosed Mia's lung function had deteriorated and, although her physician had advised her to commence sirolimus and be assessed for transplant, Mia had not started sirolimus and had cancelled two appointments with the transplant physician. She was well at the time of interview.

Patricia is a 62 year old Australian woman. Her mother is alive and father deceased. She has two siblings. Patricia lives with her husband in an urban area and has two adult children. Patricia has professional training. Her husband travelled overseas for work until his recent retirement and she worked two days a week when her husband was home. She is currently not working. She enjoys exercise, particularly walking, gym, ten pin bowling, yoga and swimming. Patricia has been hearing impaired since birth and is treated for epilepsy. She first became aware of her breathing when she was 25 and needed to stop while jogging. When she was 32 a tumour that was bleeding was removed from her kidney but not investigated further. Eight years later, when she was 40, her three year old son was diagnosed with tuberous sclerosis autism spectrum disorder and epilepsy. Patricia was herself diagnosed with tuberous sclerosis at the same time. Eleven years later, when she was 51, she experienced breathlessness going up stairs and was diagnosed with LAM after a CT scan. She started using oxygen intermittently six months after she was diagnosed and continued to work one day a week for another five years. In this period she placed her son in residential care when his behavioural problems became difficult to manage. Patricia was listed for transplant two years after she stopped work. She was using oxygen continuously by this time. She continued to exercise throughout her illness. Patricia received a lung transplant one year after being listed. She recovered without complications. Four months after transplant she had recommenced bowling and bike riding and was competing in sporting competitions after twelve months. At the time of interviews she was three years post-transplant and well and stable.

Ruth is a 64 year old Australian woman. She lives in an urban area with her husband. She has two adult children and two grandchildren. She is university educated, works four days a week in her husband's business, and also does volunteer work. She enjoys walking, swimming and travel. She had asthma as a child, a hysterectomy when she was 53, breast cancer when she was 59, and osteoarthritis in her knee. Ruth's first symptom of LAM was the sudden, acute onset of severe left flank pain. She was diagnosed with a ruptured renal

angiomyolipoma. Four years later she experienced the same problem in her right kidney. In the same year, when she was 58, a routine chest CT following treatment for breast cancer revealed LAM. Her diagnosis was confirmed by a physician expert in LAM. Since then, Ruth's angiomyolipomas have been monitored and treated intermittently with embolisation. She experiences only mild breathlessness when walking quickly or uphill. She is well and stable.

Sarah is a 36 year old Australian woman. Her parents are alive and she has two siblings. She lives with her husband and two children, aged seven and five years, in an urban area. She is university educated and works part time. Before the onset of LAM she was fit and healthy and enjoyed exercising by walking, hiking, Pilates, tennis and the gym. Other interests included painting, travel and theatre. Sarah first experienced breathlessness when she was 29 during her second pregnancy two and a half years before her diagnosis. It was attributed to her pregnancy. After the birth she continued to become breathless when exercising, coughed frequently and had an episode of haemoptysis. She was misdiagnosed as being depressed and having asthma. After two further episodes of haemoptysis, a CXR showed atypical pneumonia. After not improving, she was diagnosed following a chest CT and lung biopsy. She was 32. She received doxycycline for six months but her lung function and exercise tolerance continued to deteriorate significantly in the twelve months after her diagnosis. She started sirolimus one year later. After four months her exercise tolerance and lung function had improved markedly, and her cough and haemoptysis had resolved. She uses a treadmill at home for exercise and although she did not fit the criteria for oxygen prescription, she purchased an oxygen concentrator to use while exercising on her treadmill. Eighteen months later she remains well and stable.

Ursula is a 51 year old Australian woman. Her parents are alive and she has three siblings. She lives with her husband and three adult children in an urban area. She is high school educated and works part time. She was diagnosed with asthma when she was 29 but was otherwise fit and healthy. She enjoys walking and painting. Her first symptom of LAM was sudden breathlessness due to a pneumothorax when she was 30. A pleurodesis was performed. Although multiple lung cysts were noted at the time, no biopsy was taken. Seven years later she experienced another pneumothorax. A repeat pleurodesis was performed and she was diagnosed from the lung biopsy taken at that time. Since diagnosis, Ursula's LAM has progressed very slowly. She is well and stable and has good exercise tolerance.

Veronica is a 68 year old Australian woman. She lives with her husband in an urban area.

She has two adult children and one grandchild. She has a graduate diploma level of education and worked full time until she retired when she was 62. She has a history of hypertension and anxiety but was otherwise fit and healthy before the onset of LAM. She enjoys walking, socialising, theatre, movies and cultural events. When she was 62, a routine CT scan following the removal of skin lesions showed a mass on her left kidney. Her kidney was removed for possible cancer. Pathology showed the growth was a benign renal angiomyolipoma. Later in the same year she experienced severe breathlessness walking uphill and was found to have chylous effusions in both her lungs. A pleurodesis was performed on each lung. She was diagnosed at this time. Since her surgery, Veronica has remained stable and has good exercise tolerance.

Vidu is a 48 year old South Asian woman. Her parents are alive and live in her home country. She has three siblings who also live overseas. She lives with her husband and adult child in an urban area. She is university educated and enjoys walking, yoga and dancing. Vidu and her family lived in different countries due to her husband's work. She travelled frequently to visit her family in South Asia. When she 31, on a visit to her family, she developed sudden back pain due to pneumonia and a small pneumothorax. The following year she became breathless and fatigued walking uphill and had a second pneumothorax. She was misdiagnosed as asthma. She was diagnosed three years later, when she was 36, after a CT scan to investigate her symptoms. Vidu's lung function gradually deteriorated over the next ten years and she developed an abdominal lymphangiomyoma. At this stage she was using oxygen continuously and, later that year, was listed for transplant. A few months later Vidu was admitted to hospital twice for a pneumothorax and pleural effusion. Vidu received a lung transplant one month later, eleven years after she was diagnosed. She was 47. Her recovery was delayed by chylothoraces, a pneumothorax, and side effects from her medications. She was well and stable at the time of interview twelve months later.

2.9 Conclusion

This chapter has presented the life history methodology employed by this study to explore women's experiences of living with LAM over their life course. Firstly, the methodology was discussed in relation to Gadamer's hermeneutic philosophy and Rosenthal's (1993) biographic interpretive method of narrative analysis which provided a theoretical framework for the study. This was followed by discussion of the research process, including participant recruitment, data collection and analysis, construction of the collective life history, rigor,

and ethical considerations. Rosenthal's method was modified to analyse the participants' life story narratives and decision-making concerning this process was explained. Individual brief biographies and factual accounts of their illness, summarising the biographical accounts, introduced the participants.

The following four chapters will present the collective life history of the participants that was constructed following cross case analysis of their life stories. It is focused on the period from the onset of their symptoms to the late stage of LAM. The structure of the thesis represents the chronological sequence of the life history as follows: Chapter 3, Being Diagnosed; Chapter 4, Life after Diagnosis and Learning to Live with LAM; Chapter 5, Interacting with Healthcare Professionals; Chapter 6, Living with Advanced LAM and Experiences of Oxygen Therapy, Respiratory Failure and Lung Transplant. Each chapter includes a figure of a mind map to provide a visual representation of the themes of that chapter as a guide for the reader.

In Chapter 7, the findings of the thesis are examined in the light of existing literature and theories to extend the analysis and link the interpretations of the participants' experiences to broader issues and more general knowledge. The final chapter of the thesis discusses the implications of the findings for practice and future research, reflects on the life history method used, and presents a personal reflection on the research process and concluding remarks. The following chapter will reveal the participants' experiences of being diagnosed, from the onset of their symptoms to the immediate period after.

Chapter 3

BEING DIAGNOSED

3.1 Introduction

This chapter presents the first of four chapters of the collective life history of the participants which convey the meaning of their experiences of living with LAM across the illness trajectory. Meaning was reflected in their narratives as they prioritised their experiences and narrated at length issues they perceived to be important. They revealed how they experienced diagnosis, a long hospital admission [n=1], commencing oxygen therapy [n=7], developing respiratory failure [n=1], and lung transplantation [n=2]. These were turning points which involved significant life changes. The chapters present the participants' experiences at these turning points, in their everyday life after diagnosis, and in their interactions with healthcare professionals. They reveal how the meaning of their illness changed over time, and the process of adaptation and development of resilience which emerged as marked features of the participants' experiences.

Being diagnosed was the first turning point in their illness trajectory. Whilst a number of participants experienced variable symptoms some time before diagnosis, for most the onset of symptoms was gradual and, while they knew something was wrong, they did not associate the changes in their body with a serious condition. Being given a diagnosis not only labelled the disease but also made them aware of potential implications for their daily lives and the future. It was the formal identified point of separation from their familiar world of health and wellness to the unknown territory of illness.

This chapter presents the participants' experiences of being diagnosed and how these were affected by the rarity of LAM. Figure 3.1 displays the women's experiences from the onset of their symptoms when they became aware of changes in their bodies, through a period of knowing something else was wrong when they were misdiagnosed or treated and not investigated further, to the delivery of their diagnosis, its impact on them, and how they dealt with it. Table 3.2 (see Appendix 3) summarises the biomedical facts from onset of symptoms to diagnosis for each of the participants.

3.2 Becoming aware of a changed body

All nineteen participants described how they initially became aware of changes in their bodies. They felt a variety of changes consistent with the clinical variability characteristic of LAM. Nine women experienced a combination of symptoms including breathlessness, chest/back pain, haemoptysis, cough, wheeze, and fatigue. Ten women experienced isolated symptoms of breathlessness [n=4], chest pain/pneumothorax [n=3], haemoptysis [n=1], flank pain/renal angiomyolipoma [n=1], and abdominal bloating and pain /lymphangioliomyoma [n=1].

Thirteen women described gradually becoming aware of being more breathless than usual when they were exercising at the gym, or walking uphill or upstairs. Louise became breathless while swimming and felt *“like a truck sitting on my chest”*. Helen, Sarah and Ursula all experienced breathlessness during, or following, pregnancy. Clare’s breathlessness was preceded by feelings of exhaustion and she felt she *“must be just really unfit because I’m not working as much”*. Her breathlessness was later found to be due to a collection of lymph fluid in the lung (chylothorax).

Two women described how their breathlessness had occurred suddenly and acutely with feelings of anxiety, panic, and impending loss of consciousness. Deb thought she was having a heart attack when her breathlessness was accompanied by chest pain. She described feeling like she was *“going to pass out because I really couldn’t breathe and I was really stressed”*. Eva experienced acute severe breathlessness that made her *“panic”* and feel she *“would lose consciousness”* while flying interstate. She was diagnosed a day later as having a pneumothorax in each lung.

Anna, Vidu and Julie initially felt chest and/or back pain only due to a pneumothorax. They developed breathlessness later over a period of years as their LAM progressed. Jess experienced abdominal bloating and pain due to a lymphangioliomyoma as a result of blockages in the lymph channels. She described her lymphangioliomyoma as *“a wine bladder in my stomach...With my period it was like something was expanding...It was extremely painful”*. Five years later she also experienced breathlessness and was subsequently diagnosed with a chylothorax. Ruth’s only symptom was *“excruciating pain”* in her lower back from a ruptured angiomyolipoma on her kidney.

3.3 Knowing something’s wrong

Ten participants sought medical advice when their symptoms of breathlessness, chest and

back pain, or abdominal pain persisted. These more generalised symptoms were commonly misdiagnosed and often attributed to psychosomatic causes. Six women continued to search for an answer for their symptoms for between two and 16 years, knowing something was physically wrong. Breathlessness was misdiagnosed as asthma [n=4], chronic bronchitis [n=1] emphysema [n=1], chest infection [n=1], anxiety [n=1] and depression [n=2]. Chest and back pain were misdiagnosed as cardiac pain [n=1], nerve pain [n=1] and psychosomatic pain [n=1]. Jess's abdominal bloating and pain was misdiagnosed on four occasions as having a gut infection, psychosomatic pain, appendicitis and endometriosis.

Helen and Sarah refused to accept a diagnosis of depression and anti-depressant medication when they experienced breathlessness during and after pregnancy:

I know myself really well. I'm not depressed, I just can't breathe. Something's wrong, I'm breathless... I didn't know what was happening. (Sarah)

After my baby was born I just suddenly started becoming really breathless all the time...So I kept going back to the doctor's and he just said it was anxiety... he sent me to counselling... He wanted to put me on anti-depressants but I just refused. (Helen)

Similarly, Julie's persistent chest pain and Jess's abdominal pain were attributed to psychosomatic causes and they also refused antidepressants. Each of these women knew they were not depressed and that something else was wrong.

Before 2001 LAM was a rare and poorly understood disease and consequently diagnosis was delayed when acute problems such as pneumothorax or ruptured angiomyolipoma were treated and not investigated further. Anna, Julie, Ursula and Vidu's pneumothoraces were managed with a drain or surgical pleurodesis without further investigation between 1990 and 1998. Similarly, a thoracic surgeon performing Ursula's pleurodesis in this period observed multiple small cysts on the lung but did not take a lung biopsy.

Ruth, Veronica and Patricia initially presented with renal tumours or bleeding from these tumours. Ruth was diagnosed correctly as having a ruptured renal angiomyolipoma but was found incidentally to have LAM four years later on a follow up CT scan after breast cancer. Veronica's renal tumour was misdiagnosed as cancer resulting in the unnecessary removal of her kidney. She was diagnosed with LAM when she developed breathlessness later in the same year and was found to have a chylothorax.

Julie searched for 16 years for an answer to her persistent chest pain following a left pleurodesis for multiple pneumothoraces. Her pain was medically diagnosed as psychosomatic. Seven years after the initial surgery, during her second pregnancy, a right pleurodesis was performed for a pneumothorax and a lung biopsy was taken. However, the results of the lung biopsy were never followed up and Julie continued to have ongoing pain. After four years she developed breathlessness climbing hills and steps and was misdiagnosed as having emphysema three years later. She was finally diagnosed with LAM following a chest CT 16 years after her initial surgery. At this time her specialist sought, and located, the results of the lung biopsy taken nine years earlier and discovered that LAM had been reported at that time. Julie expressed strong feelings of shock and anger that the results had not been communicated to her and her diagnosis subsequently delayed. A medical opinion was sought from an interstate respiratory specialist who implied that Julie had not attended her follow up appointment and suggested that a diagnosis would not have changed the course of her illness. Julie felt victimised and that she was being blamed for the failure in communication:

It had actually been diagnosed then and nobody had told me... I was shocked...I was so angry...Then they wrote to someone interstate, some professor, about LAM and the treatment in the hospital. That was like getting a slap in the face...They'd even put in it, if you had been told basically it wouldn't have changed anything because it's an incurable disease. Do you think when you're pregnant was a good time to know? I just thought, that's not for you to judge at all...I just felt that everyone was blaming me.

In contrast, nine women were diagnosed correctly in twelve months or less from the onset of their first symptom of LAM. Of these, four women were diagnosed between 1998 and 2001 and five women after 2001. In Patricia's case, her initial diagnosis of tuberous sclerosis was made eight years after bleeding and removal of a tumour from her kidney. Eleven years following her diagnosis of tuberous sclerosis, she was diagnosed with pulmonary LAM within twelve months of developing her first respiratory symptom of breathlessness between 1998 and 2001. The correct diagnoses of LAM received by the women within twelve months occurred after the creation of the LAM Foundation in 1995 and associated database in 1997, and subsequent acceleration of scientific research into the condition (McCormack 2008; Tattersfield & Glassberg 2006). Respiratory specialists were the most likely physicians to access this knowledge as evidenced by the majority of correct diagnoses made by this group.

GPs (local doctors) and gynaecologists misdiagnosed participants' symptoms on twelve occasions.

Julie's experience and that of Mia, who was misdiagnosed as having asthma and bronchitis for 15 years, raise the question of whether receiving a diagnosis of a rare incurable condition at a young age is desirable given the generally slowly progressive nature of LAM. Julie would have been 32 years old if she had received her diagnosis when her biopsy was taken. She was already married and expecting her second child. Mia first experienced breathlessness when she was 19, unmarried and without children and had not established a career. The two women expressed different opinions regarding early diagnosis. Julie was adamant that it was her right to receive a correct diagnosis regardless of her stage of life. Mia, on the other hand, preferred a later diagnosis. Had she been diagnosed in her early twenties the limitations of having LAM would have meant that she would have been unable to pursue her career and its required overseas travel which she loved. She felt it may also have impacted on her ability to partner and marry:

(If) they tell you that in your 20's and you start thinking you might die... and can't have babies...you bring that baggage to a relationship... In my 20's it would have totally ruined my life. I don't think I would have married (my husband), I would have to rethink my career too early. It would have been horrible.

While Helen was angry that her breathlessness had been wrongly attributed to anxiety after her first pregnancy and not investigated further, she also acknowledged that if she had been diagnosed at that point she would have been advised not to risk pregnancy again and would not have had her second child:

I suppose there's two sides to it, you know. If I had been diagnosed after my first baby I wouldn't have been able to have my daughter and my husband was of the opinion that I probably wouldn't have been this bad because definitely the pregnancies exacerbated the whole situation.

These stories reveal the complex decision making that may be involved in investigating and diagnosing a rare incurable disease with uncertain course, and the ability to compromise lifestyle and impact on a woman's plans for pregnancy. They suggest a need for full discussion to understand a woman's life goals and priorities during the process of investigating and diagnosing LAM.

3.4 Life disrupted by diagnosis

Receiving a diagnosis of LAM was unexpected and devastating news for the majority of participants. Most had been fit and healthy with no significant past medical history before the onset of their symptoms. Deb, for example, had *“always been fit... hiking up mountains and... biking”*. Sarah *“was always a very active person...walking five or six times a week and doing weights classes”*. They did not associate their symptoms with a serious condition and did not expect to be told they had a rare, incurable, progressive lung disease they had never heard of. According to Sarah, *“there was not supposed to be something wrong”*. Aiko *“wasn’t expecting anything like this”* and *“could not believe it happened”*.

Each participant held a personal life meaning which they had accumulated over the course of their life. The women appraised their illness against their life meaning to determine the significance of the changes they were experiencing in their bodies, and the implications of their diagnosis for their future. Their life meaning encompassed their sense of self, values, beliefs, disposition, past experiences, present and future expectations, sense of life purpose, and life goals. It was shaped by their family, cultural and societal values, life experiences, and social interactions in the world. Each participant’s life meaning represented her beliefs about herself, the world, and her relatedness to the world. It was the structure of meaning which orientated her and gave her life coherence. The participants experienced levels of disruption which reflected their life meaning and the varied ways they were affected by LAM.

Participants who were only mildly affected, with minimal impact on their daily life, experienced minor disruption. Others were able to reconcile their diagnosis with the meaning of their current expectations and other life experiences. Ruth and Carol were only mildly affected by LAM, were older and already had children. Ruth was 58 when diagnosed. She had only experienced symptoms twice when renal angiomyolipomas ruptured and caused severe back and flank pain. A routine chest CT following treatment for breast cancer had shown abnormalities in her lungs. Ruth felt her diagnosis provided her with a logical explanation for her angiomyolipomas and was consistent with her expectation that she would have developed cancer during her life. She had recovered from her cancer treatment, felt well and took a positive approach to her diagnosis:

It made the angiomyolipoma fall into place... I really hadn't had anything to make me feel that my lungs were impaired... I wasn't surprised... since I'd just

been diagnosed with and treated for breast cancer which I'd fully expected would be my lot at some stage... I'm quite positive about making the most of life... I think I'd be in a different situation if my lungs were deteriorating fast...for me there's a bit of a holding pattern.

Carol was diagnosed at 57 with very mild LAM that it was predicted would cause her little problem in her life. She was well and her only symptom was a minor inconvenience of some mild breathlessness while talking. Diagnosis, while a shock, gave her an explanation for her problem:

It was a bit of a shock, but it was good to know what the problem was. I've only had probably maybe a dozen instances when I couldn't finish a sentence and I know what it is... as long as it doesn't affect me anymore that it does now, I'll be quite happy.

Ursula was diagnosed when she was 37 following a pneumothorax and pleurodesis. It was the same day that a well-known young celebrity died suddenly and tragically. She felt fortunate that she “*was still alive... we didn't feel like I was going to die in ten years because I didn't feel that bad*”.

Fifteen participants experienced diagnosis as a major disruption of their life meaning. Thirteen of these were younger women aged between 25 and 41 years old when they were diagnosed. This period was described by many as “*one of the hardest times*” and a “*struggle*” to deal with. They were told their illness was incurable. Ten women were given expected lifespans of between one and ten years, and five were told to prepare for a lung transplant from “*fairly soon*” to within ten years. This dramatic and unexpected news disrupted their expected future and life plans of having children, work, leisure and travel, and, in turn, their sense of self and being in the world

The women felt a sense of shock, uncertainty, and loss, with strong emotions of grief, sadness, fear and anger. Helen described being diagnosed as “*a huge loss and grief*”. Anna remarked that “*what happens emotionally and mentally to anyone, to me in particular, is far worse than what's going on physically*”. The rarity of LAM heightened the disruption of their diagnosis by creating feelings of isolation and loneliness. Being diagnosed with LAM constituted a turning point in life for these women because it was a point of all-encompassing change in body, mind, emotions, spirit, and social relations.

3.4.1 Feeling shell shocked

Many participants expressed the significance of receiving a diagnosis of LAM through their use of metaphor. Images of war conveyed the unexpectedness of their diagnosis, their initial feelings of shock and devastation, and the impact of LAM as an all-encompassing disruption of their familiar worlds. Anna “*was very angry and shell shocked and almost devastated...I literally felt like my whole world fell apart*”. Eva was “*so shocked... It was very difficult first of all to think... everything is turned upside down*”.

The sense of “*shell shock*” and trauma also affected partners and children. Deb’s teenage daughters were “*shell shocked*”, having only seen their mother as fit and healthy. Sarah’s whole family, including her husband, two children and parents, “*went into shell shock... and just shut everything out and that was very disturbing*”. For Sarah’s husband, her diagnosis “*was the biggest thing that happened to him*”:

It’s very traumatic to be told that your wife’s got a condition that’s going to mean that she’s not probably going to be around...We were going along with our lives and really everything was great - and someone put a bomb under him... this destroyed his life.

Diagnosis was an extreme disruption for Helen and her family. When she was diagnosed she was a 39 year old mother with a twenty month old child and a three month old baby who she was still breastfeeding. Her breathlessness during each of her pregnancies had been attributed to anxiety. She was diagnosed after being admitted to hospital with severe back pain and found to have a low oxygen saturation of 88%. A chest CT revealed advanced LAM. Helen’s life was disrupted in terms of both her future and her present life. She was told she would now need to use oxygen continuously, would have to wean her baby, would require a lung transplant soon, and was given an expected lifespan of two to five years. She described the impact of her diagnosis:

My partner just burst into tears...When he came the next day he'd obviously been crying all night, he looked terrible...They said you're going to be on oxygen now all the time. I had gone from fit and active to being on oxygen. They said you're going to have to stop breastfeeding because we're putting you on to sirolimus. I was just devastated...A double lung transplant is the only option and that will probably be fairly soon.

3.4.2 Uncertainty - the ‘not knowing’ of being diagnosed with a rare disease

Being diagnosed with a rare disease created a high level of uncertainty. The participants did ‘not know’ what this disease was, how it would progress, how long they would live, what their life would be like, if and when they would need a transplant, and what that would mean for them. Most were given their diagnosis by physicians who had either not heard of or knew very little about LAM. The manner in which many women were delivered their diagnosis added to its negative impact and exacerbated their uncertainty. Eleven women were given their diagnosis alone without support. Of these, Eva, Ursula and Aiko were told late at night while in hospital. Eva and Ursula were recovering from surgery. Louise and Sarah received their diagnosis alone by phone. Twelve women indicated they received minimum information and fifteen women a lack of emotional support from their physicians who spent only a brief time delivering the diagnosis. No explanation or discussion occurred about the possible progress of the illness. Helen’s doctor “*spent two seconds with me just saying we’re looking at lung transplant*”.

Prior to 1998 there was no information available to the participants at all apart from a limited number of published medical articles. Louise was told, “*there is no cure so just go and enjoy your life if possible*”. Margaret was told only, “*it’s just a disease that women get and you’re going to die from it...you’ll be dead by fifty*”. Anna’s physician said to her, “*You know my colleague’s daughter had this and she was gone in five years and she was about your age*”.

Louise and Deb were given the name of the disease but advised initially not to read more about it. Jess was given the name lymphangiomyomatosis by a specialist physician but spelt incorrectly so that neither she nor her GP could find information about the condition. Shortly after, by chance, she viewed a documentary on television featuring the stories of two women with LAM and recognised the similarities with her own story. She was distressed to work out her diagnosis in this way:

I just happened to watch Australian Story... it was saying, this woman has this incredibly rare lung disease...Then they said the name and it just turned out that the doctor had misspelt it and that's why we couldn't look it up... I was absolutely, absolutely shocked watching that program. It was really the worse way to find out.

Most of the participants, or their families, referred to the internet and commonly read outdated information about survival rates of between one and ten years. They were generally

not referred to reputable websites with up to date information such as the LAM Foundation. Even in the later period, 2005-2010, Helen, Clare and Eva were given little information and were not referred to LAM peer support groups. Helen was “*just left...we had nothing*”. Eva described how her physicians handed her information taken from the internet:

The diagnosis was really bad...It's 10 o'clock (at night), the day after my surgery...I was by myself... He decided to tell me that here's a sheet of paper that I found on the Internet. It's some information for you. I know nothing about this disease but you'd better start preparing for a lung transplant because you will need one within five years. That's all he told me.

The manner in which participants were delivered their diagnosis created feelings of distress, confusion, and fear. The women who had children didn't know whether they would survive to see their children grow up. Most women, having already experienced symptoms for some time, assumed the worst of a dramatically reduced lifespan. Louise felt “*my life's ending ...I just thought I'm doomed*”. Irena conveyed her distress:

It rings in my mind all the time when he said, you've got between one and ten years of life...That really upset me obviously. I just thought I'm too young to die and obviously I focused on the one instead of the ten or in between. I thought, well, is there anything that I can do, anything I can take? Apparently nothing.

There was an added layer of uncertainty and anxiety for women who were told to prepare for a lung transplant. This was particularly the case for Helen who was told she would need a transplant soon. She conveyed the uncertainty, fear and anxiety surrounding the process and timing of transplant in her war metaphor of “*ticking time bomb*”:

That's the worst thing, this ticking time bomb... I don't know what's going to happen tomorrow...There's no end date. It's not like I know that in four months' time I'm going to have a lung transplant. It could be four years, it could be 14 months, it could be four months. It's just that whole ticking - you always have that little anxiety in your head... what's going to come?

Uncertainty was reduced for participants who received their diagnosis and adequate information in a manner which suited their individual personality and needs. Sarah, Ruth and Mia consulted LAM experts (respiratory physicians with a special interest in LAM) at diagnosis, Margaret and Anna later after diagnosis. LAM experts provided the women with

reassurance and accurate, up to date information. Margaret was relieved to find out that she could hope to live for longer than she had originally been told:

He was the best and he did say... yes, there is no cure for this disease but you know some people can die quite young, some can live for more years... People get told once they've been diagnosed they've got ten years and he goes, that's a myth.

Deb felt she was given adequate, honest information and explanations and appreciated structured meetings with relevant healthcare professionals present and inclusion, with her family, in clinical decision making:

He (my physician) came back and sat down and explained the whole thing to me... (He) organised that the head of the heart-lung transplant unit... a social worker, a psychologist... were there... and brought my brother and the girls down... He had all the x-ray scans, blood results and just explained... so then they had a debate whether I should have a thoracotomy.

Vidu was a South Asian woman who received her diagnosis from her respiratory specialist of the same cultural background. She felt the information given to her was adequate. She was discouraged from seeking further information or discussing her condition with others. She was rather encouraged to lead her life as normal. Vidu felt that her doctor knew her as a person and that his advice was personally and culturally appropriate for her:

He explained everything... He told me one thing, don't talk about this with your friends... because I don't want anyone to treat you as a patient. I want you to live your life. I want you to forget about this... you have to be positive. You have to fight it. He was just giving me moral support... Lead the same life as you did before... He knows that I'm a bit of a weak person... He knew about me... He didn't want me to have contact with other LAM patients because... knowing you, if you hear about the dark side of it you will be totally negative... He is also a Buddhist so... according to our religion... he advised me... For me... I think it's good.

3.4.3 Layers of loss

The participants revealed the disruption of multiple aspects of life in the layers of loss which they experienced at diagnosis. At this turning point most focused on the future, in the loss of their expected lifespan and plans for family, work and travel. Younger women, who were

advised not to become pregnant once they were diagnosed, particularly felt a sense of loss of their life goal of having children and seeing them grow up. This was felt acutely by both women who had not yet had children and those who had. The forced loss of such a fundamental aspect of female identity struck at the core of their sense of self and many found this the most difficult aspect of diagnosis to deal with. Margaret, 34 when diagnosed, single and without children, spoke with emotion of her loss:

I actually cried more when I got told basically don't have kids than when... (my doctor) explained to me about the death ratio... All I ever wanted to do was get married and have children and that was my life's hope... and then to get told, no don't have kids... I was angry, I was really angry. (Quivery, teary voice)

Sarah, 32 when diagnosed, was married and had two children. She had “*just assumed that I'd have more than two kids*”. She and her husband struggled for a year with decision making around IVF, surrogacy and adoption:

At the time my lung function was degenerating very quickly and I... realised the IVF surrogacy process would have been overwhelming... I thought I really had better focus on my kids... Someone said to me I could adopt... It was a debate between us... We just decided it was all too much, the emotions of the whole thing would have been too much.

Irena conveyed the combined shock, shattering of life's plans and struggle in her use of ‘*battling*’ as a war metaphor in confronting her illness and the issue of not having her own child:

Do I allow this child to grow up without a mother? Do I allow my husband to have the burden on his own of raising this child? ...I just thought I was really selfish wanting a child so badly. Then I thought I was selfish not going ahead with it in the hope that I won't get worse... I was toying and battling with it.

Even if having a child was not a primary aim, diagnosis and the subsequent inability to have children represented the loss of choice and possibility as an aspect of self and normal life. While Jess was “*not really one of those women who always wanted to have a kid*”, not being able to become pregnant became “*part of not really having a real life.... it was part of not knowing if you'll be able to walk in five years or will you be able to work*”. Similarly, although Louise was 41 and had not had children when she was diagnosed, she felt that

“part of my life hasn't been expressed and I feel a bit incomplete”.

Anna and Eva reflected on the possibility of adoption and dealt with it by considering their current priorities, how they would care for a child in the context of possible deteriorating health, whether their partner would be able to care for a child on his own, and the fairness of potentially leaving a child either without a mother or as a carer for their mother. Anna prioritised her health, and Eva her marriage and Buddhist faith. Eva was able to separate her female identity from being a mother, *“I don't identify being a mother necessarily with being a woman”*. The two women described their decision-making:

I have those moments where I think I'm missing out big time. Then I think if I was to bring a child into the world, I have bad days as it is, that's not fair on the child...and I don't think it's a child's place to be the carer. (Anna)

What if I start declining again? ... What's going to happen to this poor child? Is my husband ready to take care of it on his own?... I may have a limited time to what I once thought I would have to live. So do I want to use the time raising a child? ... It's not something that is at the top of my list. (Eva)

The necessity to avoid pregnancy was not a loss for all women. Five women were post-menopausal and already had adult children. Ursula's family was complete before she was diagnosed. Mia felt that having to avoid pregnancy *“wasn't a big deal”* as her life plan had included adopting a child.

Helen experienced both a loss of career opportunities and financial loss. Her disease was already advanced and she needed to reduce her work hours when she was diagnosed:

I have worked for ten years to get to a senior position. I really like my work and it's important to me...My partner and I had this great plan that suddenly we were going to have some money ... I'm on quite a good wage if I was working full time but that's now not happening...I have to now realise my career as such at this point is on hold.

Mia, Anna and Helen were advised not to undertake air travel due to the risk of pneumothorax. This represented the loss of an expected future when holiday plans they had made with their partners would not be realised. Mia's work involved frequent travel and, as well, she was now isolated from her family overseas. Mia reflected on this loss:

The other factor that was really shocking and that had a big impact in our lives was that I travelled a lot... Travelling is one of the things I love the most...we always wanted to travel together... Boats are really expensive and you really need to take three times more time off to get somewhere... My family was so far away. It was very hard because I knew that I couldn't go and visit them as often.

Louise and Aiko felt a loss of hope and a sense of life ending. They became acutely aware of their breathing and experienced anxiety and panic. At the time they were diagnosed (before 2001) having such a rare disease exacerbated their sense of hopelessness when there seemed no prospect of a cure. They felt engulfed by feelings of blackness, depression, fear, anger, loneliness and having no control over their situation. Louise said:

I started having panic attacks... I felt like I had to make myself breathe... I remember being on a tram one day and thinking I'm going to suffocate... I remember just feeling so bleak and alone. I just felt like I'd fallen down a black hole and there was just no hope, there was no one who could pull me out of it. I was just going to die... I just thought well, there's no hope really, is there? Absolutely no hope, because no one's going to research a disease that's so rare.

Aiko considered ending her own life. She used the metaphor of “corpse” to describe both her physical appearance and loss of will to live:

When I was in hospital, I had that not much choice, not much hope. Then, I became like depression, like panic attack. Suddenly I can't breathe... I was just so struggling. I was quite angry about everything... One time I was thinking, if I'm going to die, can I commit suicide? ...I lost so much weight....a corpse - just bone and skin...I was very depressed.

3.4.4 Feeling “so alone”

Receiving a diagnosis of a rare disease was isolating on a number of levels. As a rare disease there were so few other women with LAM, particularly in the Australian context. Before 1998 there was no support group in Australia and most physicians, even in later years, did not refer their patients to the LAM Foundation or the Australian organisation after it was formed. Being given the diagnosis alone, with little information or support, was a lonely experience. Louise “*felt so alone. It was just awful*”. Helen’s doctor “*wouldn't put me in touch with anybody and I said, well could you pass my number on and ask them if they would contact me, and he never did that...it was just really frustrating*”.

Geographically, many women living in other states or rural areas were isolated from the two centres of LAM expertise in Australia as well as from other women living with LAM. This was especially pertinent to Helen who lived a considerable distance from LAM experts, was unable to fly, and was further isolated by knowing of no other women on oxygen with young children with whom she could share her experiences.

Socially, a diagnosis of LAM was isolating when most people the participants met, including their own local doctors and other healthcare professionals, had no awareness or knowledge of LAM. Other than her partner, Jess “*didn't really have anybody who understands the disease*”. A woman’s particular social circumstances could exacerbate the sense of isolation. Aiko, for example, was particularly alone and isolated by her diagnosis. Her husband was elderly and in poor health and her only child was living overseas. In addition she was culturally isolated as an Asian woman living with an Australian husband. Her parents were recently deceased and she had no real contacts with a cultural community in Australia. She described herself as a cultural “*alien*”. These feelings were only accentuated by her diagnosis:

I feel really strange... (Asian) people think I'm not typical (Asian)... but I think Australian people don't believe I'm Australian... I feel like an alien.

Not belonging anywhere... I don't have really my own group...I have no... (Asian) contact at all...

The doctor came very late in the night...just the two of us... I didn't have any family...I have to face things by myself.

3.4.5 A shifting sense of self

When they received their diagnosis the participants entered a period of transition between separation from their old life and previous state of health, and passage to a new way of being. For six participants entering transition was an existential experience in which they reflected on their existence. As they perceived life itself becoming more tenuous and uncertain they experienced an unsettling, shifting sense of self. They had an awareness of separation and uncomfortable feelings of instability, ambiguity and paradox, of being in between states and neither one thing nor the other.

The women expressed a sense of disembodiment and separation of body and self. Anna spoke of her diagnosis as being “*like an out of body experience*”. The body took on an alien identity and seemed no longer a part of the self. Jess felt “*trapped*” in this foreign body. It

had become a burden, “*I felt like I was carrying my body around*”. The uncertainty of illness and the future created in Louise an “*emotional disconnection*” that made her feel less confident and impacted on her relationship with her partner. Aiko felt she had lost herself in her disease, “*I became LAM*”.

They experienced a similar shift in their relatedness of self to the world. There was a new world of illness, hospitals and healthcare professionals they would have to negotiate within their everyday world. While they remained situated in the world there was a sense of not being fully part of it. Eva felt that the limitations of her illness removed her from familiar life:

My life's going to be like this from now on. I'm going to be someone sitting in a chair when my friends are outside, dancing or going wherever they're going... All of a sudden, from feeling like you can do everything... these are my limits now and you have to deal with that. It's quite hard.

Feeling both in the world and yet at the same time outside of it engendered a sense of unreality. Helen described life at diagnosis as “*surreal*”. An uncertain future shifted life out of balance so that it seemed less tangible. There was a perception of instability, of moving between worlds. Eva “*felt so lost*”. For Louise “*nothing felt very solid any more*”. It was like being “*on a slippery slope*”. Jess felt she had entered a “*shadow life*” of uncertainty that was out of context with her old life:

Everything becomes de-contextualised, because I couldn't plan for a future. I didn't know - in three years will I be working? Will we be able to buy a flat? Will I still be able to be walk around? Will I be dependent on ... (my partner) for everything?

The concept of liminality reflects this subjective state of ambiguity and uncertainty which the women were experiencing. Liminality was first described by Van Gennep (1960) in an anthropological study of initiation rites of passage in tribal communities. He conceptualised it as a phase of ambiguity and being at the margin of normal life during a transition or passage from one age or social situation to another. According to Van Gennep (1960), there are three phases of transition: separation from the old life, a liminal period, and passage to a stable state during incorporation. All the participants demonstrated the phase of separation as they entered transition in this immediate period surrounding their diagnosis. The themes of disruption, loss, uncertainty and isolation which they revealed were indicative of the

unsettling, indeterminate and marginal quality of this period. The women above who experienced liminality conveyed it in their metaphors of instability and darkness.

Not all participants experienced a state of liminality during their transition. Women with mild disease for whom diagnosis was minimally disruptive were able to transition quickly to a more stable physical and emotional state and adapt to their illness. The participants' experiences of liminality after diagnosis and at later turning points are explored further in the next three chapters.

3.5 Dealing with the diagnosis

The participants dealt with the disruptive effects of their diagnosis by either actively engaging with their illness or intentionally avoiding it. Each approach, in different ways, was directed at restoring a sense of normality and control. The women made decisions that were consistent with their life meaning, including their disposition, past life experiences, self-efficacy and spiritual beliefs. Being able to mobilise social support assisted them in dealing with their diagnosis.

3.5.1 Engaging with the illness

Nine participants actively engaged with their illness, taking the first steps toward adapting to it in their lives, by conducting their own research, seeking expert medical care or contacting peer support groups to find information about LAM. Clare and Eva identified themselves as having curious dispositions. Clare described her personality as "*inquisitive*" and her "*way of coping was just to find out a lot*".

Margaret, Patricia and Eva had developed resilience from their past experiences of successfully managing adversity or illness over time. Resilience represented the knowledge, skills, competence and self-efficacy beliefs they had gained through these experiences. They took a positive approach and demonstrated confidence in their ability to cope with their diagnosis. While Margaret was "*scared, worried, confused*" when she received her diagnosis, she had lived with asthma all her life and "*always had to be careful so in a way being an asthmatic has really helped me with this disease, to cope with it, cause I've always had restrictions in my life*".

Eva had experienced civil war in her country before she came to Australia alone when she only 19. She had been through three bombings and a cousin had died on the war front. She found it a "*shocking*" experience. Eva's experience gave her courage by showing her that it

was possible to survive and move on from severe adversity:

In civil war, people go through horrible stuff... you cannot fathom that someone would actually be able to survive. But you do. People move on. Just seeing that transformation, and how strong people really are. I think that teaches you you've got that inside of you as well... So it gives you a bit of courage.

Patricia's son, who was 25 at the time of interviews, had been diagnosed with tuberous sclerosis and severe autism spectrum disorder and epilepsy when he was three years old. She herself was diagnosed with tuberous sclerosis and epilepsy at the same time, and had lived with a hearing impairment all her life. Patricia had been successfully managing her own health and her son's complex medical issues and challenging behaviours for many years. Her husband regularly travelled overseas for work. Patricia received and dealt with her diagnosis alone. Her ability to manage a difficult social and medical situation over a long period of time had developed her resilience and gave her the confidence she would be able to manage her LAM. She pragmatically accepted her diagnosis and adopted a positive, proactive approach to her illness by seeking expert medical care and continuing to exercise to keep fit for transplant. For Patricia, LAM was "just another step".

3.5.2 Striving to be 'normal'

Eleven participants who were in the early stages of LAM protected their sense of self by avoiding associating themselves with being ill or anything related to LAM. They wanted to feel and be perceived as the same normal person as before their illness. They avoided thinking, talking, and seeking information about LAM to keep the disease separate and distance themselves from its foreign and frightening implications. This provided a safe space to protect the self for a time. According to Jess:

At that time I wanted the same things to be important...you want to be the same person... I just wanted to really be in a bit of denial...There will be a time when I can't disguise it. But while I can, I'll just not mention it.

Veronica could not connect with her illness, "I had difficulty applying it to myself... I didn't relate it to myself". She coped with diagnosis in a way that was usual for her, "head in the sand, bum up in the air. I think that's probably how I handle it... I didn't want to read about it... It was very good self-talking, that it's not necessarily going to happen". Deb "didn't think about the disease... I just had too much to do... it was not going to be part of my life".

Irena “*wanted to shut myself off from anything LAM related*” to preserve her emotional stability:

I just didn't want to think about it. Because I thought well I'm going to die... why should I research this?... There's no cure, nothing's going to help me. I just want my emotional state to be at a level where I can cope.

Vidu understood herself to be a person prone to negative thoughts. Avoidance allowed her to protect herself from negativity and anxiety in a way that matched her personal disposition. She didn't discuss her illness with anyone outside her family, and avoided reading about LAM and contacting other women with the condition. She forgot her LAM by avoiding thinking or talking about it and keeping busy with her everyday life:

I thought it's better to lead my life like this, the same way that I lived my life before. I thought I will forget about it because actually I didn't have... much pain... I didn't feel that much.

Avoidance manifested for Louise as recklessness, drinking too much and living for the day, believing she was to die prematurely anyway. She saw herself as fighting LAM as an enemy:

I think I had a bit of a drinking problem... that I'm invincible kind of thing. I'm just going to have a good time...I think also I felt careless because I thought I was going to die anyway... it's also a way of denying the illness, you know it's not going to get me.

Avoiding being ill was a temporary response for the majority of these participants and they later engaged with their illness as their initial feelings of shock settled. Vidu, Irena and Veronica, however, continued to practise ongoing forms of avoidance in their life after diagnosis.

3.5.3 Finding support

Social support was a crucial means of coping with diagnosis. The majority of participants were supported by partners, husbands, parents, siblings and adult children. Family provided practical and emotional support, acted as advocates, sought further information on LAM, and contacted LAM peer support groups at the time of shock and disruption. Supporting each other was an important aspect of Anna and Vidu's family and cultural values, as Anna said, “*sticking together when things get hard*”. Vidu's family gave her advice and moral support. Her brother-in-law had taken responsibility in the role of eldest brother in her

family when she was young. When she was diagnosed he acted as Vidu's advocate by communicating with her parents and health professionals, and researching necessary information on her condition. "When I was in hospital he was there. He did everything".

Not all participants received support. Aiko, as highlighted earlier, was socially and culturally isolated and did not have access to social support. Clare essentially dealt with her situation alone. Although she was living with her parents, she received little emotional support. Her father was seriously ill at the time and she modified her own emotional response out of concern for her parents, trying to be strong and positive so as not to add to their worry:

The first month I was pretty much on my own... Dad was still quite critical ...Mum and dad had been through so much - right from the word go I was trying to be positive for them...I just had to deal with it... I was trying to be strong for mum and dad. I just thought well, what will be will be. I'll just try and do the best with it...I tried not to cry either because I just didn't want her to get upset.

The isolation Jess felt in having a rare disease was exacerbated by her estrangement from her family and feelings of being unsupported by them:

On these sorts of occasions the conflicts become more...I've never had much support from them (my family)...My father was overseas...he hasn't been there at the crucial bits...I haven't spoken to my mother for about two years.

Peer support groups organised by women with LAM themselves were a potential source of information and support for newly diagnosed women. The organisations which most participants had contact with were the US LAM Foundation, founded in 1995, and the Australian support organisations, LAM Australia (1998 - 2006) and the LAM Australia Research Alliance, LARA (from 2006). They could also communicate via social media on Facebook sites.

The participants expressed a range of views on their experiences and need for contact with other women with LAM or LAM websites. Eight women chose initially not to contact a peer support group. Each of these women had either mild disease or were in the early stages of their illness with manageable symptoms. They did not want to be reminded of their illness or be confronted by women at the later stages which could evoke uncomfortable images of the potential progress of their own LAM. Deb was busy getting on with her life and keeping

LAM backgrounded. She had support of good friends and didn't feel a need to make contact with other women. *"I can't be bothered with it, it's all just too maudlin. I just don't want to know about it"*. Later she occasionally looked at the Facebook site but tended to find it *"depressing"*. Irena preferred to deal with her diagnosis on her own. She felt looking at LAM websites and having contact with other women would foreground her illness and make her constantly relive it:

I didn't want to read things because every time I did I would read so-and-so passed away, this person has been diagnosed and she's on the transplant list. It just brought it all back home so I avoided the website as much as I could.

Julie, Jess, Mia and Ursula met older women with advanced disease and on oxygen in hospital clinics and when invited to their homes. For each woman it was a confronting, frightening and depressing experience and did not assist them in dealing with their own diagnosis. They wanted to feel connected to life and health rather than sickness and the end of life which they sensed in the older women. As Julie said:

It's easy for people to say you need to support each other, but it's also very scary when you go and see someone like that, because you're seeing a bit of the future. It's actually a horrible feeling.

They would have preferred to meet someone younger and managing their condition well who could give them a more positive view of the future and hope that some sense of normality in their life was possible. Ursula wanted to *"be like the one that's going slowly... you want to be like the one that survives not the one that dies from it"*. Mia would have been *"happy with someone younger or in my same sort of age group with the same sort of situation... a positive outlook as opposed to worst case scenario"*. Jess felt that being with the women grounded her in her illness at a stage when her own breathlessness was minimal:

She could barely walk. She would walk a little bit and puff a lot. I found that really quite horrifying... They didn't understand how normal I felt. I just didn't feel like them... I could walk around and not notice particularly being breathless... I was intellectually preparing for some of the symptoms, but I wasn't actually experiencing them.

Eight women elected to contact a peer support group for information when they were diagnosed. When Louise was diagnosed prior to 1998 the LAM Foundation had only just been started. A formal support structure was not in place. Louise felt alone and isolated, *"in*

a closet all by myself". The only information available to her was what she had gleaned from medical articles in a university library. Later, after LAM Australia started, Louise met other women with LAM. Although she feared meeting them would be confronting, she found it was "*wonderful*" and reduced her feelings of isolation.

As the internet became more widely available, the participants were able to make contact with peer support groups through their own or their family's search of the internet. Frequently the LAM Foundation was the first point of contact. By 1999 the LAM Foundation was well established and women were sent an information package and referred to the Australian support group. They could also register with both the US and Australian databases which provided data to support LAM research. Although Ursula had indicated she "*didn't want to meet anyone sick when I first got diagnosed*" her physician made contact with a woman with LAM through the Australian Lung Foundation and asked her to contact Ursula. Ursula was relieved that the woman was similar to herself, well and mildly affected. The interaction was helpful and a positive experience and the two women maintained ongoing contact.

Julie and Helen, both geographically isolated, felt that the emotional support of other women with LAM was fundamental in managing their diagnosis. Helen acknowledged that the support of women from LARA (LAM Australia Research Alliance) was "*how I got through it really*". For Julie, one particular woman had "*been the best thing. She's been my support through all this*".

When women supported each other they could form relationships which lessened feelings of isolation and were encouraging and sustaining. However, because all were living with a life threatening condition, forming relationships had the potential to add to the losses experienced at diagnosis. This was compounded by the fact that, because there were so few women with the rare condition in Australia, one or two women only tended to take on the role of providing personal phone communication with newly diagnosed women. A member of LAM Australia provided crucial emotional support for Margaret and Aiko. Margaret spoke of becoming "*close*" to her and Aiko's spirits were lifted by her support. She "*cheered me up so much. Hearing from her, I thought it's just like bright light. The depression gone*". The relationship, however, came with risk of loss and this was realised when the woman who had provided such valuable support passed away from complications following lung transplant. Her death was "*devastating*" to both Margaret and Aiko and represented loss of friendship and support, confrontation of the risk of transplant, and a view of their own

potential future. In the context of a rare disease and the solo nature of the woman's support work there was not awareness among other women of Margaret and Aiko's emotional loss at this time. Consequently Aiko withdrew from contact with other women with LAM for nine years and Margaret maintained only very occasional contact. Aiko feared "*losing them again, and how do I cope with that?*"

In recent years social media and email communication have become convenient ways for women diagnosed with LAM to overcome the isolation of a rare disease by connecting with a broader range of women both within their own country and internationally to share information and experiences and create a sense of community. 'Lammies' is a closed Facebook group providing a private forum for discussion. Through this online community participants gained access to new information and first hand experiences of new treatments such as sirolimus. This assisted with decision making in regard to their own illness management and gave women the confidence to discuss the possibility of treatments with their physicians. Social media was a safe forum to discuss common personal issues such as sexual intimacy and contraception that were difficult to raise with physicians during limited appointment times. There was also an immediacy to social media so that a woman could express her feelings and receive support from women who understood when she was having a bad day. Helen, who knew of no one else in Australia caring for young children while living with oxygen therapy, particularly found Facebook helpful in connecting with other women overseas who were in a similar situation to herself:

I got on to Facebook Lammies... and that was brilliant because there were people my age or younger so I didn't think I was the only person with kids that had LAM...It's a really good site for asking questions and finding out about stuff... (When) I'm actually having a really shit day ... I need to offload... I need some support...That's really good because they were, Helen, that's fine; just come on; you'll be all right; you've just got to work through this; tomorrow you'll feel better.

Eva, Vidu and Anna drew on their personal belief systems for support during the period of diagnosis. Spiritual beliefs provided meaning and comfort at a time of suffering, direction for coping with their diagnosis, and a link to their familiar life before diagnosis. Eva had been practising meditation and had adopted the Buddhist faith two years before she was diagnosed. She credited her spiritual practice with giving her tools to enable her to cope with her diagnosis. In turn she felt her illness deepened her faith:

People that are in trouble always seek religion because it's comforting. It was no different for me... I certainly did turn to it consciously to try and make it a tool for me to deal with this better. It certainly did help. I intensified my practice because I was sick. If I wasn't sick, I probably would have been halfway on the path now.

Vidu was also Buddhist. At diagnosis, receiving her diagnosis and advice on managing her illness from a Buddhist doctor in a manner that fitted with her spiritual and cultural beliefs supported her coping. Anna's Catholic faith, an important part of her family and cultural life, acted as an "anchor" and a "safety blanket" when she was diagnosed. She found some peace in the turmoil of diagnosis through her practice of going to Mass, prayer and the rosary.

3.5.4 Deciding to conceal or disclose the diagnosis

At diagnosis LAM was an invisible illness to outsiders for the majority of participants. Although they had a serious lung condition most appeared well and healthy. Only Helen, who was prescribed oxygen therapy when she was diagnosed, bore visible signs of LAM with her nasal prongs and oxygen cylinders. The invisibility of LAM meant that participants could choose with whom they would share news of their diagnosis. While this gave them some sense of control it could also mean that people could lack understanding of the seriousness of their condition and display less empathy. This created a tension between inner fragility and an outward appearance of normality, between a need for understanding and a fear of being treated differently. According to Aiko, "*the problem is we don't look bad at all although inside we are so fragile*".

The participants did not want to be identified as a sick person or be regarded as a victim to be pitied. This was a prime motivating factor in concealing their illness from people they met. Most women chose to discuss their diagnosis only with partners, family and selected friends. As Julie said:

I don't want people to feel sorry for me. I don't want to be treated differently... So I don't like everyone to know. My friends know, my work colleagues know, but I don't run around telling everyone I've got LAM.

Looking healthy and concealing their LAM backgrounded their illness and was consistent with the coping mechanism of avoidance used by some of the women. Irena, Deb and Carol "*didn't talk much to anybody*", preferring to manage on their own and avoid worrying loved

ones.

Concealing LAM was a means of avoiding stigma and being judged as less than competent because of their illness, especially in the workplace. This was a particular concern for Jess who worked full time and was concerned about how her work performance would be assessed and her prospects for future work assignments:

I've always been afraid that I won't get interesting work and that I'll just be treated as a bit of a basket case if I make too much fuss over the condition...you don't want to be judged on having something wrong with you in the future.

Other women were selective in revealing their diagnosis at work to a few trusted colleagues or their manager only, when they felt it was necessary. This might occur when there was an atmosphere of trust or when they felt there was an obvious issue which required explanation. In Mia's view:

When it's noticeable... then I feel the need of explaining it so that they realise that it's something that could be permanent...When there's a certain relationship of trust I feel okay to tell them.

Other reasons for concealing their illness included maintaining privacy and avoiding the inevitable explanations of LAM required when revealing a rare disease to others. It was difficult to explain the condition casually to acquaintances. Often others confused LAM with asthma and Sarah and Jess found it easier not to correct that misconception. Sarah said:

I just feel awkward having a conversation with someone in a casual way...A lot of people...want it to be asthma because they understand asthma...So you pretty much agree with them...I just can't be bothered to go into an explanation.

There was also a sense that people were generally not interested in the illness or its associated problems. Patricia felt that “*some people, you can talk about it to, and other people, you just leave it on the backburner... people don't want you to be talking about your problems*”.

Participants avoided lengthy explanations by referring people to the LAM Foundation or LARA website for information and providing an information sheet about LAM. Irena's

main motivation in discussing her illness was to raise awareness of LAM in the community. Anna only shared thoughts about her illness if she felt it was of benefit to someone who might also be experiencing difficulties in their life. Eva, Ruth and Ursula felt comfortable in revealing their diagnosis to others. Ursula did not regard herself as a private person. Certain social contexts could hinder a woman from concealing her illness, for example, Clare lived in a small rural town where news of her illness was readily shared in the close-knit community. *“It was a small town everybody knew I was sick....so there's no point hiding it”*.

3.6 Summary

The participants' narratives revealed that being diagnosed with a rare, degenerative, incurable illness was a critical turning point of significant change for their lives. Receiving a diagnosis, for the majority of participants, was a shock which profoundly disrupted their lives due to the absence of a cure, the limited lifespan most were given, the uncertainty of the rate at which their illness would progress, and the subsequent impact on their daily life and expected life plans. In this period, in the context of being given a predicted maximum life expectancy of ten years, the women particularly focused on their future and the losses which this implied. For younger women, the loss of their ability to choose to have children and, for those who already had children, the possibility that they may not live to see their children grow up was an especially devastating loss and disruption of their sense of self. The shock, uncertainty, and loss which the women experienced was accompanied by strong emotions of grief, sadness, fear, anxiety and anger.

The rarity of LAM compounded the participants' sense of disruption by exacerbating the uncertainty surrounding diagnosis and creating a sense of isolation. The lack of awareness and knowledge of LAM among physicians delayed diagnosis and affected the way it was delivered. Most participants were given their diagnosis alone, often in an insensitive manner, and provided with minimum and, at times, inaccurate information and no emotional support. Most were not referred to peer support organisations. This increased the negative impact of the diagnosis itself. The women consequently had to rely on themselves or their families to access generally outdated information from the internet and eventually locate support from the peer support organisations. These findings are reflected in the results of two surveys of the experiences of almost 12,000 rare disease patients conducted by the European Organisation for Rare Diseases (EURORDIS) between 2003 and 2008 (EURORDIS 2009), a study of the experiences of Australian families living with a rare

disease (Anderson et al. 2013), and Carel's (2013b) experience of being diagnosed with LAM.

Diagnosis marked entry to a period of transition between separation from familiar life and adaptation to a new way of being. Entering transition was, for some participants, an existential state of liminality in which they were aware of a shifting sense of self and feelings of disconnection and instability, of being in the world but simultaneously outside of it. Martin-McDonald and Biernoff (2002) also utilised Van Gennep's (1960) rites of passage to explore the meaning of dialysis dependency and conceptualised transition as a liminal state of space, time and place.

The participants revealed that they interpreted being diagnosed through the lens of their personal life meaning representing their values, beliefs, life purpose and goals. Other authors have referred to this as global meaning (Park and Folkman, 1997), biographical construct (Rosenthal 1993), life scheme (Thompson and Janigian 1988), and assumptive world (Murray Parkes 1971). The participants experienced disruption as they perceived a discrepancy between their life meaning and the implications of their diagnosis for their future. Similarly, Park and Folkman (1997) claimed that people interpret a stressful event by looking for congruence between global meaning and the situational meaning of a particular experience. They proposed that a perception of incongruence is associated with discontinuity and provides a motivation to reconcile the meanings in order to restore order and coherence.

The participants strove to restore a sense of normality by either by engaging with their illness or avoiding it. Similarly, Bury (1982) claimed that the biographical disruption of chronic illness forces a person to evaluate their life plans and self-concept and attempt to normalise. The women demonstrated that resilience some had developed as a result of knowledge and self-efficacy gained through past experiences of adversity and illness reduced the disruption of diagnosis.

Family support, and, for some, personal spirituality and religious beliefs, were crucial resources which assisted the participants to cope at this time. While a number of participants sought support from the peer support organisations, others avoided associating with other women with LAM to further preserve a sense of normality and avoid being confronted by oxygen therapy and experiences of advanced disease. They preferred support that was positive, hopeful and focused on health and wellness. Communication with other women

through online communities of women living with LAM was a valuable support for participants who were particularly isolated, geographically and socially.

3.7 Conclusion

This chapter has presented the participants experiences of being diagnosed with LAM. It covered the time span from the onset of their symptoms, an interval of misdiagnosis or delayed diagnosis, through to their diagnosis and the period immediately following it. For the majority of participants, being diagnosed signified a profound disruption of their life meaning, conveyed in feelings of shock, loss, fear, grief and anger. The rarity of LAM added dimensions of isolation and uncertainty. Many were given their diagnosis in an insensitive manner, and the majority received little information or emotional support from their physicians.

Later, in their everyday lives after diagnosis, the participants learned to live with the changes associated with their illness and integrate them into their lives. Resilience developed through these processes and influenced their experiences at later turning points. The following chapter presents the participants' experiences of adapting to their illness in their everyday lives after diagnosis and the growth of their resilience during this period.

Chapter 4

LIFE AFTER DIAGNOSIS

ADAPTING AND LEARNING TO LIVE WITH LAM

4.1 Introduction

This chapter presents the participants' experiences following diagnosis to the moderate stage of their illness as they learned to live with LAM in their everyday world. The previous chapter revealed that, for the majority of women, being diagnosed with LAM was a turning point of profound disruption which was compounded by the rarity of their condition. When most of the women were given a predicted maximum lifespan of ten years they focused on their future and how their illness would impact on their life plans. Diagnosis marked their entry to a period of transition during which the participants' focus shifted to the task of learning to adjust to its impact on their everyday lives. This was a dynamic process of adaptation which enabled them to passage from transition to a new way of being as their illness became an accepted and integrated part of their lives. Through this process they developed resilience which supported them in living with the permanent uncertainty of a progressive, incurable illness. Resilience represented the meaning, knowledge and skills that evolved throughout their lives and their experiences of learning to live with LAM.

The participants' experiences after diagnosis were embedded in their social worlds. While they identified the physical effects of their illness, their narratives were concerned primarily with the personal and social impact of LAM on their lives. Many indicated that these were more difficult to deal with than their physical symptoms. Adaptation involved learning to manage their symptoms, emotions, and social relations in the everyday fluctuations of life as well as times of illness-related or significant social change. Figure 4.1 guides the reader visually through these processes. The participants revealed how they learned to live with their changed bodies and manage their relationships and responsibilities and how these were influenced by the rarity of LAM. In living with illness in a changing world they encountered new turning points and varying experiences of liminality which affected their ability to adapt. They demonstrated how they built their resilience through adaptation and the first stage of illness progression.

4.2 Learning to live with a changed body

Following diagnosis, the participants needed to learn to live with their changed bodies and acquire knowledge and skills to manage these changes over time. This involved their day to day experience of living with their illness symptoms and the limitations they imposed, as well as learning to understand and manage their rare disease. Breathlessness and fatigue were the most common symptoms of LAM which affected the majority of the participants, limiting many of their usual activities. The women regarded breathlessness as the most serious physical effect of LAM. They perceived it to be a sign of the extent of their lung disease and associated it with mortality, Being breathless could be a frightening sensation associated with anxiety and panic, particularly in the early stages of the illness when they were not familiar with how to manage it, and again at the late stage when it became more persistent. When feeling breathless their illness was in the foreground of their minds and they found it difficult to focus on wellness. According to Eva:

Breathlessness is something that is quite a scary sensation. Because it's like... dying with every breath... you can't help thinking about just the disease. It becomes your whole focus... it makes it very hard to break out of feeling helpless and unwell and really get a strong sense of wellbeing.

The women described feeling breathless when they walked up a hill or stairs or at a fast pace; performed actions such as pushing, bending over, carrying a weight or talking; were in hot or cold environments; and at night. This impeded their ability to perform home chores such as vacuuming, sweeping, mopping, and hanging out washing; leisure activities such as sports, swimming, hiking, and dancing; and other everyday activities such as work and shopping. They felt socially isolated and became observers rather than participants. This occurred when they were not included in certain activities or left behind when they could not maintain the faster pace of their friends or join in conversations while walking. Julie “used to love snorkelling. I can’t snorkel any more...I just have to watch now and take the photos”. Aiko found she *started slipping out from the group*”. Sarah said:

I stopped being able to walk with anyone because I stopped being able to walk and talk... I got upset... I used to try and exercise every day and so that was a bit of an impact. A lot of the people I used to see in that way I stopped being able to.

At interview, most of the participants appeared well, even though they had a serious lung

condition. Other people tended to assume that their breathlessness was caused by a lack of fitness or lung problems due to smoking. Ursula felt that when she walked upstairs at work “they must hear me puffing. They probably just think I’m unfit”. In Sarah’s view:

People make a real judgment. The judgment is you're young, your breathing is not good. You must have been a heavy smoker and you've caused yourself damage... I get quite offended by it. But I know that it's irrational...I didn't say anything.

The majority of participants reported that fatigue interfered with their daily lives. Whilst they did not regard it as being as serious as their breathlessness, fatigue was more persistent and difficult to manage. They could objectively monitor their breathlessness by measuring their oxygen saturation levels but fatigue, in contrast, was highly subjective and could not be measured. The women often experienced breathlessness and fatigue together and found it hard to distinguish whether their fatigue was a product of their breathlessness, an effect of their illness on their bodies, or due to ageing or the daily demands of caring for children and working. Sarah said:

I think I'm just generally a bit more tired... I just think sometimes you can't even distinguish what's what. How can you tell? ... I used to be someone that could really burn the candle at both ends and make it all work - and now I just can't...I don't know if that's just because I'm getting older.

Vidu associated being “tired” with her breathlessness and described this as her main symptom of LAM throughout her illness. She became progressively more “tired” as her illness progressed and interfered with her daily activities, *I think due to breathlessness I got tired... when I was so tired I used the oxygen.*

Fatigue was Jess’s predominant symptom after her abdominal lymphangioliomyoma resolved following treatment with sirolimus. Her low energy levels interfered with her ability to engage in activities and concentrate at work:

Fatigue probably affects me more... it's frustrating and I wish that I didn't sleep so much or feel so tired. I wish I had more energy... Not find everything so much of a struggle...One of the things that stops me exercising isn't really my lung capacity, it's my fatigue levels... the fatigue is the biggest obstacle.

Anna’s fatigue was associated with anxiety and depression which she experienced eight years after diagnosis on the anniversary of her marriage breakup. It corresponded to a

progressive decline in her lung function but was not related to specific episodes of breathlessness. She felt that her fatigue had become so unrelenting that it permeated her whole self:

It's restrictive, it's draining and I get frustrated...it makes you feel a bit down...I just want to feel ok and get that energy back...it's almost like a monotone, the energy level in your body... I'm tired, I'm tired, I'm tired, and you know it feels like it becomes everything or it becomes you.

Fatigue was rarely noted as an issue in the participants' medical records and was not addressed by healthcare professionals. It was not mentioned at all in Vidu's and Anna's medical records and only once in Jess's record but not at the time when it had the greatest impact on her life. Jess's physician dismissed her fatigue when she attempted to raise the issue with him:

I was trying to get him (her doctor) to understand how tired I am. I said, sometimes I just come home from work and I go to bed at 7 o'clock. I just have to. He said, half your luck.

The subjective and invisible nature of fatigue meant that family and friends could also lack understanding of its impact, particularly when participants looked otherwise healthy. Sarah's husband was "not so sure he believes me, he says everyone's tired." Louise felt that:

I look well, you know, I don't look skinny and I don't get breathless sitting here and so people don't understand if you feel tired...I think tiredness can often lead to a bit of depression too because you just feel flat, you know, and you don't understand why.

Sirolimus itself could contribute to fatigue as a side effect of the medication. Helen felt she was "exhausted all the time because of the immunosuppressants." Julie remarked:

I don't feel particularly well on it (sirolimus)... I started to sleep heaps more on my days off... I put the kids on the bus and I go back to sleep. That might be eight o'clock. I wake up at one o'clock in the afternoon.

Breathlessness and fatigue affected the majority of participants on a daily basis, limiting many of their regular activities. They felt breathlessness was more serious and saw it a sign of the extent of their lung disease. Fatigue, although not life threatening, was more persistent and difficult to manage. In learning to live with LAM, the women learned to manage these illness effects.

4.3 Finding motivation and direction

After the initial shock of diagnosis had settled, the majority of participants actively engaged with managing their illness in an effort to regain wellness and a sense of stability, normality and control in their lives. Each participant's personal life meaning motivated and directed their attitudes, decision-making and actions as they learned to live with LAM. When they were diagnosed most had experienced profound disruption as they perceived the stark discrepancy between their life meaning and the implications of their illness for their future. In their everyday lives following diagnosis they sought meaning in their illness that could be integrated into their life meaning to restore a sense of coherence and balance to their lives. The participants' sense of self, purpose, beliefs, disposition, family and cultural values, and past experiences, as aspects of their life meaning, were reflected in the ways they managed and adapted to their illness and their individual responses to new turning points.

The women were particularly motivated by the sense of purpose they felt in their family relationships and responsibilities, and their desire to work. Fifteen participants were married or living with partners. Seven were responsible for caring for dependent children, one for a disabled adult, one for an elderly husband, and one for an elderly parent. They wanted to be well to fulfil their responsibilities, participate in family life and see their children finish school and grow up. Sarah felt "*if I can see them (her children) grow up it'll be ok*". Mia sustained hope that she would live to fulfil her goals of travelling with her family and seeing her son get married:

I never think that I'm not going to be there in the future basically. So I just assume that I'm going to be there when he's 20, when he gets married... I want to make a trip around the world with (my husband) on a cruise, so maybe in 10 years...when we save and when (my son) is older so that we can take him with us.

All the participants were working when they were diagnosed, in roles such as company CEO, lawyer, nurse, accountant, teacher, editor, engineer, social worker, child care worker, secretary, office manager, information technology manager, and project manager. The women felt a sense of purpose in their work for satisfaction, recognition of achievement, self-esteem, and to contribute to family finances, workplace and community. Work symbolised independence, control and normality, reduced isolation through social

connections in the workplace, and balanced the losses and uncertainty of living with LAM. Helen said:

I really like my work and it's important to me. It's just my sanity as well really because if I'm at home sitting thinking about LAM too much it can get to me... It's good for your mental health to work... and financially...it keeps me going.

The majority of participants continued to work after diagnosis even though, for some, it was necessary to reduce their hours. Eleven women were working at the time of interview, four full time and seven part time. Carol and Veronica had retired as they were at retirement age. Irena, Margaret, Aiko, Patricia and Vidu had stopped work due to their illness. For Irena, “work was another way of saying well things are normal... at work I used to forget”. Anna’s attitude to work reflected her family’s strong work ethic and belief in one’s ability to get through hard times:

My mother taught me to be strong...take the day as it comes...My job’s important to me and having that financial independence... trying to keep it balanced...My lung function is terrible but if I can go to work for five days then I feel like I can function.

Having a sense of purpose enabled the participants to sustain hope for physical stability and wellness. In turn, they held hope for a life imbued with meaning and purpose despite the limitations of their illness. The women viewed hope as an essential element of living with uncertainty and moving on with life. Patricia believed “hope is so important... you’ve got to hold onto hope”. Sarah felt that “hope is the most powerful thing to have... If someone can give you a bit of hope you can actually move forward. If there’s no hope you start to just flounder”. Eva associated hope with a sense of purpose:

It's necessary to have hope. For me, hope is... a peaceful life. A life that has purpose... looking after people; really sharing the potential that you have... I would consider my greatest hope is really that I'm of some use to other people.

The participants possessed core self-efficacy beliefs which determined their confidence in their ability to manage the challenges associated with their illness. These had developed through their previous life experiences, managing challenging situations, and competently performing their work roles. When they were diagnosed, Patricia, Eva and Margaret had demonstrated high levels of self-efficacy gained from their past experiences of adversity

and illness. Sarah demonstrated self-efficacy in her belief that she was capable of managing her breathlessness when she attempted activities:

I'll try almost anything... I never know what I'm going to be able to do... Everyone said... we're going to go for a walk but if you want to stay and go for coffee... I said, no, I really want to go for a walk... I'll just go up the stairs slowly... that actually was fine and then I walked away thinking, ok well I'm really happy with myself.

In contrast, Veronica and Vidu showed lower self-efficacy and perceived themselves as lacking confidence. Both had chosen to avoid engaging with their illness when they were diagnosed. Veronica viewed herself as a gentle, timid person. She believed her lack of assertiveness may have stemmed from her early childhood living with an abusive father:

He (my father) was very, very violent... I think he had a lot of anger in him... Also emotional cruelty. He used to yell and threaten... I think it has had an effect on me, lack of confidence.

Vidu's family and cultural values determined that her close family provided her with consistent moral, practical and economic support. However, her negative disposition and her reliance on them made her lose confidence in her ability to manage her illness herself:

Always we have that bond with your parents. Even though we get married they just think that they have to look after us... she (my mother) did everything for me and even my dad did everything for me... I was not able to handle it by myself.

Eva, Patricia and Margaret showed that having an optimistic disposition and sense of humour assisted them in managing their illness. Eva identified herself as having "always been a very positive person... I've just been born like that". She felt her positive attitude contributed towards her maintaining a sense of hope. Patricia believed "you've got to have a sense of humour. You don't survive without it". Margaret stated that she had "always been half full, not half empty", and enjoyed having a joke and seeing the funny side of situations:

I'd rather be happy... It's still hard, every day I have problems breathing and I'm restricted in a lot of things I do... I can't run but I can walk slowly. I can't do certain things I used to but I can sit on a computer and design something... I'd rather look at things in a good way than a bad way...

I make jokes about it, even in the hospital... I wasn't allowed out of bed for three weeks and I had to have a catheter and I said, oh this is great... I could go out and get drunk and I wouldn't have to worry about going to the ladies (laugh). You know it's just I use really sick humour.

Throughout their illness the participants revealed their life meaning in their decision-making and their efforts to integrate the meaning of their illness and restore order to their lives.

4.4 Learning to manage

The participants learned to manage their illness through both their day to day experience of living with it, and employing active strategies. Over time they came to know their changed bodies well. They became familiar with breathlessness and when it occurred. They learned to manage it by anticipating it and avoiding or pacing certain activities, or resolve it by slowing or stopping what they were doing. Normal activities could take much longer to perform. Mia said, *"It takes me three hours to do what you would do in twenty minutes"*. Many found this frustrating but adapted over time. Sarah said:

I'm acutely aware of how far things are these days... I've made a conscious decision recently...my natural instinct is to go faster but I'm out of breath when I get there. So it's like just pace yourself a bit better... it is worth doing.

In the context of living with an incurable disease with few treatment options, many participants demonstrated their agency in taking responsibility for their health by independently engaging in active strategies and alternative therapies. The women focused on general lifestyle measures such as exercising and eating a healthy diet. Eva and Louise reported losing excess weight improved their breathing. Deb found an alkaline diet gave her more energy, *"I honestly felt like a gazelle just bouncing through the forest...It was fruit and vegetables"*, and practised breathing exercises and meditation, *"I do lots of breathing exercises. I do try to meditate... I'm slowly getting better at it and same with my breathing."*

Exercising and being fit and active had been a normal part of life for many participants before the onset of LAM, and, for Patricia, Sarah, Julie and Helen, an important aspect of their identity and life meaning. When the women accepted that they needed to slow their pace and adjust their fitness goals and expectations they were able to enjoy and feel the benefits of exercise while living with their chronic lung condition. Fifteen women exercised regularly. All walked, either outdoors or indoors on a treadmill, Patricia, Julie and Helen cycled, and Patricia continued to swim. Exercise promoted fitness, improved their energy

levels and mood, and restored a sense of normality. This increased their confidence and happiness and gave them a sense of achievement and wellbeing. Mia found that “going to the gym...was really good for my emotional stability...I felt more energetic”. Jess said:

Exercise...for me was really your sense of wellbeing... When I come home I can do a few exercises... so I have been feeling quite a bit healthier... and also started to stay up a bit later and feel a bit more like a normal person...Exercise is really crucial. If I don't exercise I get much more tired... it does make you feel stronger and happier.

Belkin et al. (2014) suggested that traditional pulmonary rehabilitation programs might benefit women living with LAM. Only Irena and Margaret reported attending a single pulmonary rehabilitation program and offered no comment about their experience. Most of the participants, in keeping with their striving for normality, chose to exercise outdoors or in environments such as gyms with other well people closer to their age.

Five participants practised yoga and felt that the associated relaxation and stretching assisted their breathing, gave them more energy and improved their mood. Patricia found it “helped with the breathing...it has a positive effect mentally”. Eva felt:

Yoga seemed to help my energy levels quite a bit... you open up your chest, and you relax... open up the lungs, and... breathe a bit better...the way you feel afterwards is just really uplifting.

Five women meditated regularly. They found it facilitated processing of difficult emotions and improved their energy levels. Mia felt that “twenty minutes of meditation is like three hours of sleep”. Clare said:

I just found it very powerful for me... it's all positive, as much as I may be going through rough times...I try to do a lot of visualisation... just letting your body be open to healing.

Seven participants decided to try alternative therapies such as bush flower essences, vitamins and supplements, traditional Chinese medicine, acupuncture, kinesiology, spiritual healing and reiki. Clare, Deb and Mia found that bush flower essences and spiritual healing were effective for emotional healing. An important aspect of these therapies was their acknowledgment of the participants’ emotional health and the long consultation time with the therapist which allowed them to talk through their emotional issues and feel listened to.

In contrast, short consultation times with physicians did not allow for expression of emotional needs. As Mia recounted:

I went to a man who advises on Australian Bush Flowers... It was like going to a psychologist basically. I sat there and talked to him for an hour and a half... I think that helped me a lot...cause I didn't really have anyone to talk to about this, cause my doctor... I can't sit and talk to him for an hour and a half, he doesn't have the time.

Engaging in creative activities such as painting, drawing and craft gave seven women a sense of wellbeing. For Aiko, painting was a means of expressing her feelings and finding a freedom in imagination that her physical condition had restricted:

I can look at myself too when I'm painting...Another world to step into, the inside of the canvas, to create something, paint something of my heart...I can't fly to see the world... so I was longing to be free to fly like a bird... I like to paint something imaginary... which I don't have with me.

The participants felt the strategies they employed gave them a sense of wellbeing, improved their breathing and energy levels, and helped settle the emotional disruption of diagnosis. There was a sense of taking some control and doing something of a healing nature for themselves. The confidence they gained from managing their illness symptoms effectively in turn strengthened their self-efficacy beliefs and self-esteem.

4.5 Living with a rare disease

Living with a rare disease stimulated the participants to become self-reliant in gathering information, and advocate for themselves to access treatment, have their needs met in their workplaces, and to raise awareness of LAM and funds for research in the community. After the initial shock of diagnosis had settled many made contact with the LAM Foundation or the Australian support organisation or became familiar with medical websites to keep abreast of current research. Jess said:

The kind of patient I wanted to be was very well informed. We read PubMed. We don't understand all the medical jargon, but we certainly try and understand as much as we can. I think it's been really crucial.

From their own research the women became aware of reports of the effectiveness of sirolimus in stabilising lung function and treating lymphangioliomyomas. Anna and Jess advocated their physicians for a treatment prescription. Anna “*had read and researched so*

much about it. I was persistent with (my doctor). I said, you know, seriously, you need to get me on this". The women who were prescribed sirolimus were vigilant and took responsibility for monitoring their drug levels. Not all physicians were aware of therapeutic levels and Helen and Julie also had to advocate for therapeutic medication doses. Helen felt she was always "dictating" to her doctors:

I said to them, what are my trough levels meant to be? ...I'd been fighting with them about even being on sirolimus...I just said I just don't feel comfortable going to three milligrams. I'd rather go to 2.5 milligrams just for a couple of weeks. So once again I'm dictating.

While the majority of participants engaged with their illness in this way, others displayed varying levels of engagement. Veronica and Irena continued to avoid websites and information related to LAM as they had done when they were first diagnosed. However, they had now made contact with other women with LAM and Irena became involved in fundraising. Vidu still avoided thinking and talking about her illness and associating with other women, and relied on her family to gather information and advocate for her.

4.5.1 Advocacy at work

Breathlessness and fatigue could interfere with the participants' ability to carry out their work commitments. Walking long distances or upstairs at work, carrying heavy loads of books, bending under desks, and needing to take time off for periods of illness were some of the issues they faced in the workplace. Jess found fatigue particularly affected her ability to concentrate at work, and open areas with higher noise levels exacerbated the problem. "I'm really tired at work...I find difficulties concentrating. (The people) I work next to... shout a lot... it distracts me more than other people". These issues were, however, not necessarily visible to employers and work colleagues.

Invisibility was, in Jess's words, a "double-edged" sword. It had the dual effect of allowing the participants privacy if they chose to conceal their illness but could also limit support in the workplace. On the other hand, revealing their illness, while engendering support, could threaten their identity as competent individuals and accentuate feelings of being different or favoured. Irena was selective in disclosing her illness to her manager and few of her colleagues only, "I didn't want people thinking I was having favours done. I can cope, I can cope, I'll be right, I'll just take things slower".

While Mia's manager accommodated her needs, including extended maternity leave and

part time work, she struggled with conflicting feelings. She wanted to work effectively but felt limited by her illness. She appreciated the support of her employer but felt, at the same time, that her capable and resilient identity was reduced, and guilty that the concessions made for her were inconveniencing her colleagues. She would have preferred to stay at home to care for her young son but needed the income that work provided. She was also concerned about the effect the limitations of her illness would have on her ability to be successful in applying for a new position:

I don't feel as competent any more... Everyone has to sacrifice themselves to make room for me...The office has made so many alternative arrangements to accommodate my needs... I want to be mature as well, and resilient. I don't want to be that typical, I can't deal with this, I can't cope, I have to quit. So it's been really hard...

You're competing against so many people when you're applying (for a job). Truth is that life's like the survival of the fittest... you struggle with the competition factor versus the equity, inclusion factor.

In contrast, when Jess advocated for more flexible arrangements to address her fatigue, she felt unsupported and stigmatised. She perceived that the subjective nature of her fatigue meant that it was discounted by her manager and accounted for her average work performance reviews. Through persistence she was finally able to negotiate working from home one afternoon a week. More flexible working arrangements had the additional benefit of promoting wellness by allowing her more time for exercise which increased her energy levels and made her feel healthier:

When I talked about my fatigue and my disease... my boss really didn't care... I would like fatigue to be taken more seriously...it's a double-edged thing, because I got quite a bad performance rating recently... obviously they view my performance through that...This is the first time I've had to go on about having a disease because it's such an unsupportive working environment...I did talk to my supervisor and she started a thing with me where I can come home on Wednesday afternoons and work from home for the rest of the afternoon...That's meant that I can exercise mid-week... So I have been feeling quite a bit healthier.

4.5.2 Advocacy in the community

Seven participants engaged in community advocacy by speaking publicly about LAM,

distributing information brochures, and raising funds to support LAM research. They did this for altruistic reasons and, in a positive sense, their activities provided a sense of purpose in their lives. However, this advocacy also raised dilemmas for the women due to the general invisibility and rarity of LAM. Women often looked healthy, even with advanced disease. In this context they were aware that, unless they appeared 'sick', they were unlikely to attract financial support for their rare condition. Often a woman using oxygen therapy would become a public face for LAM and could then feel burdened by a 'sick' identity. This was particularly the case for Clare who had used oxygen temporarily after being in hospital. She preferred to be seen as 'well' rather than 'sick':

When I returned home after the hospital... I actually felt a bit awkward because I looked fine. People didn't really see me carrying around the oxygen for the first few months, because I just did that at home... I always wanted to perceive myself as healthy and not like that... I just had guilt over people thinking I did the fundraiser for the wrong reason...five months later, she's already fine, she walks round, she doesn't even look like she's sick.

Fundraising was a challenge as, being a rare disease, there were no large fundraising bodies and the women themselves raised funds within their own communities. It was difficult to keep asking the same people for donations. Some had limited social networks or felt these had shrunk since their diagnosis. Furthermore, they felt guilty and believed that people would be suspicious when their disease was progressing very slowly and they continued to look well even though they had a life threatening condition. Aiko said:

The number of the LAM patients is so small...Even though I try to do the fundraising... I cannot keep asking the same people... I'm still not dead yet. They wonder, is she using all the money?...It's not easy to be making friends, and if I make friends and then suddenly ask for a donation, they are wondering... I don't want to do that to my friends.

Fundraising took a toll in terms of time and energy. This was particularly the case for the small number of women who were active in the peer support organisation. In addition to fundraising they kept the website up-to-date, provided support and advocated for improved resources for women with LAM. At times they used their personal resources to support their work. According to Ruth:

We don't have enough people on our committee. ...We've got so many balls in the air, there's supporting women with LAM, there's maybe producing a new

brochure, there's getting the web site updated. We're looking for other ways of funding. There's the liquid air... It's a big pressure on us...I'm really glad to be doing such an important job. But from a point of view of my energy and where I spend my time, I can feel guilty doing something for myself instead of working on something that's needed for LARA.

4.5.3 Benefiting from advocacy and medical research

Nine women were prescribed sirolimus at the time of the interviews. Sirolimus was a product of medical research funded by women living with LAM. It assisted the participants who were taking it to adapt to living with LAM due to its positive stabilising effect on their physical symptoms and subsequent improvement in their quality of life. Not all women were assessed as suitable for the medication and some, already at a late stage of the illness or not in contact with a LAM expert, as was the case with Margaret, were not offered it. Aiko had only commenced sirolimus four weeks previously so was unable to report its effects. The other eight women all experienced positive effects, including improved or stabilised lung function, and resolution of their abdominal lymphangiomyomas, chylothoraces and haemoptysis (See Appendix 3, Table 3.4). Irena said:

I'm living proof that ever since I have started taking sirolimus again, I rarely now cough up blood. I can actually go to bed now and I don't wake up every 15 to 20 minutes coughing up blood... I used to bend over and it would come out like a tap. It was so frightening...Since the sirolimus I'm feeling better.

Only three participants reported side effects. Helen and Irena's menstrual periods became irregular. Helen and Julie both experienced fatigue. Julie additionally had nausea, diarrhoea, shaking, hoarse voice and a constant cough. Helen experienced mouth ulcers, bloating, joint pain, delayed wound healing, and raised cholesterol. While these side effects were disruptive and limited the improvement in their quality of life, the women were nevertheless prepared to tolerate them for the benefits they derived from stable lung function. As Julie said:

I started on the medication...it's almost like swap one set of symptoms for all these other scary things, like increased lymphoma...(and) kidney damage...You just take it and go, that's that versus lung transplant. You weigh it like that. I don't feel particularly well on it either. I started to sleep heaps more on my days off... I get nauseous. I get diarrhoea and the shakes...I get a hoarse voice as well now... I tend to have this constant cough,

which I never had. But my breathing tests have improved...now mopping and everything doesn't seem as bad. I did actually go up a bit of a hill...If it's going to make my lung capacity better and that prolongs lung transplant, yeah, I'm just going to have to take that chance.

Most women remarked on the enormous, even miraculous, improvement in their quality of life with feelings of wellness and stability. Anna believed that “*sirolimus has given me my quality of life...the confidence and all that comes from having a better quality of life.*” Being physically stable meant that the illness moved to the background of their thoughts allowing them to move on with their lives. Although they still had to modify the pace and manner in which they performed activities there was a renewed ability to engage with every day and enjoyable activities which restored a sense of self and normality. Eva said:

They (measurements of lung function) kept declining until I started taking sirolimus. There was quite a fast decline. Within a year I lost about 30 per cent of my lung capacity...I regained most of it back. It was just a miracle...It saved my life pretty much... it's made me stable.

Importantly, the improvements in lung function and resolution of other physical symptoms made women feel that the progress of their disease had slowed. Uncertainty and hope could more comfortably coexist. Although uncertainty remained, they had hope for the future that the need for a lung transplant would be delayed, their life expectancy would improve and they would maintain quality of life. There was also a sense of hope in knowing that research into a cure for LAM was being undertaken and empowerment in being conscious of their own role in enabling research. As Sarah said:

I'm quite convinced that if I hadn't started on this medicine two years ago, I'd be very bad by now. I think I was rapidly, rapidly getting worse the first year after the diagnosis. I'd degenerated significantly...I think the key change is really the medicine and suddenly having this feeling of there will probably be a future, I think it's a huge, huge thing... I think knowing that there are people interested in it, working on it, trying to figure out how to do it, is also huge.

4.6 Accepting and adapting with the passage of time

The passage of time itself encompassed a healing quality for the participants, not as a biomedical cure, but rather coming to terms with their illness and learning to live with it over time. As they adapted to life with LAM its disruptive effects diminished. Ursula felt

she had “become a bit blasé about it, I've had it 13 years...With me it's just more annoying, it's not frightening because I'm used to it”. The women felt hope for the future when they had lived past their predicted lifespan and were physically stable. Reaching ten years, the common maximum lifespan they were given at diagnosis, was a significant milestone. As Irena said, “I don't think about it as much, because I've also beaten the odds, if you like. When they said between one and 10 years, well it is 10 years now”. The passage of time allowed Anna to reflect on her illness experience and view it in a more positive light:

As much as there are challenges with LAM...had you asked me when I was diagnosed I probably would have said it's the worst thing ever, but as time passes and the saying time heals - it does, because you have that space to reassess what's happened... I realise the more time goes on, it gives me that reflection to see it in a different way... As hard as this condition is, I'm glad what happened, happened because I think I feel more at peace with myself.

The participants revealed that, over time, they had learned to live with their changed bodies and the limitations that these imposed. They gained knowledge and skills to manage their symptoms through their own experience and the active strategies they employed. The rarity of LAM stimulated them to develop additional skills of self-reliance and self-advocacy. Managing their illness effectively reduced its disruptive effects, increased their self-efficacy, and generated positive feelings of stability and control.

4.7 Managing relationships and responsibilities

Illness disrupted the participants' relationships with their partners, mothers and friends, and their ability to carry out their responsibilities of caring for their families. In managing their relationships and responsibilities they simultaneously had to manage the unsettled emotions and identity threat they experienced alongside them. At diagnosis, their partners and parents had offered unconditional support to many participants. After diagnosis, support became more complex and engendered both positive feelings of appreciation and closeness and negative feelings of vulnerability, guilt, and being a burden.

4.7.1 Disrupted equilibrium

Living with LAM disrupted the normal equilibrium of the participants' relationships with their partners. Uncertainty made them feel vulnerable physically and emotionally, and introduced a fear for the future of their relationship as well as their health. Sarah expressed the vulnerability she felt:

I'm sure that the whole diagnosis has changed our relationship completely... I felt for the first time very vulnerable... What if he suddenly decides he doesn't want to deal with this and he leaves?... It's all the doubts that it brings out.

Fear of the possibility of an early death was a confronting topic to broach and it frequently sat unspoken and unresolved between a participant and her partner. Sarah and Mia reported that they and their partners avoided the conversation feeling it would create anxiety and worry that they could not deal with constructively. Mia said:

We talked about it (the future) a couple of times, but I think that men... avoid this kind of controversial issue. So we never really debriefed on it very thoroughly... it's too scary... we didn't feel it was going anywhere constructive. It was only making us anxious and worried about the future... It was very negative, so I think that we both agreed silently not to talk about it.

While most participants continued to receive practical support from their partners, they found it difficult to elicit emotional support when their partners were themselves emotionally affected by their illness and unable to express their emotions. Helen said:

My partner finds it very hard to talk about how he feels. As a result it really has impacted on our relationship. We do argue a heap. I initially just pushed him away... I just felt he deserved better... I didn't want him to see me deteriorate... He didn't get the job he was promised - which is a higher level - because they said let's put you in this job because it will be easier for you with what's going on at home. He's really quite depressed about it now.

The women felt their identity as being competent, independent and in control was threatened by their need for additional support and their inability to contribute to the family in the usual way. Disruption of their familiar roles created conflicting feelings including both positive emotions related to each woman's appreciation of her partner's support, and negative feelings of guilt, being a burden, and vulnerability through the loss of reciprocity in their relationship. Sarah felt her husband "was very tolerant. I actually can't fault him on it. I just felt very guilty".

Helen and Mia, having reduced their hours of work and being restricted in their ability to carry out household tasks, expressed feelings of guilt and being a burden because their partners were assuming greater financial responsibility yet reducing their own career

opportunities by spending more time on home chores and caring for their children. Mia expressed her feelings of being a burden:

I feel bad getting sick so often and then he has to compensate for that...I've always been very self-confident and really proactive... and quite a leader in many aspects of my life, and I think that since I've found that I've got LAM it has made me feel more vulnerable... I feel that I am more of a burden to (my husband), to my mum... I felt more of an asset before.

Jess felt that her illness had “*totally dominated our relationship...he's become more of a carer*”. Clare believed that the patient/carer status that endured being in hospital contributed to the breakup of her relationship:

I separated with my partner... I blame myself because... I lost a lot of confidence after I was sick, just even in my body image... I was pretty wilted away and not myself ... I think he started to not see me as attractive either. Maybe it's that patient-carer relationship, it has a big impact on you.

4.7.2 Altered sexuality

Issues of sexual relations concerned the younger participants particularly. Of the ten participants who discussed the impact of LAM on their sexual relations, nine were between 34 and 48 years old, seven were 40 or younger. One post-menopausal woman gave the perspective of a more mature woman. Of the remaining nine participants, five stated only that their sex lives were normal or satisfactory. Four of these women were older, between 51 and 68 years old. Of the other four women, one was currently not living with her elderly husband, and three women were single and not in permanent relationships. Due to the very personal nature of this topic the women's comments are not identified.

LAM had a significant impact on the participants' sexual relations. Sexual activity induced breathlessness which was uncomfortable and a source of fear and anxiety. Some women were unable to tolerate the weight of their partner on their chest. Fatigue reduced their libido and the women often felt so tired that they had little desire for sexual activity. The fear of becoming breathless and breathlessness itself was a “*passion killer*” which medicalised a personal moment of intimacy. This was accentuated by feelings of being unattractive after periods of being sick. One woman described the effect of sexual activity on her breathing:

You're just worried about your breathing. You're going, hang on, you can't push on me too much... It becomes a bit medical almost because you're going, no, hang on, just wait - I need to get my breath back or stop for a minute. It's a bit of a passion killer... (We have sex) not that often. It's terrible. Sometimes just once a month.

Two women reported experiencing persistent cough when having sex. One described how coughing affected her relationship:

It would be in the middle of a moment and I would completely lose it coughing and then I actually couldn't get my breath back and that's all very traumatic... You're a wife and... you think, well at which point am I going to turn around and say to him, just don't come near me ...It is very destructive for a relationship I think.

A third woman discussed the impact of fatigue and breathlessness on her relationship:

I just feel tired...I really have to make an effort to try to keep active and to try to get in the mood...I would honestly prefer just to sleep... once it happens, then I enjoy it but then I feel really breathless... I'm scared...It's such an intimate moment that you don't want to pull out your inhaler in the middle of it because it ruins the whole thing. So I really struggle with that... I avoid anything that will bring that attack. I'm not attractive ... I'm more passive.

Most women reported that they did not initiate sex and sexual activity was therefore reduced to between once a week and once a month or less. While partners were supportive in this situation the women felt guilty and worried that their partner's sexual needs were not being met. A post-menopausal woman found that low libido which she attributed to menopause added to that associated with LAM:

I think I've got a very low libido and I could just not have it, you know? Then I know if I don't then it impacts on the relationship and we lose that intimate connection... I don't feel like being the one who initiates it...I don't want to get breathless...After menopause... I just don't have that sort of lusty feeling anymore ...I feel guilty.

Two women reported they were able to resume normal pre-illness levels of sexual activity with improvements in lung function after they commenced sirolimus medication. Three

women and their partners managed the problem of breathlessness by adopting more comfortable positions or by expressing intimacy in other ways. Being able to communicate openly and receiving expressions of acceptance and reassurance from their partners contributed to their satisfaction. As one woman said:

I started declining, I was feeling quite guilty, not only I didn't have energy for sex, I didn't have energy for walking. So we weren't doing anything...My husband was amazing... We found other ways to be intimate without having to do things that I can't do.

Three participants commented that issues related to sex were not commonly discussed and were difficult topics to raise with their physicians in the limited time available for consultations. One woman felt that “everyone's quite prudish about it, they're not going to ask, does this and this and this happen?” Another reported that when problems concerning sex were raised on the LAM Facebook site there was a flood of responses indicating that this was a common issue. There was a sense of relief to share experiences and not feel alone with the problem:

One of the girls on the Facebook site did actually say... I really want to know how people feel about having intimacy because, she said, I'm just exhausted, I can't catch my breath...It just opened up this whole massive debate... we all had the same issue but everyone was too scared to say it.

Other authors have found, in common with this study, that sexuality was an important aspect of identity, health and quality of life for women living with long-term illness (Koch et al. 2002; Kralik et al. 2001; Wilmoth 2007). Verschuren et al. (2010), in forming a conceptual framework for chronic disease and sexuality, argued that relational skills influence how the impacts of the illness are felt as a couple. Further, Badr and Taylor (2006), in a study of spousal communication in lung cancer, found that open communication assisted both partners to cognitively and emotionally process the illness.

4.7.3 Risking pregnancy

Contraception was an important issue for the participants who were premenopausal, had been advised not to become pregnant, and who were taking sirolimus which has immunosuppressive effects. The women expressed confusion around contraception and found it difficult to access relevant information. They wished to discuss their reproductive health collaboratively with their physicians but the topic was not raised by some physicians

and others had differing opinions. Some women sought information from other women through personal conversation or Facebook. In this context it was difficult for an individual woman to decide what was the most suitable form of contraception to use:

The only time we discussed it with (the other doctor) is when he said, oh well, we can bring on early menopause...the dilemma of what you can take, because most of them are oestrogen based...I really need to talk about contraception, it's an ongoing issue on the Facebook sites... I don't know what to do about it...We need to have an answer about contraception because all the specialists have said something different.

Seven participants discussed the contraceptive method they were using. Five women risked pregnancy, three due to their partners practising withdrawal and two by using no contraception at all. Two women and their partners used condoms. Both a woman and her partner could feel the pressure of being responsible for contraception. Younger men were reluctant to undergo vasectomy as a permanent surgical procedure. Two participants reported that it was up to them to explore other options for contraception because their partners disliked using condoms. The women feared using any hormone based contraception. They tended not to consider tubal ligation because they would need to stop sirolimus prior to the procedure due to its possible effect on wound healing and also because they preferred not to have sedation during the procedure when their lung function was compromised. They expressed a high level of indecision and a need for consistent professional advice. The women discussed their decision making regarding contraception:

I did not take contraception... Condoms we'd be using but because I used to be so regular I'd look at the time of the month... withdrawal - I know that's not fool proof but it's served us well thus far... I know when I'm ovulating, I regulate it that way. (Withdrawal as contraception)

You're supposed to use contraception on sirolimus. I wouldn't even expect to be fertile now. But, nobody's actually talked about it with me. (No contraception)

We talked about it (vasectomy) but (my husband)... said no, I don't want to... I can't have the risk of being pregnant because of LAM... so we use condoms and he said, I really miss not having condoms...so I have to do something about that. ... I have no idea...Information about that... would be really good

to put out there for LAM patients...It's quite a taboo subject. Some people wouldn't dare to ask publicly. (Condoms as contraception)

Using an unreliable method or no contraception could result in added anxiety for a participant whose menstrual period was late arriving. This was particularly the case for two women whose periods had become irregular since they started on sirolimus therapy. Anxiety around contraception and possible pregnancy placed further stress on a couple's relationship. One participant reported:

Basically, the specialist (physician) said to (my partner), she can't have any more children; it would kill her; and you're going to have to get the snip... It was that whole pressure - the responsibility's on you. It's horrible for him. He had never really thought about that. I was always on the pill. Now, we're in this position where I'm not on anything...we use condoms but it's that hit and miss thing. I'm just petrified all the time that I'm going to get pregnant. It is a responsibility for him and that's another passion killer... we kind of do withdrawal...He hates them (condoms)... It still petrifies me going on to some hormones... It has caused a bit of tension between us in terms of I don't want to pressure him to have a vasectomy. He's not really ready yet and I don't really want to go on the pill.

One woman felt very emotional and had severe cramping after having the Mirena (an intrauterine device) inserted. Similarly, another found Implanon (a contraceptive implant placed under the skin) made her unacceptably highly emotional and she had it removed:

I have given up on using any chemical contraception whatsoever... I have tried the implant - the under the skin one, Implanon. That made me feel miserable. I was overly emotional... I got it pulled out after two weeks.

Although contraception is an issue of great concern to pre-menopausal women living with LAM, the only contraceptive advice offered by the European Respiratory Society Guidelines for the Diagnosis and Management of LAM is that oestrogens should be avoided, including the contraceptive pill and hormone replacement (Johnson et al. 2010). Lara et al. (2012), in a review of contraception, pregnancy and rare respiratory diseases, recognised that there is limited experience in managing the reproductive health of women with LAM and that advice regarding best contraception and pregnancy should be individualised and multidisciplinary. Thompson et al. (2008) further argued that it is important for women to

be given adequate time and space to discuss their concerns and share in decision-making regarding reproductive health.

4.7.4 Balancing caring for others and caring for self

The participants had to balance caring for their dependent children and elderly relatives with caring for their own health. Mia and Sarah revealed how they were able to care for their young children while managing the effects of their illness. Aiko, Patricia and Irena cared for elderly relatives. The particular issues which Helen faced in caring for her children while using oxygen therapy are discussed in Chapter 6, Living with Advanced LAM.

Mia and Sarah's breathlessness made it difficult for them to pick up and carry their young children. They planned and minimised the number of times they went upstairs in a day and educated their children in simple terms that they could not expect to be always lifted and carried. Mia had not found any resources to assist her in explaining her limitations to her young son. She said:

I think that being a mum with LAM you're always reminded that there is a disability there because you cannot do some things. For instance I cannot go upstairs a thousand times during the day so I really think about how I do things and the routine is around that...I'm still having to carry him up the stairs and now he's almost 14 kilos...Those are the moments where I really struggle with LAM and with motherhood... Once I get up the stairs I am so breathless... Sometimes it's really scary, you know... You think about all of these things that other mums don't have to think about...You just adapt to it.

Managing household chores caused the women to become breathlessness and added to the physical demands of mothering. Mia found “*that the housework was the toughest*”. Both women recognised that having regular help in the home allowed them to better manage caring for their children. Sarah could afford regular help in the home. Although it was an added financial burden, Mia “*decided to pay for cleaners twice a month...I came to the realisation that I need to invest in that so that I don't get as breathless...It's managing the health, literally*”. Both women were supported by their own mothers who minded the children, lifted them, and assisted with shopping and other practical tasks.

Mia and Sarah both worked. In balancing the demands of work and mothering, the women also had to care for their own health. In order to keeping these three aspects of life in balance

they adjusted their work goals and reduced their hours of work which compromised their income and career advancement. Mia struggled with achieving a balance:

Most people have to think about motherhood and professional life. We have to factor in health as well... We have an added thing to balance out...you struggle with doing that.

Mia experienced multiple periods of illness during the year. These were usually due to chest infections and often she had to be hospitalised. While she was helped by her mother and her husband, she felt she needed additional services during this period of crisis but did not know how she could access them:

I had a chest infection and the coughing was so bad that I ended up breaking a rib...I had to do all the mother things... It was so much pain that I wish I would have had some support...What you need, you know, when you get a crisis, is to get someone from the Department of Health and Ageing... get access to that support which we can't because we're not disabled as such. So that was really hard because I felt like I wasn't able to be a good mother because of that.

While modifying their activities, the women interacted with their children in ways that were consistent with their sense of self and identity. Sarah wanted her children to know and remember her as a fit, active person so continued to participate in exercise and family activities such as hiking, managing her breathlessness by slowing her pace. Mia felt her illness limited her ability to bond with her adopted son through vigorous play. She adjusted by altering her expectations and focusing on interacting with him through activities such as art and reading which she felt aligned with her sense of self:

In gymberoo I have to carry him and dance... I just warned the staff there right at the beginning, I have a lung condition... can you help me?...I guess that from the perspective of attachment it's an obstacle...I still cuddle, I do everything else...Because I love reading and I'm very artistic I think my son relates to me through that.

Aiko, Patricia and Irena had responsibilities of caring for older, dependent relatives who had poor mobility. In fulfilling these responsibilities, the women tended to ignore their own problems, pushing the elderly in wheelchairs and attending to their physical needs, even

though their breathing was compromised. Aiko's husband was 88 years old and required assistance with activities of daily living:

My husband is an elderly guy... he has heart failure. So last year was I was busy looking after him, but at the same time I became sick and then I'm being hospitalised and my oxygen level is not so good...He's alone, so I was also worrying he's not capable by himself.

Patricia travelled weekly to assist her mother with caring for her seriously ill father before he died, washing him and turning him even when she was on oxygen herself. Irena's mother had moved to a nursing home with dementia and Irena visited her daily, changing her and helping to dress her even though it made her breathless:

Because I'm constantly with mum I don't think about my situation... It's only when I have to change her, when I have to bend, the coughing starts. So that makes it a bit hard.

4.7.5 Feeling 'out of synch'

Receiving their diagnosis shifted the participants' perceptions of time. Before the onset of LAM, they had perceived their life being lived through in a linear fashion. They now felt a greater awareness of time, as Sarah said, "*I've become more aware of the time...much more aware of how long it takes to get places*". Paradoxically the women felt time had both slowed to accommodate the slowing of activities compelled by their breathlessness and fatigue, and accelerated to project them forward on their life trajectory to a stage where they were confronting their mortality, an experience they had not expected until old age. Jess felt "*as though I skipped middle age and I went straight into old age...I felt as though I'd almost accelerated*".

Their paradoxical perception of time had the effect of making the participants feel 'out of synch' with their peers and their mothers. This was particularly the case for younger participants. They felt different to their peers for whom life in the modern world continued as normal, fast paced and active. Peers were proceeding along their expected life paths, working, socialising, partnering and having children. Being forced to confront one's own mortality and dealing with issues that peers did not have to face was emotionally taxing but also a source of knowledge and wisdom usually associated with older people at the later stage of their life. Jess felt as though "*they've overtaken me in some things*" and, at the same

time, “*I know something that they don't know yet*”. Sarah had a sense of being emotionally older than her peers:

You deal with your mortality. There's a whole lot of different issues that come up that most people your age don't deal with...I felt like I'd somehow aged emotionally terribly... I almost identified more with my grandmother who's 92...That's distressing.

While they felt older than peers, younger women still wished to associate with and be identified with their own age group and this provided a further incentive to health and wellness. Deb continued to go hiking with her friends:

When I went hiking...normally I'd be the first in the pack...I've always been fit... I went with my brother and this other friend ... I couldn't quite keep up to them as hard and fast as they were, but I did it and I was really quite pleased.

While the participants felt ‘out of synch’ with their peers, conversely, most friends did not have the life experience or knowledge to understand the women’s situation. The invisibility of LAM contributed to misunderstandings and friends diminishing the impact of their illness. Jess felt “*it's impossible, if you're looking healthy, to have people really understand*”. Helen said:

I lost loads of friends because they just couldn't handle it and they just felt I was being really grumpy and I've got to pull myself together and a lung transplant is going to cure it all.

In the context of living with the potential for their own life span to be shortened, the participants adopted a different attitude to ageing than their peers. Where some of their friends bemoaned their children growing up and their own advancing years, younger participants saw growing older and children growing up as the natural progression of life which they hoped to experience themselves. Growing old, for them, was an achievement to be celebrated. Sarah said:

I know that my anxieties are that I won't see the kids grow up...It really accelerates a lot of things that most people don't think about as they're going through it...I've really changed my attitude to them growing up...When my older daughter... had her first day of school... and all these mothers are crying... I wanted to scream at these parents, it's good they're going to

school...they're supposed to get older...your whole way you see the world changes.

Younger participants compared themselves with their mothers. Their feelings of being 'out of synch' were highlighted by their mothers' ability to physically support them. They felt a discrepancy between their own physical limitations and their mothers who were strong and capable, lifting heavier weights, walking faster, and in some cases, participating in activities which they had had to relinquish since the onset of their illness. There was a keen sense of roles being reversed. Sarah reflected:

My mother...is super fit, she's in amazing shape, she's very strong... I found it stressful that it ended up being I still need her... because I should be the young, fit one at this point in time...This is all wrong you know, she's sixty years old and it's not supposed to be that way. There was a lot that we would have done together that she won't do with me anymore which I think distresses me... She almost must feel like there was a role reversal you know...and I feel left out of that...she puts a contrast on it for me which is difficult.

Mia felt, at her stage of life, she should be caring for her mother who had her own health issues, whereas her mother was still caring for her:

I always felt that at this point in our lives I would be caring for her because she had a shoulder injury and you can tell that she has grown older and that she is weaker, but in no way, I don't have to care for her as she does for me when I have all of these crises.

4.7.6 Balancing negative emotions

The participants balanced their emotions as they adapted to their illness over time. Accepting their situation, adjusting their expectations and goals, effectively managing their illness, the strategies they employed such as meditation and yoga, and finding meaning and purpose through their relationships, spirituality and altruism restored a sense of coherence and increased their positive emotions and self-beliefs. Engaging affordable home services rather than relying on family support enabled them to manage their health while fulfilling their responsibilities and reduced feelings of guilt and dependency.

Experiencing social connectedness in their incidental everyday social interactions enhanced their emotional wellbeing by directly inducing positive emotions of happiness and

contentment. These included sharing a coffee and a laugh with friends, going to the theatre, having dinner with a partner, and cuddling children and pets. Anna was “*seeing my friends a lot more...I’ll make an effort to go because I’ll always feel better after I’ve been*”. Experiences of social connectedness were a source of meaning in raising the women’s feelings of self-worth and belonging and generating a sense of normality. They differed from social support because they involved mutual feelings of companionship without the negative associations of loss of reciprocity. Margaret described how she enjoyed her simple social interactions:

I have days where I actually kill myself laughing with my friends, doing something silly or just having a coffee... Every day I wake up it’s a bonus, every day I wake up and see these two pests (her pet dogs) in my face going, feed me.

4.8 Living in a changing world – turning points and experiences of liminality

The participants lived with their illness in the context of constant change, from everyday disruptions to substantial physical, emotional or social changes. At their initial turning point of being diagnosed, six participants had experienced entry to transition as an existential state of liminality in which they felt a shifting sense of self and of separation from life, of paradoxically being in the world and yet not part of it. Liminality was a fluctuating state which resolved, persisted for extended periods, varied in intensity, or recurred in response to new turning points. The women demonstrated that change in one life dimension could instigate change in another, for example, physical changes could disrupt them emotionally and socially and vice versa. Aiko, Eva, and Anna experienced liminality as a period of personal and spiritual growth which shifted their perceptions and enabled them to complete their transition and live with contentment in the present.

4.8 1 Experiencing spiritual growth and transformation

Aiko had identified herself as socially and culturally liminal before she was diagnosed with LAM. As an Asian woman married to a Caucasian man she felt like an “*alien*” marginalised between two cultures. Being diagnosed with LAM had intensified her liminality and isolation. After diagnosis, Aiko’s liminality persisted. The death of the woman from LAM Australia who had befriended her after diagnosis and subsequent loss of her friendship and support was a turning point which made Aiko withdraw from contact with other women with LAM, “*I closed my door*”. She felt like a “*bird in a cage*”, demonstrating her

vulnerability and entrapment in liminality. *“For the last...nine years... I felt I didn’t care that the world was going on, because I was here still, but almost like everything was dead”*.

Aiko experienced two social turning points which enabled her to move from transition, resolving her liminality to finally accept and adjust to living with her illness, incorporating it into a new stage of life. Firstly, the birth of her grandchild gave her a renewed will to live and hope for a future in which she perceived the possibility of participating in the life of her grandson. Her narrative shifted from metaphors of death to language of life indicating the beginning of her movement out of transition:

I thought I’d never see her (my daughter’s) wedding, her child. Now I’m going to witness new life... I thought well, life may not be too bad. I may be able to see my grandchild go to kindergarten...I would like to give him my culture, to teach him... I started to want to live now ... I felt like why am I giving up before the time comes?

The second social turning point which completed Aiko’s movement from liminality was the sudden death of her son-in-law. Aiko’s reflection on this significant life crisis enabled her to find meaning in her existence, accept her illness, and cross the threshold of liminality to a new stage of her life. This process constituted an epiphany and spiritual passage through its profound transformation of her sense of self in the world. Her narrative reflected her transcendence of spirit above the impermanence of her body and material possessions, and her renewed desire to rise above the limitations of her illness and live in the present:

He gave me, I think, more chance to live. Live until my end...he lived until the end... I feel whatever comes, I take it, and I try to battle through... wherever I live is my home... I don't need to carry the big suitcase with me... look at (my son-in-law). When he died, he couldn't take anything...We should let it go to live light...no purpose... (just) being...I accept what has been given now. I don't have any anger. I don't have any doubt...It's maybe the peace, acceptance I find this time...I feel more stable in a way...calm.

Eva’s experience of meditation and spirituality transformed her sense of self and her perception of her illness. She referred to her intense meditation, in accordance with her Buddhist faith, as a spiritual “*tool*” which gave her mental strength and insight. This reflective space enabled her to feel a renewed sense of self-worth, accept her illness and view it as an opportunity to live in the present with greater contentment. She likened her

view of life before illness to a state of “*dreaming*”. She developed a sense of altruism in her passage through liminality. Subsequently pursuing volunteer work was empowering and gave her a sense of purpose and value:

It's changed me as a person...Meditation strengthened the way my mind works and how I feel...When you actually reflect on death and think about it, then you can much better prioritise your life ...You realise you're not going to live forever...So you do with what you've got right now, you start living more in the present day... I focused on helping others...I went searching for volunteering jobs...It really... did wonders for me because I completely forgot about myself. Yes, I can't breathe very well, but I can do these things. I'm not completely useless...I needed to get that sense of worth.

Anna had experienced liminality following a crucial social turning point of her marriage breakdown and divorce three years after her diagnosis. This had destabilised her emotionally, mentally and physically. Another critical turning point five years later shifted her perception and enabled her to move out of liminality. An acute, disabling attack of anxiety led her to discover a new personal spirituality and practice of meditation which transformed her and assisted her in an enduring way to accept her situation, balance her emotions and feel at peace:

With the meditating, with the breathing...just about not getting so caught up in what's supposed to be...trying to just be...you can feel that everything's not going to fall apart around you...it helps you deal with the LAM a bit better... In an inner peace way I think I have more of an acceptance.

4.8 2 Persisting in liminality

Jess's story revealed that her experience of liminality after diagnosis endured although it diminished in intensity. Jess viewed her illness as a “*transition into a whole other world and becoming another person*” and named her liminality as her “*shadow life*”. Being prescribed sirolimus was a physical turning point which, by resolving her abdominal lymphangiomyoma and improving her lung function, reduced the intensity of her liminality. This was evident in our second interview when she described being “*back to being able to do just everyday things*” and “*feel(ing) solid now*”. She was doing more exercise and “*felt really good*”. She felt a renewed sense of normality at a physical level but this did not transfer to the level of self.

Jess's paradoxes in her narrative illustrated how her perceptions of herself and her continuing social isolation caused her to remain in a liminal state. While she stated "*I recognise myself more and feel more like myself*", she perceived that over the five years since her diagnosis her identity had altered. "*I'd come to think of myself as a patient...a sick person*". She felt "*fit and healthy for me*" yet "*tired and sick all the time*". She thought she was "*more attractive than I have been for a few years*" but, at the same time, considered herself to be "*more of a medical body and an old body...I'm always like a 70 year old*". Although Jess tried to appear the same, "*I've never wanted to present myself as an ill person*", paradoxically she avoided social outings and felt estranged from peers. As a "*sick person*" her sense of self was anchored in her illness.

Although she was managing her illness effectively and had successfully advocated for her sirolimus prescription and more flexible work arrangements, she saw herself as lacking agency and felt she had failed to advance in her career. "*There is no way I would've wanted to end up (in this job)...which is a failure...The whole effort of getting a more demanding job... would probably be too hard for me*". Feeling physically stable and healthier had shifted her to a less intense level of liminality but her self-perceptions and her ongoing social isolation prevented her at that time from completing her passage from transition:

I think of myself in terms of other patients and other people with diseases and I feel as though normal people are quite different...I haven't gone anywhere or done anything...I have the lifestyle of an older person and I feel older... It is a form of social exclusion, because you can't talk about a lot of the same things... I haven't made personal friends... I don't have much in common with people in a lot of ways because I think they have more agency than I do.

4.8.3 Recurring liminality

Louise felt she had moved through liminality about three years after her diagnosis. At this stage she felt she had "*adjusted and... backgrounded*" her illness. Her passage had been a gradual process over time. Some years later Louise experienced a social turning point in her life which initiated a new transition and feelings of liminality that she was experiencing at the time of our second interview. The organisation she worked for had restructured and she was no longer required to perform a role she valued. This social change disrupted her sense of self. She felt unsettled and lost, no longer in her old role but not yet finding a new one:

I just really am in a funny state of mind at the moment... I can't be bothered doing anything...I'm all over the place...I'm in between states at the moment.

Social liminality also disrupted her physically. She had stopped exercising as much and felt more breathless which engendered feelings of anxiety that she was “going downhill”.

These findings, that the participants created meaning in liminality while also experiencing negative feelings, are consistent with Turner’s (1967, p.96) view of liminality. Turner formed his views from anthropological studies of tribal rituals. He saw liminality as the “betwixt and between” period of transition between stable states, which could include physical, mental and emotional states. He proposed that liminality was a stage of reflection on existence. As the participants of this current study demonstrated, Turner (1967, p.96) claimed that it was “an inward and conceptual process” of transformation with potential for growth and possibility as old patterns are undone and reconfigured, new ideas emerge, and connection with the infinite is experienced.

4.9 Building resilience

The participants’ stories revealed that they built resilience over time through their experiences of adaptation. Life after diagnosis was not static. The women lived with and adapted to their illness in constantly changing environments with the day to day disruptions of everyday life, physical changes related to their illness, and at times more significant social changes. Resilience represented the learning they had actively engaged in which allowed them to ‘move on’ from adversity and forward with life. They had developed knowledge, skills and expertise as they learned to understand the changes in their bodies, and how to independently manage these within their personal context of mind and emotions and changing social worlds. The personal learning and spiritual growth which a number of women experienced while in a state of liminality was a source of meaning and purpose which enhanced their resilience. This accumulated learning achieved competence, confidence, and mental and emotional stability to restore coherence to their lives. With each turning point and transition new knowledge and skills expanded their resilience. Resilience could be seen in the positive feelings of wellness, control and normality which the participants experienced as they integrated their illness into their lives, and in their ability to adapt to changes that occurred as their illness progressed.

Vidu, who continued to avoid anything related to LAM in her life after diagnosis, adjusted to her illness in a way that was consistent with her own life meaning and view of herself.

This was effective for her at this stage while her disease was stable. However, in not engaging in her illness and learning about it, she did not develop the same resilience as the other women. In Chapter 6, Living with Advanced LAM, Vidu demonstrated the effects of this as her illness progressed.

4.9.1 Resilience through adaptation

The participants who had passed through transition and adapted to their illness displayed qualities that revealed that they had built their resilience through their experiences. Firstly, they felt generally positive about their life and experienced subjective wellness even while living with an incurable and life threatening illness. They had come to terms with their illness and modified their goals to reflect both their new reality and their personal life meaning. There was now a sense of being able, once again, to achieve goals related to exercise, activity, work and social participation. Sarah was able to go skiing and enjoy the theatre with friends:

The skiing was for three days... it was a real win cause I thought I wouldn't do it so, as far as I was concerned, if I could go that was great...

We were at the theatre...there's two big flights of stairs...I felt so proud of myself. I got to the seat... and thought, wow, that's good. I was okay and I didn't make anyone stop.

Anna was “going out and seeing my friends a lot more...I'll initiate that, whereas I didn't before”. She felt confident to shop on her own, “I find different ways around it now”, and consider changing her job:

That little bit more confidence... I'm looking at wanting to change my work... maybe I should do something I love... I'm getting to do a lot more of what I like. ...I do feel a lot more energetic...I feel a little bit more balanced.

Secondly, they demonstrated that they had retained their agency and regained a sense of autonomy and control in their life through their successful self-management and self-advocacy. In being independent researchers of their condition and managing their illness over time, they developed competence and knowledge which raised their self-efficacy by giving them confidence in their ability to manage problems that might arise in the future. They understood their bodies well and were able to distinguish between minor issues which they could manage themselves and more serious matters that required medical attention. While they remained diligent in attending regular medical check-ups there was less need for

medical attention between reviews. Mia had mastered managing her symptoms and was able to avoid a hospital admission:

I know the symptoms, I know my body, I know what I have to do... I used a nebulizer...and prednisone and antibiotics. A little bit more puffing and looking after myself here at home... I didn't end up in the hospital... I think its practice... So that was good.

Anna showed how her confidence had grown over time:

I have a bit of sternum pain, but then I think about what I've been doing in the last week and, is it really my lungs, or is it just muscular around that area? So just as time's gone by, not to automatically think, okay it's my lungs...beforehand I would have been a lot more alarmed... Knowing that it's not something huge or major... As the time's gone, that's trying to just keep things in perspective... I got that sense of now, looking out for myself as well and knowing I can rely on myself.

Thirdly, resilient women had reconstructed normality in a personally meaningful way and restored their sense of self, balancing the negative impacts of their illness. They did this through their personal agency in self-managing their illness within their individual social contexts. Gaining a sense of control and independence contributed to feelings of normality, coherence and being oneself again. Anna no longer relied on her family for practical support:

I see them weekly or fortnightly but it's nowhere near as often...Mum used to help out with the cleaning, I got a cleaning lady so she doesn't do that anymore... I feel like I've taken back that independence as well...When I do see them it's for family things, not because I need them to do something for me...Doing a lot more on my own, which makes me feel like that's me again.

There was a feeling of mental and emotional stability in contrast to the instability they experienced at diagnosis. Some further enriched their sense of self and enhanced their resilience through the personal growth they experienced in liminality. Aiko, Anna and Eva demonstrated spiritual growth and transformation and the peace, calm and acceptance that flowed from this. Accepting their illness enabled them to remove their focus from the illness itself and, in conjunction with successful self-management, 'move on' from adversity to direct their attention to possibilities for their life and outwards towards others, engendering empathy and a motivation to altruism. Ursula "*just carried on with living... I just accept*

it...I don't think of the future with LAM, I just think of the future with me". Eva revealed how she felt transformed by her illness:

It's (having LAM) actually quite empowering too... because that's what I think really started this process of me accepting who I am and what I can do and what I can't do. In finding my own limits I became a lot happier person...stopping focusing on myself completely and actually focusing on helping others.

Sarah's reflection illustrates how she had integrated her illness into her life over time:

I think it takes a long time to process and to get comfortable with the whole concept and to understand.... I try and most days forget about it... Even if I get breathless, I don't think about it directly. I think I've just accepted that that's where I'm at... It is just part of life now and you take it on as part of your person... it's not overwhelming... I think in perspective, it's probably because it's managed to be staying relatively stable where it is... Where it seems to be hovering, for now, allows me to do most of the things that I want to do... I really feel I got a large part of me back.

Anna demonstrated how her resilience developed through reflection, personal agency and her everyday life experiences of social connectedness eleven years after her diagnosis. In the two years between her second and third interviews the anxiety and fatigue she had been experiencing had settled and she had remained physically stable. At her third interview she identified that this was a period in which "I've built up my resilience". She attributed her resilience to her young age, her personal agency and the passage of time which opened a space in which she could reflect on her experience to view it in a more positive light. While she acknowledged the role of sirolimus in improving her quality of life, it was her own agency in successfully dealing with her anxiety and reconnecting with her social world that she felt had had a greater impact on developing her resilience:

As much as it's a bit of a bitter twist, to get this thing when you're younger and I can't have children... it's a good thing for me because I've built up my resilience... I think that sirolimus has given me my quality of life... but it's also to do with, I think, having hit that anxiety wall...and to deal with that and have that reflection, reassessment... getting out again and putting myself back connected with the world...Not thinking, oh I can't do this because I have

LAM... (I'm) a lot more independent. I'm a lot happier. I think I'm just a lot more peaceful...Just okay with me.

4.9.2 Resilience and illness progression

Resilience was evident not only in the positive improvements the participants achieved but also through difficult physical and social turning points as they experienced illness progression. Margaret's story illustrates her resilience at the first significant turning point after her diagnosis when she was forced to stop work due to the progress of her illness. She demonstrated her decision-making and personal agency at this time and how her personal life meaning and social context influenced her ability to adapt to her changed situation.

In narrating her life story Margaret revealed two significant aspects of her life meaning. Firstly, she regarded herself as a "mother hen", reflecting her primary life goal of being a mother, her love of children and animals, and her sense of purpose in helping others:

I've always had it more with children and animals... because they can't defend themselves... I always stood up for my friends. If someone hurt them or insulted them they had me to deal with...I'm mother hen.

Secondly, in telling the story of a conflict with a teacher when she was at school, she identified herself as independent and a "fighter". "If I thought something wasn't right I would speak my mind...I do not stand back and let people walk over me...she had a fighter on her hands". Throughout her illness, Margaret's attitudes and actions consistently reflected these meanings.

LAM had begun to impact more seriously on Margaret eighteen months after her diagnosis when her lung capacity had dropped to 60%. Although she did not yet need to use oxygen therapy, she was forced to finish work. Stopping work represented significant loss. She lost her social world at work, her identity as a valued, expert worker, her main daily activity, and her financial independence. As a single woman living on her own, the financial impact of stopping work was considerable. Margaret could no longer afford to stay in her apartment and was forced to go on to a disability pension and to apply urgently for Department of Housing accommodation. A sense of powerlessness accompanied these losses:

The hard thing was when I had to stop work cause I was doing 80 hours a week...my breathing would go because an employment agency is very fast paced... I just couldn't do that anymore and that was getting frustrating... I was raised to be able to take care of myself... so I was very much, I'm going

to do this and no one's going to stop me, and then of course my body's stopping me.

Margaret's "fighter" and "mother hen" aspects of self and her past experience of successfully managing her asthma over time countered the disruptions and reduced choices now available to her. She demonstrated agency and maintained a sense of continuity in her life by continuing to live alone and manage her illness independently even as her health deteriorated:

I like living by myself cause I don't have anyone saying, Are you ok? Do you need anything? Cause it's like, No I'm fine, if I need anything I'll get it... I hate having to rely on people.

Margaret had viewed herself as a competent person at work and preserved this identity by presenting herself as expert in coping with LAM. As soon as she became breathless and anxious she would calm herself by reading, drawing, listening to music, or meditating. These strategies also relieved the boredom she felt being at home after she stopped work. Her optimism was evident in her positive attitude and refusal to take on a 'sick' identity or regard herself as a victim:

It's easier for me because I have the disease. I'm living with it. I understand how to cope with it... I'll have a day and I'll just cry and cry and cry. But it's a good release because it just gets all that stress out ...I'd rather be happy... I'll get the home care for cleaning because I can't do that. Yes, you need that help, that assistance. But for your own sanity and your own state of mind... you have to stop looking at yourself as a victim... That's just self-pity.

Margaret chose to direct her mothering instincts to animals. She became "mother hen" to two small pet dogs and took on the work of caring for dog rescue puppies until they were found a home. It was a role where she was in control and gained esteem through her expertise in caring for the dogs. It was consistent with her goal of being a mother and gave continuity to her life.

Margaret showed resilience in adapting to this next stage of her illness through her positive, independent attitude, her humour, her self-management strategies, and redirecting her energies in a positive and creative way towards her hobbies and her pets. Margaret's story will be explored further through later turning points in Chapter 6, Living with Advanced LAM.

4.10 Summary

This chapter has presented the participants' experiences of learning to live with LAM in their everyday lives after diagnosis at the mild to moderate stage of their illness. It was a period of transition in which they adapted and integrated their illness into their lives over time and built their resilience. Adaptation was a dynamic process of learning to manage their illness physically, and through their interactions within their social world of relationships and responsibilities, and their personal world of emotions, self and identity.

Each participant's personal life meaning motivated and directed their actions to adapt to their illness and achieve wellness and normality. In particular, the women were motivated by their prior health and fitness, their relationships, and a desire to fulfil their responsibilities, and participate in family, work, and social life. In integrating their illness into their life the participants accepted their situation and reframed their expectations and goals so that the meaning of living with their new limitations integrated with their life meaning.

As part of their life meaning, the women held core self-efficacy beliefs, derived from their previous life experiences, when they were diagnosed. Those who had successfully coped with adversity or illness in the past had high levels of self-efficacy which enabled them to more readily accept and adapt to their limitations. Lower self-efficacy beliefs posed a barrier to adaptation as those participants felt less confident in managing the challenges of living with their illness and perceived themselves as lacking agency.

Most participants actively took responsibility for their health and developed knowledge, skills and expertise as they learned to manage their illness. The rarity of LAM meant that a number of participants also developed enhanced skills of self-advocacy. The women engaged in seeking knowledge of LAM and became independent researchers. Over time they came to know their changed bodies well and learned through experience to manage their breathlessness. Fatigue was more difficult to manage and was not addressed by their physicians.

The emotional and social impact of LAM was more difficult for the participants to manage than their physical symptoms. It was hard to elicit emotional support from others when externally most appeared well and healthy. The women were in a paradoxical situation of being both providers of care and recipients of support and this generated complex, paradoxical emotions. Their relationships and responsibilities were a positive source of

meaning, purpose and motivation to manage their illness effectively and live well, and they appreciated the support they received. However, the disrupted equilibrium they experienced, in being unable to fulfil their roles in the usual way and needing extra support, threatened their identity and created negative feelings of vulnerability, guilt, and being a burden. Participants who felt able to communicate openly and perceived empathic emotional support from their partners were able to resolve negative emotions. Younger participants indicated that they received limited and often conflicting advice from their physicians on their sexual health and contraception and these were significant areas of concern and confusion. While the majority of women were supported in their workplaces when they advocated for flexible working arrangements, receiving support engendered similar conflicting feelings. Two women encountered stigma and a lack of support from their managers.

The women managed their emotions when they accepted their need to adjust their expectations and goals to accommodate their illness and focus on what was possible, including reducing their work hours or changing roles. The independent strategies they employed to care for their health also helped to balance their emotions. These variously improved their breathing, mood and energy levels. Being able to effectively manage their symptoms over time contributed to physical stability, strengthened their self-efficacy and engendered positive feelings of stability, autonomy, control and wellbeing. Sirolimus medication was an important factor for many participants in achieving physical stability and improved quality of life and subsequently positive emotions and a sense of hope. Engaging home services reduced the need for support from partners and family and associated feelings of guilt and dependency, and increased their sense of autonomy. Similarly, experiencing social connectedness created positive feelings of belonging, normality, and self-worth.

Liminality was, for some participants, a significant inner process of reconstructing their disrupted sense of self and finding meaning through personal and spiritual growth. The women who experienced this were able to accept their situation and felt a calmness and peace which allowed them to live with contentment in the present. They felt a connectedness to others and found new meaning and purpose which they expressed in altruistic activities such as volunteering work and supporting other women with LAM. These liminal experiences engendered positive feelings and emotional stability and facilitated their adaptation.

The participants lived with their illness in interacting environments of constant change. For most of the participants these were the daily fluctuations of ordinary life punctuated by

temporary isolated disruptive events related to exacerbations of their illness or social situation. Others experienced new social or physical turning points which initiated new transitions and new experiences of liminality to generate new understandings.

The participants' ability to alter their expectations and goals and act to achieve these as they managed their symptoms, emotions and relationships and responsibilities was a demonstration of agency. This finding is consistent with Bandura's (2001) social cognitive theory. Bandura (2001, p.2) defined agency as one's ability "to intentionally make things happen by one's actions". He theorised that agency was characterised by core features, including intention, self-regulation, and self-reflectiveness on one's abilities and life meaning, purpose and goals. As this current study demonstrates, Bandura viewed agency as functioning dynamically within sociostructural contexts as thought processes, emotions, biological events, behaviours and environments interact and influence one another. Further, he saw agentic action as the means by which people expand knowledge and competencies and adapt to limitation and a variety of environments. According to social cognitive theory, self-efficacy, a person's belief in their ability to exercise control over their own behaviour and environmental events, is the core of human agency.

The participants built their resilience through the process of adapting to their illness. Resilience represented the meaning, knowledge, skills, and positive beliefs they developed through this learning process and other life experiences. It enabled them to move forward with their life even while living with the uncertain, incurable and progressive nature of LAM. The women demonstrated their resilience in the mental and emotional stability they achieved through successfully managing their physical symptoms and limitations, emotions, and relationships and responsibilities. They felt a sense of wellness and control, and were able to reconstruct their sense of self and normality in a meaningful way. Resilience gave them the ability to deal with turning points of disruptive change as they occurred.

4.11 Conclusion

This chapter has presented the participants' experiences from diagnosis to the moderate stage of their illness as they learned to live with LAM in their everyday lives in a period of transition. The women demonstrated that this was a multidimensional process of adaptation which encompassed learning to live with their altered bodies and manage their emotions, relationships and responsibilities in the context of constant change, in the fluctuations of daily life and more significant changes related to their illness or social situation. As the women accomplished this, they restored their sense of self, integrated their illness into their

lives in meaningful way, and built resilience. They showed this in their ability to experience wellness, and a sense of autonomy, competence, and coherence while living with the limitations of their incurable, progressive illness.

In adapting and learning to live with LAM, the participants needed to develop communication skills in their encounters with healthcare professionals in healthcare settings. The following chapter focuses on their experiences of interacting with healthcare professionals at their regular reviews with their physicians and during hospital admissions, and how these were influenced by rarity of LAM.

Chapter 5

INTERACTING WITH HEALTHCARE PROFESSIONALS

5.1 Introduction

The previous chapter presented the participants' experiences as they learned to live with LAM in their everyday lives following diagnosis. As they adapted to their illness they developed knowledge and competence through their independent research and self-management. They brought their expertise to their encounters with healthcare professionals who frequently lacked knowledge of LAM. This chapter presents the participants' experiences of interacting with healthcare professionals, as patients in the hospital setting and at their periodic reviews with their physicians, and how their interactions were affected by the knowledge and attitudes of both parties, and the rarity of LAM.

The participants were admitted to hospital when their health status changed. This occurred for isolated surgical procedures for particular issues, periodic events such as pneumothorax or chest infections over the course of their illness, or critical events such as respiratory failure or receiving a lung transplant. The most common reason for the participants being in hospital was for a surgical pleurodesis [n=7] to treat a chylothorax [n=3] or pneumothorax [n=4]. Of these, two participants had also had a previous nephrectomy (Veronica) and partial nephrectomy (Julie) for renal angiomyolipomas. Ruth had multiple embolisations for her renal angiomyolipomas. Mia was hospitalised periodically for chest infections. The participants were also admitted for procedures unrelated to LAM, including cholecystectomy (Helen), removal of ovarian haematoma (Clare), and appendicectomy (Anna). They were in hospital for varying periods from overnight to six weeks, with the exception of Clare whose prolonged hospital stay for persistent chylothorax in both lungs lasted for five months.

While Clare was not the only participant to experience a hospital admission, her hospital stay was the longest and she experienced a prolonged period of contact with healthcare professionals in this setting. It constituted a critical event which was not only physically disruptive but also brought about significant social and emotional change for Clare. Clare's story is presented to provide insight into the hospital experience as an aspect of living with

LAM, and illuminate how the rarity of LAM affected her experience and how she developed resilience in dealing with the particular issues she faced in this context. This is consistent with the life history approach of “intensity sampling” to give a deeper understanding of specific aspects of experience (Plummer 2001, p. 133). No literature has explored the experiences of women living with LAM or other rare diseases in the hospital environment or how people show resilience in this setting.

Figure 5.1 summarises the themes related to the participants’ experiences of interacting with healthcare professionals. Firstly, the participants revealed the impact of being a patient in hospital with a rare disease and how they responded in this situation. Secondly, Clare’s long hospitalisation demonstrated how she developed resilience and experienced liminality in this context. Thirdly, the participants’ interactions with healthcare professionals are explored in relation to the power balance that existed between them and how it was influenced by the rarity of LAM and shifted as knowledge and attitudes changed. Table 3.3, Appendix 3, shows the medical and surgical treatments the participants received.

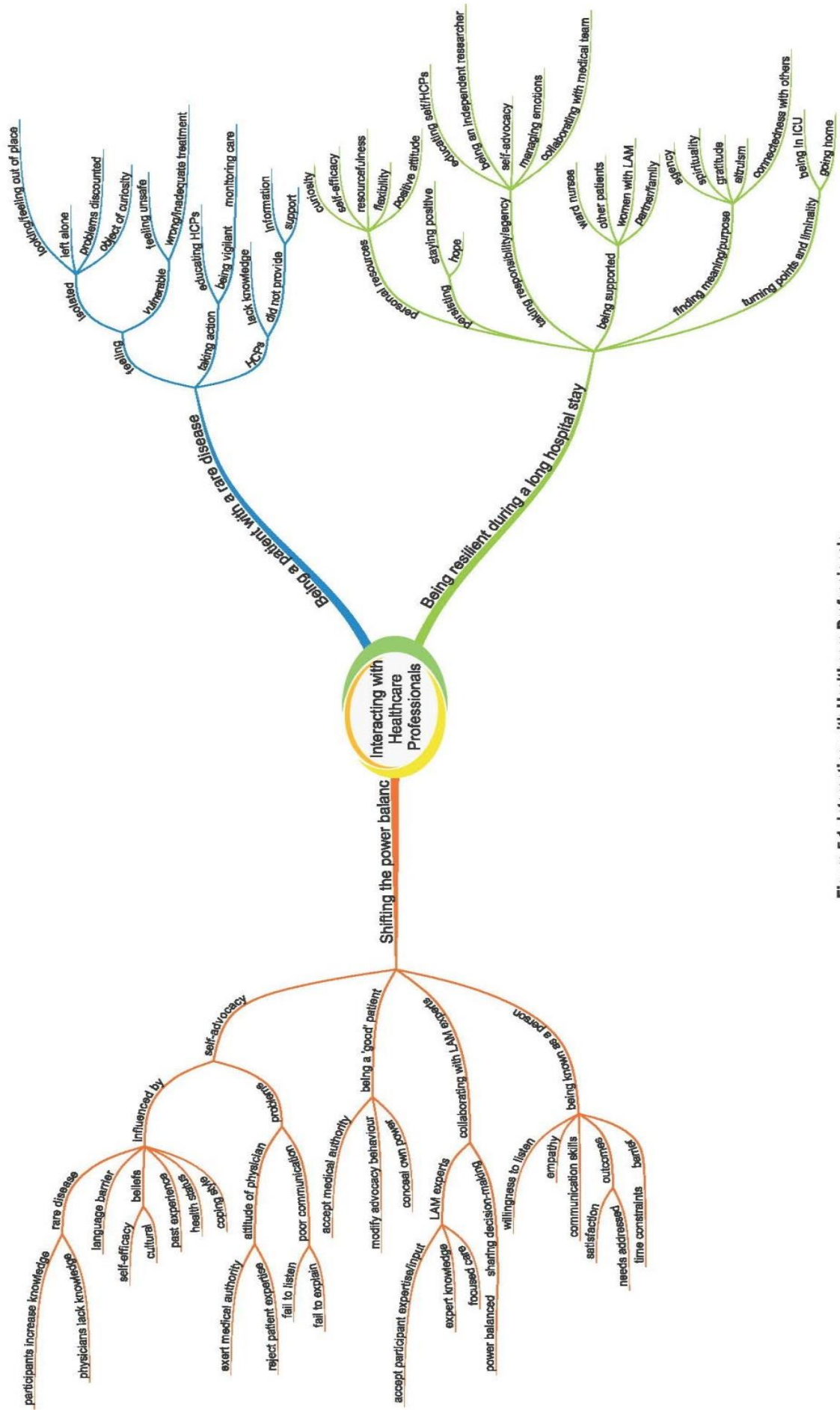


Figure 5.1: Interacting with Healthcare Professionals

5.2 Being a patient with a rare disease

Being a patient with a rare disease created feelings of isolation and vulnerability as the participants interacted with healthcare professionals in the hospital setting. The healthcare professionals caring for them frequently knew little about LAM and were unable to provide adequate information and support. There are only two medical centres of expertise in LAM in Australia and women living a distance from these were isolated and relied on their physicians to communicate with LAM experts for up to date management. When they were in hospital the participants found they needed to be alert and vigilant, and take on the role of educating the healthcare professionals. It was a stressful experience which required effort, and two participants emphasised this in their use of war metaphors. Jess referred to being in hospital as “*a battle for survival*” and Clare as “*going to war*”.

5.2.1 Feeling isolated and vulnerable

Entering the hospital setting with a rare disease was an isolating experience for the participants. Their outward appearance of good health did not correlate with the expected presentation of a person with chronic respiratory disease. As Jess said:

(The doctor) was obviously really confused to see me, because I think looking at the pictures and such an enormous mass (her abdominal lymphangiomyoma), they were expecting probably somebody... who could barely stand, who was really emaciated, obviously sick, but when he saw me, I looked pretty normal.

In this context the women looked out of place and felt their problems were discounted. They found they were ignored and left to look after themselves. Veronica felt she “*was just really left to my own devices because I never complained.*” Ruth said:

You have to look after yourself in hospital... You feel quite isolated being stuck away... You spend a lot of time alone.

Helen felt she was treated impersonally and as an object of curiosity:

The oxygen... was freaking them out a bit I think and they didn't know anything about LAM, so in between I had to keep explaining and then I just started taking leaflets every time I went. I just said, read that... Then they started whispering about it and I could hear, this lady's got some really weird, rare disease.

It was obvious to the participants that the healthcare professionals who were caring for them knew little about LAM when they seemed excessively worried and sought information from non-medical internet sources such as Wikipedia. Ruth *“had these teams of people coming around who didn't have a clue about LAM and had Wikipedia... generally there's great ignorance”*. In addition to creating a sense of isolation this made the women feel vulnerable to receiving wrong or inadequate treatment. Mia *“hoped that they would have control because you're in their hands... You feel, oh no, I don't trust (them)...They literally don't know”*. They felt unsafe when they were uncertain if healthcare professionals understood the significance of their oxygen saturations and lung function tests, or were able to manage their care appropriately. When it was possible the women preferred to always attend the same hospital where a LAM expert was available. Mia chose *“never go anywhere else because I know they have no idea what to do with me”*.

5.2.2. Being vigilant

The participants felt particularly vulnerable on general surgical wards where nursing staff could be unfamiliar with LAM-related issues and how to manage oxygen therapy and pleural drains. The women felt a need to be vigilant in monitoring the care provided to them. When Jess was admitted for treatment of her chylothorax she realised that the nurses caring for her did not understand the implications of the amounts of drainage they were observing in her drains. She felt safer with nurses who demonstrated competence and confidence:

The nurses...were always congratulating me, isn't it good, another two litres today. I was really afraid and hated to hear how much was draining...they obviously had absolutely no idea about what the thing (chylothorax) was and some of them looked quite intimidated... from then on I knew it was going to be a battle for survival at this hospital. They really didn't know what to do... There were...nurses who were really great...The difference wasn't really just that they had a nicer manner... it was that I actually felt safe with them. I felt like they weren't going to kill me. That they knew how to use all the equipment...I actually think when you're in hospital - you know everybody gets so worried about do they have the best surgeon but... I think probably the quality of the nursing and the recovery is much more important.

Helen felt a need to monitor her own oxygen saturations, explain her condition to nursing staff and advise them how to manage her oxygen therapy:

My sats were really, really bad and I was checking all the time on the monitor... I said, can't you just put me back on nasal prongs? The minute they put me back on nasal prongs my sats went right back up again... They were taking my blood pressure at the same time as taking my (sats)... They were all inaccurate, all the readings, because when I started checking them myself they were actually normal for me...It's that frustration that people didn't understand what was wrong with me and I had to explain to them.

5.2.3 Educating the professionals

The participants found themselves educating the healthcare professionals who were caring for them. It was a burden to be always having to explain their condition. Jess felt “*it's awful to be an interesting patient, because I'd rather be just quickly diagnosed and treated and out of there*”. Anna wanted to be “*a patient like any other patient*”.

Some women took brochures provided by the LAM peer support organisation to hospital and distributed them to staff to raise awareness of their condition. Veronica “*photocopied, from the internet, what it was, and up to date information, and I gave it to them... There were middle aged doctors that didn't know*”. They also made themselves available to be interviewed and examined by medical students and junior medical staff for education. Margaret said:

I don't mind taking time to explain it... I tell them to bring the trainees in and ask me 5000 questions...I give them all pamphlets...I'll say, do you want to check the lungs?...I'm sure there won't be a cure in my lifetime. But if there's something I've contributed to that maybe will help...I'm more than happy to get poked and prodded.

5.3 Being resilient during a long hospital stay

Clare's story revealed her resilience and experiences of liminality over the course of her prolonged five month hospital stay. Clare was admitted to a local hospital for persistent significant collections of chyle (effusions) in both lungs two months after her diagnosis. She was later transferred to a larger hospital interstate. Clare's doctors had not encountered a case such as Clare's before and they struggled over the course of her admission to find an effective treatment for her problem. Clare underwent various procedures and surgery (multiple drain insertions, lymphangiograms, pleurectomy and pleurodesis). She also

received all her nutrition via an intravenous infusion (total parenteral nutrition, TPN) for 104 days to reduce dietary fat and the formation of the chyle which was leaking into her lungs. None of these measures were successful. Over the course of her admission Clare had multiple medical emergency calls (Metcalls) for low blood pressure, low oxygen saturations, and breathing difficulties. She was admitted to ICU (intensive care unit) four times.

Clare's experience was characterised by uncertainty and isolation. Not only did she feel isolated by her rare condition in the hospital setting but she was also geographically isolated from her partner, family and friends in an interstate hospital more than five hours drive from her home. The complexity and rarity of Clare's particular issue added a further layer of uncertainty to the vulnerability felt by other women with LAM when they were in hospital. She was uncertain as to the outcome of her situation and her future and her doctors had no experience with her rare problem and did not know how best to manage and resolve it:

The transplant team I was under... said Clare, honestly, we don't know what to do. We've never seen a case like this...we're scratching our heads, we don't know how to stop this.

Clare needed to be resilient to endure her unusually long hospitalisation. Throughout her hospital stay she was far removed from normality due to the acute and unpredictable nature of her medical problem and her frequent Metcalls and admissions to ICU. She was constantly aware of her illness. In this context Clare's resilience enabled her to endure the instability and uncertainty of her situation.

In her life story, Clare identified that her personal traits and past experiences, as aspects of her life meaning, were resources which contributed to her resilience. Her inquisitive nature stimulated her to research information on LAM and possible solutions to her problem. Although she had not previously experienced adversity or illness, she had travelled extensively and worked in a variety of jobs and locations before her diagnosis. These experiences had developed her resourcefulness, flexibility and self-efficacy. Clare's resilience was evident in her positive attitude, her persistence, and her actions to take responsibility for her physical and emotional health. Being supported by family, nurses, other patients and women with LAM also contributed to her resilience.

5.3.1 Staying positive and persisting

Clare likened her situation to an experience she once had of skiing through an unknown

landscape. Her metaphor illustrated her persistence and determination to “keep going” in spite of the uncertainty she faced:

Some stages you get to some spots in your life - you don't have any option but to keep going because you're in the middle of nowhere...I just wanted to keep going.

In her difficult situation Clare cultivated positivity to maintain her hope that her problem would resolve and she would be discharged home and be well. Her young age motivated her to return to her previous good health. She was also motivated to manage her emotions to protect her parents from additional worry in the context of her father being seriously ill at the same time. She maintained a positive demeanour with the staff and allowed herself to cry only in private:

I just had to deal with it...I think I was so positive through the whole ordeal to some extent, and I didn't really cry much at all, I just thought, take each day as it comes...you try to be positive to the staff, but when they'd probably nip away I might have a bit of a sob.

5.3.2 Being supported

Clare was supported through her ordeal by the relationships she established with the ward staff, other patients, and two women with LAM. Over such a long period of time the hospital ward became her ‘home’ and being cared for consistently by familiar and friendly staff assisted her to cope. In contrast, her ICU experience was one to be endured. For Clare, being in ICU, with additional equipment and monitoring, signified illness and a move backwards from going home and wellness. While she appreciated that the staff did “*amazing things*” there was not the consistency and familiarity with the staff and she did not receive emotional support from them. This contrasts with Veronica’s experience in ICU after her surgical pleurodesis. Veronica was focused on her physical safety and felt “*the intensive care nurses... did their job well. You were always watched...They'd come in and look after you*”. The difference here was that Clare needed a higher level of emotional support to deal with the instability and length of her hospital stay. She felt this was better provided by ward nurses who had formed a rapport with her over time and knew her as a person:

When I was ICU... that was a really hard time when I was on the high flow and connected to everything. I couldn't do anything...ICU's different because it's a bit busier. I think I hadn't really formed a relationship with the staff as

much because they would always swap; you'd have a different staff every shift... So I found that a little bit hard, because you don't get the rapport with the staff as much.

While Clare's family and partner were unable to be with her consistently they provided moral support. In particular, Clare regarded her partner as “*overwhelmingly supportive ...my rock*”. Clare also received moral support from another patient whose advice to stay strong became a guiding mantra for her throughout her hospitalisation:

I think the biggest thing I got told by that lady was keep strong; you've got to be strong - that really rings true because you feel a bit like, sometimes, you're going to war...She really kept me determined to be a bit tougher.

Clare had made contact with the peer support organisation and was particularly supported by two other women with LAM. Firstly, they instilled hope and symbolised the possibility of wellness while living with LAM through their positive attitude and successful management of their own LAM. Secondly, they visited her frequently, were a source of information and, in the absence of any known effective treatment, played an active role in researching possible solutions to resolve her persistent effusions. Clare was inspired to be positive by one of the women in particular:

She was just so positive. She was not down about being sick. She was improving. Her lung functions had improved...She did a bit of meditation and just had her lifestyle in a good shape; so I'm like right, I'm jumping on her back and I'm going to do everything she's doing... It was good because she wasn't whingeing about it. She wasn't moping, going oh, I'm going to die... she was strong... I'd ring up and ask her all the questions.

5.3.3 Taking responsibility

Clare took responsibility for her emotional and physical health during her admission. Although confined in hospital, she set goals, remained vigilant in monitoring her body and the care being given to her, managed her emotions, actively participated in researching treatment options, advocated for herself, and collaborated with her physicians in clinical decision-making. While it was an added burden to take on the responsibility of finding her own solution and oversee the care given to her, it empowered her, gave her a sense of control, and raised her self-efficacy and self-esteem. In turn, perceiving herself as having agency in influencing the management of her health increased her resilience.

Clare had realised when she was diagnosed that there was no information or help available for women with LAM. She determined there was a need to produce brochures that would provide information for newly diagnosed women and healthcare professionals and she set a goal to raise money for this purpose. She had started organising a large fundraising event and continued to do so during her hospital admission although she was unable to attend the event herself. She also determined that she would need to independently research LAM herself and, as her effusions persisted, search for alternate options for treatment that could resolve her problem. Setting these goals gave Clare a sense of purpose and a clear focus which helped her to cope by keeping her occupied and feeling useful:

I was more clued up... I was like right, can we just perhaps do this this way? ... It probably made me more determined to find out even more. I think my way of coping was just to find out a lot... to put all my focus on that... It gave me a bit of drive... They really didn't know much either... I'm pretty inquisitive. I like to ask lots of questions, so I was always wanting to know how this worked or that worked...I did feel quite involved with it and...that made me feel a bit worthwhile in what I was doing.

Clare educated herself, accessing the internet from her hospital bed, searching for information about LAM and, with the assistance of her LAM friends, researching medical articles for possible effective treatments for her persistent effusions. They also initiated communication with other LAM experts overseas:

They're just like we don't know how to stop this. So then we looked further...We came up with a whole list of possibilities of surgeries...So at this stage - because we didn't have any options - we presented this to the surgeon.

As she acquired knowledge Clare also educated healthcare professionals about LAM. She made herself available to groups of medical students for examination and to answer their questions.

5.3.3.1. Advocating and collaborating

Clare advocated for herself in communicating with her medical teams. Firstly, she advocated to be transferred interstate to the hospital which was a centre of LAM expertise where she had greater confidence that she would be managed according to latest research. Secondly, during her long admission, in the context of her physicians struggling to find a solution to her problem, the knowledge she gained through her research empowered her to voice her

opinions. In this way she collaborated closely with her medical team in making decisions concerning her care. She advocated to restart sirolimus medication when all other treatments had failed and then to increase the initial dose which she felt, from her research, was inadequate. After receiving it the effusions gradually resolved and, although it was uncertain why, Clare and her physician believed it contributed to the positive outcome:

Sometimes in hospital I was on it (sirolimus), but they didn't want me on it very often because it lowers your immune system... even though I was begging them to put me back on it... my thought was, after lots of research, that (sirolimus) will help... They only put me on one milligram every second day. I was like it's (that dose) not going to help... towards the very end I was on it. Anyway, no-one really knows why the chyle effusion stopped, but I think it's helped... (My doctor) believes... that the sirolimus helped.

Clare was so involved with the clinical decision-making aspect of her medical care that she thought of herself as part of the medical team and this was reflected in her language:

Sometimes we aspirated the tube.

We tried Fentanyl as well.

It just kept reducing the fluid. Then this one got down to 50 mls, so we pulled the plug.

We did a laparotomy.

5.3.4 Turning points and liminality

Clare's long hospitalisation and the immediate period after discharge was an experience of liminality and multiple paradoxes. The unreal space of the hospital became 'home' to her yet was not her home. Her status was ambiguous - she was a patient yet, in researching for solutions to her confounding medical problem, at the same time thought of herself as part of the medical team. She was 'sick' but worked to organise a fundraising event for LAM research at which she could be present only in a technological space through Skype. She was present but not present. Her physical condition was unstable and she shifted unpredictably between the spaces of the ward and intensive care. Her feelings of isolation and uncertainty were indicative of her liminality.

Clare's final stay in ICU was a turning point. Believing she was improving and would be discharged soon, her ICU admission was regressive and Clare felt she had lost hope when time seemed to have lengthened with no perceivable end to her problem. It was her "lowest

point” and difficult to endure. Paradoxically, however, it was also a positive turning point in shifting her perspective by providing the impetus for her to set a new goal and plan of action to renew her hope and remain resilient:

I'd thought I was getting better and I was going to get out. It just dragged on for such a long time. I just sobbed because I was hooked up to everything...It was just like I just want to get out (of ICU). I sobbed in front of my parents as well. That was the first time they'd really seen me have a good cry, because I'd just had enough...I was begging to go back to the ward... I just couldn't bear it. I knew that I was just not improving there.

Clare decided she was going to recover by establishing a firm time frame for going home. She supported her goal through her personal spirituality and consciously renewing her positive attitude. She was supported by her parents and nurses from the ward who visited her in ICU. Her agency was evident in her actions of visiting the hospital chapel to “pray and wish” for her recovery, starting a journal to record what she was grateful for each day, and surrounding herself with positive affirmations. These actions provided a sense of control and balanced the negativity, uncertainty and powerlessness of her physical situation. Mentally and spiritually choosing hope was sustaining and contributed to her resilience. In turn, being resilient and continuing to cope maintained her hope for a positive outcome:

So after that instance I was, right, I'm going home; so everything changed... I told everyone I'm marching out in March... I've decided... I've just had enough. I don't want to be here anymore... Because you're in there for such a long time, and it's hard to remain really positive. So I started a little diary up. Whether it's the cleaner giving you a laugh... or someone making some joke. I think that's a good thing to do because... you don't want to be negative while you're there because it doesn't help... I think definitely it helped to believe that you could get better... I have this belief now that you might get worse if you don't change that belief.

After her ICU admission and treatment with sirolimus Clare's effusions gradually reduced, her drains were removed and she was discharged home in the time frame she had set for herself. While the sirolimus treatment seemed to have been the likely reason for the resolution of her problem, her positive attitude and actions sustained hope and resilience in the final stages of her ordeal.

Clare's discharge and passage from hospital was a turning point which initiated a new transition. She had experienced a separation from normality to illness and was now at a threshold where she had left the hospital world but had yet to integrate her illness to a new wellness. Positively, her discharge represented physical recovery and stability. However, paradoxically, it simultaneously shifted and intensified her liminality to an emotional and social level. She was no longer a patient yet not her "usual self" either. She felt like she was returning from "war" and had not adjusted to life outside the hospital. She lacked confidence and felt "wilted". Her relationship with her partner was confused by vestiges of patient /carer roles creating tension and eventually a breakdown of the relationship, "that patient-carer relationship, it has a big impact on you". This was a significant social turning point which Clare found more difficult than her diagnosis. "When he left I probably got tearier than what I did when I found out I had LAM". She felt marginalised among women her age by her inability to have children. She returned to live with her parents and felt stripped of relationship, work, and her own home. Clare identified her liminality at this time as being in "limbo".

It is hard because lots of friends are having their babies. You think this is just my life... when I get older I won't have that extra support around... My job's finished... it was so active, not having that either. So I'm in a bit of a limbo patch at the moment.

Although Clare experienced uncomfortable feelings in her liminality it was also a reflective period to ponder a meaning for her life going forward. As other participants had experienced, her liminality was a time of spiritual and personal growth. She felt a new sense of altruism and empathy for others, a desire to offer support to other women with LAM, and an interest in exploring other spiritually based modes of healing:

Before I was sick...I didn't have a full understanding; whereas now I know ...You see how vital having a good belief system is as well - that it affects you...like I said, I'm in that limbo stage now; what am I going to do? Hopefully, I'd like to - down the track - do something that I'm proud of, that will make a difference... I speak to other women that are diagnosed. I found that really rewarding...I'm quite open to different spiritual things. Probably more so after (my experience)... it just opens your eyes up to life itself... just open to other ways that people can maybe be healed...there's potential that you can help your body a little bit more, whether it's just mind over matter or just feeling good

inside yourself.

Clare demonstrated a number of factors that promoted her resilience. Her motivation and personal resources of curiosity, flexibility, resourcefulness, self-efficacy and a positive attitude stimulated her actions to take responsibility for her health. She retained agency even in the context of her unstable physical condition and, in the process, gained knowledge and skills and reinforced her self-beliefs. In addition, she maintained hope, found meaning and purpose, and felt positive emotions in her agency, spirituality, gratitude, and connectedness with others. Supporting these findings, Kashdan and Rottenberg (2010) proposed that curiosity motivates a person to be engaged and find meaning in their actions when in situations of complexity and uncertainty. They suggested, furthermore, that this enhances a person's psychological flexibility to promote resilience and the ability to adapt to changing demands. Emmons and McCulloch (2003) found, in a study of gratitude and wellbeing, that gratitude facilitates coping with stress and adversity and benefits emotional health by increasing positive emotions, optimism and a sense of connectedness to others. Tang and Anderson (1999), in two qualitative studies of women living with diabetes, reported that when their agency was diminished by illness the women reclaimed it by relying on themselves, understanding their bodies and taking responsibility for self-care.

5.4 Shifting the power balance

The participants' narratives revealed a shifting power balance in their interactions with healthcare professionals. Power related to the differing abilities of both the participants and their physicians to influence and control behaviour and health outcomes affecting the participants. The women's stories demonstrated a relationship between knowledge, power, attitude, and the rarity of their condition. The power balance between the participants and their physicians shifted with changes in knowledge and attitude.

In the previous chapter, the participants showed that they had increased their power following diagnosis in a number of ways. Firstly, in living with a rare disease, they had developed knowledge, skills and expertise in managing LAM through their own experience of living with it, their independent information gathering, their ability to advocate for themselves, and their collaboration with medical researchers. Secondly they had shown agency in engaging in these activities and increased their self-efficacy beliefs. Thirdly, they possessed subjective knowledge of their own needs and preferences. These factors enabled them to collaborate with their physicians and influence decision-making concerning their own health outcomes. They took responsibility for monitoring their health, including their

oxygen saturation levels, lung function results and sirolimus levels, and wished to have adequate time to discuss these with their physicians.

Physicians' power lay in their biomedical knowledge and designated authority within the healthcare system. While LAM experts possessed expert knowledge of LAM, many physicians the participants encountered lacked this knowledge. In contrast, the participants had increased their own knowledge. The resulting difference in knowledge levels shifted the power balance between the participants and their physicians. However, physicians retained greater power in interactions with the women through their control of prescribing treatments, ordering investigations, and following up results. In this context many participants felt they needed to advocate for themselves in order to receive optimal treatment.

5.4.1 Advocating for their health

The participants advocated for where to be treated, appropriate investigations and management, full explanation of results, improved communication between physicians, and access to sirolimus therapy. In advocating for themselves they further increased their own power and shifted the power balance to improve their health.

Not all participants engaged in self-advocacy. Their ability to self-advocate correlated with their self-efficacy and cultural beliefs, past experience, language barriers, and health status. Helen identified herself as *“an advocate for my own health”*. She was young, university educated, confident, and experienced in advocating for clients in her work. She was not afraid to speak up for herself and advocated for information, investigations, and inclusion in discussion. She also actively promoted better communication between different physicians to improve equity in treatment for women with LAM regardless of where they lived. *“The biggest things are lack of communication between my state and the rest of Australia. I'm determined to get that changed.”* Similarly, Jess, Julie, Mia and Ruth were experienced communicators in their work and Patricia had a long history of dealing with healthcare professionals as a result of her son's disability. They did not hesitate to advocate for appropriate treatment and investigations.

Veronica and Aiko, while highly competent in their work, did not feel as confident in their dealings with healthcare professionals and accepted the medical advice given. Aiko, who was Asian, was not confident to discuss her issues in English. *“Whenever I have the check-up...I have to go by myself. I have to be careful about if I misunderstand the English.”*

Veronica, who regarded herself as timid and “*not a strong character*”, had lower self-efficacy:

Whatever they did I accepted because I presumed they knew more than I did. Some people are like that, more accepting. Other people are challenging...and ask lots of questions and things. I didn't because I didn't know what was wrong with me.

Margaret, at the late stage of her illness at the time of interview, was too unwell to engage in collaborative decision-making.

When advocating for themselves, the participants revealed that physicians who lacked knowledge of LAM were less accepting of their expertise and tended to exert their power in paternalistic-style interactions. This was the case for Jess and Mia when, familiar with the management they needed, they advised hospital doctors how they should be treated. Jess “*had... the worse registrar and intern...who just couldn't believe anything that I told them...They didn't know anything about LAM but they couldn't believe that I did*”. Mia understood the treatment regime when she was admitted for her chest infections. She communicated this but found it uncomfortable to be issuing instructions to doctors:

Every time I'm in the emergency room, most doctors have no idea what LAM is, no nurses know what LAM is...Just having to explain over and over again to everyone just to get a prescription for the antibiotics, it took 30 minutes trying to persuade a doctor to give me one. I think that lack of awareness obviously has an impact... I feel really strange telling the doctor what to do with me.

Similarly, when supporting another woman with LAM in hospital, Louise felt “*they (the doctors) wouldn't listen to us... (they were) patronising, you know... there's a sort of a hierarchy that I was very unaware of to start with*”. Eva said:

What my friend's (who is a doctor) presence meant is that we could actually connect to all the surgeons more, because we had no respect from those, being just patients. So whatever we suggested was just a layman's view of things, and dismissed pretty much instantly...some of the doctors are so arrogant...If I've got the disease, at least listen to me, hear me out.

Helen and Julie were geographically isolated interstate from the two centres of LAM expertise. They felt disadvantaged when their physicians did not initiate appropriate

investigations or explain results of tests adequately, did not know the therapeutic levels of sirolimus, and gave conflicting information. Helen said:

I'm at a disadvantage. I'm the worst in this state...I don't know anything... I don't know if I should be having pneumonia shots... I've got all these questions and... I can't fly (to see doctors interstate).

Julie was concerned, having been treated for a renal angiomyolipoma in the past that more may have developed. Her doctor did not initiate an investigation and his response to Julie's request for a scan to detect if any were present suggested that he did not take her seriously and agreed to her suggestion only to relieve her anxiety:

When I saw... (my doctor) I said to him, do you think I should have a kidney scan? This can be part of it. I said, I'd hate to miss something... because they could burst too... He said, oh yeah, if it makes you feel better.

Julie was frustrated that her physician was unable to explain her sirolimus levels to her:

I check my sirolimus (level)... (my doctor) doesn't really know that much about it. All the doctors don't... The doctors just tell me what it (my sirolimus level) is because they don't know what it's meant to be. So they just go, there's your level.

Helen was unable to interact collaboratively with her physicians who were not LAM experts. Their interactions were characterised by a paternalistic style of communication, and, at times, Helen felt she needed to assert herself over issues. The frustration and stress she experienced as a result exacerbated the disruption and uncertainty she felt in being recently diagnosed with advanced disease. Helen perceived a lack of communication between her physicians and LAM experts and found they had set opinions about stopping her sirolimus and seemed unable to assess if her sirolimus levels were therapeutic. She felt she had to tell them what to do and argue against their treatment options:

Initially I was only put on one milligram (of sirolimus) and I had to fight and fight and fight and I've only just gone on to two milligrams... The two males (physicians) have this very set opinion about sirolimus... I tell them what to do. So when I came off sirolimus (for my gall bladder surgery) ... they said look, we don't think we're going to put you back on it and I said well, you are... I had to do this whole presentation to him about why I wanted to go back on

sirolimus... In my situation with two young kids... I can't take that risk of deteriorating. I stabilised for a year on it.

In addition, Helen felt she was not informed of additional aspects of managing her health, for example monitoring her bone density and having annual flu vaccination. She was frustrated by medical time pressures and her allocated appointment time being inadequate to ask questions:

I didn't really feel like he was doing much for me... I was telling him everything... I was asking about vitamins and bone density and all that I'd learned from the other women and he wasn't really saying anything about it...He's very much time checking and I had all these questions ... He never really even gave me my full results of the evaluation... He hadn't read them obviously... My trough levels, what are they meant to be? I don't know... You (the doctor) could not speak to (the other doctor)...interstate and ask?...I'm sick of this whole bureaucracy around physicians not talking to each other.

Vidu was satisfied with paternalistic-style interactions. She felt this suited her negative disposition and was culturally appropriate in the case of her South Asian Buddhist specialist who, she felt, knew her well as a person (see Chapter 3, Being Diagnosed, p.68).

Jess's story of her interaction with a physician in attempting to access sirolimus therapy revealed shifts in the power balance. Sirolimus is a product of research funded internationally by women with LAM. However, although the participants were partners in research, their physicians controlled access to sirolimus through their authority to prescribe and organise the additional approval and funding required for its use. From her own research Jess knew that sirolimus was a possible treatment for the abdominal lymphangioliomyoma which was making her bloated and causing her distress. She advocated her physician for access to the treatment over a prolonged period of 18 months. She felt she wasn't listened to and that her concerns were dismissed:

I really began this whole campaign with... (my doctor) to try and get this drug which I'd read about on the internet... But... (the doctor) never got it. He was very dismissive... every time I went down there I was always trying to make him understand how desperate I was to get rid of this abdominal mass and how much it affected my life... He never listened.

Jess was finally approved for sirolimus but when she was about to start treatment approval

was withdrawn and she was offered a course of radiotherapy and doxycycline instead, neither of which resolved her problem. The reason for withdrawal was not clearly communicated to her and, being aware of another patient at the same hospital who had received sirolimus, she could not understand why she was denied the therapy. After further enquiries Jess sought the opinion of another physician at the hospital, also a LAM expert, who, in a reasonable time frame, organised sirolimus for her. It was an effective treatment. Her abdominal symptoms resolved over twelve months and she had a 60% improvement in her lung function. She could not understand the reason for conflicting opinions:

The huge crisis was that my partner was looking on PubMed as we both often did. He saw that... (my doctor) had published this article about giving sirolimus to one of the patients. So I emailed him and I asked why he hadn't given it to me, because I'd been going on about it for so long... He sent this... letter to me and it said... you can't be on sirolimus. It's not available. You cannot take it for this purpose... I couldn't believe that this had happened. If he had explained it to me and said that this other patient met our criteria better... but he'd always told me that you just can't have it... I find it odd that you can go to the same hospital and get different treatment depending on who you see. He was saying to me, you can't get it...when, really, he could've just referred me to... (the other doctor).

Jess's interaction with the first physician she consulted was paternalistic. It revealed how poor communication shifted the power balance, and affected Jess's quality of life and clinical outcome. The physician's failure to listen to Jess's perspective and communicate the reasons for the initial delay in approval for sirolimus and its subsequent withdrawal diminished Jess's power by limiting the effectiveness of her advocacy and diminishing her sense of control when she was unable to influence decisions concerning her health. Furthermore, it created additional uncertainty and reduced her quality of life when her problem remained unresolved.

5.4.2 Being a 'good' patient

The participants understood that their physicians controlled access to treatment. In this context, they retained respect for medical authority and reported a need to modify their own advocacy behaviour. They exercised diplomacy in their interactions with their physicians and other healthcare professionals in order to maintain friendly relations, ensure ongoing care and access to treatment, and be perceived as a 'good' patient. After Jess was finally

prescribed sirolimus, she feared having her treatment withdrawn, *“I wouldn't want to complain because I don't want them to stop my drugs... I don't want to be a difficult patient”*. Aiko didn't want to be seen to be *“rock(ing) the boat”* if she sought a second opinion. Ruth observed the conflict between self-advocacy and the need to temper advocacy with diplomacy:

You need to be your own advocate... and so often it's hard and you don't want to upset anybody by speaking out in a way that might be thought of as rude when you're in their hands.

Clare judged that making suggestions to nursing and medical staff were best done tactfully so as not to usurp her role as a patient:

I had a very good relationship with all the specialists at the hospital, because they'd let me have a bit of a say and investigate and find papers...as long as I was tactful and just said, look, this is what we've found...it had to be from a substantial hospital...from a recognised specialist.

This was particularly the case when healthcare professionals who had no knowledge of LAM or her case were caring for her. Clare described how she had to provide instructions as to her management in a sensitive manner when she was having difficulty breathing and becoming distressed. An on call nurse who was unfamiliar with Clare's case interpreted her distress as anxiety and tried to calm her. Clare, however, knew that her symptoms were due to a blockage in her drain and had to advocate for herself and give instructions to the nurse even while she was distressed:

A lot of the times my sats (oxygen saturation) would still read not too bad, even though I was gasping... I remember being a bit frustrated because I might have an on call nurse who was not normally on the ward... You've just got anxiety, love... you'll be fine, just calm down... and I'd already known - because I'd had a couple during the week - that it was a clog (blockage in her drain), but you've got to be a bit diplomatic. You don't want to go well, I know. You've got say well, this happened the other day, I think this is what it could be.

Helen, in contrast, was less concerned with diplomacy and being a 'good' patient. She was focused on resolving her issues and more forthright in her advocacy when dealing with her physicians who were not LAM experts. Having advanced disease, she felt pressed to put forward her own case in the absence of expert care and other advocates. This can be seen in

her interaction with a psychiatrist consulted when she was feeling anxious about the possibility of receiving a transplant:

He was going, you seem quite anxious... how would you feel about anti-anxiety drugs? I said... there's no way I'm going on any medication. He goes, what about compliance with drugs with the lung transplant team? You've got to be compliant... I said, firstly, I've initiated this meeting with you. Secondly, I know what I need for myself and I will be compliant... I am dedicated but I'm not going on any more medication than I need to be...I'm an advocate for my own health. I'm on trial drugs so I'll be saying what I feel I need.

5.4.3 Collaborating with LAM experts

At interview, sixteen of the 19 participants had come under the care of LAM experts who reviewed them every six months. Of these, four women travelled or had relocated to be managed by a LAM expert. Julie arranged a government subsidy to travel interstate annually to be reviewed by one. In between these visits she consulted a general respiratory physician.

Three participants were not under the care of LAM experts. Margaret was now too sick to travel to see her LAM physician and was managed by a local respiratory physician. Deb and Helen lived long distances from LAM centres of expertise and Helen was unable to fly. Deb was reviewed by a general respiratory physician. At the second interview, Helen was being seen by a team of respiratory physicians, one of whom had recently moved to her city and had an interest in LAM. She was hopeful that her assessments and treatment would now be more focused.

The participants' stories showed that power shifted to be more mutually shared when their physicians accepted their expertise and collaborated with them in decision-making. This was dependent on the attitude of the physicians, as demonstrated by Jess's story, and, although not uniformly the case, was more likely to occur with LAM experts. The participants generally experienced a high level of collaboration and satisfaction when they were cared for by LAM experts. LAM experts were cognisant of latest research and were able to arrange sirolimus therapy where appropriate and monitor drug levels. Their care was focused and their knowledge and expertise engendered positive feelings of confidence and trust. There was no need to continually explain their disease. In Aiko's opinion:

That supportive system available here... is good. I don't need to explain everything every time I open up my mouth...It seems like he's under control with

my disease...I feel more positive...I really feel very confident... They are all caring. Keep eye on me.

The women felt their issues were understood and their own expertise accepted. Clare's story showed that being able to influence clinical decision-making and her health outcomes increased her self-efficacy and sense of control and, in turn, enhanced her resilience during her long hospital stay. They received appropriate assessments and investigations and these physicians were prepared to explain results and discuss treatment options collaboratively. Sarah said:

(In my altitude study) they said I actually didn't need the oxygen any more (to fly). Then I got nervous and said, well I'd still like it please, and my doctor said, well you don't really need it... We had a bit of negotiation about that, so I had it as emergency oxygen in case.

The participants found that LAM experts were accessible and would respond to email questions. Ruth was "able to ask about anything and... (my doctor's) wonderful about answering questions." They would also contact hospital staff to advise on appropriate care when a woman alerted them she was in hospital. As Mia said:

I'm so reliant on... (my specialist) because I feel that I can trust being in his hands. Like for instance when I'm in the emergency room I always SMS and tell him and then he finds a way to get there, to call and to give instructions on what to do with me because it's always like that... because they don't know.

5.4.4 Being known as a person

The participants considered that knowing each woman as a person was an important aspect of communication and collaborative care in order to understand their individual priorities when deciding on treatments. Aiko appreciated that her physician "will talk to the patient". Some women, such as Veronica, preferred a gentle positive approach while others, such as Jess, liked "frank talking". Jess felt that knowing a person as an individual allowed the physician to "work out what kinds of conversations people want to have." She said:

I think doctors probably don't need to understand much about the personality of the patient in a way, because they're thinking more about the symptoms. But, the doctors who do - my GP (local doctor) or(my physician) - are always, I think, better at conveying information and also receiving information...to even

know what your priorities are.

Underlying knowledge of the person was an attitude of empathy and a willingness to listen. Helen felt a sense of relief and hope from her communication with her new physician who had an interest in LAM and demonstrated these qualities:

I just felt for the first time some hope that someone was listening to me and taking it seriously...He spent ages with me...I think he is just really empathetic and even though he's busy and he's a top specialist...he was interested.

Jess's story of how she accessed sirolimus illustrated the impact of a physician's ability to listen and know her as a person. The first physician Jess consulted did not take her perspective into account and failed to elicit knowledge of her as a person. According to Jess, the physician lacked empathy and did not listen to her, and consequently did not understand that her priority was the resolution of her abdominal lymphangiomyoma which had markedly reduced her quality of life. The physician focused instead on her lung disease. Clinically her abdominal mass remained a problem and her quality of life had not improved. In contrast, the second LAM expert Jess consulted listened to her viewpoint, gained knowledge of her as a person, understood her priority, focused on its resolution and facilitated appropriate treatment. Her quality of life increased significantly and she achieved an excellent clinical outcome when her lymphangiomyoma resolved and her lung function improved.

A physician's ability to communicate skilfully and discuss how to live life well within the parameters of their chronic illness was important to the participants when there was no cure and, for some, no treatment options other than oxygen therapy. Aiko's LAM expert offered hope by considering how she could live fully without a transplant as her disease progressed. Aiko said:

He said, better to be living with your own organs... So we will probably focus on how we're going to make you a little bit more normal life than bedridden. You can still enjoy your life... without transplantation, which is a very, very clear focus.

The participants revealed that time was a desirable but limited resource in knowing a woman and her priorities. While they generally had a good rapport with their LAM specialists they were aware of their busy schedules, and their allocated appointment times were often inadequate to discuss issues more fully or bring up minor, but nevertheless concerning,

matters. Ruth reported there was “usually not time for chatting”. The limited time of medical appointments was constraining and could inhibit discussion, collaboration or understanding a woman fully. Aiko felt that:

Sometimes doctors and patient don't talk too much... consultation time is, that's that, and we will see you six months' time. Not enough to get to know each other...Doctors are sometimes very blunt. Just fact is fact...I don't think they've got the time to discuss.

The participants' experiences have demonstrated that the power balance in their interactions with their physicians related to the ability of each to influence and control behaviour and health outcomes affecting the participants. The power balance shifted in relation to knowledge and attitudes. The rarity of LAM meant that the participants increased their knowledge and power but many physicians lacked knowledge. Physicians' power, however, tended to remain constant while the participants' power shifted in response to the attitudes of their physicians. Physicians who lacked knowledge of LAM retained power in their authority to control treatment and investigations. Although the participants increased their power as their knowledge and expertise increased, they concealed it by modifying their advocacy behaviour so as not to challenge the authority of their physicians. The participants' power was reduced when physicians exercised their power with a paternalistic attitude and communication style. The women were most empowered and more satisfied when their physicians shared power, accepted their knowledge and expertise, listened to know their priorities, and were willing to collaboratively share decision-making.

5.5 Summary

The rarity of LAM had a significant impact on the participants' experiences in their interactions with healthcare professionals. It was associated with a notable increase in their illness burden when healthcare professionals lacked knowledge, and did not communicate clearly or provide adequate information or support. When they were in hospital these factors generated feelings of isolation and vulnerability. In this context the women demonstrated agency by taking responsibility for their health and outcomes through vigilance, self-advocacy, and taking on the role of educating healthcare professionals. Having expert knowledge was empowering for the participants in the health care setting. It facilitated meaningful communication with healthcare professionals to narrow the power differential between medical authority and their position as vulnerable patients.

A prolonged length of stay in hospital, as demonstrated by Clare's experience, exacerbated these effects and generated a greater need for resilience and emotional support than in a short hospital stay. Clare showed that her personal attributes, positive attitude and agency in researching, gaining knowledge, and advocating for herself, as well as being supported by others, enabled her to be resilient. Her inclusion in collaborative decision-making in hospital reduced her feelings of isolation, vulnerability and uncertainty and increased her self-efficacy and sense of control. In addition to these factors, Clare revealed that the personal growth and spirituality she developed through periods of liminality during and immediately after her long hospital stay contributed to her resilience.

The participants approached their interactions with healthcare professionals with knowledge they had acquired of their disease through their own research and illness experience, and communication skills gained from past life experience. This had the effect of reducing the power difference between them and their physicians. However, adopting the role of expert in their self-advocacy efforts with their physicians challenged traditional patient/physician roles. Budysh et al. (2012), in an exploratory study of the experiences of patient-physician interaction in rare diseases, found this to be the case and identified four patterns of patient/physician interaction: paternalistic where the physician directed decision-making; collaborative involving equal information exchange and mutual decision-making; patient-directed where the physician lacked expertise and was educated by the patient; and confrontational involving conflict.

At diagnosis, interactions for the majority of women were directed by their physicians due to the women's lack of knowledge at that stage. As they acquired knowledge and developed their own expertise, the majority of participants participated actively in their health care and adopted a collaborative style of interaction with their physicians. Julie and Helen experienced different patterns of interaction with different physicians. This generally corresponded to the physician's level of knowledge and their willingness to acknowledge a woman's expertise. Collaborative interactions were more likely when a woman's physician was a LAM expert. In the process of collaborating the participants enhanced their own communication skills and gained a greater sense of partnership with their physicians and control in influencing their own health outcomes.

Interactions with physicians who lacked expert knowledge of LAM were patient-driven in the case of Helen who was isolated from centres of expertise, and Clare during her admission for a gynaecological procedure. At times the women needed to be assertive in these

interactions and rejected medical advice on the basis of their own knowledge. Brashers (2000, p.387) termed this “mindful nonadherence”. Helen, for example, explicitly identified herself as her own health advocate and demonstrated assertiveness in advocating for expert medical care, variation in her sirolimus dose, and discussion of medical investigations and issues of concern such as contraception. She rejected proposals for antidepressant treatment and the inducement of early menopause for the purpose of contraception.

Some participants were satisfied with physician-directed interactions. These included women with mild disease; those who coped by avoidance; women who were not confident in voicing an opinion or for whom language was a barrier; and women at the late stage of the illness who were too unwell to engage in collaborative interactions. Other issues of communication included misunderstanding due to looking healthier than the expected clinical picture of a person with degenerative lung disease in the case of Jess, and conflicting medical opinions, as experienced by Helen in relation to contraception. Budysh et al. (2012) similarly found that interactions were influenced by a person’s health status and that conflicts could arise when there was medical uncertainty due to a person’s appearance not corresponding with the typical clinical picture. A number of women voiced the opinion that the limited time allocated to medical consultations constrained communication, even in collaborative interactions. This aligns with the findings of A.E. Beisecker and T.D. Beisecker (1990) who, in a study of rehabilitation patients’ information-seeking behaviours, demonstrated that a longer interaction time of greater than 19 minutes with their physician was necessary for a patient to engage in communication behaviour such as asking questions in order to gain information and participate in decision-making.

According to Foucault (1977, p. 27) “power and knowledge directly imply one another”. The participants showed that power and knowledge were directly related, and also that their ability to exercise their power depended on how their knowledge was received by their physicians and the physicians’ attitudes to sharing decision-making. Physicians retained greater power in interactions with the participants due to their control of prescribing treatment and investigations. The majority of physicians lacking expertise in LAM did not accept the knowledge of participants. The participants, recognising the power imbalance, modified their own behaviour so that they could receive optimal treatment. While the majority of participants advocated for themselves, they attempted to communicate in ways that were acceptable to physicians and did not challenge their authority. The exception was Helen who, with advanced disease, felt a more urgent need to advocate strongly and

challenge her physicians in order to have her needs met.

The value of patient knowledge/expertise in partnership with healthcare professionals has been recognised in reducing symptom severity, and improving confidence, resourcefulness, self-efficacy, and sense of control for patients with chronic illness (Department of Health UK 2001). As a consequence, expert patient programmes have been introduced in the UK (Shaw & Baker 2004), and in Australia as the Sharing Health Care Initiative (Department of Health 2005). However, as this current study has found, patient expertise is not always accepted by physicians. Similarly, Thorne et al. (2000), in a study of the experiences of people living with Type 1 diabetes and environmental sensitivities, found that healthcare professionals' attitudes influenced interactions with healthcare professionals. They reported that healthcare professionals held on to the role of expert, and controlled information and who qualified for health services. Likewise, Henwood et al. (2003), in a study of the information practices of midlife women seeking to know about hormone replacement therapy for the relief of menopausal symptoms, found that shared decision-making was constrained by some participants' choice to leave decision-making to their physician who they trusted, others not disclosing their own knowledge for fear of challenging the authority of their physician, and physicians' reluctance to engage in collaboration and consequent dismissal of the women's views. Shaw & Baker (2004) suggested that physicians reject patient expertise because they associate it with demanding, unreasonable and time-consuming behaviour even though it leads to improved outcomes. The findings of this current study suggest that improving patient-physician interactions is complex and requires a shift of medical cultural attitudes. This was similarly proposed by Tattersall (2002) in discussion of chronic disease management. Likewise, Ellis-Hill et al. (2008) and Tang and Anderson (1999) called for healthcare professionals to be reflexive of and address power differentials inherent in their encounters with patients.

Physicians having expert knowledge of LAM did not alone provide the best outcomes for the participants. When they supplemented their expert knowledge with subjective knowledge of each woman as a person, taking into account her individual perspective, care was focused. This achieved higher levels of satisfaction for the participants and improvement in their illness management. Communication skills of empathy and an ability to listen enabled healthcare professionals to elicit this subjective knowledge while time constraints could hinder the process.

5.6 Conclusion

This chapter has presented the participants' experiences of interacting with healthcare professionals, as patients in the hospital setting and at their periodic reviews with their physicians, and how their interactions were affected by the knowledge and attitudes of both parties, and the rarity of LAM. The participants demonstrated that being a patient with a rare disease in the hospital setting created feelings of vulnerability and isolation when healthcare professionals lacked awareness and knowledge of LAM. In this context the participants were vigilant in monitoring their care and educating the healthcare professionals. Being in hospital for an extended period created a need for resilience. Clare's personal qualities and spirituality, maintaining hope and a positive attitude, taking an active role in increasing her own knowledge and educating healthcare professionals, advocating and collaborating with healthcare professionals, and being supported by nurses, family and other women with LAM contributed to her resilience. Being an active participant in her care was empowering and created meaning by giving her a sense of purpose and enhancing her positive self-beliefs.

A power balance existed between the participants and their physicians. This represented their differing abilities to influence the health outcomes of the participants. The power balance shifted in relation to knowledge and attitudes. While the participants increased their power through their own increased knowledge and self-advocacy efforts, physicians retained greater power due to their authority to control investigations and treatments. The ability of the participants to collaboratively share in decision-making concerning their care depended on the attitude of their physician. The women modified their own behaviour so as not to usurp medical authority or jeopardise their care. The exception was Helen who, with advanced disease, felt a more pressing need to exert her own power. The women's experiences demonstrated that their satisfaction and management of their illness were most improved when power was more equally balanced. This was the case when both they and their physicians acquired expert knowledge, physicians took time to listen to the participants and gain subjective knowledge, and when power was mutually shared and physicians and participants collaborated in decision-making.

The following chapter will explore the participants' experiences of living with LAM as their illness progressed to the advanced and late stage, including their experiences of living with oxygen therapy, respiratory failure, and receiving a lung transplant. Their hospital experiences and interactions with healthcare professionals at that stage are considered in

that chapter.

Chapter 6

LIVING WITH ADVANCED LAM - EXPERIENCES OF OXYGEN THERAPY, RESPIRATORY FAILURE AND LUNG TRANSPLANT

6.1 Introduction

The previous chapter presented the participants' experiences of interacting with health care professionals in the hospital setting and at their regular reviews with their physicians. This chapter explores the experiences of the participants who were living with advanced LAM. Their illness had progressed to the stage where they had lost a significant amount of their lung function and now required oxygen therapy for exercise or other activities, either intermittently when they required or continuously. The need to commence oxygen therapy was a turning point of disruptive change. Seven participants described how having to use oxygen affected them and how they adapted and learned to live with it.

Margaret, Patricia and Vidu reached the late stage of advanced LAM. Margaret had advanced disease in both her lungs and her kidneys and was not suitable for a lung transplant. She had been hospitalised for life threatening respiratory failure. Patricia and Vidu received lung transplants. These participants revealed the impact of advanced disease on their lives, how they faced their mortality, their resilience, and their experiences of liminality at these major turning points and times of transition.

Figure 6.1 provides a visual summary of the themes of this chapter related to the progressive stages of living with advanced LAM. Firstly, the participants revealed the meaning of oxygen as a therapy of paradox, with both positive benefits and negative limitations, and the issues they faced in accessing their oxygen supplies. Secondly, the participants' experiences of living with late stage LAM are presented in relation to their everyday lives and at turning points of being hospitalised for respiratory failure and listed for transplant. Thirdly, the women who received a transplant experienced a period of transition as they adapted to living with their new lungs. Fourthly, the participants revealed how they felt transformed by their

experiences of living with late stage LAM and receiving a transplant. Through each turning point the women demonstrated how they developed the resilience which enabled them to live with their struggle and suffering.

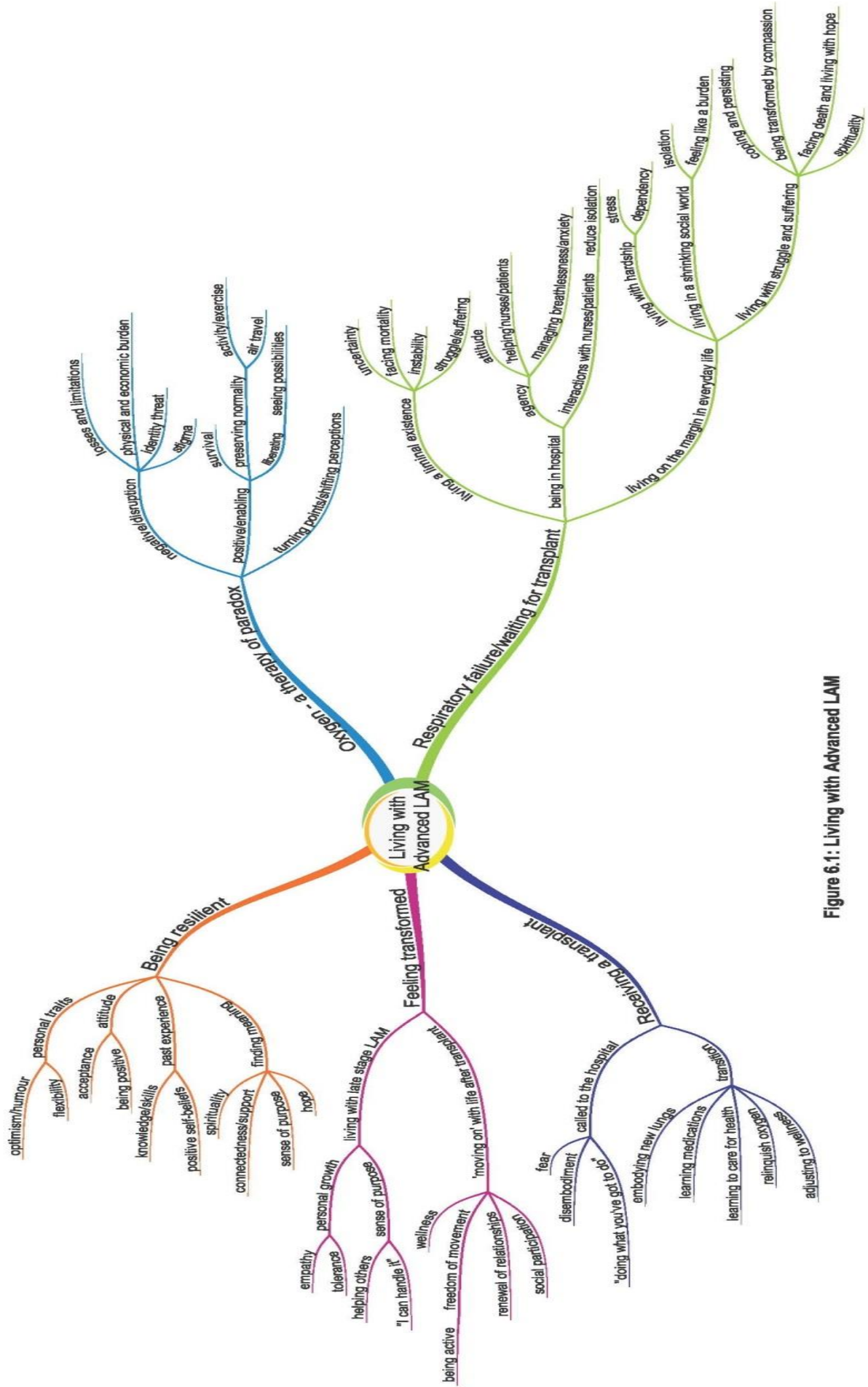


Figure 6.1: Living with Advanced LAM

6.2 Oxygen – a therapy of paradox

Commencing oxygen therapy was a critical turning point which marked significant progression of their disease and added a new dimension of disruption to the participants' lives. The majority of participants using oxygen were initially prescribed it intermittently, using it at night and when needed during the day to support exercise and activity. Over time they progressed to continuous oxygen use as their illness advanced. Five women in the study were using oxygen at the time of the interviews, Helen and Margaret continuously, and Irena, Aiko and Sarah intermittently. Helen, whose disease had progressed rapidly, required continuous oxygen therapy from when she was first diagnosed. Patricia and Vidu, who had received lung transplants, had used oxygen continuously in the twelve months prior to their transplants. Clare had used oxygen both intermittently and continuously over a period of six months while hospitalised and in the immediate period after her discharge but now no longer required it. Sarah was the only participant who used oxygen voluntarily even though her LAM was not at the advanced stage.

Oxygen therapy held paradoxical meanings for the participants according to their stage of illness and their own particular context and life meaning. Positive meanings were associated with having some element of choice regarding when to use their oxygen, being able to conceal it from others, and using it to independently engage in activities. Negative meanings were associated with the physical presence and visibility of the oxygen equipment and a loss of freedom when they needed to use oxygen for mobility and all activity. The experiences of seven of the women who had used or were currently living with oxygen therapy illustrate these meanings and how shifts in perception influenced their resilience.

Intermittent oxygen therapy was less disruptive for the participants who commenced it some years after diagnosis. In this context their disease had progressed slowly allowing them time to adjust to living with breathlessness and certain limitations. Irena commenced intermittent therapy five years after diagnosis, Aiko eleven years and Vidu nine years. According to Aiko:

I'm not worrying about handling the portable oxygen, because my life for 11 years has been limited. So having the portable oxygen 24/7 may not make much of a difference.

Intermittent oxygen was a supportive therapy for the women. It improved their sleep, facilitated exercise and activity, and gave them the ability to undertake air travel. This was

particularly important for Vidu in maintaining the close social connection with her family overseas who were her primary source of support. Additionally, intermittent oxygen therapy allowed the women some sense of control through having the freedom to choose when they would use it outside their home. They were then able to conceal their oxygen use from public view and avoid stigma and discussion around their condition. This had the positive effect of achieving a level of normality. While being able to choose when to use their oxygen made it easier for the women to tolerate it, it also meant that the manner in which they used it, due to misunderstandings, did not always provide them with the greatest physical benefit. Irena and Vidu used their oxygen after rather than during activity, negating its supportive benefit. Irena said:

I don't use it as often as I should... As soon as I get back from mum - I don't take it there with me, it's just a hindrance rather than a help, ... I'll have a bite to eat, clean up... Then I might just sit down and just get the wind get back into me... That's when I'll put it on because when I'm active it bothers me.

Patricia was uncertain if it was legal to use oxygen while driving and waited until she felt very fatigued before stopping the car to use some oxygen:

I wasn't sure whether it was legal to drive with it or not...I did, on a few occasions, drive with it, but then I always made sure I had rest periods, because... I got fatigued very easily ... I had to have rest periods to stop and close my eyes, have some oxygen and go.

Clare thought oxygen was addictive:

I think the other thing with oxygen is the more you have it, the more reliant you get on it as well. So you try and limit it a bit.

Irena and Vidu had both adopted a strategy of avoiding anything related to LAM when they were diagnosed and applied this strategy to their intermittent oxygen use. They avoided using their oxygen where possible, kept it hidden, and avoided discussion with others. For both women, this was consistent with how they viewed themselves. Irena felt she was capable of coping without oxygen, while Vidu saw herself as prone to negative thoughts.

Irena found using oxygen uncomfortable and the cylinders were heavy so she did not perceive it to be beneficial. She also had a strong sense of self-efficacy and autonomy. Her life experience reinforced her view of herself as strong, and a “doer... (who) can get things

done” on her own. She had adapted to living with breathlessness in the five years since her diagnosis and was confident in deciding independently to continue to manage by pacing herself as she had become expert in doing. *“I know that I can walk up to a certain amount, then I stop, then I keep walking. I pace myself and I think I can cope.”* Irena presented herself as one who met adversity in a positive way, *“always smiling, that’s how I’d like to see myself”*. Keeping LAM backgrounded through avoidance helped her to remain optimistic:

I’ve always been like this - even before the LAM, if anything affected me I dealt with it on my own. I didn’t necessarily talk too much about it, because for me talking about it, you just live it all the time and relive it. I just think well, if I can handle it - and I’ve been pretty good.

Over time this was likely to be a negative strategy as her medical record showed that her oxygen saturation dropped to only 80% on her six minute walk test, indicating that she was placing stress on vital organs by not using her oxygen when active. Avoidance also meant losing fitness because she also avoided exercise. Although it assisted Irena to cope psychologically and adjust to her illness, avoidance was not likely to be beneficial over the course of her illness.

Vidu saw herself as *“a bit of a weak person...if I think I will be negative, I will get negative thoughts.”* She hid her oxygen even from her family to avoid negative thoughts and being identified as a sick person. She didn’t want to be treated as an invalid and denied the opportunity to do anything for herself which would further limit her freedom, control and independence, already restricted by her disease:

I just didn’t want to see them see me with oxygen...I didn’t want to listen to any stories. I just wanted to avoid things like, my god what’s wrong ...I didn’t want to talk about it...She (my husband’s sister) treated me like she didn’t want me to stand even...I felt bad. I thought at that time, good I didn’t tell my friends.

In avoiding using their oxygen Irena and Vidu did not adapt to it. They had not accepted and integrated their oxygen as a part of everyday life nor developed the knowledge and skills to manage it effectively. Vidu’s story later in the chapter will reveal the effect of avoidance on her resilience and ability to manage her illness when she progressed to late stage prior to transplant.

Intermittent oxygen therapy was less disruptive because the participants retained control

over its use. However, this meant that they did not always adhere to it due to misunderstandings about its use, inconvenience, discomfort, and to avoid making their illness visible and consequently avoid stigma and a sick identity.

Becoming less well and progressing to continuous oxygen therapy caused a high level of disruption to the participants' lives. They could no longer avoid using their oxygen. To maintain their mobility while living with advanced disease, they would now need to learn to live with their oxygen and use it for the rest of their lives or until transplant. Continuous oxygen therapy was a paradox in that it disrupted their lives but, at the same time, enabled them to remain active.

6.2.1 Disruption - the negative meaning of oxygen

The negative meanings of continuous oxygen were associated with its disruptive effect on the participants' lives. The women experienced additional losses and limitations due to both the physical and visible presence of oxygen equipment, and their advanced lung disease. Using continuous oxygen therapy felt like a burden and, being visible to others, created feelings of being stigmatised. It was a new period of transition and adjustment.

6.2.1.1. Losses and limitations

Physically, continuous oxygen therapy indicated to the participants that they had lost a significant amount of lung function. The women found their activities and ability to access certain spaces, already restricted by their breathlessness, were further limited by their oxygen. At home, oxygen cylinders and the concentrator took up space and the women's freedom and range of movement was limited by the length of their oxygen tubing. Helen had high functional mobility and was able to walk about independently with her oxygen. Margaret, however, whose disease was further advanced, was functionally very limited and needed to use a motorised scooter to get about. She could not access other social spaces such as movie cinemas and cafes with steps. Camping and going to beaches and swimming pools were logistically difficult to manage. Margaret "*loved the beach... I can't go to the beaches the way I used to, I need the oxygen which I can't have around the sand.*" Going out meant having to carry oxygen cylinders, tubing and spare batteries and the women needed to plan their outings to ensure an adequate supply of oxygen. Helen expressed her loss:

We loved camping, and now we can't do it. I just got rid of all my exercise equipment... I don't want it as a reminder of what I can't do... We'll replace it by oxygen concentrators and tubing and batteries.

The women's physical losses and limitations generated social losses. Margaret and Helen had lost most of their friends who they felt did not understand their situation and found it too hard to accommodate them in social arrangements. Margaret said:

That's the hard thing too, your social life. I mean I've lost basically 95% of my friends because it's too hard for them to want to keep in contact.... I was devastated because I've always done everything I can to help my friends.

The two women felt the visible and constant presence of their continuous oxygen impinged on their identity and sense of self. Helen had always viewed herself as a fit and active person. The limitations posed by her advanced disease and oxygen therapy meant she was unable to engage in her fitness activities in the usual way. She experienced an altered sense of self:

I just cried because I just wanted to be able to go to boot camp, I feel terrible about my weight and people just don't get how I can't do that. I feel like saying, imagine... how you'd feel in yourself if that's your whole thing you really love doing, and for me that's what it was, doing my exercise. I loved it, it was my de-stressor.

The visibility of their oxygen tubing threatened their female identity by making them feel less attractive. Margaret felt rejected by potential partners:

Now no one really wants to go out with me cause the first thing they see is the nasal prongs and like something's wrong with me cause as soon as you've got nasal prongs you've got no brains...

Helen worried about what her partner thought of her when he looked at her with oxygen tubing and felt that oxygen and breathlessness intruded on intimate moments:

You feel terrible about yourself... I hate looking at myself in the mirror especially when I've got oxygen on... You've got this tube attached to you... I'm half the time thinking, oh god, what's he thinking about when he looks at me?

6.2.1.2 The burden of oxygen therapy

The women felt that their oxygen was a physical and economic burden. Physically, oxygen cylinders were literally and metaphorically a heavy weight. Oxygen was a financial burden although it was medically necessary. It was not funded equally in all Australian states and the participants reported different levels of government subsidy. Some states subsidised all

oxygen equipment including a large concentrator, while others, as in Margaret's case, provided oxygen cylinders only for a fee. Margaret was a single woman on a disability pension. She reported that she was paying \$400 a month (almost one fortnight's payment of her pension) for her oxygen cylinders. Helen and her partner had to undergo expensive kitchen renovations to change from gas to electricity.

The women did not receive funding for portable oxygen concentrators which facilitated their ability to participate in exercise and activity and, in turn, social engagement. They reported that they could not afford portable concentrators which cost between six and seven thousand dollars. Margaret and Vidu's parents paid for their portable oxygen concentrators while Helen's mother paid for a large stationary concentrator for home. This in itself created a feeling of being a burden to family. The women would have preferred to use liquid oxygen which was light and convenient but this was not available. As Helen said:

In the UK they have liquid oxygen so you have a big tank and you fill it up yourself and they're much lighter and it's a much better system. I don't know why they don't have it here.

The burden the women felt was increased by the inadequacy of the criteria used to assess their oxygen requirements as younger people. Margaret and Helen reported that their oxygen needs were assessed on the basis of spot checks of their oxygen saturation levels at rest and did not take into account their function and activity level. According to Margaret, her oxygen saturation at rest was not low enough to ensure funding for her oxygen even though functionally she clearly demonstrated she needed it to manage activities of daily living. Helen was a young mother of two preschool aged children and working two days a week. Her assessed requirements for oxygen did not take into account her exercise and activities at home, work and caring for her children and were inadequate to meet her requirements. It was only when she assertively advocated for herself that she was able to access the extra oxygen she needed:

They take your sats (oxygen saturation level), oh your sats are really good, and they say, you can have three bottles of oxygen for the week and I'm like, that's not going to work for me... I'm 40, I've got two young kids, I work two days, I need oxygen to survive... I said I need another bottle of oxygen. She goes, well you can't have it... I said well, you know, forget it, I'm just going to phone the oxygen company... and just say I need more oxygen, I've run out, and I said, I'll just get my specialist involved, and they're like, no, no, no, don't do anything...

later she goes, you can have your six bottles.

Helen displayed resilience in organising her oxygen supply. As an articulate, well-educated, professional woman, her self-efficacy was evident in her belief in her own ability to solve her problems and reach her goal of obtaining adequate supplies of oxygen. She showed self-determination in deciding her oxygen requirements, and self-reliance and self-advocacy in being able to voice her needs and negotiate the bureaucracy of the supply system. While the necessity of relying on her own abilities so early in her illness was a psychological burden compounding the physical burden of her illness and the therapy itself, it also enhanced her sense of autonomy, control, and confidence in her capacity to manage any ongoing health issues.

Helen was the only participant to be living with oxygen therapy while caring for young children. Her illness and oxygen therapy added layers of complexity and challenge to her already demanding role of mothering two preschool aged children while also working. Helen was still adjusting to having a new baby when she was diagnosed. She left hospital experiencing the emotional disruption and loss of being forced to wean her daughter and accommodate oxygen equipment. At the time of our interviews her children were three years and sixteen months old respectively. The fatigue associated with Helen's illness meant that she lacked the energy to manage her two active children and this was exacerbated by nightly disrupted sleep due to her children's frequent waking. She was perpetually exhausted. *"Some days at the end I'm exhausted...I just totally give up, I literally have not one ounce of energy in my body to even make dinner"*. It was a difficult cycle to break and increased her sense of struggle and burden.

Oxygen interfered with Helen's ability to lift her children and participate in and enjoy activities with them. At home it was difficult for her to move quickly enough to prevent falls and break up fights between the children. Attending medical appointments with two children was not possible on her own. Pushing a stroller affected her breathing, making even going for a walk with her children too hard. She expressed the burden of her oxygen:

I get really bad shoulders from lugging the oxygen around all the time, especially if I've got Emma (pseudonym) in one hand and the oxygen in the other. It really hurts. It's really heavy because I'm carrying 15 kilos.

The children also restricted Helen's ability to manage her health. They played with her oxygen tubing and the settings on her concentrator causing her oxygen saturation levels to

drop. Although she had a treadmill it was difficult to exercise independently at home when the children were around and she rarely used it. Constantly supervising her children around her equipment added to the burden of her oxygen:

People are always treading on my oxygen... they're always switching my concentrator on and off... or they're changing the setting...The other night... I was thinking, I just really don't feel like my breathing is brilliant... I just felt awful. Then I suddenly thought, my God, I hope my oxygen's up because Emma's been fiddling with the dial.... it had been basically turned down to zero. So I had no oxygen for nine hours and I've been running around.

Helen's illness and periods of hospitalisation disrupted her children, particularly "affecting (my son) badly". He resisted going to day care after Helen had been in hospital and displayed aggressive behaviour towards other children while there. At home he had nightmares, "gets very angry at me...or just panics". Helen felt frustrated and isolated when she searched but found no resources to assist her in explaining her illness, oxygen therapy and hospitalisation to her young children. She knew of no other women in her position, either locally or nationally, with whom to share her experiences. In this context, she found online communication with women overseas was a vital source of social support.

Helen experienced a spectrum of emotions in her mothering role. She loved her children and was grateful that she had them before she was diagnosed. At the same time she felt guilty and frustrated at the restrictions her breathing and oxygen imposed on her ability to interact with her children. "I feel like I'm not doing enough for the kids because I'm limited to what I can do now because of the oxygen." She felt "sometimes like a bit of a failure or I'm not doing a good enough job" as a mother. Uncertainty and fear as to whether she would be alive to see her children grow up was a constant presence:

You just think I don't know what's going to happen tomorrow...I'm stressing about the transplant now...What happens if I don't make it? What happens if I have rejection? I want to know the figures about life expectancy after transplant... I just need to know because I've got the kids and I want to see them get up to their 20s.

6.2.1.3 Feeling stigmatised

The participants experienced stigma in public spaces, commonly in relation to their use of a disability parking scheme which allowed them to park their car in specially designated

parking spots that were closer to facilities and shopping centre entrances in order to avoid walking long distances and becoming breathless. Although they were wearing oxygen, their appearance of being young and otherwise well meant that other people assumed they were misusing the disabled parking spot and they frequently experienced disparaging looks or verbal abuse, chiefly from other disabled or elderly people. Helen described the stigma she received and how she responded:

I think it's because I've got the kids and I don't look sick...I get it all the time... This woman started screaming from her ute, what are you doing parking in the disabled bay? We need that...so I just got my leaflet and said, you know what lady, I'd love to swap lives with you. Read this and educate yourself before you start making judgments... Sometimes I feel I have to actually make myself worse than I am, especially when I'm shopping.

Patricia, Helen and Aiko were excluded from yoga classes because of their oxygen. Patricia said, “I used to do yoga, even with my oxygen, until the person that was taking the class said to me one day, I think you should give it up”. Margaret felt that she attracted unwanted stares, looks of pity and people deliberately crossing her path:

Being in the scooter... going along the aisles (of the supermarket) and... it was so blatant... this woman kept staring Some look at me with pity.... A lot of them do it out spite, especially to walk in front of me. Because I'm young and on oxygen in a scooter I must have done something to myself to be like this.

The participants responded to stigma in a number of ways - “fighting” back verbally (Margaret), ignoring (Patricia, Helen, Margaret), handing out information brochures about LAM to raise awareness (Helen), giving up the parking space (Patricia), parking further away (Irena) or exaggerating symptoms of breathlessness to avoid stigma (Helen). Irena even pretended to limp to be visible and appear acceptable. Margaret could ignore stigma when she felt well but retorted angrily when she felt unwell:

*I wasn't feeling well, I hadn't slept, and then things get on top of me a lot easier...She goes, what the bloody hell do you think you're doing parking in a disability spot?... I just turned around and said...Would you like to see my*** (swearing) lungs?... cause I can't*** (swearing) breathe.*

Helen felt that her manager made assumptions about her work performance because of her oxygen. The stigma she experienced was more subtle and took the form of inflexible work practices, being overlooked for positions, and being made to feel ignored, isolated and ostracised. She was denied the flexibility to work from home so that her oxygen could be more easily accommodated, and felt that her opinions and skills were discounted. In contrast to her ability to advocate for herself in other situations, Helen felt restricted in what she could say, fearful of losing her job in the context of needing the income and accumulated benefits it provided. She also enjoyed her work, *“it keeps me going”*:

I'm on quite a good wage if I was working full time but that's now not happening ...I have to stay there because of the benefits...I'm scared about leave...I'm never consulted about anything...I kept coming up with ideas, they were just like oh no... that wouldn't work... I thought I just want to resign from this, I can't do this anymore. It's just too hard. I'm not being respected as an experienced worker, I'm being devalued. I'm basically just seen as nothing, just like a cleaner. That's how I was feeling... normally I would change jobs, I wouldn't stay in a team like that.

6.2.2 Enabling - the positive meaning of oxygen

The participants were able to perceive positive meanings for their continuous oxygen as they accepted it as part of their everyday life, adapted to it and learned to manage it. In a positive sense, oxygen enabled them to not only survive, but also participate in everyday life at this advanced stage of their illness. Being able to engage in exercise, work, and social activities allowed them to preserve a sense of normality and control.

6.2.2.1. Staying alive

Staying alive was the enduring meaning of oxygen for the participants at the late stage of their illness. Margaret's lung function was severely impaired and she had experienced respiratory failure. Her life depended on her oxygen. Initially she had felt self-conscious about it but, over time, so intimately attached to it in all that she did, she embodied it. *“(The oxygen) is just me...I need it to breathe.”* She accepted it and it became a silent extension of herself. In sustaining life, oxygen was a therapy of hope for quality of life, for Patricia, Vidu and Helen until transplant or, in Margaret's case, until the end of her life.

6.2.2.2 Preserving normality

Rather than focusing on the limitations of oxygen therapy, Patricia concentrated on the

possibilities for activity that oxygen enabled and put these into action. Throughout the course of her illness Patricia had adopted a pragmatic attitude. She had demonstrated her resilience in capably managing her own health and her intellectually disabled son's complex medical and social needs for five years while using intermittent oxygen therapy, as well as working part time. Seeing her oxygen as enabling allowed her to fulfil her responsibilities, continue to exercise, and remain socially connected. In turn this provided a sense of normality, maintained her stamina and positive mood, and preserved her identity. She continued walking, swimming, going to the gym every day (walking on the treadmill, doing light weights), ten pin bowling, yoga and bike riding using oxygen:

I walked and I went to gym every day and just got on the treadmill, I had my oxygen and I did light weights - I still ten pin bowled... I started riding with my oxygen on my bike...I swam...

I went on a cruise with my mother-in-law... and I took about six bottles of oxygen in a shopping trolley and I went through every last bit ...I got around - I had my oxygen in my backpack...I got off every day...I walked around the town.

Oxygen also enabled Patricia to participate in caring for her father when he was seriously ill and she applied humour to attending to her father's needs even while her own breathing was compromised:

When Dad was first sick, I remember looking after him one day... I said to him, now, Dad, just stay there, don't move... the top half of his bottom was nearly over the bed, and I said to him, Dad...you and I will have to take a puff each of the oxygen to get you back up...I got him on the bed. I managed to.

6.2.2.3 Feeling liberated

Sarah's story illustrated her creative thinking and ability to see possibilities for using oxygen. Oxygen was liberating for Sarah. Although she did not medically need to use it, and because she had the financial resources available to privately purchase an oxygen concentrator, Sarah uniquely advocated for an oxygen prescription in order to be able to exercise more intensely on her treadmill at home and maintain her identity as a fit and active person in order to be a role model for her daughters. "*I didn't want it (LAM) to define me*". For Sarah, oxygen positively meant empowerment and liberation in taking control of her exercise and walking at a faster rate:

I said to him (my husband), I want to get an oxygen concentrator at home and a treadmill and I want to walk with oxygen. He really didn't like the idea at all...I said, no, I need to move and I'm not able to move fast enough on my own steam... I feel frustrated that I can't go and it's really annoying me, and I don't want my kids to ever think that I'm not a very active person. I want them to see that because that's who I always was...In the end I got it... I just thought it was very liberating ...if I can do what I want, then why wouldn't I?

The participants demonstrated that oxygen therapy was a paradox, in both disrupting their lives and enabling them to be active. Negatively, they experienced a loss of freedom and friendships as their oxygen equipment and advanced disease restricted their movement and access to social spaces. Oxygen therapy was a physical and economic burden. Its visibility threatened their identity and created feelings of being stigmatised in public spaces and workplaces. Positively, oxygen therapy enabled the women to stay alive and participate in exercise, work, and social activity, in turn allowing them to preserve a sense of normality and control.

6.2.3 Turning points and shifting perceptions

Vidu and Helen demonstrated how their perceptions of their oxygen therapy shifted and influenced their resilience. Vidu experienced a turning point when she changed from intermittent to continuous oxygen in the year before her transplant, ten years after her diagnosis. This represented significant illness progression and negative change. With the constant presence of oxygen, and experiencing increasing breathlessness and periods of anxiety and panic, she was no longer able to avoid her illness but continued to avoid showing people her oxygen. However, to maintain this strategy, she now had to confine herself to her home and avoid face to face social interactions. Avoidance now meant social isolation rather than normality.

Vidu demonstrated a lower level of resilience at this transition. While avoidance had been an effective means of coping earlier in her illness, it was now an isolating strategy. Additionally, in relying heavily on her family for support, Vidu had not developed personal resources to assist her in adjusting to the change. She entered a liminal phase of feeling trapped and socially marginalised. She was uncertain if she would survive. She used metaphors of chains and death to demonstrate her loss of freedom and feelings of being imprisoned by her breathlessness and oxygen. She felt she was giving up on life and had thoughts of dying. Her perception was one of suffering and, with limited resilience, she

struggled to endure:

I couldn't breathe then, I was so tired... I was a bit isolated... I didn't go anywhere, I was just chained inside... I didn't want to go, because I can't... I just didn't want them (her neighbours) to see me with oxygen... Sometimes I was scared... I just thought why should we suffer like this? It's better if I die now.

Helen showed how her agency and self-advocacy shifted her perception to enable her to view her oxygen as having positive benefits. Helen had entered a liminal phase when she was diagnosed due to the early rapid progression of her disease and the uncertainty of not knowing how quickly she would need a transplant. Although sirolimus medication had stabilised her physically she still felt emotional and mental disruption due to her inability to access adequate social support. Helen's agency was apparent in her determination of her need for additional support and her advocacy in independently organising affordable home services:

The company that does my oxygen prescription, I had a big meltdown with them... I just said, there's no services and you don't help me at all really... So they got a healthcare assessment... It was a bit tricky because I didn't really fit the criteria... I said I can do it (housework) on oxygen but off oxygen I couldn't. So I think that's how I got through the system. So then they started home care services.

Accessing home help was a significant positive turning point which shifted Helen's perception of her oxygen as a burden to one of seeing it as enabling. Helen's first narrative was focused on a negative view of her life on oxygen as one of struggle and heavy burden. Her second narrative, in contrast, had a positive focus of moving beyond coping to adapting to her situation. In doing so the intensity of Helen's liminality was lessened. Home help restored Helen's energy, gave her time to herself and enhanced her optimism and resilience. She still lived with uncertainty but, with the additional social support, she felt greater emotional and mental stability and could now see possibilities for her life. She took control and decided independently to return to exercise at the gym:

I've got a really wonderful woman that comes in... (She) now comes to (the kids') swimming lessons... So I'm achieving that and that's something I never thought was going to happen - to give my kids the opportunity... Then she does an hour's cleaning. She's babysat a couple of times so my partner and I've actually got out... I now have a bit more energy so I can do a bit of cooking...

I just decided...I was feeling just so depressed because I wasn't exercising - because I love exercising. So I just thought I'm just going to phone my own gym...I just said, These are my worries, I am on oxygen, I really hope you can look beyond that. I'm responsible for my own oxygen.

By exercising at the gym Helen regained a vital aspect of her identity and, although she could not exercise in the same way as before her diagnosis, exercise elevated her mood, improved her self-image, and gave her a sense of achievement and normality. The benefits Helen was feeling flowed on to her family so that family interactions were also more positive:

I just felt so normal...even though I've got the oxygen I just feel really normal...I've built it up (riding her bike) from four minutes right up to eighteen now. I just check my oxygen...I just feel so good. It just makes me feel happy and I feel really like I've achieved something...

I went to the gym and then I said to my partner, let's go for a walk. That was something we always used to do all the time with the kids. It was just lovely... my partner and I actually talked normally without arguing. It was just a little bit of normality without focusing on LAM.

Helen's actions in organising her oxygen supplies, mobilising home services, and managing her health increased her self-efficacy and feelings of autonomy and competence. In adapting to her illness, Helen demonstrated resilience in her acceptance of both "good days" and "bad days" in her life with LAM and her confidence that she had developed strategies for managing the "bad days" and moving on from them:

I have good days all the time...I think it's just time goes by and you just get more used to adapting. I'm not going to let this thing beat me... I think you're always going to have bad days... I just Facebook or talk to... (my friend with LAM)...Then... I think it's not worth doing that for more than two days... it's just not worth it for your own health and everything. So I just can pull myself (up) - luckily I know, and I just go to the gym.

Helen and Vidu demonstrated the connection between resilience, coping style and perceptions of illness or wellness. Vidu's avoidance style of coping, lack of experience in learning to manage her illness, and lower resilience prevented her from seeing the positive effects of her oxygen and she associated it with illness, restriction and isolation. Helen's

resilience increased as her perspective shifted to view her oxygen as enabling possibilities of experiencing wellness when she mobilised adequate social support.

6.3 Living a liminal existence – respiratory failure and waiting for a transplant

The late stage of LAM marked the progression of the participants' illness to respiratory failure or lung transplant. At this stage the women experienced a marked decline in their function and mobility, persistent breathlessness, anxiety, and needed to use oxygen continuously. Only three participants reached this stage. Margaret experienced respiratory failure and Patricia and Vidu each received a lung transplant. Respiratory failure, being listed for transplant, and receiving a transplant were major turning points when the women tangibly faced mortality. Their stories revealed their struggle and suffering, resilience, and experiences of liminality and personal transformation. While there were commonalities in the women's experiences they were, at the same time, very individual according to each woman's own physical condition, existing resilience, personal resources, social context and coping style. Spirituality and social support were significant factors which enabled them to be resilient and live with their suffering.

The late stage of LAM was a transition to new life after transplant or end of life from respiratory failure. It was a liminal period in which the participants moved from a relatively stable physical state to one of instability. Turning points marked entry to this transition. For Margaret, two life threatening hospital admissions for respiratory failure and a complicated pneumothorax were critical events that indicated severe disease and forced her to evaluate her life as she faced the very real possibility of her own death. Being listed for transplant intensified the liminality associated with Vidu's oxygen therapy. It was a turning point which ushered in a heightened period of uncertainty and waiting. At this stage of their illness, life was precarious as the women hovered between life and death. It was a shadow existence at the margins of physical and social life. Their life was characterised by struggle and a shrinking social world as their ability to interact socially became more limited. They struggled to carry out the simplest of tasks. Illness dominated the foreground of their consciousness as they became acutely aware of their breathing. Struggling to breathe was frightening and they experienced episodes of anxiety and panic.

Liminality at this stage was a time of struggle and suffering. There was, however, a marked difference in the women's perceived levels of suffering which corresponded to their

individual levels of resilience. Patricia and Margaret had demonstrated high levels of resilience throughout their illness while Vidu had a low level of resilience due to her usual practice of avoidance. Patricia only alluded to her struggle and did not convey a sense of suffering. Margaret relayed her struggle and suffering but emphasised her positive perspective and ability to cope with her situation. In contrast, Vidu conveyed the deepest sense of suffering and difficulty in enduring.

Being listed for transplant increased Vidu's suffering by adding a further dimension of heightened negativity, uncertainty and waiting to the liminality she felt in using oxygen continuously. Her blood group was rare and the possibility of finding donor lungs was even more uncertain for her. She felt that dying would end her suffering. Vidu used her strategy of avoidance to cope with this stage. Although she was unable to avoid her own suffering she avoided discussing the severity of her condition to limit her negative thoughts and lessen her family's worry:

I didn't want to show them (her husband, daughter, parents) that I am suffering like this...I was avoiding that. I didn't want to discuss it because I knew...if I don't get a lung then they would be upset...I thought actually it's better if while I was sleeping if that (dying) happens...so that no one will feel it. Even I will not suffer.

6.3.1 Being in hospital

Margaret's life threatening hospital admissions were both turning points and a paradox. At these turning points Margaret's life was threatened by her deteriorating physical condition. However, paradoxically, her hospital experience represented success in surviving a life threatening situation and was also socially positive. She felt less isolated and part of the hospital community through her interactions with other patients and nursing staff. These interactions maintained some continuity of her sense of self and balanced the impact of her physical decline.

Margaret demonstrated resilience in response to the disruption of her physical deterioration and being in hospital by choosing an attitude that was consistent with her personal identity. She gained a sense of control by presenting herself as a "fighter", and a sense of purpose and feelings of self-worth by being an expert and "mother hen" to other patients and nursing staff. Her "fighter" self was apparent as she told of her stubbornness and attempt to continue to self-manage her breathlessness when the ambulance arrived to transport her to hospital,

even though she “*could hardly breathe and talk...and was just grey and...completely blue on the lips*”.

Margaret presented herself as an expert in managing anxiety and breathlessness by advising other patients experiencing these problems how to manage them. Her medical notes record that in fact Margaret was not able to manage her symptoms as usual. Anxiety triggered multiple medical emergency calls for respiratory distress and she required medication to settle. She did not mention that anxiety had been a problem for her in hospital. This omission was interpreted as her attempt to maintain a positive attitude and reinforce her identity and ability to cope with adversity by choosing to present this aspect of herself in her narration:

When I was in hospital this time there was a lady across from me... she was having problems breathing and I know what it feels like to have the panic attacks...I said, Breathe in when I breathe in, and breathe out when I breathe out, Mary... I could see her starting to relax cause her panic was going from her face... within five minutes she was fine.

In her relations with nursing staff Margaret presented herself as an expert, a fun, positive person, and a “*mother hen*” by providing brochures and education about LAM, enjoying humorous banter with the nurses and striving to “*help*” them by making few demands on their time herself and by keeping an eye on the other patients:

Three o'clock in the morning... they're (the nurses) having their coffee break and we were talking so I got to know them really well...I had to laugh because one came in and... he goes, I've got you tonight, and I went, Oh you poor bugger (laugh) and he goes, No we fight over who gets you... because when you buzz you've actually got something wrong or you're helping someone else.

Vidu was hospitalised for a pneumothorax two months after she was listed for transplant. In contrast to Margaret who was active in the hospital context despite her physical condition, Vidu emphasised the suffering she was experiencing at that time. Vidu had not developed strategies to manage her symptoms and had a correspondingly lower level of self-efficacy and resilience. She took a more passive role of depending on her family and nursing staff to assist her in even basic activities and managing her anxiety. She was highly anxious, “*always asking for oxygen... (and) couldn't go to the toilet alone.*” Members of her healthcare team recorded her high level of anxiety. Nurses documented that she was “*agitated and anxious, SpO₂ (oxygen saturation) 82%, requiring assistance with ADLs*

(activities of daily living)”, and the psychiatry team that she “experiences panic symptoms - has become increasingly dependent on family for perceived safety.” Vidu said:

I wanted someone so I was just insisting whether I could keep someone (from my family) because I know the nurses are really busy and I can't depend on them... I was so panicked then. I thought I will die.

6.3.2 Living on the margin

Margaret's narratives described her struggle to manage everyday life as a single woman living alone with severe disease. She was living a marginal existence physically, socially and financially. At home, following her discharge from hospital, Margaret struggled to manage activities of daily living. Physically, she was experiencing persistent pain due to her lung disease and kidney growths and was only able to move about indoors using her scooter. Her breathlessness interfered with her ability to shop, prepare meals and eat and drink. She gave a vivid description of the difficulty she had in making simply a salad and a cup of coffee.

When dad was here... I don't think he realised how bad I was until he saw me when I came out of hospital... I was just cutting up vegetables for a salad...I had sweat, like a tap on my head. Sweat was just coming off me and I was fighting to breathe. He goes, you're going grey. He goes, stop, go on your nebuliser, and he finished it. He didn't realise the energy it takes for me...even to make a cup of coffee...and I'm out of breath...

...Every winter I have lung infections for longer. They're more severe. I have to drive my scooter into the bathroom... I drove straight in, turned the seat sideways, and to lift my bottom from there onto the toilet I was fighting to breathe.

The metaphor of “*fighting*” illustrated this struggle that was part of her daily life. She referred to “*fighting to breathe*” and February as the “*killer month*”. Her “*fighter*” identity responded to the struggle, “*fighting*” not only to breathe but also to maintain some level of independence and continue to live alone in her own home:

I hate having to rely on people... I'm so bloody stubborn and so independent.

6.3.2.1 Living with hardship

Margaret was disadvantaged financially, and suffered economic hardship. The cost of

additional medications stretched her budget to the limit. She could not afford fresh fruit and vegetables, or high energy supplements that may have been useful for her to ensure an adequate nutritional intake. Social support was crucial for Margaret's resilience at home at this stage. She depended on the practical and financial support of her mother in helping to pay for her medications and providing home cooked meals for her. Her financial worries were one of the main sources of stress for her at this stage and further exacerbated her physical symptoms:

Usually I'll be spending about maybe \$90 or so a month (on medicine) - that's when I'm healthy. When I'm not healthy then you've got the antibiotics and everything else. I get, like, \$500 a fortnight. That's food, electricity, medicine... I can't afford fruit and vegetables...they cost a fortune...That stresses me like you would not believe...and when I stress about that my body tenses and then my breathing goes.

6.3.2.2 Living in a shrinking social world

Margaret's social world was shrinking. Breathlessness, pain, immobility, and her inability to afford to continue running her car meant social isolation. The most difficult aspect of her illness to deal with was “*not being able to get around by myself that much...and not having anybody in my life*”. She was unable to continue with her dog rescue work and lost her social interaction with visiting potential owners and the puppies. Her identity and sense of self were threatened by the loss of this mothering role and sense of expertise and accomplishment she gained through that work:

I cried. God, I cried (when I gave up dog rescue)... not just because I was giving them a chance at life. but seeing the people when they came to pick them up... I still miss my dog rescue... with children or animals, I have the patience of a saint ... I had (one dog) trained in a week.

Although she still lived alone, growing dependency altered Margaret's regular social relationships. Her family and close friends continued to support her but, for Margaret, her relationship with her mother and her friendships were becoming overlaid with a sense of being a “*burden*” causing her to lose reciprocity in her relationships. Margaret was particularly concerned about the toll her illness was taking on her mother. Her “*mother hen*” identity was evident as she attempted to balance the relationship by reversing the mother/daughter roles and taking on a mothering role herself, protectively limiting the amount of time her mother spent with her. Dependency was also balanced by continuing to

provide care for her dogs.

I get down because I feel like I'm a burden. Mum's 67 this year so I don't want her to do too much for me...I said, live your life... I have to, she's the one who has to drop the apron string, not me (laugh), you know it's the other way around.

Vidu was similarly socially isolated by her illness and depended on her family for practical and financial support. She rarely went out during the day due to breathlessness and fatigue and, by choice, to avoid revealing her oxygen use. She washed herself by a sponge bath. Her husband showered her on weekends and attended to shopping and other chores. Vidu was less physically impaired than Margaret and was able to walk slowly and cook at home. She also had more financial resources at her disposal. Her husband was working and so she was able to afford a cleaner, running a car, and adequate supplies of fresh food. Being able to drive a car with her oxygen if she chose gave her greater mobility than Margaret. Although Margaret was the most socially isolated by her illness and her circumstances she demonstrated greater resilience than Vidu who isolated herself further by her avoidance strategy.

6.3.3 Living with struggle and suffering

The women drew on their personal and social resources to live with the struggle and suffering associated with this liminal late stage of the illness. The accumulated knowledge and skills they had gained over the course of their life and through their illness experience contributed to Margaret and Patricia's resilience. Vidu struggled to endure her suffering but a social turning point shifted her perspective to enable her to endure until she received her transplant. Each of the women experienced spiritual and personal growth and were supported by their family and close friends. Family relationships, and, in Margaret's case her pets, continued to give their lives meaning and a sense of purpose. These were a source of hope and crucial elements in living with their suffering.

6.3.3.1 Coping and persisting - Patricia

Patricia's resilience was evident in her ability to persist through her struggle with breathlessness. She did not perceive herself as suffering. Patricia continued to present herself in her narrative as managing her health at the late stage of her illness, the year before her transplant, in the same pragmatic way she had since her diagnosis. She emphasised her

positive perception of herself as independent, persisting and coping in spite of her severe disease. Her younger son, husband and close friends provided social support in this period. “*I just tried to hang onto what I had, and my friends were quite supportive.*” Her regular exercise and spirituality were instrumental in supporting her resilience and ability to cope while waiting for transplant:

I needed oxygen more and more - I couldn't bike ride. The dog was 15, had hip dysplasia...Him and I struggled round the block every night - we walked...I struggled with the breathing, and had the oxygen tank...everybody knew me as the lady with the limpy dog and the oxygen...

I became very spiritual...I bought a lot of angels ... I went into the Catholic Church...and lit some candles ... I think it helps with coping...I meditated... it's a stillness, a quietness, a calmness.

She remained hopeful by choosing to avoid thoughts of death and the possible consequences of a transplant:

You don't think about it (dying), because if you think about it, it just makes it worse... I think keeping active and walking and going to the gym - just keeping yourself busy, and then resting when you need to. I still had to cook and all of that for myself.... It's just a matter of do it, you know... The outcome is going to happen, whatever. I was lucky. I was blessed to get a transplant.

6.3.3.2 Feeling transformed by compassion – Vidu

While her suffering continued until her transplant, Vidu simultaneously experienced a turning point of positive transformation as a result of compassion extended to her by her family, doctors, nurses and other patients. Vidu’s South Asian chest specialist encouraged her Buddhist spirituality. He “*meditates a lot...he was always telling me, just meditate, be positive.*” Similarly her father gave her “*moral support*” by reminding her of their Buddhist beliefs, “*he was just telling me to do good things, be kind to people, help whenever you can...then it will be easier for you to cope...think positively.*” Her husband “*was praying...he was encouraging me.*”

In hospital, her doctors and nurses provided compassionate care and offered positive messages and prayers. One nurse “*was telling me I’m always praying for you. You will get*

a lung as soon as possible. If you get a lung, don't worry...Even the doctor... he said don't worry...we're praying for you." Vidu met other patients who had received lung transplants. She saw they were *"so active and doing this and that."* They gave her a belief that she would be well after a transplant. *"Don't worry, if you get a lung you don't have to suffer like this. You are going to get a lung. Think positively."*

The compassion offered to Vidu had the powerful effect of transforming her negative feelings of giving up life and wishing for death into positive feelings of happiness, a renewal of her will to live, hope to receive a transplant, and an outward turning to others through her desire to extend loving kindness to all:

It gave me hope when I saw these transplanted people and when they were talking about the good side of it. I got positiveness and I was just thinking that I wanted my life back. I wanted to fight for it... I think at that stage I was giving up everything... So when someone said that they prayed for me... I felt that I had to live. I can do a lot more for other people if I live not just to waste my life like this... I was just happy about people because they are praying for me.

Compassion enhanced Vidu's resilience. While she remained in a negative situation, positively she had been transformed mentally and spiritually. Through her practice of loving kindness meditation, she was paradoxically elevated from her physical reality to a spiritual plane of calm and detachment which enabled her to endure her physical suffering:

To calm myself down... I always sent loving kindness to other people, at first to myself and then to others...I forgot about my breathing ...I didn't get angry... It's really a calm feeling and you feel good about yourself and others... you are not getting attached to everything...

I was so happy now, I just wanted to go - I was waiting - I was looking forward to it...I was not scared about the transplant because at that stage I knew that this was the last option for me. If not there's no point in living like this...There's no point in suffering like this.

6.3.3.3. Facing death and living with hope – Margaret

Facing the end of her life was a major issue confronting Margaret. She introduced the subject of death early in her first interview with *"I was in high dependency cause I flat lined three*

times”, and reflected again on her mortality at the beginning of her second interview. She could not hope for a transplant and lived with uncertainty as to how much time she had left. While death occupied a prominent position in Margaret’s consciousness after her life threatening hospital admissions, at the same time she vigorously continued to be engaged with life and have hope for the future.

Margaret discussed the end of her life openly and frankly. She had reflected on this stage of her life and confronted death philosophically and practically. Her personal spiritual belief in reincarnation, a core aspect of her personal life meaning, supported her and enabled her to remain resilient by adding meaning to her life and death and providing a sense of hope. She distinguished between the existential fear of death and the emotion of feeling scared and anxious when unable to breathe. She was not afraid of death, believing in time as infinite, with “*no beginning...no end*”, and offering “*continual, continual learning*”. Margaret believed her death held a purpose and meaning of new life and learning:

I’ve still got a lot more to learn so I think I’ll be reincarnated...I’ve got no fear.

I mean, when you can’t breathe, yeah, you get scared. But that’s an emotion.

Practically, death would represent an end to Margaret’s physical suffering. She had developed a deeper knowledge of her body over the course of living with LAM and she felt that she would know when death was at hand. Her positive experience of the care she had received during her recent hospital admissions made her believe that she would be kept comfortable and allowed to die peacefully:

When it’s time for me to say goodnight my body will let me know...I’ll be happy because I’ll be in so much pain or in so much strife trying to just draw breath that it’ll be like a godsend. No more pain. No more sweating, literally sweating taking a breath...What’s there to be scared of? ...You go to sleep peacefully, especially in hospital...with morphine.

Margaret also saw death as a release for her family of the burden of caring for her and her own sense of being a burden:

I don’t want to be a burden on them. When I was in hospital I did feel like a burden...That’s why I was, like, if I’d just gone they wouldn’t have to worry about anything.

She demonstrated her agency by acting to relieve her family of decision-making regarding her end of life issues. She had discussed with them at what point she no longer wished to be

kept alive with medical treatment and had made notes about her preferred funeral arrangements. Caring for her family in a way that was consistent with her identity as a competent, organised person and “*mother hen*” gave her a sense of control and mediated her feelings of being a burden:

I was just doing notes of what I really want... so then mum can just have this thing and go, okay, that's what she wants... I just like being prepared. It's the PA in me... They all know if I get to a certain way with my health and I can't talk for myself, I've told them at which point basically to pull the plug... Non-responsive, less than a 40 per cent chance of recovery... why put the family through it?

Although she was living in a seemingly hope-less situation with no hope of either a cure for her condition or of receiving a lung transplant, Margaret maintained a sense of hope and purpose for the present as well as the future. Hope lay in her ability to imagine possibilities for her life. She believed it was possible to experience wellness and enjoyment in simple pleasures and her relationships with her family, close friends and pets. Furthermore, she believed it was possible for her to realise her chosen role of “*mother hen*” by helping others. Margaret acknowledged the anxiety and sadness she felt but demonstrated hope in her positive attitude, her optimism and her independent choices:

I think with any disease... out of some really bad adversity whether it be this, whether it be cancer, whether it be you have to have a limb amputated... there's a future. There's still, you wake up in the morning and you get sloppy kisses from your dogs. It's so nice.

Realistically Margaret approached the physical future with “*six month plans because I never know*” but hoped “*to make it to... (her) 50th*”, four years away. Hope was present in her setting herself a goal for six months “*to be as healthy as possible...and enjoy myself...I have my really good days*”.

6.4 Receiving a transplant

Patricia and Vidu's experiences of transplant aligned with their previous illness experience. Patricia remained grounded and practical. When she received the phone call to go to the hospital she did not think about possible outcomes and pragmatically took the attitude that “*you just automatically do what you've got to do.*” Vidu felt shocked to receive her call. She was thrust into unreality, experiencing a sense of disembodiment - “*I knew at that*

moment I was not the real me". She shifted between a state of "fear" and being "not that scared". She tried to hold on to some sense of reality by reverting to her mothering role, preparing breakfast for her daughter before she left for the hospital.

Receiving a transplant was a positive turning point in the women's lives which initiated a change in state from suffering and limitation to renewal and wellness. The immediate postoperative period after transplant constituted a time of transition in which the women adapted to living with their new lungs and learned their medication regime and caring for their health after transplant.

Patricia's resilience and preoperative fitness facilitated a smooth postoperative recovery. She progressed well with no complications and had no difficulty relinquishing her oxygen after her surgery. She had regarded it as an aid only to her survival and activity preoperatively and had not embodied it. As she had accepted her need for oxygen then, postoperatively she accepted that she now no longer needed it. *"I didn't have it."*

Her sense of humour helped her to remain resilient in the postoperative period. She had viewed her illness experience through a humorous lens and she was able to see humour in the postoperative hallucinations she experienced due to her medication:

When I was in the ward after I had been in intensive care, I had really ruthless legs, and the drugs were sending me high... I'm sure I saw the pilot out there and the helicopter. I thought it was coming to take me, and I was struggling to get out of bed... The staff were telling me to keep my legs to myself in intensive care, and I said, I can't help it, they've got a mind of their own.

Embodying her new lungs was a more gradual process for Vidu. During the postoperative transition she experienced nausea, vomiting, bowel problems, chyle leaks and feelings of depression related to new medications as her body adjusted to profound physical change. Vidu also found it difficult to wean from her oxygen therapy and become used to the freedom of breathing only air. She had embodied oxygen therapy during her period of suffering. Although it was itself a metaphor for feeling "chained" and lost freedom, it was also paradoxically a metaphor for life as survival. At a time when feelings of illness were part of her transition experience, it was therefore difficult for her to release the oxygen and its powerful meaning of survival. *"It took some time for me to get rid of oxygen."* As she moved through the transition period and wellness became her dominant experience, she was able to integrate and embody her new lungs, let go of oxygen for survival, and experience

the full positive transformation of transplant and its meaning of life as possibility, renewal, hope and the future.

6.5 Feeling transformed

The three women all felt transformed through their experience of liminality at the late stage of LAM. All experienced personal growth, a deepened spirituality and feelings of empathy and being open to other people and new experiences. Facing death made them less fearful of the end of life and enabled them to accept their human condition of impermanence and live calmly and contentedly in the present in whatever time they had. For Vidu and Patricia, personal growth was also associated with adaptation to a new stable state and moving on with life after transplant. Margaret, in contrast, remained in an unstable state of late stage disease where her illness was foregrounded. Her personal growth enhanced her resilience and enabled her to continue living with struggle with positivity and hope. The transformative growth and learning that took place in late stage liminality became part of each woman's personal life meaning and strengthened her resilience as a personal resource to be mobilised at future turning points.

6.5.1 A transformation of learning and growth

Margaret felt that her experience of living with LAM had been an opportunity for personal growth. As her disease had progressed and her physical and social world shrank, her inner world had expanded, providing space and time for learning and growth. Through this lens, Margaret saw LAM as a "*blessing in disguise*" which broadened her perspectives as she "*learned*" to slow down in attitude as well as physically, be calmer and "*don't... sweat the big stuff anymore*". She felt that her awareness of others grew as LAM "*opened... (her) eyes more*" to "*prejudice, not just me, people towards other people*" and so develop her sense of empathy and tolerance for others consistent with her "*mother hen*" identity:

I've become calmer. Don't stress as much...I truly believe I was given this as a learning curve for some reason...maybe to calm me down. Maybe to make me sit back and not rush through life...Don't take things too seriously. Let go. Don't worry what other people think or say.

Margaret believed her life with LAM held purpose. "*I've always believed you're here for a reason. I believe I've got this disease cause I can handle it*". She hoped to contribute to the understanding of LAM by sharing her experience of living with her illness with healthcare professionals and helping other women live with LAM by showing them "*they're not alone*

if they have this disease” :

LAM is not something that's well known so if there were doctors or nurses that you said... I'll bring a LAM patient along and you can ask questions, I'll be there... Show them the positive...take out the negative.

6.5.2 Moving on with life after transplant

Vidu and Patricia were renewed physically and socially by their transplants. Through the transition after surgery they adapted to their new stable state. Each woman regarded her transplant as a gift and took responsibility for caring for her health.

Patricia was transformed physically and psychologically from the “*lady with the limpy dog*” to a “*social butterfly*”. The butterfly metaphor represents her metamorphosis from the “*struggle*” of slow, limited mobility and breathlessness to “*freedom*” of movement, exercising and socialising, unimpeded by illness, oxygen cylinders and tubing. She regarded her transplant as a “*gift*” that “*you want to make sure you make the most of.*” Physically, “*having a transplant has allowed me to go back to living how I used to live... being really active.*” Patricia, in valuing her transplant, desired to live life fully. She enjoyed her social life, resumed activities she had enjoyed before transplant, learned new ones and participated in sporting competitions:

We've had our transplants and we're moving on...When you have had these things, you don't want to be staying at home sitting in a chair. I think you want to be out. I'm a social butterfly... now I just want to do what I want to do... I'm going to go and learn to play racquetball..., when you've had no complications and the drugs are working well... you have to keep your side of the bargain, and do the right things by yourself.

Receiving a transplant changed Patricia as a person. She felt she had “*become more open to things, more open to suggestions... more patient.*” Patricia maintained a pragmatic attitude towards her future. She understood that a long life was not guaranteed after her lung transplant. Her hope was directed towards wellness in the present by focusing on being as active and engaged with life as she could:

You've got to look at it this way, I've had three and a half years, and each one is a bonus, isn't it?...The expectation can't be there, because nobody knows

when their time is to go... I think you've got to say, you're blessed to have what you've got... lucky enough to get a transplant.

Vidu's transplant extended the transformation she had experienced prior to her surgery. Her negativity was replaced by a positive attitude, happiness, a sense of hope and looking toward the future. She experienced a renewal of her social relationships, particularly with her husband and daughter, and no longer felt a need for the support of her parents. At the time of her interviews she felt she had lived "well" in the eleven months since her transplant and she appreciated the new quality of her life. She had let go of her fear of dying and could live positively in the present. She now saw life as the possibility of action, enjoyment and satisfaction. She felt a sense of gratitude and a strong imperative to help other people. She acted to organise voluntary work at a retirement village and this was a source of satisfaction. She was undertaking a business course and had made plans to seek part time work in addition to her work as a volunteer. She exercised by walking and had taken up yoga. There seemed almost to be a sense of urgency not to "waste" any more time at home but to "do what I want to do". After coming so close to death time was more precious now. Vidu could now foresee a future and had a goal of travelling with her husband:

Now I go for walks and I do everything...I'm really grateful...I don't mind dying tomorrow even because I suffered a lot...At least this 11 months I spent well...I'm trying to find voluntary work because I just want to help people...I feel like I have to do something... I don't want to waste my time... I feel really happy... Everything has changed, the way that I think... now I try to think the good side of it. Earlier it was the other way, mainly because of the illness.

The late stage of advanced LAM was a transition to transplant or death from respiratory failure. The participants experienced it as a liminal existence of instability, uncertainty, struggle and suffering in which they confronted their own mortality and experienced personal growth and transformation. They found meaning in their spirituality and close relationships and retained agency in their active efforts to manage the effects of their illness. This contributed to their resilience and enabled them to live with struggle and suffering.

6.6 Summary

The participants who were living with advanced LAM experienced two levels of illness at this stage. Commencing oxygen therapy marked the first stage of advanced illness while

periods of respiratory failure and being listed for a lung transplant signified the second later stage of severe disease and disabling breathlessness. Most participants used oxygen intermittently initially and progressed to continuous use as their lung function deteriorated. Oxygen therapy represented a paradox as it held both negative and positive meanings for the participants. Negative meanings were associated with the visibility and physical presence of the oxygen equipment and the effects of advanced disease. Oxygen therapy added a new dimension of disruption to everyday life and the participants who needed to use oxygen continuously experienced losses and limitations, threat to their identity, physical and economic burden, and stigma.

Intermittent oxygen therapy was less disruptive as the participants could choose when they used it. This allowed the women who had earlier coped by avoiding their illness to avoid using their oxygen where possible and hide it from others to avoid its negative effects. The participants demonstrated misunderstandings about how they should use their oxygen to gain benefit from it, including using it after, rather than during activities and while driving, and believing they might become addicted if they used it frequently.

Helen and Margaret revealed that the measures applied to assess their oxygen requirements did not take into account their function and activity levels. Helen had to advocate strongly for herself in order to receive adequate supplies of oxygen. It was difficult for the women to access the lighter, portable means of oxygen delivery which they favoured. Portable oxygen concentrators, which would facilitate their ability to improve their health and quality of life through participation in exercise, work, and social activities, were expensive and not subsidised. Liquid oxygen which was lighter, more affordable and suited to exercise and activity was not available in Australia. There were also no educational resources available to support mothers using oxygen therapy and caring for young children.

The participants' view of their oxygen shifted as they accepted it, adapted to it and learned to manage it. They were then able to perceive it positively as helping them to survive at the late stage, and enabling them to be active. Although Sarah did not medically require oxygen, she regarded it as liberating and used it creatively to allow her to exercise more intensely, and retain her identity as a fit and active person. Accessing social support in the form of affordable home services shifted Helen's perception of her life with oxygen as struggle and burden to a more positive view of seeing her oxygen as enabling possibilities for exercise

and activity which aligned with her sense of self. Her mood, self-image and family relationships improved and she felt a greater sense of control and normality.

The participants strove for normality even while living with advanced disease. This contrasts with Pollock BarZiv's (2005) study of lung transplant experiences of women with LAM who adopted a 'sick role' when using supplemental oxygen. In common with this current study, Kelly and Madden (2014), in a critical interpretative synthesis of 42 studies of respiratory patients' perceptions of their oxygen therapy, reported that patients perceived their oxygen both positively and negatively. Likewise, Adams (2008), in a study of five rural patients' perceptions of oxygen therapy, found the patients adapted to their oxygen, accepting that it was both restrictive and enabled greater independence. Kelly and Madden (2014) were unable to elucidate why some patients adapted better than others. This study has found that participants with more advanced disease were able to perceive greater benefits in their continuous oxygen therapy and adapted to it more readily. This was also the case for resilient participants who had an optimistic disposition, higher self-efficacy beliefs, and a strong sense of self and purpose.

The participants using oxygen intermittently had more difficulty adhering to their oxygen. Similarly, Katsenos and Constantopoulos (2011), in a review of patient compliance with long term oxygen therapy in COPD, reported that poor adherence was associated with misunderstandings about oxygen use, perceptions of limitation, bulky and heavy equipment, discomfort, fear of addiction, lack of support, and stigma. According to Goffman (1963), stigma relates to an individual's undesirable attributes or appearing different to what is expected. The stigma which the participants of this current study experienced related to their appearance as young, well-looking women who, although using oxygen, did not fit the expected appearance of a person with a disability. Carel (2013b), as a young woman living with LAM, also reported experiencing stigma when using oxygen.

The participants were highly motivated to engage in exercise and preferred lightweight portable oxygen concentrators to facilitate their physical activity. Encouraging exercise is important for people with respiratory conditions as, according to Watz et al. (2014), in an official European Respiratory Society Statement on physical activity in COPD, regular physical activity reduces decline in lung function and hospital admissions. Furthermore, Dal Negro and Hodder (2012) have argued that lightweight, portable oxygen promotes exercise, mobility, social interaction and quality of life for people requiring long term oxygen.

Funding policy in Australia is influenced by research into the older COPD population. Casaburi et al. (2012), for example, in a study of 22 patients with advanced COPD, found that activity levels did not increase when lightweight oxygen devices were used over a six month period. The lack of funding for portable concentrators does not take account of younger, active people's need for affordable, lighter, portable devices.

The participants reported differences in funding for their oxygen. Similarly, Serginson et al. (2009), in a retrospective observational study of domiciliary oxygen therapy in Australia, found that funding varied across states and access to portable oxygen was inequitable. They recommended a national oxygen register to enable a more equitable supply of oxygen equipment. Five years later, the Thoracic Society of Australia and New Zealand, in the clinical practice guideline for adult domiciliary oxygen therapy, was still calling for this policy change (McDonald et al. 2014). This document also stated that the spot checks of oxygen saturation, which Margaret and Helen reported, are not a suitable means of assessing ongoing oxygen needs.

At the late stage of their illness, the impact on each participant's life was more severe as they contended with persistent breathlessness, and increasing impairment of their function and mobility. Their physical condition was unstable and they underwent periods of hospitalisation. Respiratory failure and being listed for transplant were turning points which initiated a persistent liminality of uncertainty, waiting, and living on the margin physically and socially. Struggle, anxiety and suffering were significant aspects of their experience at this time of transition to either death or transplant. As the women reflected on their mortality, they experienced a deepened spirituality, personal growth and transformation. Receiving a transplant was the turning point which signified Patricia and Vidu's passage from the transition of late stage to stability and wellness as they adapted to living with their new lungs.

Margaret's experience of economic hardship at late stage is reflected in Essue et al.'s (2011) cross-sectional study of 218 patients with COPD attending a Respiratory Ambulatory Care Service in Australia. The authors reported that 78% of the respondents could not afford basic living costs due to their health care expenses. The cost of medications, medical consultations, home oxygen, transport and their inability to access adequate social support contributed to their economic stress. Essue et al. (2011) argued changes in public policy are

required to address these factors and reduce economic hardship for people living with chronic illness.

The participants demonstrated resilience in their ability to accept their situation, adapt to life with continuous oxygen therapy, and live with struggle and suffering. They revealed that personal traits of optimism, humour, and flexibility; self-esteem and self-efficacy beliefs; knowledge, skills and competence acquired from past experiences; and having a sense of hope and purpose were factors which contributed to their resilience. For the women at late stage, finding meaning in their life and spirituality, experiencing connectedness with others, and receiving practical and moral support from family and close friends were particularly important in assisting them to live with their suffering and remain resilient. The participants showed, by setting goals and influencing their situation through their attitude and actions, that it was possible to retain a sense of agency and autonomy despite their difficult circumstances and physical limitations.

The levels of disruption and suffering which the participants perceived corresponded to their individual levels of resilience. Higher levels of resilience were associated with lower perceived levels of suffering and vice versa. Patricia showed that continuing to exercise as she was able throughout her illness elevated her resilience. It was protective for her at late stage, enabling her to cope effectively, perceive less suffering, and make a smooth transition through recovery after transplant. Vidu's avoidance was associated with her lower level of resilience and greater suffering at the late stage of her illness.

6.7 Conclusion

This chapter has presented the participants' experiences of living with advanced LAM through the turning points of commencing oxygen therapy, experiencing respiratory failure, and receiving a lung transplant. Oxygen therapy was a paradox, in both disrupting the participants' lives and enabling them to be active. Negatively, the women felt continuous oxygen therapy was a burden. The visibility of oxygen equipment threatened their identity and engendered feelings of being stigmatised. The women identified problems related to public policy which increased their sense of burden, including inequities in oxygen funding between states, lack of funding for lighter portable oxygen concentrators, assessment criteria for oxygen supplies that did not take their function and activity into account, and a lack of educational resources to support mothers with young children. Positively oxygen therapy enabled the women to stay alive and participate in exercise, work and social activities.

Accepting and learning to manage their oxygen while receiving adequate social support allowed the participants to perceive its benefits.

The participants demonstrated resilience in their experiences of living with advanced LAM. The women who had developed positive self-beliefs, knowledge and skills throughout their lives and in the course of learning to live with their illness had become resilient and they drew on this as a resource at the advanced stage of their illness. Their resilience influenced their ability to accept their oxygen therapy and adapt to it, and their perceptions of their ability to cope with persistent breathlessness and disabling limitations at late stage.

The participants continued to build their resilience as a dynamic process. Mobilising affordable home services, continuing to manage their illness effectively, and participating in work and social life while living with oxygen therapy increased the women's self-efficacy and positive feelings of autonomy and competence. Experiences of spirituality and personal growth, connecting socially with others, choosing to maintain hope and a positive attitude, and receiving compassionate support were sources of meaning and purpose and positive feelings which enhanced their resilience at late stage. As an outcome, resilience improved the participants' ability to manage their health, and enabled them to experience wellness while living with the effects of advanced disease.

These last four chapters have revealed two interrelated concepts weaving through the collective life history, an overarching narrative of resilience, and the influence of the rarity of LAM on the women's experiences. The narrative of resilience encompasses the turning points the participants experienced in living with LAM, beginning with the disruptive life change of their diagnosis and its impact on their expected life plans, sense of self and how they related to the world; the transition period and process of adaptation in which they learned to live with change, integrate their illness into their lives, and restore a sense of self, coherence and purpose; and the influence of the rarity of LAM on their resilience narrative. The following chapter will discuss the findings of the thesis in relation to resilience in the context of a rare condition and in the light of existing literature and theories.

Chapter 7

DISCUSSION

7.1 Introduction

The previous four chapters presented a life history of women living with LAM to provide an understanding of their experiences from the onset of symptoms to the late stage of their illness. The participants showed that they lived with LAM in a context of constant change, from the day to day fluctuations of everyday life to turning points of significant change related to their illness or social world. In their narratives the women revealed diagnosis, a long hospital admission, commencing oxygen therapy, experiencing respiratory failure, and receiving a transplant as turning points when they experienced significant life disruption.

While each participant's life was unique, the women demonstrated resilience as an overarching theme in their experiences of living with LAM. Analysis revealed that resilience was the complex learning that evolved over the course of each woman's life through adaptation to situations of change and adversity, and as a process of everyday life. As a learning outcome, it enabled them to experience wellness while living with the uncertainty and limitations of their rare, incurable condition, and, for those whose illness advanced, cope with the progressive deterioration in their health. Resilience learning was multidimensional and involved the growth of meaning, knowledge and competence. It was a dynamic process influenced by changes in their health status and social contexts and the rarity of their illness. As it accumulated through life it formed a resource that could be drawn on as new turning points occurred.

Resilience has been described as a complex concept that is more easily recognised than defined (Butler 1997). Within a large body of literature examining resilience in varying populations (children, adolescents, the elderly, the chronically ill, minority groups, communities) and contexts (loss, trauma, poverty, life span, organisations, social systems, ecosystems), there is no agreed definition of the concept (Haase & Peterson 2013). Additionally, there is a limited range of literature exploring resilience in chronic illness (Edward 2013). While many studies use the terms resilience and resiliency interchangeably, Luthar et al. (2000, p.546) distinguished between ego-resiliency as a "personality

characteristic of the individual” and resilience as “a dynamic developmental process”. This latter description of resilience applied to the participants’ experiences of living with LAM and resilience is the term used in this thesis.

The participants’ experiences highlighted that resilience and learning were interrelated as they adapted to their illness. Adaptation was a response to specific turning points during a period of transition. It was a way of dealing constructively with change and incorporated problem-focused learning and self-learning. In adaptation the participants’ learning was focused on problems related to their illness. It was a process of acquiring knowledge and skills to manage their symptoms, relationships, responsibilities, emotions and identity in their individual contexts, and communicate with healthcare professionals in healthcare settings. The rarity of LAM stimulated the participants to develop increased knowledge and enhanced communication skills through their need for self-reliance in seeking information, and self-advocacy in accessing expert management and treatment. Self-learning developed in the state of liminality as they reflected on their existence, created meaning, and reconstructed their sense of self through their experiences of spirituality and personal growth. Everyday life was a process of resilience learning by increasing their competence and self-efficacy as they dealt with minor disruptions and the fluctuations of daily life, and by generating meaning and positive emotions in their incidental experiences of social connectedness and enjoyable activities.

Resilience as a learning outcome was the meaning, knowledge and competence the participants gained through these processes, allowing them to integrate their illness into their life. Knowledge and competence in self-managing their illness, in a practical sense, enabled them to participate in family, work and social life, and was a source of meaning in facilitating connectedness with others and generating positive self-beliefs. Finding meaning in their new reality involved perceiving life having value and purpose, and a sense of autonomy and self-efficacy in being able to independently determine life goals, make decisions, adopt attitudes and behaviours, and act to meet their goals. Change became less disruptive and they were able to experience a sense of coherence and wellness.

Women whose illness progressed to late stage drew on resilience they had accumulated through life as a resource. Resilience was also an ongoing process as they created meaning in their experiences of spirituality and personal growth. This learning enabled them to live with struggle and suffering while waiting for transplant or until end of life.

Previous research has associated resilience with adaptation. According to Luthar et al. (2000) and Cicchetti (2010), resilience is a process or capability which enables a person to cope successfully with or adapt positively to adversity. Zautra et al. (2010), in contrast, suggested that resilience is an outcome of successfully adapting to adversity. Common factors and characteristics of resilience identified in the literature (see Table 7.1) included active engagement, motivation, positive reappraisal (finding benefit in one's situation and reframing issues), optimism, competence, self-efficacy, spirituality, growth, learning, and finding meaning. Resilience was also regarded as occurring in the context of person/environment interactions over time or across the life span and was facilitated by a supportive social environment.

This chapter will discuss the findings of the study in relation to resilience, the participants' experiences of living with LAM as a rare disease, and the life history method. It will present a broad concept of resilience as both a learning process and outcome in the context of a rare, chronic illness. Discussion is guided by a model of resilience (Figure 7.1) constructed to provide a visual summary of resilience in the collective life course of the participants and their experiences over time. Horizontally, the model shows the turning points of their illness and social world across their life course. Vertically, it displays the learning processes and outcomes of adaptation and resilience that developed during periods of transition following turning points, the personal and social resources which influenced the process, and processes of everyday life which contributed to their resilience.

The discussion focuses specifically on the following dimensions of resilience learning: turning points; transition; the process of adaptation, including problem-focused learning and self-learning; self-advocacy in the context of living with LAM as a rare disease; how avoidance coping influenced resilience learning; resilience as a dynamic process; living with struggle and suffering at late stage; and the significance of hope and meaning in the process of resilience. Following this discussion, the findings are discussed in relation to the interpretation of experience using life history and Gadamer's (2004) hermeneutic philosophy. Finally, the findings of the study are summarised in relation to the specific aims of the study.

Table 7.1: Resilience factors and characteristics

Resilience Factors	Resilience Characteristics	Resilience Literature
Personal Resources	Optimism	Trivedi et al. 2011; Sturgeon & Zautra 2010; Zautra et al. 2010
	Hardiness	Trivedi et al. 2011; Bonanno 2004; Kobasa et al. 1982
	Empathy	Holaday & McPhearson 1997; Zautra et al. 2010; Norman 2000
	Curiosity	Holaday & McPhearson 1997
	Flexibility	Holaday & McPhearson 1997
	Sense of humour	Holaday & McPhearson 1997; Norman 2000; Vaillant 2003
	Creativity	Richardson 2002; Vaillant 2003; Simonton 2000
	Hope	Butler 1997; Richardson 2002; Gillespie et al. 2007; Horton & Wallander 2001; Haase et al. 2014
	Social competence	Holaday & McPhearson 1997
	Personal control	Holaday & McPhearson 1997; Zautra et al. 2010
	Positive emotions	Ong et al. 2006; Zautra et al. 2010; Cohn et al. 2009; Sturgeon & Zautra 2010; Tugade et al. 2004
	Perseverance	Allen et al. 2011
	Spirituality	Trivedi et al. 2011; DeSantis et al. 2013; Richardson 2002; Nelson-Becker 2006; Hunter-Hernandez et al. 2015; Haase et al. 2014
	Self-efficacy	Trivedi et al. 2011; Holaday & McPhearson 1997; Zautra et al. 2010; Norman 2000
	Self-esteem	Trivedi et al. 2011; Haase et al. 2014
Social Resources	Supportive social environment	Haase et al. 2014; Kralik et al. 2006; Rabkin et al. 1993; Wyman et al. 2000; Zautra et al. 2011; Henderson 1998
Adaptive coping	Active engagement	Trivedi et al. 2011; DeSantis et al. 2013; Kralik et al. 2006; Schanke & Thorsen 2014; Zautra et al. 2010
	Person/environment interactions	Egeland et al. 1993; Gillespie et al. 2007; Butler 1997; Ungar 2011; Resnick 2011; Waller 2001; Edward & Warelw 2005; Polk 1997; Tusaie & Dyer 2004
	Dynamic	Luthar et al. 2000; Gillespie et al. 2007; Cicchetti 2010; DeSantis et al. 2013
	Over time/life span	Trivedi et al. 2011; Egeland et al. 1993; Gillespie et al. 2007; Robertson & Cooper 2013
	Acceptance	Sturgeon & Zautra 2010; Zautra et al. 2010; Nelson-Becker 2006
	Growth	Richardson 2002; Kralik et al. 2006; Sturgeon & Zautra 2010; Nelson-Becker, 2006
	Learning	Kralik et al, 2006; DeSantis et al, 2013; McAllister & McKinnon 2009
	Competence	Norman, 2000; Gillespie et al, 2007; DeSantis et al, 2013; Holaday & McPhearson 1997
	Motivation	DeSantis et al. 2013; Gillespie et al. 2007; Richardson 2002; Resnick 2011; Norman 2000
	Positive reappraisal (finding benefits)	Affleck & Tennen 1996; DeSantis et al. 2013; Kralik et al. 2006; Sturgeon & Zautra 2010
	Finding meaning	Kralik et al. 2006; Sturgeon & Zautra 2010; Fife 2005
	Positive adaptation to adversity	Luthar et al. 2000; Waller 2001; Gillespie et al. 2007; Herrman et al. 2011; Cicchetti 2010
	Altruism	Norman 2000; Richardson 2002
	Wellness	Carel 2013a; Zautra et al. 2011

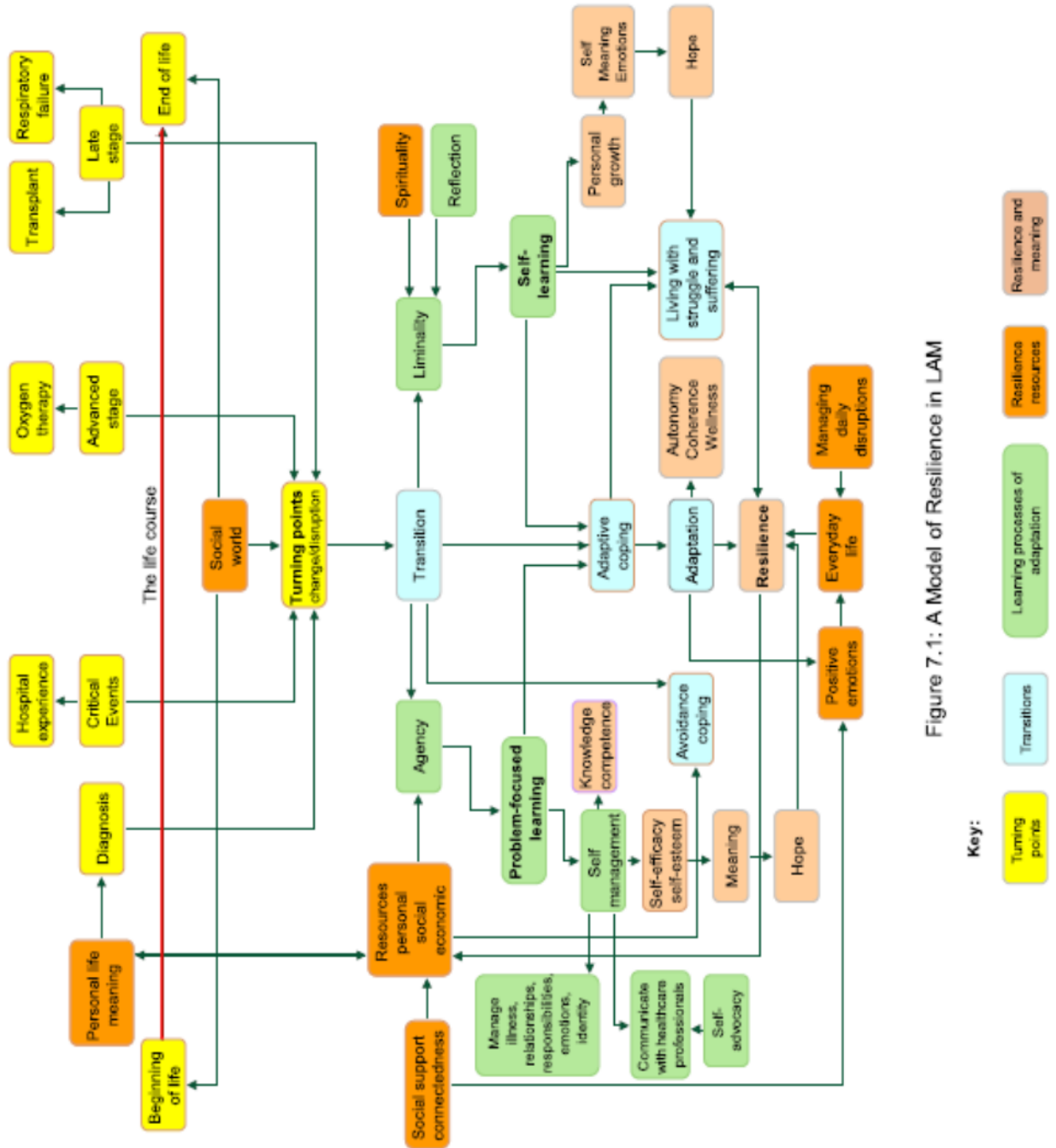


Figure 7.1: A Model of Resilience in LAM

7.2 Turning points

In living with LAM, the participants experienced turning points as significant life changes related to their illness or social world. Turning points could signify either negative or positive change. The women revealed illness-related turning points as being diagnosed, a long hospital admission, commencing sirolimus therapy, starting intermittent oxygen therapy, advancing to continuous oxygen therapy, being listed for transplant, receiving a transplant, and experiencing respiratory failure.

Fifteen participants, as fit and healthy women with no significant medical histories, experienced being diagnosed as a turning point of profound life disruption. Physically they experienced changes in their bodies which impacted on their expected life plans, their sense of self and their interactions in the world. They evaluated the meaning and significance of the changes they were experiencing through the lens of their personal life meaning which incorporated their values, beliefs, disposition, sense of self, past experiences, and future expectations. Their appraisal of their diagnosis as a disruption of their familiar world, personal meaning and sense of coherence reflected Bury's (1982, p. 169) concept of chronic illness as a "biographical disruption" of "the structures of everyday life", including a person's assumptions, beliefs, behaviours, and sense of self. The women's feelings of shock, loss, uncertainty, isolation and shifting sense of self revealed their sense of disruption.

The feelings of disruption expressed by the participants were also felt by other women diagnosed with LAM (Carel 2013b; Pollock-BarZiv 2005) and people with pulmonary fibrosis (Duck et al. 2015; Schoenheit et al. 2011; Swigris et al. 2005), pulmonary hypertension (Kingman et al. 2014) and women with scleroderma (Oksel & Gunduzoglu 2014). The women in this current study described emotions of fear, sadness, grief and anger. These were similarly reported by Carel (2013b), Hoy (2016), and Pollock-BarZiv (2005) and people with pulmonary hypertension and pulmonary fibrosis (Armstrong et al. 2012; McDonough et al. 2011; Swigris et al. 2005).

Bury's (1982) concept of biographical disruption was not true for all participants. Four women experienced their diagnosis as less disruptive and were able to adjust more quickly to their illness due to the meaning they attributed to their situation, their life expectations, and the context of their diagnosis. Patricia interpreted her diagnosis as a continuation of her

experience of tuberous sclerosis, her son's multiple medical issues, and her regular contact over many years with healthcare professionals and the healthcare system. Ruth felt her diagnosis was consistent with her expectation that she would develop cancer, and her own recent experience of breast cancer. Carol felt very little impact on her daily life or expected future. Ursula reconciled her diagnosis contextually by comparing her situation with that of a well-known young celebrity who died tragically and suddenly at the same time. These experiences are consistent with Williams' (2000) argument that the onset of illness is not disruptive for all and that the level of disruption depends on the meaning a person attributes to their situation, their context, timing, purpose, and current expectations. A diagnosis of chronic illness might, for some individuals, represent "biographical continuity... reinforcement" or confirmation when their illness aligns with their past biographical experience (Williams 2000, p. 52).

Patricia, Margaret and Eva who had respectively experienced illness, adversity and the disruption of civil war, were able to interpret their diagnosis in the context of these past experiences by perceiving their illness as a challenge they could manage rather than a threat. Similarly, Williams (1984) argued that people engage in a process of "narrative reconstruction" in which they interpret the meaning of their illness in the light of their past experiences in order to reaffirm a sense of purpose and future and "repair the rupture between body, self, and world". Pound et al. (1998), in a study of older working class people's experience of stroke, found that, although their stroke was a crisis, they perceived it as not an unusual event in their lives. Pound et al. (1998) theorised that their participants' responses may have been mediated by their past experiences of hardship over the course of their long lives and the skills they may have developed over time to adapt to new circumstances and difficulties.

The concept of resilience as lifelong learning proposed by this study provides an understanding of the mechanisms underlying the participants' responses to their illness. Through their past experiences Patricia, Margaret and Eva had learned knowledge and skills in managing illness and difficult situations. This learning strengthened their self-efficacy beliefs and confidence in their ability to manage challenges in their lives. It constituted a resource of resilience which facilitated their initial adaptive response to their diagnosis of attempting to have their needs met by seeking the information, support and expert care they required to effectively manage their health. Resilience learning in the participants' later experiences of living with LAM is discussed further below in relation to adaptation, living

with a rare disease, avoidance coping, the dynamic process of resilience, everyday life, living with struggle and suffering at late stage, and hope and meaning.

7.3 Transition

Turning points initiated a period of transition in which the women learned to live with change. Transition was complex and dynamic as they experienced variations through transition, physically with acute illness events, and subjectively with fluctuating day to day experiences of their illness in their individual changing social environments. This study identified three transitional experiences, adaptive coping, avoidance coping, and living with struggle and suffering.

After the shock of diagnosis had settled, the majority of participants engaged in adaptive coping and the learning processes of adaptation. In contrast, Vidu's experience demonstrated that avoidance coping was associated with her negative personal disposition and the availability of strong family support which, consistent with family and cultural values, encouraged her reliance on family to manage illness-related issues. The late stage of the illness was a transition to transplant or end of life, marked by turning points of being listed for transplant and respiratory failure. At this stage women coped with struggle and suffering.

Transition frameworks have been developed which relate to illness transitions (Chick and Meleis 2010; Kralik and van Loon 2010; Meleis et al. 2000; Schlossberg 1981; Schumacher 1994). They generally include a critical event initiating significant disruptive change, personal and environmental factors influencing perception of change and the process of transition, and adaptation as an outcome of transition. In signifying adaptation as the outcome of passage through transition, current models do not account for the alternate transitional experiences observed in this current study. Kralik and van Loon (2010) suggested that studies of transition and chronic illness are limited by their short-term focus, and that longitudinal studies are needed to examine transition in relation to the passage of time. By examining the adaptive process across the illness trajectory, this study expands current knowledge of transition by differentiating between varying transitional responses, their effects over time, and their relationship to resilience.

7.4 Adaptation – a process of resilience learning

Adaptation, for the participants, was a process of learning to live with their illness, re-establishing their sense of self, and building resilience over time as they developed

knowledge and skills in managing their illness and found meaning in their situation. It involved motivation, active engagement and learning processes of problem-focused and self-learning. Their personal resources of pre-illness health and fitness, social networks, work, self-efficacy beliefs and interests shaped their personal life meaning, motivated them and influenced their perceptions and decision-making as they engaged in the process of adaptation. Personal qualities of optimism, curiosity, flexibility, and spirituality were additional resources for some. They were motivated to restore coherence to their lives, regain their sense of autonomy and competence, fulfil their responsibilities and goals, and experience connectedness through participation in family, work and social life. (See summary of participants' personal and social resources in Appendix 3, Table 3.1). These findings reflect Bury's (1982) claim that the responses people make to chronic illness involve a search for meaning, a re-evaluation of self, and the mobilisation of cognitive, material and social resources.

7.4.1 Problem-focused learning

Learning involved thinking and problem-solving to manage their physical symptoms and the impact of these on their emotions, identity, relationships and responsibilities. The women demonstrated agency by intentionally acting to manage these illness-related concerns. They reframed their goals and expectations, found new ways of performing familiar tasks, and adopted management strategies and new roles to accommodate their illness and balance their emotions in meaningful ways. They learned to negotiate the healthcare system, source information and support, communicate with healthcare professionals, and self-advocate for treatments, services and flexibility in their workplaces. These processes produced knowledge and skills in managing their illness, and were sources of meaning by engendering positive feelings of confidence, autonomy and control to increase their self-efficacy and self-esteem.

The participants' social environments influenced their ability to build their resilience as they managed their physical and emotional health. The women mostly needed practical social support at turning points, especially diagnosis, and late stage, and during acute illness events and periods of hospitalisation. Their resilience was facilitated by support, such as affordable external home services, which encouraged their autonomy and agency, and having access to expert medical care, online LAM resources and communities, and supportive and flexible workplaces. At all stages of their illness, experiencing connectedness with others increased the women's resilience by generating meaning in positive feelings of self-worth and

belonging. In this regard, Ong and Allaire (2005), in a study of the effect of social connectedness and positive emotions on cardiovascular function in older adults, found that social connectedness extended positive emotions and conferred biological resilience by buffering the effects of daily negative emotions.

The participants expressed a need for emotional support and many relied on their own strategies to manage their emotions. These included exercise, meditation, yoga, creative pursuits such as art, and alternative therapies. Their ability to manage their emotions was enhanced by receiving empathic emotional support and communicating openly with their partners. This was difficult for some participants when their partners were themselves in need of emotional support. In limited appointment times the women did not receive emotional support from their physicians.

The strategies adopted by the participants to improve their emotional wellbeing have been identified by other authors. Trivedi et al. (2011), in discussion of resilience in chronic illness, proposed that people who recover from the negative emotional impacts of chronic illness are resilient. They found that exercise improved emotional wellbeing through achieving lifestyle goals and resultant feelings of mastery and improved self-efficacy. Likewise, mindfulness meditation (Elias 2012), and yoga (Fulambarker et al. (2012) were reported to have positive effects on symptoms and quality of life for people with respiratory conditions.

The participants' actions to independently manage their illness can be referred to as self-management. In a metasynthesis of studies of self-management in chronic illness, Schulman-Green et al. (2012, p.136) defined self-management as "a dynamic process in which individuals actively manage a chronic illness". This study found, similar to Schulman-Green et al.'s (2012) findings, that self-management related to transition to integration of the illness into life, and was associated with knowledge acquisition, learning skills of illness management and communication with healthcare professionals, mobilising resources including social support, processing emotions, finding meaning, and personal growth. The findings of this current study extend the concept of self-management by demonstrating its association with resilience in the learning processes and outcomes which developed as the participants learned to manage their illness.

7.4.1.1 Living with a rare disease - self-advocacy and resilience

This study illuminates how the rarity of LAM influenced the participants' illness experiences. The participants experienced problems associated with the attitudes and limited knowledge and awareness of both healthcare professionals and members of the community, and the low numbers of women living with LAM in the Australian context. These factors relating to the rarity of LAM influenced the process of their diagnosis, their interactions with healthcare professionals in the healthcare system and their social relations in the community. In response, many participants engaged in self-advocacy to have their healthcare needs met and this contributed to their resilience.

Medical professionals' lack of knowledge of LAM was the main cause of the participants' negative experiences of searching for a diagnosis, including delayed diagnosis, being misdiagnosed, and, for some, multiple medical consultations and investigations. These experiences reflected those of Carel (2013b), women with LAM in the US (Pollock-BarZiv et al. (2005), and other people with a variety of rare diseases (EURORDIS 2009; Feinberg et al. 2013; Garau 2016; Garrino et al. 2015; Gordon 2013; Molster 2016; Rare Diseases UK 2016; Rare Voices Australia 2013; Vitale 2005). Four women in this current study reported their symptoms being attributed to depression or psychosomatic causes. This was also the case for people with pulmonary hypertension (Armstrong et al. 2012) and pulmonary fibrosis (Duck et al. 2015). These experiences demonstrated the need to raise awareness among medical professionals of considering a diagnosis of rare disease when encountering difficult to diagnose symptoms.

The majority of participants received their diagnosis alone and were given scant information and emotional support, exacerbating their feelings of uncertainty and isolation in being diagnosed with a rare, potentially life limiting illness. This was also reported by Carel (2013b) and other people with rare diseases (Collard et al. 2007; Duck et al. 2015; EURORDIS 2009; Garau 2016; Garrino et al. 2015; Gordon 2013; Jefferies & Clifford 2009; Pollock-BarZiv 2005; Rare Diseases UK 2016; Rare Voices Australia 2013). The participants read and received from some physicians outdated information, sourced from unreliable internet sites such as Wikipedia, concerning survival rates of between one and ten years from onset of symptoms of LAM. This was similarly the case for Carel (2013b) and Hoy (2016) as recently as 2014. These negative experiences of being diagnosed indicate physicians' lack of both knowledge and empathy and, as reported by Huyard (2009), their failure to meet the moral needs of their patients.

Participants who were isolated geographically or by their health status felt marginalised in the healthcare system. They experienced difficulties in accessing expert care, appropriate information, explanations, investigations and treatment, advice concerning social services, and adequate supplies of oxygen. This was compounded by poor communication between physicians and LAM experts, and other healthcare professionals. These findings were also reported for people living with other rare diseases by Rare Voices Australia (2013) and in the EURORDIS (2009) and Rare Diseases UK (2016) surveys. Molster et al. (2016), in a survey of 744 Australian adults living with various rare diseases on their healthcare experiences, found their respondents' healthcare needs were not being met. These people felt their treatment was not holistic and there was a lack of communication between specialist physicians. Molster et al. (2016) suggested that integrated and coordinated care and diagnosis for people with rare diseases in Australia was constrained by the fragmented nature of the structures of the Australian healthcare system, and that there was a need for multidisciplinary centres of expertise in rare disease in Australia. Supporting this proposal, Garrino et al. (2015), in a qualitative study exploring the impact of a rare disease on patients' lives, found that after diagnosis the rarity of the disease was not problematic for people who attended a multidisciplinary Regional Centre for Rare Diseases in Italy. These people reported satisfaction with their care, that their information needs were met, and that they were able to focus on adapting to living with their illness.

Healthcare professionals' lack of knowledge and awareness of their rare disease motivated the participants to be self-reliant in gathering information, and to engage in self-advocacy to have their healthcare needs met. The women demonstrated two levels of advocacy. At an individual level, they advocated physicians for expert medical management, investigations, information, and access to treatment, and individual community members socially and in the workplace to raise awareness of LAM and seek support for their needs. At a collective level, in conjunction with the peer support group, they advocated in the community by fundraising to support LAM research, speaking publicly to raise awareness of LAM, and providing support to other women living with LAM.

Self-advocacy was a problem-focused learning process which enhanced the women's resilience by expanding their knowledge and communication skills to develop their expertise in self-management of their illness. Successful self-advocacy created meaning by increasing their self-efficacy and restoring a sense of autonomy, competence and control. Congruent with these findings, De Santis et al. (2013), in a study of resilience in the context

of HIV infection, found that the ability to master self-advocacy was an important aspect of mastering the disease, gaining a sense of control, and being resilient. Similarly, Hagan and Donovan (2013), in a concept analysis of self-advocacy among people with cancer, associated self-advocacy with learned communication, information-seeking and problem-solving skills and a drive to resolve difficulties which reflected personal values, beliefs and goals. They reported improved self-concept, a sense of control, and adaptation as long term effects of self-advocacy. Likewise, Jonikas et al. (2013), in an investigation of patient self-advocacy among those who received a peer-led mental illness self-management intervention, found that self-advocacy skills could be learned and when practised increased the participants' hopefulness, quality of life and psychiatric symptoms.

Ayme et al. (2008), in a medical essay on patient empowerment and rare diseases, claimed that people with a rare disease have been empowered by their advocacy. This was the case for the participants collectively through their collaboration with researchers, fundraising efforts, and participation in clinical trials. Individually, however, self-advocacy was more complex and could be constrained by the attitude of physicians and personal factors. While the women's power to influence their health outcomes in their interactions with their physicians was strengthened by the expertise they developed over time, physicians retained greater power due to their authority to order investigations and prescribe treatments. The ease and effectiveness of the participants' self-advocacy was dependent on the attitudes and willingness of their physicians to accept their knowledge and share decision-making. Physicians with expert knowledge of LAM were more willing to engage collaboratively with the participants while those lacking knowledge were less inclined to accept the women's self-advocacy.

This issue was accentuated in the acute hospital setting when the women experienced greater vulnerability during acute illness events. The women's advocacy took the form of being vigilant in monitoring their care and educating the health professionals. However, there was a wider power differential between themselves and their care providers in this context and, perceiving a need for diplomacy, they modified their self-advocacy when communicating and offering opinions. Consistent with these findings, Finn (2008, p.191), in an ethnographic study of empowerment in the alpha-1 antitrypsin rare disease community, reported that physicians remained in control and that empowerment for patients meant participating in managing their illness in a subordinate role to their physicians.

Participants of this current study who were assertive and persisted in their advocacy efforts had a high level of self-efficacy and were motivated by a sense of increased need and urgency. This was demonstrated by Helen who had been diagnosed with advanced disease, and Clare during her long hospital admission for persistent chylous effusions. Both Helen and Clare had become well-informed about their rare disease through their own research efforts. Advocating successfully to have their needs met gave the women a sense of satisfaction and control and enabled them to better manage their health. Similarly, Brashers (2000, p.386), in a study of AIDs activism and communication patterns between people with HIV/AIDs and healthcare professionals, found that both collective advocacy and self-advocacy were associated with education about the illness, taking an assertive/confrontational stance toward healthcare, and “mindful nonadherence” to physicians’ orders.

Individually, some participants reported that the lack of knowledge of LAM in the community and their general appearance of being healthy meant that, at times, they encountered a lack of understanding for their issues and felt stigmatised in their social networks, workplaces, and the general community. This was similarly reported by other women living with LAM in relation to using oxygen therapy in the community (Carel 2013b; Pollock-BarZiz 2005). Yorke (2014), in a qualitative study of the impact of living with pulmonary hypertension, found that their participants’ feelings of being stigmatised was compounded by the invisible nature of the condition and others’ lack of knowledge of the disease.

Collectively, in the Australian context, the very small number of women with LAM meant that there were few available to run the peer support organisation, fundraise and support other women. These roles took time and energy and, at times, involved the use of personal resources. While collective advocacy provided a sense of purpose and empowerment, it could also be a burden. Women who engaged in fundraising could feel a sense of guilt in asking friends and community members for donations when, due to the invisibility of LAM, they appeared healthy. No studies have reported on experiences of collective advocacy in the community for people with rare diseases.

Not all participants engaged in self-advocacy. Hagan and Donovan (2013), in a study exploring ovarian cancer survivors’ experiences of self-advocacy, suggested that further research is needed to identify people who are unable to self-advocate. This study has distinguished personal factors which limited the ability of some participants to self-

advocate. These included their disposition, self-efficacy beliefs, health status, competence in speaking English, and coping style. For example, women who had a negative disposition, less confidence, lower levels of self-efficacy, and who adopted an avoidance style of coping tended not to self-advocate and their interactions were directed by their physicians. This was also the case at late stage for women such as Margaret who struggled with breathlessness and limited mobility. She was unable to travel to see a LAM expert and so was isolated from expert care, and could not self-advocate or access potential treatments.

This study has revealed the complexity of self-advocacy in the context of living with a rare disease and how engaging in self-advocacy helped build the participants' resilience. In a positive sense, the women's resilience and sense of empowerment were enhanced when they were able to engage in collaborative relationships with their healthcare providers and share decision-making. Negatively, having to assume responsibility for research and educating healthcare professionals, and being forced to take an assertive stand in interactions could be a burden, particularly in the acute care setting. According to Huyard (2009), difficult rare disease experiences were caused by the failure of healthcare professionals to meet the moral needs of their patients rather the rarity of the disease itself. This current study found, in the Australian context, that the rarity of LAM did exert an influence on the participants' difficult experiences through the knowledge, awareness, and attitudes of both healthcare professionals and community members. These affected the women's experiences in the health system and their broader social relations.

7.4.2 Self-learning in liminality

For six participants, self-learning created meaning and reconstructed their sense of self through a process of reflecting on their existence in the transitional state of liminality. Although this was a state of negative feelings of instability and ambiguity, it was also a space in which they broadened their self-understanding through their experiences of positive personal growth and deepened spirituality. This learning assisted them in managing their emotions by producing feelings of calm, peace and contentment and a desire to live in the present, and created meaning in enhancing their feelings of self-worth and renewing their sense of self. While it was an inward process, personal growth simultaneously enabled them to look beyond themselves and feel a sense of connectedness with others which was a further source of meaning and purpose. Eva, for example, expressed this as empathy and altruism in her desire to become involved in volunteer work. Reed (1991) defined this process of expanding the self inwardly in reflection and outwardly by reaching out to others as self-

transcendence, and recognised that, for people living with a life-threatening or terminal illness, it was associated with wellbeing. Similarly, Coward (2000) found that self-transcendence was associated with finding meaning and improved quality of life for women with breast cancer. Not all participants of this current study experienced transition as a liminal period. Women with very mild disease, for example, who felt less disrupted by their diagnosis, adapted and passed through transition without experiencing a sense of existential threat.

Liminality has been identified in other studies of chronic conditions, including the frail elderly (Nicholson et al. 2012), the disabled (Murphy et al. 1988), those living with AIDS dementia (Kelly 2008), HIV/AIDs (Bruce et al. 2014), chronic kidney disease (Molzahn et al. 2008), chronic pain (Jackson 2005), and cancer (Blows et al. 2012; Little et al. 1998). Sabo (2014), exploring the experience of caregivers of stem cell transplant recipients, associated liminality with transformation and creating meaning. Similarly, Little et al. (1998) found that meaning was constructed and reconstructed during sustained liminality. Most studies, however, associated liminality with negative feelings, including uncertainty, loss, disruption, fear, grief, and isolation. Kralik and van Loon (2010, p.29) described “limbo” as a commonly experienced phase of transition associated with suffering, isolation and powerlessness. This current study has increased understanding of both liminality and resilience by uncovering the meaning-making processes of liminality as mechanisms of resilience while living with chronic illness.

7.4.3 Resilience as an outcome of adaptation

Resilience was an outcome of the participants’ adaptive learning when they moved through transition and integrated the reality of their illness into their lives. It related to acceptance of their situation and the ability to experience wellness while living with their incurable progressive illness. Wellness was associated with positive feelings of normality, coherence and stability as they participated in social life and found meaning in their situation which aligned with their life meaning. For some participants this was enhanced by personal growth.

The women had retained their agency in being able to determine and act to achieve their goals, and regained a sense of autonomy, competence and control through their self-management and self-advocacy. The knowledge and skills which they acquired increased their self-efficacy, giving them confidence that they could manage day to day fluctuations in their health as they arose. Life was no longer dominated by their illness. It assumed a background position and there was a sense of being able to move on with everyday life.

Congruent with these findings, other studies of chronic illness have found that people with chronic illness can move from a focus on illness to one of wellness as they adapt to their illness. Paterson (2001), in a metasynthesis of qualitative research on chronic illness, found wellness in the foreground was associated with illness knowledge and skills, support, appreciation of life and close relationships, supporting others and spirituality. Whittemore and Dixon (2008), in a study exploring how adults with chronic illness integrate the illness experience into their life, additionally found managing emotions was an important aspect of moving from living an illness to living a life. Further, according to Ellis-Hill et al.'s (2008) life threads model, a person can manage their emotional responses to acquired disability and move on with life by recreating a positive sense of self.

According to Carel (2013a), resilience for people living with illness correlated with personal growth, living in the present, and experiencing wellbeing. Haase et al. (2014), in discussion of a Resilience in Illness Model, associated resilience with confidence/mastery, self-transcendence and self-esteem. The experiences of the participants of this current study of moving on with life contrast with Belkin et al.'s (2014, p.3) finding that the "primary psychological experience" of living with LAM was one of "getting stuck with LAM".

7.5 Avoidance coping

Avoidance coping was an outcome of transition which enabled adjustment and a sense of control and normality while a participant's illness remained relatively stable. However, it was associated with lowered resilience as their illness progressed. Vidu demonstrated that, in coping by avoidance, she had not engaged in the learning processes of adaptation. The strong social support she received from her family limited her resilience through her over reliance on them. She did not develop the knowledge and skills of self-management nor the personal resources of competence, confidence and self-efficacy associated with adaptive learning. Subsequently, she lacked the resilience to sustain her coping as her illness advanced. Vidu's perception was one of illness and she felt restricted and isolated by her need for continuous oxygen therapy. At the late stage, in the months before her transplant, she perceived her situation to be one of profound suffering which she struggled to endure.

Congruent with these findings, Stanton et al. (2001) asserted that coping by avoidance temporarily reduces the effects of severe stress but, in the long term, is associated with poor psychological adjustment and impaired coping, and that adjustment is impeded by support that results in dependence and lack of control. Vidu's situation revealed that social support which encourages autonomy and agency is more effective in building resilience than support

which fosters dependency. This finding is supported by Weinstein et al.'s (2012) claim that environments that support people's need for autonomy, competence and relatedness facilitate their ability to adapt to new situations and challenges. Similarly, Sturgeon and Zautra (2010), in a study of resilience in chronic pain, reported that resilience develops in environments that reward efforts to achieve growth and recovery.

7.6 The dynamic process of resilience

The participants adapted to their illness and built resilience as a dynamic process shaped by changing environments and individual perceptions of illness or wellness over time. Although LAM as a disease process followed a linear trajectory from the initial onset of symptoms to progressive breathlessness and loss of lung function requiring oxygen therapy and, at the late stage, transplantation or eventual respiratory failure, the women experienced the illness differently. The women's physical manifestations and progress of LAM varied from minimal and stable symptoms to rapidly progressing disease with marked impact on daily life, and disease which advanced over time to the late stage. Likewise, the process of adaptation and building resilience was a continuous and fluctuating process over their life course which developed individually both temporally and experientially for the women. It was influenced by their personal life meaning, social context, and past experiences. Some adapted more quickly, others experienced a longer period of adjustment. Resilience learning that accumulated through life influenced their perceptions of their situation and facilitated their ability to adapt and deal constructively with their individual level of change and experience a sense of wellbeing.

Changes in the participants' health or social situation could constitute turning points and shift their perceptions either positively or negatively. Some participants felt their wellness and resilience was diminished when they encountered new disruptions which they felt they did not have the resources to deal with. This initiated new periods of transition and adaptation. For example, losing a valuable work role created emotional and physical disruption for Louise and shifted her perspective negatively. Conversely, wellness and resilience increased when they perceived changes as positive. This was the case for Aiko who found meaning in a renewed will to live, sense of purpose, and connectedness with her family after the birth of her grandson. Similarly, organising affordable home services renewed Helen's sense of autonomy and shifted her perception of her oxygen therapy from being restrictive to enabling her to engage in enjoyable activities and quality social interactions with her family.

Eight participants experienced commencing sirolimus therapy as a positive turning point. It stabilised their lung function and resolved other issues, including chylothorax and lymphangioliomyomas, giving the women physical stability and improved function. Their resilience increased and quality of life improved as they found meaning in their renewed capacity to participate in social life, sustain hope for a longer life, and to achieve goals.

Resilience could fluctuate with shifting perceptions of wellness and illness in everyday life. Jess, for example, although managing her illness competently, and physically stable on sirolimus, was socially isolated and experienced persistent fatigue which foregrounded her illness so that she primarily viewed herself as a sick person lacking agency. This perception limited her self-efficacy beliefs and resilience. When exercising her dominant perspective was one of wellness. When she felt the effects of fatigue it shifted to illness.

Resilience could develop at any stage. This was demonstrated by Vidu when, at the late stage of her illness, she received compassionate support from healthcare professionals and other patients during a hospital admission. This turning point enabled her to find a positive meaning in her situation and shifted her perception of her suffering. Perceiving empathy lessened her distress, enabled her to view her life as having value and purpose, and motivated her to reach beyond herself and connect with others by practising loving kindness meditation. This spiritual practice reduced her anxiety, enabled her to better manage her breathlessness and, in being freely chosen, gave her a sense of autonomy, balancing her dependency on others. Her positive feelings of hope and self-worth increased her resilience and enabled her to endure her suffering. Vidu's experience is consistent with Gillespie et al.'s (2007) assertion that resilience can be developed at any stage. Likewise, Fredrickson et al. (2008) found that a loving kindness meditation intervention could induce a wide range of positive emotions to build personal resources of self-acceptance and feelings of competence in managing life situations.

The participants' experiences of changes in their health and social context showed that they were able to shift their perceptions to wellness and increase their resilience when they found meaning in their situation. This occurred when they felt a sense of purpose, gained a sense of autonomy and agency, and experienced connectedness with others. Their experiences of shifting perceptions of wellness and illness aligns with Paterson's (2001) Shifting Perspectives Model of Chronic Illness which holds that illness changes, social situations and life events can shift perspective towards either illness or wellness. The findings of this current study extend Paterson's model by linking it with resilience. Being able to cope with

or adapt to changed social circumstances and find positive meaning in them increased feelings of wellness and enhanced the participants' resilience. Perceptions of illness and a loss of agency lowered their resilience.

7.7 Resilience as a process of everyday life

The participants built their resilience not only through the process of learning to manage their illness but also as a process of everyday life through their daily experiences of positive interactions and dealing with the fluctuations of day to day life. Resilience here was derived from their experiences of competence, meaning and positive emotions. They felt positive emotions in their daily incidental experiences of happiness, humour, work and social connectedness, as well as their chosen strategies of exercise, creativity, yoga, meditation, spirituality, and altruistic activities such as volunteer work, fundraising for research and supporting other women with LAM. These activities were a source of meaning by increasing beliefs of self-worth, self-efficacy and self-esteem, and life having value and purpose. Positive emotions counteracted their negative emotions and enhanced their feelings of wellbeing.

Luthar et al. (2000) suggested that significant or severe adversity is a core condition of resilience, but the concept has since been expanded to include the notion that resilience develops also through everyday life disruptions (Allen et al. 2011; Richardson 2002). This study revealed that resilience accumulated throughout life in everyday experiences as well as through the process of adapting to negative change and adversity. Masten (2001, p.226) referred to the "ordinariness" of resilience, and Wyman et al. (2000, p.155) to "cumulative competence", suggesting resilience and learning accumulate through life.

7.8 Living with struggle and suffering

The late stage of living with LAM was a significant turning point. Being listed for transplant and experiencing respiratory failure marked entry to a transition of a persistent state of liminality between life and death, characterised by struggle and suffering. Bruce et al. (2014, p. 37) referred to the uncertainty of living with a life-threatening illness as a "pervasive liminality". This transition revealed resilience as a resource, a way of being, and an ongoing process.

As a resource, resilience influenced the women's perceptions of their situation, reflecting their degree of confidence in their ability to cope. Margaret and Patricia, for example, had high levels of resilience. By late stage they had developed a stable self-concept and a strong

sense of personal control and self-efficacy beliefs. They appraised their situation as one of struggle rather than suffering. This contrasted with Vidu who, with lower resilience, perceived herself as suffering.

Resilience as a way of being was evident in their choice of attitude, ability to find meaning and purpose in their situation, sustain hope, and apply the self-management strategies they had learned over time. Their selection of strategies were meaningful in reflecting their sense of self and personal goals. For example, Patricia continued to exercise gently, Margaret persevered with living independently and managing her breathlessness by distracting herself with art activities, music and the companionship of her pets. Vidu practised loving kindness meditation to diminish her suffering. Margaret and Patricia had optimistic dispositions and chose to use humour to maintain a positive attitude. Maintaining a sense of social connectedness and finding meaning in ordinary experiences contributed to the women's resilience.

Margaret's use of humour was an important strategy to sustain her positive emotions and balance her negative feelings of anxiety in her experience of respiratory failure. Expressing humour when interacting with nurses while in hospital, and emphasising the positive aspects of her experience as she narrated her life story was a strategy which Langston (1994) identified as "capitalizing". He found it amplified positive effects and boosted self-esteem. Margaret also overestimated her personal control in managing her breathlessness in hospital, not acknowledging the multiple medical emergency calls made for her acute anxiety. Taylor and Armor (1996) termed this effect "positive illusions" and suggested that it was a way of actively restoring a sense of self during extreme adversity when self-beliefs are threatened. They further asserted that such illusions were contained as an individual gradually processed and integrated negative information. This was the case for Margaret who recognised and incorporated her possible mortality in later formulating an end of life plan. This action created meaning in being an expression of her agency, autonomy and control. Nelson-Becker (2006) similarly found that terminally ill older adults valued independence through the process of dying. In experiencing both positive and negative emotions Margaret demonstrated the emotional complexity which Ong and Bergman (2004) and Ong et al. (2006) stated is characteristic of high resilience people who are able to experience and regulate both negative and positive emotions and maintain a boundary between them. Similarly, Tugade et al. (2004), Moskowitz (2010), and Bonanno (2004) found that people who demonstrate resilient traits and have built resilient resources throughout life are able to

use emotional knowledge to regulate their emotions and enhance coping efforts during negative experiences of adversity.

Resilience continued to develop at late stage through the women's experiences of spirituality. Spirituality was a source of meaning, purpose, and growth through their ongoing self-learning. This strengthened their self-efficacy, self-esteem and self-worth and generated positive emotions to assist them in balancing their anxiety. Spirituality additionally provided connectedness with a higher power which was a source of support, comfort and hope. The three women felt an expansion of their inner world and a connectedness to others through their spiritual practices and expanded feelings of empathy. This enabled them to transcend their limitations and live with their struggle and suffering.

Spirituality has been recognised as a resilience factor more generally (De Santis et al. 2013; Kuhlman 2007; Richardson 2002) and for people living with terminal illness (Nelson-Becker 2006; Vanistendael 2007). Fredrickson (2002) furthermore stated that spirituality contributes to positive emotions through finding positive meaning. Reader (2010) linked spirituality to liminality and learning. He referred to spiritual resilience as the capacity to remain in the uncertainty of liminality through openness and flexibility in order to develop the learning of self-understanding.

7.9 Hope and resilience

Hope was the participants' ability to perceive meaning in their changing circumstances and encompassed their beliefs, perceptions of future possibilities, sense of purpose, and goals. Hope motivated and assisted the women in coping with an uncertain future. It was both a resource and a way of being resilient in the context of living with their incurable, progressive condition.

Sarah saw hope as essential to successful adaptation and moving on with life. Eva, Irena and Margaret associated hope with meaning, and a peaceful and purposeful existence. Over her long period in hospital Clare's resilience was evident as she sustained hope for the resolution of her chylothorax through her practice of positivity and gratitude. Women with children hoped to survive to see their children grow up. Medical research was a source of hope for a cure. The effectiveness of sirolimus treatment in stabilising lung function gave women hope for a longer life.

Spirituality, social support and self-efficacy beliefs supported hopeful thinking at late stage. Patricia and Vidu sustained hope for a transplant. Margaret hoped for survival and quality

of life even though a transplant was not a possibility for her. She maintained a positive belief in her self-worth and ability to achieve her goals of health, quality of life, learning, and connecting with and helping others. Hope was, at the same time, realistic in her awareness of her mortality, planning for her end of life, and her faith in the possibility of a peaceful death. Similarly, in a study of families living with chronic illness, Mattingly (2010) found hope was a practice of creating meaning in the presence of suffering. Hickey (1986) and Borneman et al. (2014) found that hope can coexist with fear and uncertainty for people with cancer and that this is related to realistic awareness of their situation.

Previous research supports the connection between hope and resilience. Gillespie (2007), in a concept analysis of resilience, identified hope as integral to resilience. Similarly, Ong et al (2006), in a study of daily stress and emotion in older adults, found that hope was an important source of resilience and reduced the intensity of stress. According to Snyder's hope theory (2002), hope is a person's perception of their ability to find pathways to their goals and be motivated by agency to use them. Further, supporting the connection between resilience and learning, Snyder (2002) asserted that hope is learned through life in social interaction and is an enduring way of thinking applied to particular situations. The participants highlighted the cumulative effect of hope and resilience through the mechanism of meaning. Having hope contributed to their resilience by being a source of meaning and motivation to cope with their illness, and hope was also facilitated through the process of finding meaning as an aspect of resilience learning.

7.10 Meaning and resilience

Through their experiences, the participants highlighted that meaning, learning and resilience were interrelated. Their personal life meaning motivated and directed their engagement with their illness and, in turn, resilience learning was a process of making meaning as they gained competence in self-management, developed spirituality and personal growth, and experienced social connectedness in everyday life. The positive self-beliefs, hope, and sense of life having value and purpose that were generated in these processes enabled them to integrate the reality of their illness into their life, re-establish a sense of self, experience wellness, and cope with illness progression and other life changes.

Visser (2008), defined learning as interacting constructively with constantly changing environments throughout life. Visser (2008) and Land et al. (2014) argued that learning involves the creation of meaning and self through life experience and develops in the context not only of significant change but also through the fluctuations of everyday life. As the

participants' experiences attested, learning, according to Visser (2008) and Patel and Yoskowitz (2005), has been associated with adaptive, health-related behaviours and reflects beliefs, values, emotions, context, and past knowledge and experiences in individual decision-making. This concept of learning identifies the connection between resilience and learning revealed by this study and encompasses the dynamic, developmental and transactional character of resilience as well as the meaning, knowledge and competencies associated with it.

The participants were motivated by their relationships and their desire to experience autonomy, coherence, wellness, connectedness with others, and fulfil their responsibilities competently. They found meaning when they had learned to live with their illness and mastered managing it, and were again able to achieve goals and participate in work and social life. These findings are consistent with self-determination theory, articulated by Deci and Ryan (2000), which holds that people have a basic psychological need for autonomy, competence and relatedness. The participants demonstrated the view of Weinstein et al. (2012) that meaning was found when their goals and actions satisfied these needs.

Resilience was multidimensional. The participants' experiences revealed a constant interplay of meaning, agency, and interactions in their changing physical, emotional, spiritual, and social environments. Wong and Wong (2012) proposed that programs to build youth resilience should be comprehensive and meaning-centred to address the multidimensional nature of resilience. They identified dimensions of resilience as interactions with changing environments, behaviours, motivation, social relations, emotions, spirituality, and cognition. These dimensions were demonstrated by the participants of this current study and relate to meaning and learning. Likewise, Edwards (2013), in a review of chronic illness and resilience, distinguished biological, psychological, emotional, social and spiritual domains of resilience.

This study has demonstrated that meaning developed across the course of the participants' lives, shaped their perceptions, decision-making, and experiences throughout the course of their illness, and was integral to their learning, adaptation and resilience. Frankl (1984) claimed that a person's main motivation in life was to find meaning and they did this by doing deeds, experiencing or creating something, encountering others, and in the attitude they chose to suffering. The participants similarly found meaning in their attitudes, agency, experiences, and interactions as they adapted to their illness. Other authors agree that the formation of self and meaning involves agency through one's choice of action and attitude

(Land et al. 2014; Murray Parkes 1971; Dewey 1916), and that a sense of agency is associated with resilience (Wong and Wong 2012).

The participants demonstrated resilience as an outcome of their adaptive learning in their ability to experience positive feelings of wellness while living with their illness. The wellness they felt as they integrated their illness into life was associated with finding meaning that was congruent with their personal life meaning of self, values and beliefs. Wellness included functioning optimally within the limitations of their illness and feeling a sense of emotional wellbeing. Wellness, according to Lightsey (1996, p.591) is “a broad range of positive outcomes, including physical and psychological health”, with psychological health equivalent to subjective wellbeing.

In considering the role of positive emotions and meaning in building resilience and wellbeing, it has been suggested that positive emotions precede experiencing life as meaningful. King et al. (2006) found a strong correlation between positive emotions and meaning in life and claimed that positive emotions predispose people to feeling life has meaning. Similarly, Ong et al. (2010, p.89) claimed that positive emotions are the primary mechanism by which everyday experiences build resilience by promoting “adaptive flexibility”. Using Fredrickson’s (1998) broaden-and-build theory of positive emotions, Tugade and Fredrickson (2007) proposed that the effects of incidental experiences of positive emotions accumulate over time to enhance wellbeing, build resilience and form a personal resource.

In contrast, this study argues that the participants’ resilience, in the context of living with a rare, chronic illness, was closely related to the process of finding meaning, and they experienced wellness and positive emotions as an outcome of this process. This echoes Frankl’s (1966) assertion that happiness is an effect of meaning being fulfilled. The participants, for example, during liminal periods of uncertainty and shifting sense of self, felt positive emotions and greater emotional stability when they found meaning through their experiences of spirituality and personal growth. Similarly, Margaret’s strategy of “capitalizing” during her experience of respiratory failure might also be viewed as her effort to create meaning by expressing her optimism and competence as a reflection of her sense of self. In accordance with these findings, Park et al. (2008), in a study of meaning-making in cancer survivors, reported that the process of making meaning produced an outcome of psychological wellbeing.

Day to day experiences which built the participants' resilience throughout their lives were a source of both positive emotions and meaning. While their experiences of happiness in daily life gave rise to positive emotions in a present sense, it was finding meaning in their experiences that made an enduring contribution to their construction of self, wellbeing and resilience throughout life. This finding is explained by a closer examination of wellbeing. King and Hicks (2012) distinguished between hedonic wellbeing as a subjective feeling state of pleasure or happiness, and eudaimonia as the experience of values and meaning in life. Reflecting the enduring quality of meaning, Baumeister et al. (2013, p.505) claimed that "happiness was largely present-orientated, whereas meaningfulness involves integrating past, present and future" and is related to acts that reflect the self and positively help others.

It is conceivable that meaning and positive emotions may act synergistically to increase wellbeing and resilience, and the dominance of each may vary in different contexts at different stages of life. King et al.'s (2006) finding that positive emotions predicted meaning, for example, was based on research with university undergraduate students and meaning as related to personal goals. Finding meaning in a broader sense and experiencing eudaimonic wellbeing may be more important in building resilience in contexts such as the onset of chronic illness where a person's life meaning has been disrupted by their diagnosis.

Previous research related to older adults and chronic illness has connected learning with the cognitive and competency aspects of resilience. Lewis and Harrell (2012, p.348), in discussion of resilience in older adults, conceptualised resilience as "continued competence across the lifespan". Optimism and positive emotions, associated with resilience, have been shown to be skills that can be learned through behavioural and cognitive interventions (Moskowitz, 2010; Seligman, 1991) and loving-kindness meditation interventions (Fredrickson et al. 2008). In relation to chronic illness, Kralik et al. (2006, p.199) stated that resilience was associated with knowledge and skills and could be strengthened by participatory action research as "a process of reflection, learning and action focused toward overcoming adversity". Further, according to Rosenbaum (1990), skills and behaviours which influence a person's ability to adopt and maintain health practices are learned and he referred to these as learned resourcefulness. Likewise, Trivedi et al. (2011, p.181) asserted that resilience is "an ability that can be fostered". This study has presented a broader concept of resilience learning which, while inclusive of knowledge and competence as essential to illness management, identifies the creation of meaning as integral to building resilience in the context of chronic illness.

7.11 Interpreting the meaning of the experience of living with LAM through life history

This life history study revealed the meaning of the participants' experiences of living with LAM through levels of interpretation and the reconstruction of the temporal order of their experiences in the construction of the life history. Gadamer's (2004) hermeneutic philosophy and the view that "human life is a process of narrative interpretation" (Widdershoven 1993, p.2) was reflected in the women's life stories. On a personal level, the participants' lives involved constant change which they interpreted against their personal life meaning. This background meaning of their personal, family, society and cultural values and understandings, consistent with Gadamer's (2004) concept of "prejudice", influenced how they interpreted their experiences and interactions and informed their perspectives and view of the world. Gadamer (2004) referred to a person's perspective as their "horizon" and asserted that understanding is always the fusing of past and present horizons, a "fusion of horizons". Similarly, meaning constantly evolved and shifted as the participants interpreted situations they were experiencing and fused their present perspectives/ horizons with their past perspectives. Engaging in the learning processes of adaptation enabled them to interpret their illness in a more positive light and integrate its meaning into their life meaning to build their resilience and move from an experience of disruption to one of wellness. Interpreting an experience negatively could lower their experience of wellbeing and resilience.

The research process involved further levels of interpretation. The participants deepened their understanding of their experiences as they reflected on them and interpreted them in the telling of their life stories in the context of the interviews. Moreover, my own background/prejudices and the "standpoint" I took of openness and privileging the participants' accounts (Frank 2000) influenced the conduct of the research, my interpretations of their told stories, and the presentation of the interpreted stories in the thesis.

Constructing the collective life history of the participants concerned the issue of temporal order as well as interpretation. The order of the women's stories as they told them was subjective rather than temporal and linear. The women ordered them according to the significance they placed on different experiences and events from their present perspective, and, in the context of the interviews, as a response to the main purpose of the research inquiry to uncover the meaning of living with LAM.

The collective life history was a co-created construction. It represented both the participants' interpretation and telling of their experiences as they constructed their life stories in interaction with me in the interviews, and my interpretation and reconstruction of these told stories through the analysis undertaken. Constructing the collective life history in chronological order connected the women's experiences and, in Mishler's (1995, p. 96) words, "reconstructed the order of the told". The presentation of the participants' shared story in this way has provided an understanding of their experiences of living with LAM as a progressive illness over time. Mishler (1995, p.90) referred to this model of narrative analysis as "reconstructing the told from the telling".

Through these levels of interpretation and the construction of the life history this research has uncovered and enriched the meaning of the women's experiences and demonstrated Gadamer's hermeneutic view that understanding continually evolves with new experiences and interpretations. Readers of the research will develop meaning further as they bring their own prejudices and horizons to their interpretations of the research findings.

7.12 Summary of key findings

This study set out to answer the research question: what are women's experiences of living with LAM over their life course? Using life history the study has explored and captured a comprehensive view of these experiences. The findings of the study are summarised in relation to its specific aims of increasing understanding of the meaning of women's experiences of living with LAM over time across the illness trajectory, understanding how the rarity of LAM affected the women's experiences, and identifying issues of concern for women living with LAM. This knowledge fulfils the aim of the study to enhance the existing body of nursing knowledge to inform the practice of nurses and other healthcare professionals. It thereby may facilitate their ability to improve the care and support provided to women with LAM.

7.12.1 The meaning of women's experiences of living with LAM over time

- The women experienced turning points of significant life change related to their illness or social world over time.
- Turning points of negative disruptive change were:
 - Being diagnosed
 - Critical illness events such as being in hospital for treatment of pneumothorax, chylothorax or renal angiomyolipoma

- Starting oxygen therapy and advancing to continuous oxygen therapy
- Being listed for transplant – signified the late stage of LAM
- Experiencing respiratory failure
- Stopping work due to illness progression
- Social changes such as a death in the family, losing a valued work role
- Positive turning points included:
 - Starting sirolimus therapy – stabilised lung function, resolved lymphangiomyomas and chylothorax, and provided a sense of hope.
 - Receiving meaningful social support e.g. affordable home services
 - Receiving compassionate care at late stage
 - Receiving a transplant
 - Social change such as a birth in the family or adopting a child.
- Being diagnosed was associated with a sense of life disruption and feelings of shock, loss, uncertainty, isolation and a shifting sense of self, and emotions of fear, sadness, grief and anger.
- Diagnosis was less disruptive for women who had experienced significant illness or adversity through their lives, or who were mildly affected by LAM with minimal impact on their daily life.
- Turning points initiated a period of transition in which the women learned to live with changes they experienced physically, personally, emotionally and socially.
- Living with LAM was characterised by three transitional experiences:
 - Adaptive coping
 - Avoidance coping
 - Living with struggle and suffering at the late stage of LAM – a transition to transplant or end of life
- Adaptation was a process of learning to live with change. It involved:
 - Mobilising personal resources of personal qualities, existing resilience, and spirituality; economic resources; and social resources of social support and connectedness. Participants preferred support that was positive and focused on wellness.
 - Problem-focused learning to self-manage their illness, including managing symptoms, relationships, responsibilities, emotions and identity, and communicate with healthcare professionals through personal agency and self-advocacy.

- Self-learning in the transitional state of liminality created meaning and reconstructed a sense of self through a process of reflecting on their existence. It involved negative feelings of instability and ambiguity, and positive experiences of personal growth and deepened spirituality which assisted in managing their emotions and realising a renewed sense of self and purpose, expressed as empathy and altruism. Not all experienced liminality as an aspect of adaptation.
- Using oxygen therapy at the advanced stage of LAM held paradoxical meanings for the women. Meaning shifted with shifts in their perception at turning points.
 - Negative meaning of disruption, associated with themes of loss and limitation, burden, and feeling stigmatised.
 - Positive meaning of enabling, associated with staying alive, preserving normality, and feeling liberated.
- The participants developed resilience through their experiences of living with LAM over time.
 - Resilience was the complex learning that developed throughout life as a result of adaptation to situations of change and adversity, and as a process of everyday life.
 - Resilience learning involved making meaning of change, and gaining knowledge and competence in managing their illness over time.
 - Everyday life experiences built resilience through managing minor daily disruptions and experiencing positive emotions, for example in experiences of social connectedness.
 - Resilience was a dynamic process which shifted with changes in the women's individual health and social world, and reflected personal beliefs and perceptions. The women experienced different levels of resilience at different stages of their illness according to their individual situations of change and perception of need.
 - The outcome of resilience learning was the ability to experience wellness and positive beliefs of self-efficacy, self-esteem and self-worth, and later to cope with struggle and suffering at late stage.
- Avoidance coping enabled some to adjust to change while their illness remained stable. It was associated with not engaging in the learning processes of adaptation and, at late stage, a lower level of resilience and perception of suffering.

- The late stage of LAM was characterised by living with struggle and suffering. Resilience was a resource which influenced the women’s ability to cope with their worsening symptoms and shaped perceptions of their suffering. It was an ongoing process through experiences of spirituality, personal growth, and compassion.

7.12.2 How the rarity of LAM affected the experiences of women living with LAM

- The process of diagnosis was associated with periods of misdiagnosis, delayed diagnosis, and multiple medical consultations due to the lack of knowledge of LAM amongst physicians.
- Many participants received their diagnosis in an insensitive manner:
 - Lack of empathy and emotional support
 - Minimal outdated and inaccurate information sourced from unreliable internet sources was provided
 - Lack of referral to the peer support organisation.
- The rarity of LAM created feelings of uncertainty and isolation.
- Women isolated from centres of LAM expertise had difficulty accessing appropriate expert care, information and treatment, and contact with other women with LAM. There was a lack of communication between general physicians, LAM experts, and other healthcare professionals.
- There was a need for women to be self-reliant in seeking information, and self-advocate to access appropriate care, treatment, investigations and follow-up of investigations.
- The participants’ interactions with physicians were characterised by a shifting power balance influenced by knowledge and attitudes:
 - Women became expert in knowledge of their rare disease and managing their illness.
 - Physicians with less knowledge of LAM tended to be less accepting of their patient’s expertise and less willing to share decision-making.
 - Physicians expert in LAM were more likely to be collaborative and share decision-making with the participants.
- In the hospital setting, women felt a need to be vigilant and educate healthcare professionals. Their self-advocacy was tempered by their increased vulnerability in this setting and their perception of needing to be a “good patient”.

- There was a lack of knowledge and awareness of LAM in the community which created a constant need to explain the illness to others and, at times, feelings of being stigmatised in public places and workplaces.
- The rarity of LAM meant it was difficult to attract funds for research. There were few women available to run the peer support organisation, support other women with LAM, and advocate in the community through fundraising and public speaking. These activities took time and energy and were felt to be a burden. Raising funds through friendship networks while looking healthy created feelings of guilt.

7.12.3 Issues of concern for women living with LAM

- The limited knowledge and awareness of LAM among healthcare professionals and in the community.
- Difficulties receiving a timely diagnosis and accurate, appropriate information and emotional support.
- The limited number of physicians with expertise in LAM in Australia and poor communication between physicians created difficulties in accessing expert care, and appropriate information, investigations and treatment for women isolated geographically or by their health status.
- The need to be vigilant in monitoring their care and educate healthcare professionals in the hospital setting was a burden.
- Fatigue, a problem for some women, was not addressed by healthcare professionals.
- Lack of ongoing emotional support and advice regarding social services.
- Sexuality and contraception were significant issues for pre-menopausal women. The women felt sexuality was a difficult subject to raise with physicians. They received inadequate and inconsistent advice concerning contraception.
- Inequities in funding for oxygen therapy across different states
- Lack of funding for portable oxygen concentrators which would facilitate engagement in exercise, work and social activities.
- Non-availability of liquid oxygen
- Need for further education on managing supplementary oxygen therapy.
- Need for educational resources suited to younger women and for children whose mothers are living with LAM and using oxygen therapy.
- Some expressed a desire for a health coaching style of support after diagnosis.

- Lack of public funding for research into LAM created a need to fundraise while managing their illness.
- Few women were available to run peer support organisation, fundraise, and support other women living with LAM – need for community support with these activities.

7.13 Conclusion

This chapter has discussed the findings of the study in relation to resilience which emerged as the overarching theme of the women's experiences of living with LAM across their life course. This research extends previous concepts of resilience by adopting a broad view of resilience as learning. The participants revealed that this enabled them to manage their physical, mental, emotional and spiritual health while living with LAM in their individual changing social contexts throughout the course of their illness. The women emphasised their emotional and social concerns and the importance of achieving wellness by gaining autonomy and a sense of coherence. The concept of resilience as lifelong learning identifies the mechanisms by which the participants reached these outcomes as the interplay of meaning, agency, knowledge, competence, and positive emotions, in times of change and adversity as well as times of stability. Finding meaning was a particularly significant aspect of their resilience learning.

This research has demonstrated that resilience is not an inbuilt quality but is accessible to anyone and can develop at any stage at a level suited to a person's individual situation and their perception of their needs. As such, resilience learning is a constructive, strength-based concept which healthcare professionals can utilise in planning interventions to improve people's ability to manage and live well with chronic illness in the context of their life. The following chapter concludes the thesis. It will discuss the significance of the findings for practice in more detail, reflect on the life history method used in the study, consider limitations and directions for future research, and offer a personal reflection on the research journey.

Chapter 8

CONCLUSION

8.1 Introduction

This chapter concludes the thesis. Specific findings of this research, including issues of concern to the participants, are discussed in relation to their implications for practice. The limitations of the study are acknowledged and possibilities for future research are suggested. I reflect on the life history method that has been used and offer a personal reflection on the research journey.

Consistent with its aims, this study has explored and captured a comprehensive understanding of the experiences of women living with LAM over time, from the onset of symptoms to the late stage of their illness, and how these were affected by the rarity of LAM. Diagnosis, a long hospital admission, starting oxygen therapy, receiving a transplant, and experiencing respiratory failure were significant turning points of disruptive change for the participants.

This study illuminated resilience as an important aspect of the participants' experiences of living with LAM across their life course. Resilience was revealed as the complex learning that developed throughout life as a result of both adaptation to situations of change and adversity, and as a process of everyday life. Resilience learning involved making meaning of change, and gaining knowledge and competence in managing their illness over time. The outcome of these processes was the ability to move from an experience of disruption to one of wellness, and later to cope with struggle and suffering if their health deteriorated to a late stage. In everyday life, resilience accumulated as positive beliefs of self-efficacy, self-esteem and self-worth. These were generated as they experienced both the fluctuations and positive aspects of daily life. Resilience was a dynamic process which shifted with changes in the women's individual health and social world, and reflected their personal beliefs and perceptions. While dispositional qualities of optimism, curiosity, flexibility and resourcefulness facilitated the participants' resilience, learning processes played a more significant role.

The rarity of LAM had paradoxical influences on the participants' experiences. Negatively, it created uncertainty and feelings of being stigmatised and isolated. Accessing appropriate medical care, information, and treatment was a problem for women if they were unable to travel to specialist centres for management of their LAM. Additionally, needing to educate healthcare professionals and advocate for services and funds for research was perceived to be a burden. Positively, having a rare disease increased the women's knowledge and communication skills as a consequence of their self-reliance and self-advocacy. While this empowered them to engage in collaborative relationships with researchers and their care providers, shared decision-making regarding their medical care was dependent on the attitude of their physician. When decision-making was shared, a sense of control and feelings of autonomy and competence were enhanced. In turn, their satisfaction with their medical management improved.

8.2 Implications for practice

The participants of this study revealed a number of issues related to their experiences of living with LAM that have implications for practice and public policy. These may also be relevant to people living with other chronic conditions.

In common with other rare diseases, the period from onset of symptoms to diagnosis was a time of delay, misdiagnosis and uncertainty due to physicians' lack of awareness and knowledge of LAM. There remains a need to raise awareness among physicians of the possibility of a woman having LAM when presenting with unexplained breathlessness and reduced exercise tolerance, rather than attributing symptoms to other causes. A woman's experience of being diagnosed improved if she was provided with accurate information and explanation by an expert physician.

The issue of unequal power balances in patient/physician relationships is well recognised in the literature. This study has demonstrated the power differential was reduced when both patients and physicians increased their knowledge about LAM, and when physicians listened to and acknowledged the patient's understanding of their illness. This tended to be the case with LAM experts who were generally more willing to engage in collaborative interactions with their patients and accept their input in decision-making. The findings of this study reinforce the need for all healthcare professionals to be constantly mindful of power balances in their interactions with patients.

Nurses have a role to play in addressing the concerns of women living with LAM and helping improve their healthcare experiences. Nurses may encounter these women or other people with rare diseases in community and hospital inpatient and outpatient settings as they are treated for their condition or undergo investigations. Within a multidisciplinary team, a nurse specialist can potentially undertake a leadership role coordinating care for these patients. There are at present no multidisciplinary centres of expertise in rare disease in Australia (Molster et al. 2016). While a specialist nursing role may potentially be funded if such a centre was established, in the context of the difficulty in attracting government funding for research into rare diseases, it is unlikely that a specialist nursing position would be publicly funded in the current Australian healthcare system. Rather, it is feasible that a nursing role supporting women with LAM and people with other rare lung diseases could be encompassed within the scope of practice of respiratory nurse consultants or nurse practitioners practising throughout Australia. Specialist nurses would therefore be available to patients with rare diseases and the healthcare professionals caring for them at a more local level.

A specialist nursing role would involve acting as a conduit to physicians, nurses and other service providers. The nurse could advocate to facilitate appropriate investigations, treatment, oxygen supplies and equipment, as well as affordable home help. In addition, the nurse could provide specific education on LAM to nurses who may be caring for these women in hospital or community settings or direct them to appropriate educational resources. In this way, a specialist nurse can raise awareness of LAM and other rare diseases among nurses and other healthcare professionals who may have infrequent contact with these patients. The specialist nurse can assist them in developing skills to access accurate literature on both the biomedical facts as well as people's lived experiences of rare conditions to increase their knowledge of issues of concern for these people and inform their practice when needed.

Women can be isolated from healthcare centres proficient in managing LAM by distance or their poor health status. They are consequently disadvantaged in their ability to access appropriate care and treatment. This includes sirolimus medication which the participants in this study reported as significantly enhancing their quality of life. Establishing pathways of consultation and communication using technology, such as phone, email, internet, and social media, may improve dialogue between women, their general physicians, nurses, and LAM experts. This may enable women who are isolated to access expert medical and

nursing management without relocating and travelling long distances. In addition, a specialist nurse could incorporate technology to provide educational resources for healthcare professionals in smaller and remote health facilities if they are caring for women with LAM.

Turning points were periods when the participants needed most support. From the time of diagnosis, an educated nurse could provide information, education, resources, and emotional support to women, their partners, and families. The nurse may provide an opportunity for open discussion, as well as advice and support, regarding specific areas of concern such as fatigue, emotional and sexual health, contraception, and managing the impact of illness on relationships, friends, and family and work responsibilities. When necessary, referrals could be made to other members of the healthcare team, for example, psychologists, dieticians and social workers. The nurse may also respond to women seeking advice on more minor matters not warranting a medical appointment but nonetheless of concern to them. Additionally, the nurse could connect women with peer support groups if desired, being sensitive to connecting them with women who are at a similar stage of LAM and managing their illness well.

The understanding of the learning processes of adaptation and resilience provided by this study can guide an informed nurse in supporting women with LAM to independently manage their illness. In the context of LAM as a chronic illness, resilience incorporated a cognitive and practical aspect of learning the skills necessary to manage the illness, and an emotional and spiritual component of making meaning to re-establish a sense of self and relatedness to the world in the context of living with illness. Resilience was enhanced by care focused on health, wellness, autonomy in illness management, and participation in shared decision-making. Active listening enables a patient to voice their concerns and reveal their personal life meaning, goals, priorities, past experiences, and social context. Knowing these, the nurse can provide individualised care and assist a woman to identify her own strengths and coping style, and reframe her goals to reflect her new reality. An understanding of resilience can assist healthcare professionals in encouraging their patients, particularly those with an avoidance style of coping, to engage in the learning processes which build resilience.

Managing their emotions was an important aspect of the participants' resilience. This study uncovered self-learning as a mechanism of emotional stability and showed how it occurred as the women found meaning in their experiences and developed positive self-beliefs. The

nurse can utilise this understanding to support patients in managing their emotional health and social interactions throughout their illness. For example, the nurse can be aware of and identify when a woman is experiencing liminality, such as through her use of metaphor, and provide additional emotional support and reassurance of the positive benefits of this self-learning for integrating illness into life. Many women try alternative therapies and the nurse needs to be open to discussing these options.

The concept of resilience learning revealed by this study may additionally inform specialist nurses and other healthcare professionals involved in health coaching for people living with chronic illness and disability in a variety of healthcare settings. According to Palmer et al. (2003), health coaching facilitates learning in health education and health promotion to enable a person to achieve their health-related goals and wellbeing. It includes emotional support and assistance in developing self-management skills (Bennett et al. 2010). This study provides an understanding of the factors that influenced the participants' motivation, as well as the processes involved in learning to self-manage their health and achieve wellness. This may be relevant to the motivational interviewing and goal-setting that Olsen and Nesbitt (2010) claimed are involved in health coaching.

An informed nurse can provide education to women living with oxygen therapy, particularly those using oxygen intermittently who demonstrated misunderstandings regarding their oxygen use. While attending a pulmonary rehabilitation program in a hospital clinic is an option that can be offered to women with LAM, these are attended mainly by older people with COPD. The participants indicated they preferred exercising at home, outdoors or with people their own age at a gym. The nurse can teach and assess women's skills in self-monitoring their breathing and oxygen saturations to enable them to exercise in these environments. This encourages personal autonomy and self-management, and enhances resilience. In addition, educational material currently available on living with chronic respiratory disease is directed to people with COPD and there is a need both for resources suitable for younger women, and to assist mothers living with oxygen therapy in educating their children. Within the acute care setting, the contrasting experiences of Patricia and Vidu explain why some patients find it so difficult to relinquish their oxygen after transplant and emphasises the need for nurses to show patience and empathy in educating these patients. An expert nurse who has been caring for women prior to transplant can liaise with hospital nurses to inform them of a patient's previous coping style so that any difficulties can be anticipated and managed.

This study has revealed the need to address policy issues concerning oxygen therapy in the Australian context. These include inequities in funding for oxygen in different states, non-availability of liquid oxygen, and lack of funding for portable concentrators which facilitate exercise and activity. Most policy is based on research regarding older people with COPD and does not take account of the needs of the younger LAM population who in this study showed they were highly motivated to engage in exercise, work and social activity. Furthermore, assessment criteria determining oxygen supplies needs to take into account the function and activity levels of younger women. Factors of inequity need to be addressed so as not to compound the hardship which single women living alone, who are particularly vulnerable, can face in the advanced stages of their illness. A nurse coordinating care could maintain contact with these women and assess their health and need for additional services and support.

The findings of this study have uncovered a role for an expert nurse in providing collaborative, person-centred care to support women living with LAM to manage their illness effectively throughout their life. In addition, using technology, a specialist nurse can inform and educate nurses caring for women with LAM in other locations. Further to this, the understanding of resilience illuminated by this research may assist nurses in facilitating a patient's engagement in the learning processes which build resilience. These findings may be applicable also to people living with other rare and chronic conditions.

8.2.1 Summary of recommendations for practice

It is proposed that a specialist nursing role in rare lung disease is developed within the scope of practice of a respiratory nurse consultant or nurse practitioner. It is recommended that a specialist nurse:

- Co-ordinate care of women with LAM or other rare lung diseases within a multidisciplinary team, referring women to multidisciplinary healthcare services when needed.
- Establish pathways of communication using technology to improve dialogue between women living with LAM, general physicians, nurses and LAM experts.
- Provide education on LAM to nurses and healthcare professionals. Assist them in developing skills to access accurate literature on rare disease that can inform their professional practice. Incorporate technology to provide educational resources for healthcare professionals in smaller and remote health facilities.

- Provide appropriate information, education, resources and emotional support to women with LAM, their partners and families at diagnosis, later turning points and as needed throughout their illness.
 - Provide education to women living with oxygen therapy.
 - Develop educational resources suitable for women living with LAM, including mothers living with oxygen therapy in educating their children.
- Develop an understanding of the learning processes of adaptation and resilience to support women with LAM to independently manage their illness and achieve their health-related goals and wellbeing.
- Connect women diagnosed with LAM with peer support groups if desired.
- Liaise with hospital nurses when patients are admitted to hospital to inform them of a patient's coping style so that this can be incorporated into individual care plans.
- Maintain contact with women at the late stage of LAM and assess their health and need for additional services and support.
- Advocate policy makers to address issues of:
 - Inequities in funding for oxygen in different states of Australia
 - Non-availability of liquid oxygen
 - Lack of funding for portable oxygen concentrators

8.3 Reflection on the method

The life history method used in this study has been effective in achieving its aim of exploring women's experiences of living with LAM across the life course. The strength of this method was its holistic character and ability to capture the complexity of multiple aspects of illness experience, including the biological, physical effect of LAM; personal meaning and social context; and the temporal dimension of the participants' experiences. It revealed the dynamic interaction between these aspects of human experience in response to the day to day changes of everyday life, as well as significant life changes related to illness progression or social context.

Constructing the collective life history of the participants around turning points and significant aspects of their illness revealed how the meaning of their experiences changed over time, as well as issues of concern and their changing support needs. It also allowed the participants' experiences to be situated in historical time to reveal the influence of events

on the women's experiences, including the establishment of the LAM Foundation, and the discovery of the pathogenesis of LAM and subsequent effectiveness of sirolimus as a treatment for LAM. As well as common themes, life history demonstrated individual differences and complexities and paradoxes in the women's experiences and how they were influenced by the rarity of LAM. The life course and contextual character of life history provided new insights into resilience in the context of chronic illness and demonstrated its multidimensional, transactional and developmental qualities.

Accessing the participants' medical records and constructing the biographical accounts of their illness provided, as Rosenthal (1993) intended, a database and factual details with which to compare the women's subjective narratives. It revealed meaning in the different emphases the participants' placed on aspects of their experience compared to their physicians, and the absence of areas of significance, such as fatigue and sexuality, in their medical records. This lends support for qualitative research of this nature to inform healthcare professionals so that management is focused and meets patients' needs.

Life history is intensely personal. Taking an empathic stance and allowing the women's stories to permeate me meant there was a need to be reflexive and manage my own responses to the participants' situations. As a nurse researcher, at times it was difficult separating myself as nurse from myself as researcher. For example, witnessing Margaret's vulnerability and hardship at the late stage of LAM elicited a response of wanting to care for her and improve her situation. I gave her the contact details of the social worker who had agreed to be available while interviews were being conducted so that she might explore options for additional services and funding, but, as a researcher, I was not entitled to provide her with nursing care. I dealt with these conflicting feelings by reflecting on them, writing about them, and discussing them with my research supervisors.

This life history study was hermeneutic as it was deeply infused with meaning, making meaning, and shifts in meaning as the participants' experienced their illness in changing circumstances over time, and interpreted their lives and experiences as they narrated their life stories. Reflecting on their lives allowed some, in turn, to reach deeper levels of meaning in their experiences, a process which they indicated was therapeutic in giving them a fresh perspective on their illness. It was, in Gadamer's (2004) words, a "fusion of horizons". The collective life history within this thesis represents an additional level of meaning as my perspectives and interpretations were fused with those of the participants. The reader of this research will further enrich the meaning with their own perspectives.

8.4 Limitations and directions for future research

This study concerned the experiences of women living with LAM in the Australian context. The nineteen participants recruited to the study represent 18% of women known to be currently living with LAM in Australia. However, the numbers are small and only three participants experienced the late stage of the illness. While this study has provided valuable insights into the experience of living with LAM, the findings cannot be generalised to other contexts.

Further research is required to explore the experiences of women living with LAM in other contexts and at the advanced and late stages of the illness. The participants identified that their partners, children and parents were emotionally and physically affected by their diagnosis. No studies have sought the perspectives of family members and this remains an area to be explored. Future research, eliciting the views of both patients and healthcare professionals, could examine the applicability and usefulness of technology in providing access to expert medical and nursing management for women with LAM who are geographically isolated.

Resilience emerged as a finding of this study and could form a focus of research into people's experiences of other chronic illnesses. Additionally, resilience learning, as conceptualised in this study, could be examined for its applicability to both health coaching interventions and training for nurses and other healthcare professionals conducting health coaching.

8.5 A Personal reflection

Characteristic of life history research, this study has been a complex and winding research journey. There were no simple formulas to follow and few examples of life history studies in chronic illness research. Yet it seemed an approach that would suit an exploration of human experience that extended over the course of a person's life. The rarity of LAM made the process more challenging with the difficulties in recruitment, ethical issues, and logistical factors of time, distance and expense as discussed earlier.

Despite the turns in the road, conducting this life history research has been rewarding and deeply satisfying. It was a privilege to meet each participant and share her life story. I have always considered listening and talking to patients to be the best aspect of my nursing role. As a researcher, to be able to do so uninterrupted and completely focused on an individual woman was a rare and special experience. My intention was to provide a deeper

understanding of the women's experiences, to represent each woman's unique story truthfully while revealing the common threads of their narratives in a way that would inform healthcare professionals who may care for them so they may be better supported throughout their illness. Life history has been a pathway towards achieving that goal.

This research has also been a deeply personal experience for me. It is not possible to enter people's lives and become immersed in their stories without being touched. Analysing the women's narratives and writing my findings was itself a liminal experience. It involved separating myself from familiar life and becoming, in Turner's (1967, p.95) words, a "liminal persona". There is the physical act of taking oneself to a quiet place on one's own to think and write. It is a journey that no one else can take and this makes it a lonely time of isolation. Much of normal life is put aside, on hold. The liminality of being in the women's lives was an unsettling and at times emotional experience and I needed to draw on my own personal resources and cultivate my own resilience. It has been a time of professional and personal learning – gaining understanding, knowledge and research skills, finding meaning, and experiencing personal growth through exploring the women's experiences. I have gratitude that I have been able to take this journey and give the women's voices a place in the literature on LAM and chronic illness.

8.6 Concluding remarks

This study explored the experience of women living with LAM across the life course. The women's experiences were complex and varied, reflecting the differing manifestations, stages and progress of their illness, and their personal life meaning and individual social contexts. While each woman's experience was unique, the majority of participants positively adapted to their illness over time and developed resilience in the process. Exploring the women's experiences in the context of their whole lives revealed that resilience was the learning that evolved over the course of their lives through both adaptation to situations of change and adversity, and in their everyday experiences. The participants demonstrated that their learning was associated with acquiring knowledge and competency in managing their illness and health, finding meaning in their experiences, and personal growth. It was influenced by their social interactions, changes in their social environment, and the rarity of their illness. These learning processes generated positive self-beliefs and positive emotions which enabled the women to experience wellness while living with LAM, and accumulated as a resource they drew on to cope with struggle and suffering in the late stages of their illness.

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APPENDIX 1

Ethics Documents

Appendix 1.1

Ethics Approval for Honours Thesis



St Vincent's Hospital

16 October 2008

Denise Haylen
Cardiothoracic Unit
St Vincent's Hospital
Darlinghurst NSW 2010

A facility of
St. Vincents & Mater Health Sydney

St. Vincent's Hospital Sydney Ltd
ABN 77 054 038 872
390 Victoria Street
Darlinghurst NSW 2010 Australia

T +61 2 8382 1111
F +61 2 9332 4142
www.stvincents.com.au

Dear Denise

SVH File Number: 08/163

Title: Living with lymphangioliomyomatosis – A life history.

Thank you for submitting the above project for review. Based on the information you have provided and in accordance with the following NHMRC guidelines; National Statement 2007 – Section 5 Institutional Responsibilities and “When does quality assurance in health care require independent ethical review?” (2003), this project has been assessed as **low risk** and is therefore exempt from full HREC review.

I am pleased to advise that the on **16 October 2008** the HREC Executive on behalf of the Executive Director granted authorisation for the above project to commence at **St Vincent's Hospital**.

The documents approved for this project are:

Participant Information sheet and Consent Form version2: 15 October 2008
Research proposal (no version/date)

Please note the following conditions of approval:

1. This approval is valid for five years, and the Committee requires that you furnish it with annual reports on the projects progress beginning in **October 2009**. Please notify the HREC Executive in writing when this project is completed.
2. The Investigator will immediately report anything which might warrant review of ethical approval of the project in the specified format, including unforeseen events that might affect continued ethical acceptability of the project and any complaints made by participants regarding the conduct of the project.
3. Proposed changes to the research protocol, conduct of the research, or length of approval will be provided to the HREC Executive for review, in the specified format.
4. The HREC Executive will be notified, giving reasons, if the project is discontinued before the expected date of completion.

Please note that for multi-site projects authorisation needs to be obtained from each participating institution.



The University of Sydney

NSW 2006 Australia

Human Research Ethics Committee

www.usyd.edu.au/ethics/human

Manager: Gail Briody

Telephone: (02) 9351 4811

Fax: (02) 9351 6706

Email: gbriody@usyd.edu.au

Deputy Manager: Marietta Coutinho

Telephone: (02) 9036 7566

Email: mcoutinho@usyd.edu.au

Human Secretariat

Telephone: (02) 9036 9308

(02) 9036 9309

(02) 9036 7274

Facsimile: (02) 9036 9310

Mailing Address:

Room 313, Level 3, Old Teachers College – A22

4 November 2008

Dr. Murray Fisher
Faculty of Nursing and Midwifery – M02
88 Mallett Street
The University of Sydney.
m.fisher@usyd.edu.au

Dear Dr. Fisher,

Title: *Living with Lymphangiomyomatosis - A Life History Project*
(Ref. No. 11449)

Honours Student: *Denise Haylen*

Your application was reviewed by the Executive Committee of the Human Research Ethics Committee (HREC), and in doing so has ratified your study to include the Honours student, Ms. Denise Haylen.

The Executive Committee acknowledges your right to proceed under the authority of *St. Vincent's Hospital Human Research Ethics Committee*.

Please note, this ratification has been given only in respect of the ethical content of the study.

Any modifications to the study must be approved by *St. Vincent's Hospital Human Research Ethics Committee* before submission to the University of Sydney Human Research Ethics Committee.

Yours sincerely

Gail Briody
Manager
Ethics Administration

Appendix 1.2

Approval for amendment to Research Protocol and Participant Information and

Consent Form to convert from Honours thesis to PhD thesis



St Vincent's Hospital

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www.stvincents.com.au

12 May 2010

Denise Haylen
Cardiothoracic Unit
St Vincent's Hospital
390 Victoria St
Darlinghurst NSW 2010

COPY

Dear Denise

SVH File Number: 08/163

Title: Living with lymphangioliomyomatosis – A life history.

Thank you for your letter submitting a request for an amendment dated 6 April 2010 to the above project. This was considered by the St Vincent's Hospital HREC at its Executive meeting held on 5 May 2010. This HREC is constituted and operates in accordance with the National Health and Medical Research Council's National Statement on Ethical Conduct in Human Research (National Statement) and the CPMP/ICH Note for Guidance on Good Clinical Practice. No HREC members with a conflict of interest were present for review of this amendment.

I am pleased to advise that the documents reviewed and approved at the meeting were:

- Protocol Version 2: dated 17 March 2010
- Participant Information Sheet and Consent Form Version 4: dated 17 March 2010

For multi-site projects reviewed by the HREC after 1 July 2007 a copy of this letter must be forwarded to all Principal Investigators at every other site conducting the project for submission along with a copy of the approved documents to the relevant Research Governance Officer.

Should you have any queries about your project please contact the Research Office, Tel: 8382-2075, email research@stvincents.com.au. The HREC Terms of Reference, Standard Operating Procedures, *National Statement on Ethical Conduct in Human Research (2007)* and the *CPMP/ICH Note for Guidance on Good Clinical Practice* and standard forms are available via the research/education link on the St Vincent's Hospital website: internal address: <http://exwwwsvh.stvincents.com.au> external address: <http://wwwsvh.stvincents.com.au>

Yours sincerely

Sarah Charlton
HREC Executive Officer
Research Office

Appendix 1.3

Participant Information and Consent Form

Revocation of Consent Form



PARTICIPANT INFORMATION SHEET AND CONSENT FORM

Living with Lymphangiomyomatosis – A Life History Project

Invitation

You are invited to participate in a research study into the experience of living with lymphangiomyomatosis (LAM) over the life course from onset of symptoms to the present.

The study is being conducted by:

- Denise Haylen, Registered Nurse, Cardiothoracic Unit at St Vincent's Hospital; Doctor of Philosophy (Nursing) candidate, The University of Sydney.
- Dr Murray Fisher, Sydney Nursing School, The University of Sydney.
- Dr Jo Patching, Sydney Nursing School, The University of Sydney.

Before you decide whether or not you wish to participate in this study, it is important for you to understand why the research is being done and what it will involve. Please take the time to read the following information carefully and discuss it with others if you wish.

1. 'What is the purpose of this study?'

The purpose is to discover what has been your experience of living with lymphangiomyomatosis as a rare lung disease. This project is being undertaken because to date no studies have examined in depth the personal perspectives of women living with LAM and the impact of this disease on their life. This research project seeks the views of a range of women living with LAM. It aims to increase knowledge of the experience of living with LAM, improve understanding of how the rarity of this disease affects the illness experience, identify needs of women with LAM and improve support for these women.

2. 'Why have I been invited to participate in this study?'

You are eligible to participate in this study because you are over 18 years of age, have been diagnosed with LAM and have a broad range of experience of the disease.

3. 'What if I don't want to take part in this study, or if I want to withdraw later?'

Participation in this study is voluntary. It is completely up to you whether or not you participate. If you decide not to participate, it will not affect the treatment you receive now or in the future. Whatever your decision, it will not affect your relationship with the staff caring for you.

If you wish to withdraw from the study once it has started, you can do so at any time without having to give a reason.

4. 'What does this study involve?'

If you agree to participate in this study, you will be asked to sign the Participant Consent Form.

This study will be conducted over 3 years.

If you agree to participate in this study, you will then be asked to take part in 2 or 3 interviews. Each interview will require you to spend around one to two hours speaking with Denise Haylen. A third interview will be conducted to clarify or further explore issues raised in the previous interviews if required. The interviews will take place at a time and location that is suitable to you. With your consent, each interview will be audiotaped and transcribed.

In addition, the researchers would like to have access to your medical record in order to correlate your medical history with your life story.

5. 'How is this study being paid for?'

This project is being supported by the Heart/Lung Program at St Vincent's Hospital. No funding is being provided by other means.

6. 'Are there risks to me in taking part in this study?'

The telling of personal life stories can have a beneficial effect for participants. However, it is possible that you may become upset when telling your story. During the interview, should you become distressed, the interviewer will pause or stop the interview. You would be given time before being offered the opportunity to continue after a period, continue on another occasion or withdraw from the study. You may terminate the interview and/or withdraw from the study at any time. The interviews are being conducted by the Researcher who works in the area of cardiothoracic nursing and is well versed with the possible range of responses relating to this condition. Professional counselling and support will also be available at no cost should you require it.

7. 'Will I benefit from the study?'

This study aims to further nursing and medical knowledge and may improve future support for women with LAM, however it may not directly benefit you.

8. 'Will taking part in this study cost me anything, and will I be paid?'

This study has no financial support and no funds are available to cover transport costs of the participant to attend the interviews. If you should wish to participate in the study the Researcher is willing to conduct the interviews at a location convenient to you. There will be no additional costs to you other than your time.

9. 'How will my confidentiality be protected?'

Of the people treating you, only Professor Allan Glanville who is supervising Denise Haylen at St Vincent's Hospital, will know whether or not you are participating in this study. Any identifiable information that is collected about you in connection with this study will remain confidential and will be disclosed only with your permission, or except as required by law. Audiotapes and transcripts of interviews will not contain any information that will allow you to be identified. A pseudonym (made up name) will be used in your records of interviews. Only the researchers named above will have access to your details and results. These and audiotapes and transcripts will be held securely at the University of Sydney in a locked filing cabinet in Dr Murray Fisher's office.

10. 'What happens with the results?'

If you give us your permission by signing the consent document, we plan to discuss/publish the results in peer-reviewed journals and in presentations at conferences or other professional forums. Completion of this research study will form part of the requirements for a Doctor of Philosophy Degree for Denise Haylen. Results may also be presented to the HREC of St Vincent's Hospital and the University of Sydney for monitoring purposes.

In any publication, information will be provided in such a way that you cannot be identified. Results of the study will be provided to you, if you wish.

11. 'What should I do if I want to discuss this study further before I decide?'

When you have read this information, Denise Haylen will discuss it with you and any queries you may have. If you would like to know more at any stage, please do not hesitate to contact her on:

Phone: 9351 0587 via Dr Murray Fisher at The University of Sydney

Email: dhaylen@stvincents.com.au

Please feel free also to contact:

Dr. Murray Fisher
Sydney Nursing School
University of Sydney, NSW 2006
Phone: 9351 0587
email: mfisher@nursing.usyd.edu.au

12. 'Who should I contact if I have concerns about the conduct of this study?'

This study has been approved by St Vincent's Hospital HREC. Any person with concerns or complaints about the conduct of this study should contact the Research Office who is nominated to receive complaints from research participants. You should contact them on 02 8382 2075 and quote 08/163.

The conduct of this study has also been authorised by the University of Sydney HREC. Any person with concerns or complaints about the conduct of this study may also contact the Senior Ethics Officer, Ethics Administration, University of Sydney on (02) 9351 4811 (Telephone); (02) 9351 6706 (Facsimile) or gbriody@usyd.edu.au (Email) and quote reference number 11449

**Thank you for taking the time to consider this study.
If you wish to take part in it, please sign the attached consent form.
This information sheet is for you to keep.**



CONSENT FORM
Living with Lymphangioliomyomatosis – A Life History Project

1. I,.....
of.....
agree to participate as a subject in the study described in the participant information statement set out above.
2. I acknowledge that I have read the participant information statement, which explains why I have been selected, the aims of the study and the nature and the possible risks of the investigation, and the statement has been explained to me to my satisfaction.
3. Before signing this consent form, I have been given the opportunity of asking any questions relating to any possible physical and mental harm I might suffer as a result of my participation and I have received satisfactory answers.
4. I consent to my medical record being used as a reference source for clarification of facts in my life story.
5. I understand that I can withdraw from the study at any time without prejudice to my relationship to the University of Sydney or St Vincent's Hospital.
6. I agree that research data gathered from the results of the study may be published, provided that I cannot be identified.
7. I understand that if I have any questions relating to my participation in this research, I may contact Denise Haylen or Dr Murray Fisher on telephone 9351 0587, who will be happy to answer them.
8. I acknowledge receipt of a copy of this Consent Form and the Participant Information Statement.

Complaints may be directed to the Research Governance Officer, St Vincent's Hospital, who may be contacted on 02 8382 2075. Complaints may also be directed to the Senior Ethics Officer, University of Sydney, on 02 9351 4811 (telephone), 02 9351 6706 (facsimile) or gbriody@usyd.edu.au (email).

Signature of subject Please PRINT name Date

Signature of witness Please PRINT name Date

Signature of investigator Please PRINT name Date



The University of Sydney



St Vincent's Hospital

Living with Lymphangiomyomatosis – A Life History Project

REVOCAION OF CONSENT

I hereby wish to **WITHDRAW** my consent to participate in the study described above and understand that such withdrawal **WILL NOT** jeopardise any treatment or my relationship with the University of Sydney, St Vincent's Hospital or my medical attendants.

Signature

Date

Please PRINT Name

The section for Revocation of Consent should be forwarded to

Dr. Murray Fisher
Sydney Nursing School,
University of Sydney, NSW 2006
Phone: 9351 0587
email: mfisher@nursing.usyd.edu.au

Appendix 1.4

Interview Schedule

Living with Lymphangiomyomatosis – A Life History Project

Interview Schedule [Version 2 - 17 March 2010]

The interviews will be conversation/narrative style (tell me about.....) and will focus on the following areas:

Interview 1

- How life was leading up to diagnosis
- The process of diagnosis
- How life has changed with diagnosis.

Interview 2

- How life is now
- What the issues are for you living with a rare disease
- Further exploration of issues raised in Interview 1 or this interview.

Appendix 1.5

Approval for amendment to Research Protocol and Letter of Invitation
following complaint by physician to HREC



St Vincent's Hospital

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9 December 2010

Prof Alan Glanville
Thoracic Medicine
Xavier Level 4
St Vincent's Hospital
390 Victoria St
Darlinghurst NSW 2010

Dear Alan

COPY

SVH File Number: 08/163

Title: Living with lymphangioliomyomatosis – A life history.

Thank you for your letters dated 22 November 2010 and 24 November 2010 responding to the HREC request for an amendment to the above project regarding a complaint received on the 15 November 2010. The HREC Executive Committee wishes to thank you for your quick response to this matter.

The amendment was considered by the St Vincent's Hospital HREC at its Executive meeting held on 1 December 2010. I am pleased to advise that the documents reviewed and approved at the meeting were:

- Protocol Version 3: dated 23 November 2010
- Recruitment letter version 24 November 2010

This HREC is constituted and operates in accordance with the National Health and Medical Research Council's National Statement on Ethical Conduct in Human Research (National Statement) and the CPMP/ICH Note for Guidance on Good Clinical Practice. No HREC members with a conflict of interest were present for review of this amendment.

Should you have any queries regarding the above please contact the HREC Executive Officer on 8382 2075. The HREC Terms of Reference, Standard Operating Procedures, *National Statement on Ethical Conduct in Human Research (2007)* and the *CPMP/ICH Note for Guidance on Good Clinical Practice* and standard forms are available on the Research Office website: www.stvincents.com.au/researchoffice or internal at <http://exwwwsvh.stvincents.com.au/researchoffice>

Yours sincerely,

Sarah Charlton
HREC Executive Officer
Research Office
D/2010/17306

CC: Denise Haylen
Dr Murray Fisher

Continuing the Mission of the
Sisters of Charity

Appendix 1.6

Approval for amendment to Research Protocol to allow recruitment through the

LAM Australia Research Alliance



St Vincent's Hospital

8 March 2011

Denise Haylen
Cardiothoracic Unit
St Vincent's Hospital
390 Victoria St
Darlinghurst NSW 2010

A facility of
St. Vincents & Mater Health Sydney

St. Vincent's Hospital Sydney Ltd
ABN 77 054 038 872
390 Victoria Street
Darlinghurst NSW 2010 Australia

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www.stvincents.com.au

COPY

Dear Denise

SVH File Number: 08/163

Title: Living with lymphangioleiomyomatosis – A life history.

Thank you for your letter dated 22 February 2011 submitting a request for an amendment to the above project. This was considered by the St Vincent's Hospital HREC at its Executive meeting held on 3 March 2011. This HREC is constituted and operates in accordance with the National Health and Medical Research Council's National Statement on Ethical Conduct in Human Research (National Statement) and the CPMP/ICH Note for Guidance on Good Clinical Practice. No HREC members with a conflict of interest were present for review of this amendment.

I am pleased to advise that the document reviewed and approved at the meeting was:

- Protocol Version 4 dated 21 February 2011

Should you have any queries about your project please contact the Research Office, Tel: 8382-2075, email research@stvincents.com.au. The HREC Terms of Reference, Standard Operating Procedures, *National Statement on Ethical Conduct in Human Research (2007)* and the *CPMP/ICH Note for Guidance on Good Clinical Practice* and standard forms are available on the Research Office website: www.stvincents.com.au/researchoffice or internal at <http://exwwwsvh.stvincents.com.au/researchoffice>

Yours sincerely

Sarah Charlton
HREC Executive Officer
Research Office

CC: Denise Haylen
D/2011/2921

APPENDIX 2

Tables of literature related to living with LAM and rare disease

Table 2.1 Literature related to the experiences/perspectives of women living with LAM

Author Year Country	Aims	Design	Participants	Findings
Belkin et al. 2014 US	Capture patients' perceptions of how LAM affects their lives	Qualitative study 7 semi-structured focus groups, data analysed for themes using content methods and reflexive team analysis.	37 women aged 34-68 years living with LAM (0.4-39 years) who were attending LAMposium in Cincinnati 2013 (5 focus groups of 5 women each, 2 focus groups of 6 women)	Experiencing and managing symptoms – fatigue prominent and severely impairing Psychological experience one of “getting stuck with LAM”: frustration and resentment, worry and fear, loss of identity, embarrassment and guilt. Some adapted and showed defiance and resilience.
Carel et al. 2010 UK	Describe personal patient journey	Personal narrative of illness experience with comments by two physicians		Short description of experience. Expanded narrative contained in her book (2008/2013) – two editions
Carel 2013b UK	Reflect on broad experience of illness using author's personal experience	Book of personal narrative and philosophical reflection using phenomenological framework (mainly Merleau-Ponty and Heidegger)		At diagnosis, lack of information, empathy and support, need to do own research. Post diagnosis HCPs generally showed lack of understanding and empathy. Need to become own advocate. Post diagnosis was process of adapting, transformation, and learning to live in the present. Author was able to experience health and wellbeing.
Cohen et al. 2005 Canada	Provide update of clinical picture of LAM	Quantitative study Cross-sectional questionnaire survey analysed statistically	448 women on US LAM Foundation database 59 women listed with UK LAM Action. Participants were from US, UK, Canada and other countries	More women over 40 are being diagnosed with LAM Fatigue was common symptom, affected quality of life, overlooked by physicians Women with TSC-LAM were younger than non-TSC LAM

				<p>patients, had more symptoms, were more likely to have AML, had less dyspnoea</p> <p>There was a relationship between patient reported symptoms and reduced lung function results.</p> <p>Authors conclude research is needed to explain and reduce fatigue experienced by women with LAM</p>
<p>Cohen et al. 2009 Canada</p>	<p>Determine pregnancy and health outcomes in LAM to assist counselling patients re pregnancy</p>	<p>Quantitative study. Questionnaire survey analysed statistically</p>	<p>448 women on US LAM Foundation database 59 women listed with UK LAM Action</p>	<p>Women with LAM can have uncomplicated pregnancies. Respiratory complications and adverse pregnancy outcomes more common for those who experience exacerbation of LAM or are diagnosed during pregnancy. Pregnancy may affect lung function long term but did not increase dyspnoea or reduce quality of life.</p>
<p>Haylen et al. 2009 Australia</p>	<p>To gain an understanding of LAM and, in particular, the lived experience of persons living with LAM</p>	<p>Literature review</p>		<p>4 studies using quantitative methods were found that sought women's views on some aspects of living with LAM (QoL, measurement of respiratory function, clinical management, dyspnoea and fatigue).</p> <p>No qualitative studies exploring the experience of living with LAM were located at that time.</p>
<p>Hoy 2016 US</p>	<p>To raise awareness of LAM</p>	<p>Personal narrative of author's experience of living with LAM as a trainee physician</p>		<p>Author was medically trained, partner also a physician – access to expert facilities and</p>

				<p>investigations. Diagnosed in six days. Diagnosis – emotional rollercoaster (feelings of fear, sadness, denial, confusion, anger, grief); “kept going” as usual with life to retain sense of normality. Adjusted in year after diagnosis – accepted new normalcy (benefited from sirolimus – stabilise lung function; research gave hope; feelings of gratitude and optimism)</p>
<p>Pollock-BarZiv 2005 Canada</p>	<p>To explore and describe the lung transplant (Tx) experiences of women with LAM, including QoL, psychosocial impact, and specific needs, concerns and issues of women with LAM who are transplant candidates or recipients</p>	<p>PhD thesis, mixed methods study Quantitative: survey-statistical analysis. Qualitative: in-depth interviews, grounded theory</p>	<p>31 waitlisted and 43 transplanted women answered surveys. 11 post transplant women were interviewed</p>	<p>For most delays in diagnosis, misdiagnosis, multiple doctors and tests before diagnosis received. Deterioration in physical function prior to Tx led to changed social roles and perceptions of changed identity. Most were unable to adjust. Study focused on women with severe disease. Lung transplantation improved QoL of women with LAM. Difficulties pre-transplant: impaired function, fatigue, anxiety, isolation. Post transplant issues: depression, adverse drug effects</p>
<p>Pollock-BarZiv et al. 2005 Canada</p>	<p>To evaluate the relationship between objective and subjective functional status</p>	<p>Quantitative cross sectional study including survey of demographic data, objective lung function tests, and in survey subjective Functional Performance Inventory</p>	<p>31 lung transplant candidates and 43 women post transplant from LAM Foundation database</p>	<p>Certain aspects of spirometry/6 min walk do not correlate well with patients’ perceptions of their function. Considering subjective aspects of function may focus</p>

				interventions and improve QoL
Ryu et al. 2006 US	To describe the clinical characteristics of women with pulmonary LAM	Quantitative observational study using demographic data, lung function tests, laboratory results and quality of life instruments (SF-36, SGRQ)	230 women with LAM enrolled in the US LAM Registry between 1998 and 2001	Women with LAM experience impaired QoL. SF-36 mental and physical component scores were worse than the normal population but better than people with COPD. Results suggested both groups develop coping mechanisms
Vafamand 2014 US	To investigate the knowledge, attitude and practices of women living with sporadic LAM and the relationship of these to quality of life	PhD thesis Quantitative study. Data from online survey questionnaires. Analysed statistically. Transtheoretical model used to describe the relationship between variables.	143 sporadic LAM patients registered with the LAM Foundation	There was a significant positive relationship between attitude, lifestyle practices and QoL, and a negative relationship between knowledge and QoL
Walker et al. 2015 US	To develop and conduct initial testing of ATAQ-LAM (A Tool to Assess Quality of Life in LAM).	Phase 1. Guided by qualitative content analysis of the transcripts from a focus group study (Belkin et al. 2014) and clinical experience, a pool of 56 items was developed comprising a preliminary version of ATAQ-LAM (A Tool to Assess Quality of life in LAM) Phase 2. Item reduction and validation of tool	A convenience sample of 69 LAM patients were recruited at the 2014 LAMposium in Chicago, Illinois, to test tool.	ATAQ-LAM ("A Tool to Assess Quality of Life in LAM"), a 32-item questionnaire to assess HRQL in patients with LAM was developed. Higher ATAQ-LAM score = more impaired HRQL Subjects requiring supplemental oxygen had higher ATAQ-LAM scores than subjects not requiring supplemental oxygen; ATAQ-LAM scores were significantly higher for subjects with the lowest FEV1% compared with subjects with the highest FEV1% values (measure of lung function).
Young et al. 2006 US	To evaluate patients' perspectives regarding	23-item questionnaire	314 respondents from LAM	One third of patients believed their physicians did not consider their

	pneumothorax (PTx) treatment		Foundation database	<p>preferences for PTx management. LAM patients and their physicians held different views, most patients favouring more conservative management initially. Authors concluded understanding patients' perspectives will facilitate shared decision-making and may ultimately improve clinical outcomes in LAM for PTx</p>
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Table 2.2 Literature related to the experiences/perspectives of people living with a rare lung disease – pulmonary hypertension (PAH) and idiopathic pulmonary fibrosis (IPF)

Author Year Country	Aim	Design	Participants	Findings
Armstrong et al. 2012 UK	To investigate the patient's experience of the trajectory to diagnosis of PAH and inform provision of care	Qualitative study using in-depth interviews and pictorial representations Thematic analysis	30 patients with PAH (18F) 26-80 yrs (mean 56 yrs)	Lack of awareness of HCPs led to misdiagnosis and delays in diagnosis. Multiple tests and doctors. Emotional impact – frustration, anger, uncertainty Poor communication .Patients perceived lack of empathy when they received diagnosis. Lack of information. Internet used to search for information Lack of guidelines for diagnosis.
Belkin & Swigris 2014 US	To better understand the experiences and perceptions across the spectrum of IPF severity.	Literature review	5 studies reviewed	Delayed diagnosis, misdiagnosis, multiple medical consultations. Diagnosis: lack of competence of HCPs, emotional support, information and resources. QoL impaired; feelings of depression, anxiety and isolation. Higher levels of satisfaction reported when treated at centre of excellence.
Collard et al. 2007 US	To describe the attitudes and experiences of patients with IPF regarding diagnosis and management of their disease	A survey of 52 defined-choice and open-ended questions	1448 respondents who were members of the Coalition for Pulmonary Fibrosis (CPF)	Lack of information and resources at diagnosis. Less than half of respondents felt well-informed about treatment options, the role of supplemental oxygen, pulmonary rehabilitation, and transplantation. Need for improved patient education.
De Vries et al. 2001 The Netherlands	To study the relationship between QOL, depressive symptoms, and breathlessness in patients with IPF	Quantitative study using World Health Organisation Quality of Life Assessment instrument	Forty-one IPF patients (aged 38-80 years; 15 M, 26 F) and 41 healthy persons matched for age and sex	QoL was impaired in domains of "physical health" and "independence". Subjective breathlessness was associated with depressive mood and QoL. It was not associated with objective breathlessness measures. Major QoL problems were fatigue, mobility, activities of daily living and working capacity.

Duck et al. 2015 UK	To understand the perceptions, needs and experiences of patients with IPF	Qualitative study Semi-structured interviews Framework Analysis (Ritchie & Spencer 1994)	17 patients (7M, 10F) with moderate to severe IPF and 6 of their informal carers 12 patients were using oxygen therapy	Experience of struggle and loss Struggle to get diagnosis (delayed diagnosis, misdiagnosis, symptoms trivialised), lack of information Disruption - World turned upside down Loss of independence, control, social roles, sense of self, spontaneity Struggle to live with IPF (breathlessness, cough, fatigue, oxygen). Need for support. Hoping for effective treatment. Oxygen enabled activities, sense of control Need for specialist ILD centres and nurses
Flattery et al. 2005 US	To describe patient experiences of living with PAH and identify factors that may have an impact on QOL.	Qualitative study Semi-structured interviews Phenomenology (Colizzi's 7-step process of analysis)	11 participants (8F, 3M) 40-72 years with PAH from outpatient setting of tertiary care hospital	Participants experienced uncertainty related to illness but learned to cope with uncertainty (seeking information, making memories, humour, spirituality, finding support) and move on with their lives (adjusted to treatment, resumed life's activities)
Kingman et al. 2014 US, Germany, UK, Brazil, France, Italy, Korea	To better understand the patient's perspective of PAH, including the impact of living with PAH, disease management and treatment.	Qualitative ethnographic study using observational video footage, supplemented by field notes and patient diaries	34 patients with PAH, 5 with chronic thrombo-embolic PH in 7 countries, 10M 29F, 19-91 years	Delayed diagnosis Increasing dependency as disease progressed – change in social roles. Access to medication improved QoL Invisible nature of PAH -
Matura et al. 2012 US	To determine how people with PAH are using an online Discussion Board	Qualitative descriptive methodology was used to analyze a convenience sample of self-identified patients with PAH. Internet posts to an online Discussion Board were analyzed for common themes.	549 people with PAH who posted to the online board (142 F – 92%, 19-78 years where demographic detail available)	Used online board to seek guidance re treatments and testing, validate feelings, support through communication. Adjusted to living with PAH by refocusing their lives (paced activities, dealt with employment and interests) – sense of control
McDonough et al. 2011 US	To describe symptoms experienced by PAH patients and the impact these symptoms on their lives.	Qualitative descriptive study Telephone interviews analysed for themes	10 patients from PAH clinic of large academic medical centre (7F 3M, 38-81	Experience of symptoms: Holding back (fear, anticipation of symptoms worsening, treatment effects) – lack of spontaneity How they adapted to change: Redefining life (activity restrictions – continuous oxygen,

			years, avg 65 years)	IV medications - uncertainty, making the best of it) – incorporated illness and treatment into lives, learned to live with symptoms
Overgaard et al. 2016 Denmark	To increase knowledge of life with IPF for patients and carers	Qualitative descriptive study. In-depth dyadic interviews with IPF patients and caregivers. Five-step analysis for themes from framework method	29 patients (15 M, 10 F), 24 caregivers Recruited from specialist clinics at two university hospitals	Prolonged period of misdiagnosis Diagnosis – stages of shock, denial, acceptance Became alert to symptoms. Oxygen experienced as a limitation and necessity. Loss of social role with increasing dependency on caregiver Adapted coping strategies: Tried to maintain as normal a life as possible while preparing for death. Avoided talking about death
Pulmonary Hypertension Association Europe 2016 France, Germany, Italy, UK, Spain	To explore the impact of PAH on the lives of patients and carers	Mixed methods study 1. Qualitative one-to-one interviews with patients and carers – themes identified 2. Quantitative survey using online or postal questionnaires – informed by qualitative interviews	1. 25 PAH patients 15 carers (5 countries) 2. 326 patients (avg age 52 years 74% F) 129 carers (avg age 52 years, 52% F) Recruited from local PAH patient organisations	Problems: breathlessness, fatigue, dizziness, syncope, peripheral oedema, chest pain PAH had significant impact, on daily life, serious limitations, : physical activity (difficulty walking short distances), employment and work (loss of income), social life (invisibility of condition, lack of understanding, isolation), relationship issues (decrease in sexual relations, loss of libido) Wide range of negative emotions (fear, guilt, loss, worry, feelings of uselessness and frustration) – lack of psychological support
Schoenheit et al. 2011 US	To gain insights regarding the perspectives of people living with IPF on the diagnostic process, disease education, emotional well-being, and quality of life	Qualitative study In-depth interviews	45 IPF patients (22 M, 23 F, avg age 67 years) from France, Germany, Italy, Spain, UK	Delayed diagnosis (2-12 years), initially symptoms dismissed, misdiagnosis, many doctors. Diagnosis – lack of empathy, emotional support, and perceived competence of HCPs, insufficient time given for questions. General lack of awareness and understanding in community Post diagnosis: greater satisfaction when treated in recognised centre of excellence. Unmet needs: access to specialist care, educational resources, family support/counselling, patient advocacy and public education.

				Loss of income, feelings of being a burden, difficulty maintaining social relationships
Swigris et al. 2005 US	To gain insight into patients' perspectives on how IPF affects their lives and QoL	Qualitative study Focus groups and individual interviews. Analysed for conceptual categories of IPF-related QoL using Nvivo	20 IPF patients (13 M, 7 F, 44-82 years)	QoL impaired by cough, breathlessness, fatigue, oxygen therapy: Sleep disturbance, need for planning and preparation, loss of income, loss of independence, feelings of being unattractive, decreased libido and sexual activity, social participation curtailed. Life turned upside down – need to adjust life goals and refocus lives. Feelings of fear, worry, anxiety and panic
Taichman et al. 2005 US	To assess health-related QoL (HRQoL) in patients with PAH	Quantitative cross sectional survey of HRQoL	155 outpatients with PAH (81% F aged 18 to 84 years)	Patients had severe impairments in both physical and emotional domains of HRQoL. It is associated with functional status.
Yorke et al. 2014 UK	To explore the experience of living with PAH	Qualitative study, semi-structured one-to-one interviews, thematic analysis	30 people with PAH through Pulmonary Hypertension Association (18 F, 12 M, aged 26-80 years)	Having invisible, rare illness → lack of knowledge of HCPs, lack of understanding from others (legitimacy of illness not recognised, stigma), difficulty claiming sickness and disability benefits (anxiety, frustration) Treatment: a burden, need to adjust, financial burden, side effects, new treatment engendered optimism and sense of control, perception that educational needs and individual circumstances not taken into account by HCPs when prescribing treatments

Table 2.3 Comparison of characteristics of LAM, idiopathic pulmonary fibrosis and pulmonary hypertension

	Lymphangiomyomatosis (LAM)	Idiopathic Pulmonary Fibrosis (IPF)	Pulmonary Hypertension (PAH)
Disease	Rare, multisystem disease characterised by progressive cystic lung disease (Ryu et al. 2006)	Rare, progressive, fibrotic interstitial lung disease (Overgaard et al. 2016)	Rare disease characterised by progressive narrowing of the pulmonary arteries leading to right heart failure and eventually death (Kiely et al. 2013)
Population	Prevalence of 3.3-7.7 per million Affects almost exclusively women (pre- and post-menopausal) (Harknett et al. 2011; Taveira-DaSilva & Moss 2015)	Prevalence of 13 per million in Denmark Affects men and women (Overgaard et al. 2016)	Incidence of 1.3-3 per million per year (idiopathic PAH) - higher incidence of PAH caused by severe lung and heart disease Affects men and women (Kiely et al. 2013)
Cause	Non-inherited gene mutations (Taveira-DaSilva et al. 2006)	Unknown (Overgaard et al. 2016)	Genetic mutations (idiopathic and familial PAH), mechanical obstruction (chronic thromboembolic PAH - CTEPH), severe lung and heart disease, multiple/unclear factors (Kiely et al. 2013)
Diagnosis	HRCT Lung biopsy (Johnson et al. 2010)	HRCT Lung biopsy (Overgaard et al. 2016)	Clinical evaluation, exercise tests, biochemical markers, echocardiogram, haemodynamic assessments (European PAH Association 2015)
Symptoms	Breathlessness, fatigue, recurrent pneumothoraces, chylous effusions, lymphangiomyomas, angiomyolipomas (kidney and abdomen), cough, chest pain, chyloptysis, wheeze (Taveira-DaSilva & Moss 2015; Ryu et al. 2006; LAM Foundation 2015)	Breathlessness, dry cough, fatigue, anxiety, depression (Overgaard et al. 2016)	Breathlessness, fatigue, dizziness, syncope, chest pain, abdominal distension, peripheral oedema, anxiety, depression (European PAH Association 2015)
Treatment	Medication (sirolimus/everolimus) Oxygen therapy Lung transplantation (Taveira-DaSilva & Moss 2014)	Oxygen therapy (Schoenheit et al. 2011)	Medication (calcium channel blockers, prostanoids, endothelin receptor antagonists, phosphodiesterase-5 inhibitors), Treatment of underlying condition, Oxygen therapy Lung transplantation (Kiely et al. 2013)
Median survival time	29 years (variable rate of progression) from onset of symptoms No cure (Oprescu et al. 2013; Taveira-DaSilva & Moss 2014)	3-5 years No cure (Overgaard et al. 2016)	2.8 years untreated > 7 years with treatment No cure (Benza et al. 2012)

Table 2.4 Literature related to the experiences/perspectives of people living with a rare disease

Author Year Country	Aim	Design	Participants	Findings
Budysh et al. 2012 Germany	To describe the experiences of patient–physician interaction in rare diseases, to develop a typology of interaction patterns and to explore the antecedents of these interaction patterns, with a special focus on role behaviour	Qualitative study, semi-structured interviews, thematic analysis	107 participants with 6 rare diseases (73 adults, 34 parents of ill children; 42 F, 65 M), mean age 36.2 years	The low prevalence of the disease and HCPs lack of expertise created challenges for treating these diseases. A patient-directed interaction pattern with physicians was identified: patients became experts in their disease and drove the information-seeking and treatment processes. Traditional patient-physician roles were altered and tended to be resisted by physicians rather than patients.
Caputo 2014 Italy	To identify common themes that characterized the illness experience and quality of life of patients affected by rare disease (RD).	Quantitative with qualitative component. Content analysis of patient’s written illness stories (avg 700 words) using Emotional Text Analysis (Carli & Paniccia, 2002). Statistical analysis with text analysis software (T-LAB)	32 adult patients (8 M, 24 F) with 4 RD diagnoses, recruited from database of Italian National Centre for Rare Diseases,	Thematic domains: hopelessness, need for autonomy, search for normalcy, expectations of recovery. Latent themes: the relationship with social and medical HCPs, adjustment processes to disease and social limitations, self-beliefs and coping.
Cohen & Biesecker 2010 US	To summarize and integrate research findings to help elucidate how healthcare providers can more effectively enhance the QoL of patients affected with rare genetic conditions.	Systematic literature review	58 QoL studies of 30 rare genetic conditions	Although genetic conditions can negatively affect individuals’ lives, having a genetic condition does not necessarily entail poor QoL. Psychological wellbeing, coping, and illness perceptions, influence QoL.
Dockser Marcus 2010	Illness narrative describing author’s mother’s experience of gallbladder cancer and search for effective treatment	Personal narrative		Funding for rare cancer research comes from money raised by patients themselves. Patient advocates can speed up therapies.

EURORDIS 2009 Europe	To collect information on the healthcare needs and expectations of RD patients and their families to inform public health decision-making	Book presenting the results of the EurordisCare Survey Programme (3 surveys between 2002 and 2008)	12,000 people with 18 rare diseases in 17 European countries. Surveys conducted by patient organisations	RD patients face barriers to equitable health care: (1) lack of scientific knowledge of their disease, (2) lack of access to correct diagnosis, (3) delays in diagnosis, (4) lack of appropriate multidisciplinary healthcare, (5) lack of quality information and support at the time of diagnosis, (6) undue social consequences, (7) inequities and difficulties in access to treatment, rehabilitation and care, (8) dissatisfaction with and loss of confidence in medical and social services and (9) rejection by health professionals.
Feinberg et al. 2013 Canada	To understand the patient experience of having a rare malignancy (neuroendocrine tumours (NET)).	Qualitative study Telephone interviews Grounded theory	18 participants (8 F, 10M, aged 45-77 years) recruited through specialised NET clinic	Themes: (1) difficulty with obtaining a diagnosis; (2) difficulty finding appropriate information about NETs from physicians; (3) difficulty finding treatment centres with knowledge of NETs and (4) difficulty finding disease specific support. Attending a specialised clinic was a positive experience – information needs met.
Garau 2016	Letter to the Editor of Orphanet Journal of Rare Disease describing the experience of author's mother and her family when diagnosed with a rare cancer, leiomyosarcoma	Personal narrative		Story illustrates the main challenges that people with rare diseases and their families face: delay in diagnosis, lack of appropriate support and information, and impaired access to treatment.
Garrino et al. 2015 Italy	To explore the impact of the rare disease on patients' lives and the experience of the health professionals caring for the patients, and also to highlight the positive and the critical aspects of the health care services.	Qualitative study Autobiographical narrative interviews Phenomenology	22 patient participants (16 F, 6 M aged 21-79 years) with a variety of rare diseases who attended a Regional Centre for Reference of Rare Diseases	Diagnosis: delayed diagnosis, multiple doctors and tests, hospital admissions, sense of isolation and disorientation, uncertainty. Post diagnosis rarity not seen as problem – focus on symptoms and accepting and learning to live with disease (stay positive, try to live a normal life, alter habits) – improves QoL. Satisfied with care at RD Centre – information needs met.
Gordon 2013 US	To understand the lived experience of men with penile	PhD thesis Qualitative study – Heidegger's	13 men with penile cancer aged 41-78 years	Rarity influenced diagnosis: period of misdiagnosis and lack of information and support.

	cancer, a rare cancer	hermeneutic phenomenology Analysis: Colaizzi's (1978) method		Study mainly focused on the experience of living with the disease itself.
Huyard 2009 France	To establish (1) to what extent people with RD think that their disease's rarity causes particular difficulties, (2) to what extent these difficulties relate to other causes than rarity (i.e. other characteristics of the disease or other components of the illness experience), (3) to what extent the rarity of the disease may relate to other components of their experience	Qualitative study Semi-structured interviews Data analysis drew on French pragmatic sociology	29 patients and 15 parents of children with 6 rare diseases	Participants did not regard rarity as the major source of their difficulties when HCPs behaved in a way that met their moral needs. Difficult rare disease experience was related to the failure of HCPs to meet these expectations.
Jaeger et al. 2015 Sweden	To investigate the experiences of adults living with different rare diagnoses, how they perceived their difficulties, needs and participation in everyday life, and to identify some common issues and problems.	Descriptive qualitative study 4 focus groups of 4 RD Content analysis	1. 9 F aged 22-57 years 2. 9 F 2 M aged 18-67 years 3. 5 F 5 M aged 18-39 years 4. 8 M aged 17-69 years	Main problems related to: Lack of knowledge among HCPs and healthcare providers Lack of knowledge and understanding in the community affected activity and participation Lack of communication and cooperation within healthcare system and between medical HCPs, and social insurance system
Jefferies & Clifford 2009, 2011 UK	Two papers from single study exploring the experience of women with cancer of the vulva who underwent surgical treatment	Qualitative study using hermeneutic phenomenological approach (Heidegger, van Manen), framework analysis	13 British women aged 27-53 years, 6 months to 5 years post surgery	Rarity caused lack of awareness and knowledge: sense of isolation, few to share experiences, search for information and control of symptoms, delay in diagnosis, invisibility (of condition and lack of awareness). Need for more information and emotional support. Impact of disease: varying levels of disruption/change physically, socially, emotionally

Jessop 2014 UK	To explore the extent to which people with rare diseases structured their narrative accounts of their search for diagnosis as a quest	Deductive narrative typology using Booker's (2004) schema to search for quest structure within 16 narratives of people living with a variety of RD published 2007-2013 in BMJ as 'A Patient's Journey'.		There was a quest for diagnosis. GPs acted as helpers, consultant doctors as opponents (factor of ignorance). Diagnosis represented the final ordeal. Any boon came later as the person accepted and adapted to their illness.
Joachim & Acorn 2003 Canada	To understand, from the individual's perspective, the experience of living with scleroderma.	Qualitative study – 2 focus groups of people living with scleroderma recruited through the Scleroderma Association. Data pooled and analysed thematically	Group 1: 4 women aged 46-58 years, diagnosed 2-14 years. Group 2: 9 women aged 53-79 years, diagnosed 5-17 years.	Physical manifestations were both visible and invisible. Disclosure- for invisible was managed to minimise stigma. Living with disease: impacted on activities of daily living. Perception of living a normal life, able to cope (some used humour), hoped for more effective treatments. Living with a rare disease: makes experience more complex, face lack of understanding and knowledge, burden of having to explain condition to others.
Molster et al. 2016 Australia	To explore the healthcare experiences of Australian adults living with rare diseases	Quantitative study. Online survey via the networks and mailing lists of the Office of Population Health Genomics, within the Western Australian Department of Health and the four peak bodies in the Australian rare and genetic disease sector: Rare Voices Australia, Genetic Alliance Australia, Genetic Support Network Victoria and the Genetic and Rare Diseases Network. The survey link was initially distributed to over 300 patient support groups across Australia, as well as individuals registered with any of the collaborating organisations	744 respondents with various RD (568 F, 176 M aged > 18 years)	Healthcare needs were not being met. Experienced delayed diagnosis (50%), misdiagnosis (50%), and multiple medical consultations (66%). Perception of treatment not being holistic (lack of communication between specialities). Experiences influenced by inefficiencies/gaps in diagnostic process (may be contributed to by structural aspects of healthcare systems – fragmentation in the Australian context. Respondents were more likely to use health services and wished to be involved in research into their condition.

<p>Oksel & Gunduzoglu 2014 Turkey</p>	<p>To explore individuals' personal experiences of living with scleroderma and present a view of scleroderma patients and their nursing care.</p>	<p>Exploratory descriptive qualitative study using phenomenology (Colaizzi)</p>	<p>20 women living with scleroderma, average age 50.85 years</p>	<p>Themes: 1. Self-perception (lowered self-esteem, fear, anxiety). 2. Relationship role: altered family and social process, altered role performance. 3. Activity/exercise – activity intolerance, fatigue, physical difficulties. 4. Sexual dysfunction – sexual lives disrupted due to disruption in self-respect, altered body image, physical symptoms, altered attitude of partner.</p>
<p>Rare Disease UK 2016</p>	<p>To provide insight into the patient and family experience of RD</p>	<p>Survey of 85 questions conducted in 2015 (followup to survey conducted in 2010)</p>	<p>1203 respondents form UK with over 450 different rare diseases</p>	<p>Little had changed since previous survey: Patients still experiencing difficulties in diagnosis (misdiagnosis – some as psychological, delayed diagnosis, multiple consultations), accessing information (left alone to do their own research), receiving appropriate coordinated care, accessing treatments and finding out about research. Patients become expert and educated HCPs. Rare disease affects many aspects of an individual's life including their social, educational and employment opportunities. Increased awareness and understanding, including adjustments where needed, can help to reduce this impact.</p>
<p>Rare Voices Australia 2013</p>	<p>To highlight the experience of living with a rare disease.</p>	<p>27 personal vignettes illustrating aspects of living with a rare disease.</p>	<p>Australian people and their families living with a variety of rare diseases</p>	<p>Diagnosis often difficult and delayed, misdiagnosis is common. Access to testing is difficult, expensive and inconsistent from one state or territory to the next. RD patients are not always seen as sick - lack of understanding of what is a RD impacts individuals in their daily lives. Less than 1% of the Australian rare disease population can access an effective therapy. Clinical trials are difficult to access. Multiple medical consultations cause financial struggle. There is no national RD registry in Australia. Information limited and varies from one state/territory to another.</p>

<p>Vitale 2005 US</p>	<p>To explore the experience of living with the rare disease, mycosis fungoides (a skin lymphoma)</p>	<p>PhD thesis Qualitative phenomenological study</p>	<p>12 people living with mycosis fungoides (7 F, 5 M aged 34 – 79 years, diagnosed 4-23 years)</p>	<p>Rarity: lengthy period of misdiagnosis and ineffective treatment -delivery of diagnosis mixed (limited information, wait for referral, multiple health providers, fragmented service, treatment delayed) -unmet information needs → quest for information (internet, peer support group, emotional distress, frustration, disappointment -difficulty finding others with disease -lack of understanding – unpredictable reactions from other (stigma) -became experts -frustration with perceived limited research -process of long term adjustment, striving for normality.</p>
<p>Wallenius et al. 2009 Sweden</p>	<p>To describe the physical, psychosocial, emotional and financial consequences of having a rare diagnosis from the individual and the family perspective.</p>	<p>Quantitative study Questionnaire survey of members of Rare Diseases Sweden Association</p>	<p>1660 respondents from 30 RD organisations (62% F, aged <6 years to >65 years, most 21-64 years)</p>	<p>- Problems searching for support within healthcare system (lack of knowledge of HCPs) – cooperation limited -Complex nature of some rare diseases necessitated contact with multiple HCPs -clinical pathways not available for RD -family stressed – need for support -treated arbitrarily by social service system -added financial burden for some -meeting others through support group was a positive experience.</p>

APPENDIX 3

Biographical account of Margaret

Margaret is a 45 year old woman, born in 1965 in Europe. She has a sister three years older. Her family moved to Australia when she was seven years old. Margaret developed asthma when she was three years old triggered by animal danders such as horses and cats. She also had a history of menorrhagia, pre-menstrual tension, benign breast lumps, benign skin lesions, oesophagogastric erosions and gastro oesophageal reflux disease. She had fibroids and kidney stones removed between the ages of 40 and 45. She was an ex-smoker, having smoked 15 per day for 20 years until she was 37, three years after she was diagnosed with LAM.

Margaret experienced a gradual onset of LAM. She found it harder to do things from 27 years of age. She was first investigated for abdominal pain and increasing shortness of breath while climbing 12 stairs in 1998, aged 33 years. She was able to walk only slowly on the flat. She had a wheeze, was productive of sputum, coughed up some blood and experienced central chest pain and daily renal pain. She was diagnosed with LAM in 1999, aged 34 years, following a kidney ultrasound and a CT of the thorax and abdomen which showed lung cysts and multiple renal angiomyolipomas.

Margaret attended a LAM clinic three years after she was diagnosed. Her LAM was progressing slowly but at that time she was still working and was independent with activities of daily living. She was single and lived alone. She stopped work four years after diagnosis and moved to a Department of Housing villa in an outer urban suburb one year later. In the same year she completed a Pulmonary Rehabilitation Programme at a Respiratory Ambulatory Care Clinic. At this time she was able to walk her dog daily, exercise on a treadmill for 15 minutes a day at 2.2 km/hr and use hand weights at home. She was noted to have good knowledge of her disease and support from her family (mother, step-father and sister) and close friends.

Seven years post diagnosis, Margaret's dyspnoea on exertion had increased and she was now unable to walk her dog. She also had blood in her urine and pain due to her kidney lesions. She commenced home oxygen and was on a disability pension. Over the following three years her LAM slowly progressed. Nine years after diagnosis, she was noted to be unsuitable for transplant due to multi organ system problems. Medications included puffers (ventolin, atrovent, seretide, bricanyl, spiriva), prednisone, losec, perindopril (for hypertension), effexor, MS Contin and endone.

Eleven years post diagnosis, five months prior to her first interview for this study, she was admitted to a local hospital with acute respiratory failure and an acute exacerbation of asthma

following a four day history of fever, productive cough and increasing dyspnoea. Vital signs were respiratory rate 40/min, blood pressure 220/100, heart rate 140/min, SpO₂ 95% on 30% venturi mask. Blood gases: pH 7.295, pCO₂ 49.1, pO₂ 79.5 HCO₃ 23.1. She was admitted to a high dependency unit and treated with hourly nebulisers, IV hydrocortisone, IV and oral antibiotics, and oxygen therapy to keep her SpO₂ 88-92%. She refused non-invasive ventilation. Multiple medical emergency calls were made for increasing respiratory distress, mostly triggered by anxiety. Morphine and lorazepam were administered with good effect. An NFR (not for resuscitation) order was in place. She was discharged home 17 days after admission. It was noted that she had borderline pulmonary hypertension and that her SpO₂ was 67% on room air. This was the last entry as a discharge letter in her medical record at the hospital where she had attended a LAM clinic.

APPENDIX 4

**Tables summarising the social and medical backgrounds
of the participants**

Table 4.1 Social background

Participant (nationality)	Education	Family	Living Arrangements	Identified Spiritual Beliefs	Work	Pre-illness health/ disposition/ experiences/ interests
Aiko (Asian)	University	Only child, parents deceased Married, husband 88, one child, one grandchild	Lived with husband, later with daughter, city	Personal spirituality	retired	Fit and healthy Death of son-in-law Yoga/painting
Anna (Australian, Parents European)	University	Parents, 2 siblings (close family) Divorced post diagnosis, new partner	Lives with partner and pet dog, city	Christian, Personal spirituality	FT (full time)	Fit and healthy. Fashion, makeup, walking, sewing
Carol (Australian)	Business college	3 siblings. Divorced, remarried and now widow. 2 adult children, 2 grandchildren	Lives alone, city	Christian	Retired, Voluntary work	Fit and healthy, Walking, singing, yoga, craft
Clare (Australian)	Diploma	Parents, 1 sibling. Single, no children	Lives with parents, rural area	Personal spirituality	PT (part time)	Fit and healthy, inquisitive nature, Separated from partner after diagnosis, Walking, painting
Deb (Australian)	Professional training	Parents deceased. 5 siblings Divorced, 3 adult children	Lives alone, regional town	-	FT	Fit and healthy, Hiking, camping, gardening
Eva (European)	University	Parents separated, mother, sibling in Australia. Married, no children	Lives with husband, city	Buddhism	FT	Fit and healthy, Inquisitive nature, Escaped civil war and came to Australia aged 19, Meditation, yoga, waling
Helen (European)	University	Father deceased, mother and 2 siblings live overseas. Partner, 2 children (3 yrs, 16 months)	Lives with partner and children, city	-	PT	Fit and healthy, Gym, yoga, bike riding

Irena (Australian, European parents)	University	Parents (mother in nursing home), 1 sibling (close family) Married, 1 stepchild	Lives with husband and stepchild, city	Christian	Retired	Fit and healthy, Optimistic nature, Mother recently diagnosed with Alzheimer's Dancing, walking (pre LAM) Cooking
Jess (Australian)	University	Father deceased, mother and stepfather divorced. 1 brother. Estranged from mother. Partner, no children.	Lives with partner, city	-	FT	Fit and healthy Bike riding (pre LAM) walking, reading, television
Julie (Australian)	Business college	Parents. Husband, 2 adolescent children	Lives with husband and children, rural town	-	PT	Snorkelling, swimming, hiking, sports (pre LAM); Walking, bike riding
Louise (Australian)	University	Parents deceased, 2 siblings. Partner, 2 adult stepchildren	Lives with partner and pet dogs, city	-	Currently not working	Fit and healthy, Walking, art reading, pets, gardening
Margaret (European)	High school	Parents divorced, mother remarried, one sibling. Single	Lives alone with pet dogs, city	Reincarnation	Retired, previous dog rescue work	Asthma since childhood, optimistic, sense of humour, Art, knitting, music, meditation
Mia (South American)	University	Parents separated, mother in Australia, father overseas. Married, one adopted child since diagnosis	Lives with husband and child, city	-	PT	Asthma, bronchitis, Gym, travel, art, photography
Patricia (Australian)	Professional training	Mother alive, father deceased. Married, 2 adult children.	Lives with husband, city	Personal spirituality	retired	TS epilepsy, hearing impaired Optimistic, flexible nature, Son – TS autism – lives in supported accommodation. Husband travelled overseas for work. Exercise (walking, gym, bowling, swimming)
Ruth (Australian)	University	Married. 2 adult children, 2 grandchildren.	Lives with husband, city	Formal religion	PT, voluntary work	Fit and healthy, walking

Sarah (Australian)	University	Parents, sibling (close family) Married, 2 children (7 and 4 years)	Lives with husband and children, city	Formal religion	PT	Fit and healthy, Gym, hiking, pilates, walking, tennis, skiing, painting, travel
Ursula (Australian)	High school	Parents, sibling. Married, 3 adult children	Lives with husband and children, city	-	PT	Fit and healthy, Painting, walking
Veronica (Australian)	Professional training	Married, 2 adult children, 1 grandchild	Lives with husband, city	-	Retired	Fit and healthy, Gentle, timid nature, walking
Vidu (South Asian)	University	Parents, 2 siblings live overseas, (close family). Married, 1 adult child.	Lives with husband and child, city	Buddhism	Currently not working	Fit and healthy, Negative disposition, Walking, yoga

Table 4.2: Onset of symptoms to diagnosis

Participant (age at interview)	Onset of symptoms	Misdiagnosed (by)	Diagnosis of LAM (age at diagnosis)	Diagnosed by	Years from onset to diagnosis
Aiko (64)	Breathlessness some years pre Dx, Yr 0 [1998-2001] pneumonia, low SpO ₂ , lung biopsy, renal AMLs		Yr 0 lung biopsy (52)	Respiratory physician	< 1
Anna (37)	Yr 0 [Pre 2001] (L) PTx Yr +1 (R) PTx, mediastinal PTx Yr +2 (L) PTx, pleuradesis/lung biopsy Yr +5 breathlessness Yr +8 abdominal lymphangioliomyoma, Haemoptysis		Yr 2 lung biopsy (25)	Surgeon	2
Carol (66)	Yr 0 [Post 2001] breathless while talking	asthma	Yr 0 chest CT (57)	Respiratory physician	< 1
Clare (34)	Yr 0 [Post 2001] fatigue, breathless on exertion, CXR – pleural effusion (chyle)	chest infection (GP)	Yr +1 chest CT (32)	GP	1
Deb (52)	Yr 0 [Post 2001] breathless walking uphill, chest pain	cardiac origin (GP)	Yr +1 chest CT, lung biopsy (49)	Respiratory physician	1
Eva (38)	Yr 0 [Post 2001] breathless, chest pain, ↓exercise tolerance Yr +1 severe breathlessness during flight, bilateral PTx/pleuradesis/lung biopsy Yr +3 abdominal lymphangioliomyomas	Yr 0 nerve pain (GP)	Yr +1 lung biopsy (32)	Respiratory physician	1
Helen (40)	Yr 0 [Post 2001] breathless 1 st pregnancy Yr +1 breathless 2 nd pregnancy Yr +2 cough/back pain during labour, severe back pain 3/12 later (SpO ₂ -88%)	Yr 0 anxiety; Yr +1 depression (gynaecologist)	Yr +2 chest CT (39)	Respiratory registrar	2
Irena (47)	Yr 0 [1998- 2001] intermittent haemoptysis 6/12		Yr 0 HRCT scan (37)	Respiratory physician	<1

Jess (41)	Yr 0 [Post 2001] abdominal bloating Yr +4 abdominal bloating/pain with period Yr +5 abdominal pain Yr +8 breathlessness, wheeze, chylothorax	Yr 0 gut infection (GP); psychosomatic pain (GP); Yr +4 appendix, Endometriosis (gynaecologist)	Yr +5 abdominal ultrasound, lung biopsy (36)	General surgeon, confirmed by LAM expert	5
Julie (46)	Yr 0 [Pre 2001] chest/back pain – multiple PTx Yr +1 pleuradesis, ongoing shoulder pain Yr +7 chest pain 2 nd pregnancy, PTx, pleuradesis/lung biopsy Yrs +7-+11 ongoing pain Yr +11 breathless steps/hills Yr +14 severe bronchitis, ↓lung function Yr +16 renal AML	Yr +1 CT-small cysts – NAD; Yrs +7-+11 pain psychosomatic; Yr +14 emphysema (respiratory specialist)	Yr +16 chest CT (results of Yr +7 lung biopsy showing LAM found, had not been communicated) (41)	Respiratory physician	16
Louise (57)	Yr 0 [Pre 2001] thoracic back pain Yr +1 breathless swimming, haemoptysis – bronchoscopy NAD Yr +3 bronchoscopy NAD		Yr +3 chest CT (41)	Junior doctor	3
Margaret (45)	Yr 0 [1998-2001] abdominal pain, breathless stairs, wheeze, haemoptysis, chest/kidney pain		Yr +1 CT chest / abdomen – LAM Kidney ultrasound- multiple AMLs (34)	Respiratory physician	1
Mia (38)	Yr 0 [Pre 2001] Breathless/cough for 13 years Yr +13 breathlessness and cough worsened	Asthma, chronic bronchitis (respiratory specialists)	Yr +15 HRCT (34)	Respiratory physician	15
Patricia (62)	Yr 0 [Pre 2001] bleeding (L) kidney, r/o tumour Yr +8 diagnosis of tuberous sclerosis Yr +19 breathless stairs [1998-2001]		Yr +19 HRCT (51)	Respiratory physician	< 1 (from onset of respiratory symptoms)
Ruth (64)	Yr 0 [Post 2001] acute (L) flank pain – ruptured AML Yr +4 acute (R) flank pain – ruptured AML		Yr +4 chest CT following breast cancer (58)	Respiratory physician	4
Sarah (36)	Yr 0 [Post 2001] breathless on exertion and fatigue during and after 2 nd pregnancy, cough, haemoptysis Yr 0 - +2 altered menstrual periods, ongoing breathless on exertion, haemoptysis	Depression (gynaecologist) Asthma (GP)	Yr +3 chest CT, lung biopsy (32)	Respiratory physician (LAM expert)	3

Ursula (51)	Yr 0 [Pre 2001] breathless during 3 rd pregnancy, PTx x 2, pleuradesis – multiple cysts, no biopsy Yr +7 – PTx, pleuradesis, lung biopsy		Yr +7 lung biopsy (37)	Respiratory physician	7
Veronica (68)	Yr 0 [Post 2001] CT post r/o skin lesion – renal AML, (L) nephrectomy; breathless uphill post discharge – bilateral chylous pleural effusions – bilateral pleuradesis, thoracic duct ligation		Unknown (not documented in medical record) (62)	oncologist	< 1
Vidu (48)	Yr 0 [Pre 2001] sudden back pain – PTx Yr +1 breathless/fatigue uphill, PTx Yr +5 ongoing breathlessness/fatigue on exertion Yr +14 abdominal lymphangioliomyoma	Yr +1 asthma (respiratory specialist)	Yr +5 chest CT (36)	Respiratory physician	5

KEY

Dx diagnosis

GP local doctor

(L) left

NAD no abnormalities detected

PTx pneumothorax

(R) right

r/o removal of

SpO2 oxygen saturation

AML angiomyolipoma

Note: Specific dates are not supplied to preserve privacy. Each participant's year of onset of their symptoms is designated as Year 0, pre or post 2001. Subsequent milestones are labelled as +years from this initial point in time. The year 2001 is significant as it represents the time when the LAM Foundation was well established, and knowledge and awareness of LAM had increased through 40 research projects and 38 published articles (LAM Foundation 2015).

Table 4.3: Medical and surgical treatments

Participant	Problem	Surgery/ Procedure	Doxycycline	Sirolimus	Oxygen
Aiko				Yes, current	Yes, intermittent
Anna	Pneumothorax, haemoptysis, abdominal lymphangioleiomyoma	Pleuradesis, lung biopsy		Yes, current	
Carol					
Clare	Chylothorax	Pleuradesis, pleurectomy		Yes, current	Yes, intermittent for 6 weeks
Deb		Lung biopsy	Yes, current		
Eva	Pneumothorax, abdominal lymphangioleiomyoma	Pleuradesis		Yes, current	Intermittent, now not using
Helen				Yes, current	Yes, continuous
Irena	Haemoptysis			Yes, current	Yes, intermittent
Jess	Chylothorax, abdominal lymphangioleiomyoma	Lung biopsy, Radiotherapy, Pleuradesis	Yes, stopped	Yes, current	
Julie	Renal angiomyolipoma, pneumothorax	Partial nephrectomy, Pleuradesis, lung biopsy		Yes, current	
Louise			Yes, stopped	Yes, stopped	
Margaret	Renal angiomyolipomas				Yes, continuous
Mia					
Patricia	Renal angiomyolipoma	Lung transplant			Pre transplant
Ruth	Renal angiomyolipomas	Embolisations			
Sarah		Lung biopsy	Yes, stopped	Yes, current	Yes, intermittent (not medically required)
Ursula	Pneumothorax	Pleuradesis			
Veronica	Renal angiomyolipomas, chylothorax	Nephrectomy, Pleuradesis			
Vidu	Pneumothorax, abdominal lymphangioleiomyoma	Lung transplant			Pre transplant

Summary

Treatment	Number
Pleuradesis	7
Pleurectomy	1
Nephrectomy/partial nephrectomy	2
Lung transplant	2
Doxycycline	4 (3 stopped, 1 current)
Sirolimus	9
Oxygen continuous current	2
Oxygen intermittent current	3
Embolisation	1

Table 4.4: Effects of sirolimus

Participant	Lung function	Abdominal lymphangiomyoma	Haemoptysis	Chylothorax	Quality of life	Side Effects
Aiko						Commenced 4 weeks ago – too soon to assess effects
Anna	improved	resolved	resolved	N/A	improved	Nil
Clare	improved	N/A	N/A	resolved	improved	Nil reported
Eva	improved	almost completely resolved	N/A	N/A	improved	Nil
Helen	stabilised	N/A	N/A	N/A	less improvement due to side effects	Fatigue, bloating, mouth ulcers, delayed wound healing, joint pain, raised cholesterol, irregular menstrual periods
Irena	stabilised	reduced in size	resolved	N/A	improved	Irregular menstrual periods
Jess	improved	resolved	N/A	resolved	improved	Nil
Julie	improved	N/A	N/A	N/A	less improvement due to side effects	Fatigue, hoarse voice, cough, shaking, nausea, diarrhoea
Sarah	improved	N/A	resolved	N/A	improved	Nil

APPENDIX 5

Glossary

Asthma, a respiratory disorder characterised by recurring episodes of shortness of breath, wheezing, coughing and thick airway secretions.

Chyle, the cloudy products of digestion containing mainly fats carried from the small intestine in the lymphatic system.

Chyloptysis, coughing up chyle.

Chylothorax/ chylous pleural effusion, a collection of chyle in the pleural space of the lungs.

COPD, chronic obstructive pulmonary disease, lung disease involving increased airway resistance, for example, emphysema or chronic bronchitis.

CT scan, computerised axial tomography, a medical imaging procedure which produces cross-sectional images of the body using x-rays and digital computer technology. High resolution CT scanning (HRCT) is used to investigate the lungs for the presence of cysts indicative of LAM.

Emphysema, an abnormal condition of the lungs, characterised by over inflation and destructive changes in the walls of the air sacs (alveoli) at the end of the small airways of the lung. It results in a loss of lung elasticity and a decreased exchange of oxygen and carbon dioxide in the lung. A person with emphysema may experience shortness of breath and frequent respiratory tract infections.

GP, general practitioner, local doctor.

Haemoptysis, coughing up blood.

HCP, healthcare professional

ICU, intensive care unit

Intercostal catheter (ICC), a chest tube inserted into the pleural space between the lining of the lung and the lining of the chest wall to remove air and fluid from this space and to restore normal intrapleural pressure so that the lung can re-expand.

Lymphangiogram, a special x-ray of the lymph nodes and lymph vessels.

Lymphangiomyomas, a chyle-filled masses occurring most frequently in the peritoneum, pelvis or mediastinum. They can cause abdominal pain and bloating.

Mediastinal, pertaining to the mediastinum, the space in the chest between the lungs.

Nephrectomy, the surgical removal of one or both kidneys.

Peritoneum, the membrane lining the pelvic cavity and covering the abdominal organs.

Pleurectomy, a surgical procedure done to remove part of the pleura, the linings of the lung.

Pleurodesis, the creation of a fibrous adhesion between the pleura (the lining of the lung and the lining of the chest wall) to obliterate the pleural space. It is performed surgically by abrading the pleura or by inserting a sterile irritant into the pleural space and is used to treat recurrent spontaneous pneumothorax.

Pneumothorax, the presence of air in the pleural space, that is, the space between the lining of the lung and the lining of the chest wall. Normally the pressure in this space is less than atmospheric and the lung is expanded. When air enters the space and the pressure is equal to that of the atmosphere the lung collapses. A spontaneous pneumothorax occurs when air accumulates in the pleural space without an apparent preceding event.

Renal angiomyolipoma, a benign tumour of the kidney containing smooth muscle and fatty tissue.

Sirolimus, an immunosuppressant medication which has been found to stabilise lung function and reduce the size of chylous effusions, lymphangiomyomas, and angiomyolipomas in relation to LAM.

Six minute walk test (6MWT), a test measuring the distance a person is able to walk over six minutes on a flat surface. It is used to test exercise tolerance in chronic respiratory disease.

Spirometry, laboratory evaluation of the air capacity of the lungs to assess lung function by means of an instrument called a spirometer.

SpO₂, oxygen saturation. A measurement of the percentage of oxygen carried by haemoglobin in the red blood cells compared to the total amount of haemoglobin in the blood. Normal SpO₂ is between 95 and 100%. SpO₂ can be measured with a pulse oximeter placed on the finger.