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The Coping Strategies of Alpha-1 Deficient Patients and Their Family Caregivers

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Abstract

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MS, Walden University, 2009

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Dissertation Submitted in Partial Fulfillment

of the Requirements for the Degree of

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Abstract

Alpha-1 antitrypsin deficiency, for which there is no cure, is one of the most lethal genetic diseases among the European White population. Early and proper diagnosis is challenging, as understanding how both alpha-1 antitrypsin deficient patients and their family caregivers cope with their related stressors on a daily basis. This qualitative phenomenological study incorporated the biopsychosocial approach, the Bowen family systems theory, and the family systems model of illness to examine the experiences of Alpha-1 patients and their family caregivers related to living with the disease and coping strategies. Participants were recruited with the assistance of a national organization that serves the study population. Sixteen participants, (8 alpha-1 deficient individuals and 8 primary family caregivers) were interviewed. Data were analyzed to identify codes and resulting themes. The data analysis led to the identification to the following themes: (a) shock; (b) misdiagnosis; (c) lack of awareness; (d) lengthy multiple medical evaluations necessary to be appropriately diagnosed; (e) caregiver shock; (f) caregiver despair in watching their loved ones struggle; and (g) keeping busy and a positive family dynamic because factors help the participants cope with the challenges of alpha on a daily basis. The results provide an understanding of the complexities of alpha-1 deficiency and how it impacts the lives of alpha-1 deficient participants and their family caregivers. This information might be used by alpha-1 community members and health care practitioners to bring about social change by creating additional resources needed to improve communication between the health care system and alpha-1 community, as well as assist alpha-1 patients and caregivers in managing the disease.

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Dedication

This dissertation is dedicated to my brother Douglas Bruscino, who died in May of 2016 from the complications of Alpha-1 Antitrypsin Deficiency. He has been my light and driving force to complete the dissertation process and go forth to spread awareness and research to the rest of the Alpha-1 community.

Acknowledgments

I would like to thank the faculty who have helped me each quarter throughout my Walden experience but especially my dissertation committee, Dr. Stadtlander and Dr. Napoli, who have given me the guidance and confidence needed to achieve my PhD. Dr. Stadtlander has helped me gain the knowledge needed to complete the study, as well as provided the stability necessary to pursue future endeavors, and Dr. Napoli opened up my mind to the possibilities of psychometrics. In addition, I would like to thank my mom, who has been there through both the rough and smooth periods of my entire school career.

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Chapter 1: Introduction to the Study

Chronic obstructive pulmonary disease (COPD) is an umbrella term used to describe progressive lung disease including emphysema, chronic bronchitis, asthma, and some forms of bronchiectasis (COPD Foundation, 2017). Alpha-1 antitrypsin deficiency (AAT-D) is a relatively common genetic disorder that is related to COPD and caused by a deficiency of the alpha-1 protein in the bloodstream (COPD Foundation, 2017). People with AAT-D, which is often misdiagnosed are predisposed to COPD and liver disease, researchers have found that without the alpha-1 protein, white blood cells begin to harm the lungs and lung deterioration occurs (Anariba, 2017). AAT-D is a deficiency characterized by the development of COPD and/or liver damage in adults between the ages of 20 to 50 years old or liver disease in children (Anariba, 2017). Individuals will not know that they have the disease unless their doctor recognizes the specific signs and symptoms and asks for the designed blood examination which directly tests for the AAT-D (Alpha-1 Association, 2015). The challenges faced by patients in receiving proper diagnosis not only affect AAT-D patients but also their family caregivers.

Furthermore, the daily challenges involved in caring for AAT-D individuals place strains on caregivers' physical and/or emotional well-being, social lives, and financial situations (Lee, Lum, Xiang, Ungvari, & Tang, 2010). In the present study I examined the complexity and challenges both AAT-D patients and their family caregivers encounter daily and the most common coping methods used to best manage their stressors. In Chapter 2, I further discuss how I drew from biopsychosocial model (1977), Bowen's (1974) family systems theory and the family systems model of illness (Rolland,

Emmanuel, & Torke, 2017) to elicit a deeper insight into the experiences AAT-D patients and their family caregivers.

Background

Alpha-1 antitrypsin is a protein that is normally found in the lungs and bloodstream and which helps protect the liver and lungs from outside toxins and hazards as well as from diseases, such as cirrhosis, emphysema, and chronic COPD (de Serres, Bianco, & Fernandez, 2003). AAT-D is one of the three most common lethal genetic diseases among adult European White, affecting one out of 3,000 to 5,000 individuals (Anariba, 2017). Severe AAT-D affects an estimated 70,000 to 100,000 individuals in the United States with approximately 25 million people carrying at least one deficient gene (Anariba, 2017). World- wide, European White constitute approximately 117 million carriers, and 3.4 million are affected (Anariba, 2017). Although AAT-D is most commonly found in individuals of Northern European and Iberian descent, it has been identified in all populations (Anariba, 2017).

Common symptoms of AAT-D are reoccurring upper respiratory infections, weight loss, tachycardia when standing up, coughing up sputum, wheezing, increased breathlessness, skin problems (panniculitis), short-term memory problems, chronic pain, and sleep disturbances (Alpha-1 Awareness, 2017). Current medical treatment protocols involve the use of inhalers, nebulizers, corticosteroids, oxygen therapy, Lasix, CPAP machine, short-term pulmonary rehabilitation, and lung transplants for the most severe cases (Anariba, 2017). There are a variety of gene combinations that physicians use to determine the severity of the symptoms which are discussed in Chapter 2.

Early onset panacinar emphysema is an indication that an individual may have AAT-D and should be tested (Anariba, 2017). However, in the early stages of being symptomatic, it takes an average of seven to eight years to be properly diagnosed (Campos, Wanner, Zhang,& Sandhaus, 2005). The physical demands that are placed on AAT-D patients, as well as the severity of the symptoms and the uncertainty of AAT-D, have an impact on the psychological state of AAT-D patients.

High levels of depression and anxiety are common among AAT-D patients as they cope with challenges of AAT-D (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015). However, family caregivers are also affected by the disease, according to researchers. The disruptions of family life, the tasks and activities associated with providing care, the length of time caregiving for the patient, as well as the physical condition of the patient, may heighten the degree of stress for the family caregiver (Lee et al., 2010). Caregivers for COPD patients may observe their family member with COPD experience progressive dyspnea, physical disabilities, altered cognition, memory disturbances, and behavioral changes that will further strain the family caregiver's overall life (Lee et al., 2010). How AAT-D patients and their family caregivers experience and cope with the complexities and challenges of AAT-D is further discussed in Chapter 2.

Problem Statement

AAT-D is a disease characterized by the development of lung disease and/or liver damage in adults between 20-50 years old (Alpha-1 Association, 2015). Individuals will not know that they have the disorder unless their doctor views the specific signs and

symptoms and asks for the designed blood examination which directly tests for alpha -1 deficiency (Alpha-1 Association, 2015). Some of the most common symptoms of this disease are wheezing, weight loss, reoccurring respiratory infections, insomnia, short term memory loss, shortness of breath, and tachycardia upon standing (Alpha-1 Association, 2015). There is no cure for the disease, only management, which involves the use of bronchodilators; corticosteroids; augmentation infusions once a week as a goal to replace the missing enzyme that is needed to help preserve the liver and lungs; and, in the most severe cases, lung transplants (Alpha-1 Foundation, 2017). Hence, the treatment measures are determined by the combination of inherited genes. For instance, one copy of each gene is inherited from each parent to create two copies of the alpha-1 antitrypsin (AAT) gene which may come in three forms: M, S, and Z (Suri, 2015). However, the amount of AAT that is produced and the severity of the disorder are determined by the combination of the genes that were developed (Suri, 2015).

Since 1963, researchers have striven to unveil the complexity of AAT-D and how to better detect and manage the disease. However, it has been a slow process, and it was not until 1989 that augmentation therapy was first administered within the United States and 2007 that the first genetic counseling was offered (Alpha-1 Awareness, 2017). Although researchers continue to discover the underlining issues with alpha-1 deficiency, the challenge remains in early and proper diagnoses. For example, AAT is often undetected or misdiagnosed with only 5% of cases being diagnosed in the estimated 100,000 affected individuals in the United States, especially in its early stages, the disease also takes an average of seven to eight years after symptoms develop to be

properly diagnosed (Campos et al., 2005). COPD is an umbrella term used to describe various respiratory disorders, such as bronchectasis, chronic bronchitis, asthma, emphysema, and Alpha-1 deficiency. In particular, Alpha-1 deficiency is a genetic subtype of COPD (Hoth et al., 2015).

Previous researchers have examined the interactions between family caregivers and their patients. For example, researchers have focused on coping strategies, such as problem-solving strategies, emotional-cognitive strategies, and stress management strategies, among COPD patients and family caregivers (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015); COPD patients' experiences (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015 ;), how dyspnea in COPD patients may increase anxiety level, (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015), how the severity of the symptoms may impact alpha-1 patients' quality of life, (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015), and how illness uncertainty may increase anxiety levels (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015). In addition, a study was conducted to examine and compare family caregivers of people with early stages of COPD to those family caregivers of people with advanced COPD, using a cross-sectional design with a convenience sample (Figuerido, Gabriel, Jacomes, Cruz, & Marques, 2014). The author, Figuerido et al., discovered that family caregivers of people with early COPD used fewer coping strategies compared to those family caregivers of advanced COPD individuals (Figuerido et al., 2014). However, the problem-focused coping strategy proved to be the most helpful between the two groups

(Figuerido et al., 2014). In another study, an analysis was performed to examine from a phenomenological approach the experiences of individuals living with COPD (Barnett, 2004). The author, Barnett found that the breathlessness described by the COPD patients was considered the most difficult symptom which to lead to anxiety and the feeling of a lost role within the patient's family (Barnett, 2004).

Dyspnea has been associated with general anxiety, and it appears that it may influence activities of daily living (DeVoogd et al., 2011). Hoth et al. (2015) examined illness uncertainty, such as ambiguity and complexity, among alpha-1 deficient-associated COPD patients was examined. The authors suggested that uncertainty about physical symptoms (ambiguity) induced depression, anxiety, and breathlessness while decreasing alpha-1 deficient patients' quality of life (Hoth et al., 2015). In contrast, uncertainty about treatment and the health care system (complexity) did not have an independent effect on any of the outcomes (Hoth et al., 2015). Although these findings suggest an association between increased dyspnea with anxiety and the types of coping strategies used among those with COPD, the studies have limitations. For instance, Figuerido et al. (2014) used quantitative measures and elicited limited perspectives from family caregivers on coping strategies. The authors of the second and third studies, Barnett (2004) and de Voogd et al. (2011), recruited either participants they knew or recruited individuals from a pulmonary rehab program, suggesting that the results may not represent all COPD patients and leading to a selection bias. Finally, the authors of the fourth study, Hoth et al., had a reduced sample. For example, the study initially had 1,727 alpha-1 subtype COPD participants, but only 407 completed the study (Hoth et al.,

2015). Hence, the study may not properly represent entire alpha-1 associated COPD population.

Further qualitative research appears to be needed to better understand the complexity of alpha-1 deficiency and how psychological distress and disease specific anxieties are placed on both the alpha-1 deficient patients and their family caregiver. Moreover, an analysis of the specific coping strategies used by both alpha-1 deficient patients and their family caregivers should be further investigated in order to gain a deeper insight into the alpha-1 deficient patient and their family caregiver's personal experiences living and managing the illness. Many Alphas appear to require caregivers to help assist with their treatments, activities with daily living, and finances. Hence, in this qualitative study, I assessed common themes and coping strategies used across the alpha-1 community and how the family unit is impacted daily. For instance, I probed how family caregivers manage their finances while taking care of an alpha-1 deficient family member. In addition, I evaluated both the Alpha-1 deficient individual's and the family caregiver's emotional responses to the complexity of the illness and the added demands of financial circumstances.

Purpose of the Study

The purpose of this qualitative study was to examine the experiences of being an Alpha-1 patient and a family caregiver for someone living with Alpha-1 deficiency. In addition, I sought to examine coping strategies among alpha-1 deficient patients and their family caregivers.

Research Questions

I sought to answer four research questions (RQ):

RQ1. How do AAT-D patients describe living with their illness?

RQ2. How do the family caregivers who live with AAT-D patients describe their experiences?

RQ3. What are the common coping strategies among AAT-D patients?

RQ4. What are the common coping strategies among the family caregivers?

Theoretical Foundation

The current study involved in-depth interviews given to Alpha-1 patients and their family caregivers based upon the biopsychosocial model, Bowen's family systems theory, and the family systems model of illness. The biopsychosocial model offers a broad view of disease and attributes disease outcomes to the intricate variable interaction of biological factors, such as genetics; psychosocial factors, such as mood and behavior; and social factors involved with family (Lakhan, 2006). The model also allows for understanding of the factors' complex interactions in health, illness, and health care delivery (Lakhan, 2006).

Bowen's (1974) family systems theory relies on the patterns that develop in families in order to defuse anxiety and bridges the patient's disease state with psychology in order to treat the whole patient. Moreover, it stresses the concept of wellness where the state of being in good health is accompanied by good quality of life and strong relationships as cited in (Lakhan, 2006).

The family systems model of illness uses the family as the interactive focal point and pivots on the concept of a systemic interaction between an illness and family that evolves over time (Rolland, Emmanuel, & Torke, 2017). The model distinguishes three dimensions. First, it incorporates psychosocial types of health conditions, based on the pattern of onset, course, outcome, disability, and level of uncertainty (Rolland et al., 2017). Second, the major developmental phases throughout the disease process are observed (Rolland et al., 2017). Third, key family system variables, such as organization, communication, family life course development, belief systems, and outlook, are also analyzed (Rolland et al., 2017). I further discuss the study's theoretical foundations in Chapter 2.

Nature of the Study

For this study I used the qualitative method and a phenomenological design based on individual interviews. Latham (2017) suggests that a minimum of 12 homogenous participants is needed to ensure saturation. Although, additional participants will not provide any added insight, 15 participants are suggested by Latham as a minimum for most qualitative interview studies with homogenous groups (Latham, 2017). For this reason, I recruited a total of 16 participants, eight of whom had been diagnosed with alpha-1 deficiency and eight of whom were caregivers for alpha-1 deficient individuals (siblings, parents, or spouses). I recruited participants by advertising on alpha-1's social media support page and sending out e-mails to the alpha-1 community with the help from the director of the national organization of alphas. In-depth interviews were incorporated to gain a deeper understanding of the complexity of the illness and how coping strategies

may differ across Alpha-1 patients and their family caregivers. In addition to highlighting the common coping strategies used by Alpha-1 deficient patients and their family caregivers, study findings provide insight into the complexity of the illness from a psychological perspective.

Definitions

Alpha-1 antitrypsin deficiency: An inherited disease which is passed from parents to their children through their genes. It may result in serious lung disease in adults and/or liver disease at any age (Alpha-1 Foundation, 2017).

Burden of care: Disruptions of family life, the tasks and activities associated with providing care, the length of time caring for the patients, as well as the physical condition of the patients which may heighten the stress of the caregivers (Lee et al., 2010).

Caregiving burden: The distress that caregivers feel as the result of providing care. It is specific to the complexity and the degree of the disease care and varies from anxiety and depression to other emotional and more general responses which may heighten sleep disturbances, fatigue, and pain of the caregivers (Rha, Park, Sang, Lee, & Lee, 2015).

Chronic obstructive pulmonary disease: An inflammatory condition of the lungs that causes the airflow of the lungs to become blocked (Mayo Clinic, 2017).

Dyspnea: A term used to describe individuals who experience difficulty breathing (Alpha-1 Foundation, 2017).

Assumptions

I had several assumptions when conducting the study. For instance, during the interview process, I assumed that the participants would be truthful in their responses and would appropriately represent the alpha-1 community.

Scope and Delimitations

In this qualitative study, I recruited participants who had been diagnosed with AAT-D, who currently did not smoke, and who lived in Florida. Choosing these criteria helped keep the sample as homogeneous as possible and prevented external variables, such as other COPD issues, smoking, or various weather conditions from potential altering the results. In addition, I only evaluated family caregivers who lived with their AAT-D relative order to establish a homogenous sample and limit external factors from influencing the data.

The population of interest was AAT-D individuals and their family caregivers who live in Florida. I recruited the sample through the Alpha-1 Association's Facebook page and e-mail system. Therefore, the sample consisted of participants who had access to a computer, used Facebook, and were members of the Alpha-1 Association's Facebook group. I met participants in a public location (such as a library) close to where I live if there were participants who were too ill or who did not live near me, I administered phone interviews. The AAT-D participants had separate interviews from their caregivers to ensure that their caregivers would not be able to influence their answers, and vice versa.

Limitations

I interviewed 16 participants who lived in the southern part of the United States who had access to a computer and were a part of the National Foundation for Alphas Facebook group. Eight participantshad been diagnosed with AAT-D and did not currently smoke; the other eight participants were the primary family caregiver for their AAT-D family member. Because participant recruitment was limited to only those participants who have access to a computer, a Facebook account, and membership in the National Foundation for Alphas Facebook group, findings may not be generalized toto the broader proportion. In addition, the specificity of recruiting participants from the southern portion of the United States may have resulted in participants being different from other demographics. Results also may not be generalizable across the alpha community due to the small sample size and because people who chose to volunteer for the study may have had a different perspective than those who chose not to volunteer. Moreover, the use of a phenomenological approach and interpretive data limits generalizability compared to a quantitative study which provides data that can be measured, expressed and clearly defined (Sven, A., 2014).

Significance

The present study filled the gap in understanding by focusing on the coping mechanisms both Alpha-1 patients and their caregivers use daily. In turn, the study provided a deeper insight into the complexity of alpha-1 deficiency, coping strategies, and family unit in which possible additive care could be a part of the standard medical protocols. For example, a monthly visit with a specialized therapist could consult both the

patient individually as well as the family as a whole. Additionally, behavioral tools could be designed to fit the needs of each patient in order to help his or her severity of the illness. Moreover, this research exhibited a critical analysis to serve a better role in defining how to bring forth increased awareness and detection of the illness through gathering the personal experiences on how each of the patients was diagnosed and how long it took to appropriately be diagnosed and placed on a treatment plan. Hence, discovering the common denominator from the alpha-1 patients' medical history helped develop a plan of action needed to improve the attention Alpha-1 deficiency.

Summary

AAT-D is characterized by the development of COPD and/or liver damage in adults between the ages of 20-50 years old or liver disease in children. Thus, no one will ever know they have it unless doctors can recognize the specific signs and symptoms and ask for the designed blood examination which directly tests for AAT-D (Alpha-1, Foundation, 2017). Previous research has shown that there is a high rate of undetected or misdiagnoses of AAT-D (Campos et al., 2005). Moreover, the uncertainty and complexity of COPD has been exhibited to be stressful for both AAT-D patients and their family caregiver, as well as the degree of importance the interaction between the COPD patient and their family caregiver has on how they cope with the disease's demands placed on the family unit (Barnett, 2004; deVoogd et al., 2011; Figuerido et al., 2014; Hoth et al., 2015; Stenzel et al., 2015). However, additional research needs to discover the complexity of AAT-D associated COPD and the disease's specific anxieties that are placed on both the AAT-D patient and their primary family caregiver. The proposed

qualitative study searched to understand both the AAT-D patient and their family caregiver's experiences, as well as uncovered the most common coping strategies used by the AAT-D patient and their family caregiver daily. This chapter will be followed by a review of the pertinent literature in Chapter 2. Chapter 3 will follow with a description of the study design, participants, procedures, assessments used and how any information gathered will be assessed.

Chapter 2: Literature Review

Introduction

(COPD) is an umbrella term used to describe progressive lung disease including emphysema, chronic bronchitis, asthma, and some forms of bronchiectasis (COPD Foundation, 2017). (AAT-D) is a relatively common genetic disorder that is often undiagnosed. AAT-D is a disorder that is related to COPD which is caused by a deficiency of the alpha-1 protein in the bloodstream; people with AAT-D are predisposed to COPD and liver disease (Anariba, 2017). Without this protein, white blood cells begin to harm the lungs, and lung deterioration occurs (Anariba, 2017). AAT-D is a deficiency that is characterized by the development (COPD)and/or liver damage in adults between the ages of 20 and 50 years old or liver disease in children (Alpha-1 Awareness, 2015). Individuals will not know they have the disease unless their doctor recognizes the specific signs and symptoms and asks for the designed blood examination which directly tests for alpha-1 deficiency (Alpha-1 Awareness, 2015). Although there is no cure for the disease, management of the illness entails the use of bronchodilators, corticosteroids, and augmentation infusions once a week as a goal to replace the missing enzyme that is needed to help protect the liver and lungs from hazards (Alpha-1 Foundation, 2017). Since 1963, researchers have striven to unveil the complexity of the disease and how to better detect and manage the disease. However, it has been a slow process, and it was not until 1989 that augmentation therapy was first administered within the United States and 2007 when the first genetic counseling was offered (Alpha-1 Awareness, 2017).

Caregiving refers to the experience and tasks involved in providing assistance to a family member who is no longer independent (Lee et al., 2010). In providing care, caregivers experience a high level of burden, according to researchers. Hence, burden of care is considered the extent to which caregivers perceive their emotional and/or physical health, social life, and financial status to suffer as the result for caring for their relative (Lee et al., 2010). Moreover, the disruptions of family life, the tasks and activities associated with providing care, the length of time caregiving for the patient, as well as the physical condition, of the patient may heighten the degree of stress for the family caregiver (Lee et al., 2010). Caregivers for COPD patients may observe their family member with COPD experience progressive dyspnea, physical disabilities, altered cognition, memory disturbances, and behavioral changes that will further strain the family caregiver's overall life (Lee et al., 2010).

Despite researchers' continued efforts to discover the underlying issues with alpha-1 deficiency, the challenge still appears to remain in how to better detect, manage, and treat the patient's individual mental and physical needs, as well as their patient's family as a whole unit. Moreover, the common coping strategies that help alpha-1 patients and their caregivers live a better quality of life, as well as prevail over the challenges from the disease each day, still appear to further need researched. Previous researchers appeared to have focused on how illness uncertainty, depression, and anxiety play a role in both COPD patients and their caregivers' quality of life. In addition, past literature has suggested that specific psychosocial resources, coping styles, and CBT have an impact on the way COPD patients manage their illness. However, few researchers

have specifically examined how alpha-1 deficient-associated COPD patients' commonly use coping strategies that help them live a better quality of life, according to my review of the literature. Furthermore, there has been little focus on how alpha-1 patients' family caregivers (spouses and/or children) cope with their personal challenges daily. In the current study I examined both alpha-1 patients and their caregivers' coping strategies and evaluated factors that helped them manage the illness better than others. In this chapter, I will describe my literature search strategy and provide an overview of my theoretical foundation before revising relevant literature. I used literature mostly from COPD studies because there appears to have been fewer studies that have focused on AAT-D-related COPD.

Literature Search Strategy

I used several resources to conduct the literature review. I searched the PsychInfo database, nursing journals and the national library of medicine using the general search terms *COPD* and *Alpha-1 antitrypsin deficiency* as the root of all inquiries. With these terms, other search words such as *anxiety*, *depression*, *coping strategies*, *alpha-1 deficient patients*, and *family caregivers* were used to narrow the search. The articles that were found from these search strategies helped to produce additional resources and authors the general search did not discover. The online Walden University Library provided many of the needed articles for this review. Additionally, Alpha-1 Awareness and Alpha-1 Foundation provided additional resources that helped describe the complexity of the disease, its treatments, and overall prognosis.

Theoretical Foundation

The current study involved in-depth interviews given to Alpha-1 patients and their family caregivers based upon both the biopsychosocial model (Engel, 1977) and Bowen's (1974) family systems theory. The biopsychosocial model focuses on disease outcomes to the intricate variable interaction of biological factors, such as genetics; psychosocial factors, such as mood and behavior; and social factors involved with family (Lakhan, 2006). Clinicians use Bowen's family systems theory (Bowen, 1974) to analyze the patterns that develop in families in order to defuse anxiety and bridge the patient's disease state with psychology in order to treat the whole patient. Moreover, Bowen's thery stresses the concept of wellness where the state of being in good health is accompanied by good quality of life and strong relationships (Lakhan, 2006).

Over the years, medical providers have mainly utilized the biomedical model, which focuses on the physical causes of disease (Lakhan, 2006). However, in the 1970s an American psychiatrist George Engel was one of the first practitioners to focus on implementing patient-centered practices to address the psychological and social dimensions of patients' health concerns within the health care system (The "Biopsychosocial Approach," 2006). Engel developed the biopsychosocial model which considers biological, psychological, and social factors and their complex interactions in understanding health, illness, and health care delivery ("The Biopsychosocial Approach," 2006). While the biomedical approach takes a reductionist view, the biopsychosocial approach recognizes that different clinical scenarios may be best understood scientifically

at several levels of the natural systems continuum ("The Biopsychosocial Approach," 2006).

For instance, Block and Dorstyn (20105) used the biopsychosocial model was to evaluate the resilient levels of individuals who have been diagnosed with Multiple Sclerosis. The researchers used the biopsychosocial model to examine the interplay between the individuals' coping skills, behaviors, and thoughts, as well as social and environmental resources (Block & Dorstyn, 2015). As a result, the study helped bring forth some insight into how these individuals can achieve optimal functioning. For example, psychological variables, like self-efficacy, directly and significantly contributed to resilient levels in the sample (Block & Dorstyn, 2015). Additionally, disability-specific variables, such as fatigue and physical independence in conjunction with social support, exhibited influential resilience indirectly (Block & Dorstyn, 2015).

Studies have also shown that illness-related variables, such as time of diagnosis and severity, play an influential role in the individual's coping mechanisms. For example, (COPD) patients have exhibited an association between passive coping and poorer physical health (Manne, 2002). I used the biopsychosocial modelin the current study to evaluate coping strategies from the perspectives of alpha-1 patients and their family caregivers, as well as provide a deeper insight into the complexity of the interaction between family caregivers, patients' illness, the behavior, environment, and social experiences.

Bowen Family Systems Theory

The Bowen family systems theory of human behavior views the family as a synergistic emotional support unit which uses system thinking to describe the complex interaction within the family. Hence, families affect their members' thoughts, feelings, and actions. It is the connectedness and reactivity which makes the functioning of the family members interdependent (The Bowen Center, 2017). Bowen recognized this theory and its eight interlocking concepts, such as triangles (involves a three person relationship which may cause tension that shifts around the three relationships), differentiations of self, nuclear family emotional process, family projection process, multigenerational transmission process, emotional cutoff, sibling position, and societal emotional process (The Bowen Center, 2017).

Studies have examined how coping strategies and quality of life among the chronically ill and their caregivers may be synergistic with each other. For instance, in one study that used Bowen's Family System Theory to focus on patients and caregivers for those individuals with a complex neurological and palliative diagnosis found that family was an important emotional unit, suggesting that the stronger underlying attachment bonds between family members the more resilient they were under emotional pressure (Draper et al., 2013). Furthermore, other studies in which evaluated patients and their caregivers' relationship was a study that examined how lung transplant patients and their caregivers perceived their quality of life. Results proposed that while patient and caregiver quality of life (QOL) were related to each other, individual coping was exhibited (Myaskovsky, Dew, Switzer, McNulty, DiMartini, & McCurry, 2004). More

specifically, in families living with chronic illness, strong reciprocal effects existed between patient's and caregiver's QOL; however, the caregivers' coping strategies differed from the patients' well-being (Myaskovsky et al., 2004). In turn, incorporating the biopsychosocial model and Bowen's Family System Theory helped evaluate both the Alpha-1 patients' and their family caregivers lived experiences, as well as their coping strategies used, which connected the disease state from the patients with their perspectives involved within their family dynamics.

Family Systems Model of Illness

When patients are faced with the challenges of a debilitating disease, family caregivers must take the lead in both making medical decisions and advocating for the patients. The family systems approach to clinical practice has become essential to the understanding and treatment of individual and relational disorders (Rolland et al., 2017). Research evidence supports involving families in assessment and treatment and for the mutual influence of family functioning, health, and physical illness, as well as the usefulness of family centered interventions with chronic conditions. The FSI model provides a useful framework in clinical practice and research with families dealing with chronic and life threatening diseases (Rolland et al., 2017). Thus, taking the family as the focal point, this model can base the concept of a systematic interaction between an illness and family that evolves over time. Moreover, the FSI model can be used to analyze the association between the psychosocial demands of the disorder over time and the family style of how they function, cope, and adapt to the progressive changes of the chronic disorder (Rolland et al., 2017).

For example, a qualitative research study was conducted using the Family Systems Illness Model in order to better understand the experiences of families dealing with COPD, predominantly in the chronic phase of the illness. The findings showed that restraints in the family social life represented a significant lifestyle change for both patients and their family caregivers, secondary to the symptomatic challenges of COPD (Gabriel et al., 2014). The results exhibited that patients felt deprived from family and social activities as a consequence of the progressive COPD related fatigue, sleep disturbances, breathlessness, and difficulty in mobility. Hence, leaving them to feel sad, lonely, or abandoned (Gabriel et al., 2014). There was also data that exhibited how the COPD patient's extended family members and/or caregiver felt isolated from society.

Momentary breathlessness, long-term oxygen therapy and treatment regimen adherence place high demands on family caregivers which limited the caregivers own social activities and relationships (Gabriel et al., 2014). Gabriel et al. (2014) suggested that the family caregivers tend to define themselves based solely upon their family member's illness and personal experiences. Thus, as recommended by the FSI model, interventions should be implemented to help family members cope with the demands of the disease, as well as raise community awareness, enhance social networks and support while enabling self-care (Gabriel et al., 2014).

COPD patients often have other diseases and are at an increased risk for contracting infections, having anxiety, and/or depression, and are frequently hospitalized. The primary goal for most health care policies is to reduce the number of hospital stays and limit long term care services which places an increased demand on family members

to care for the COPD patients at home (Gautum, Werner, Lucas, 2012). According to previous studies from Norway and other Western countries, the main caregivers to the chronically ill are spouses and children (Gautum et al., 2012). The next section explored the previous research that has been examined for mostly COPD patients and their caregivers, as well as AAT-D patients and their caregivers which created the foundational literature necessary to help the current study focus on the coping strategies from both AAT-D patients and their caregivers' perspectives used daily.

Literature Related to Key Concepts and/or Variables

Like many chronic diseases, Alpha-1 antitrypsin deficiency not only places stress on the patient's physical and psychological quality of life but also impacts their caretakers. The following literature review provided an insight into the complexity of alpha-1 antitrypsin deficiency and the coping strategies used among alpha-1 deficient patients and their caretakers.

Alpha-1 Antitrypsin Deficiency

Alpha-1 antitrypsin (AAT) is a protein that is normally found in the lungs and bloodstream and helps protect the lungs from diseases, such as emphysema and chronic obstructive pulmonary disease (COPD). This disease is related to a high risk for development of jaundice in infants, liver disease in children and up to 50% of adults with AAT are affected by liver disease (de Serres, Bianco, & Fernandez, 2003). AAT-D is one of the three most common lethal genetic diseases among adult white persons, affecting one per 3,000 to 5,000 individuals. Severe AAT-D affects an estimated 70,000 to 100,000 individuals with approximately 25 million people who carry at least one

deficient gene (Anariba, 2017). World- wide, white persons constitute approximately 117 million carriers and 3.4 million are affected. Although, AAT-D is most commonly found in individuals of Northern European and Iberian descent, it has been identified in all populations (Anariba, 2017).

Some people either do not make enough of the AAT or create abnormal types of it, which may cause AAT deficiency (Thompson & Yoneda, 2017). Specifically, AAT deficiency is a heritable, autosomal recessive metabolic disease that results in the synthesis and secretion of defective AAT. Some of the abnormal AAT is retained as pathological polymers within the endoplasmic reticulum of the hepatocytes, resulting in a low plasma concentration. In turn, this deficit usually is insufficient to ensure a lifetime protection of the lungs from the proteolytic damage of neutraphil elastase, causing early onset of panlobular pulmonary emphysema (de Serres et al., 2003). AAT deficiency is suspected to promote asthma, bronchiectasis, systemic vasculitis, relapse panniculitis, rheumatoid arthritis, intracranial arterial dissection, and multiple sclerosis (de Serres et al., 2003).

On the other hand, if one were to receive only one changed gene then they will not have the disease but be a carrier. However, some people who are carriers may still experience mild symptoms of alpha-1 antitrypsin deficiency (Thompson & Yoneda, 2017). Studies suggest that AAT-deficiency carrier phenotypes (PiMS and PiMZ) and deficiency allele phenotypes (PiSS, PiSZ, and PiZZ) are suspected to make subjects susceptible to a variety of other adverse health related effects (de Serres et al., 2003).

There is a variety of gene combinations in which determines the severity of the symptoms. For instance, the most common allele is called M, producing normal levels of AAT. Most people in the general population have two copies of the M allele (MM) in each cell (de Serres et al., 2003). Other versions of the gene lead to reduced levels of AAT, such as S allele in which produces moderately low levels of the AAT protein and the Z allele that produces little to no AAT. Thus, individuals with two copies of the Z allele (ZZ) in each cell are likely to have AAT-deficiency and those with the SZ combination have an increased chance for developing lung disease (de Serres et al., 2003). It is estimated worldwide that 161 million people have one copy of the S or Z allele and one copy of the M allele in each cell (MS or MZ). Individuals with an MS or SS combination usually produce enough AAT to protect the lungs. However, people with MZ alleles have a slight increased risk of impaired lung or liver function (de Serres et al., 2003).

Alpha-1 antitrypsin deficiency was discovered in 1963 by Laurell and Erisksson (Anariba, 2017). Laurell and Eriksson found that in five of the 1500 serum protein electrophoreses (SPEP) submitted to his laboratory in Sweden had the absence of the band of alpha-1 protein. Moreover, Laurell and Eriksson discovered that three of the five patients were diagnosed with emphysema at a young age, leading them to develop the cardinal clinical features of AAT-deficiency with absence of the AAT and early onset emphysema (Anariba, 2017). For example, people whose bodies do not produce enough of the protein AAT are more likely to develop emphysema typically between the ages 30-40 years old. Physicians should suspect AAT-deficiency if the patient develops

emphysema at 45 years of age or younger, develops emphysema without having any recognized risk factors, such as smoking, chemical fumes, or pollutants over a long period of time, x-rays show less density in the lungs than normal, have unexplained liver disease, necrotizing panniculitis, bronchectasis without any cause, or asthma that does not completely resolve after the use from regular breathing treatments (Fahy, Laureaus, & Week, 2014). Although AAT is a common genetic condition, less than 10% of severely deficient individuals are currently identified (Anariba, 2017). People with AAT-deficiency are predisposed to COPD and liver disease, such as cirrhosis and hepatocellular carcinoma in children and adults.

Alpha-1 antitrypsin deficiency can occur in all ethnicities, but it is most commonly seen in white or European descent individuals. Early onset panacinar emphysema is an indication that an individual may have Alpha-1 antitrypsin deficiency and should be tested (Anariba, 2017). Alpha-1 antitrypsin deficiency (AATD) is considered one of the three most lethal genetic diseases among white adults, affecting one per 3000 – 5000 individuals and about one to five percent of those patients diagnosed with chronic obstructive pulmonary disease are estimated to have alpha-1 antitrypsin deficiency (Anariba, 2017). Severe AATD affects an estimated 70,000 -100,000 individuals and approximately 25 million people carry at least one deficient gene. However, less than 10% of severely deficient individuals have been properly identified (Anariba, 2017).

There is no cure for the disease, only management, which involves treatment measures from the use of bronchodilators, corticosteroids, augmentation infusions to lung

transplants in the most severe cases (Alpha-1 Foundation, 2017). Moreover, pulmonary rehabilitation that includes exercise training, nutritional counseling, education, energy conserving techniques, breathing strategies, and psychological counseling along with the standard medical treatment is advisable (National Heart, Lung, & Blood Institute, 2017). Furthermore, patients should be directed to avoid lung irritants, such as pollen, ash, air pollution, wood burning stoves, paint and cleaning fumes, as well as to maintain a healthy nutritional plan involved in smaller meal portions, vegetables, fruits, whole grains, lean protein, and fat free or low-fat dairy products (National, Heart, Lung, & Blood Institute, 2017). In addition, patients would be recommended to learn how to reduce stress through meditation, yoga, and breathing exercises as a goal to positively cope with the disease.

Since 1963, researchers have strived to understand the complexity of the disease and how to better detect and manage the disease. However, it has been a slow process and it was not until 1989 that augmentation therapy was first administered within the United States and in 2007 the first genetic counseling became offered (Alpha-1 Awareness, 2012). Although researchers continue to discover the underlining issues with alpha-1 deficiency, the challenge still remains in early and proper diagnoses. For example, AAT is often undetected or misdiagnosed with only 5% of cases being diagnosed in the estimated 100,000 affected individuals in the United States, especially in its early stages and takes an average of seven to eight years after symptoms develop to be properly diagnosed (Campos et al., 2005).

Chronic Obstructive Pulmonary Disease (COPD) is an umbrella term used to describe various respiratory disorders, such as bronchectasis, chronic bronchitis, asthma, emphysema, and Alpha-1 deficiency. In particular, Alpha-1 deficiency is a genetic subtype of COPD (Hoth et al., 2015).

Previous related research has examined the interactions between family caregivers and their patients, for example, research has focused on the coping strategies, such as problem-solving strategies, emotional-cognitive strategies, and dealing with the stress consequences strategies between COPD patients and family caregivers, COPD patients' experiences, how dyspnea in COPD patients may increase anxiety levels, how the severity of the symptoms may impact alpha-1 patients' quality of life, and how illness uncertainty may increase anxiety levels (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015).

Further qualitative research is needed to better understand the complexity of alpha-1 deficiency and how psychological distress and disease specific anxieties are placed on both the alpha-1 deficient patients and their family caregiver. Moreover, an analysis of the specific coping strategies used from both alpha-1 deficient patients and their family caregivers should be further investigated in order to gain a deeper insight into the alpha-1 deficient patient and their family caregiver's personal experiences living and managing the illness. Many Alphas require caregivers to help assist with their treatments, activities with daily living, and finances. Hence, in the proposed qualitative study I will examine common themes and coping strategies used across the alpha-1 community and how the family unit is impacted on a daily basis. For instance, I will look

deeply into how the family caregiver manages their finances while taking care of an alpha-1 deficient family member. In addition, I will evaluate both the Alpha-1 deficient individual and their family caregiver's emotional responses to the inter-relation of the complexity of the illness and the added demands of financial circumstances.

Impact of Illness Uncertainty on COPD Patients

The perceived uncertainty about the current symptoms and future prognosis is the challenging part of living with a chronic illness. The uncertainty illness theory states that uncertainty occurs when a person is unable to understand the meaning of the illness and predict the likelihood of the treatment, as well as the prognosis (Hoth et al., 2015). Hoth et al. (2015) analyzed illness uncertainty, such as ambiguity (physical symptoms) and complexity (treatment and healthcare system) among alpha-1 deficient associated COPD patients and found that uncertainty about physical symptoms (ambiguity) induced depression, anxiety, breathlessness while decreasing alpha-1 deficient patients' quality of life. In contrast, uncertainty about treatment and the healthcare system (complexity) did not have an independent effect on any of the outcomes stress. As alpha 1-deficiency is closely related to COPD, and little research has specifically examined alpha 1, the literature related to COPD will also be examined.

The trajectory of COPD is unpredictable and shows a slow decline interspersed with acute exacerbations of the physical symptoms, such as re-current respiratory infections and severe dyspnea. Moreover, the combination of the uncertainty of the exacerbations and the increased fear of death becomes apparent. Recent studies show

that the diagnosis of a life-threatening illness, such as COPD can trigger end-of-life fears through the awareness of death salience (Stenzel et al., 2015).

In a study conducted by Stenzel, Vaske, Kuhl, Kenn, and Rief, (2015) the researchers documented that the diagnosis of a life threatening illness, like chronic obstructive pulmonary disease (COPD) can trigger end-of-life fears. Early studies have shown that these fears can play critical roles in a COPD patient's life. In particular, the authors investigated the relevance of socio-demographic variables, illness severity, psychological distress, and disease specific anxiety as predictors of end-of-life fears in COPD (Stenzel et al., 2015). They found that illness severity was not a predictor of end-of-life fears but gender and psychological distress explained augmented differences. When disease specific anxieties were included as additional predictors, psychological distress was no longer significant. On the other hand, disease specific anxieties mediated the association between psychological distress and end-of-life fears (Stenzel et al., 2015). In the next section, studies will be highlighted to provide additional information on how the challenges of alpha-1 deficiency play a role in alpha-1 deficient individual's coping mechanisms. Understanding the importance of illness uncertainty and how it plays a role in the overall perception of both alpha-1 patients and their family caregivers may be the key to better managing the patients and their family caregivers' quality of life. These studies are useful for the current study because they are a foundation that will help to understand the effects on AAT-D's uncertain challenges and how the degree of uncertainty may be a critical factor in how the patient and their family caregiver perceive the patient's health related quality of life.

Coping Mechanisms of Alpha-1 Deficient Individuals

Coping with breathlessness is a complex and multidimensional challenge for people with COPD which involves interacting physiological, cognitive affective, and psychosocial dimensions (Jorgensen, Lombborg, Dahl, & Pedersen, 2014). People suffering from COPD most often develop a variety of strategies to cope with breathlessness, which may either negatively or positively influence not only their breathing abilities but also their overall quality of life (Jorgensen et al., 2014). The next few studies explored and captured the multidimensionality of how people cope during daily living with the COPD challenges.

In research conducted by Cooney et al. (2013), the authors studied how breathlessness influenced COPD individuals' activities of daily living and their overall perception of life. The authors found that at the initial stage of COPD, the individuals were able to co-exist with the COPD symptoms. However, as the disease progressed, the unpredictability of it strained the co-existence and even produced an uncontrollable co-existence depending on the degree of the breathlessness (Cooney et al., 2013). In another study, the authors Sunde, Walstad, Bensten, Wagnen, Rustoen, and Henriksen, (2014) analyzed the possible impact of a COPD home care model has on COPD individuals and found that it exhibited a positive progressive attempt to integrate the competencies and behaviors of the patients and the community nurses to provide follow-up care in accordance with the guidelines for interventive care. In turn, the authors suggested that the next step is to evaluate the COPD home care model's ability to assist both healthcare workers and planners to improve the management of COPD, reduction of exacerbations

and improve the quality of life and coping among patients with COPD (Sunde et al., 2014).

The impact of stress caused by a chronic disease can have consequences on daily functioning and quality of life. Adaptation to a chronic illness depends not only on the degree of the disease but also on the individual's capacity to evaluate and react to the stressors associated with the disease. A study conducted by Hesselink et al. (2004) examined the association between psychosocial coping resources and coping style with health related quality of life (HRQoL) in asthma and COPD patients separately. There were 220 recruited adult asthmatics and 53 COPD patients. The data was collected by a pulmonary function test, face to face interviews, and questionnaires about psychosocial coping resources (self-efficacy, mastery, self-esteem, and social support), coping styles (avoidant, rational, and emotional) and HRQoL (Hesselink et al., 2004). The results showed that there were significant associations between coping resources, coping style, and HRQoL in both asthmatics and COPD patients. For example, in both asthma and COPD patients, a more emotional coping style was independently associated with poor HRQoL. Asthmatics that exhibited a poor pulmonary function, showed less efficacy and mastery feelings, and a more avoidant coping style was considered independently related to poor HRQoL, whereas a more rational coping style was linked to poor HRQoL in COPD (Hesselink et al., 2004).

Moreover, COPD patients scored lower on psychosocial coping resources, higher on coping scales and poorer on HRQoL compared to asthmatics. In addition, asthmatics who had low confidence and less mastery showed poorer HRQoL. The psychosocial

resources used among the COPD patients did not exhibit an independent association with HRQoL. However, since the number of COPD patients was considered small, caution with the results was considered and the authors suggested that further research is needed in order to directly connect self-efficacy, self-esteem, and mastery to HRQoL (Hesselink et al., 2004).

Furthermore, the degree of COPD patients' symptoms may play a critical role in how they cope with their illness. Thus, it is the mind body connection that may lead the COPD patient to react in a particular manner. The authors, Jorgensen et al. (2014), conducted a grounded theory study to explore the nature of coping, specifically with breathlessness from 12 patients with moderate to severe COPD. In particular, the study examined COPD patients' physiological and behavioral parameters that could distinguish between the four coping types, such as overrater, challenger, underrater, and leveler (Jorgensen et al., 2014). As a result, the data exhibited a relation between the physiological condition and coping behavior among people with COPD living with breathlessness. However, the authors suggest that the results should be tested on a new population involving education, social, economic, and cultural aspects within a longitudinal study with more participants (Jorgensen et al., 2014). In the following section, studies will explore how CBT may manage the anxiety and depression levels of COPD patients. In the current study, I evaluated the effects of a progressive chronic long term illness, AAT-D, and how the uncertain challenges of the illness may place a strain on both the alpha-1 patients and their family caregivers' perceptions of their quality of life.

CBT, Coping, and COPD Patients

Chronic disease is associated with high levels of uncertainty. Patients need to change their behavior as part of a new lifestyle of self-care. They also endure debilitating and demanding treatments (White, 2001). These are a few factors that make adjustments to chronic illness psychologically demanding. Previous research has found cognitive behavioral therapy (CBT) to be effective in managing chronic physical illness. In particular, the cognitive therapy framework is suited to address the problems associated with chronic disease, such as mood disorder and fatigue (White, 2001). Additionally, CBT has proven efficacy to promote active self-management and help patients establish collaborative relationships with health care staff, as well as build the skills needed to manage the psychological problems linked with chronic illness (White, 2001). Thus, the following studies will evaluate the effectiveness of CBT has on COPD patients' psychological challenges.

Hynninen, Bierke, Pallesen, Bakke, and Nordus (2010), examined the effect of CBT in groups of COPD patients of both sexes with co-morbid clinically significant anxiety and depression. There were 51 participants and the main outcome measures were comprised of the Beck Anxiety Inventory and Beck Depression Inventoryn11. Measures of health status, sleep, age, and sex were also included as secondary outcomes (Hynninen et al., 2010). The findings indicated that CBT may provide rapid symptom relief for COPD patients with clinically significant anxiety and depression. However, women exhibited higher anxiety and depression levels with non-significant improvements with the aid of CBT compared to males. In addition, the younger patients appeared to have

more anxiety and depression compared to the older participants. However, both responded well to CBT (Hynninen et al., 2010). Poor sleep patterns revealed non-significant improvements with the use of CBT. Thus, COPD patients may reflect feelings and fatigue during the day as a consequence of the lung disease and possible reduced sleep quantity caused by breathlessness (Hynninen et al., 2010).

Some studies have examined the psychological needs of COPD patients. COPD patients experience disabling physical symptoms, such as breathlessness, chronic cough, and sputum production, a large proportion of patients also experience psychological distress (Baraniak & Sheffield, 2011). In particular, anxiety and depression may become prevalent. A systematic review and meta-analysis have reported that clinical anxiety and depression account approximately for 36%-40% of COPD patients which significantly shows a reduction in the patients' quality of life (Baraniak & Sheffield, 2011). A study was performed to evaluate the efficacy of psychologically based interventions, such as cognitive behavioral therapy, psychotherapy, and a progressive muscle relaxation for patients who struggle with COPD. The study was predominately comprised of COPD patients with moderate to severe disease that experience existing morbidity of mild to moderate anxiety and/or depression (Baraniak & Sheffield, 2011). The authors suggested that the interventions did not appear to be effective in reducing psychological morbidity in COPD population (Baraniak & Sheffield, 2011).

Due to the chronic and progressive character to the disease, patients are not only physically limited but often show reductions in both their psychological and social functioning which can feedback on the course of the disease, as well as on the social

environment (Leupoldt, Fritzsche, Truebu, Meuret, Ritz, 2012). Previous studies have analyzed psychotropic medications but the evidence to support COPD patients' psychological needs has been weak.

Leupoldt et al. (2012) examined a few studies which provided an overview on findings on psychosocial factors and behavioral medical approaches and found that CBT to be beneficial for COPD patients. Specifically, aspects including behavioral activation, enhancement of competency through skill building exercise, cognitive re-structuring, psychoeducation, problem solving techniques, sleep management skills, self-instruction training, and stress management have also exhibited positive outcomes as additives to the CBT (Leupoldt et al., 2012). The use of relaxation techniques, like yoga have revealed beneficial effects to calm the breathlessness of COPD patients. In addition, in a few smaller studies, biofeedback training has suggested some potential effects to oxygen saturation and ventilation patterns. Attentional distraction has been proposed as useful in reducing the preserved level of exertional dyspnea. However, studies specific to COPD patients have shown to be conflicting (Leupoldt et al., 2012). Other studies recommend that there may be a link between COPD patients' physiological and behavioral pathways. Although beneficial effects of some behavioral medical intervention has been demonstrated in COPD, future research is suggested to study the effects of distinct components of these interventions as a goal to link the importance of the psychological and sociological aspects to COPD patients' quality of life (Leupoldt et al., 2012).

The most common physical symptoms of COPD patients are breathlessness, cough, and excessive sputum production. However, additional physical consequences,

such as systematic inflammation, nutritional challenges, musculoskeletal dysfunction, fatigue, low activity, and poor exercise capacity may occur which will often lead to psychological distress (Farver-Vestergaard & Zachariae, 2015). Thus, throughout literature, it is suggested that the physical and psychological effects of the disease work synergistically together which catalyzes the exacerbation rates of the physical and psychological symptoms in a continuum (Farver-Vestergauard & Zachariae, 2015). In turn, psychosocial research has been conducted in an attempt to discover a positive supplement to medical and physical treatment for COPD patients.

Farver-Vestergaurd and Zachariae (2015) conducted a systematic review and a meta-analysis of controlled trials that evaluated the effects of psychosocial interventions on psychological and physical outcome of 1,361 COPD patients. The results revealed statistically significant effects of psychosocial interventions on both physical and psychological outcomes in COPD patients when compared with passive or active control groups (Farver-Vesterguard & Zachariae, 2015). Regarding the effects of the different types of psychosocial intervention being used with COPD patients, the moderation analyses showed that only CBT significantly increased psychological outcomes and the mind-body intervention significantly increased the physical outcomes (Farver-Vestergaurd & Zachariae, 2015). On the other hand, duration of the treatments exhibited an insignificant effect on the patients' physical and psychological outcomes. For example, the data exhibited non-significant results for both CBT and mind-body intervention on physical and psychological outcomes. The authors propose one explanation could be the deteriorating effects of the COPD patients' physical and

psychological condition over time. Therefore, the authors recommend to further examine the most appropriate duration time of treatment. In the current study, I analyzed the coping strategies of AAT-D patients and evaluated the common themes that have a critical role in impacting the patient's quality of life.

Age and Diagnosis of AAT-D- Associated COPD

Studies have found that age plays a role in how COPD patients manage their disease. A study conducted by Holin, Plautcan, Ford, Sandhaus, Strand, Strange, and Wamboldt, (2014) examined the impact of age on both the psychological and clinical outcomes among individuals with AATD-associated COPD. There were 468 individuals with AATD ages 32-84 that completed questionnaires at baseline and then at one and two year follow-up. Age was examined as a predictor of depression, anxiety, and health related quality of life, and breathlessness at all three time points using linear mixed models (Holin et al., 2014). The authors indicated that younger individuals reported more symptoms of anxiety over a two year period, regardless of relationship status. On the other hand, younger individuals reported more symptoms of depression, worse health related quality of life, and more breathlessness only if they were single. Thus, age was not associated with depression, health related quality of life, or breathlessness among individuals who were in a couple relationship (Holin et al., 2014).

The authors suggested that being in a relationship may be an important social resource for younger individuals with AATD- associated COPD. Hence, being married positively influences psychological distress and future studies may strive to analyze other social resources, such as the relationships with family and friends as additional systems

that may play a role in one's psychological and health related quality of life (Holin et al., 2014).

There is evidence suggesting that psychosocial resources in the form of social support may play an important role in managing and slowing of the progression of a variety of chronic degenerative diseases. In persons with COPD, receiving positive social support, interaction from spouse/partner, friend, extended family, and/or support group may be associated with reduced hospitalizations, better health status and promotion, disease management behavior, and engaging in physical exercise (DiNicola, Jilian, Gregorich, Blanco, Katz, 2013). Moreover, previous research has posited that the quality of the relationship may have an impact on the overall quality of life of the COPD patient (DiNicola et al., 2013).

DiNicola et al. (2013) examined the contribution of perceived social support to the presence of anxiety in persons with COPD. A sample of 452 COPD participants completed telephone surveys. Thus, the study used a cross-sectional design which measures included the anxiety subscale of the Hospital Anxiety and Depression Scale, five social support subscales from a positive and negative social exchange scale, a COPD Survey Scale, and the Geriatric Depression Scale (DiNicola et al., 2013). Positive and negative social support were defined as positive and negative social interactions. Both positive and negative aspects of social support emerged as significant and independent predictors of patient anxiety.

Two negative aspects of social support, the perceived use of insensitive and unsympathetic responses by social networks, were associated with greater anxiety in

COPD patients, whereas, stronger support from social networks exhibited fewer depressive symptoms (DiNiCola et al., 2013). On the other hand, practical help received from others increased patients' anxiety by making salient their needs and dependence of others. However, other factors such as the type of relationship (spouse, friend, extended family member, or co-worker) and quality of relationship were not evaluated within the study. The authors suggested that the type and quality of the relationship may have an influential factor for COPD patients (DiNiCola et al., 2013). Although the study provided an association between greater support with decreased depression and less support with greater depression, the relationship between depression and anxiety in chronic lung disease remains unclear (DiNiCola et al., 2013). Even though I did not specifically examine how age plays a role in the AAT-D patient's perception of life, these studies provided an insight into the timing of being diagnosed with a long term illness and how it affects a patient's perception of quality of life.

Family Caregivers

Family caregivers take on an important role with little to no training and with limited resources. They are required to provide a broad range of assistance which includes monitoring and managing the treatment and symptoms of the disease, medication administration, emotional support, as well as assistance with personal care and financial support (Rha et al., 2015). Caregiving burden is described as the distress that caregivers feel as the result of proving care. It is specific to the complexity and degree of the disease care and varies from anxiety and depression to other emotional and more general responses. Caregiving burden encompasses physical burden, such as sleep

disturbances, fatigue and pain (Rha et al., 2015). The emotional support provided by the caregiver to the chronically ill patient is considered as difficult psychological tasks. Moreover, missing work because of the caregiving responsibility and caring for others besides the chronically ill patient could be considered a social burden. The financial burden could be consequent from paying high medical expenses and losing income and savings (Rha et al., 2015). The following studies exhibited the challenges family caregivers face taking care of chronically ill patients, such as cancer, COPD, and AAT-D.

Family Caregivers and Cancer Patients

Family caregivers play a critical role in caregiving. In particular, family caregivers (FCs) of people with cancer often feel overwhelmed as a result of the intense responsibility associated with managing the patient's symptoms, providing them with emotional support, and assisting in various care related tasks (Lee et al., 2015). In turn, the high demand of care may increase the risk of the caregiver's physical and emotional decline, as well as the challenges placed on their social and financial status. Sleep problems commonly impact the physical and mental health of FCs. Previous studies have shown that between 45% and 95% of caregivers report mild to severe sleep disturbances (Lee et al., 2015).

Lee et al. (2015), suggests that sleep disturbances of perceived or actual alteration of sleep patterns. The criteria for sleep disturbance include sleeping for less than seven hours, sleep latency of more than 20 minutes, waking up more than six times per night, napping for more than two hours, and sleep efficacy of less than 80%. Lee et al. (2015)

conducted a study to assess sleep disturbance using both subjective and objective measures. There were 238 FCs of terminally ill cancer patients who initially participated and a total of 176 FCs who completed the study. The results showed that caregivers of patients with advanced cancer had more sleep disturbances compared to those caregivers of patients in the earlier stages of cancer (Lee et al., 2015).

This study provides an insight into the complexity of the challenges family caregivers face while caring for the chronically ill and will help me focus my study on the family caregiver's stressors of the physical (sleep disturbances), emotional (caring for a loved one), and the social (isolation from the outside world) and financial (high expenses and fewer hours at work) obstacles on a daily basis.

Family Caregivers and COPD Patients

Severe and progressive dyspnea is one of the most debilitating and frequent complaints of patients with COPD. Thus, pervasive dyspnea can be frightening to both the patient and the caregiver which can lead to high stress levels (Lee et al., 2010). COPD can result in physical disability, altered cognition and memory disturbances due to the lack of oxygen to the brain, as well as behavioral changes that may further increase the demand of care for caregivers. Besides the increased care for the physical symptoms of the patient, caregivers also have to be their decision maker, financial manager, deal with the patient's attitudes and irritability, and assist with their activities of daily living (Lee et al., 2010).

Lee et al. (2010) conducted a study to examine the health related quality of life and burden of caregivers of patients with COPD and to identify associated relevant

factors. The study involved 81 eligible Chinese caregivers who completed a caregiver survey on HRQOL, caregiving burden, and other biopsychosocial factors. The results suggested that the psychosocial well-being of caregivers was associated with their physical health, anxiety and depression levels, and social support. In particular, emotional distress was exhibited in COPD caregivers with little to no support or assistance (Lee et al., 2010). Thus, those caregivers who experienced a low level of daily emotional support perceived caregiving in a more negative manner which lead to depression over time (Lee et al., 2010). This study provides an in-depth view from a caregiver's perspective on caregiving to COPD patients and how the emotional and social support may be as important to the caregiver as the COPD patient. It was useful to my study because it helped me focus on the experiences of the caregiver which I may build upon to understand the needs of the caregiver to AAT-D patients.

Family Caregivers and Coping With COPD Patients

The demands of COPD can place a significant burden on family members. COPD is an incapacitating disease which progresses, and the exacerbations and comorbidities of the disease severely affect the patients' physical and social dimensions of life (Jacomeedo, Gabriel, Cruz, & Marques, 2014). The gradual disability experienced by patients contributes toward the dependence on family assistance which becomes the central and support. In turn, the increased demand on the family caregiver develops into elevated levels of stress (Jacome et al., 2014).

Jacome et al. (2014) examined the psychological health of family caregivers and patients with COPD. The main findings suggested that family caregivers presented

clinically significant anxiety and depressive symptoms, their perceived burden, female gender, older age, and patients' activity limitations were significant predictors of their distressing symptoms. The results also indicated that female caregivers had statistically significant higher levels of anxiety and depression than male caregivers (Jacome et al., 2014). However, no statistically significant differences were found for both anxiety and depression considering the quality of relationship between the family caregiver and the COPD patient. There was a statistically significance in depression among the family caregivers who cared for patients with advanced COPD compared to those patients who were in the early stages of COPD (Jacome et al., 2014). On the other hand, anxiety showed no statistically significance among family caregivers of either early stages of COPD or advanced stages of COPD (Jacome et al., 2014).

Lee et al. (2010) examined HRQOL and the burden of COPD caregivers and to identify the relevant factors associated with the burden of care. The study included a total of 81 caregivers that completed a caregiver survey on HRQOL, caregiving burden, and other biopsychosocial factors. Descriptive statistics, correlations, and multiple linear regression models were used to analyze the data. The authors suggested that the psychological well-being of caregivers was associated with their physical health, anxiety symptoms, and social support. Emotional distress was encountered in COPD caregivers with little or no support and assistance (Lee et al., 2010). However, there were some limitations to the study. For instance, the cross-sectional study may not reveal the effect on the caregiving burden and caregivers' QOL (Lee et al., 2010). Additionally, the use of only Chinese participants may not be representative of the majority of COPD caregivers.

Thus, cultural perspectives may play a role in how they share their feelings and communicate with each other.

Caring for a family member with a progressive chronic disease like COPD can put a strain on finances. Not only are there high costs for the traditional medical treatments on a daily basis but also on the family's personal finances. Hence, the cost of the electrical bill may be high due to the oxygen therapies and lowered air conditioning temperatures needed to make the COPD patient more comfortable. Additionally, the loss of work hours from both the patient and family caregiver places a strain on the family's income. Research shows in the United States, individuals with COPD receive an additional 5.1 hours a week of informal care compared to those without lung disease which represents an annual cost of $2 billion dollars (Figueiredo et al., 2014). Thus, these numbers indicate that the family caregivers provide a vital source of care to their relatives. As a result, the combination of the increased demands on finances and caring for a loved one impacts the caregivers' psychological well-being (Figueiredo et al., 2014).

As an example, Figueiredo et al. (2014), conducted a study to analyze the subjective burden of family caregivers of people with early and advanced COPD. The study was from a cross-sectional perspective, involving a total of 167 family caregiver with 113 caring for people with early COPD and 54 caring for those COPD patients with advanced stage. The authors suggest that family caregivers of people with advanced COPD experienced more stress compared to those caring for early stages of COPD. Caregivers of advanced COPD also reported higher depression and a worse self-rated

mental health compared to their counterparts (Figueiredo et al., 2014). Moreover, COPD severity was found to be a significant predictor of the caregivers' subjective burden, as well as the hours per week spent on caregiving tasks. The frequency of anxiety symptoms was found to be similar in both groups, underlying the uncertainty and unpredictable nature of the disease (Figueiredo et al., 2014). Furthermore, significant correlations among financial consequences, anxiety and depression scores, and self-rated health were discovered, positing the stressful entity of financial implications of COPD. The authors propose that the financial strain is mainly related to expensive medication, oxygen therapy, along with the loss of the care recipient's income due to early retirement forced by the disease (Figueiredo et al., 2014). In the current study, I examined the stresses of everyday life for the family caregivers, such as the physical and emotional strains on watching and taking care of their loved ones with AAT-D, as well as the financial and social struggles they face daily. In addition, I evaluated the coping strategies of the family caregivers and examined the common coping strategy that manages their stresses daily.

Family Caregivers (Spouses) and COPD

Patients experience and deal with changes caused by the disease and its treatment, as they do with the increasing dependence on others. Thus, individuals with COPD have to cope with social, practical, and existential challenges that affect the well-being of both themselves and their relatives. Living with a person who suffers from COPD has both a physical and mental impact (Ek, Ternestedt, Andershed, & Sahlberg- Blom, 2011). Previous research has suggested that family caregivers suffer from fatigue and live in

constant fear of the ill person's impending death. Ek et al. (2011) conducted a study to examine couples who experience living together with a partner who has advanced COPD. The study involved four couple over an eight month period using repeated qualitative interviews. As a result, Ek et al. (2011) suggested that the phenomenon of living as a couple when one partner has advanced COPD was found to consist of one main theme, living with the disease and one's spouse is a new changeable life rhythm. In particular, the underlying common denominators were uncertainty, living with a changed intimate relationship (less physical intimacy), and finding new ways of living together. These studies provide background information to further analyze the perceptions of the family caregivers, such as spouses and children. In particular, how each of the family caregivers manage not only their physical and mental health but also their family member struggling with AAT-D.

Family Caregivers (Spouses), AAT-D, and Communication

Both spouses exhibit similar beliefs, attitudes, and behaviors which are often exposed to the same experiences. For instance, a couple may receive the news that their partner has been diagnosed with alpha-1 deficiency and share the same negative feelings (Smith, et al., 2014). Thus, it is a synergistic effect of feelings and emotions on a daily basis. However, spouses may talk with each other to cope with their shared reactions which may lead to altered stress levels.

Smith et al. (2014) conducted a study to examine the intrapersonal and interpersonal influences in 50 couples. Their findings suggested as the couples felt more negative emotions about AAT-D, there were increasingly higher levels of stress.

Additionally, for spouses, communication about AAT-D and stress were positively related. Those who tested positive for AAT-D identified their spouses as their primary source of support. However, spouses may be stressed because they may not feel that they are being supportive enough or they may suppress their own personal worries and uncertainty of the illness which creates heightened stress for the spouses. In turn, Smith et al. (2014) suggest that the spouses may need assistance in communicating their own feelings. Moreover, the data indicated that patients with AAT-D communicated more about their feelings and emotions on the challenges they face with their disease with their spouse who felt less social and family isolation stigma as compared to spouses who perceived a stronger sense of social and family isolation stigma. Smith et al.'s study provides the current study additional information on how important open communication of feelings and emotions are to the management of the fear and uncertainty levels brought on by the challenges of AAT-D. In the current study, communication between the AAT-D individual and their family caretaker (spouse) showed as one of the key elements that helps both the AAT-D individual and their caretaker cope with the stresses of the complexity and uncertainty of the illness.

Summary

` AAT-D is a genetic disorder with a high risk factor for the development of COPD, affecting most commonly the white population (de Serres et al., 2003). AAT-D is often undetected or misdiagnosed, taking an average of seven to eight years after symptoms develop to be properly diagnosed (Campos et al., 2005). In addition, the uncertainty and complexity of COPD has shown to be stressful for not only the patient

but also their family caregiver (Stenzel et al., 2015). Moreover, previous research has shown that the interaction between the COPD patient and their family caregiver has an altering result in how they cope with the disease's physical and psychological demands on a daily basis (Barnett, 2004; de Voogd et al., 2011; Figuerido et al., 2014., Hoth et al., 2015).

What is not known, however, is the complexity of AAT-D associated COPD and the disease's specific anxieties that are placed on both the alpha-1 deficient patient and their family caregiver. The current research examined and determined the challenges of proper diagnosis. For example, what are the most common coping strategies used across the alpha community that helps both the alpha-1 patient and their family caregiver manage the stressors on a daily basis? Is there one specific coping mechanism or a combination of factors that influences the patient's and their caregiver's overall management of the illness, as well as the caregiver's daily personal and social aspects? Chapter 3 provides information on how this study was performed, how the participants were identified, the questions that were asked, and how the information was organized and analyzed.

Chapter 3: Research Method

Introduction

In Chapters 1 and 2 I discussed how AAT-D is a genetic disorder that is a high-risk factor for developing COPD, which most commonly impacts the European White population (de Serres et al., 2003). In addition, I described the high rate of undetected or misdiagnosed AAT-D (Campos et al., 2005). Moreover, I reviewed research indicating that the uncertainty and complexity of COPD is stressful for both the AAT-D patient and their patient's family caregiver (Barnett, 2004; de Voogd et al., 2011; Figuerido et al., 2014; Hoth et al., 2015; Stenzel et al., 2015).

The reviewed research also highlights, the importance of the interaction between the COPD patient and the patient's family caregiver on how both parties cope with the disease's demands (Barnett, 2004; de Voogd et al., 2011; Figuerido et al., 2014; Hoth et al., 2015; Stenzel et al., 2015).

As I noted, what is not known, however, is the complexity of AAT-D-associated COPD and the specific anxieties that are of both the alpha-1 deficient patient and the patient's family caregiver. Understanding the experiences of both the AAT-D patient and the patient's family caregiver and discovering the most common coping strategies used by the AAT-D patient and the patient's family caregiver may bring forth a deeper insight into the daily challenges faced by patients and family caregivers. In this chapter I outline the qualitative method I used in the study and present the research questions, methodology, design, as well as the overall data analysis.

Research Design and Rationale

Research Questions

RQ1. How do AAT-D patients describe living with their illness?

RQ2. How do the family caregivers who live with AAT-D patients describe their experiences?

RQ3. What are the common coping strategies among AAT-D patients?

RQ4. What are the common coping strategies among the family caregivers?

Research Methodology and Rationale

I used qualitative methodology for the study. Previous researchers appeared to have focused on the challenges COPD patients face on a daily basis, as well as how the severity of their symptoms may impact their health-related quality of life. In addition, researchers have focused on how COPD patients' caregivers may manage the daily challenges from psychological and physical perspectives. In contrast, I examined the specific complexities of AAT-D associated-COPD and explored AAT-D patients' experiences and coping strategies in conjunction with their family caregiver's experiences and coping strategies. Selection criteria for the eight AAT-D patient participants were that they had be diagnosed between the ages of 20 and 50 years old and did not currently smoke, so other confounding factors can be eliminated. The family caregiver who was chosen was considered to be the primary caregiver, which could be a spouse, child, or sibling of the AAT-D patient.

Research Design and Rationale

I chose phenomenology as the research design to examine the experiences of AAT-D patients and family caregivers. I conducted a phenomenological inquiry to reveal unified experiences across the same population and described a phenomenon that exists in people's minds that is most appropriately explained through the perspective of those who have faced similar experiences (Barken, Thygensen, & Soderhamn, 2018; Creswell, 2014). Some previous researchers have incorporated a phenomenological approach to better understand the lived experiences of COPD patients. Barken et al. (2018), for instance sought to describe the quality of life among patients challenged with COPD; the researchers included participants in a telemedical intervention after hospitalization from their disease exacerbation. The findings showed that telemedical intervention can reduce the perceived limitations of living with COPD by eliciting support from health professionals, strengthening clinical insight, and developing a mutual clinical language that may help keep clear and open communication channels between patients and health professionals (Barken et al., 2018). For this reason, I selected a phenomenological approach to examine the lived experiences of both the family caregiver and the caregiver's and AAT-D family member.

Role of the Researcher

I observed the participants' behaviors while conducting interviews. I conducted 16 total interviews, one interview for each of the eight AAT-D patients and one interview for each of the eight family caregivers. There were no personal or professional relationships between the AAT-D patients or family caregivers and me. However, a few

of my family were members of the Alpha-1 Association and were precluded from participating in the study. To help limit possible bias in the research, I approached the participants neutrally, without personal bias, and kept a journal where I related any emotions I experienced during the interview process. Moreover, I was not engaged in social conversations or shared personal experiences about AAT-D with the participants during the entire study. I also provided nonleading interview questions. The participants chose the most appropriate and comfortable environment in which to be interviewed. If the participants began to experience physical and/or emotional challenges during the interview process, they were free to leave the study and reconvene at a later time. Notes were provided and/or adjusted within the results.

Methodology

Participant Selection Logic

I interviewed 16 total individuals, of whom eight were AAT-D patients and eight were family caregivers. I selected participants using purposive sampling. The AAT-D participants were diagnosed with AAT-D between the ages of 20 to 50 years old and did not report smoking at the time of the study. The AAT-D patient's caregiver was a spouse, child, or sibling who was the primary caregiver. Participants were recruited through Facebook and/or e-mails within the Alpha community that lived in the southern part of the United States. Discussing the study with alpha-1 community members helped me to identify AAT-D patients and their family caregivers who might participate in the study. I used Facebook and/or e-mail to contact interested participants to confirm study participation and begin the interview process. A letter describing the study was sent to

the Alpha-1 community and potential volunteers. This letter can be found in Appendix A.

Instrumentation

The purpose of this study was to examine the daily experiences of AAT-D patients and their family caregivers face. In addition, I analyzed coping strategies among both AAT-D patients and their family caregivers. The goal was to discover the most common coping strategies used among AAT-D patients and their family caregivers helps to manage daily stressors. I posed four broad research questions (see Chapter 1) to better understand participants' experiences. The study involved one interview administered to each AAT-D patient and one interview to each family caregiver. Each interview focused on the participant's life history and encompassed the participant's detailed reflection on his or her experiences. However, if a participant indicated being overwhelmed, either physically or emotionally, I stopped the interview process and finished it at another time when the participant was ready to continue. Specific interview questions are listed in Appendix B.

Procedures for Recruitment, Participation, and Data Collection

The following procedure served as a sequential guide to recruit and inform participants, collect, and analyze data, and validate findings. Contact was made via telephone to the person in charge of the National Foundation of Alphas to provide information about the study to the alpha community. An informative letter detailing the nature of the study was sent to the person in charge of the National Foundation of alphas, requesting assistance in recruiting the participant. An additional letter was sent to the

Alpha-1 community requesting permission to recruit and conduct the study using the community members. Advertising the nature of the study was through the Alpha-1 community Facebook page, emails, and phone. Thus, any male and female AAT-D patients and their family caregivers who were interested in participating in the study was informed to contact the researcher to schedule an initial interview and a follow-up telephone call was made if there is not contact within one week of informative meeting.

During the individual interviews, each participant was given a copy of the letter describing the proposed study and signed the Consent Form. The interview included asking questions listed throughout the interview in Appendix B. After the interviews were completed, the audiotapes were transcribed verbatim and analyzed according to the steps at the end of the chapter. The interviews were conducted in an individual format to provide further validation that the results depict their experiences.

Data collection. Data was collected through an interview process. The interview focused on the background of the participant with the intent to put the participant's experience in context. This interview involved building a rapport, signing the consent form, and gathering information about the individual's life up to the present time. Stadtlander (2015) recommended placing the participant's experience in the context of the topic. Information gathered during the first interview provided insight into the complexity of AAT-D and how it has impacted the AAT-D patient and their family caregiver from the time of diagnosis to the present time. The interview also entailed describing the nature of the study and shared the researcher's personal experiences related to AAT-D. The purpose of sharing this information assisted in building a rapport

with the participants.

The interview also addressed the details of the participant's present experience by asking focused questions that stem from the research questions in the study. You may view the interview questions in Appendix B.

To address the third and fourth research questions, each AAT-D participant was asked to describe specific coping strategies that they feel helps them manage both their physical and psychological state on a daily basis most efficiently and how often they engage in these coping strategies. Each question for the interview process is listed in Appendix B.

Lastly, the interview entailed having the participants describe the meaning of their experience as being an AAT-D diagnosed patient or the sole family caregiver to these family members. Additionally, the questions served to validate the overall meaning for the group of participants discovered during the analysis of data. The interviews were conducted in a location selected by the participant that is free from distraction and ensures privacy. Each interview was tape recorded and transcribed. Additionally, notes were taken to document nonverbal communication and provide details of the environment.

Data was organized by creating files of the transcribed interviews. Files and recordings were maintained in a locked filing cabinet in the researcher's home office. Once the data was transcribed the process of analyzing the information began.

Data Analysis Plan

Once the data was organized, I obtained an overall understanding of what type of information the data is providing. The goal of the first step was to begin to understand what type of information the data is conveying. The second step was to begin highlighting the statements in the text that have specific meaning and importance to the phenomenon being studied. In this case, the proposed research attempted to extract statements to understand the complexity of AAT-D, how it impacts both AAT-D patients and their family caregivers, and if there is one or a combination of coping strategies that best manages the stressors exhibited by AAT-D patients and their family caregivers on a daily basis. Each presented experience helped bring forth the necessary variables that have the potential to be labeled. Thus, the most influential factors to the participants' experiences were necessary to understand and those factors that were not considered an impact was eliminated. Labeling the statements identified as necessary to understanding the phenomenon was the third step in understanding both AAT-D patient and their family caregivers' challenges experienced daily.

For the proposed study, I began labeling the underlying meaning factors in the form of physically and psychologically sensitive expressions. These expressions represented the physical demands, feelings, emotions, and actions concerning how the participants have and continue to experience the phenomenon. The statements were reframed by using meanings to describe the everyday ordinary language concerning the phenomenon. The final step analyzed the data involved in developing the individual and then group descriptions of the experience. Constructed what it meant for each participant

to experience the challenges presented by AAT-D and then connected each participant (AAT-D patients and family caregivers) to the experience that helped explain the essence of the phenomenon as a family unit.

Issues of Trustworthiness

The purposed research findings verified, which is common in qualitative studies. Morse, Barnett, Mayan, Olson, and Spiers (2002), posits that the use of verification strategies helps to ensure the reliability and validity within a qualitative study. The purposed research employed triangulation to help ensure credibility and the use of rick, thick descriptions to convey the findings which employed the appropriate strategies to establish transferability. Creswell (2014) recommends that triangulation will help build a cohesive justification of themes and if themes are found throughout the study based upon the perspective from the participants, then validity to the study has been increased. Thus, one of the goals for the purposed study was to discover the most common and beneficial coping strategies used across both the AAT-D patients and their family caregivers that helps manage their stressors most optimally daily.

Moreover, the use of rich, thick description across the detailed shared experiences from the challenges of AAT-D among both the AAT-D patients and their family caregivers helped add to the validity of the findings and conveyed the results more realistically (Creswell, 2014). The proposed study had verbatim transcripts that provided contextual and descriptive information. There were process notes in the margins of the transcript that highlighted the meaning and directed quotes from participants beneath physical and psychological expression. Each of the aforementioned method of analysis

was important in providing a rick, thick description. Moreover, dependability and confirmability was accomplished through audit trails. For instance, I kept a detailed written record of everything done in the study, whom I met with, and the specific topics that were discussed. Thus, the transcribed verbatim transcripts and the additional detailed notes about the conducted study and surrounding environment, whom I met with, and the degree of the explained results from the data collected helped decrease the level of biased research, increasing the connection between the findings and the focus of the study.

Ethical Procedures

The participants in the study were adult male and female volunteers who were free to choose whether to participate. There was no known harm associated with participating in this study. If a participant experienced harm or difficulty associated with participating in this study, a referral to local services was made. If any participants felt any emotional distress from the questions presented in the study, a number for the hotline or the genetic counseling center provided by the Alpha-1 community was provided. Each member completed a consent form and confidentiality was protected. Files, audiotapes, and transcripts were stored in a locked cabinet in the researcher's home office. Only the researcher and those selected to assist in validating results had access to the transcripts. All the data was maintained on a password protected flash drive and/or hard drive with limited access to the information. Identifying information was removed from transcripts prior to data validation and all data will be destroyed after five years.

Summary

The current study examined the complexity of AAT-D and how it impacts both the AAT-D patients and their family caregivers on a daily basis. In particular, the analysis involved a qualitative approach with a phenomenological design as a goal to gain a deeper insight into the personal experiences of the AAT-D patients and their family caregivers. Moreover, one interview per participant was conducted while maintaining ethical procedures and established reliable and valid pertinent data to the study. Chapter 4 will provide information describing the location of the study, how often I met with the participants and the length of time both for the individual interviews, as well as for the total study. In addition, how I recorded and transcribed the interviews will be explained. Moreover, the details on how I analyzed the qualitative data will be exhibited. Lastly, I will describe how I implemented the strategies and plans discussed in Chapter 3.

Chapter 4: Results

Introduction

There is little previous research about the lived experiences of Alpha-1 patients and their primary caregiver and how both parties cope in their daily lives, according to my review of the literature. My aim in the current study was to examine the experiences of being an alpha-1 patient and a family caregiver for someone living with alpha-1 deficiency. In addition, I sought to examine AAT-D patients and caregivers' coping strategies. I used a descriptive phenomenological method which gave me the opportunity to examine each participant's experiences as a whole while maintaining the participant's perspectives. The individual interviews were informed by the research questions that I developed and used to gather the necessary data. Chapter 4 includes a detailed description of the study, the setting, and the participants' demographic information. The chapter also includes an overview of the data collection and data analysis procedures, evidence, trustworthiness, and the study results. I conclude Chapter 4 with a summary of key points made in the chapter.

Setting

I conducted the study with 16 participants, of whom eight were alpha-1 deficient patients and eight were primary caregivers from the southern part of the United States. All the alpha-1 participants met the study's requirements of having been diagnosed with Alpha-1 deficiency and not currently smoking all the caregivers met the study's requirements of being the primary caregiver for the Alpha-1 deficient patient. I contacted K. Erickson (personal communication, March 14, 2018), who is the associate director of

the Alpha-1 Foundation, via telephone to explain the study and ask for her assistance in sending out e-mails to the alpha-1 community. Participants who were interested in my study contacted me via e-mail. I e-mailed the interested participants the informed consent and scheduled a time and day to conduct the interviews. The prospective participants signed and sent back the informed consent.

After receiving the informed consent form, the prospective study participants detailed information about the study and the steps I would take to conduct the interview. All prospective participants gave consent. The interviews were conducted over the phone and recorded. Neither during nor after the completion of the interview did any of the participants appear to be distressed. The interviews lasted 30 to 60 minutes, and there were no apparent problems. I did not conduct a pilot study. After the interviews, I transcribed and typed the interviews. Once the interviews were completed, I thoroughly reviewed the interview transcripts and encrypted them. I e-mailed them to my dissertation chair, Dr. Stadtlander, to review the accuracy of the typed transcripts. I kept each participant's information confidential by using an identification code for each participant and securing data.

Demographics

All eight alpha-1 participants were diagnosed with a specific genetic code of Alpha-1 deficiency and used various medications to help them manage their symptoms. They reported that they did not currently smoke. Additionally, the eight primary caregivers in the study were either the patient's spouse or significant other who primarily

cared for the alpha. Table 1 displays the demographic information for each of the alpha-1 participants.

Table 1

Main Study Demographics

Participant	Genetic code	Medications	Counseling
AAT-D 1	SZ	Weekly infusions, oxygen, and nebulizers	None
AAT-D 3	ZZ	Weekly infusions, oxygen, and nebulizers	None
AAT-D 5	ZZ	Weekly infusions, oxygen, and nebulizers	None
AAT-D 8	ZZ	Weekly infusions, oxygen, and nebulizers	None
AAT-D 9	ZZ	Weekly infusions, oxygen, and nebulizers	None
AAT-D 11	ZZ	Weekly infusions, oxygen, and nebulizers	None
AAT-D 13	ZZ	Weekly infusions, oxygen, and nebulizers	None
AAT-D 15	ZZ	Weekly infusions, oxygen, and nebulizers	None

Table 2 displays the demographic information for each of the primary caregivers.

Table 2

Primary Caregiver Demographics

Participant	Relationship to alpha participant	Counseling
Caregiver 2	Wife	None
Caregiver 4	Wife	None
Caregiver 6	Husband	None
Caregiver 8	Husband	None
Caregiver 10	Husband	None
Caregiver 12	Husband	None
Caregiver 14	Husband	None
Caregiver 16	Wife	None

Data Collection

The study began after I received approval from Walden University's Institutional Review Board (approval #0124061), and I completed data collection in October 2018. The process from recruitment to the completion of the data collection took approximately four months. Participants communicated with me via e-mail to consent to study participants and to schedule interview times. I conducted the interviews by phone. All the participants engaged in the interviews from their homes. At the beginning of each interview I explained to the participant that the interview would be recorded and the information given would be kept confidential. I asked that during the interview each participant be separated from the other participant to help secure the honesty of the answers. However, four participants out of the 16 needed to be near their family member in the study due to hearing difficulties. All 16 participants voluntarily agreed to be interviewed and recorded using a digital recorder.

The interview instrument consisted of six open-ended questions for the eight AAT-D participants and nine open-ended questions for the primary family caregivers. I digitally recorded all 16 interviews, and the participants answered their questions in a session between 30 to 45 minutes. The interview duration was based upon the amount of detailed information was provided. A notebook was kept by my side where I made notes as cues to follow-up with additional questions and clarifications. Follow-up questions were used to address the research questions and to collect further data regarding the participants lived experiences with AAT-D. All 16 interviews were completed in its entirely the first time. At the end of each interview, I thanked the participants for participating in my study. I transcribed and coded each digital recording. The hard copies of the transcriptions and the written notes have been stored in a locked filing cabinet.

Data Analysis

I used a phenomenological method for this study and kept the focus on the phenomenon of the alpha-1 deficient participant and their primary caregiver's coping mechanisms and the lived experiences identified by each participant. For data analysis, I used the eight interview transcripts for the AAT-D and eight interview transcripts for the caregivers, which were then typed into a typed Microsoft Word table. Each interview response to the questions were read multiple times to formulate codes, emergent themes, and sub-themes. The table was helpful to me in methodically developing codes one at a time. I developed a legend at the bottom of each table explaining the symbol and color highlight and what theme they represented.

To formulate the codes, I highlighted partial comments associated with each interview question and labeled "C1" in parentheses to indicate as code 1. Each interview question included diary notes, which I placed on the table and highlighted in the same color to the matching code as an identifier. The same color highlight diary note would follow the partial comment which were included in the coding formulation. I performed a thematic analysis to obtain the emergent themes by reading and re-reading the identified codes for each of the interview questions for both the alpha-1 deficient participants and the primary caregivers. Validation for commonality between the four research questions was conducted and a careful analysis showed eight common themes associated with the four research questions. For example, five themes (shock, lack of awareness from the healthcare community and patient, misdiagnosis, took an average of five to seven years to properly get treated, and physical limitations) emerged in answers to RQ1. Themes for RQ2 had one similarity identified in RQ1 with an added aspect from the caregiver's perspective (shock and despair to see their loved one struggle). Finally, themes for RQ3 and RQ4 were the same (Keeping busy and family).

To determine an in-depth meaning of the themes, I referenced the constructs of the Biopsychosocial Theoretical Model, Bowen's Family Systems Theory, and the Family Systems Model of Illness (Engel, 1977; Bowen, 1974; Rolland, Emmanuel & Torke, 2017). In the Results section, I present the results, the themes developed along with the support from the participants' statements, and any detailed variances.

Triangulation

I achieved triangulation by asking each participant the same interview questions which allowed me to build a cohesive justification of themes (Creswell, 2014). I developed Microsoft Word tables for each interview question to sort the data and discover the most common themes. The identified common themes came from the participant's responses to the administered questions. The responses were analyzed for similarities and differences which helped to decrease bias. Moreover, the use of rich and thick descriptions from the shared experiences of the AAT-D and their caregiver helped to increase the validity of my study.

Evidence of Trustworthiness

The research findings for this qualitative study were verified to ensure the credibility, transferability, dependability, and confirmability. Morse, Barnett, Mayan, Olson, and Spiers (2002), posit that the use of verification strategies helps to ensure the reliability and validity within a qualitative study. This section presents the steps taken to demonstrate the evidence of trustworthiness. Ensuring the validity in my study, I discovered the most common and beneficial coping mechanisms used across both the AAT-D participant and their primary caregiver that helped manage their stressors most optimally on a daily basis. In addition, I used detailed descriptions of the shared experiences from the challenges between the AAT-D participants and their caregiver which helped to convey the results in a realistic manner. Moreover, the dependability and confirmability were accomplished through audit trails. For example, along with the verbatim transcripts, I kept detailed written information about the conducted study,

surrounding environment, whom I met with, and the degree of the explained results from the collected data helped to decrease the level of biased research, increasing the connection between the findings and the focus of the study.

Results

The study findings are presented in this following section where I address the four research questions:

RQ1: How do AAT-D patients describe living with their illness?

RQ2: How do the family caregivers who live with AAT-D patients describe their experiences?

RQ3: What are the common coping strategies among AAT-D patients?

RQ4: What are the common coping strategies among the family caregivers?

Data was collected from eight alpha-1 deficient participants and eight primary caregivers. In this section, there are eight emergent themes that I present, along with examples from participants. All the names of the participants associated with the quotes used within the study are kept confidential. The research questions were created to examine the experiences of being an Alpha-1 patient and a family caregiver for someone living with Alpha-1 deficiency, as well as the coping strategies used on a daily basis. All interviews were conducted from the privacy of their homes through the telephone and the participant's consent was acquired before the interview. The interview instrument consisted of six questions for the alpha-1 participants and nine for the primary caregivers that addressed the four research questions. Each participant's responses were coded to exhibit the relevant information.

The interview responses to question 1 for the alpha-1 participants addressed the date of their diagnosis, how long it took to be properly diagnosed, the challenges they may have experienced, and their overall perception of their diagnosis. The interview responses to question 2 addressed if there has been any individual or group counseling. The interview responses to question 3 addressed how they cope with their disease, such as meditation, prayer, family. The interview responses to question 4 addressed how they managed all the household duties and finances. The interview responses to question 5 addressed was what their overall experience living with alpha-1 has been. The interview responses to question 6 addressed was how they thought their primary caregiver was managing their illness. The interview questions 1, 4, and 5 addressed RQ1. The interview responses to questions 3 and 6 addressed RQ2 and RQ3. The interview responses to question 1 for the caregivers addressed their connection to the alpha-1 participant. The interview responses to question 2 addressed the challenges and positive aspects of being the primary caregiver.

The interview responses to question 3 addressed their experience of their family member being diagnosed with alpha-1 deficiency. The interview responses to question 4 addressed their experience as the caregiver. The interview responses to question 5 addressed the responsibilities of the household duties and finances. The interview responses to question 6 addressed if they have been to any individual or group counseling. The interview responses to question 7 addressed how they are feeling physically and psychologically. The interview responses to question 8 addressed how they cope with being the primary caregiver. The interview responses to question 9

addressed how they think their alpha-1 family member is managing their illness. The interview questions 1, 2, 3, 4, 5, 6, and 7 addresses RQ2. The interview questions 8 and 9 addresses RQ4.

RQ1. The first research question was: How do AAT-D patients describe living with their illness? I present the five themes that emerged from the participants' responses about how AAT-D patients describe living with their illness.

Theme 1: Shock

The first theme that emerged from RQ1 was: "shock." The one response that was common among the eight AAT-D participants was that they have never heard of such a disease and was surprised at the debilitating nature of the illness. Excerpts from the participants' responses to support this theme are presented below.

One participant, AAT-D 1 (male), noted "I was never sick before and then all of sudden I started getting consistent bouts of bronchitis…when I was diagnosed it was shock." AAT-5 (Female), commented, "You know my diagnosis was funny…I was never sick in my life until I turned 50 and was constantly sick…my diagnosis came as a shock." AAT-D 9 (Female), stated "It was when I was just turning 50…it came to me as a shock." AAT-D 15 (Male), described "…yeah it came to me as a complete shock."

Theme 2: Lack of Awareness

The second theme that emerged for RQ1 was: Lack of awareness of the disease from both the healthcare community and AAT-D patient. The participants shared their experiences on how the healthcare community was unaware of the disease and how to manage it. Alpha-1 deficiency appeared to be an unheard of illness.

One participant AAT-D 5 (Female), reported:

> The Radiologist suggested that in her whole time of practice no patient came back with a positive alpha-1 test…she had absolutely no information for me no direction, no brochures, no recommendations for a pulmonologist. She said was don't drink or smoke because I have never seen this.

AAT-D 7 (Female), commented "my doctor was very sad and upset and he could not find any literature on it…only that it was a fatal genetic lung disease…he said that he would try to find out about this disease."

AAT-D 11(Female), explained:

> I was getting foot surgery because of a fall but I could not shake the bronchitis, so the foot surgeon ordered a chest x-ray which revealed that I had COPD but I was too young to be diagnosed with that…so I did my research and brought it back to my primary physician and that is when he tested me for alpha-1 deficiency.

Theme 3: Misdiagnosis

The third theme that emerged from RQ1 was: misdiagnosis. The most common response from the eight AAT-D participants was that they were initially diagnosed with COPD/ Emphysema.

> AAT-D 3(Male) stated: I was surprisingly diagnosed with Emphysema with approximately half my lung function gone and I have never been a smoker… the PA asked the doctor did you check him for alpha-1?... the doctor leafed through my file and no joke said umm oops.

AAT-D 5 (Male) explained:

> …so we went to the doctor and they first thought I had RA…I just kept getting sicker and sicker and sicker…I stopped at the Mayo Clinic in Jacksonville and they ran a bunch of tests but did not run an alpha-1 test…they found I also had granulomatosis…however I was still short of breath and the doctors could not understand why I was short of breath…I then got sent to a gastroenterologist because my liver enzymes were high and they did not know why…it was there that they ran an alpha1 test which came back positive.

AAT-D 7 (Female) declared:

> I decided to see an allergy specialist because I was so sick all the time… all my life I was told that I had asthma… I went to see a new doctor and he had just got back from a conference that talked about a rare disease called Alpha-1 deficiency and he said that I looked like I was going to die so he decided to run an alpha-1 test… he looked sad and very upset and told me that there was no literature about this disease but only one article which said that it was a fatal genetic lung disease… he said he would do his best to find out more about it…

AAT-D 11(Female) commented:

> …I had bronchitis that I just could not shake…I was ordered a chest X-ray to be safe and that is when it said I had severe COPD. So my primary doctor told me that it is important to not smoke and how we needed to find out more about why my lungs are so bad…well I went home to look up an old reference Mayo Clinic guide from 1981 and that is where it said in a small paragraph under emphysema that adults ages 30-50 can have early onset emphysema due to alpha-1

> deficiency…so I brought this information back to my doctor where they tested me for it and sure enough that was what was going on…

AAT-D 13 (Female) explained:

> I had going through a few bouts of being misdiagnosed with bronchitis and so forth… I was 36 years old at the time and they told me I had COPD but in my 30s that did not explain anything to me at the time and just sent me to a pulmonologist… he did a test right off the bat and called me in and said that I have alpha-1 deficiency…

Theme 4: Lengthy Diagnosis Process

The fourth theme that emerged from RQ1 was: time of diagnosis was lengthy. The most common responses among the AAT-D participants was how it was a lengthy process to be appropriately diagnosed. AAT-D 1 (Male) declared "it took about two years to be properly diagnosed." AAT-D -3 (Male) suggested "it took about a year to be properly diagnosed." AAT-D 5(Male) claimed "…if they would have ran the test back in 2010 instead of 2014 I would have had more lung function…" AAT-D 7 (Female) commented "…it took him a whole year to get properly diagnosed." AAT-D 11(Female) said "…from the initial symptoms to get properly diagnosed was 17 years…" AAT-D 13 (Female) exclaimed "…it took a couple of years to be properly diagnosed…" AAT-D 15 (Male) declared "…oh gosh it took approximately five years to get properly diagnosed…"

RQ2. The second research question was: How do the family caregivers who live with AAT-D patients describe their experiences? I present one similar theme to RQ1

with an added aspect from the caregiver's perspective. The most common responses from the caregivers were shock and despair from watching their loved ones struggle.

Theme 5: Shock

The two themes that emerged for RQ2 was caregiver shock and despair in watching their family struggle with the disease. The participants shared their experiences they had in the initial diagnosis of their spouse and how it is a challenge to watch them struggle with the disease on a daily basis. Caregiver 2 (Female) commented "It was a shock obviously…" Caregiver 8 (male) stated "When she was initially diagnosed with it, I had never heard of it…" Caregiver 10 (Male) suggested "was a surprise." Caregiver 12 (Male) declared "…I don't know anything about oxygen and I never have even heard of it…" Caregiver 14 (Male) stated "Never heard of it before". Caregiver 16 (Female) claimed "I never heard of it before."

Theme 6: Despair

Theme six that emerged for RQ2 was despair watching their loved one go through the challenges of living with Alpha-1 deficiency on a daily basis. Caregiver 2 (Female) stated "…I have my responsibilities to do and I just do it. I love him When you are married for that many years, he is your buddy and I love."

Caregiver 4 (Female) explained:

> Well it is horrible and our life stopped. We have no life at all. We can't travel or do anything physical. We have become more or less homebodies and I hate it with every ounce of my being but I have no choice and I have been married to him for

> 42 years…it's a horrible disease and it is horrible to watch someone go through it and it's horrible being a part of it and all the things he has to do to stay alive…

Caregiver 8 (Male) stated "…there is a lot I help her with…it makes me feel better if I am able to be of assistance to her…"

Caregiver 12 (Male) declared:

> The challengers about being a caregiver. Um, with three kids probably trying to keep your wits together. Because it's another strain on me just to go and get the remote of the entertainment center instead of her getting up to get it. She needs me to get it, I have normal lungs. Just the little small things you know, it's definitely that middle challenge, by the end of the day when I'm tired and aggravated, not so much for the person, but for yourself, knowing that I'm tired and she needs that I'll get it or if the kids need something I'll get it, you know…

Theme 7: Keeping Busy

Theme seven, keeping busy emerged for both RQ3 and RQ4. The caregiver participants shared their experiences how maintaining a busy schedule is a way to cope with the illness. The majority of the participants' responses to coping was attempting to maintain a busy lifestyle through work and/or activities. AAT-D 5 (Female) stated "keeping busy." AAT-D 7 (Female) commented "I like to stay real busy. I like to swim and work is so busy…Yeah I try to stay real busy so I do not think about it." AAT-D 9 (Female) explained "I am trying to just live a normal life like not try to make that the center of everything try to go places and do things…" AAT-D 13 (Female) suggested "I work as much as possible and stay on the go." AAT-D 15 (Male) declared "My farm, I

raise cattle for beef." Caregiver 4 (Female) noted I just do what I have to do…" Caregiver 8 (Male) commented "I have a number of projects that I am working on, welding, carpentry, stuff life that I manage to fill my days." Caregiver10 (Male) explained I do walk…I walk every day for about 2 miles with a couple of friends and we have a good time…I ride my Harley…" Caregiver 12 (Male) suggested "I have some hobbies, golf is one of my releases…wood working…and yard work…"

Theme 8: Family

Theme eight, family emerged for both RQ3 and RQ4. The participants shared their experiences on how family helps them coping with the illness on a daily basis. AAT-1 (Male) stated "My wife has probably been the key to my coping. She has been phenomenal, and we have been married for 58 years…" AAT-D 11 (Female) explained "…what motivates me the most is the kids. Having young kids at home and wanting to be around for them…" Caregiver 2 (Female) declared "…we do everything together…you cope and move forward." Caregiver 6 (Male) suggested "…she is my wife and that is all I need…do not feel like I need to cope." Caregiver 14 (Male) declared "No I just cope, as long as I get to hold her, I am just fine."

Summary

The purpose of this study was to study the lived experiences of Alpha-1 deficient participants and their primary caregiver, as well as the coping mechanisms used by both the alpha-1 deficient participant and their family caregiver. In chapter 4, I presented a review of the data collected and how the data was managed and analyzed. The 16 participants declared that they understood the Informed Consent before the start of the

interview. The responses from the interviews examined the lived experiences of alpha-1 participants and their primary family caregiver and how they coped with the illness on a daily basis. A descriptive phenomenological method was used, and the appropriate steps were incorporated to ensure that the lived experience among the alpha-1 community was expressed accurately. In response RQ1, most of alpha-1 participants commented how "shock" was the initial feeling to being diagnosed with the illness. They had never heard of the illness and were surprised at its debilitating nature. Lack of awareness was another common response represented by both the alpha-1 participant and healthcare community.

The participants shared their experiences on how the healthcare community was unaware of the disease and how to manage it. Additionally, the majority of the alpha-1 participants explained how they were initially misdiagnosed with COPD/Emphysema. Lastly, for RQ1 was the lengthy time for proper diagnosis. The most common responses from the participants was how it took time and the correct tests to be appropriately diagnosed and placed on the correct plan of action. Reponses to RQ2 revealed caregiver shock and despair in watching their loved one struggle with alpha-1 deficiency daily. The participants shared their experiences they had in the initial diagnosis of their spouse and how it is a challenge to watch them struggle with the disease. Responses from the participants to RQ3 and RQ4 demonstrated that keeping busy and family helps the caregivers cope with the illness. Chapter 5 presents the interpretation of the research findings, limitations, recommendations, social change implications, and conclusion.

Chapter 5: Discussion, Conclusions, and Recommendations

Introduction

The purpose of the present study was to examine the lived experiences of Alpha-1 deficient patients and their family caregiver, as well as the coping mechanisms used by patients and caregivers. I used the descriptive phenomenological method to gain a deeper insight into the shared experiences of the alpha-1 patients and their primary family caregiver. Specifically, I used this method to gather individual perceptions from both the alpha-1 patient and their family caregiver, as well as shared perceptions regarding managing alpha-1 deficiency and the challenges participants have faced from the initial diagnosis to the present.

In reviewing previous literature, I found a lack of information on the lived experiences of alpha-1 deficient patients and their family caregiver related to the coping mechanisms used to manage the stressors. Previous researchers have mainly focused on the uncertainty and complexity of how COPD can be stressful for not only the patient but also the patient's family caregiver (Stenzel et al., 2015). Moreover, previous research has shown that the interaction between the COPD patient and the patient's family caregiver affects how both parties cope with the disease's physical and psychological demands (Barnett, 2004; de Voogd et al., 2011; Figuerido et al., 2014., Hoth et al., 2015).

However, what is unknown, according to my review of the literature, is the complexity of AAT-D-associated COPD and the specific disease-related anxieties of both the alpha-1 deficient patient and the patient's family caregiver, as well as the coping mechanisms used by both. Thus, I conducted this study to better understand the lived

experiences of alpha-1 deficient patients and their family caregivers and how they manage their stressors daily. I formulated four research questions to address the specific goals for this study:

RQ1. How do AAT-D patients describe living with their illness?

RQ2. How do the family caregivers who live with AAT-D patients describe their experiences?

RQ3. What are the common coping strategies among AAT-D patients?

RQ4. What are the common coping strategies among the family caregivers?

I administered six interview questions to the eight alpha-1 deficient participants and nine interview questions to the eight primary family caregiver participants, several themes emerged from participants' responses. The key findings for RQ1 were that the alpha-1 deficient participants declared how shock, lack of awareness, misdiagnosis, and the process that it took to properly be diagnosed were all challenges they experienced. They reported that (a) they have never heard of such a debilitating illness before being diagnosed, (b) neither they nor the health care community were familiar with the disease, (c) they were initially diagnosed with COPD, and (d) being appropriately diagnosed and managed was a lengthy process. The main findings for RQ2 were caregiver shock and despair in watching their family member struggle with the illness. The key findings for RQ3 and RQ4 were coping by keeping busy and family. The participants shared how maintaining a busy schedule and family support system were critical elements in coping with the challenges of alpha-1 deficiency.

Interpretation of the Findings

Alpha-1 antitrypsin deficiency was discovered in 1963 by Laurell and Erisksson (Anariba, 2017) who found that five of the 1,500 serum protein electrophoreses (SPEP) submitted to their laboratory in Sweden had the absence of the band of alpha-1 protein. Moreover, Laurell and Eriksson discovered that three of the five patients were diagnosed with emphysema at a young age, leading them to develop the cardinal clinical features of AAT-deficiency with absence of the AAT and early onset emphysema (Anariba, 2017). For example, people whose bodies do not produce enough of the protein AAT are more likely to develop emphysema typically between 30-50 years old. Physicians should suspect AAT-deficiency if the patient develops emphysema at 45 years of age or younger; develops emphysema without having any recognized risk factors, such as smoking or exposure to chemical fumes or pollutants over a long period of time; x-rays show less density in the lungs than normal; or the patient has unexplained liver disease, necrotizing panniculitis, bronchectasis without any cause, or asthma that does not completely resolve after the use of regular breathing treatments (Fahy et al., 2014).

COPD is an umbrella term used to describe progressive lung disease including emphysema, chronic bronchitis, asthma, and some forms of bronchiectasis (COPD Foundation, 2017).AAT-D is a relatively common genetic disorder that is often undiagnosed; people with AAT-D are predisposed to COPD and liver disease (Anariba, 2017). Some previous researchers studying COPD have examined the interactions between family caregivers and their patients. For example, researchers have focused on the coping strategies, such as problem-solving strategies, emotional-cognitive strategies,

and stress management strategies, used by COPD patients and family caregivers (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015) COPD patients' experiences (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015); how dyspnea in COPD patients may increase anxiety levels (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015); how the severity of the symptoms may impact alpha-1 patients' quality of life (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015); and how illness uncertainty may increase anxiety levels (Barnett, 2004; de Voogd et al., 2011; Figueirdo et al., 2014; Hoth et al., 2015). The results of the present study showed the complexities of alpha-1 deficiency and the challenges the alpha-1 community face daily. My findings indicated that the alpha-1 deficient participants were previously unaware of the disease and were often misdiagnosed. The time from the initial symptoms to the appropriate diagnosis proved to be a lengthy process. Previous researchers have not reported these findings, according to my review of the literature.

The results of this study showed that the both the alpha-1 patient and the patient's primary caregiver used a variety of coping strategies to manage the stressors of the illness on a daily basis. Coping strategies such as staying busy with hobbies and work and having a supportive family dynamic were the key components in managing the stressors of alpha-1 deficiency for the majority of the participants in this study. I identified eight themes from the participants' answers regarding living with alpha-1 deficiency on a daily basis: shock, lack of awareness from both the Alpha-1 health care community and alpha-1 deficient patients, misdiagnosis, lengthy diagnosis, caregiver shock, despair in watching

their loved ones struggle with the disease, as well as keeping busy and family relationships to help cope with the obstacles of alpha-1 deficiency on a daily basis.

The eight alpha-1 deficient participants reported that they had never heard of the disease before being diagnosed with it and were surprised at the debilitating nature of the illness. Several of the alpha-1 deficient participants also mentioned that they were never sick until the abrupt symptoms, such as difficulty breathing while performing their normal activities or continuous bouts of bronchitis and pneumonia. In addition, all eight of the alpha-1 deficient participants reported that their doctors or physician assistants stated that they were unaware of the disease and how to properly manage it. Previous research has shown that there is a high rate of undetected or misdiagnosed AAT-D and that it takes an average of seven to eight years to be properly diagnosed (Campos et al., 2005). The present study confirmed those challenges of AAT-D with five out of the eight alpha-1 participants stating they were misdiagnosed with COPD, bronchitis, asthma, or some other illness. The participants also reported that it took anywhere from a year to 12 years to be properly diagnosed. Previous researchers have examined the effect that the type and quality of a relationship has on the COPD participants' overall psychological state; their research suggests that having more support from their partners helped to decrease patients' stress levels while having less support resulted in higher stress levels (DiNiCola et al., 2013). In the current study I focused on AAT-D participants and found that all eight alpha-1 participants were managing their psychological stressors with the positive support from their family.

In the present study, seven out of the eight caregiver participants experienced shock when their spouse was diagnosed with alpha-1 deficiency. Only one out of the eight caregiver participants was not surprised with the diagnosis because her husband's brother was diagnosed earlier with the genetic illness. Prior related research has found that pervasive dyspnea can be frightening to both the patient and the caregiver which can lead to high stress levels (Lee et al., 2010). The current study suggested that caregivers exhibited despair in watching their loved ones struggle with the disease. All eight participants stated that it is a terrible disease which makes it difficult for them to see their spouses manage the challenges of alpha-1 deficiency. However, the degree of the alpha-1 participants' symptoms, how involved the caregivers were to their spouse, and the depth of their relationship with their spouse all played a role on how they managed their stressors.

Previous research has shown that caregivers for COPD patients that observe their family member with COPD experiencing progressive dyspnea, physical disabilities, altered cognition, memory disturbances, and behavioral changes will further strain the family caregiver's overall life (Lee et al., 2010). Caregiver burden is described as the distress that caregivers feel as the result of providing care which is specific to the complexity and degree of the disease care (Rha et al., 2015). However, the present study found that seven out of the eight caregivers did not feel like it was a challenge to take care of their spouses. They expressed that is a natural aspect to take care of the spouse. Several of the participants were married for more than 20 years. Only one out of the eight participants found every aspect of the disease while taking care of their spouse a

horrible experience and if she would have known her spouse would be as sick prior to marriage, she would have never married him.

The biopsychosocial model (Engel, 1977) was used in the present study to examine how experienced biological factors, such as genetics and dyspnea could be the possible reason for the anxiety and uncertainty levels of the participants. All eight of the alpha-1 participants reported that they have and continue to feel anxious when their breathing difficulty levels increase. Fifteen out of the 16 participants felt anxious during the initial diagnosis because of the unknown genetic characteristics of the disease. Previous research has shown that the perceived uncertainty about the current symptoms and future prognosis is the challenging part of living with a chronic illness. Hoth et al. (2015) analyzed illness uncertainty, such as ambiguity (physical symptoms) and complexity (treatment and healthcare system) among alpha-1 deficient associated COPD patients and found that uncertainty about physical symptoms (ambiguity) induced depression, anxiety, breathlessness while decreasing alpha-1 deficient patients' quality of life.

In the present study, only one out of the eight couples did not feel anxious during the diagnosis because of the known properties of the illness from prior family experiences. The current study found that keeping a busy schedule, such as working within the alpha-1 community and hobbies helped the 15 out of 16 participants to cope with the stressors in their daily lives. The Bowen Family Systems Theory (2017) and the Family System of Illness (Rolland et al., 2017) were used in the current study which showed that the strength of the family interaction was an intricate part in how both the

alpha-1 deficient and caregiver participants coped with the illness. Fifteen out of 16 participants suggested that it was the family unit that kept them going in life.

Limitations of the Study

I intended to recruit eight alpha-1 deficient participants who were diagnosed with alpha-1 deficiency that did not currently smoke and eight primary family caregivers who lived in the southern portion of the United States through a mass email sent out to the alpha-1 community describing my study from the Alpha-1 community's associative director. The total process time for recruiting participants was approximately four months and at the half-way point I placed a reminder email for the associative director of the Alpha-1 community about the study.

A limitation of the study was three out the eight couples could hear each other's interview questions because of hearing difficulties which may have caused them to withhold their true answers to the interview questions. Another limitation was that the participants were from different geographic areas which may have provided a variety of perceptive experiences. Although the study had a few limitations, the background information, framework of the study, as well as the detailed description of the study may be used to further investigate and compare future studies using the same populations.

Recommendations

The present study increased the understanding of the lived experiences and coping mechanisms of alpha-1 deficient and their primary caregivers. However, future research may further investigate the effects for the patient and the caregiver of participating in a support group. Does the support group reduce feelings of burden on the caregiver? It also

may provide a normalizing effect for both caregiver and patient. Another recommendation would be to examine the siblings who are not considered an alpha and how it plays a role in their psychological state on a daily basis. In particular, the future study could analyze if they have "survivor guilt" and the aspects that impact their lives. I also recommend conducting a study on the depth of alpha-1 awareness across the healthcare community and how it can be improved. How can the alpha-1 community bring more education and awareness to the healthcare personnel in order to achieve a quicker and more accurate diagnosis? Another recommendation is to examine how AAT-D patients identify with the disease. Does the illness identify them? In addition, does gender play a role in how AAT-D patients manage their stressors or is there a biological aspect that exposes differences in the degree of physical and/or psychological symptoms?

Implications

Many opportunities exist to make a positive change across the alpha-1 community. The findings from the current study can positively add to the current body of literature used to understand the lived experiences of alpha-1 deficient participants and their primary caregiver, as well as their coping mechanisms. It was apparent that shock, lack of awareness, misdiagnosis, and the lengthy time of diagnosis proved to be a few of the challenges among the alpha-1 deficient participants and caregiver shock. Additionally, despair in watching their loved ones struggle on a daily basis was a central issue from the caregivers. However, maintaining a busy schedule and engaging in a positive family environment showed to be positive coping mechanisms for both the alpha-1 deficient participants and their caregivers.

Conclusion

The present descriptive phenomenological study focused on the phenomenon of alpha-1 deficiency and investigated the lived experiences of the alpha-1 deficient participants and their caregivers, as well as their coping mechanisms. The experiences of the alpha-1 deficient participants and their caregivers were expressed through recorded phone interviews and provided accurate descriptions that established significant themes that led to a better understanding of the phenomenon of alpha-1 deficient participants and their primary caregivers while gaining a deeper insight into the coping mechanism used.

The current study extends the previous knowledge of the lived experiences of alpha-1 deficient participants and their primary caregivers, as well as the coping strategies used. As a result of this study, the phenomenon of the complexities of alpha-1 deficiency and the life experiences of alpha-1 deficient participants and their caregivers were shown to be challenges. However, being active within the alpha-1 community, engaging in hobbies, and having a supportive family dynamic proved to be positive aspects in coping with the stressors.

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Appendix A: Letter Sent to Participant

Date

Name of Participant
Address

Dear (Name),

My name is Nicolette Bruscino and I am a doctoral candidate at Walden University. I am conducting dissertation research on the complexity and challenges AAT-D places on both the AAT-D individual and their family caregiver, as well as the coping mechanisms used from both the AAT-D individual and their family caregiver on a daily basis. There are a vast number of studies detailing the difficulties of COPD and how to better manage the illness, as well as the obstacles faced on the primary caregiver to COPD individuals. What is not known, however, are the specific challenges of AAT-D associated COPD individuals and their family caregivers, as well as the most optimum coping strategies used to better manage the stressors from both the AAT-D individual and their family caregiver on a daily basis.

I realize that your time is important to you and I appreciate your consideration to participate in this study. In order to understand your experience we need to meet for one interview for approximately one hour. The interview can be held at a location of your choosing and will not require you to do anything you don't feel comfortable doing. The interview is designed to simply get to know you and learn about your experience of being an AAT-D individual or the sole family caregiver of the AAT-D individual. All information gathered during our interview will be kept strictly confidential.

Please contact me at your earliest convenience to schedule a date and time that we can meet. My telephone number is XXX. You can also email me at XXX. I look forward to hearing from you.

Nicolette Bruscino
Doctoral Candidate
Walden University

Appendix B: Interview Questions AAT-D Participants

1. Tell me about your diagnosis.
    A. When was the date of your diagnosis?
    B. How long did it take to get properly diagnosed?
    C. Where there any challenges of being diagnosed?
    D. What is your personal outlook on your diagnosis?
2. Tell me about any individual or group counseling you have had.
3. How do you cope with your disease? For example, some people use distractions, prayer, or meditation.?
4. How do you manage all the household duties and finances?
5. What has been your experience living with AAT-D?

Family Caregivers

1. What is your connection to the AAT-D participant? Are you the primary caregiver?
2. Tell me about the challenges of being a caregiver. What are the positive aspects of being a caregiver?
3. Tell me about your experience about your family member being diagnosed with AAT-D.
4. Tell me about your experience as a caregiver.
5. Tell me about the responsibilities of the household duties and finances.
6. Tell me about any individual or group counseling you have had.
7. Tell me how you are feeling physically and psychologically.
8. How do you cope being the primary caregiver?