Engaging Individuals with Lived Experiences: Identifying the Needs and Priorities of Informal Caregivers for People Living with Systemic Sclerosis

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April 2020

A thesis submitted to McGill University in partial fulfillment of the requirements of the degree of Doctor in Clinical Psychology

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Abstract

People with rare disorders often require support from an informal caregiver. Among individuals with a rare disorder, the caregiver role is typically fulfilled by a family member, friend, or a loved one. Caregivers of people with rare disorders face the same challenges as all caregivers (e.g., balancing multiple roles), as well as additional difficulties related to a lack of information and available resources in the rare disease context. In Canada, the majority of caregivers to individuals with a rare disorder report feeling unsupported by Canada's healthcare system and that they are negatively impacted emotionally and financially due to their caregiver role. The objective of this research was to (1) identify studies that have examined the potential benefits and feasibility of implementing psychosocial interventions among caregivers to a person with a rare disorder, (2) conduct group discussions among caregivers of people living with scleroderma (SSc), a rare, chronic autoimmune disorder, to develop survey items focused on the challenges experienced and preferences for support services, and (3) evaluate the importance of identified challenges and support service preferences through an online survey of caregivers to persons with SSc.

Study one was a scoping review that identified (1) perceived and tested benefits of participating in psychosocial interventions for caregivers of a person with a rare disease and (2) barriers and facilitators of establishing and maintaining these interventions. The findings of the review suggested that psychosocial interventions for caregivers to a loved one with a rare disease may help to decrease caregiver's feelings of isolation, stress, and burden. Establishing and maintaining these interventions, however, may be challenging over time, given the rarity of the disorders.

Study two involved conducting a series of structured group discussions with informal caregivers of persons with SSc to determine the challenges faced by these individuals and their preferences for support services that could be developed. This work also aimed to identify stakeholder priorities through engaging people with lived experience in the research design process. The group discussions resulted in

the identification of 61 unique challenges and 18 unique support services, which provided items for use in a survey to sample a larger group of caregivers.

Study three used an online survey to evaluate the importance of challenges experienced by an international sample of informal caregivers to persons with SSc and identified priorities for support services. Caregivers rated the importance of various challenges faced through their role and the likelihood of using different types of support services if they were to be developed and available. Caregivers reported that supporting their care recipient with emotional difficulties and physical discomfort were the most important challenges they faced. They indicated that they would be most likely to use informational resources that are provided soon after diagnosis in online or hard-copy format. They were less interested in support services delivered in groups of caregivers.

In sum, psychosocial interventions are perceived as a helpful resource among caregivers to persons with a rare disease. Adapting interventions to meet the specific needs of caregivers was a commonly identified facilitator for initiating and maintaining these interventions. Tailoring existing interventions to address challenges identified as important by caregivers to a person with SSc, such as the difficulty supporting their care recipient's emotional challenges and physical discomfort, could address the needs and preferences identified by caregivers to persons with SSc.

Résumé

Les personnes atteintes de maladies rares ont souvent besoin du soutien d'un aidant naturel. Chez les personnes atteintes d'une maladie rare, le rôle du soignant est généralement joué par un membre de la famille, un ami ou un proche. Les aidants naturels des personnes atteintes de maladies rares sont confrontés aux mêmes défis que tous les soignants, ainsi qu'à des difficultés supplémentaires liées au manque d'informations et de ressources disponibles. Au Canada, la majorité de ces aidants déclarent ne pas se sentir soutenus par le système de santé canadien et se sentir lésés financièrement et émotionnellement. Les objectifs de cette recherche étaient (1) d'identifier les études qui ont examiné les avantages potentiels et la faisabilité de la mise en œuvre d'interventions psychosociales auprès des aidants de personnes atteintes d'une maladie rare, (2) de mener des discussions de groupe avec des aidants de personnes atteintes de sclérodermie (ScS), une maladie auto-immune chronique rare, afin d'élaborer les items d'un sondage axé sur les défis rencontrés et les préférences en matière de services de soutien, et (3) d'évaluer l'importance des défis et préférences par le biais d'une enquête en ligne auprès des aidants de personnes atteintes de ScS.

La première étude était une étude de portée qui a permis d'identifier (1) les avantages perçus et testés de la participation à des interventions psychosociales pour les aidants de personnes atteintes d'une maladie rare et (2) les obstacles et les stratégies liés à la mise en place et au maintien de ces interventions. Les résultats ont suggéré que les interventions psychosociales pour les aidants d'un proche atteint d'une maladie rare peuvent contribuer à réduire les sentiments d'isolement, de stress et de fardeau. La mise en place et le maintien de ces interventions peuvent toutefois s'avérer difficiles au fil du temps, compte tenu de la rareté des maladies.

La deuxième étude a consisté à mener des groupes de discussion structurée avec des aidants naturels de personnes atteintes de ScS, afin de déterminer les défis auxquels ces personnes sont confrontées et leurs préférences quant aux services de soutien qui pourraient être élaborés. Les discussions de groupe ont permis d'identifier 61 défis et 18 services de soutien uniques, qui ont ensuite été consolidés dans un sondage envoyé

à un large échantillon d’aidants.

La troisième étude a utilisé ce sondage en ligne pour évaluer l'importance des défis rencontrés par les aidants naturels de personnes atteintes de ScS de partout à travers le monde, et a identifié les priorités en matière de services de soutien. Les aidants ont évalué l'importance des différents défis rencontrés dans le cadre de leur rôle et la probabilité d'utiliser différents types de services de soutien. Les aidants ont indiqué que le soutien à un proche souffrant de difficultés émotionnelles et de malaises physiques étaient les défis les plus importants auxquels ils étaient confrontés. Ils ont indiqué qu'ils seraient plus susceptibles d'utiliser des ressources d'information fournies peu de temps après le diagnostic, en ligne ou sur papier. Ils étaient moins intéressés par les services de soutien fournis par l’entremise de groupes d'aidants.

En résumé, les interventions psychosociales sont perçues comme une ressource utile par les aidants de personnes atteintes d'une maladie rare. L'adaptation des interventions ayant pour objectif de répondre aux besoins spécifiques des aidants a été identifié comme une stratégie importante pour la mise en place et le maintien de ces interventions. L'adaptation des interventions existantes ayant pour but de répondre aux défis identifiés comme importants par les aidants de personnes atteinte de ScS, tels que la difficulté à soutenir un proche souffrant de difficultés émotionnelles et de malaises physiques, pourrait répondre aux besoins et aux préférences identifiés par les aidants de personnes atteintes de ScS.

Acknowledgements

I would like to extend my deepest gratitude to my supervisor, Dr. Brett Thombs for his direction, feedback, flexibility and support throughout my doctoral degree. Thank you for your guidance and input during the conception, development, and completion of my doctoral thesis and the projects outside of this work. I am very appreciative of the opportunities you have provided me with as they will shape my career in the field of mental health. Your support throughout my graduate studies has been unwavering.

I would also like to thank all of the co-authors and my thesis committee members (Drs. David Vachon and Irv Binik) for their collaboration and support throughout the development of the manuscripts included in this thesis. Your input initiated important discussions while improving the quality of my work. I would also like to thank the thesis examiners who are taking the time out of their busy schedules to provide input and expertise on this work.

I would like to extend my sincere appreciation to the current and former members of Dr. Thombs Research Team, especially Andrea Carboni-Jiménez and Mara Cañedo-Ayala who have supported each study within my thesis. Without their assistance and collaboration this work would not have been possible.

In addition, I would like to acknowledge personal funding support from the Fonds de Recherche du Québec-Santé Doctoral Award and the Canadian Institutes of Health Research (Vanier Canada Graduate Scholarship and Health Systems Impact Fellowship).

To my amazing cohort – Paige Ethridge, Mallory Frayn, Paul Grunberg, Emily Moore, Sarah Peters, and Sarah Schell, and our honorary cohort member, Ali Farrell-Reeves, while I am not sure how we made it this far, I'm confident that bringing candy to afternoon classes was definitely a contributing factor. Thank you all for the never-ending support, the laughs, and the collaborative efforts to get through every stage of classes, clinical work, and research, together. There is no one else I can imagine being surrounded by these past 5 years. My graduate school experience was also heavily supported by Chantale Bousquet, many faculty members in the department, clinical supervisors, and upper year students who helped guide the PhD process as role models (and endless sources of knowledge and support).

Finally, I would like to thank Aaron Gartner, Taya Gartner, Megan Smith, Taren Byrne, Cassi Connolly, Lindsay Deveaux, Stephanie Gumuchian, Sarah Howorth, Lisa Jewett, Melissa Nikkel, Jacqueline Oliver, Erin van der Hoff, and Jenn Wira. Each of you have helped me make some of my best memories that took place simultaneously with my schooling. Thank you each for your positivity, love, and encouragement through my degree(s).

Contribution to Original Knowledge

The present doctoral thesis provides original contributions to the literature that increase the understanding of the experience caring for a loved one with a rare disorder. To date, most research focuses on the caregiving role to persons with common medical conditions (e.g., cancer, dementia) and very little research has considered the experience of caring for a loved one with a rare disorder, despite the unique challenges that are faced as a result of the rarity of such diseases (e.g., a lack of available resources and information). Each article in the current thesis provides a unique and novel contribution to the literature with the goal of improving the understanding of caring for an individual with a rare disorder.

Study 1 provided an overview of interventions that have been tested among caregivers to individuals with a rare disorder. Interventions that have been tested among this populations were absent from the literature, as was an understanding of the benefits and feasibility of implementing interventions among caregivers of persons with a rare disorder. This review identified the interventions that have been implemented and tested in a rare disease context to serve as a basis for the development of future interventions targeting caregivers to persons with a rare disorder. This work also identified gaps in the literature and provided directions for future research, such as highlighting the need to increase the sustainability of interventions and adapt interventions based on the unique needs of a rare disease community.

Study 2 provides an original contribution to the literature by focusing on the experience of caring for a loved one with scleroderma (systemic sclerosis, SSc), a rare chronic autoimmune disorder. Prior to the current thesis, only one previous study had included caregivers of persons with SSc, and this study had conducted qualitative analyses to identify common concerns among 13 caregivers. Study 2 involved generating a comprehensive list of the specific challenges faced by caregivers, and the support services that caregivers would potentially use if they became available. This study also provided a novel

contribution to the literature by integrating people with lived experience into the development of survey items through use of the nominal group technique.

Study 3 is the first study of caregivers to persons with SSc that presents quantitative data. A survey that included 202 caregivers from Canada, US, France, and Australia was conducted and serves as a needs assessment to understand the preferences that caregivers to persons with SSc have for the development of support services. Caregivers reported that supporting their care recipient with emotional and physical difficulties were the most important challenges they face. Informational resources provided soon after diagnosis in online or hard-copy format were reported as being most likely to used, if available. This dissertation fills an important need in the SSc community and provides a novel understanding of the interventions that have been tested among caregivers to persons with a rare disorder that could be adapted for use among caregivers to persons with SSc in order to decrease their reported challenges and fulfil their preferences for support services.

Contribution of Authors

The three manuscripts included in the present thesis are original research. I am the first author on all three papers, as I contributed to the conception and design of each paper; conducted the acquisition, analysis, and interpretation of data; completed the original draft of each paper; incorporated critical feedback from co-authors and reviewers; and prepared and submitted the final draft of each paper. The first manuscript is co-authored by Andrea Carboni-Jiménez, Mara Cañedo-Ayala, Kimberly A. Turner, Matthew Chiovitti, Alexander W. Levis, and Dr. Brett D. Thombs. Ms. Carboni-Jiménez, Ms. Cañedo-Ayala, and Ms. Turner each contributed to the acquisition, analysis, and interpretation of data; revising the paper; and final approval of the manuscript. Mr. Chiovitti and Mr. Levis contributed to the acquisition of data; revising the paper; and final approval of the manuscript. Dr. Thombs contributed to the conception and design of the paper; the analysis and interpretation of data; supporting me in drafting and revising the paper; and final approval of the published version.

The second manuscript is co-authored by Mara Cañedo-Ayala, Kimberly A. Turner, Stephanie T. Gumuchian, Dr. Vanessa L. Malcarne, Dr. Mariët Hagedoorn, Dr. Brett D. Thombs, and the Scleroderma Caregiver Advisory Team. Ms. Cañedo-Ayala contributed to the analysis and interpretation of data; drafting the manuscript; revising the paper, and final approval of the published version. Ms. Turner contributed to the analysis and interpretation of data; revising the paper; and final approval of the published version. Ms. Gumuchian contributed to the acquisition of data; the analysis and interpretation of data; revising the paper; and final approval of the published version. Dr. Malcarne, Dr. Hagedoorn, and the Scleroderma Caregiver Advisory Team contributed to the analysis and interpretation of data; revising the paper; and final approval of the published version. Dr. Thombs contributed to the conception and design of the paper; the acquisition of data; the analysis and interpretation of data; supporting me in drafting and revising the paper; and final approval of the published version.

The third manuscript is co-authored by Mara Cañedo-Ayala, Andrea Carboni-Jiménez, Kimberly

A. Turner, Marie-Eve Carrier, Julie Cumin, Dr. Vanessa L. Malcarne, Dr. Mariët Hagedoorn, Dr. Brett D. Thombs and the Scleroderma Caregiver Advisory Committee. Ms. Cañedo-Ayala, Ms. Carboni-Jiménez, Ms. Turner, Ms. Carrier, Ms. Cumin, Dr. Malcarne, Dr. Hagedoorn, and the Scleroderma Caregiver Advisory Team contributed to the acquisition of data; revising the paper; and final approval of the published version. Dr. Thombs contributed to the conception and design of the paper; the acquisition of data; the analysis and interpretation of data; supporting me in drafting and revising the paper; and final approval of the published version.

Chapter 1

General Introduction

Literature Review

Rare Disorders

Approximately 400 million individuals in the world are living with a rare disease (Global Genes, 2019), including 1 in 12 Canadians (Canadian Organization for Rare Disorders, 2015). A disease is considered rare if it affects fewer than one in 2000 people (Orphanet, 2012) and while the prevalence of any given rare condition is low, cumulatively, there are more than 7,000 rare diseases (Canadian Organization for Rare Disorders, 2015). As compared to diseases that are more prevalent (i.e., "common chronic diseases"), people living with a rare disease experience substantially greater reductions in quality of life (Bogart & Irvin, 2017).

In Canada, rare diseases account for 4.6% of total years of life lost in society. Comparatively, the percentage of total years of life lost most common diseases is substantially lower; for example, the total years of life lost is 2.6% from diabetes and approximately 1.4% from infectious diseases (pre-COVID-19) (Standing Committee on Health, 2019). This demonstrates the disproportionately high impact that rare conditions have on the health of Canadians (Standing Committee on Health, 2019). In 2018, Canada's House of Commons Standing Committee on Health commissioned a review of the barriers faced by Canadians with a rare disease. That report highlighted barriers to diagnostic tests for rare disorders, difficulties obtaining medication, and financial strain incurred by patients, their families, and the healthcare system (Standing Committee on Health, 2019). Attempts to develop and test new, more affordable, and more effective medications are complicated by the relatively small number of patients with a given disease, limiting the ability to conduct clinical trials. These barriers are in

line with literature reviews reporting unique challenges that are faced by individuals living with a rare disorder (Adams, Miller, & Grady, 2016), including economic, emotional, social, and physical consequences of coping with a rare disease (Adams et al., 2016; Shire Report, 2013).

In addition to direct health implications, living with a rare disorder can pose important daily challenges and can be socially isolating. The difficulties associated with a rare disorder often begin prior to a formal diagnosis, as receiving an accurate diagnosis can take several years (Adams, Miller, & Grady, 2016). During the process of searching for an accurate diagnosis, many individuals describe feeling hopeful at the chance of obtaining a medical explanation for their symptoms, while simultaneously feeling fearful, uncertain and frustrated due to a series of misdiagnoses occurring over an extended time period (Shire Report, 2013). Adverse experiences with the health care system are common in the context of rare disorders, and negative experiences often continue as patient expectations and needs go unmet. Non-specialist health professionals, who are the point of entry into the system, typically, are not knowledgeable about rare diseases, competent to diagnose them, or certain as to when a specialist should be involved (Budych, Helms, & Schultz, 2012). Many individuals living with a rare condition have recounted their experiences of being misdiagnosed, undergoing unnecessary treatments, and being referred to psychological services due to health professionals suspecting somatic symptom disorder (Kole & Faurisson, 2009), prior to receiving the correct diagnosis. Personal accounts of over 12,000 people living with rare diseases describe an overwhelming lack of knowledge about rare disorders in society and in the health care system, resulting in diagnostic mistakes and delays, and a lack of high quality treatment (Kole & Faurisson, 2009; Molster et al., 2016). Once a diagnosis of a rare disorder is confirmed, prognosis is uncertain, treatments may be unavailable or expensive, and support resources are often inaccessible or unaffordable (Adams et al., 2016).

As a result, rare diseases significantly impact the health and lifespan of people living with a rare disorder as well as their family (Shire Report, 2013), and many people with a rare disease and their family members experience depression, anxiety, stress, isolation, or worry based on an uncertain future, a lack of information, and a lack of treatment options (Shire Report, 2013).

Informal Caregivers in the Context of Rare Disorders

Given the complex and burdensome nature of a rare disease, many persons diagnosed with a rare disease require some level of care (Adams, Miller, & Grady, 2016). In rare diseases, the caregiver role is often fulfilled by a family member or friend on an informal basis. Informal caregivers are caregivers who typically do not receive training and who are unpaid for the role (National Alliance for Caregiving, 2015; Reinhard, Given, Petlick, & Bemis, 2008). Among caregivers to a loved one with a rare disease, 50% of a national sample in the US reported feeling they had no choice but to take on the caregiving role (National Alliance for Caregiving, 2018). Informal caregivers are heavily relied upon for rare diseases, given the lack of disease-specific resources available (Adams et al., 2016).

Informal caregiving can be a rewarding role, with positive aspects including increased closeness with the care recipient and pride from understanding the care recipient's rare disorder (National Alliance for Caregiving, 2018; Hilgeman, Allen, DeCoster, & Burgio, 2007). Often, however, caregiving is perceived as demanding, complex and stressful, with just one in 5 caregivers in Canada reporting coping 'very well' (The Change Foundation, 2019). From the onset of symptoms, family members or friends may immediately begin accompanying a care recipient to numerous appointments, facilitating transportation, helping with instrumental activities of daily living (e.g., housework, grocery shopping), and advocating to professionals on

behalf of the care recipient (National Alliance for Caregiving, 2015). In addition to facing challenges traditionally associated with caregiving such as balancing multiple responsibilities and helping with activities of daily living (Stenberg, Ruland, & Miaskowski, 2010), informal caregivers to those with a rare disease describe taking on complex roles, including researcher, advocate, and care coordinator to support the care recipient (Shire Report, 2013). Taking on these responsibilities alters the daily lives of caregivers. In the US, caregivers to an adult with a rare disease spend an average of 29.7 hours per week providing care (National Alliance for Caregiving, 2018). When no treatment is available for a care recipient, an average of an additional 12 hours per week (41.7 hours per week) is reportedly spent providing care (National Alliance for Caregiving, 2018).

The burden of caregiving can accumulate from adapting to a new role, disease duration, uncertainty of the disease progression, and unpredictability of symptom flareups (Long, Moriarty, Mittelman, & Foldes, 2014). A lack of information about the disease, few resources available, and limited information for caregivers in rare disease settings further complicate the caregiving experience (Pelentsov, Laws, & Esterman, 2015). These challenges result in an increased need for social connections of individuals caring for a loved one with the same disease (Pelentsov et al., 2015). Due to the rarity of these diseases, however, caregivers are unlikely to meet someone going through the same caregiving experience, resulting in increased isolation (Maril, 2012).

Carrying out caregiving responsibilities while experiencing isolation can amplify the physical, economic, and psychological implications of the caregiver role (National Alliance for Caregiving, 2018; Pearlin, Mullan, Semple, & Skaff, 1990). Caregivers to a loved one with a rare disease experience a decline in their own physical health, increased stress and depression, and

decreased subjective well-being and self-efficacy (Adams, Miller, & Grady, 2016; National Alliance for Caregiving, 2018). In Canada, a 2019 national report found that 75% of Canadians caring for a loved one with a rare disease report social isolation and two-thirds (66%) of reported feeling unsupported by Canada's healthcare system (Canadian Organization for Rare Disorders, 2019). Ninety percent of these caregivers reported that their financial situation has been negatively impacted by their care recipients' rare disorder, as almost half of caregivers reported missing 6 or more days of work per month due to caregiving responsibilities. These stressors result in decreased self-reported mental health of caregivers of individuals with a rare disease in Canada, yet, caregivers have been described as the most critical component of the rare disease community, as our national healthcare system relies upon the care provided by these individuals to maintain health system capacity (Canadian Organization for Rare Disorders, 2019).

Scleroderma and Informal Caregiving

Approximately 16,000 Canadians are affected by systemic sclerosis (SSc), also known as scleroderma, which is a rare chronic autoimmune disease (Bernatsky et al., 2009). SSc is a degenerative disorder (Abraham & Varga, 2005). On average, incidence rates of SSc in the United States are 20 cases per million per year (Mayes et al., 2003). Disease onset typically occurs between ages of 30 and 50 years and 80% of people with SSc are women (Mayes et al., 2003). There is currently no cure for SSc, and from time of diagnosis, the median survival is approximately 11 years which was considerably less than one study found for a sample matched for age, sex, and race (ratio of observed to expected survival 63% at 10 years) (Mayes et al., 2003). SSc tends to be more severe among African Americans and Native Americans as compared to White Americans (Gelber et al., 2013). Patients with SSc experience a range of

problems that significantly affect their quality of life, including gastrointestinal symptoms, Raynauds syndrome, respiratory problems, fatigue, pain, pruritus, symptoms of depression, and disfiguring changes in appearance (Denton & Khanna, 2017). Few treatments to alleviate the symptoms associated with SSc exist, and the combination of disfigurement to highly visible parts of the body, a sequalae of physical complications, and psychosocial distress can make daily life difficult.

Given the range of symptoms and associated difficulties that impact persons diagnosed with SSc, as well as the chronicity of the condition with no cure and substantial uncertainty, many people with SSc receive substantial care from a family member or friend (Lopez-Bastida, Linertova, Oliva-Moreno, Posada-de-la-Paz, & Serrano-Aguilar, 2014; Maril, 2012). There is little available research, however, regarding informal caregivers of people with SSc (Pelentsov, Laws, & Esterman, 2015). Aside from economic analyses highlighting the financial burden of SSc caregivers due to absenteeism from work secondary to caring responsibilities, prior to my research, only one study, an unpublished thesis, had examined the experience of caregivers in SSc settings (Maril, 2012). This study interviewed 13 informal caregivers (8 females, 5 males) to those with SSc and collected qualitative data regarding the caregiving experience. Key themes that emerged from these interviews included the negative effect that caregiving has on emotions (e.g., increased feelings of stress), existential concerns (e.g., considering mortality of the care recipient and trying to find meaning) and an overall complexity in managing SSc due to the uniqueness of the disease, causing additional difficulties and feelings of isolation (Maril, 2012). This study also provided recommendations for future research of caregivers to persons with SSc to build upon the results from the qualitative work, including, (1) conducting quantitative and mixed-methods studies with larger sample sizes, (2) conducting research to guide intervention

development based on resources available for other rare disorders, and (3) recruiting male caregivers to better represent the expected demographic characteristics of caregivers to those with SSc, since most people with SSc are female (Maril, 2012). These suggested areas for future research align with policy recommendations established by national organizations of rare disease caregiver research in Canada and the US, which highlight the need to enhance the health and well-being of caregivers to persons with rare disorders through disease-specific psychosocial interventions (National Alliance for Caregiving, 2018; The Change Foundation, 2019).

In order to support the design and development of appropriate psychosocial interventions that can best meet the needs of caregivers to people with SSc, engaging caregivers in the research design process is essential. Involving people with lived experience and health care professionals in research development from conceptualization to knowledge translation is necessary for effective program development and in generating research priorities relevant to stakeholder groups (i.e., caregivers, care recipients, policy makers, and researchers) (Breault et al., 2018; Manns et al., 2014). Given an absence of research assessing disease-specific challenges and preferences for psychosocial interventions among caregivers to persons with a rare disease, including those with SSc, the current thesis aimed to develop a program of research in collaboration with people with lived experience caring for individuals with SSc. Applying this research within the context of SSc is critical due to the severity of this condition, the lack of available interventions to support caregivers, and the engagement of the SSc community in contributing to and participating in research.

Literature Summary and Research Objectives

The research conducted to date among caregivers to persons with a rare disorder has demonstrated that caregivers are often fully or partially responsible for organizing and sustaining

their care recipients' medical care. To understand the demands of caregivers, it is important to understand the disease with which caregivers are involved (Vitaliano, Young, & Zhang, 2004), highlighting the need for disease-specific research programs that engage people with lived experience. Caregivers to individuals SSc are at a heightened risk of experiencing negative psychosocial and physical consequences of caregiving due to the disease rarity and severity, and the lack of available treatment options. Aside from one previous thesis with 13 caregivers to individual with SSc, however, the experience of SSc caregivers has not been explored. This absence of research is a central barrier in the development and initiation of services to improve the well-being of caregivers to persons with SSc, as disease-specific caregiving challenges are currently unknown. This precludes the ability to adapt currently available caregiver interventions. Further, for a rare disease such as SSc, identifying and understanding the caregiver experience could aid in bringing awareness and knowledge of this disease and the complex reality of managing a rare disorder to physicians, clinicians, and society, while generating the necessary knowledge to allow for useful intervention development.

The aim of the present research was to understand the experience of caring for someone living with SSc and identify caregiver priorities by embedding people with lived experience into the research process. Conducting practical research that can allow for the creation of research priorities to support caregivers has been absent from the literature and is an important area of focus given the overwhelming lack of support reported by Canadian caregivers to individuals with a rare disease. As such, three independent studies were conducted and included in this thesis. First, a scoping review was conducted to explore the 1) perceived benefits of participating in psychosocial interventions for caregivers of individuals with a rare disease, and 2) barriers and facilitators of establishing and maintaining these interventions. Next, a study was conducted to

develop a caregiver generated list of 1) challenges experienced by informal caregivers of people living with SSc, and 2) their preferences for types of support services that could potentially be developed. Finally, a study was performed through the administration of an online questionnaire in order to 1) evaluate the importance of different challenges experienced by informal caregivers to persons with SSc and 2) identify priorities for support services that could potentially be developed.

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Chapter 2

Manuscript 1 (The Patient – Patient-Centered Outcomes Research – Under Review)

Submitted to The Patient – Patient-Centered Outcomes Research: Rice, D. B., Carboni-Jiménez, A., Cañedo-Ayala, M., Turner, K. A., Chiovitti, M., Levis, A.W., & Thombs, B. D. (2020). Demonstrated and Perceived Benefits and Factors that Influence the Ability to Establish and Maintain Services for Informal Caregivers of People with Rare Diseases: A Scoping Review.

Demonstrated and Perceived Benefits and Factors that Influence the Ability to Establish and Maintain Services for Informal Caregivers of People with Rare Diseases: A Scoping Review

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No specific funding was received for this review. DBR is supported by a Vanier Graduate Scholarship through Canadian Institutes of Health Research. ACJ is supported by a Canadian Graduate Scholarship (CGS – M) through Canadian Institutes of Health Research unrelated to the present study. KAT was supported by a Fonds de Recherche Québec - Santé (FRQ-S)

masters training award unrelated to the present study. No funding body had any input into the any aspect of this review.

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ABSTRACT

Background: Psychosocial interventions can be an important resource for caregivers. Little is known, however, about the benefits of participating in psychosocial interventions for caregivers of patients with rare diseases and the factors that may influence the establishment and maintenance of these services. The objective of this scoping review was to map the literature reporting (1) benefits of participating in psychosocial interventions for caregivers of someone with a rare disease and (2) the barriers and facilitators of establishing and maintaining them.

Methods: CINAHL and PubMed were searched in December 2018. Studies in any language that described benefits of participating in psychosocial interventions for caregivers or the barriers and facilitators of establishing and maintaining these interventions were eligible for inclusion.

Results: There were 2,257 titles and abstracts reviewed. Thirty-four studies met inclusion criteria for the review. Interventions were behavioural or psychological, supportive, educational, or multi-component. All (100%) included studies reported on benefits of participating in psychosocial interventions, 14 (41%) reported on facilitators and 19 (56%) reported barriers of establishing and maintaining interventions. Benefits that were most common included statistically significant improvements in emotional states (e.g., stress) and caregiver burden, and narrative reports of the intervention being helpful. Four themes for facilitators of establishing and maintaining interventions were identified (i.e., characteristics of the intervention, characteristics of intervention delivery, providing resources, support provided outside of intervention) and 4 themes of barriers were identified (i.e., intervention misaligned to caregiver needs, ability to make time for intervention, practical barriers, emotional barriers).

Conclusions: Psychosocial interventions for caregivers to a loved one with a rare disease may be a helpful source of support for many caregivers. Future research should design interventions for caregivers that take into account the facilitators and barriers to establishing and maintaining such interventions.

Keywords*:* Rare Diseases; Psychosocial Interventions; Scoping Review; Caregivers

INTRODUCTION

Informal caregivers are people who provide ongoing support for a family member or friend in need of care due to a health condition (Reinhard, Given, Petlick, & Bemis, 2008). They are caregivers who are not compensated monetarily for providing care, and most do not receive formal training (Reinhard et al., 2008). Nonetheless, the level of care that they provide can be substantial (Reinhard et al., 2008) and may include physical, practical and emotional aspects of care (Candy, Jones, Drake, Leurent, & King, 2011). Tasks involved vary across situations, but may include assisting with transportation, activities of daily living (e.g., feeding, dressing), managing medication and household activities (e.g., chores, meal preparation, paying bills), negotiating work and school environments, and emotional support (Stenberg, Ruland, & Miaskowski, 2010; Turner & Findlay, 2012).

Significant burden on informal caregivers is common. A population-based survey of over 1200 informal caregivers from the United States found that 58% reported moderate to high levels of burden, defined by the amount of time spent providing care and the degree of dependency of care recipients (National Alliance for Caregiving, 2015). On average, caregivers reported that they provide 24 hours of care per week with approximately 1 in 4 providing 41 hours or more (National Alliance for Caregiving, 2015). A systematic review of burden among informal caregivers of patients with cancer found that greater reported burden was associated with a higher likelihood of caregivers reporting physical health problems (e.g., pain, headache, muscle tension), social problems (e.g., difficulties paying bills, balancing multiple roles, feelings of being unappreciated), and emotional problems (e.g. anxiety, worry, fear of leaving the patient alone) (Stenberg et al., 2010).

Little is known about the experiences and challenges faced by informal caregivers of patients with rare diseases (Shire, 2013). It would be expected that they experience the same challenges faced by carers of patients with more common diseases. Additionally, they may need to navigate challenges related to gaps in knowledge about the rare disease of the person for whom they care, as well as more limited support resources than are available for people with more common diseases (Doyle, 2015; Gowran et al., 2015; Kole & Faurisson, 2009; European Organisation for Rare Diseases, 2005). Caregivers regularly attend medical appointments and provide transportation for patients, which can involve travelling long distances for some people with rare diseases. Obtaining an accurate diagnosis and making treatment decisions often involves multiple consultations with different doctors (Schieppati, Henter, Daina, & Aperia, 2008; Stenberg et al., 2010). Unlike common medical disorders, many caregivers of a patient with a rare disease have never met another individual caring for a patient with the same disease (Maril, 2012). This can result in significant isolation for informal caregivers of a family member or friend diagnosed with a rare disease (European Organisation for Rare Diseases, 2005).

Many informal caregivers of patients with common medical disorders rely upon informational and emotional support interventions offered by professionals, volunteers, or peers. Psychoeducational and supportive interventions have been developed with the goal of improving caregiver well-being (Nelis, Quinn, & Clare, 2007). Psychoeducational interventions may offer information about a patient's diagnosis, implications of an illness, caregiving skills, and support networks (Banningh et al., 2013). Psychosocial interventions may include professionally provided services or peer support options, such as support groups, which can provide emotional support by bringing together individuals facing similar disease-related or caregiving challenges to empower one another through social contact and support (Aymé, Kole, & Groft, 2008; Nelis et

al., 2007). A meta-analysis found that professionally led support groups for informal caregivers of patients with dementia led to improvements in psychological well-being, caregiver burden, and social consequences (e.g., social support, relationship with the patient) (Chien et al., 2011).

In common medical conditions, psychosocial services to support caregivers may be available through the health care system or advocacy groups and are organized and delivered by knowledgeable professionals (Brereton, Carroll, & Barnston, 2007; Chien et al., 2011). In rare diseases, on the other hand, where professional support services are not typically available, patients and caregivers sometimes organize their own informational and emotional sources of support (Doyle, 2015; Reimann, Bend, & Dembski, 2007). Establishing and maintaining these types of interventions, however, pose challenges related to the small number of patients or caregivers affected by any rare disease and their wide geographical distribution.

The establishment and maintenance of effective support services for informal caregivers of rare disease patients requires an understanding of the reasons why people may use these services and what they hope to obtain from them, as well as factors that influence the ability to establish and maintain informational and emotional support services in a rare disease context. Thus, the objective of this scoping review was to identify and map evidence on the (1) demonstrated and perceived benefits of psychosocial interventions for caregivers of patients with rare diseases and (2) the barriers and facilitators to initiating and maintaining these interventions.

METHODS

A scoping review is a "form of knowledge synthesis that addresses an exploratory research question aimed at mapping key concepts, types of evidence, and gaps in research related to a defined area or field by systematically searching, selecting, and synthesizing existing

knowledge" (Colquhoun et al., 2014). A scoping review is rigorous like a systematic review but addresses broader topics and includes relevant studies regardless of their study design (Arksey & O'Malley, 2005). A protocol for the methods for this scoping review were drafted prior to beginning the review. The methods were not posted online. The methods applied drew upon recommendations by Arksey and O'Malley (Arksey & O'Malley, 2005), as well as subsequent refinements by Levac, Colquhoun, and O'Brien (Levac, Colquhoun, & O'Brien, 2010) and Colquhoun et al. (Colquhoun et al., 2014). As recommended in these publications, we utilized a five-stage methodological framework: (1) Identifying the research question, (2) Identifying relevant studies, (3) Selecting studies, (4) Charting the data, and (5) Collating, summarizing, and reporting results (Arksey & O'Malley, 2005; Colquhoun et al., 2014; Levac et al., 2010). The reporting of this review followed the Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) reporting guidelines, please see Appendix A (Tricco et al., 2018).

Identifying the research question

Rare disease organizations have identified supporting caregivers as an important priority. In a national Canadian survey of caregivers to individuals with a rare disease, 66% of respondents reported feeling unsupported by the healthcare system (Canadian Organization for Rare Diseases, 2019). To guide this scoping review and the development and implementation of support services for caregivers, we defined the following research question: What are the (1) demonstrated and perceived benefits of psychosocial interventions for caregivers of patients with rare diseases and (2) barriers and facilitators to initiating and maintaining these interventions.

Identifying relevant studies

Articles published in any language that described tested or perceived benefits of an intervention intended to support caregivers of people with rare diseases or the facilitators and barriers of establishing and maintaining these interventions were eligible for inclusion. We did not set any methodological restrictions in our eligibility criteria in order to avoid missing important or significant data, which is consistent with standard scoping review methods (Arksey & O'Malley, 2005; Colquhoun et al., 2014; Levac et al., 2010). Articles were eligible if they included informal caregivers of people diagnosed with a rare disease based on Orphanet's "List of rare diseases and synonyms in alphabetical order" (March 2016, available at https://osf.io/2cd7w/?view_only=d5b686b288ab46e081db815ff39f8512), which includes diseases with prevalence rates of 1 person in 2,000 or less in European countries. The only exception to this was when an article was conducted in a non-European setting where the disease may or may not be rare (e.g., tuberculosis in Ethiopia); in these instances, we determined the disease's prevalence rate in the country in question based on data available on the World Health Organization's website (https://www.who.int/health-topics/) (World Health Organization, 2015). If the intervention was conducted in a country where the disease's prevalence rate is 1 person in 2,000 or less, then the article was included; however, if it was conducted in a country where the disease's prevalence rate is greater than 1 person in 2,000, then the article was excluded. Articles about interventions intended for caregivers of people without rare diseases were excluded, even if some participants in the studies described may have cared for persons with a rare disease.

For the purpose of this study, any intervention intended to support informal caregivers of a person with a rare disease was included. Informal caregivers were defined as a family member or friend helping to care for a person diagnosed with a rare disease. Examples of eligible interventions included strategies for reducing emotional distress, decreasing burden, or providing

education to support caregivers. Eligible interventions could be delivered in any format, including in-person interventions, written materials, or the use of the telephone or internet and could be delivered in groups (e.g., support groups), or individually (e.g., individual psychotherapy). Interventions that were delivered to informal caregivers but that were intended to impact outcomes only of the person with the medical condition, but not the caregiver, were excluded. Interventions delivered to informal caregivers and that involved outcomes for both the person with the medical condition and for the informal caregiver were included.

In order to identify potentially relevant publications that described relatively current versions of support services for caregivers of people with rare diseases, we searched PubMed and CINAHL through the EBSCOhost platform from January 2000 through December 2018. A medical librarian developed the search strategy and performed the search. To develop the search strategy, we extracted the names of rare diseases listed in Orphanet's March 2015 "Rare Disorders and Cross-References" dataset (available at https://osf.io/2cd7w/?view_only=d5b686b288ab46e081db815ff39f8512). The list included names of disorders, groups of disorders, subtypes, and synonyms and totalled 20,169 unique terms. To manage the size of the search, we excluded names of groups of disorders and synonyms, leaving 6,999 unique rare disorders and subtypes. We then combined these disorder names with terms relevant to informational and psychosocial interventions (Nelis et al., 2007). The complete search strategy can be found in Appendix B.

Selecting studies

The results of the search were downloaded into the citation management database RefWorks (RefWorks, RefWorks-COS, Bethesda, MD, USA), and duplicate references were identified and removed. Following this, references were transferred into the systematic review

software DistillerSR (Evidence Partners, Ottawa, Canada). Using this software, we assessed the eligibility of each reference through a two-stage process. First, two investigators independently reviewed the titles and abstracts of all articles identified through the search strategy. If either investigator deemed an article potentially eligible based on the inclusion criteria, then two investigators completed a full-text review, independently. Disagreements after full-text review were resolved by consensus, with a third investigator consulted when necessary.

Charting the data, and collating, summarizing, and reporting results

Two investigators independently extracted data from each included study and entered it into a standardized Excel spreadsheet. For each publication, we extracted the following information: (1) first author; (2) publication year; (3) study design (4) country where the intervention took place; (5) disease; (6) number, mean age, and gender or sex of caregivers; (7) intervention description; (8) control group intervention description, if applicable; (9) type of intervention; (10) participants included in the study (i.e., caregiver only, caregiver and care-recipient, or caregiver, care-recipient and family); (11) intervention delivery format (e.g., group, individual); (12) individual that delivered the intervention (e.g., peer); (13) mode of intervention delivery (i.e., in person, online, telephone, multi-delivery); (14) whether the intervention was rare disease-specific; (15) actual or perceived benefits of the intervention; (16) facilitators of the intervention; and (17) barriers of the intervention. For articles on interventions that included both patients and informal caregivers, only data from informal caregivers were extracted. Disagreements were resolved by consensus, and a third investigator was consulted when necessary.

Quantitative findings related to benefits, barriers, or facilitators to establishing and maintaining rare disease support services for caregivers were extracted, including any results

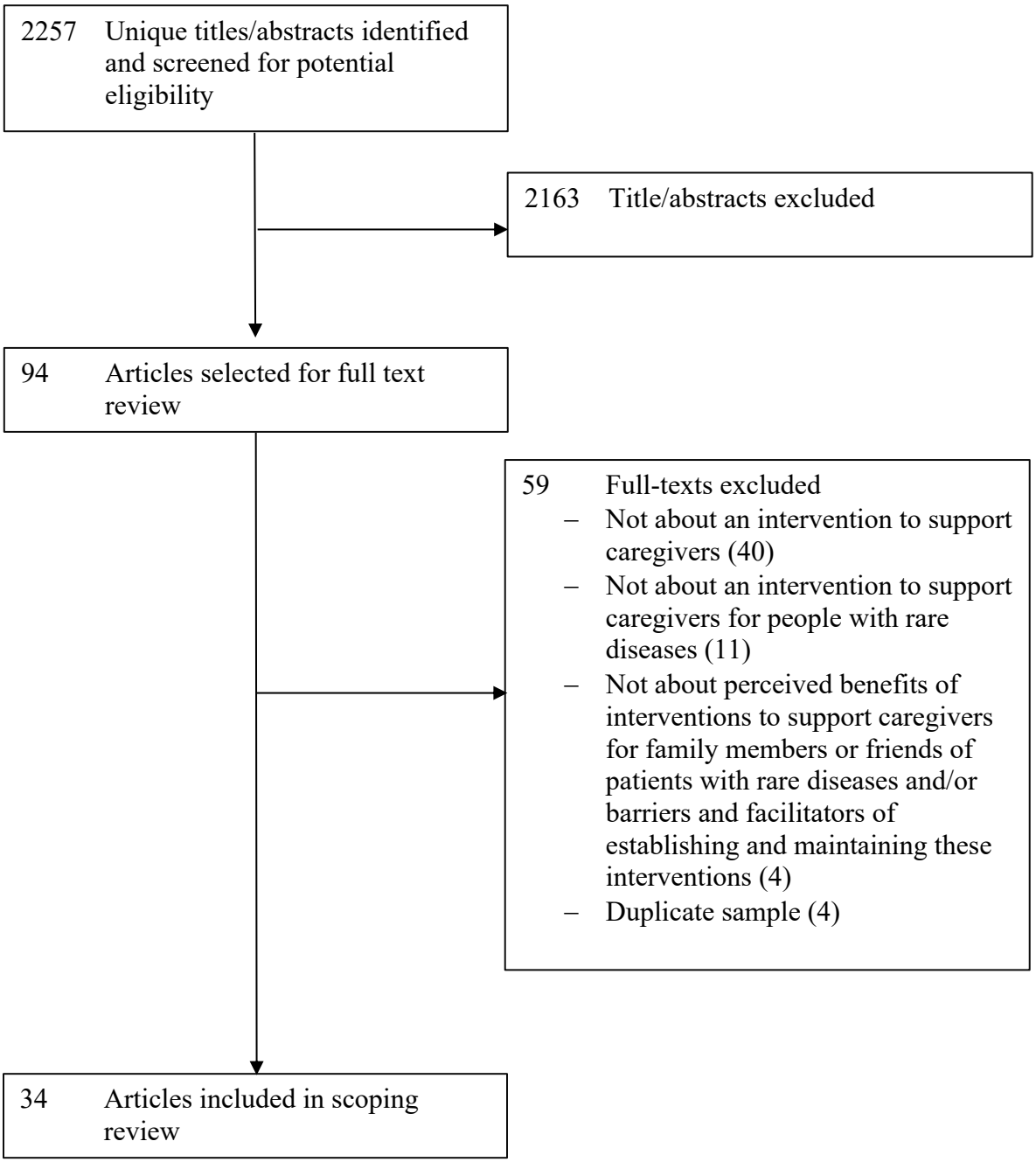
from statistical tests or aggregate data that were reported. All reported results in each eligible study, including significant and non-significant findings were extracted and included in results tables. Only statistically significant findings, however, were categorized as "tested benefits" for mapping perceived and tested benefits. Evaluation of benefits included between group comparisons where available (e.g., any study that compared an intervention to a control group) or within group differences if the study only compared pre and post scores. If effect sizes were not reported, standardized mean differences (SMDs) were calculated using study data (i.e., sample sizes, means, standard deviations, t-values, degrees of freedom) when necessary data were provided (Becker, 1999; Lipsey & Wilson, 2001). Baseline and first post-intervention follow-up points were used to calculate pre-post changes. In the case that only baseline and a later follow-up time point were available, this information was used.

Qualitative results of perceived benefits, barriers, and facilitators to establishing and maintaining rare disease support services for caregivers were extracted and included any reports in results presented in narrative format or solely descriptive results that did not include comparative statistical tests. These findings were categorized using conventional content analysis (Hsieh & Shannon, 2005). Two investigators independently identified key themes in the qualitative findings without using preconceived categories. Themes generated were discussed between reviewers until consensus was achieved. Subthemes were generated by one investigator and reviewed by a second investigator. Content analysis was also used to identify key themes for quantitative findings, such as classifying the type of outcomes reported. All results were collated and grouped based on the type of intervention studied. Results were presented in tabular format.

RESULTS

The database search yielded 2257 unique titles and abstracts. Of these, 2163 were excluded after title and abstract review, leaving 94 publications for full-text review. A total of 34 publications (A'Campo, Spliethoff-Kamminga, & Roos, 2012; Alankaya & Karadakovan, 2015; Aoun, Chochinov, & Kristjanson, 2015; Bevans et al., 2010; Bevans et al., 2014; Bozkurt et al., 2014; Cipolletta, Gammino, Francescon, & Palmieri, 2018; Cox et al., 2012; Dowling et al., 2014; Dunlop, Kent, Lashley, & Caruana, 2016; Elliott & Berry, 2009; Elliott, Brossart, Berry, & Fine, 2008; Fidika et al., 2015; Gormley, Duff, Brownlee, & Hearnshaw, 2014; Langer et al., 2012; Laudenslager et al., 2015; Leenaars, Denys, Henneveld, & Rasmussen, 2012; Li et al., 2017; Lindell, 2008; Manne, Mee, Bartell, Sands, & Kashy, 2016; Marconi et al., 2015; Mazanec et al., 2017; Mioshi, McKinnon, Savage, O'Connor, & Hodges, 2013; Moola, Henry, Huynh, Stacey, & Faulkner, 2017; Raj et al., 2015; Rexilius, Mundt, Megel, & Agrawal, 2002; Rodgers et al., 2007; Rotondi, Sinkule, & Spring, 2005; Schulz et al., 2009; Sheija & Manigandan, 2005; Stewart et al., 2001; Van Groenestijn et al., 2015; Videaud, Torny, Cartz-Piver, Deschamps-Vergara, & Couratier, 2012; Wade et al., 2010) met the inclusion criteria and were included in the scoping review (see Figure 1). A list of studies excluded at full-text level, with reasons, is provided in Appendix C.

Figure 1. Flow diagram of publication selection process.



Publication Characteristics

All 34 included publications were primary research studies, 17 (50%) of which were trials of interventions that included a control group (A'Campo et al., 2012; Dowling et al., 2014; Elliott & Berry, 2009; Elliott et al., 2008; Langer et al., 2012; Laudenslager et al., 2015; Li et al., 2017; Lindell, 2008; Manne et al., 2016; Mazanec et al., 2017; Mioshi et al., 2013; Raj et al., 2015; Rexilius et al., 2002; Schulz et al., 2009; Sheija & Manigandan, 2005; Van Groenestijn et al., 2015; Wade et al., 2010). The sample size in studies ranged from 4 to 475 caregivers (median = 28). Twenty-one publications (62%) were from North America (Bevans et al., 2010; Bevans et al., 2014; Cox et al., 2012; Dowling et al., 2014; Dunlop et al., 2016; Elliott & Berry, 2009; Elliott et al., 2008; Langer et al., 2012; Laudenslager et al., 2015; Leenaars et al., 2012; Lindell, 2008; Manne et al., 2016; Mazanec et al., 2017; Moola et al., 2017; Raj et al., 2015; Rexilius et al., 2002; Rodgers et al., 2007; Rotondi et al., 2005; Schulz et al., 2009; Stewart et al., 2001; Wade et al., 2010) nine (26%) were from Europe (A'Campo et al., 2012; Alankaya & Karadakovan, 2015; Bozkurt et al., 2014; Cipolletta et al., 2018; Fidika et al., 2015; Gormley et al., 2014; Marconi et al., 2015; Van Groenestijn et al., 2015; Videaud et al., 2012), two (6%) were from Australia (Aoun et al., 2015; Mioshi et al., 2013), and two (6%) were from Asia (Li et al., 2017; Sheija & Manigandan, 2005). Six publications included caregivers of individuals with hematopoietic stem cell transplantation (Bevans et al., 2010; Bevans et al., 2014; Langer et al., 2012; Laudenslager et al., 2015; Manne et al., 2016; Rexilius et al., 2002), 4 included caregivers of individuals with amyotrophic lateral sclerosis (Alankaya & Karadakovan, 2015; Cipolletta et al., 2018; Marconi et al., 2015; Van Groenestijn et al., 2015), 4 included caregivers of individuals with spinal cord injury (Elliott & Berry, 2009; Elliott et al., 2008; Schulz et al., 2009; Sheija & Manigandan, 2005), 3 included caregivers of individuals with cystic fibrosis (Fidika et

al., 2015; Gormley et al., 2014; Moola et al., 2017), 3 included caregivers of individuals with traumatic brain injury (Raj et al., 2015; Rotondi et al., 2005; Wade et al., 2010), 2 included caregivers of individuals with frontotemporal dementia (Dowling et al., 2014; Mioshi et al., 2013), 2 publications included caregivers of individuals with HIV/AIDS (Li et al., 2017; Stewart et al., 2001), and 1 publication reported on a sample that combined caregivers of individuals with traumatic brain injury and spinal cord injury (Rodgers et al., 2007). The remaining 9 publications involved caregiving to persons with a rare disease that was not reported on in any other included study (A'Campo et al., 2012; Aoun et al., 2015; Bozkurt et al., 2014; Cox et al., 2012; Dunlop et al., 2016; Leenaars et al., 2012; Lindell, 2008; Mazanec et al., 2017; Videaud et al., 2012). Characteristics of included publications and all extracted study findings are summarized in Tables 1-4.

Table 1. Study Characteristics and Interventions - Behavioural/Psychological Interventions

First Author, Year Country	Type of Disease	Demographics of Included Caregivers	Population, Intervention Description, Objectives	Control Group Intervention	Intervention Delivery Format	Intervention Use and Satisfaction	Tested Benefits	Facilitators	Barriers
Trials									
Dowling 2014 USA	Frontotemporal Dementia	**Life Enhancing Activities for Family Caregivers:** N= 12 Mean Age: 59 Percent Female: 75% **Control:** N= 12 Mean Age: 60 Percent Female: 64%	Population: Caregivers of individuals with frontotemporal dementia. Intervention: A generic intervention, "Life Enhancing Activities for Family Caregivers (LEAF)", which involved positive emotion skill building interventions, was delivered by clinical nurses and a psychologist, through five 1-hour weekly individual sessions either in person or through videoconference. Gratitude, mindfulness, positive reappraisal, personal strengths and goals, and altruistic behaviors were taught and were intended to be applied and recorded in logs outside of sessions. Objectives: To determine the feasibility of the intervention, improve caregiver's positive affect, mood, and decrease stress and burden.	Five 1-hour individual sessions with a facilitator. Sessions involved reviewing affect diaries that participants were keeping, and discussing a theme (e.g., diet and exercise) but did not involve a didactic component.	Individual face-to-face and/or online delivered by professionals.	The two most favourably ranked skills learned in the intervention were "noticing positive events" and "gratitude".	There were six caregiver outcomes. Four of them showed a statistically significant improvement, and two of them did not. A significant difference between groups was found in favour of the intervention group on the Differential Emotions Scale with decreased negative affect scores ($p < 0.05$; partial eta squared = 0.36) and increased positive affect scores ($p < 0.05$, partial eta squared = 0.30), the Perceived Stress Scale ($p < 0.05$; partial eta squared = 0.48) and the Zarit Burden Interview ($p < 0.05$; partial eta squared = 0.26). No statistical significance was found between groups for Neuropsychiatric Inventory or the Center for Epidemiologic Studies Depression scale.	Sessions were delivered through videoconference, which allowed caregivers to stay at home and eliminated barriers such as finding patient supervision, distance, caregiver health, and financial strain.	Not Reported
Elliott 2008 USA	Spinal Cord Injury	**Overall:** Mean Age of Females= 47 Mean Age of Males= 60 Percent Female: 89% **Problem-Solving Intervention:** N=32 **Control:**	Population: Caregivers of individuals with spinal cord injury. Intervention: A generic intervention focused on problem-solving skills delivered by coordinators with doctorates in clinical psychology through videoconference sessions. One face-to-face in-home session lasting 2 to 3 hours was provided to train caregivers to use the videoconferencing software and to	Education provided by telephone at scheduled intervals and as needed based on participant needs. Monthly face-to-face videoconferencing to discuss educational materials.	Individual face-to-face, online, and by phone, delivered by professionals.	Not Reported	There were six caregiver outcomes, none of which were significantly different between groups. No statistically significant differences were found on the Inventory to Diagnose Depression, Social Problem-Solving Inventory-Revised, the Satisfaction with Life Scale, or on the General Health, Social Functioning,	Not Reported	Not Reported

		N=29	provide information about problem-solving. The remainder of sessions were given once a month over 1-year, and involved caregivers developing solutions to problems encountered in different caregiver scenarios. Objective: to evaluate the feasibility of the intervention and assess improvements in social problem-solving skills, quality of life, and depressive-symptoms.				or Mental Health subscales of the Medical Outcomes Study Short Form Health Survey.		
Langer 2012 USA	Hematopoietic Stem Cell Transplantation (HSCT)	**Emotional Expression:** N= 28 Mean Age: 51 Percent Female: 71% **Control:** N= 29 Mean Age: 57 Percent Female: 72%	Population: Caregivers of people undergoing HSCT. Intervention: A rare-disease adapted intervention, emotional expression, was delivered to caregivers of people going through HSCT. A total of three 10-minute talking sessions were delivered, where caregivers were instructed to talk (into an audio-recorder) about their deepest thoughts and feelings related to their care-recipient's transplant as well as their experience as a caregiver. An experimenter was also present in the room. Objectives: To decrease negative affect.	Three 10-minute sessions of discussing caregivers' daily schedule.	Individual face-to-face, with a professional.	Caregivers in the intervention group rated exercises as significantly more helpful, meaningful, revealing of their emotions and noted sharing more information (all $p < 0.05$) than caregivers in the control group. There was no significant between groups difference of if caregivers they would recommend the intervention.	Not Reported	Not Reported	Timing of the intervention was 50 days post-patient transplant, which caused less people to participate in the intervention because of other caregiving responsibilities. A writing experience was described as possibly being more accessible than one delivered in-person.
Laudenslager 2015 USA	Allogeneic Hematopoietic Stem Cell Transplantation (AHSCT)	**Stress Management:** N= 74 Mean Age: 52 Percent Female: 76% **Control:** N= 74 Mean Age: 55 Percent Female: 76%	Population: Caregivers of people undergoing AHSCT. Intervention: A rare-disease adapted psychoeducation and relaxation exercises, PsychoEducation, Paced Respiration and Relaxation (PEPRR). A total of 8 semi-structured one-on-one weekly sessions were delivered by a social worker starting 2 weeks post-transplant. Sixty-75 minute sessions covered topics such as coping-skills training, stress management, and ways to improve	Treatment as usual.	Individual face-to-face, with a professional.	Most caregivers found the intervention acceptable and were willing to complete it (70%).	There were 10 caregiver outcomes, five of which were significant. A statistically significant difference was found between caregivers receiving the intervention and those receiving treatment as usual in the Perceived Stress Scale ($p = 0.04$; effect size = 0.39), the Center for Epidemiological Studies Depression Scale ($p = 0.02$; effect size = 0.46), the State-Trait Anxiety	Sessions took place at the hospital when caregivers were visiting their patients during hospitalization or at the clinic during appointments. Integrating flexibility into the intervention (e.g., varying times for starting, ability to pause intervention) was an important facilitator. Sessions	There was a high drop-out rate due to the seriousness of the illness, time constraints, lack of interest, living far away, scheduling difficulties, and experiencing conflict with the care recipient.

			mental and physical well-being. A workbook and respiration relaxation device were provided. Caregivers were instructed to use the device for 15 minutes at least 4-5 times per week. Objective: To determine if the intervention significantly impacted physiological and psychological variables (e.g., perceived stress, cortisol awakening response, symptoms of anxiety and depression, and distress).				Inventory ($p = 0.01$; effect size = 0.66), the Profile of Mood States-Total Mood Disturbance ($p = 0.04$; effect size = 0.39), and the Caregiver Distress Composite score ($p = 0.02$; effect size = 0.45). No statistically significant differences between groups were found in the Caregiver Reaction Assessment, the Pittsburgh Sleep Quality Index, the Short Form Health Survey-36 Physical and Mental components, the Impact of Events Scale, and the Composite Caregiver Well-being score (all $p > .05$).	were also tailored to the specific needs of the caregivers.	
Lindell 2007 USA	Idiopathic Pulmonary Fibrosis	**Program to Reduce Symptoms and Improve Lifestyle Management:** N= 10 Mean Age= 63 Percent Female: 76% **Control:** N= 8 Mean Age= 67 Percent Female: 54%	Population: Caregivers of idiopathic pulmonary fibrosis. Intervention: A rare-disease specialized Program to Reduce Symptoms and Improve Lifestyle Management (PRISIM) focused on the improvement of lifestyle, and was provided to patients and caregivers. Six group sessions lasting 2 hours in length were delivered by trained nurses and an advanced care planning instructor. Each session addressed topics (e.g., exercise, cognitive behavioural techniques) through educational material. Homework was assigned based on the topic discussed in each session. Objectives: To decrease levels of anxiety, depression, perceived stress, and improve perceptions of health-related quality of life.	Usual care and a book.	Group face-to-face, patients and caregivers, delivered by professionals.	Caregivers reported feeling less isolated and enjoyed participating in a research study.	There were five caregiver outcomes, one of which was significant. Caregivers in the intervention group reported significantly lower scores than caregivers in the control group on the Perceived Stress Scale ($p = 0.02$; $d = -1.96$). There were no statistically significant differences between groups on the Beck Anxiety Inventory, Beck Depression Inventory, SF-36 Short Form Medical Outcomes Study Form Version 2 Physical Component, and SF-36 Mental Component (all $p > .05$).	Not Reported	Barriers of this study related to attendance. Many of the participants had to travel 2-3 hours to arrive to the intervention.
Manne 2016 USA	Hematopoietic Stem Cell Transplantation (HSCT)	**Parent Social-Cognitive Intervention Program:** N= 110	Population: Parents of children undergoing HSCT. Intervention: Adapted for a rare-disease context, focused on	Parents received a DVD, pamphlet, offer of respite care, and a walkie-talkie to	Individual face-to-face, delivered by professionals.	Most caregivers reported using the CD-ROM provided (86%). On average,	There were four caregiver outcomes, including the Beck Anxiety Inventory, the Beck Depression Inventory, the Impact of	Not Reported	There was a very high drop-out rate due to the timing of the intervention

		Mean Age: 37 Percent Female: Not Reported **Control:** N= 108 Mean Age: 38 Percent Female: Not Reported	cognitive and social processing strategies. Delivered 2-3 weeks post-transplant, through five 1-hour individual sessions delivered over 2 to 3 weeks by graduate level therapists. A CD-ROM and workbook supplemented sessions. Objectives: To reduce traumatic distress, depression, and anxiety symptoms, and to increase overall well-being.	communicate with their child.		caregivers rated the intervention fairly well with an average score of 3.5 out of 5.	Events Scale, and the Mental Health Inventory, none of which significantly differed between groups post-intervention.		which occurred 1-week post-transplant. Caregivers reported being too overwhelmed to complete the program. Caregivers reported wanting to focus all their time and energy on their child (93%), not wanting to leave their child alone (82%), and already having adequate support (59%).
van Groenestijn 2015 Netherlands	Amyotrophic Lateral Sclerosis	**Cognitive Behavioural Therapy:** N= 10 Mean Age: 57 Percent Female: 70% **Control:** N= 5 Mean Age: 53 Percent Female: 60%	Population: Caregivers to those with amyotrophic lateral sclerosis. Intervention: A rare-disease adapted cognitive behavioural therapy (CBT) intervention, was delivered to both caregivers and people living with amyotrophic lateral sclerosis. The intervention was delivered by psychologists in the format of 1-hour long sessions. Over a 16-week period, participants received 5 to 10 sessions. Sessions were delivered individually or in dyads and content were tailored according based on participants' needs. Objective: To compare the effects of cognitive behavioural therapy versus usual care on caregivers' overall health, quality of life, psychological distress (i.e., depression and anxiety), and caregiver strain.	Usual Care	Family (caregiver and patient) face-to-face, delivered by professionals.	Not Reported	There were three caregiver outcomes, two of which were significant. There were statistically significant differences favoring the intervention groups on the Health Survey Short Form "mental component summary" ($p < 0.05$) and the Caregiver Strain Index ($p < 0.05$). No significant between group difference was found on the Hospital Anxiety and Depression Scale scores ($p>.05$).	Depending on the patients' and caregivers' perceived problems, sessions were held individually (patient or caregiver) or together (patient and caregiver). Sessions were also tailored to the needs of the patient and caregiver.	Not Reported

Quasi-Experimental Studies

Mioshi 2013 Australia	Frontotemporal Dementia	**Structured Group Program:** N= 9 Mean Age: 59 Percent Female: 100% **Control:** N= 12 Mean Age: 63 Percent Female: 67%	Population: Caregivers to those with frontotemporal dementia. Intervention: A generic intervention, involving teaching of cognitive appraisals and coping strategies. The intervention was delivered weekly by two researchers specialized in frontotemporal dementia, a total of 15 weeks. Each session lasted 2 hours and involved caregivers being taught to identify stressors, develop a coping strategy, and seek support. Objective: To decrease caregiver burden and reaction to patient behaviour, and psychological distress (i.e., depression, anxiety, and stress) through coping strategies such as problem-solving.	Treatment as usual.	Group face-to-face, delivered by professionals.	Caregivers reported learning problem-solving strategies, allowing for a higher sense of control and self-efficacy.	There were four caregiver outcomes, three of which were significant. Only caregivers in the intervention had statistically significant reduction in burden as measured by the Zarit Burden Interview ($p < 0.05$), their reaction to challenging patient behavior as measured by the Cambridge Behavioral Inventory-Revised ($p < 0.05$), increase in the Coping Skills Questionnaire humour subscale ($p < 0.05$). No significant difference between pre and post intervention scores were found for depression, anxiety, and stress ($p > 0.05$) as measured by the Depression, Anxiety, and Stress Scale. A greater number of caregivers in the intervention group increased their functional response scores during problem-solving tasks (63%) than caregivers in the control group (13%).	Not Reported	Twelve caregivers were unable to attend because of travelling distance, ill health, or inability to arrange alternative caregivers in their absence.
Rexilius 2002 USA	Autologous Hematopoietic Stem Cell	**Overall Mean Age:** 52 **Healing Touch Group:** N= 10 Percent Female: 90% **Massage Therapy Group:** N= 13 Percent Female: 85%	Population: Caregivers to those with autologous hematopoietic stem cell. Intervention: A generic massage therapy and healing touch therapy intervention. A total of six 30-minute sessions were provided over a 3-week period by a certified massage therapist or certified healing touch practitioner. Objective: To evaluate its feasibility and to reduce reported levels of anxiety, depression,	A researcher visited for 10 minutes twice a week and asked "How are you doing?"	Individual face-to-face, delivered by professionals.	Caregivers expressed that the intervention provided time off from caregiving (62%), helped them feel relaxed (54%), and energized (8%). In the healing touch intervention, most caregivers expressed that the intervention provided time to	There were eight caregiver outcomes compared within groups. For five outcomes, caregivers in both intervention groups showed a statistically significant decrease in the Beck Anxiety Inventory ($p = 0.03$) as compared to the control group. Caregivers in the massage therapy group had significantly lower Center for Epidemiological Studies Depression Scale scores (p	Sessions were delivered in the hospital to reduce transportation issues.	Caregivers found scheduling difficult as they reported feeling obligated to stay with the patient. They also described being bothered by external noises during the healing touch intervention (e.g., doors

		Control Group: N= 13 Percent Female: 46%	fatigue and perceived caregiver burden.			be alone and relax. Caregivers also reported relief from pain (20%).	= 0.002) than the control group, and the Multidimensional Fatigue Inventory-20 components: general fatigue (p = 0.03), reduced motivation (p = 0.02), and emotional fatigue (p = 0.004) were substantially improved among the massage group as compared to the intervention group. Three outcomes did not show a statistically significant reduction between the intervention and control group for the Multidimensional Fatigue Inventory-20 components: physical fatigue, activity, and in the Subjective Burden Scale (all p > 0.05).		opening and closing).

Pre-Post Studies

Aoun 2015 Australia	Motor Neuron Disease	N=18 Mean Age = 60 Percent Female = 72%	Population: Caregivers to those with motor neuron disease. Intervention: A generic dignity therapy intervention involving discussing issues that are important toward the end of life, was administered. A psychologist delivered 3-7 sessions of in-person dignity therapy to caregivers and their family member with motor neuron disease. Objective: To test the acceptability and feasibility of dignity therapy and its effect on caregiver burden, depression, anxiety, and hopefulness.	Not Applicable	Family, caregivers, and patients face-to-face, delivered by professionals.	Caregivers reported that dignity therapy was helpful for their family member (89%), they would recommend dignity therapy to other patients/caregivers (78%), the generativity document will continue to offer them and their family comfort (72%), dignity therapy was as important as any other aspect of health care (60%), and dignity therapy helped prepare them for	There were four caregiver outcomes, none of which showed statistical significance. No statistically significant decrease was found between pre and post scores on the Zarit Burden Interview, Hospital Anxiety and Depression Scale anxiety or depression subscales, or the Herth Hope Index (all p>.05).	Not Reported	Caregivers stated that they wished the intervention would have been done earlier after the diagnosis. Authors of this article reported that the intervention was costly (e.g., therapists travel, transcribers).

						their partner's end of life (50%).			
Bevans 2010 USA	Allogeneic Hematopoietic Stem Cell Transplantation	N= 8 Mean Age = 54 Percent Female = 63%	Population: Caregivers to those with allogeneic hematopoietic stem cell transplantation. Intervention: A rare-disease adapted social problem-solving therapy, which focuses on topics related to problem orientation and problem-solving skills was administered. This intervention was delivered to patient-caregiver dyads by a clinician. Dyads completed a total of 4 sessions before, during and after Allogeneic Hematopoietic Stem Cell transplant. Objective: To evaluate the feasibility of the intervention and any changes in problem-solving skills, psychological distress, family functioning, and symptom distress.	Not Applicable	Family, caregivers, and patients face-to-face, delivered by professionals.	Participants reported being very satisfied with the individual sessions. Participants noted the intervention provided: "an opportunity to talk", "expert information", "creative thinking", and forced the dyads to communicate.	There were three caregiver outcomes, none of which were statistically significant. There was no statistically significant change between pre and post test scores on the Social Problem-Solving Inventory-Revised, the Symptom Distress Scale, or the Brief Symptom Inventory (all $p > 0.05$).	Not Reported	Caregivers described the home care guide as being too long and repetitive to information provided from usual care. Clinicians reported that limited caregiver availability for sessions was challenging.
Bevans 2014 USA	Allogeneic Hematopoietic Stem Cell Transplantation	N= 72 Mean Age = 53 Percent Female = 72%	Population: Caregivers to those with allogeneic hematopoietic stem cell transplantation. Intervention: A rare-disease adapted intervention that focused on problem-solving skills and problem-solving education was delivered to both caregivers and patients receiving an Allogeneic Hematopoietic Stem Cell Transplantation. The intervention included three 1-hour sessions delivered by nurses and social workers with the Objective: To improve self-efficacy and reducing distress.	Not Applicable	Family, caregivers, and patients face-to-face, delivered by professionals.	Not Reported	There were two caregiver outcomes, both of which resulted in statistically significant improvement. A statistically significant increase was found in Cancer Self Efficacy Scale-transplant scores ($p = 0.03$; $d = 0.27$) and a statistically significant decrease was found in the 18-item Brief-Symptom Inventory scores ($p = 0.01$; $d = 0.40$) when comparing pre-intervention to post-intervention scores.	Not Reported	Not Reported
Cox 2012 USA	Acute Lung Injury	**Study 1:** N= 23 Mean Age = 66 Percent Female = 57% **Study 2**:	Population: Caregivers to those with acute lung injury. Intervention: A rare-disease adapted intervention, focused on coping skills training, and was	Not Applicable	Family, caregivers, and patients, telephone-delivered by professionals.	All caregivers reported applying the coping skills they learned. They reported that the skills were useful, especially with	There were three caregiver outcomes. There was an improvement reported for all three measures when comparing pre-to-post intervention scores on the Hospital Anxiety and	The intervention was telephone-based, making it less expensive, more likely to be covered by insurance and health	Not Reported

		N= 7 Mean Age = 52 Percent Female = 57%	developed to teach caregivers how to help their loved one apply and implement coping strategies to help manage acute lung injury. This was delivered to patient-caregiver dyads by a clinician over 12 weeks with sessions being delivered by phone for 30-minute telephone sessions that included patient-caregiver dyads. Objective: To evaluate the feasibility and acceptance of the coping skills intervention, and the impact on psychological distress (i.e., symptoms of anxiety, depression, and post-traumatic stress).			issues concerning stress and disabilities.	Depression Scale, scores on the Post-Traumatic Symptom Scale, and self-efficacy scores. Significance was not tested.	systems, and did not require caregivers to travel.	
Fidika 2015 Germany	Cystic Fibrosis	N= 23 Mean Age = 37 Percent Female= 91%	Population: Parents of children with cystic fibrosis. Intervention: A rare-disease adapted intervention involving web-based psychological support. This 9-week intervention involved caregivers engaging in cognitive behavioural writing therapy, once a week for 45 minutes. Caregivers wrote about a topic and received feedback from psychotherapists within 48 hours. Three main treatment topics covered were: anxiety and fear-related thoughts, sharing care responsibility, and increasing self-care Objective: To evaluate the feasibility of this web-based intervention, and determine if it resulted in improved coping skills and quality of life by reducing levels of anxiety, fear, and depression.	Not Applicable	Individual, online-delivered by professionals.	Not Reported	There were five caregiver outcomes, all of which were statistically significant when comparing pre to post test scores. Significant reductions in symptoms of anxiety as assessed by the Hospital Anxiety and Depression Scale ($p = 0.01$; $d = 2.06$), depressive symptoms as assessed by the Center of Epidemiologic Studies Depression Scale ($p = 0.02$, $d = 0.72$), and fear of disease progression as assessed by the Fear of Progression questionnaire ($p < 0.01$; $d = 1.11$). A statistically significant increase was found in caregivers' quality of life as assessed by the Ulm Quality of Life Inventory for Parents of Chronically Ill Children total score ($p = 0.01$; $d = 0.76$) and its domains, including emotional stability ($p < 0.01$; $d = 1.49$), self-development ($p = 0.02$; $d =$	Not Reported	One participant reported problems with the setting and structure of the program.

							0.62), and well-being (p = 0.01; d = 0.83), and in caregivers' coping skills as assessed by the Coping Health Inventory for Parents social support, self-esteem, and psychological stability components (p = 0.04; d = 0.51).		
Qualitative Studies									
Marconi 2016 Italy	Amyotrophic Lateral Sclerosis	N= 18 Mean Age= 58 Percent Female = Not Reported	Population: Caregivers to those with amyotrophic lateral sclerosis. Intervention: Rare-disease adapted meditation sessions delivered by two professionals on a weekly-basis, for a total of 8 weeks. Each session emphasized accepting the discomfort and physical limitations associated with the disease and focusing instead on available resources and abilities. Objective: To promote acceptance and increase quality of life.	Not Applicable	Caregiver and patient face-to-face, delivered by professionals.	Caregivers reported improvements in well-being, relaxation, emotional self-regulation, acceptance, consciousness, breathing, sleeping, and relationships. The group setting was described as helpful to raise awareness about the care recipient-caregiver dynamic, and improve feelings of support. Participants also reported having better coping skills in relation to anxiety and depression. All participants expressed a will to continue with the intervention if possible.	Not Reported	Not Reported	Not Reported
Moola 2016 Canada	Cystic Fibrosis	N= 8 Mean Age= 37 Percent Female = Not Reported	Population: Caregivers to those with cystic fibrosis. Intervention: A rare-disease adapted “Cystic Fibrosis Chatters” intervention was provided. A total of four 90-minute in person	Not Applicable	Caregiver and patient face-to-face, delivered by professionals.	Caregivers described the intervention as being highly beneficial, and ideally should be incorporated into	Not Reported	Not Reported	Not Reported

		sessions were delivered over a 4-month period. All sessions involved counseling that was grounded in empathic listening, dialogue, health-related goal setting, and sessions were influenced by cognitive behavioural, behavioural self-regulation, and motivational interviewing techniques. The goal of this study was to determine how caregiving-related thoughts, feelings, behaviours, health, and overall quality of life are impacted by the counselling program. Objective: To test the feasibility of this caregiver counselling intervention for the cystic fibrosis population.	routine cystic fibrosis care. They appreciated being asked about their feeling and liked that it was relaxing and not rushed. Participants reported benefiting from their relationship with the therapist, as they felt a strong sense of rapport and mentorship which helped improve mental wellbeing. Caregivers also reported improved time management skills. Participants reported wanting to continue with the counseling sessions after the intervention had been terminated.

1. Total sample mean age provided only.

Table 2. Study Characteristics and Interventions – Support Focused Interventions

First Author, Year Country	Type of Disease	Demographics of Included Caregivers	Population, Intervention Description, Objectives	Control Group Intervention	Intervention Delivery Format	Intervention Use and Satisfaction	Tested Benefits	Facilitators	Barriers
Trials									
Sheija 2005 India	Spinal Cord Injury	**Support Groups:** N= 19 Mean Age: 33 Percent Female: 100% **Control:** N= 17 Mean Age: 39 Percent Female: 88%	Population: Spouses caregiving to those with spinal cord injury. Intervention: Generic support groups ("RISE UP") were delivered to spouses of people with spinal cord injury. A total of seven 1-hour long support group sessions were held. Support groups took place over a 2-week period. Each session covered a core topic, which was discussed and explained through various techniques (e.g., role play) and ended with a relaxation activity. Topic-related home tasks were also assigned to caregivers. Objective: To improve upon the overall health, depression and anxiety levels, and quality of life of caregivers.	No Group Sessions	Group, face-to-face, with a professional.	Not Reported	There were seven caregiver outcomes, all of which had statistically significant differences between the intervention and control groups on the 12-item General Health Questionnaire ($p = 0.02$; $d = -0.8$), Hospital Anxiety and Depression Scale anxiety ($p < 0.01$; $d = -1.03$) and depression ($p < 0.01$; $d = -2.02$) components, and the World Health Organization Quality of Life physical ($p < 0.01$; $d = 1.17$), psychological ($p < 0.01$; $d = 1.85$), social relation ($p < 0.01$; $d = 1.3$), and environmental ($p < 0.01$; $d = 1.01$) components.	Caregivers were stratified into groups based on their language preferences.	Not Reported
Qualitative									
Cipolletta 2018 Italy	Amyotrophic Lateral Sclerosis	**Partners:** N= 6 Mean Age: 66 Percent Female: 44% **Children:** N= 6	Population: Caregivers to those with amyotrophic lateral sclerosis. Intervention: Generic support groups were held, one for caregivers that were partners and one for caregivers that were adult children. A total of 10 support	Not Applicable	Group, face-to-face, with a professional.	All caregivers reported being satisfied with the support groups and said that they would recommend them to other caregivers. Caregivers stated that the support group sessions	Not Reported	Having support groups arranged by a supervisor or conductor was helpful as opposed to having peers arranging meetings.	Caregivers found it hard to participate in the support groups due to time management issues. Travel was also a barrier and

		Mean Age: 42 Percent Female: 83%	group sessions were provided, each lasting 1.5 hours. Support groups were facilitated by psychologists and sessions did not have predetermined topics as to better accommodate to the caregivers' needs. Objective: To explore whether differences between family caregivers who are partners or children of people with amyotrophic lateral sclerosis may differ and to integrate the role of caregiving within their lives.			were very helpful and allowed them to share fears and concerns with others going through a similar experience. Caregivers also reported feeling less alone. Caregivers also expressed benefits from listening to other caregivers' experiences.			many caregivers wished sessions were offered closer to their area so as to meet and connect with people of the same community.
Stewart 2001 Canada	Hemophilia and HIV/AIDS	N=4 Mean Age = Not Reported Percent Female = 75%	Population: Caregivers to people living with hemophilia and HIV/AIDS. Intervention: A rare-disease adapted support focused intervention provided informational, affirmational, and emotional support for caregivers. Telephone delivered support group sessions were delivered weekly for a total of 12 weeks. Sessions lasted on average 105 minutes. During each session, participants were encouraged to freely discuss any topic of their choice related to caregiving and the disease. At the end of the session, facilitators suggested coping strategies and other sources of social support. Objective: To evaluate the feasibility of the intervention, better understand problems encountered by caregivers, and reduce feelings of loneliness by increasing social support.	Not Applicable	Group, by telephone, delivered by professionals.	Caregivers found the intervention to be a source of support by helping diminish feelings of isolation and loneliness, and improving communication, relationships, and confidence. Caregivers felt like the intervention provided affirmational, informational, and emotional support.	Not Reported	Group members received written information about teleconferencing and instructions for re-joining the teleconference if they had to leave for part of a meeting or were disconnected.	All of the participants were disappointed with the length of the intervention (12 weeks) and wished it was longer.

Table 3. Study Characteristics and Interventions – Educational Interventions

First Author, Year Country	Type of Disease	Demographics of Included Caregivers	Population, Intervention Description, Objectives	Control Group Intervention	Intervention Delivery Format	Intervention Use and Satisfaction	Tested Benefits	Facilitators	Barriers
Pre-Post									
Alankaya 2015 Turkey	Amyotrophic Lateral Sclerosis	N=30 Mean Age = Not Reported Percent Female = 90%	Population: Caregivers to those with amyotrophic lateral sclerosis. Intervention: Rare-disease specialized educational intervention was provided based on caregivers' self-identified needs. Using a one-to-one card sort, caregivers selected 3 areas where they needed help. A rare disease specialized caregiver booklet and an individual educational power-point session was delivered to caregivers based on their identified needs. Objective: To decrease caregiver burden and improving quality of life.	Not Applicable	Group, face-to-face, patients and caregivers, delivered by professionals.	Not Reported	There were two caregiver outcomes, including one that showed a statistically significant improvement and one that did not. A statistically significant decrease was found between pre-test and post-test scores on the Caregiver Burden Interview ($p = 0.01$, $d = 0.28$). There was no statistically significant difference found between pre and post test scores on the Duke Health Profile ($p = 0.12$, $d = -0.39$).	Not Reported	Not Reported
Qualitative									
Gormley 2014 UK	Cystic Fibrosis	N=50 Mean Age = Not Reported Percent Female = Not Reported	Population: Parents to children living with cystic fibrosis. Intervention: A rare-disease specialized educational intervention was delivered at an annual educational event, in which several disease specific tools were provided, such as 30-minute group presentations, small-group discussions, and question-and-answer sessions with a multidisciplinary team.	Not Applicable	Group, face-to-face, delivered by professionals.	All caregivers reported being satisfied or very satisfied with the topics covered, presentation styles, and hospital location (100%). The majority of caregivers also reported being satisfied or very satisfied with the duration (92%) and day of the event (84%). Following the intervention, caregivers also described benefiting from feeling less isolated	Not Reported	Not Reported	Participants reported an issue with the start time (6pm) and reported that this was too early and too rushed for people working. Caregivers also reported that it would have been helpful to have information in advance about the event program,

	Objective: To evaluate the educational programs offered and increase future participation in the program.	and having contact with others who understand the difficulty of caregiving.	and have ice-breaker activities to promote conversation among caregivers

Table 4. Study Characteristics and Interventions – Multi-Component Interventions

First Author, Year Country	Type of Disease	Demographics of Included Caregivers	Population, Intervention Description, Objectives	Control Group Intervention	Intervention Delivery Format	Intervention Use and Satisfaction	Tested Benefits	Facilitators	Barriers
Trials									
Elliott 2009 USA	Spinal Cord Injury	**Problem Solving Training:** N= 30 Mean Age: 42 Percent Female: 90% **Control:** N= 30 Mean Age: 43 Percent Female: 73%	Population: Caregivers of individuals with spinal cord injury. Intervention: A rare-disease adapted multi-focused intervention was delivered to enhance problem-solving skills for caregivers through three face-to-face sessions provided over the first year of caregiving. Sessions focused on rare-disease specific problem-solving and were individualized based on each caregiver's needs as identified through a card sort activity completed in session one. Sessions lasted 2-3 hours. Caregivers also received booster sessions on an as-needed basis and were mailed additional disease adapted educational information if needed. Objectives: To decrease the use of dysfunctional problem-solving styles, improve health-related quality of life, and decrease symptoms of depression.	Usual access to outpatient clinic staff and information provided by rehabilitation staff concerning spinal cord injury management.	Individual face-to-face and home based delivered by professionals.	Not Reported	There were three caregiver outcomes, one of which was significant. A statistically significant difference between groups was found in dysfunctional problem-solving as measured by the Social Problem Solving Inventory-Revised ($p < 0.05$; $d = -0.94$) in favour of the intervention group. No statistically significant difference between groups was found when comparing scores on the Inventory to Diagnose Depression ($p > 0.05$), or the Short Form Health Survey-36 "physical functioning" ($p = 0.07$; $d = 0.73$) and "social functioning" ($p = 0.09$; $d = 0.44$) components.	Not Reported	Not Reported
Li 2017 China	Human Immunodeficiency Virus	**Together for Empowerment Activities:** N= 237 Mean Age: 41 Percent Female: 68% **Control:** N= 238 Mean Age: 43 Percent Female: 62%	Population: Caregivers of individuals with human immunodeficiency virus. Intervention: A rare-disease specialized multi-focused intervention, Together for Empowerment Activities, was provided to people living with human immunodeficiency virus and their family members. Caregivers participated in group sessions over the course of two months. Sessions involved interactive group	China's standard of care: three weekly didactic group sessions that focused on healthy daily routines, antiretroviral drug adherence and side effects, nutrition, and personal and family hygiene.	Family, caregivers and patients, group-based, face-to-face delivered by professionals.	Not Reported	There were two caregiver outcomes, one of which was significant. Statistically significant differences were found between the intervention and control groups on the Zung Self-Rating Depression Scale ($p < 0.01$). No statistically significant differences were found between the intervention and control groups on the Perceived Caregiver Burden Scale ($p > 0.05$).	Not Reported	Not Reported

			activities, such as games, role play, blanket-making, and talent shows. Ten reunion sessions were also provided. Objectives: To improve human immunodeficiency virus-related challenges at the individual, family, and community level.						
Mazanec 2017 USA	Multiple myeloma	**Overall:** N=12 Mean Age = 55 Percent Female = 73%	Population: Caregivers of individuals with multiple myeloma. Intervention: A general behavioural and educational focused intervention was delivered to patients and their caregivers where they were provided psychoeducation about living with a chronic illness, information booklets, and links to the American Cancer Society Website. The behavioural component of the intervention involved home-based walking to be done independently or as a dyad. A step goal based on national guidelines was provided as was a pedometer and a calendar to reinforce walking activity. Objectives: To assess the feasibility of the intervention and to obtain preliminary data on the effectiveness of the intervention on anxiety, activation for self-management, fatigue, depression and health-related quality of life in patients with multiple myeloma and their caregivers.	The control group received the same educational resources as the intervention group, but were not given pedometers, counseling, or tailored instructions for walking activity.	Families, caregiver and patient, face-to-face and home-based delivered by professionals.	Caregivers in the intervention group reported being highly satisfied with the intervention. Out of a maximum score of 10 for acceptability, the mean score for caregivers was 8.8 (standard deviation = 1.2). Follow-up interviews revealed that the component that caregivers found the most beneficial was the relational aspect of the intervention.	There were eight caregiver outcomes, none of which were significant. Score improvements were not higher for the intervention group compared to the control group on the Patient-Reported Outcomes Measurement Information System, short forms of depression, anxiety, fatigue and Health Related Quality of Life, the Caregiver Patient Activation Measure, and the "Caring for Oneself" subscale of the Caregiver Inventory.	To promote the use of the educational and walking program, participants received booster telephone calls.	There was a very high drop-out rate due to scheduling challenges, time limitations, treatment demands, emotional distress caused by the diagnosis, and lack of interest.
Raj 2015 USA	Traumatic Brain Injury	**Overall:** Percent Female: 95% **Internet-based Interacting Together Everyday:**	Population: Caregivers to children living traumatic brain injury. Intervention: A rare-disease specialized multi-focused intervention in the format of a web-based program, Internet-based Interacting Together Everyday: Recovery After Childhood TBI (I-	Website with links to many available resources (e.g., support groups, services for children with disabilities).	Individual face-to-face, online, and in person, delivered by professionals.	Not Reported	There were four caregiver outcomes, none of which were significant. No statistically significant differences were found between the TBI intervention group and the control group on the Global Severity Index of the Symptom Checklist-90-Revised, Center	Information package and website instructions were provided. Computers and high speed internet were also provided/refunded	Not Reported

		Recovery After Childhood TBI: N=20 Mean Age: 33 **Control:** N=17 Mean Age: 33	InTERACT), involved training parents in stress management, anger control, and provided education about pediatric TBI. Ten core sessions, and up to four additional sessions were delivered over a 4 to 6 month. Sessions were initially scheduled weekly, and later occurred bimonthly. An initial face-to-face session was given to orient participants with the program, and remaining sessions were delivered online. The first part of each session consisted of a self-guided online intervention addressing concerns related to the disease (e.g. behavior management following traumatic brain injury, dealing with anger, establishing rules). The second portion of each session was a videoconference call with the therapist, where caregivers discussed the topic learned in the online session. Objective: To reduce distress, depression, parenting stress, and increase self-efficacy.				for Epidemiological Studies Depression Scale, the Parenting Stress Index, and the Caregiver Self-Efficacy Scale (p >0.05).	if not available to caregivers. Supplemental sessions were provided if needed.	
Schulz 2009 USA	Spinal Cord Injury	**Dual Treatment:** N= 57 Mean Age: 51 Percent Female: 84% **Caregiver-Only Intervention:** N=56 Mean Age: 54 Percent Female: 75% **Control:** N= 60 Mean Age: 53 Percent Female: 68%	Population: Caregivers of people with spinal cord injury. Intervention: A rare-disease adapted multi-focused intervention which implemented cognitive behavioural strategies. The caregiver-only intervention was delivered over a 6-month period in the format of seven 60-90 minute sessions. Five sessions were delivered at home, and two of them were delivered by phone. In addition, five phone support group sessions with other caregivers were facilitated. A spinal cord injury information booklet and an information telephone system were also provided for participants to practice learned skills at home. Sessions focused on topics related	Information and three “check-in” telephone calls.	Individual or with caregivers and patients, face-to-face and by telephone, with a professional.	Not Reported	There were six caregiver outcomes, two of which were significant. There were also five caregiver-care recipient outcomes, one of which was significant. Caregivers in the intervention group reported significant improvement compared to the control group in health symptoms ($p = 0.01$, $d = -0.24$) and social integration ($p < 0.01$, $d=0.84$). No statistically significant differences were found between groups ($p > 0.05$) on the Zarit Burden Interview, the Center for Epidemiological Studies Depression Scale, self-care problems, and satisfaction with support. Caregivers and	Caregivers were provided with a screen phone and a notebook that contained standard information on spinal cord injury, the aging process, caregiving and community resources, and instructions on how to use the screen phone.	Not Reported

			to environmental and personal stress, health and self-care, access to formal and informal support, and emotional well-being. The caregiver component of the dual-target intervention was identical to the caregiver-only intervention, however, the intervention was adapted to also address care recipients' needs. Objectives: To improve levels of emotional well-being, communication, self-care, physical health, and social support.				care recipients in the dual intervention reported significant improvement compared to the control group on health symptoms ($p = 0.01$, $d=-0.50$). There were no statistically significant differences between the groups on the Center for Epidemiological Studies Depression Scale, self-care problems, satisfaction with support, and social integration.		
Wade 2012 USA	Traumatic Brain Injury	**Teen Online Problem-Solving:** N= 21 Mean Age: 41 Percent Female: Not Reported **Control:** N= 20 Mean Age: 42 Percent Female: Not Reported	Population: Caregivers of teenagers living with traumatic brain injury. Intervention: A rare-disease adapted multi-focused intervention, teen online problem-solving (TOPS), was delivered. The intervention provided a total of 10 core web-based sessions delivered over a 6-month period. The first session, however, was delivered in person. All sessions were divided in two portions, each lasting 45-60 minutes. The first part consisted of individual disease specific self-guided problem-solving skills, information, and interactive exercises. The second part involved reviewing the online content with a therapist. Supplemental sessions addressing less common problems or specific caregiving problems were also provided if requested. Objective: To test the efficacy of the online intervention and improve social problem-solving skills, distress, and caregiver depression.	Control caregivers were encouraged to use 1 hour more of web links to brain injury associations and educational resources.	Families, caregiver and patient, online and in-person delivered by professionals.	Caregivers in the intervention group rated the intervention as being helpful and enjoyable. Caregivers also felt they had more knowledge about the disease, understood their child better, and felt less stressed after the intervention. Caregivers rated certain aspects of the intervention as moderately to extremely helpful, including materials on problem solving, communication, behaviour management, and anger control. Caregivers found the intervention to neither be too short or too long (80%).	There were three caregiver outcomes, none of which were significant. No differences between the intervention and control groups were found ($p > 0.05$) on the Social Problem Solving Inventory - Revised Short Form, the Center for Epidemiological Studies Depression Scale, or the Global Severity Index of the Symptom Checklist.	All participating families were provided with high speed internet access, when available. Any family without an existing home computer was given one to use for the duration the study. Participants were trained for the online-program utilization and supplemental sessions addressing family concerns were given if needed. The telehealth approach also reduced travel time and fuel costs.	Unexpected technical difficulties with online software (e.g., sound issues with some of the video content).
Pre-Post Studies									
A'Campo 2012 Netherlands	Huntington's Disease	N=28 Mean Age = 56	Population: Caregivers to those with Huntington's Disease.	Not Applicable	Caregivers, and patients group-based,	Caregivers described the intervention as a	There were four caregiver outcomes, including one that showed a statistically	Not Reported	More than 33% of caregivers described the

		Percent Female = 57%	Intervention: A rare-disease adapted education program, the Patient Education Program for Huntington's disease (PEP-HD), was delivered in eight group sessions through two 90-minute weekly sessions. The education sessions covered several topics such as social support, self-monitoring, relaxation techniques, stress management, and information seeking. Objective: To improve caregivers' quality of life and coping strategies.		face-to-face, delivered by professionals.	positive experience that was useful in daily life. Most caregivers rated the timing of the intervention positively. Additionally, caregivers reported that the content of the intervention was easy to understand and rated the "stress management" session as most valuable.	significant improvement, and two that did not. A significant decrease between pre and post test scores was found in psychosocial burden on the Belastungsfragebogen Parkinson Angehörigen kurzversion questionnaire ($p = 0.02$; $d = -0.32$). No statistically significant differences were found in pre and post scores on the Hospital Anxiety and Depression Scale, the 36-item Short Form Health Survey Questionnaire, and the Utrecht Coping List ($p > 0.05$).		intervention as tiresome and 19% of caregivers reported that the program was difficult to follow.
Bozkurt 2014 Turkey	Osteogenesis Imperfecta	N= 16 Mean Age = 35 Percent Female = 69%	Population: Caregivers to those with osteogenesis imperfecta. Intervention: A rare-disease specialized psychoeducational intervention focused on enhancing psychosocial adjustment through ten 3-hour weekly group sessions. Each session included emotional support, coping skills, and education about the disease. The intervention targeted caregivers by attempting to improve coping and problem-solving strategies. Objective: To improve coping and problem-solving strategies.	Not Applicable	Caregivers, group-based, face-to-face, delivered by professionals.	Caregivers (94%) reported receiving current and adequate information during the intervention. Caregivers (75%) also reported that they experienced positive changes including psychological changes (56%), social changes (50%), and economic difficulties (13%) due to the disease, after receiving the intervention. There was also, however, an increase in physical difficulties reported by caregivers (69%).	There were four caregiver outcomes, three of which resulted in significant improvement. Caregivers showed a statistically significant decrease at follow-up on the Burden Interview ($p < 0.01$, $d = -0.41$) and the Psychosocial Adjustment to Illness Scale-Self Report ($p = 0.01$, $d = -0.42$). A significant decrease was also found on the "helpless" ($p = 0.05$, $d = -0.63$) and "face-saving" ($p = 0.01$, $d = -0.73$) components of the Coping Strategies Scale, however, the "self-confident" ($p = 0.11$; $d = 0.64$), "optimistic" ($p = 0.62$; $d = 0.14$) and "seeking social support" ($p = 0.64$; $d = 0.06$) subscales did not show statistically significant differences between pre-intervention and follow-up. There were no significant differences in follow-up Problem-Solving Inventory scores ($p = 0.72$, $d = -0.21$).	Not Reported	Not Reported
Dunlop 2016 USA	Progressive Supranuclear Palsy	N= 11 Mean Age= Not Reported Percent Female = 64%	Population: Caregivers to those with progressive supranuclear palsy.	Not Applicable	Families, caregiver and patient, delivered by	Five themes emerged as intervention outcomes: (1) enhanced patient	There were two caregiver outcomes, one of which was significant. A significant improvement was found in caregiver pre-test and post-test	Not Reported	Not Reported

			Intervention: A rare-disease specialized multi-focused telehealth nursing intervention, the Cure Progressive Supranuclear Palsy Care Guide, focused on providing educational information and emotional support to families. An individualized intervention was delivered to help caregiver-patient dyads manage the disease based on their specific needs (e.g. providing information on palliative care, introducing them to support groups, suggesting home modifications). Objective: To enhance caregiver knowledge, decrease caregiver strain, and build a support network.		telephone by professionals.	and caregiver knowledge, (2) improved day-to-day management, (3) development of an awareness of resources, (4) decreased dependence on resources, and (5) addressing caregiver needs.	knowledge assessment scores ($p < 0.01$; $d = 1.2$). No statistical significant reduction was found between pre and post Caregiver Strain Index scores ($p > 0.05$).		
Leenaars 2012 Canada	Fetal Alcohol Spectrum Disorder	N= 186 (families) Mean Age = 46 Percent Female = 88%	Population: Families, including caregivers to those with fetal alcohol spectrum disorder. Intervention: A rare-disease specialized multi-focused intervention that aims to help families cope with raising a child diagnosed with fetal alcohol spectrum disorder, the Coaching Families program, was evaluated. The program is individualized based on the needs of each family. Professional mentors help families by providing support, education, advocacy, and referrals in relation to the disease. The length of mentorship varied from months to years, depending on each family's needs, as did the amount of time spent between families and mentors. Objective: To evaluate the mentorship program and determine if it results in reductions in familial stress and needs and increases caregiver self-care and goal-setting strategies.	Not Applicable	Family, caregivers, and patients face-to-face, delivered by professionals.	Caregivers reported that they were satisfied with the intervention (98%) and that they would return to the program if needed (99%). Some caregivers also reported that the intervention helped them parent better (32%), understand their children and the disease better (28%), and feel less stressed, more patient, and more positive (15%). Most caregivers reported that they did not experience any problems with the intervention (66%).	There were three caregiver outcomes, all of which were found to be statistically significant when comparing pre intervention to post intervention ratings. Caregiver needs ($p < 0.01$, n squared = 0.45), and stress significantly decreased ($p < 0.01$, n squared = 0.35) while perceived goal achievement significantly increased following the intervention ($p < 0.01$, n squared = 0.66).	Not Reported	Some caregivers reported that they felt the facilitator did not fully understand what it was like to have a child with the disease.
Rodgers 2007 USA	Spinal Cord Injury and Traumatic Brain Injury	N= 28 Mean Age = 47 Percent Female = 86%	Population: Caregivers to those with a spinal cord injury or brain injury.	Not Applicable	Family, caregivers, and patients, face-to-face	Participants in the traumatic brain injury group expressed positive	There were six caregiver outcomes, one of which was significant. A statistically significant decrease was found	Socialization time was added for all participants.	Not Reported

			Intervention: A rare-disease adapted multi-focused intervention, multiple-family group treatment, was delivered to caregivers and individuals with either a traumatic brain injury or a spinal cord injury. The intervention began with individual sessions with each dyad, and then educational workshops were provided. Lastly, the intervention included 90-minute group sessions which included 4-8 families, led by two clinicians over a 12-18 month period. Sessions were delivered bi-monthly or monthly, and focused on problem-solving skills related to socialization, identifying problems, and finding solutions to these problems. Objective: To reduce caregiving burden, depression, and anger, and increase quality of life, social support, and coping skills.		delivered by professionals.	feelings related to the intervention, especially regarding meeting others going through the same experience, learning useful information, and learning new coping strategies. Caregivers also expressed having a better sense of organization, improved ability to express their feelings and set limits. Participants in the spinal cord injury group expressed learning coping strategies and skills in managing medical complications. In addition, participants learned more about caregiver burden, and along with their caregivers, developed strategies to better manage it. Participants expressed feeling less isolated as a result of the intervention, and were grateful for the information learned.	over time on the Caregiver Burden Inventory ($p < 0.01$). There were no significant differences found over time ($p > 0.05$) on the Interpersonal Support Evaluation List, revised Ways of Coping checklist, Quality of Life Interview, the Center for Epidemiologic Studies Depression Scale–Global Distress, and the Anger-Expression Inventory.		
Rotondi 2005 USA	Traumatic Brain Injury	N= 17 Mean Age = 46 Percent Female= 100%	Population: Wives of spouses with traumatic brain injury. Intervention: A rare-disease specialized multi-focused intervention, Web Enabled Caregiver Access to Resources and Education (WE CARE), which involved online access to resources and education, was provided for 6-	Not Applicable	Individual, online-delivered by professionals.	The intervention components rated as most useful were the support group, community resource library, and questions and answers library. Caregivers rated the intervention as being	Not Reported	Computers and internet access were provided if needed. If people wanted to continue using the intervention after the 6 months, participants were helped to find free	Not Reported

			months. The online resource included modules based on needs of caregivers of people living with a traumatic brain injury. The software offered an online support group, question and answer forum, a reference library, and a list of community events and resources. Objective: To evaluate the feasibility of the intervention and improve caregiver quality of life.			extremely wonderful (100%) and not at all difficult (75%). In addition, caregivers rated the intervention as moderately to extremely easy to use (84%), very to extremely satisfying (75%), and very to extremely helpful (75%). Caregivers expressed higher levels of acceptance, understanding, support, and increased motivation while using the intervention. Caregivers also expressed lower levels of anger, loneliness, stress, and worry while using the intervention.		or low cost refurbished computers.	
Videaud 2012 France	Posterior Cortical Atrophy	N= 4 Mean Age =Not Reported Percent Female= 50%	Population: Caregivers to those living with posterior cortical atrophy. Intervention: A rare-disease adapted multi-focused psychoeducational intervention delivered over a 1 year period. Six 2-hour long sessions were held every two months. Home-based sessions were also held. All sessions included both caregivers and care-recipients as participants, except for session 3, which split up the 2 groups for free discussions on their experience and emotional health. Each session covered a different topic and provided problem-solving skills and solutions to daily problems associated with the disease.	Not Applicable	Caregivers and patients, group, delivered face-to-face, by professionals.	Caregivers reported feeling less isolated after the group sessions and they were able to form friendships.	There were four caregiver outcomes, two of which were significant. Caregivers showed a statistically significant improvement in pre and post test scores in their knowledge about the disease and their anxiety levels. Caregiver quality of life did not improve, and the quality of the caregiver-care recipient relationship worsened following the intervention.	Not Reported	Not Reported

	Objective: To evaluate its feasibility, and to improve overall quality of life, levels of anxiety, perceived knowledge, and caregiver-care recipient relationship.	

Characteristics of Interventions

Sixteen publications described behavioural or psychological interventions (Aoun et al., 2015; Bevans et al., 2010; Bevans et al., 2014; Cox et al., 2012; Dowling et al., 2014; Elliott et al., 2008; Fidika et al., 2015; Langer et al., 2012; Laudenslager et al., 2015; Lindell, 2008; Manne et al., 2016; Marconi et al., 2015; Mioshi et al., 2013; Moola et al., 2017; Rexilius et al., 2002; Van Groenestijn et al., 2015), three publications described support focused interventions (Cipolletta et al., 2018; Sheija & Manigandan, 2005; Stewart et al., 2001), two publications described an educational intervention (Alankaya & Karadakovan, 2015; Gormley et al., 2014), and 13 publications described interventions that involved more than one component (A'Campo et al., 2012; Bozkurt et al., 2014; Dunlop et al., 2016; Elliott & Berry, 2009; Leenaars et al., 2012; Li et al., 2017; Mazanec et al., 2017; Raj et al., 2015; Rodgers et al., 2007; Rotondi et al., 2005; Schulz et al., 2009; Videaud et al., 2012; Wade et al., 2010). Twenty-six interventions (76%) were either adapted (n=17, 50%) (A'Campo et al., 2012; Bevans et al., 2010; Bevans et al., 2014; Cox et al., 2012; Elliott & Berry, 2009; Fidika et al., 2015; Langer et al., 2012; Laudenslager et al., 2015; Manne et al., 2016; Marconi et al., 2015; Moola et al., 2017; Rodgers et al., 2007; Schulz et al., 2009; Stewart et al., 2001; Van Groenestijn et al., 2015; Videaud et al., 2012; Wade et al., 2010) or were specifically developed for the rare disease (n=9, 26%) (Alankaya & Karadakovan, 2015; Bozkurt et al., 2014; Dunlop et al., 2016; Gormley et al., 2014; Leenaars et al., 2012; Li et al., 2017; Lindell, 2008; Raj et al., 2015; Rotondi et al., 2005). The remaining 8 studies (24%) (Aoun et al., 2015; Cipolletta et al., 2018; Dowling et al., 2014; Elliott et al., 2008; Mazanec et al., 2017; Mioshi et al., 2013; Rexilius et al., 2002; Sheija & Manigandan, 2005) delivered generic interventions not adapted for the needs of rare disease caregivers.

Sixteen interventions (47%) were for caregivers only (Bozkurt et al., 2014; Cipolletta et al., 2018; Dowling et al., 2014; Elliott & Berry, 2009; Elliott et al., 2008; Fidika et al., 2015; Gormley et al., 2014; Langer et al., 2012; Laudenslager et al., 2015; Manne et al., 2016; Mioshi et al., 2013; Raj et al., 2015; Rexilius et al., 2002; Rotondi et al., 2005; Sheija & Manigandan, 2005; Stewart et al., 2001), 14 (41%) were for both caregivers and care recipients (A'Campo et al., 2012; Alankaya & Karadakovan, 2015; Bevans et al., 2010; Bevans et al., 2014; Cox et al., 2012; Dunlop et al., 2016; Lindell, 2008; Marconi et al., 2015; Mazanec et al., 2017; Moola et al., 2017; Rodgers et al., 2007; Schulz et al., 2009; Van Groenestijn et al., 2015; Videaud et al., 2012), and 4 (12%) were for families, caregivers, and care recipients (Aoun et al., 2015; Leenaars et al., 2012; Li et al., 2017; Wade et al., 2010). Twenty-two interventions (65%) were delivered in person (A'Campo et al., 2012; Alankaya & Karadakovan, 2015; Aoun et al., 2015; Bevans et al., 2010; Bevans et al., 2014; Bozkurt et al., 2014; Cipolletta et al., 2018; Gormley et al., 2014; Langer et al., 2012; Laudenslager et al., 2015; Leenaars et al., 2012; Li et al., 2017; Lindell, 2008; Manne et al., 2016; Marconi et al., 2015; Mioshi et al., 2013; Moola et al., 2017; Rexilius et al., 2002; Rodgers et al., 2007; Sheija & Manigandan, 2005; Van Groenestijn et al., 2015; Videaud et al., 2012), four interventions (12%) were delivered online (Fidika et al., 2015; Raj et al., 2015; Rotondi et al., 2005; Wade et al., 2010), three interventions (9%) were delivered by telephone (Cox et al., 2012; Dunlop et al., 2016; Stewart et al., 2001), and five interventions (15%) were delivered in with more than one modality (Dowling et al., 2014; Elliott & Berry, 2009; Elliott et al., 2008; Mazanec et al., 2017; Schulz et al., 2009). All interventions were delivered by a professional.

Perceived benefits of rare disease support services for caregivers

All 34 included publications (100%) reported on tested or perceived benefits of caregiving interventions (A'Campo et al., 2012; Alankaya & Karadakovan, 2015; Aoun et al., 2015; Bevans et al., 2010; Bevans et al., 2014; Bozkurt et al., 2014; Cipolletta et al., 2018; Cox et al., 2012; Dowling et al., 2014; Dunlop et al., 2016; Elliott & Berry, 2009; Elliott et al., 2008; Fidika et al., 2015; Gormley et al., 2014; Langer et al., 2012; Laudenslager et al., 2015; Leenaars et al., 2012; Li et al., 2017; Lindell, 2008; Manne et al., 2016; Marconi et al., 2015; Mazanec et al., 2017; Mioshi et al., 2013; Moola et al., 2017; Raj et al., 2015; Rexilius et al., 2002; Rodgers et al., 2007; Rotondi et al., 2005; Schulz et al., 2009; Sheija & Manigandan, 2005; Stewart et al., 2001; Van Groenestijn et al., 2015; Videaud et al., 2012; Wade et al., 2010). Possible benefits that were described most often were related to statistically significant improvements in emotional states (e.g., reduced symptoms of stress) and reductions in caregiver burden and qualitative descriptions of helpfulness of the delivered interventions.

Thirteen unique themes of tested (statistically significant) and perceived (narratively described) benefits were identified from quantitative and qualitative study results (see Table 5). Seven themes were identified through both qualitative descriptions of perceived benefits and quantitative tests of benefits (i.e., improvements in: physical health, dyadic relationship, existential concerns, emotional states, general skills, self-efficacy, knowledge), while two were only reported via quantitative findings (i.e., burden, quality of life and well-being) and four via qualitative descriptions (i.e., resources, social relationships, support, financial stability) (see Table 5 and Table 6).

Table 5. Themes of Tested and Perceived Benefits, Barriers, and Facilitators of Initiating and Maintaining Interventions for Caregivers (n=34)

	Benefits		Barriers		Facilitators	
	Qualitative	**Quantitative**	**Qualitative**	**Quantitative**	**Qualitative**	**Quantitative**
Behavioural / Psychological Interventions	Physical health Existential concerns Resources General skills Dyadic relationship Isolation Emotional states Social relationships Support Self-efficacy	Self-efficacy Emotional states Burden Existential concerns Quality of life & well-being General skills Dyadic relationship	Intervention misaligned to caregiver needs Practical barriers Ability to make time for intervention	N/A	Specific characteristics of the intervention Specific characteristics of intervention delivery Providing resources	N/A
Support-Focused Interventions	Social relationships Support General skills Self-efficacy	Quality of life & well-being Physical health Emotional states	Intervention misaligned to caregiver needs Practical barriers	N/A	Specific characteristics of the intervention Providing resources Support provided outside of intervention	N/A

Educational Interventions	Social relationships*	Burden	Ability to make time for intervention Emotional barriers Intervention misaligned to caregiver needs	N/A		N/A
Multi-Component Interventions	Emotional states Financial stability Knowledge General skills Resources Dyadic relationship Social relationships Support	Burden General skills Emotional states Knowledge Quality of life & well-being Physical health	Ability to make time for intervention Intervention misaligned to caregiver needs Practical barriers	N/A	Specific characteristics of the intervention Specific characteristics of intervention delivery Providing resources Support provided outside of intervention	N/A

**Note:* Benefit reported as occurring after the educational intervention was delivered. Caregivers noted that they made relationships with other caregivers after having received the intervention.

Table 6. Benefit, Barriers, and Facilitators of Initiating and Maintaining Interventions for Caregivers (n=34).

Benefits from Quantitative Results

Themes	Sub-Themes	Tested Benefits
Emotional States	Distress Mood	Affect (negative and positive) Anxiety symptoms Depression symptoms Distress Mood Parental distress Perceived stress Psychosocial distress Trait anxiety
Quality of Life & Well-Being	Mental wellbeing Quality of life Social integration	Mental wellbeing Quality of life Quality of life (physical, psychological, social relationships, environmental) Social integration
Burden	Caregiver burden Psychosocial burden	Caregiver burden Psychosocial burden
General Skills	Caregiver needs Coping Goal achievement Problem solving Psychosocial adjustment to illness	Caregiver needs Coping Coping strategies (helpless and face-saving components) Goal achievement Problem solving Psychosocial adjustment to illness
Physical Health	Health	Fatigue Health
Dyadic Relationship (Caregiver and Care Recipient)	Reaction to care recipient behaviour	Reaction to care recipient behaviour
Existential Concerns	Fear of disease progression	Fear of disease progression

Self-Efficacy	Self-efficacy	Self-efficacy
Knowledge	Knowledge of the disease	Knowledge of the disease

Benefits from Qualitative Results

Themes	Sub-Themes	Perceived Benefits
Resources	Available resources	Provided and receive information from an expert Support from intervention documents Awareness of resources Decreased need of resources
Social Relationships	Social connection Relationships with others	Isolation Social connection Loneliness Social changes Relationships with others
Support	Informational support Emotional support	Affirmational support Informational support Emotional support Support
Physical Health	Overall health	Overall health Relief from pain Sleeping Breathing
Financial Stability	Economic stability	Economic stability
Dyadic Relationship (Caregiver and Care Recipient)	Communication between the dyad Understanding between dyad	Communication between the dyad Understanding care recipient point of view Understanding their children Awareness about relationship dynamics Understanding the care recipient
Existential Concerns	Preparation for partners end of life Sense of purpose	Preparation for partners end of life Sense of purpose or meaning
Emotional States	Perceived stress Anger Well-being Emotional self-regulation Relaxation Energy	Stress management Perceived stress Anger Worry Psychological well-being Mental well-being

		Well-being Emotional self-regulation Mood Anger control Feeling relaxed Feeling energized Motivation Relaxation
General Skills	Creative thinking Organization Disclosing information Parenting Consciousness Problem solving Time management Coping Understanding Gratitude skills Preparedness to address own needs Communication Acceptance Express feelings	Creative thinking Day-to-day management Disclosing information Parenting Consciousness Problem-solving strategies Organization Ability to set limits Problem solving Time management Coping skills Coping strategies Understanding Noticing positive events Gratitude skills Preparedness to address own needs Communication Acceptance Express feelings Identifying emotions Time away from caregiving
Self-Efficacy	Self-efficacy	Self-efficacy Regained sense of control over life Confidence
Knowledge	Knowledge of the disease	Knowledge of the disease

Understanding the disease

Facilitators from Qualitative Results

Themes	Sub-Themes	Perceived Facilitator
Characteristics of the Intervention	Flexibility in the intervention Professionally organized intervention Allowing caregivers to socialize	Sessions tailored to the specific needs of caregivers Intervention organized individuals based on language preference Flexibility in the intervention (e.g., delay or postpone sessions) Having professionals organize the intervention Provided socialization time for the caregivers
Characteristics of Intervention Delivery	Accessibility of intervention	Virtual format of intervention (i.e., telephone or online) Session location where care recipient receives care
Providing Resources	Providing intervention resources Providing intervention training	Providing equipment to caregivers to participate (e.g., computers) Information packets provided Providing resources on how to find affordable equipment if caregivers wished to continue with the program once the intervention ended Instructions and/or training provided for online interventions
Support Provided Outside of Intervention	Additional services provided	Individuals received support between sessions (e.g., support by telephone) Individuals received supplemental sessions if needed

Barriers from Qualitative Results

Themes	Sub-Themes	Perceived Barrier
Intervention Misaligned to Caregiver Needs	Dislike of intervention content or structure Lack of need or interest in attending services Dislike of intervention facilitation style	Dislike of, or uninterest in intervention (structure or content) Intervention perceived as being too tiresome/long/burdensome Lack of information prior to sessions Not needing additional support Lack of understanding from professional leading intervention
Ability to Make Time for Intervention	Decreased interest or ability to attend intervention	Limited time to attend intervention Scheduling difficulties (i.e. difficulty scheduling times to attend support service) Not wanting/not able to spend time away from care recipient (e.g., due to severity of illness)
Practical Barriers	Suboptimal intervention delivery Unsustainable intervention delivery	Inconvenient timing of intervention delivery (i.e. should have been provided earlier or later after diagnosis) Cost Lack of sustainability (intervention not available after study) Accessibility issues (e.g., too far away) Delivery difficulties (e.g., technical problems for online or phone interventions, noises for in person interventions)
Emotional Barriers	Emotional difficulties engaging fully in intervention	Difficulty feeling comfortable with other caregivers Talking about caregiving causes distress

Behavioural or Psychological Interventions (Table 1). Sixteen studies described behavioural or psychological interventions (e.g., problem solving therapy, emotional expression interventions) (Aoun et al., 2015; Bevans et al., 2010; Bevans et al., 2014; Cox et al., 2012; Dowling et al., 2014; Elliott et al., 2008; Fidika et al., 2015; Langer et al., 2012; Laudenslager et al., 2015; Lindell, 2008; Manne et al., 2016; Marconi et al., 2015; Mioshi et al., 2013; Moola et al., 2017; Rexilius et al., 2002; Van Groenestijn et al., 2015). Nine studies included a control group (RCTs = 7; quasi-experimental studies = 2) (Dowling et al., 2014; Elliott et al., 2008; Langer et al., 2012; Laudenslager et al., 2015; Lindell, 2008; Manne et al., 2016; Mioshi et al., 2013; Rexilius et al., 2002; Van Groenestijn et al., 2015), 5 studies were pre-post designs without a control group (Aoun et al., 2015; Bevans et al., 2010; Bevans et al., 2014; Cox et al., 2012; Fidika et al., 2015), and 2 studies sought qualitative information about perceived benefits of an intervention (Marconi et al., 2015; Moola et al., 2017).

Among RCTs, sample sizes ranged from 15 to 218 total participants (median = 57). Significant between-group reductions in perceived stress (n = 3/3 studies) (Dowling et al., 2014; Laudenslager et al., 2015; Lindell, 2008) were found favoring the intervention groups among all RCTs that reported this outcome. Interventions resulting in reductions in stress included aspects of psychoeducation and were delivered weekly either individually or in a group, and focused on (1) improving positive emotion through practicing gratitude, mindfulness, and altruistic behaviours, among other positive psychology techniques; (2) teaching coping skills, stress management, and relaxation techniques; and (3) improving lifestyle and disease management through educating caregivers and patient about the disease, end of life care, and cognitive behavioural techniques. In most cases, the 7 RCTs that tested group differences on health

outcomes or mental health symptoms did not generate statistically significant differences, including for general mental health (n = 4/5 studies non-significant) (Elliott et al., 2008; Laudenslager et al., 2015; Lindell, 2008; Manne et al., 2016; Van Groenestijn et al., 2015), symptoms of anxiety (n = 3/3 non-significant) (Lindell, 2008; Manne et al., 2016; Van Groenestijn et al., 2015), symptoms of depression (n = 5/6 studies non-significant) (Dowling et al., 2014; Elliott et al., 2008; Laudenslager et al., 2015; Lindell, 2008; Manne et al., 2016; Van Groenestijn et al., 2015), and general health (n = 3/3 studies non-significant) (Elliott et al., 2008; Laudenslager et al., 2015; Lindell, 2008).

Two quasi-experimental studies that did not assign participants to comparison groups randomly included sample sizes of 21 and 36 participants, respectively (Mioshi et al., 2013; Rexilius et al., 2002). The tested interventions included providing massage therapy or a healing touch intervention (Rexilius et al., 2002) and a cognitive appraisal intervention (Mioshi et al., 2013). The study that assessed massage therapy and healing touch reported between group differences, where reductions in negative mental health outcomes (i.e., symptoms of anxiety, depression and fatigue) were found favoring the massage therapy group. No significant differences were found for caregiver burden. In the study that delivered a cognitive appraisal intervention, within group differences were reported separately for the experimental and control groups. Only caregivers in the intervention group demonstrated statistically significant reductions in caregiver burden, improved reactions to care recipient behavior, and increased coping skills. Neither the intervention nor the control group improved significantly on measures of mental health (i.e., stress, depression, anxiety).

Five pre-post studies included sample sizes ranging from 8 to 72 total participants and reported within group differences (Aoun et al., 2015; Bevans et al., 2010; Bevans et al., 2014;

Cox et al., 2012; Fidika et al., 2015). Interventions included therapeutic writing (Fidika et al., 2015), problem-solving therapy (Bevans et al., 2010; Bevans et al., 2014), coping skills training (Cox et al., 2012), and general counselling (Aoun et al., 2015). Results for within group differences differed across studies, with no mental health outcomes (e.g., symptoms of depression, anxiety, self-efficacy, quality of life) demonstrating significant or non-significant results in more than 2 studies (see Tables 1 and 5). The two qualitative studies described perceptions of outcomes of weekly meditation (n = 18) (Marconi et al., 2015) and four sessions of counselling (n=8) (Moola et al., 2017), respectively. These studies described caregiver reported improvements in mental health outcomes such as emotional self-regulation, ability to relax, acceptance, and increased support, among other outcomes. They also reported that participants indicated that they obtained practical skills (i.e., coping skills and time management skills).

Support Focused Interventions (Table 2). Three studies delivered support focused interventions (i.e., in-person or telephone support groups). One study included a control group (RCTs = 1) (Sheija & Manigandan, 2005), and 2 studies sought qualitative information about perceived intervention benefits (Cipolletta et al., 2018; Stewart et al., 2001). In the RCT (N = 36), statistical tests were conducted to compare outcomes between those assigned to an in-person support group and to no support group (Sheija & Manigandan, 2005). Seven support groups sessions were delivered by professionals over a two-week period and included role-play and relaxation techniques. Statistically significant effects were found for all outcomes measured, including general health, symptoms of depression and anxiety, and quality of life, all favouring the intervention group. The two qualitative studies included a support group delivered in person (10 sessions total, frequency of sessions not reported) (Cipolletta et al., 2018) and one delivered

by telephone (weekly for 12 weeks) (Stewart et al., 2001). Caregivers that received both interventions reported being satisfied with the support groups and noted that they found the groups helpful to decrease feelings of loneliness and isolation. Caregivers receiving the in-person support group also indicated that they benefitted from sharing fears and concerns and listening to other caregivers' experiences. Caregivers that received the telephone support group reported improved communication, relationships, and confidence. They also noted that the intervention provided affirmational, informational, and emotional support (see Tables 2 and 5).

Educational Interventions (Table 3). Two studies delivered educational interventions which incorporated didactic learning (Alankaya & Karadakovan, 2015; Gormley et al., 2014). One was a pre-post study without a control group and 1 study sought qualitative information about the benefits of an intervention. The pre-post study included 30 caregivers while the qualitative study included 50 caregivers. The pre-post study provided individualized education in the form of power-point sessions and a booklet based on caregiver identified needs (Alankaya & Karadakovan, 2015). A significant improvement was found between pre and post scores for caregiver burden but not caregiver reported general health. The qualitative study delivered an educational event at an annual meeting, which included presentations, small group discussions, and question and answer sessions with clinicians (Gormley et al., 2014). All caregivers in the study reported being satisfied or very satisfied with the topics covered, the presentation, and the location of the event. More than 80% of caregivers also reported being satisfied or very satisfied with the duration and day of the event. Caregivers described feeling less isolated due to interacting with other caregivers (see Tables 3 and 5).

Multi-Component Interventions (Table 4). Thirteen studies delivered multi-component interventions (e.g., interventions that included both psychoeducation and behavioural

components) (A'Campo et al., 2012; Bozkurt et al., 2014; Dunlop et al., 2016; Elliott & Berry, 2009; Leenaars et al., 2012; Li et al., 2017; Mazanec et al., 2017; Raj et al., 2015; Rodgers et al., 2007; Rotondi et al., 2005; Schulz et al., 2009; Videaud et al., 2012; Wade et al., 2010). Six studies, all RCTs (Elliott & Berry, 2009; Li et al., 2017; Mazanec et al., 2017; Raj et al., 2015; Schulz et al., 2009; Wade et al., 2010), included a control group, and the remaining 7 studies were pre-post designs without a control group (A'Campo et al., 2012; Bozkurt et al., 2014; Dunlop et al., 2016; Leenaars et al., 2012; Rodgers et al., 2007; Rotondi et al., 2005; Videaud et al., 2012). Among RCTs, sample sizes ranged from 12 to 475 total participants (median = 51). Few statistically significant between-group improvements were found for any outcomes measured in the RCTs. Symptoms of depression were measured most frequently, and the majority of studies found no significant difference between groups (n=5/6 studies non-significant) (Elliott & Berry, 2009; Li et al., 2017; Mazanec et al., 2017; Raj et al., 2015; Schulz et al., 2009; Wade et al., 2010), when comparing intervention groups that incorporated problem solving, interactive groups, psychoeducation and exercise, stress management, and/or cognitive behavioral therapy components to control groups. Other commonly reported measures for comparing the intervention and control groups found few statistically significant differences for general health (n= 4/5 studies non-significant) (Elliott & Berry, 2009; Mazanec et al., 2017; Raj et al., 2015; Schulz et al., 2009; Wade et al., 2010), perceived burden (n=2/2 non-significant) (Li et al., 2017; Schulz et al., 2009), problem solving (n=1/2 studies non-significant) (Elliott & Berry, 2009; Wade et al., 2010), and symptoms of anxiety (n=1/1 non-significant) (Mazanec et al., 2017). The pre-post studies had sample sizes ranging from 4 to 186 caregivers. Results for pre-post analyses demonstrated a significant improvement in caregiver burden (n = 3/4 studies statistically significant) (A'Campo et al., 2012; Bozkurt et al., 2014; Dunlop et al., 2016; Rodgers

et al., 2007) after receiving interventions that involved group psychoeducation and emotional or social support. No other patterns were found among pre-post within group findings, as the remaining outcomes collected (e.g., caregiver strain, quality of life, perceived stress) were measured in just one study. Narrative benefits were also described in 9 of the 13 studies testing a multicomponent intervention and included feeling less isolated, improved mood, and enhanced caregiver knowledge, (see Tables 4 and 5).

Facilitators and barriers of establishing and maintaining rare disease support services for caregivers

Fourteen publications (41%) described facilitators of establishing and maintaining the interventions (Cipolletta et al., 2018; Cox et al., 2012; Dowling et al., 2014; Laudenslager et al., 2015; Mazanec et al., 2017; Raj et al., 2015; Rexilius et al., 2002; Rodgers et al., 2007; Rotondi et al., 2005; Schulz et al., 2009; Sheija & Manigandan, 2005; Stewart et al., 2001; Van Groenestijn et al., 2015; Wade et al., 2010), and 19 (56%) described barriers (A'Campo et al., 2012; Aoun et al., 2015; Bevans et al., 2010; Cipolletta et al., 2018; Devine et al., 2016; Fidika et al., 2015; Gormley et al., 2014; Langer et al., 2012; Laudenslager et al., 2015; Leenaars et al., 2012; Lindell, 2008; Manne et al., 2016; Marconi et al., 2015; Mazanec et al., 2017; Mioshi et al., 2013; Moola et al., 2017; Rexilius et al., 2002; Stewart et al., 2001; Wade et al., 2010) (see Tables 1-6). No studies conducted statistical tests on facilitators or barriers. Four themes of facilitators were identified through qualitative results of studies, including, (1) characteristics of the intervention, (2) characteristics of intervention delivery, (3) providing resources, (4) support provided outside of intervention. Examples of facilitators within each theme included, (1) tailoring sessions to caregiver needs (characteristics of the intervention), (2) telephone or online delivery of intervention (characteristics of intervention delivery), (3) providing equipment (e.g.,

computers) to caregiver (providing resources), (4) receiving support between sessions (support provided outside of intervention). Facilitators were reported within behavioural or psychological, support-focused, and multi-component interventions and were found to be similar between the types of interventions (Table 2). Studies testing educational interventions did not report any facilitators to establishing and maintaining support services.

Four themes of barriers were identified through analysis of qualitative results, including, (1) interventions being misaligned to caregiver needs, (2) the inability to make time for participation in an intervention, (3) practical barriers (e.g., cost, continuity, accessibility, technical problems with intervention), (4) emotional barriers (see Tables 1-6). Examples of barriers within each theme included, (1) the intervention being perceived as being too tiresome/long/burdensome (intervention misaligned to caregiver needs), (2) limited time to attend an intervention (inability to make time for intervention), (3) accessibility issues, such as intervention being too far away (practical barriers), (4) talking about caregiving causes distress (emotional barriers). Barriers were reported within all intervention types, including, behavioural or psychological, support-focused, educational, and multi-component interventions and were similar between the types of interventions (Table 2).

DISCUSSION

A total of 34 publications, including 17 studies (50%) with a control group, examined behavioural or psychological, support-focused, educational, or multi-component interventions for caregivers of people with a rare disease. The majority of interventions were behavioural or psychological and were delivered in person. All interventions were delivered by a professional. Commonly tested benefits that resulted in statistically significant improvements were reductions in self-reported stress and caregiver burden. These improvements were found after receiving

behavioural or psychological interventions (e.g., teaching stress management and coping skills, cognitive behavioural therapy) and support-focused interventions (i.e., support groups). Health and mental health symptom outcomes were rarely found to significantly improve among caregivers after receiving any of the interventions reviewed. Qualitative studies reported many perceived benefits including decreased feelings of isolation, benefits from listening to other caregivers' experiences, and an increase in coping skills.

Facilitators and barriers to establishing and maintaining these interventions were reported in close to half of all included studies (41% and 56%, respectively). Commonly reported facilitators included characteristics of the intervention (i.e., sessions tailored to the needs of caregivers) and its delivery (e.g., delivering the intervention online or by telephone). Commonly reported barriers included interventions being misaligned to caregiver needs (e.g., lack of understanding from professional leading the intervention) and practical barriers of the intervention (e.g., accessibility issues such as the intervention being too far away).

Many studies have reported on benefits from interventions among caregivers to individuals with common diseases. For example, a recent Cochrane review and meta-analysis assessed the effectiveness of psychosocial interventions delivered by health professionals to caregivers of individuals living with cancer (Treanor et al., 2019). Similar interventions (e.g., psychoeducational) were tested in 19 studies included in the review and in line with our findings; little to no improvements were found for outcomes of depression, anxiety, and caregiver health. Minimal improvements of quality of life were found post-intervention (standardized mean difference = 0.29), however, all included trials were rated as having a high risk of bias, and the authors concluded that there is an immediate need for rigorous trials in order to draw firm conclusions on the effectiveness of psychosocial interventions delivered by health professionals

for caregivers of an individual living with cancer. The Cochrane review did not find any RCTs that measured caregiver satisfaction with the intervention, and the review did not assess outcomes such as stress or caregiver burden, which were the outcomes most often found to significantly improve in studies included in our review.

Reduced isolation was also a common theme among studies in our review. This finding aligns with regional and disease-specific recommendations for rare diseases (Martin et al., 2019; Critical Care Services Ontario, 2017). For example, psychosocial recommendations were recently developed for individuals caring for a loved one with a rare disorder, epidermolysis bullosa, where the complexity of the disease was highlighted, as was the need to stimulate social participation to prevent patients and caregivers from feeling isolated (Martin et al., 2019). These guidelines recommend that patients and their families have access to a supportive network (e.g., a support group) to optimize social wellbeing and provide a sense of feeling understood. Consistent with these goals, three studies which delivered support-focused interventions in our scoping review found benefits including statistically significant improvements in health, symptoms of depression and anxiety, and quality of life as compared to a control group, while perceived benefits included decreased feelings of loneliness and isolation and the receipt of affirmational, informational, and emotional support. Receiving social support may be a key construct needed for caregivers to those with a rare disorder to provide a sense of being understood, alongside educational resources.

The findings of this scoping review suggest that psychosocial interventions may be an important resource for caregivers to a loved one with a rare disorder, but establishing and sustaining these interventions may be challenging over the long-term, given the rarity of the disorders and the unique needs described by caregivers to those with a rare disease (Adams,

Miller, & Grady, 2016). Rare disease organizations may be able to increase the feasibility and accessibility of interventions by considering peer-led interventions, including those available online. There is currently no evidence from trials on the effectiveness of such interventions for caregivers to individuals with a rare disease, but, the facilitators of establishing and maintaining psychosocial interventions highlighted in our current review may allow for the informed development and testing of such novel interventions in close collaboration with caregivers with lived experience and rare disease organizations.

Limitations

There are several limitations that should be considered when interpreting the results of this study. First, our search was restricted to publications in PubMed and CINAHL; therefore, relevant information from grey literature, such as rare disease websites, could have been missed. Second, most of the publications reviewed included small sample sizes, which can result in high levels of imprecision and overestimate possible intervention effectiveness. Third, although rare diseases share many commonalities, there are important differences that must be considered, such as prevalence, age of onset, and disease severity (European Organisation for Rare Diseases, 2005). Given the wide range of rare diseases present in this review, specific rare disease characteristics must be considered when interpreting the results of this study. Fourth, as this is a scoping review, we did not assess the risk of bias for individual studies, potential risk of publication bias among included studies, or attempt to synthesize effect estimates.

Conclusions

Psychosocial interventions may be an important resource for individual's caregiving for a loved one with a rare condition. These interventions may help to reduce stress and caregiver burden while decreasing feelings of isolation. There is a limited understanding of the facilitators

and barriers that can help to establish and maintain these interventions; however, providing interventions that addresses caregivers' unique needs through accessible platforms (e.g., online) may decrease the barriers that exist for establishing and maintaining these interventions. The findings of this scoping review present an overview of the various interventions that have been tested among caregivers to individuals with a rare condition and provide a preliminary understanding of interventions that may help support caregivers, especially within the many rare diseases where no research has been conducted.

DECLARATIONS

Funding: No specific funding was received for this review. DBR is supported by a Vanier Graduate Scholarship through Canadian Institutes of Health Research. ACJ is supported by a Canadian Graduate Scholarship (CGS – M) through Canadian Institutes of Health Research unrelated to the present study. KAT was supported by a Fonds de Recherche Québec - Santé (FRQ-S) masters training award unrelated to the present study. No funding body had any input into the any aspect of this review.

Conflicts of Interest: The authors do not declare any conflicts of interest.

Author Contributions: DBR made substantial contributions to the conception and design, acquisition, analysis, and interpretation of data, and was involved in drafting the manuscript. ACJ, MCA, KAT, made substantial contributions to the acquisition, analysis, and interpretation of data, and were involved in drafting the final manuscript. AL made substantial contributions to the acquisition of data, and was involved in drafting the final manuscript. MC made substantial contributions to the acquisition of data, and was involved in drafting the final manuscript. Dr. Thombs made substantial contributions to the conception and design, acquisition, analysis, and interpretation of data, and was involved in drafting the final manuscript. All authors read and approved the final manuscript. Dr. Thombs is the overall guarantor.

Ethics Approval: Ethics approval was not necessary for this study as it did not contain participants.

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Linking Manuscripts One and Two

Prior to conducting research with caregivers to persons with SSc, an understanding of interventions that had been applied and tested among caregivers to persons with a rare disease more broadly, was necessary. Mapping the available literature as a foundation for understanding the perceived and tested benefits and sustainability of interventions among informal caregivers of people with a rare disease was also sought to ensure that the current thesis built upon the understanding of caregiving in the context of rare disorders. As such, the first manuscript was a scoping review examining the benefits, barriers, and facilitators for caregivers to people with a rare disease participating in psychosocial interventions.

This scoping review identified 34 studies, including 17 trials with a control group, that tested psychosocial interventions among caregivers to persons with a rare disorder. Interventions were perceived as being helpful to caregivers and often resulted in statistically significant reductions in stress, burden, and feelings of isolation; however, few differences in mental health symptoms (e.g., depression) or health outcomes (e.g., physical health) were found. A common component of interventions that resulted in caregiver benefits were sessions being tailored to the specific needs of caregivers. Importantly, in line with the goals of this thesis, none of the included studies assessed caregivers to people with SSc. Further, 97% of all included studies were comprised of more female caregivers than male caregivers. Caregiving in the context of SSc, however, may differ, as SSc disproportionately affects women, whereby the caregiving role is more likely to be undertaken by males.

Following this review, the importance of identifying the specific needs and preferences among caregivers to individuals with SSc was evident to allow for interventions to be adapted to this group of caregivers. Moreover, the ability to infer potential effectiveness of interventions

was limited due to important differences between the rare conditions included in the review and SSc. Altogether, this scoping review highlighted potential benefits from implementing psychosocial interventions among caregivers, while further emphasizing the need to understand the specific priorities from the perspectives of informal caregivers of persons living with SSc, in order to aid in the development of a research program to enhance the experience of these caregivers. As such, we conducted a study with caregivers of persons with SSc to gain a preliminary understanding of the challenges faced and the intervention preferences of informal caregivers of people with SSc.

Chapter 3

Manuscript 2

(Published in BMJ Open: Rice, D. B., Cañedo-Ayala, M., Turner, K. A., Gumuchian, S. T., Malcarne, V. L., Hagedoorn, M., & Thombs, B. D. (2018). Use of the nominal group technique to identify stakeholder priorities and inform survey development: an example with informal caregivers of people with scleroderma. *BMJ open*, *8*(3), e019726)

Use of the Nominal Group Technique to Identify Stakeholder Priorities and Inform Survey Development: An Example with Informal Caregivers of People with Scleroderma

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Word Count: 3994

ABSTRACT

Objectives: The Nominal Group Technique allows stakeholders to directly generate items for needs assessment surveys. The objective was to demonstrate the use of Nominal Group Technique (NGT) discussions to develop survey items on: (1) challenges experienced by informal caregivers of people living with systemic sclerosis (SSc) and (2) preferences for support services.

Design: Three NGT groups were conducted. In each group, participants generated lists of challenges and preferred formats for support services. Participants shared items, and a master list was compiled, then reviewed by participants to remove or merge overlapping items. Once a final list of items was generated, participants independently rated challenges on a scale from 1 (not at all important) to 10 (extremely important) and support services on a scale from 1 (not at all likely to use) to 10 (very likely to use). Lists generated in the NGT discussions were subsequently reviewed and integrated into a single list by research team members.

Setting: SSc patient conferences held in the United States and Canada.

Participants: Informal caregivers who previously or currently were providing care for a family member or friend with SSc.

Results: A total of 6 men and 7 women participated in the NGT discussions. Mean age was 59.8 years (standard deviation = 12.6). Participants provided care for a partner (n = 8), parent (n = 1), child (n = 2), or friend (n = 2). A list of 61 unique challenges was generated with challenges related to gaps in information, resources, and support needs identified most frequently. A list of 18 unique support services was generated; most involved online or in-person delivery of emotional support and educational material about SSc.

Conclusions: The NGT was an efficient method for obtaining survey items directly from SSc caregivers on important challenges and preferences for support services.

Keywords: Systemic sclerosis; scleroderma; nominal group technique; caregiver

INTRODUCTION

Surveys provide a feasible method for gathering and prioritizing input from large numbers of stakeholders in order to inform program development (O'Haire et al., 2011). A number of approaches can be used to develop survey items for the purpose of needs prioritization. Examples include adapting a pre-existing questionnaire originally designed for use with other groups, gathering expert opinions, and using theoretical definitions of a construct to guide item generation (Delisle et al., 2016; Francis et al., 2004; Romine, Sadler, Presley, & Klosterman, 2014; Thomson et al., 2009). These approaches do not explicitly integrate perspectives of stakeholders, however, and risk identification of program goals that may not be ideally aligned with the needs of the target group (Asadi-Lari, Tamburini, & Gray, 2004). Qualitative focus groups and individual patient interviews may also be used to generate items. These methods, however, can be time- and resource-intensive and may overly emphasize input from some participants and not others (Smithson, 2000). Furthermore, they require researchers to translate identified themes into survey items, but do not directly generate survey items.

The Nominal Group Technique (NGT) is an approach that can be used to structure group discussions in a way that allows stakeholders to directly generate items for a needs assessment survey. The NGT method facilitates generation of survey items in a straight-forward, cost-efficient manner by directly soliciting stakeholder input on survey items to address specific research questions (Delbecq, van de Ven, & Gustafson, 1975; Harvey & Holmes, 2012). In the context of needs assessment, it can be used to create a priority list of challenges that need to be addressed, along with potential solutions. When the NGT is used, a specific question is presented to the group of participants, then participants individually and silently generate lists of examples in response to the question. Following this, each participant shares each of her or his items one at

a time in a round-robin format until all items are shared, which results in a compiled list of items from the entire group. After the comprehensive list is compiled, group discussion occurs among participants and items are removed, reworded, or added to the list. Finally, participants vote or rank the items generated in terms of importance or relevance (Harvey & Holmes, 2012). The NGT has been used previously for preliminary item generation and to provide direction for survey development with stakeholder groups that include patients and family members of persons impacted by health conditions (Chasens & Olshansky, 2008), including people with type 2 diabetes and multiple sclerosis, and caregivers of people with Parkinson's disease (Chasens & Olshansky, 2008; Harvey & Holmes, 2012; Kleiner-Fisman, Martine, Lang, & Stern, 2011; Kremer et al., 2016). In addition to its efficiency and ability to directly incorporate stakeholder input into surveys, the collaborative nature of the NGT may increase stakeholder ownership of research and increase the likelihood that programs that are developed effectively address stakeholders' most important needs (Vella, Goldfrad, Rowan, Bion, & Black, 2000).

Informal caregivers are people who provide support for a family member or friend in need of care due to a health condition (Hughes, 2008). There is only limited research on informal caregivers in rare diseases, despite the important role they fill and the emotional and practical challenges they face (Adelman, Tmanova, Delgado, Dion, & Lachs, 2014). Many patients with rare diseases have substantial care needs, but there are often few specialized resources available to them through the healthcare system. As such, informal caregivers for patients with rare diseases, may undertake a substantial role in caregiving (Kole & Faurisson, 2009; Rode, 2005; World Health Organization, 2005).

Systemic sclerosis (SSc, scleroderma) is a rare chronic autoimmune disorder, characterized by vascular damage, inflammatory system activation, and excessive production of collagen

(Bernatsky et al., 2009). Only one doctoral thesis has considered the experiences of informal caregivers of people with SSc. This unpublished thesis was a qualitative study that included 13 caregivers (Maril, 2012). Consistent with caregiving in other diseases, SSc caregivers who participated in the study reported that their experience as a caregiver involved having to manage additional tasks (e.g., household chores), increased negative feelings (e.g., guilt) and personal stress, and relationship changes (e.g., relational strain with the person with SSc) (Maril, 2012). The specific challenges faced by caregivers of persons with SSc, however, have not been studied systematically.

Developing resources for informal caregivers for persons with SSc may help them manage their role and reduce burden, but information is required on the challenges they face and their preferred support resources. Gathering information from a large number of caregivers is best accomplished via a survey. The objective of this study was to use the NGT in a series of discussions to develop survey items to assess: (1) challenges experienced by informal caregivers of people living with SSc, and (2) their preferences for types of support services that could potentially be developed.

METHODS

Participants and Procedures

We conducted three NGT discussions at the national patient conferences of the Scleroderma Foundation of the United States (two groups) and Scleroderma Canada (one group). Eligible participants were people who had provided unpaid care in the past 12 months to a friend, family member, or partner with SSc. It was not required that the caregiver live with the person diagnosed with SSc. Potential participants were emailed study announcements by the Scleroderma Foundation and Scleroderma Canada prior to the conferences. Participants who

expressed interest in the study were then contacted by email by the study coordinator who provided them with details about the study, including information about the date and time of NGT discussions, researchers' credentials, study goals, research questions, and the end goal for the project. Prior to each group, paper copies of the consent form were provided to participants, and they were given the opportunity to ask questions about the study.

Prior to beginning the NGT discussions, participants completed a brief demographic questionnaire that was used to describe participant characteristics. The questionnaire included items about the participants' age, sex, race/ethnicity, employment status, and information about the person for whom they provided care, including SSc diagnosis subtype and years since diagnosis. They also provided caregiving information, including type and length of relationship to the person with SSc, tasks undertaken as part of caregiving, and time spent providing care.

The three NGT discussions ranged in length from 90 to 120 minutes and were conducted in July 2016 (Scleroderma Foundation) and September 2016 (Scleroderma Canada). The three groups were held in private hotel conference rooms and were moderated by two members of the research team. The first and second groups were moderated by the principal investigator, a male professor trained in clinical psychology and knowledgeable about SSc (BDT) and a female doctoral student in clinical psychology knowledgeable about SSc (DBR). The third group was moderated by DBR and a female doctoral student in counselling psychology with experience in SSc research (STG). A female research assistant was present as an observer in the third group. All moderators had previous experience with discussion-based research. In addition, moderators pilot tested the interview guide among members of a research group from Montreal, Quebec prior to conducting the caregiver NGT discussions. The pilot test did not result in changes to the protocol, but it resulted in increasing the time allotted for the planned NGT discussions.

Participants were informed that the objectives of the NGT discussions were to: (1) develop a list of challenges they faced as informal caregivers, and (2) develop a list of caregiver support resources that would be helpful to them, as well as the ideal format for delivering these services.

After the NGT procedures were explained, participants were presented with the first research question: “Think about the challenges you have faced since taking on a caregiving role to somebody close to you with SSc”. After being presented with this question, participants were asked to list on a piece of paper the challenges that they have experienced while helping to care for a family member, friend, or partner with SSc. Participants developed a list of challenges individually without consultation with other group members. Once completed, participants shared one item at a time from their list in a round-robin format. Each member shared an item in turn until all members had a chance to share an item, then the process began again until every item on each participant’s list had been shared. Participants were instructed not to repeat items that were verbatim to items provided by others but to share any items that seemed to differ, even if only minimally. Participants’ answers were typed on a computer and projected onto a screen as they were provided so that the list could be viewed by the moderators and participants as it developed. Once all items had been shared, group discussion was used to clarify the meaning and wording of items and to remove or merge overlapping items. A consensus process was used, which involved asking participants if they agreed with the wording of items and, for example, if participants thought an item would be better separated into two items. Items were edited based on group feedback until agreement was reached for all decisions.

In each group, once a list of unique items was agreed upon, one of the moderators printed the list of items. Participants then rated the importance of each challenge listed on a scale from 1 to 10, with 1 representing challenges that they did not perceive as personally important to them

in their role as an informal caregiver and 10 representing extremely important challenges. After each participant had rated the items, the moderator collected the ratings. Item ratings were collected to inform the removal of items that may have been suggested, but were not especially relevant to caregivers.

Next, participants were presented with the second research question: "Think about services that could be put in place to provide better support to SSc caregivers. What programs, services, or supports would be helpful in your role as a caregiver? How would these programs, services or other supports operate?" Participants were asked to write down on a piece of paper any support services that they thought would be helpful and the way in which these services could be delivered. The same process used for answering the first research question was then applied to this research question, and a final master list of support services was developed. As with the first research question, each idea for a support service that was generated was rated independently by each participant on a scale from 1 to 10, with 1 representing support services that they believed they would be unlikely to use and 10 representing services they believed they would very likely use.

For both research questions, as participants shared their items, if clarification was necessary, probes were used to gain a clearer understanding of the challenges and support services stated, (e.g., "Can you elaborate on that?" see Appendix D for interview guide).

Data Analysis

Descriptive analyses were conducted to present demographic data. Many of the challenges and support services items were generated in more than one group. Thus, the research team created a master list that combined all generated items, identified items that overlapped between

groups, and merged overlapping items. Mean scores were calculated for each unique item. All analyses were conducted with SPSS version 22.0 (Chicago, IL).

The master list of potential survey items that integrated responses from all three NGT discussions was distributed to members of the research team, including the Scleroderma Caregiver Advisory Team, which was comprised of 8 informal caregivers of people with SSc. All research team members reviewed the list of items and associated means. The team then made recommendations to remove items that were too vague or repetitive and had the opportunity to suggest new items. An iterative process was used to reword or remove any unclear items, and to incorporate any additional items deemed relevant and important, until consensus on a final list of items was reached.

To categorize challenges, qualitative content analysis was employed (Mayring, 2000). First, relevant literature was reviewed to consider pre-established categories for caregiver challenges. A previous systematic review of 192 articles categorized consequences of caring for people living with cancer into four categories (Stenberg, Ruland, & Miaskowski, 2010). The four categories were used as a starting point to generate categorizations for the challenge items in our study. Challenges were first categorized by two members of the research team who, in collaboration with the principal investigator, developed definitions and rules for each category and refined or added categories as necessary (see Appendix E). Another member of the research team, blind to the initial categorization process, then used the definitions and rules to independently categorize the challenges.

RESULTS

Participant and Caregiving Characteristics

A total of 13 informal caregivers of persons with SSc (7 female, 6 male) participated in one of the three NGT discussions (Scleroderma Foundation Group One = 2 female; 0 male; Scleroderma Foundation Group Two = 3 female; 2 male; Scleroderma Canada Group = 2 female, 4 male). None of the participants who came to the group sessions declined to participate or dropped out prior to completing the study.

Participant sociodemographic and caregiving situation characteristics are presented in Table 1. Caregivers ranged in age from 28 to 76 years (mean = 59.8 years, standard deviation [SD] = 12.6). Most caregivers were employed full-time (n = 5) or retired (n = 7); one caregiver was unemployed. All five caregivers who worked reported that providing care interfered with their job. Participants provided caregiving for a partner (n = 8), parent (n = 2), friend (n = 2), or child (n = 1). Caregivers had provided care for an average of 8.9 years (SD = 7.8; range = < 1 year to 25 years) with an average of 10.2 hours per week of care provided (SD = 9.9; range = 1 hour to 25 hours).

Table 1. **Sociodemographic Characteristics of 13 Nominal Group Technique Discussion Participants**

Variable	
Caregiver Characteristics	
Female, *n (%)*	7 (53.8)
Age in years, *mean (standard deviation)*	59.8 (12.6)
Relationship status, *n (%)*	
Never married	1 (7.7)
Married	8 (61.5)
Living with partner in committed relationship	1 (7.7)
Separated or divorced	0 (0.0)
Widowed	3 (23.1)
Highest level of education, *n (%)*	
Secondary or high school	2 (15.4)
Some College or university	3 (23.1)
University degree	6 (46.2)
Postgraduate degree	2 (15.4)
Current occupational status, *n (%)*	
Unemployed	1 (7.7)
Retired	7 (53.8)
Employed full-time	5 (38.5)
Providing care has interfered with my job (of the 5 employed), *n (%)*	5 (100.0)
Scleroderma subtype of patient, *n (%)*	

Limited or CREST SSc	5 (38.5)
Diffuse SSc	7 (53.8)
Unknown	1 (7.7)
Age of care recipient, *mean (standard deviation)*	54.8 (15.3)
Years since care recipient's diagnosis, *mean (standard deviation)*	12.1 (8.6)
Years of providing care for care recipient, *mean (standard deviation)*	8.8 (7.9)
Relation to care recipient, *n (%)*	
Parent	2 (15.4)
Child	1 (7.7)
Partner	8 (61.5)
Sibling	0 (0.0)
Friend	2 (15.4)
Length of relationship with care recipient, *mean (standard deviation)*	29.2 (16.8)
Hours spent caring per week, *mean (standard deviation)*	10.2 (9.9)
Caregiving Tasks	
Transportation, *n (%)*	8 (61.5)
Activities of daily living, *n (%)*	9 (69.2)
Housework, *n (%)*	8 (61.5)
Preparing meals, *n (%)*	5 (38.5)
Managing finances, *n (%)*	2 (15.4)
Attending appointments, *n (%)*	10 (76.9)
Shopping, *n (%)*	8 (61.5)
Medical tasks, *n (%)*	2 (15.4)

Arranging other services for care recipient, *n (%)*	2 (15.4)
Other, *n (%)*	2 (15.4)

Note: SSc = systemic sclerosis

NGT Discussions – Challenge Items

The three groups generated 24, 27, and 38 original caregiving challenge items for a total of 89 (Appendix F), although there were duplicate items across groups. Of the 89 original items, 16 items received a mean score for importance between 8.0 and 10.0; 27 items were between 6.0 and 7.9; 39 items were between 4.0 and 5.9; and 7 items were rated less than 4.0.

After completion of the groups, the 89-items were distributed to members of the research team and the SSc Caregiver Advisory Team, resulting in the rewording, removal or combining of items. There were 55 unique challenge items remaining after this editing process. Six items were added by the research team and the SSc Caregiver Advisory Committee, resulting in a total of 61 identified challenges (Table 2).

Table 2. Reduced and Categorized List of Caregiver Generated Challenges and Item Means

Item	Original Item Number Prior to Item Reduction[a]	Mean Rating of Challenge Importance (1-10)	Number of Participants Who Rated the Item
Physical health concerns			
Experiencing fatigue and physical exhaustion	3, 84	6.3	8
Having trouble sleeping	8	5.0	2
Taking care of my health		Not applicable[b]	
Financial problems and work or employment problems			
Balancing caregiving and demands associated with my job	2	6.0	2
Having to take days off from work due to caregiving responsibilities	1, 63	4.3	8
Managing the cost of drugs and medical care	30	8.5	2
Managing loss of income due to my care-recipient's inability to work	31, 72	6.4	8

Role strain			
Balancing caregiving and other family responsibilities	10	6.5	2
Managing last minute changes due to the unpredictability of the disease	64	3.8	6
Having to do all of the winter chores alone due to my care-recipient's sensitivity to cold temperatures	79, 80	5.0	6
Having to handle all of the household chores on my own	45	6.0	5
Being unable to help address my care-recipient's pain or discomfort	5	4.0	2
Finding time for myself	11, 17	5.5	2
Having to learn new skills and abilities because my care-recipient can no longer do certain tasks	76	4.3	6
Having to make difficult medical decisions	51	5.0	5
Information, resources, and support needs			
Not having information about how to be a good caregiver	37	9.0	2

Not being able to find any answers as to why my care-recipient got scleroderma	28	8.5	2
Not having access to a caregiver support group	36	10.0	2
Not knowing other people who understand what I'm going through	38	9.0	2
Navigating healthcare issues while travelling	75	7.0	6
Planning trips and excursions while managing limitations, such as needing wheelchair access or other considerations	73	6.0	6
Having difficulty finding reliable and accurate information about scleroderma	20, 87	6.4	8
Having difficulty understanding important information about scleroderma and its treatment	21, 54	5.1	7
Having difficulty helping my care-recipient gain access to knowledgeable health providers	22, 23	7.5	2
Navigating the medical system	24	8.0	2

Interacting with medical, insurance, and social service agencies to address the needs of my care-recipient	29, 42, 61, 62	5.5	7
Interacting with health professionals who are not knowledgeable about scleroderma	47, 55	5.4	5
Managing rushed, inconsiderate, or insensitive behavior from health professionals	48, 52	5.1	5
Trying to find useful devices to help my care-recipient with activities of daily living	82	4.0	6
Finding assistance for things that my care-recipient used to do	19	5.0	2
Fear, anxiety, and uncertainty			
Being fearful that I will be left alone	27	8.5	2
Constantly worrying about my care-recipient's limitations	43	5.8	5
Feeling uncertain about the progression of my care-recipient's scleroderma	16, 89	8.0	8
General emotional difficulties			
Feeling helpless	12	7.0	2

Feeling hopeless	13	3.0	2
Managing my negative emotions towards my care-recipient	7	4.0	2
Managing my stress and relaxing	9	7.5	2
Managing my negative emotions	86	8.2	6
Guilt about leaving my care-recipient alone	32	6.5	2
Feeling ashamed to think about my own well-being or needs		Not applicable[b]	
Emotional difficulties of the care recipient			
Understanding the emotional needs of my care-recipient	66, 67	8.0	6
Knowing what to do about my care-recipient's guilt	83	5.5	6
Providing emotional support to my care-recipient on challenging days	65	8.0	6
Managing resentment from my care-recipient towards me	6	4.0	2
Managing my care-recipient's anger about having scleroderma	50	4.2	5
Managing my care-recipient's feelings of depression	57	4.0	5
Managing my care-recipient's thoughts of ending her or his life	60	3.0	5

Managing the disappointment or frustration of my care-recipient when she or he cannot take part in activities	81	6.5	6
Changes in relationship dynamics with care recipient			
Understanding when my help isn't wanted or needed	39, 44	5.7	5
Helping my care-recipient set reasonable limits on activities that have become difficult due to scleroderma	40	6.4	5
Providing needed help when my care-recipient doesn't want it or resists it	58, 71	5.9	11
Being patient with the care-recipient		Not applicable[b]	
Finding the balance between interfering and providing care	68, 77	6.8	6
Helping my care-recipient feel useful despite her or his physical limitations	59	5.8	5
Feeling a sense of loss because of activities we can no longer do together	4, 15	5.8	2
Accommodating my care-recipient's diet restrictions when we eat out	85	4.2	6

Discussing emotions or worries concerning scleroderma with my care-recipient		Not applicable[b]	
Dealing with loss of physical intimacy with my care-recipient		Not applicable[b]	
Changes in social interactions			
Noticing others' lack of knowledge and awareness about scleroderma	14, 25, 26	9.2	2
Managing social limitations, such as missing events or having to leave events early	69	5.7	6
Enjoying myself when spending time with friends without my care-recipient		Not applicable[b]	

a = items from original lists available in Appendix F

b = item generated from Scleroderma Caregiver Advisory Team

Using a modified set of the categories used in a previous study (Stenberg et al., 2010), the 61 challenges were grouped into 9 categories (see Appendix E for category definitions). There was 88% agreement of item categorization between raters. The definitions and rules were used in order to reach a consensus for the placement of each item. As shown in Table 2, the 9 categories included physical health concerns (n = 3 items); financial problems and work or employment problems (n = 4 items); role strain (n = 8 items); information, resources, and support needs (n = 15 items); fear, anxiety, and uncertainty (n = 3 items); general emotional difficulties (n = 7 items), emotional difficulties of the care recipient (n = 8 items); changes in relationship dynamics with care recipient (n = 10 items); and changes in social interactions (n = 3 items).

NGT Discussions – Support Service Items

The three groups generated 13, 13, and 15 original items reflecting preferred support service options (see Appendix G). Among the 41 items, 12 received a score reflecting likelihood of using the service between 9.0 and 10.0; 6 between 8.0 and 8.9; 15 between 6.0 and 7.9; and 8 less than 6.0. There was a high degree of duplication of items across groups. Thus, the 41 total items were reduced by the research team to 18 unique items, and these items were reviewed and edited by the research team, as necessary (Table 3). These 18 items included support services delivered online (n = 7); by telephone (n = 2); hard-copy resources (n = 2); and in-person delivery (n = 7).

Table 3. Reduced List of Caregiver Generated Support Services and Item Means

Item	Original Item Number Prior to Item Reduction[a]	Mean Rating of Service Importance (1-10)	Total Number of Participants Who Rated the Item
1. Caregiver internet-based chat group, forum, or social network site without professional moderator	7, 20	9.0	6
2. Caregiver internet-based chat group, forum, or social network moderated by a knowledgeable healthcare provider	8, 37	7.0	8
3. Caregiver-led breakout groups at patient conferences	9	9.5	2
4. Professionally led breakout groups at patient conferences	10, 27	8.4	8
5. Internet-based psychological and emotional self-help tools	12	10.0	2
6. One-to-one peer support (e.g., the ability to call another caregiver on the phone)	13	9.5	2
7. Professionally led in-person caregiver support group	2, 14	7.8	6
8. Caregiver-led in-person caregiver support group	1, 15, 31	7.3	12

9. Professionally led telephone-based support groups for caregivers	4, 16	5.8	6
10. Caregiver-led telephone-based support groups for caregivers	3, 17	6.0	6
11. Professionally led internet-based, live interaction (teleconference, Skype) caregiver support groups	6,18	7.7	6
12. Caregiver-led internet-based, live interaction (teleconference, Skype) caregiver support group	5, 19, 35	7.5	12
13. Caregiver newsletter	21	9.5	4
14. Retreat for caregivers	22	7.0	4
15. Online educational sessions for caregivers to help understand scleroderma and its impact on families	11, 23	8.8	6
16. Information package/pamphlet about scleroderma for caregivers of newly diagnosed patients	29	7.3	6
17. Information about scleroderma on an online reputable website for caregivers of newly diagnosed patients	30, 34	8.1	6

18. Conference caregiver educational sessions and workshops provided by a knowledgeable healthcare provider	Not applicable[b]

a = items from original lists available in Appendix G

b = item generated from Scleroderma Caregiver Advisory Team

DISCUSSION

In the present study, we used a novel NGT method to identify challenges faced by informal caregivers of persons with SSc and potential support services to address these challenges. Based on the quantity and variety of items that caregivers generated, NGT discussions were effective for developing items for our planned needs assessment survey of SSc caregivers. Use of the NGT enabled caregivers to directly share their perspectives and provided a mechanism for direct caregiver input in the development of survey that will be distributed to a larger sample of caregivers to persons with SSc.

A final list of 61 survey items that reflect unique challenges related to caregiving in SSc was generated. These challenges included physical health concerns; financial problems and work or employment problems; role strain; information, resources, and support needs; fear, anxiety, and uncertainty; general emotional difficulties; emotional difficulties of the care recipient; changes in relationship dynamics with the care recipient; and changes in social interactions. The largest number of items reflected challenges related to unmet information, resource, and support needs. Caregivers also generated items that reflected 18 support services that could be delivered to help address difficult aspects of caregiving. Support services that were delivered online, by telephone, in-person, and through hard-copy resources were all identified by caregivers as being potentially helpful. Online or in-person delivery of support services that focused on providing education and emotional support were the most common suggestions.

To our knowledge, this is the first study to gather information on challenges from informal caregivers of people living with SSc using NGT discussions. Our findings, however, can be compared to a thesis that included individual interviews with 13 caregivers of persons with SSc (Maril, 2012). Several of the challenges generated during the NGT discussions that

related to general emotional difficulties are similar to themes identified in the thesis, including guilt, frustration, and stress. Further, financial strain and career adjustments, relational strain, and difficulty managing multiple roles were reported as a challenges of caregiving in our NGT discussions and in previously reported interviews (Maril, 2012).

Our results can also be compared to studies of caregivers to persons with more common diseases, including Huntington's disease, cancer, and Alzheimer's disease. Caregivers to persons with more common diseases have described several similar challenges, including understanding and managing the patient's medical needs, changes to the patient-caregiver relationship, managing disease related problems, and concerns about the care recipient not receiving adequate care (Coristine, Crooks, Grunfeld, Stonebridge, & Christie, 2003; Mahoney, 1998; Stenberg et al., 2010; Williams et al., 2010). Our findings should also be considered in the context of rare diseases. Previous literature has suggested that rare diseases pose additional challenges for both the caregiver and the person living with the disease (Doyle, 2015; Gowran et al., 2015; Kole & Faurisson, 2009). Our results are in line with this research as the most commonly reported challenges related to the rarity of the disease and difficulty obtaining necessary information, resources, and support.

There are currently no formal resources in place for SSc caregivers, and no previous research has administered support service interventions among caregivers to persons with SSc. There are, however, many support services that have been developed to lessen the burden associated with caregiving in common diseases (Boots, Vugt, Knippenberg, Kempen, & Verhey, 2014; Pinquart & Sörensen, 2006; Selwood, Johnston, Katona, Lyketsos, & Livingston, 2007; Thompson et al., 2007), and some of these may be able to be adapted for SSc caregivers. For example, caregiver support groups, teleconference-based interventions, and educational sessions

(Bevans et al., 2014; Bormann et al., 2009; Chien et al., 2011; Neuharth-Pritchett & Getch, 2016; Walsh & Schmidt, 2003) have been provided for caregivers of persons with dementia, asthma, allogeneic hematopoietic stem cell transplantation, and cancer and were identified in our study as support services that SSc caregivers would be likely to use.

Strengths and Limitations

The present study has important strengths. For example, no other studies have used NGT discussions as a way of eliciting information from caregivers of people with rare diseases. Applying this methodology allowed us to generate a robust list of potential survey items in the words of stakeholders themselves and provided initial quantitative ratings of the importance of challenges and likelihood of service use for the items generated. Further, our research team included our Scleroderma Caregiver Advisory Team, which was comprised of caregivers to persons with SSc. This allowed for the incorporation of stakeholder input at each stage of the research and will aid in the facilitation of follow-up survey work and the eventual implementation of the findings. An advantage of the NGT compared to standard focus groups is that focus groups use open-ended questions to solicit discussion, and researchers must then develop survey items from the themes that were discussed. The NGT allowed for direct caregiver input that did not require researchers to interpret and extrapolate from what participants shared in the discussion to generate survey items.

There are limitations to consider when interpreting the results of this study. First, the NGT discussions were held at national patient conferences. This may have influenced the characteristics of participants and the generalizability of results, as only caregivers attending the 2016 American or Canadian conferences were eligible for participation in the groups. Specifically, our sample may have over-included caregivers who are well connected to the SSc

community. Second, participants only rated items one time in the groups. NGT discussions sometimes ask participants to re-score items after discussing the results of the first round of scoring, similar to a Delphi Process (Hsu & Sandford, 2007). Participants were only asked to rate items once and ratings were completed independently rather than discussed with other participants. Previous studies, however, have found that when asked, participants state that item ratings would generally remain unchanged if asked to rate items a second time (Ng & Sargeant, 2012). Thirdly, participation in the NGT discussions was restricted to caregivers who were literate. Lastly, 4 challenges were removed from our list of unique items due to the vague content of these items. During the NGT discussions, gathering additional specifications about the challenges may have been helpful to ensure that the items elicited from participants were retained. Despite these limitations, the study provides important information that can be used to inform the development of a survey to determine caregivers' most important challenges, and the support services that caregivers would be most likely to use.

Conclusion

The NGT was an efficient approach for gathering caregiver input to aid in the development of survey items. We found that caregivers of persons with SSc face many challenges and have substantial unmet needs. Some of the most important challenges identified were related to information, resources, and support needs. A range of possible support services were identified, with caregivers reporting being most likely to use services delivered in-person or through online platforms for education and emotional support, including internet-based psychological and emotional self-help tools. The findings from the present study suggest that programs offered online may result in a greater likelihood of caregiver participation. The results of the present study will be used to construct a survey that will be disseminated online to a larger number of

SSc caregivers in order to better understand the relative importance of the challenges identified and the likelihood of use of possible support services. The results of the present study, combined with survey results, will be used to develop SSc caregiver support services.

Authors' Contribution

DBR and BDT were responsible for the study conception. DBR, STG, and BDT contributed to data collection. DBR, MCA, KAT, STG, VLM, MH, BDT, and the Scleroderma Caregiver Advisory Team contributed to data analysis and interpretation. DBR, MCA, and BDT contributed to drafting the manuscript. All authors provided a critical revision of the manuscript and approved the final version of the manuscript. BDT is the guarantor.

Funding Statement

This research was supported by a grant from the Rare Disease Foundation and the BC Children's Hospital Foundation (Funding Reference Number 173-16; PI Rice). Ms. Rice was supported by a Vanier Graduate Scholarship. Ms. Gumuchian was supported by a Fonds de Recherché Santé Québec Doctoral Award. Dr. Thombs was supported by a Fonds de recherche du Québec – Santé researcher salary award. Authors had full access to the data and take responsibility for the integrity of the data and the accuracy of the data analysis. No funding body had any role in the design, collection, analysis or interpretation of data; in the writing of the manuscript; or in the decision to submit the manuscript for publication.

Competing Interests Statement

The authors have read and understood the BMJ policy on declaration of interests and declare that they have no competing interests.

Data Sharing Statement

No additional data are available.

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Linking Manuscripts Two and Three

The second manuscript in this thesis aimed to engage caregivers in developing items that were personally relevant to them and could be included in a future survey. Using a nominal group technique with groups of caregivers for people with SSc, conducted in North America, 13 caregivers independently generated lists of (1) challenges experienced in their roles as informal caregivers of people living with SSc, and (2) their preferences for support services that could potentially be developed. After removing duplicates and revising the list with an SSc Caregiver Advisory team, a final list of challenges experienced by informal caregivers of people with SSc included 61 unique items. The final list of potential support services targeting informal caregivers of people with SSc included 18 unique items.

This study provided the foundation for developing a survey that could be disseminated internationally among caregivers to a loved one with SSc, in order to understand the needs and preferences of caregivers with a larger, more diverse sample of individuals. A survey was necessary to draw conclusions regarding the relative importance of challenges that were identified from focus groups. Further, to consider the likelihood that caregivers would use any support service options that were elicited through the focus groups, surveying a greater sample was necessary. An online questionnaire containing the finalized lists of challenges and services collected from the second study was administered to informal caregivers of people with SSc in three different continents to explore the importance of challenges and the preferences for support services among caregivers.

Chapter 4

Manuscript 3

(Published in Disability and Rehabilitation: Rice, D. B., Cañedo-Ayala, M., Carboni-Jiménez, A., Carrier, M. E., Cumin, J., Malcarne, V. L., ... & Scleroderma Caregiver Advisory Team. (2019). Challenges and support service preferences of informal caregivers of people with systemic sclerosis: a cross-sectional survey. *Disability and Rehabilitation*, 1-7.)

Challenges and Support Service Preferences of Informal Caregivers of People with Systemic Sclerosis: A Cross-sectional Survey

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This is an Accepted Manuscript of an article published by Taylor & Francis in *Disability and Rehabilitation* on January 29, 2019, available online: https://doi.org/10.1080/09638288.2018.1557268

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Funding Statement: This research was supported by a grant from the Rare Disease Foundation and the BC Children's Hospital Foundation (Funding Reference Number 173-16; PI Rice). Ms. Rice was supported by a Vanier CIHR Graduate Scholarship. Dr. Thombs was supported by a Fonds de recherche du Québec – Santé researcher salary award. No funding body had any role in the design, collection, analysis or interpretation of data; in the writing of the manuscript; or in the decision to submit the manuscript for publication.

Competing Interest Statement: All authors declare that they have no competing interests.

Word Count: 3,78

ABSTRACT

Purpose: To evaluate the importance of different challenges experienced by informal caregivers to persons with systemic sclerosis (SSc) and identify priorities for support services that could be developed.

Materials and Methods: Caregivers of people with SSc from three continents completed an online questionnaire to rate the importance of possible caregiver challenges and likelihood of using different forms of support services. Importance of challenges and likelihood of using support services were rated from 1 (not important; not likely to use) to 4 (very important; very likely to use).

Results: 202 informal caregivers completed the survey (79 women, 123 men). Mean age was 58 years (standard deviation = 13). The most important challenges were related to supporting the care recipient with emotional difficulties and physical discomfort. Caregivers indicated that they would be more likely to use support services that involved online or hard-copy information resources, including those provided soon after diagnosis, compared to support that involved interacting with others.

Conclusions: Supporting the care recipient in managing emotional difficulties and physical discomfort were important challenges among caregivers. Interventions delivered through hardcopy or online resources, including those delivered soon after the care recipient's diagnosis, were rated as being most likely to be used by caregivers.

Key Indexing Terms: Scleroderma, rare disease, caregivers, surveys, questionnaires

INTRODUCTION

Systemic sclerosis (SSc), also known as scleroderma, is a rare chronic autoimmune disease. SSc is characterized by abnormal fibrotic processes and excessive collagen production, which results in skin thickening, damage to internal organs including the lungs, kidneys, and gastrointestinal tract, as well as vascular implications (Abraham & Varga, 2005). Women comprise more than 80% of cases (Mayes et al., 2003).

Patients with SSc experience diverse challenges that affect their quality of life, including gastrointestinal symptoms, respiratory problems, fatigue, and changes in appearance (Shah & Wigley, 2013). In addition to disease manifestations, persons diagnosed with SSc face difficulties related to the rarity of the disease. As compared to more common diseases, challenges include an uncertain prognosis, limited treatment options, difficulty accessing specialists, geographic distance from treatment centres, and a lack of disease-specific support resources (Adams, Miller, & Grady, 2016; Shire Report, 2013).

Many persons with rare diseases, including SSc, rely on support from informal caregivers (Lopez-Bastida, Linertova, Oliva-Moreno, Posada-de-la-Paz, & Serrano-Aguilar, 2014; Maril, 2012). Informal caregivers are typically family members who do not receive training or payment for their role (National Alliance for Caregiving, 2015). Little is known about the experiences of informal caregivers of people with rare diseases, and we identified only one study of informal caregivers of people with SSc (Maril, 2012). In that study, a doctoral thesis, 13 informal caregivers were interviewed, and the emotional challenges of caregiving of a person with SSc were emphasized (Maril, 2012).

Developing resources that address challenges faced by informal caregivers for persons with SSc could help reduce the negative consequences associated with caregiving. To develop

relevant resources, an understanding of the types of challenges that are most important to caregivers of those with SSc and the support services they would be most likely to use is required. Thus, we conducted a series of nominal group technique (NGT) discussions with 13 SSc caregivers to generate survey items that reflected challenges faced by informal SSc caregivers and the caregivers' preferences for types of support services that they believed would be useful (Rice et al., 2018). We worked with the caregivers and developed a list of 61 unique challenges in the domains of physical health concerns; financial and work or employment problems; role strain; the need for information, resources, and support; fear, anxiety, and uncertainty; general emotional difficulties; emotional difficulties of the care recipient; changes in social interactions with others; and changes in relationship dynamics with the care recipient. A list of 18 potentially useful support services was also generated, including both online and in-person support methods (Rice et al., 2018). The objective of the present study was to assess the frequency and importance of the 61 challenges and of caregivers' preferences for the 18 types of support services identified via the NGT discussions. To do this, we disseminated a survey, developed based on our NGT discussions, to an international sample of caregivers of persons with SSc.

METHODS

Participant Sample and Procedure

Informal caregivers of a person diagnosed with SSc were recruited to anonymously complete an online questionnaire with the survey tool *Qualtrics*, between December 2016 and June 2017. To be eligible for the study, participants had to indicate that they currently or previously provided unpaid care for a friend or family member with SSc. Participants had to be 18 years or older and fluent in English or French. Participants were recruited through SSc patient

organizations, including Scleroderma Canada, the Scleroderma Foundation of the United States, Scleroderma and Raynaud's UK, the Association des Sclérodermiques de France, and the Scleroderma Association of New South Wales, Australia. Recruitment also occurred through emails and posts on SSc-related websites and other social media venues. Advertisements were also emailed to people with SSc participating in an ongoing internet-based cohort (Kwakkenbos et al., 2013).

Respondents who accessed the survey website could complete the survey online in English or French. After clicking on the survey link, respondents were shown a consent form that described study objectives and survey instructions. Respondents were given the option to consent by clicking an arrow to continue the survey or to close their browser and not participate. This study was approved by the Research Ethics Committee of the Jewish General Hospital, Quebec, Canada.

Measures

Caregiver, Care Recipient, and Caregiving Characteristics.

Caregivers provided their age, gender, country of origin, race/ethnicity, relationship status, highest level of education achieved, current and past occupational status, and household income. They also provided care recipient information, including age, gender, SSc subtype (diffuse or limited), and years since diagnosis, and caregiving characteristics, including years of caregiving, relationship with the care recipient, length of relationship with the care recipient, hours a week providing care, and types of activities with which they assist or assisted the care recipient.

Challenges Associated with Caregiving

A 61-item questionnaire to measure challenges associated with caregiving was developed from three NGT discussions that involved caregivers to persons with SSc (Rice et al., 2018). The

use of the NGT allowed SSc caregivers to directly share challenges that they experience. The list of challenges was reviewed and revised by a SSc Caregiver Advisory Team prior to finalizing the questionnaire for the present study (Rice et al., 2018). The challenges questionnaire developed from the discussion items includes challenges that were previously (Rice et al., 2018) grouped thematically into 9 categories to facilitate ease of reviewing similarly themed items: (1) physical health concerns (3 items); (2) financial problems and work or employment problems (4 items); (3) role strain (8 items); (4) information, resources, and support needs (15 items); (5) fear, anxiety, and uncertainty (3 items); (6) general emotional difficulties (7 items); (7) emotional difficulties of the care recipient (8 items); (8) changes in social interactions with others (3 items); and (9) changes in relationship dynamics with the care recipient (10 items). Caregivers rate each item from 1 (“not important”) to 4 (“very important”) based on the perceived importance of the challenge to them.

Support Service Preferences for Caregivers

An 18-item support services questionnaire was previously developed through NGT discussions at the same time the challenges questionnaire was developed and using the same method. This questionnaire assesses the support services that caregivers to persons with SSc believe they would be most likely to use. Example items include “caregiver-led breakout groups at patient conferences” and “caregiver newsletter”. Caregivers rate items from 1 (“not likely”) to 4 (“very likely”) based on the caregiver’s perceived likeliness of using the support service. Likelihood of using each of the 18 support services was evaluated separately.

Statistical Analyses

Descriptive analyses were performed for demographic variables. Continuous variables were presented as means and standard deviations (SDs), and categorical variables were presented as

percentages and counts. Frequencies were presented for all items included in the challenges and support service questionnaires. Since the purpose of the questionnaire was not to develop a measure with scoring properties, but rather to identify important challenges for caregivers, we did not calculate Cronbach's alpha or conduct analyses of measurement properties. Given previously identified gender differences for caregiving (Adelman, Tmanova, Delgado, Dion, & Lachs, 2014), after having identified study objectives we decided to consider potential gender differences in demographic factors and survey responses using chi-square tests. We compared women and men on challenges (important or very important versus not important or somewhat important) and support service preferences (likely to use or very likely to use versus not likely to use or somewhat likely to use). The Hochberg Sequential Method was used to adjust for multiple comparisons (Hochberg, 1988) and results are presented with confidence intervals. All statistical analyses were performed using SPSS Statistics, version 22.0 (Chicago, IL).

RESULTS

Sample Characteristics

A total of 262 people who indicated that they were past or current informal caregivers of a person with SSc accessed the survey. Of these, 202 (77%) completed the entire survey and were included in this study (see table 1). The majority of caregivers were from North America (74%) or Europe (23%). The mean age was 57 years (SD = 14 years), and 123 were male (61%). Caregivers were providing care for a partner (72%), parent (12%), child (7%), sibling (4%) or friend (5%). They were employed (49%), retired (40%), students (2%), or homemakers, unemployed, on disability, or on a leave of absence (11%). Twenty-eight percent of caregivers had pursued postgraduate degree, 27% completed a university degree, 24% completed some university, and 21% completed primary to high school education. Caregivers provided a mean of

14 (SD = 13) hours of care per week. The mean age of care recipients was 58 years (SD = 13) and approximately half were diagnosed with diffuse SSc (50%).

Table 1. Sociodemographic information among 202 informal caregivers.

Variable	Men	Women
Number, *n (%)*	123 (60.9)	79 (39.1)
Age, *mean (standard deviation)*	60.6 (12.5)	51.8 (15.4)
Relationship status, *n (%)*		
Never Married	3 (2.4)	15 (19.0)
Married	107 (87.0)	47 (59.5)
Living with partner in committed relationship	9 (7.3)	5 (6.3)
Separated or divorced	3 (2.4)	7 (8.9)
Widowed	1 (0.8)	5 (6.3)
Occupational status before caregiving, *n (%)*		
Employed	90 (73.2)	56 (70.9)
Retired	26 (21.1)	10 (12.7)
Students	1 (0.8)	5 (6.3)
Other	6 (4.9)	8 (10.1)
Current occupational status, *n (%)*		
Employed	55 (44.7)	40 (50.6)
Retired	59 (48.0)	21 (26.6)
Students	0 (0.0)	4 (5.1)
Other	9 (7.3)	14 (17.7)
Highest level of education obtained, *n (%)*		
Primary to high school	32 (26.0)	11 (13.9)

Some college or university	29 (23.6)	19 (24.1)
University degree	26 (21.1)	28 (35.4)
Postgraduate degree	36 (29.3)	21 (26.6)
SSc subtype, *n (%)*		
Limited scleroderma or CREST	47 (38.2)	21 (26.6)
Diffuse scleroderma	57 (46.3)	43 (54.4)
Unknown or not specified	19 (15.4)	15 (19.0)
Age of person with SSc, *mean (standard deviation)*	58.8 (12.5)	56.7 (14.6)
Years since care recipient's diagnosis, *mean (standard deviation)*	13.2 (7.5)	11.0 (6.1)
Years of providing care for care recipient, *mean (standard deviation)*	31.9 (12.5)	28.0 (13.2)
Relation to person with SSc, *n (%)*		
Parent	3 (2.4)	12 (15.2)
Child	3 (2.4)	21 (26.6)
Partner	115 (93.5)	31 (39.2)
Sibling	1 (0.8)	6 (7.6)
Friend	1 (0.8)	8 (10.1)
Other	0 (0.0)	1 (1.3)*
Length of relationship with care recipient, *mean (standard deviation)*	22.8 (6.3)	23.4 (5.7)

Hours of care per week, *mean (standard deviation)*	13.9 (12.9)	14.9 (14.2)
Caregiving Tasks, *n (%)*		
Transportation	83 (67.5)	38 (48.1)
Housework	99 (80.5)	52 (65.8)
Preparing meals	72 (58.5)	47 (59.5)
Managing finances	45 (36.6)	21 (26.6)
Shopping	87 (70.7)	50 (63.3)
Medical tasks	39 (31.7)	30 (38.0)
Arranging other services for care recipient	8 (6.5)	15 (19.0)
Other	31 (25.2)	35 (44.3)

SSc = systemic sclerosis
* Cousin

Responses to Challenges and Support Services Questionnaires

Challenges Questionnaire

Item Responses. Table 2 shows the percentage of caregivers who rated challenges as "important" or "very important" for the 61 items grouped into the 9 challenge categories. Overall, item means ranged from 2.0 to 3.2. The percentage of respondents who rated challenges as "important" or "very important" ranged from 31% to 92%. The item that was rated highest (92%) was "providing emotional support to my care recipient on challenging days". The item with the lowest percentage (31%) of "important" or "very important" ratings was "feeling ashamed to think about my own well-being or needs".

Support Services Questionnaire

Table 3 shows the percentage of caregivers who rated each of the 18 items as a support service that they were "likely to use" or "very likely to use". Overall, the mean of item ratings ranged from 1.6 to 2.7. The percentage of respondents who rated services as "likely to use" or "very likely to use" ranged from 15% to 59%. The item that was rated highest (59%) was a hard-copy resource for caregivers ("caregiver newsletter"). The item with the lowest percentage (15%) of at least "likely to use" ratings was a telephone-based resource, "caregiver-led telephone-based support group for caregivers".

Table 2. Frequencies for challenge items rated as important and very important among 202 informal caregivers.

	Total Sample		Men versus Women		
	Item Mean (SD)	Important or Very Important n (%)	Men (N = 123): Important or Very Important n (%)	Women (N = 79): Important or Very Important n (%)	95% CI Lower Limit – Upper Limit
Physical health concerns					
1. Experiencing fatigue and physical exhaustion	2.3 (1.2)	79 (39.1)	40 (32.5)	39 (49.4)	2.3 - 30.8
2. Having trouble sleeping	2.2 (1.2)	73 (36.1)	39 (31.7)	34 (43.0)	-2.9 - 25.4
3. Taking care of my health	2.9 (1.1)	134 (66.3)	75 (61.0)	59 (74.7)	-0.4 - 26.5
Financial problems and work or employment problems					
4. Balancing caregiving and demands associated with my job	2.3 (1.2)	86 (42.6)	47 (38.2)	39 (49.4)	-3.5 - 25.4
5. Having to take days off from work due to caregiving responsibilities	2.0 (1.1)	64 (31.7)	35 (28.5)	29 (36.7)	-5.4 - 22.2

6. Managing the cost of drugs and medical care	2.5 (1.1)	107 (53.0)	62 (50.4)	45 (57.0)	-8.2 - 20.8
7. Managing loss of income due to my care recipient's inability to work	2.1 (1.1)	81 (40.1)	43 (35.0)	38 (48.1)	-1.4 - 27.3
Role strain					
8. Balancing caregiving and other family responsibilities	2.6 (1.2)	109 (54.0)	62 (50.4)	47 (59.5)	-5.7 - 23.2
9. Managing last minute changes due to the unpredictability of the disease	2.5 (1.1)	95 (47.0)	54 (43.9)	41 (51.9)	-6.7 - 22.3
10. Having to do all of the winter chores alone due to my care recipient's sensitivity to cold temperatures	2.5 (1.1)	103 (51.0)	64 (52.0)	39 (49.4)	-12.0 - 17.2
11. Having to handle all of the household chores on my own	2.2 (1.2)	74 (36.6)	41 (33.3)	33 (41.8)	-5.7 - 22.6
12. Being unable to help address my care recipient's pain or discomfort	3.1 (0.8)	170 (84.2)	100 (81.3)	70 (88.6)	-4.2 - 17.4
13. Finding time for myself	2.5 (1.2)	103 (51.0)	51 (41.5)	52 (65.8)	9.5 - 37.7

14. Having to learn new skills and abilities because my care recipient can no longer do certain tasks	2.5 (1.2)	100 (49.5)	60 (48.8)	40 (50.6)	-12.8 - 16.4
15. Having to make difficult medical decisions	2.7 (1.1)	130 (64.4)	76 (61.8)	54 (68.4)	-7.8 - 20.0
Information, resources, and support					
16. Not having information about how to be a good caregiver	2.6 (1.2)	101 (50.0)	56 (45.5)	45 (57.0)	-3.4 - 25.5
17. Not being able to find any answers as to why my care recipient got scleroderma	2.3 (1.1)	85 (42.1)	51 (41.5)	34 (43.0)	-12.7 - 16.1
18. Not having access to a caregiver support group	2.1 (1.2)	69 (34.2)	30 (24.4)	39 (49.4)	10.7 - 38.4
19. Not knowing other people who understand what I’m going through	2.2 (1.1)	75 (37.1)	37 (30.1)	38 (48.1)	3.6 - 31.9
20. Navigating healthcare issues while travelling	2.9 (1.1)	139 (68.8)	82 (66.7)	57 (72.2)	-8.5 - 18.5

21. Planning trips and excursions while managing limitations, such as needing wheelchair access or other considerations	2.6 (1.2)	114 (56.4)	70 (56.9)	44 (55.7)	-13.2 - 15.8
22. Having difficulty finding reliable and accurate information about scleroderma	2.6 (1.1)	102 (50.5)	59 (48.0)	43 (54.4)	-8.3 - 20.8
23. Having difficulty understanding important information about scleroderma and its treatment	2.6 (1.2)	105 (52.0)	62 (50.4)	43 (54.4)	-10.7 - 18.4
24. Having difficulty helping my care recipient gain access to knowledgeable health providers	2.5 (1.2)	106 (52.5)	61 (49.6)	45 (57.0)	-7.4 - 21.6
25. Navigating the medical system	2.8 (1.1)	134 (66.3)	79 (64.2)	55 (69.6)	-8.8 - 18.7
26. Interacting with medical, insurance, and social service agencies to address the needs of my care recipient	2.7 (1.1)	126 (62.4)	74 (60.2)	52 (65.8)	-8.8 - 19.3

27. Interacting with health professionals who are not knowledgeable about scleroderma	2.8 (1.0)	135 (66.8)	76 (61.8)	59 (74.7)	-1.2 - 25.7
28. Managing rushed, inconsiderate, or insensitive behavior from health professionals	2.6 (1.1)	113 (55.9)	59 (48.0)	54 (68.4)	5.7 - 33.7
29. Trying to find useful devices to help my care recipient with activities of daily living	2.6 (1.1)	114 (56.4)	65 (52.8)	49 (62.0)	-5.5 - 23.1
30. Finding assistance for things that my care recipient used to do	2.6 (1.2)	104 (51.5)	51 (41.5)	53 (67.1)	10.8 - 38.8
Fear, anxiety, and uncertainty					
31. Being fearful that I will be left alone	2.5 (1.2)	108 (53.5)	65 (52.8)	43 (54.4)	-13.1 - 16.0
32. Constantly worrying about my care recipient's limitations	2.6 (1.1)	102 (50.5)	58 (47.2)	44 (55.7)	-6.2 - 22.8
33. Feeling uncertain about the progression of my care recipient's scleroderma	3.0 (0.9)	145 (71.8)	86 (69.9)	59 (74.7)	-8.9 - 17.4
General emotional difficulties					

34. Feeling helpless	2.8 (1.1)	128 (63.4)	71 (57.7)	57 (72.2)	0.1 - 27.5
35. Feeling hopeless	2.5 (1.2)	107 (53.0)	56 (45.5)	51 (64.6)	4.2 - 32.6
36. Managing my negative emotions towards my care recipient	2.6 (1.1)	118 (58.4)	68 (55.3)	50 (63.3)	-6.6 - 21.9
37. Managing my stress and relaxing	2.9 (1.0)	138 (68.3)	77 (62.6)	61 (77.2)	0.7 - 27.1
38. Managing my negative emotions	2.6 (1.1)	111 (55.0)	64 (52.0)	47 (59.5)	-7.3 - 21.6
39. Guilt about leaving my care recipient alone	2.6 (1.1)	103 (51.0)	58 (47.2)	45 (57.0)	-5.0 - 24.0
40. Feeling ashamed to think about my own well-being or needs	2.1 (1.1)	63 (31.2)	26 (21.1)	37 (46.8)	11.7 - 39.0
Emotional difficulties of the care recipient					
41. Understanding the emotional needs of my care recipient	3.2 (0.8)	169 (83.7)	100 (81.3)	69 (87.3)	-5.6 - 16.3
42. Knowing what to do about my care recipient’s guilt	2.9 (1.0)	134 (66.3)	77 (62.6)	57 (72.2)	-4.6 - 22.6
43. Providing emotional support to my care recipient on challenging days	3.2 (0.7)	185 (91.6)	110 (89.4)	75 (94.9)	-3.8 - 13.4

44. Managing resentment from my care recipient towards me	2.3 (1.2)	82 (40.6)	45 (36.6)	37 (46.8)	-4.3 - 24.5
45. Managing my care recipient's anger about having scleroderma	2.6 (1.1)	118 (58.4)	71 (57.7)	47 (59.5)	-12.8 - 15.9
46. Managing my care recipient's feelings of depression	2.9 (1.0)	144 (71.3)	86 (69.9)	58 (73.4)	-10.2 - 16.3
47. Managing my care recipient's thoughts of ending her or his life	2.3 (1.1)	102 (50.5)	57 (46.3)	45 (57.0)	-4.2 - 24.8
48. Managing the disappointment or frustration of my care recipient when she or he cannot take part in activities	3.0 (1.0)	146 (72.3)	86 (69.9)	60 (75.9)	-7.6 - 18.5
Changes in social interactions with others					
49. Noticing others' lack of knowledge and awareness about scleroderma	2.9 (1.1)	138 (68.3)	72 (58.5)	66 (83.5)	11.4 - 36.6
50. Managing social limitations, such as missing events or having to leave events early	2.4 (1.2)	83 (41.1)	45 (36.6)	38 (48.1)	-3.0 - 25.7

51. Enjoying myself when spending time with friends without my care recipient	2.5 (1.2)	91 (45.0)	46 (37.4)	45 (57.0)	4.7 - 33.4
Changes in relationship dynamics with care recipient					
52. Understanding when my help isn't wanted or needed	2.8 (1.1)	117 (57.9)	67 (54.5)	50 (63.3)	-5.9 - 22.7
53. Helping my care recipient set reasonable limits on activities that have become difficult due to scleroderma	3.1 (0.9)	165 (81.7)	98 (79.7)	67 (84.8)	-7.0 - 15.9
54. Providing needed help when my care recipient doesn't want it or resists it	2.7 (1.2)	111 (55.0)	63 (51.2)	48 (60.8)	-5.2 - 23.5
55. Being patient with my care recipient	2.9 (0.9)	152 (75.2)	93 (75.6)	59 (74.7)	-11.5 - 14.2
56. Finding the balance between interfering and providing care	2.9 (1.1)	127 (62.9)	77 (62.6)	50 (63.3)	-13.7 - 14.6
57. Helping my care recipient feel useful despite her or his physical limitations	3.0 (0.9)	154 (76.2)	92 (74.8)	62 (78.5)	-9.4 - 15.7

58. Feeling a sense of loss because of activities we can no longer do together	2.6 (1.1)	107 (53.0)	55 (44.7)	52 (65.8)	6.3 - 34.5
59. Accommodating my care recipient's diet restrictions when we eat out	2.4 (1.2)	88 (43.6)	49 (39.8)	39 (49.4)	-5.1 - 23.8
60. Discussing emotions or worries concerning scleroderma with my care recipient	2.9 (1.0)	127 (62.9)	73 (59.3)	54 (68.4)	-5.4 - 22.4
61. Dealing with loss of physical intimacy with my care recipient*	2.7 (1.1)	67 (57.3)	53 (56.4)	14 (60.9)	-19.8 - 26.0

Note: CI = confidence interval; *item was optional.

Comparisons Between Women and Men

As shown in table 1, there were significant demographic and caregiving differences between women and men for caregiver age, years of caregiving, and years since the care recipient received their diagnosis. Men were significantly older than women, had been caregiving for longer, and had care recipients who had been diagnosed for a longer time. Current occupational status and relationship to care recipient also differed. A greater proportion of men (48%) were retired compared to women (27%), and men were more often caring for a partner (76%) compared to women (29%). Twenty-seven percent of women were caring for a child whereas just 2% of men were caring for a child.

As shown in table 2, the proportion of women who rated challenges as "important" or "very important" were higher than for men on 59 of 61 items. There were statistically significant differences, after adjusting for multiple comparisons, on five challenges, including "finding time for myself", "not having access to a caregiver support group", "finding assistance for things that my care recipient use to do", "feeling ashamed to think about my own well-being or needs", and "noticing others' lack of knowledge and awareness about scleroderma" as "important" or "very important". For each of these items women rated the challenge as being more important than men.

A greater percentage of women than men rated each of the 18 support service items as "likely" or "very likely" to use. These differences were not statistically significant (see table 3).

Table 3. Frequencies of support service items rated as likely and very likely to use among 202 informal caregivers.

	Total sample		Male versus Female Caregivers		
	Item Mean (SD)	**Likely to Use or Very Likely to Use n (%)**	**Male Caregivers (N=123): Likely to Use or Very Likely to Use n (%)**	**Women (N = 79): Likely to Use or Very Likely to Use n (%)**	**95% CI Lower Limit – Upper Limit**
1. Caregiver internet-based chat group, forum, or social network site without professional moderator	1.8 (1.0)	48 (23.8)	25 (20.3)	23 (29.1)	-3.8 - 22.0
2. Caregiver internet-based chat group, forum, or social network moderated by a	2.3 (1.1)	86 (42.6)	42 (34.1)	44 (55.7)	6.8 - 35.3

knowledgeable healthcare provider					
3. Caregiver-led breakout groups at patient conferences	2.0 (1.0)	59 (29.2)	28 (22.8)	31 (39.2)	2.8 - 30.0
4. Professionally led breakout groups at patient conferences	2.2 (1.1)	74 (36.6)	36 (29.3)	38 (48.1)	4.4 - 32.6
5. Conference caregiver educational sessions and workshops provided by a knowledgeable healthcare provider	2.4 (1.1)	94 (46.5)	54 (43.9)	40 (50.6)	-7.9 - 21.1
6. Internet-based psychological and emotional self-help tools	2.1 (1.1)	74 (36.6)	37 (30.1)	37 (46.8)	2.4 - 30.6
7. One-to-one peer support (e.g., the ability to call	1.9 (1.0)	54 (26.7)	27 (22.0)	27 (34.2)	-1.0 - 25.7

another caregiver on the phone)					
8. Professionally led in-person caregiver support group	2.1 (1.1)	73 (36.1)	41 (33.3)	32 (40.5)	-6.9 - 21.4
9. Caregiver-led in-person caregiver support group	1.9 (1.0)	53 (26.2)	30 (24.4)	23 (29.1)	-8.1 - 18.2
10. Professionally led telephone-base support group for caregivers	1.8 (1.0)	41 (20.3)	20 (16.3)	21 (26.6)	-1.7 - 23.1
11. Caregiver-led telephone-based support group for caregivers	1.6 (0.9)	30 (14.9)	16 (13.0)	14 (17.7)	-5.8 - 16.5
12. Professionally led internet-based, live interaction	1.9 (1.0)	55 (27.2)	25 (20.3)	30 (38.0)	4.2 - 31.0

(teleconference, Skype) caregiver support group					
13. Caregiver-led internet-based, live interaction (teleconference, Skype) caregiver support group	1.7 (0.9)	43 (21.3)	19 (15.4)	24 (30.4)	2.5 - 27.8
14. Caregiver newsletter	2.7 (1.0)	118 (58.4)	68 (55.3)	50 (63.3)	-6.6 - 21.9
15. Retreat for caregivers	1.8 (1.0)	48 (23.8)	24 (19.5)	24 (30.4)	-1.8 - 24.0
16. Online educational sessions for caregivers to help understand scleroderma and its impact on families	2.4 (1.1)	98 (48.5)	52 (42.3)	46 (58.2)	1.1 - 29.9
17. Information package/pamphlet about	2.5 (1.1)	109 (54.0)	61 (49.6)	48 (60.8)	-3.6 - 25.1

	scleroderma for caregivers of newly diagnosed patients					
18.	Information about scleroderma on an online reputable website for caregivers of newly diagnosed patients	2.7 (1.1)	111 (55.0)	60 (48.8)	51 (64.6)	1.0 - 29.4

Note: CI = confidence interval.

DISCUSSION

We surveyed 202 informal caregivers of people with SSc to determine the most important challenges and the likelihood that caregivers would use different types of support services, if available. Challenges that were most consistently rated as being "important" or "very important" were related to difficulty addressing the emotional needs of the care recipient (e.g., "providing emotional support to my care recipient on challenging days") and feeling unable to lessen the care recipients discomfort (e.g., "being unable to help address my care recipient's pain or discomfort"). Alternatively, items related to financial, work, and employment difficulties were the least likely to be rated as being important.

Among support services that were included in the survey, at least 50% of caregivers rated being "likely" or "very likely" to use information-based support services, including those provided soon after SSc diagnosis (e.g., "information about scleroderma on an online reputable website for caregivers of newly diagnosed patients"). On the other hand, few informal caregivers rated interactive support services such as support groups or peer support as resources that they would "likely" or "very likely" use (e.g., "caregiver-led in-person caregiver support group").

Only five challenges had statistically significant differences between women and men, controlling for multiple comparisons. Women and men differed substantively and statistically significantly in terms of demographics and the characteristics of their care recipients. For example, the majority of men cared for a partner; whereas, women often cared for a partner, child, or parent. Given the large number of items and the relatively small numbers of women and men in certain characteristics where they differed (e.g., relation to care recipient), we did not conduct multivariable analyses, and it is possible that some of the differences identified may

reflect factors unrelated to gender. In addition to being more likely to rate challenges as at least "important", women rated being more likely to use the 18 support service items.

Prior to our NGT study, the only previous study on caregivers of people living with SSc that we identified was a doctoral thesis involving individual interviews with 13 caregivers (Maril, 2012). Consistent with the present study, the thesis emphasized difficulties of caregivers in addressing the emotional difficulties of care recipients.

Caregivers who completed our survey indicated that they preferred information-based supports. We have not identified studies of supportive interventions tested among SSc caregivers, however, there are websites that provide information for informal SSc caregivers (Scleroderma Foundation, n.d.; Scleroderma Quebec, 2016). Many different kinds of support services have been developed and tested among caregivers to persons with more common diseases (Boots, de Vugt, van Knippenberg, Kempen, & Verhey, 2014; Dobson, Upadhyaya, McNeil, Venkateswaran, & Gilderdale, 2001; Pinquart & Sörensen, 2006), including newsletter resources, information packages, support groups, and psychotherapy tailored for caregivers. In a survey of 188 caregivers to elderly individuals, similar to our findings, caregivers reported being more interested in a newsletter developed for caregivers than interventions that require face-to-face or simultaneous virtual contact, such as support groups or help from a volunteer (Colantonio, Kositsky, Cohen, & Vernich, 2001).

The use of information packages delivered online or through hard-copy resources has been previously studied. For example, among caregivers of children with special needs, a "Keeping it Together" (KIT) information package has been tested in Canada and Australia (Stewart et al., 2006; Stewart, Galvin, Froude, & Lentin, 2010). This package included information about accessing resources and communicating information to care recipients. In Canada, the utility of

the KIT was evaluated among 440 parents of children with special needs. After using the KIT, parents' perceptions of their ability, confidence, and satisfaction in using information in different settings significantly improved (Stewart et al., 2006). Tailoring similar interventions to caregivers of persons with SSc may help to alleviate burden associated with the challenges faced in their caregiver role.

Web-based psychoeducation interventions delivered soon after diagnosis have also been tested among informal caregivers to persons with more common health conditions such as cancer. For example, a program that was originally nurse-delivered was adapted to be applied online for patients newly diagnosed with cancer and their family members (Northouse et al., 2014). The web-based intervention provided information and support tailored to the needs of patients and their caregiver (e.g., communicating with each other). Thirty-eight dyads accessed the online program which included three sessions. After delivery of the intervention, significant reductions in emotional distress were found among patients and caregivers, as well increased quality of life and perceived benefits of caregiving (Northouse et al., 2014). Participants reported being satisfied with the program usability but wanted additional content. Caregivers in our study rated support services that could be used independently and in their home higher than support services that involved other caregivers or professionals, suggesting that developing web-based psychoeducational content for caregivers of persons with SSc may be an intervention that caregivers would be likely to use.

Important limitations should be considered when interpreting study findings. First, participants were recruited through an ongoing SSc patient cohorts, patient organizations, and social media websites. These recruitment methods could have resulted in an over representation of caregivers who are actively involved in their care recipient's diagnosis. Second, this survey

was distributed online among caregivers that were fluent in English or French, therefore only caregivers with a computer and internet access could participate. Lastly, our study surveyed the challenges and support services preferences of caregivers but did not explore why certain challenges were deemed more important than others, or why the support service preferences were selected as being likely or unlikely to be used. Understanding the rationale for these preferences could help in better tailoring support services for caregivers to persons with SSc.

In sum, caring for a person with SSc can be a challenging role. The most important challenges to caregivers involved supporting their care recipients with their emotional difficulties and physical discomfort. Caregivers indicated that they would be most likely to utilize hardcopy and internet-based information resources. Providing a caregiver information newsletter in addition to reliable information about SSc may help to address caregiver needs. Future research should consider the challenges and support service preferences that caregivers have identified to develop and test interventions that positively impact the caregiving experience.

Acknowledgements

We thank our Scleroderma Caregiver Advisory Team Members: Marcia Greiten, BA, Goleta, USA; Christy McCaffery, Philadelphia, USA; Judi McDonald, Bedford, Canada; John Michalski, Macomb, USA; Mathieu Ross, Quebec City, Canada; Lisa Spinney, Berlin, USA; Terry Stacey, Oshawa, Canada; Linda Tarantino, Larkspur, USA.

Funding

This research was supported by a grant from the Rare Disease Foundation and the BC Children's Hospital Foundation (Funding Reference Number 174-16; PI Rice). Ms. Rice was supported by a Vanier CIHR Graduate Scholarship. Dr. Thombs was supported by a Fonds de recherche du Québec – Santé researcher salary award. No funding body had any role in the design, collection, analysis or interpretation of data; in the writing of the manuscript; or in the decision to submit the manuscript for publication.

Authors' Contribution

DBR, and BDT were responsible for the study conception and design; data acquisition, analysis, and interpretation; and drafting and revising the manuscript. DBR, MCA, and ACJ were responsible for data acquisition, analysis, and interpretation. DBR and BDT drafted and revising the manuscript. MEC and JC were responsible for acquisition of data. VLM, MH, and the Scleroderma Caregiver Advisory Committee were responsible for interpretation of data analysis. All authors reviewed, provided critical input, and approved the final version of this manuscript.

Declaration of Interest Statement: The authors report no conflicts of interest.

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Chapter 5

General Discussion

Summary of Main Findings

The purpose of this thesis was to develop an understanding of the experience of caring for a loved one diagnosed with SSc. This work aimed to consider the role of informal caregivers to persons with SSc by building upon available evidence that has focused on caring to individuals with any rare disorder; and through engaging people with lived experience in the research design process. Mixed methods research was conducted to collect diverse information including outcomes of previously conducted studies of interventions to support caregivers through a scoping review, a series of focus groups and a cross-sectional survey of caregivers to persons with SSc.

The aim of the first manuscript in this thesis was to systematically review and map the available literature reporting on 1) the benefits of participating in psychosocial interventions delivered to caregivers to a loved one with a rare disorder and 2) the barriers and facilitators of establishing and maintaining these interventions. Thirty-four publications, including 17 trials were identified and included in the review. All included studies reported on tested or perceived benefits of caregivers to a loved one with a rare disorder participating in a psychosocial intervention. Reported reductions in stress, caregiver burden, and feelings of isolation after participating in a psychosocial intervention were found, however, few improvements in physical or mental health (i.e., symptoms of depression or anxiety) were reported. For identified benefits to occur and for intervention delivery to be sustained requires consideration of the facilitators and barriers to establishing and maintaining interventions for caregivers to a loved one with a rare disease. Engaging caregivers in the design of these interventions (e.g., tailoring sessions to specific caregiver needs) while reducing identified barriers (e.g., accessibility issues of the

intervention) may enhance the sustainability of psychosocial interventions for caregivers. Overall, the findings of this review highlight the positive impact that psychosocial interventions delivered to caregivers can have in improving stress, burden, and isolation, while identifying the challenges associated with establishing and maintaining these interventions over time, given the rarity of the disorders.

The objective of the second manuscript was to use structured group discussions through use of the nominal group technique with caregivers to people with SSc in order to develop survey items that assess: (1) challenges experienced related to caregiving in SSc and (2) preferences for types of support services that could potentially be developed. Thirteen caregivers in 3 separate group sessions from Canada and the US generated a list of challenges where a lack of information, resources, and support was identified most often. A list of support services was also generated and those that could be delivered online or in person were identified most often, while fewer hard-copy resources or phone delivered services were generated. The use of the nominal group technique allowed study participants to share their perspectives and provided a mechanism for direct caregiver input during the development of survey items for caregivers to persons with SSc.

The third manuscript involved disseminating a survey online to caregivers of people with SSc from 3 continents in order to evaluate the importance of challenges and the priorities for support services that had been identified in the focus groups. Two hundred and two informal caregivers completed the survey (61% male, 39% female) where items related to supporting the care recipient with their emotional difficulties and physical discomfort were reported as being most important to caregivers. Caregivers indicated that they would be most likely to use online or hard-copy support services that involved information resources and less likely to engage in services that involved interacting with others. Delivering services soon after diagnosed that can

be accessed independently were rated as more likely to be used, as compared to services that would be available later within the course of the disease. Caregivers generally rated the importance of challenges higher than the likelihood of using support services. While our survey did not ask why caregivers chose the support services that they did, the quantity and importance of challenges identified, suggests that caregivers may feel overwhelmed, and incorporating additional in person commitments could be perceived as adding to the multiple tasks that they are already responsible for. The findings from this survey provide evidence that can be used to develop support services that could directly address aspects of caring found to be most challenging and for the delivery of services to align with caregiver preferences.

Implications of Findings and Directions for Future Research

Overall, results from the present thesis extend and enhance existing research on the experience of caregivers to people living with SSc. The findings from the present research suggest that numerous unique and important challenges are faced when caring to a loved one with a rare disease, such as SSc. Findings also suggest that psychosocial interventions that are designed to be disease-specific, can be an important source of support for caregivers. Firstly, this research identified and synthesized available literature related to interventions that have been studied among caregivers to a loved one with a rare disease. This allows caregivers, researchers, and health system decision makers to have access to an inclusive list of available interventions that could be implemented and offered to caregivers providing support to individuals with a rare disorder. It also presents an overview of the facilitators and barriers that impact the sustainability of interventions, which is especially important given the rarity of conditions and the lack of resources and services that are integrated into the healthcare system. Unlike psychosocial interventions for caregivers to individuals with a common medical illness, where educational

materials or support groups for caregivers to a loved one with cancer are often available within the healthcare system or through well-resourced national or regional organizations; addressing the barriers to maintaining interventions for rare disorders is necessary the sustain services. Secondly, this thesis built on the limited understanding of the experiences among informal caregivers to a loved one with SSc. The various challenges faced by caregivers and the importance of these challenges suggests that services that are developed to address the difficulty supporting the care recipient in managing emotional difficulties and physical discomfort delivered soon after the care recipient's diagnosis may be most likely to be used among caregivers to a loved one with SSc. Thirdly, this work expands the ways in which engaging people with lived experience can occur within rare disease research. Involving people with lived experience in rare disease research is especially important and allows for improved targeting of outcomes that are relevant to patients and caregivers (Young et al., 2019). A previous systematic review of patient engagement in rare disease research found that people with lived experience are typically engaged in research by contributing to outcome selection, recruitment, promoting research through patient networks, and providing financial support for research infrastructures (Forsythe et al., 2014). This thesis extends engagement within rare disease research by directly eliciting survey items from caregivers. This may provide an additional opportunity for engaging people with lived experience in rare disease research in future studies.

As the number of rare disorders continues to increase, and the need to manage and support these chronic conditions continues to rise, promoting the mental and physical well-being of caregivers is essential to consider for the well-being of the individuals themselves, their care recipients, and the health system (Tracy, Nickell, & Upshur, 2019). Given the pronounced lack of support that Canadian caregivers to individuals with a rare disease have reported in national surveys (Canadian Organization for Rare Disorders, 2019), developing and implementing

interventions that are disease specific and easily accessible to caregivers may improve the well-being of caregivers.

Findings from the current work present important future directions for SSc organizations which may strive to decrease the burden faced by informal caregivers. A recent study conducted alongside (but not included in) the present thesis involved conducting a systematic review and meta-analysis of the burden reported by caregivers for individuals with any chronic disease, excluding cancer or disease primarily impacting cognitive impairment (Carboni-Jimenez, Rice, Chiovitti, Canedo-Ayala, & Thombs, 2019). This review included 84 studies of caregivers to people living with various chronic diseases and had reported Zarit Burden Index (ZBI) scores, as this is a measure of perceived caregiver burden. One study of caregivers to a loved one with SSc was included in the review and had been conducted as part of the survey associated with the current thesis (Cañedo-Ayala et al., 2020). These caregivers had the highest reported burden scores (ZBI-12 mean = 13.5, standard deviation [SD] = 9.8) as compared to all other studies that collected the ZBI-12, which included studies of caregivers in which the care recipients had a primary diagnosis of essential tremor, spinal cord injury, cirrhosis, amyotrophic lateral sclerosis (ALS), or heart failure. Within all other conditions, lower burden scores were reported by caregivers, with ZBI-12 scores ranging from a mean of 6.4 (SD = 8.4) to 12.4 (SD = 7.9), as compared to SSc caregivers where the mean ZBI-12 scores was 13.5. Taken with the current thesis findings, this further highlights the importance of developing and providing access to information-based services to lessen the intensity of caregiver burden and the numerous challenges faced among caregivers to persons with SSc.

The studies conducted as part of this thesis addressed gaps that had been highlighted in previous research and identified directions for future program development and research. The potential development of an online resource, such as a website that can be accessed soon after

diagnosis, should incorporate content that is educational and addresses ways to manage the difficulty supporting a care recipient's emotional challenges and physical discomfort. Hard copy resources were also rated as being more likely to be used among caregivers to those with SSc. Creating hard copy resources that are available at a specialist health care providers office may provide an opportunity to give patients and caregivers immediate access to information and a link to an educational website specific to SSc. Considering these findings within future program development may help to enhance the utility and sustainability of an intervention, while addressing the specific challenges and preferences identified by caregivers. Further, gaps in research were identified with the current work. Future research that accounts for limitations of the included studies and replicates the present research is also needed. For example, studies that assess the caregiving experience among individuals who are not well connected with national organizations may be important in order to consider the variability of experiences for those less engaged in the SSc community. Future research that extends subgroup and comparative analyses would also allow for considering differences in caregiver needs based on demographic and caregiver characteristics such as race/ethnicity, the number of years one was been a caregiver, and the relationship between the caregiver and care recipient. Finally, conducting pre-post studies, including high-quality randomized controlled trials with an adequate sample size would allow researchers to assess the efficacy of implementing a specific intervention for caregivers to persons with SSc.

Limitations

There are limitations that should be considered when interpreting the present research. Firstly, given the nature of a scoping review, results were synthesized from studies that were conducted across various rare diseases, applied different study design methods, and collected

various outcome measures. Next, although two researchers extracted outcomes of interest, extracting accurate and complete facilitator and barrier data required researchers to interpret the data, as studies did not consistently label specific findings as a facilitator or barrier.

For studies involving primary data collected from focus groups and the survey constituted convenience samples that were recruited from SSc patient conferences, patient cohorts, patient organizations, SSc newsletters, and social medial websites. These recruitment methods may have resulted in samples that were overrepresented by caregivers actively involved in the SSc community; therefore, these results may not reflect what would be found within a community setting. These studies were also restricted to caregivers living in countries with a "very high" human development index, specifically, those living in Canada, the United States, France, the United Kingdom, and Australia, and findings may not generalize to caregivers living in other regions.

Another limitation relevant to each manuscript was that the grouping and categorization of qualitative information, including the narratively reported benefits, facilitators, and barriers within the scoping review, and the challenges and support preference item groupings were ultimately subjective. However, these groupings and themes were reviewed iteratively by a minimum of two members of the research team, and for the focus group and survey responses, groupings were reviewed by all members of the research team which included representatives from a Caregiver Advisory Committee, and clinicians and researchers with expertise in SSc. Thus, we feel confident that the groupings provide a reasonable structure for interpreting study findings.

Conclusion

The aim of the present research was to understand the experience of caring for someone living with SSc and to conduct a needs assessment to identify caregiver priorities while embedding people with lived experience into the research process. The research intended to fulfil previously identified gaps in caregiver research by guiding intervention development through determining the resources available for caregivers of other rare disorders, conducting quantitative studies with an adequate sample size, and including male caregivers to better represent the demographic characteristics of caregivers to those with SSc. These objectives were achieved by (1) determining psychosocial interventions tested among caregivers of individuals with a rare disease, (2) conducting structured groups using the nominal group technique where caregivers to people with SSc generated a list of items related to challenges associated with caregiving and the preferences for support services that could be developed, and (3) determining the relative importance of the identified challenges and the likelihood of using support services through conducting a survey with a large sample of caregivers.

The findings from the present research highlight the significant challenges faced by individuals that take on the role of an informal caregiver to a loved one living with a rare disorder, including SSc. At the same time, results demonstrate the potential benefits of implementing psychosocial interventions when caregiving in the context of a rare disorder. This research also demonstrates that despite the obstacles in conducting research in a rare disease environment, engaging caregivers in the research process and obtaining a high level of participation may be possible when connected to a rare disease community and recruiting from multiple countries to account for the low prevalence within any given location.

Altogether, the present findings provide a helpful starting point in the development of interventions to address challenges faced by caregivers to those with SSc. Reviewing content of

interventions that have been successfully implemented and maintained with favourable results among caregivers to care recipients with other rare disease groups may allow for existing interventions to be adapted for caregivers to a loved one with SSc. Developing resources that closely align with the challenges most important to caregivers would also likely improve the utility of a newly developed intervention. This thesis considered the unique experience of caring for a loved one with a rare disorder as compared to a common disorder. This work also emphasizes the need to consider how the healthcare system and society can better support the needs and challenges of caregivers to a loved one with a rare disease.

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Appendix A. Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) Checklist

SECTION	ITEM	PRISMA-ScR CHECKLIST ITEM	REPORTED ON PAGE #
TITLE			
Title	1	Identify the report as a scoping review.	1
ABSTRACT			
Structured summary	2	Provide a structured summary that includes (as applicable): background, objectives, eligibility criteria, sources of evidence, charting methods, results, and conclusions that relate to the review questions and objectives.	23
INTRODUCTION			
Rationale	3	Describe the rationale for the review in the context of what is already known. Explain why the review questions/objectives lend themselves to a scoping review approach.	4-6
Objectives	4	Provide an explicit statement of the questions and objectives being addressed with reference to their key elements (e.g., population or participants, concepts, and context) or other relevant key elements used to conceptualize the review questions and/or objectives.	6
METHODS			
Protocol and registration	5	Indicate whether a review protocol exists; state if and where it can be accessed (e.g., a Web address); and if available, provide registration information, including the registration number.	6
Eligibility criteria	6	Specify characteristics of the sources of evidence used as eligibility criteria (e.g., years considered, language, and publication status), and provide a rationale.	7-8
Information sources*	7	Describe all information sources in the search (e.g., databases with dates of coverage and contact with authors to identify additional sources), as well as the date the most recent search was executed.	8-9
Search	8	Present the full electronic search strategy for at least 1 database, including any limits used, such that it could be repeated.	Appendix B
Selection of sources of evidence†	9	State the process for selecting sources of evidence (i.e., screening and eligibility) included in the scoping review.	9
Data charting process‡	10	Describe the methods of charting data from the included sources of evidence (e.g., calibrated forms or forms that have been tested by the team before their use, and whether data charting was done independently or in duplicate) and any processes for obtaining and confirming data from investigators.	9-10
Data items	11	List and define all variables for which data were sought and any assumptions and simplifications made.	9-10
Critical appraisal of individual sources of evidence§	12	If done, provide a rationale for conducting a critical appraisal of included sources of evidence; describe the methods used and how this information was used in any data synthesis (if appropriate).	N/A
Synthesis of results	13	Describe the methods of handling and summarizing the data that were charted.	10-11
RESULTS			

SECTION	ITEM	PRISMA-ScR CHECKLIST ITEM	REPORTED ON PAGE #
Selection of sources of evidence	14	Give numbers of sources of evidence screened, assessed for eligibility, and included in the review, with reasons for exclusions at each stage, ideally using a flow diagram.	Appendix C
Characteristics of sources of evidence	15	For each source of evidence, present characteristics for which data were charted and provide the citations.	12; Tables 1-4
Critical appraisal within sources of evidence	16	If done, present data on critical appraisal of included sources of evidence (see item 12).	N/A
Results of individual sources of evidence	17	For each included source of evidence, present the relevant data that were charted that relate to the review questions and objectives.	Tables 1-6
Synthesis of results	18	Summarize and/or present the charting results as they relate to the review questions and objectives.	13-18
DISCUSSION			
Summary of evidence	19	Summarize the main results (including an overview of concepts, themes, and types of evidence available), link to the review questions and objectives, and consider the relevance to key groups.	18-21
Limitations	20	Discuss the limitations of the scoping review process.	21
Conclusions	21	Provide a general interpretation of the results with respect to the review questions and objectives, as well as potential implications and/or next steps.	22
FUNDING			
Funding	22	Describe sources of funding for the included sources of evidence, as well as sources of funding for the scoping review. Describe the role of the funders of the scoping review.	23

JBI = Joanna Briggs Institute; PRISMA-ScR = Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews.

* Where *sources of evidence* (see second footnote) are compiled from, such as bibliographic databases, social media platforms, and Web sites.

† A more inclusive/heterogeneous term used to account for the different types of evidence or data sources (e.g., quantitative and/or qualitative research, expert opinion, and policy documents) that may be eligible in a scoping review as opposed to only studies. This is not to be confused with *information sources* (see first footnote).

‡ The frameworks by Arksey and O'Malley (6) and Levac and colleagues (7) and the JBI guidance (4, 5) refer to the process of data extraction in a scoping review as data charting.

§ The process of systematically examining research evidence to assess its validity, results, and relevance before using it to inform a decision. This term is used for items 12 and 19 instead of "risk of bias" (which is more applicable to systematic reviews of interventions) to include and acknowledge the various sources of evidence that may be used in a scoping review (e.g., quantitative and/or qualitative research, expert opinion, and policy document).

From: Tricco AC, Lillie E, Zarin W, O'Brien KK, Colquhoun H, Levac D, et al. PRISMA Extension for Scoping Reviews (PRISMAScR): Checklist and Explanation. Ann Intern Med. 2018;169:467–473. doi: 10.7326/M18-0850.

Appendix B: Search Strategy

PubMed
(caregiv* OR care giv* OR "carer" OR "carers") OR "Caregivers"[Mesh] OR "Home Nursing"[Mesh]
AND
("intervention"[Title/Abstract] OR "trial"[Title/Abstract] OR "training"[Title/Abstract] OR "counselling"[Title/Abstract] OR "counseling"[Title/Abstract] OR "support"[Title])
AND
[Each of the 7 sets of rare disease terms (See DisSub1000.txt)]
AND
Entrez date: 2000/01/01 to present
Limit to Humans
No limits by language, publication type

CINAHL
1. (MH "Caregivers")
2. (MH "Home Nursing")
3. (caregiv* or care giv* or "carer" or "carers")
4. S1 or S2 or S3
5. ("intervention" or "trial" or "training" or "counselling" or "counseling" or "support")
6. [Each of the 7 sets of rare disease terms]
7. S4 AND S5 AND S6
Limiters: Published Date: 20000101-20181231

PsycINFO
1. CAREGIVERS/
2. Home care/
3. (caregiv* or care giv* or carer or carers).mp.
4. 1 or 2 or 3
5. (intervention or trial or training or counselling or counseling or support.mp.
6. [Each of the 7 sets of rare disease terms]
7. 4 and 5 and 6
8. limit 7 to yr= "2000 – 2018

Appendix C. List of Studies Excluded at Full-Text Level

Reference	Exclusion Criteria
A'Campo LE, Wekking EM, Spliethoff-Kamminga NG, Stijnen T, Roos RA. Treatment effect modifiers for the patient education programme for Parkinson's disease. International Journal of Clinical Practice. 2012 Jan;66(1):77-83.	Not rare disease caregivers
Bishop D, Miller I, Weiner D, Guilmette T, Mukand J, Feldmann E, Keitner G, Springate B. Family Intervention: Telephone Tracking (FITT): a pilot stroke outcome study. Topics in Stroke Rehabilitation. 2014 Mar 1;21 Suppl 1:S63-74.	Not rare disease caregivers
Brewer HM, Smith JA, Eatough V, Stanley CA, Glendinning NW, Quarrell OW. Caring for a child with juvenile Huntington's disease: helpful and unhelpful support. Journal of Child Health Care. 2007 Mar;11(1):40-52.	No eligible intervention
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Appendix D. Focus Group Guide

Note: *All participants must be informal caregivers of a patient diagnosed with scleroderma. Each focus group will have the same set of interview questions (please see below). All focus groups will be conducted at patient conferences and will be led by two moderators. Individuals will be emailed the consent form and demographics questionnaires prior to the focus groups. Demographics questionnaires will be collected prior to beginning the focus groups.*

General Introduction, Ground Rules and Opening Questions

Welcome to our session. We want to thank you all for taking the time to join us in talking about your experiences helping to care for a loved one with scleroderma. My name is Danielle Rice and I will be leading the focus group today along with Dr. Brett Thombs. We're both from McGill University in Montreal, Canada. We would like to develop, test, and make available effective support services for informal caregivers, like yourselves, in scleroderma. In order to develop effective support services, we need to begin by understanding the challenges, specific needs, and preferences you have regarding support services. The focus groups we are holding today are a first step. We are going to have discussions like this with additional groups today and within the next few months.

You were invited to participate because you help to provide care for a family member or friend with scleroderma. In our discussions today there are no wrong answers, but rather differing points of view. Please feel free to share your point of view even if it differs from what others have said. Although you may not agree with what others have said, we ask that you listen respectfully as others share their views and that there is only one person speaking at a time. We also ask that you turn your cell phones on silent while we are having our discussion today.

You've probably noticed the audio recorders that we have here. We're tape recording the

session because we don't want to miss any of your comments. People often say very helpful things in these discussions and we can't write fast enough to get them all down. Although we will be on a first name basis during our discussion, we won't use any names in our reports. We would also like for everyone to keep the discussions that we have today in this room. The discussions that we have should not be shared with others to respect the confidentiality of everyone participating. Does anyone have any questions?

Questions for Informal Scleroderma Caregivers Focus Groups:

Well, let's begin. We've placed name cards on the table in front of you to help us remember each other's names. Let's find out some more about everyone by going around the table. Tell us your name, where you live and one surprising thing about yourself."

Now we would like to ask some questions about your experiences as caregivers. We have 3 main questions about challenges, support needs, and preferences about possible ways to provide support for informal caregivers like you. For each question, the question will be posed to everyone and then each of you will have a few minutes to write your responses down on paper. After everyone has written down their responses, we will go around the table and ask people to share one item at a time from their list until all items have been heard. Once all items have been shared we will discuss why certain items were identified. Then we will have one large list of responses. We will print the list of these responses, and each of you will have the chance to rate the importance of each item for you.

The first question today is about the challenges you have experienced when helping to care for your family member or friend with scleroderma. Think about the challenges you have faced since taking on a caregiving role to somebody close to you with scleroderma (*write this on board)*. For example, some caregivers have described challenges related to providing

transportation to appointments, taking off time at work or balancing caregiving with their other responsibilities. Others describe more emotional challenges.

(*We will also write this question down on a board for all participants to* see). Please take a few minutes and write these challenges down on a piece of paper. When you're finished, we will share these challenges with everyone.

Give participants time to make a list of challenges (approximately 10 minutes).

Now I'd like everyone to share one item on your list and we will continue going around in this format until all items on each of your lists have been included. We ask for you to only share items that haven't been mentioned yet. *(One of the moderators will write down each item on a list for everyone to see).*

(Once all items have been shared). Now let's all take a look at this list, are there any items that you think can be combined or removed? If so, why? (*Group discussion is appropriate at this stage for clarification and discussion of the list).* Do you think there are any important challenges that we're missing here?

(One of the moderators will have typed out the final list and printed a copy for each participant with a rating scale, 1-10 → with 1 meaning not at all important and 10 meaning extremely important).

We'd like all of you to look at your printed copy of the list that was developed and rate each item on a scale of 1 to 10, with 1 representing challenges that are not important to you personally, and 10 being challenges that are extremely important to you in your role as an informal caregiver.

Finally, we'd like to ask you about your preferences for potential support services, and the ideal format for these services. Think about services that could be put in place to provide better support to scleroderma caregivers. What programs, services/and or supports would be helpful in

your role as a caregiver? How would these programs, services and/or other supports operate? For example, some caregivers attend support groups. The format of these support groups may be in person, online or telephone based. Other caregivers have suggested that educational services in lecture format for caregivers would be beneficial when a loved one is diagnosed with scleroderma.

(*We will also write this question down on a board for all participants to* see). Please take a few minutes and write down on a piece of paper support services that you think would be helpful, and the way in which these services could be applied. When you're finished, we will share the list of support services with everyone.

Give participants time to make a list of supportive services (approximately 10 minutes).

Now I'd like everyone to share one item on your list and we will continue going around in this format until all items on each of your lists have been included. We ask for you to only share items that haven't been mentioned yet. *(One of the moderators will write down each item on a list for everyone to see).*

(Once all items have been shared). Now let's all take a look at this list, are there any items that you think can be combined or removed? If so, why? (*Group discussion is appropriate at this stage for clarification and discussion of the list).* Do you think there are any important support services that we're missing here?

(One of the moderators will have typed out the final list and printed a copy for each participant with a rating scale, 1-10 → with 1 meaning would not likely use and 10 would very likely use).

We'd like all of you to look at your printed copy of the list that was developed and rate each item on a scale of 1 to 10, with 1 representing supportive services that you would not likely use personally, and 10 being supportive services that you are very likely to use.

Conclusion

End by saying; We've come to the end of our focus group now. We'd like to thank you for your participation today. Does anyone have any final questions?

Appendix E. Category Descriptions

Physical health concerns: challenges related to the physical health or limitations of the caregiver, including sleep problems and fatigue.

Financial problems and work or employment problems: challenges associated with financial struggles, work scheduling, and work-associated changes.

Role strain: challenges associated with role fulfillment or the perceived inability to complete key roles, including family roles, understanding or managing the caregiving role, managing many tasks at once (excluding challenges specific to paid employment), and challenges in learning new skills to fulfill the caregiving role. Does not include challenges related to the quality of the relationship with the care recipient or challenges when interacting with the care recipient, changes to social interactions with friends, or challenges associated with work and employment.

Information, resources, and support needs: challenges related to obtaining or accessing information or supportive resources needed by caregivers. This may involve information that would be obtained from medical professionals, other informal caregivers, legal systems, or health care systems, such as information on how to obtain health insurance, where to obtain caregiving resources, or how to obtain information about managing the care recipient's limitations. Challenges related to interactions with health professionals are included.

Fear, anxiety, and uncertainty: challenges involving feelings of fear, anxiety, or uncertainty about the future, including the unpredictability of the care-recipient's disease; fear about medical procedures; or fear-related thoughts or emotions.

General emotional difficulties: challenges involving negative feelings, or negative affect other than fear and anxiety, such as negative feelings directed towards the self (e.g., guilt).

Emotional difficulties of the care recipient: challenges involving helping the care recipient with emotions and feelings, but not including challenges related to how emotional difficulties affect the interaction of the caregiver and care recipient relationship.

Changes in relationship dynamics with the care recipient: challenges related to changes or difficulty in the quality of the relationship with the care recipient, including changes to interactions, communication, and relationship dynamics and changes in intimacy and sexual issues between caregiver and care recipient partners. Does not include challenges related to negative feelings or anxiety directed towards the self or the care recipient.

Changes in social interactions: items that involve changes or difficulty in social interactions with others outside of the family and leisure activities, but not including changes in activities core to the caregiver and care recipient relationship.

Appendix F. List of All Original Caregiver Generated Challenges and Item Means

Item	Focus Group Number	Mean Rating of Challenge Importance (1-10)	Number of Participants That Rated the Item
1. Having to take time away from work	1	4.0	2
2. Balancing caregiving and work demands	1	6.0	2
3. Fatigue and physical exhaustion	1	5.0	2
4. Inability to do the things we used to do as a couple	1	6.5	2
5. Inability to help address pain	1	4.0	2
6. Resentment from care recipient directed at caregiver	1	4.0	2
7. Managing frustration and anger towards loved one	1	4.0	2
8. Difficulty sleeping	1	5.0	2
9. Difficulty managing stress and relaxing	1	7.5	2
10. Managing multiple caregiving responsibilities	1	6.5	2
11. Finding time for myself	1	8.5	2
12. Feeling helpless	1	7.0	2

13. Feeling hopeless	1	3.0	2
14. Others not recognizing my care recipient's illness and needs	1	9.0	2
15. Losing things that we liked to do together	1	5.0	2
16. Worry about an uncertain future	1	9.0	2
17. Can't get away to do things that I like to do	1	2.5	2
18. Lifestyle changes in my own life to accommodate my care recipient's needs	1	6.5	2
19. Finding help for things that my care recipient used to do	1	5.0	2
20. Ability to find reliable and accurate information about scleroderma	1	3.5	2
21. Difficulty understanding important information about scleroderma	1	2.5	2
22. Finding the right medical person	1	7.5	2
23. Getting access to a knowledgeable rheumatologist	1	7.5	2
24. Navigating the medical system	1	8.0	2
25. Insensitivity of others	1	9.0	2
26. Lack of awareness of others about scleroderma	1	9.5	2
27. Fear that I will be left alone	1	8.5	2

28. Not being able to find any answers to why this happened	1	8.5	2
29. Managing insurance restrictions for getting needed care	1	8.5	2
30. Expense of drugs and medical care	1	8.5	2
31. Financial considerations due to loss income	1	7.0	2
32. Guilt about leaving my care recipient alone	1	6.5	2
33. Guilt that I could have done something differently that would have prevented scleroderma	1	5.0	2
34. Resisting tendency to overprotect or “smother” my care recipient with attention	1	7.5	2
35. Allowing my care recipient to do things for himself or herself when appropriate	1	5.0	2
36. Not having access to a caregiver support group	1	10.0	2
37. Not having information about how to be a good caregiver	1	9.0	2
38. Not knowing other people who understand what I’m going through	1	9.0	2
39. Knowing how much to help	2	5.8	5
40. Being able to help set limits and establishing boundaries on activities	2	6.4	5

41. Being able to step back and give my care recipient freedom to be independent	2	6.6	5
42. Negotiating access to needed medications	2	4.8	5
43. My care recipient's limitations are always on my mind	2	5.8	5
44. Understanding when my help isn't wanted or needed	2	5.6	5
45. Having to handle all of the household chores on my own	2	6.0	5
46. Dealing with my care recipient's denial of the effects of the disease	2	5.2	5
47. Interactions with doctors who are not knowledgeable about scleroderma	2	4.6	5
48. Rushed, inconsiderate, insensitive behavior from doctors	2	5.0	5
49. Advocating for my care recipient's needs	2	5.6	5
50. Dealing with my care recipient's anger about having scleroderma	2	4.2	5
51. Having to make difficult major medical decisions	2	5.0	5
52. Anger at medical professionals who don't treat my care recipient appropriately or respectfully	2	5.2	5
53. Having to play the role of the "mother" or authority figure to my adult care recipient	2	6.0	5

54. Frustration about knowledge about scleroderma and its treatment	2	6.2	5
55. Lack of understanding by doctors on how treatments affect scleroderma patients differently than other patients	2	6.2	5
56. Being too overbearing or involved when my care recipient can do it	2	5.8	5
57. Dealing with my care recipient's depression	2	4.0	5
58. Providing needed help when my care recipient doesn't want it or resists it	2	5.4	5
59. Helping my care recipient feel useful despite real physical limitations	2	5.8	5
60. My care recipient's suicidality	2	3.0	5
61. Frustration negotiating with government agencies to get needed support	2	5.2	5
62. Dealing with medical, insurance, and disability related paperwork	2	5.2	5
63. Having to take days off from work (i.e., appointments, taking care of children)	3	4.3	6
64. Managing last minute changes in plans (i.e., unpredictability of the disease)	3	3.8	6
65. Providing emotional support on challenging/bad days (i.e., due to increased disability, emotional need)	3	8.0	6

66. Understanding the emotional needs of the patient (i.e., what is going on with the patient's emotions)	3	9.3	6
67. Getting open and honest responses from patients about their emotions/feelings	3	6.7	6
68. Finding the balance between interfering and providing care	3	7.7	6
69. Social implications/limitations (i.e., having to leave early)	3	5.7	6
70. Transitioning from the role of primary caregiver, to someone with a less prominent role	3	3.5	6
71. Frustrations with offering suggestions and them not being considered	3	6.3	6
72. Financial implications/stressors (i.e., budget changes, job changes, days off)	3	6.2	6
73. Accommodating trips and excursions (i.e., having wheelchair access, limits to what one can do)	3	6.0	6
74. Reminding and making sure my care recipient is wearing adequate clothing for the weather	3	5.0	6

75. Navigating healthcare issues while travelling (i.e., making sure you always have access to medication, coverage, ability to communicate about the disease)	3	7.0	6
76. Learning new skills/abilities to compensate for a loss of these roles/responsibilities originally assumed by my care recipient (i.e., cooking, cleaning)	3	4.3	6
77. If the care recipient does not let the caregiver help them, or being unsure of how much help the care recipient's needs	3	5.8	6
78. Trying to find the balance between providing my care recipient with support and letting them do things on their own	3	5.7	6
79. Engaging in winter activities (i.e., not possible because of the cold)	3	5.8	6
80. Having to do all of the winter activities alone (i.e., playing with kids, walking the dogs)	3	4.2	6
81. Dealing with emotional struggles of my care recipient when they cannot take part in activities	3	6.5	6
82. Trying to find useful daily devices (in the kitchen, etc.)	3	4.0	6

83. Financial guilt (i.e., one person having to work and cover more bills, communicating about this)	3	5.5	6
84. Fatigue	3	6.7	6
85. Eating in restaurants (i.e., accommodating diet restrictions)	3	4.2	6
86. Managing your own emotions as a caregiver (i.e., mental health challenges, sickness, stress)	3	8.2	6
87. Learning information about the disease (i.e., initial flood of information following diagnosis)	3	7.3	6
88. Dealing with further medical complications (i.e., lung transplants)	3	5.7	6
89. Dealing with the unpredictability of the disease (i.e., uncertainty about progression)	3	7.7	6

Appendix G. List of All Original Caregiver Generated Support Services and Item Means

Item	Focus Group Number	Mean Rating of Challenge Importance (1-10)	Number of Participants That Rated the Item
1. Caregiver-led face-to-face support group	1	6.0	2
2. Professionally-led face-to-face support group	1	7.5	2
3. Caregiver-led phone-based support group	1	4.0	2
4. Professionally led phone-based support group	1	4.0	2
5. Caregiver-led internet-based support groups	1	5.5	2
6. Professionally led internet-based support groups	1	5.5	2
7. Caregiver internet-based chat groups, forums, or social network site	1	9.0	2
8. Internet-based chat group or forum with knowledgeable healthcare provider	1	10.0	2
9. Caregiver-led breakout groups at patient conferences	1	9.5	2
10. Professionally led breakout groups at patient conferences	1	9.0	2
11. Internet resource site designed for caregivers	1	10.0	2

12. Internet-based psychological and emotional support tools (e.g., guidance on cognitive tools, use of journaling)	1	10.0	2
13. One-to-one peer support (e.g., ability to call somebody on the phone)	1	9.5	2
14. Professionally led in-person support groups for caregivers	2	8.0	5
15. Caregiver-led in-person support groups for caregivers	2	7.8	5
16. Professionally led telephone-based support groups for caregivers	2	6.8	5
17. Caregiver-led telephone-based support groups for caregivers	2	7.0	5
18. Professionally led internet-based support groups for caregivers	2	8.8	5
19. Caregiver-led internet-based support groups for caregivers	2	9.3	5
20. Controlled chat room or forum for caregivers	2	9.0	5
21. Caregiver newsletter	2	9.5	5
22. Retreat for caregivers	2	7.0	5
23. Online workshop for caregivers to help understand scleroderma and its impact on families	2	8.3	5
24. Information on resources to help with caregiving burden	2	8.8	5
25. Tools or resources on managing interactions with healthcare providers	2	9.3	5

26. Tools to raise awareness about scleroderma	2	9.8	5
27. Specific sessions or workshops focused on caregiver needs (i.e., at conferences)	3	8.5	6
28. Financial support for families without insurance (i.e., from pharmaceutical companies, from special funds specific for patient support)	3	5.0	6
29. Information package/pamphlet about scleroderma for newly diagnosed patients (covering all aspects of the disease, not just the long term outcomes/shocking content)	3	7.3	6
30. Information about scleroderma on an online reputable website for newly diagnosed patients (covering all aspects of the disease, not just the long term outcomes/shocking content)	3	7.8	6
31. Loosely structured local in person peer support group meetings (i.e., with some structured activities)	3	7.3	6
32. List of names of other scleroderma caregivers in your area (i.e., contact them for support, meetings)	3	5.7	6

33. Increased access to and funding for medical support devices (i.e., wheelchairs, oxygen)	3	6.8	6
34. Online informational resources explaining the different physiological symptoms that might occur	3	8.3	6
35. Online support groups (via teleconference, skype) for caregivers	3	7.0	6
36. Online support groups (via teleconference, skype) for patients and caregivers	3	5.7	6
37. Physicians providing online medical advice and consults	3	6.0	6
38. Networking resource with other patients, caregivers, physicians	3	6.8	6
39. Information on how to accommodate people with scleroderma when they attend scleroderma related events (i.e., on hotel websites, in conference program)	3	4.2	6
40. Educating physicians/doctors/medical community about scleroderma (i.e., how to speak with and educate medical community about scleroderma specific issues)	3	6.8	6
41. Helping professionals know more about scleroderma and the patient's specific needs related to scleroderma (i.e, via business card)	3	7.5	6