Distribution Agreement

In presenting this thesis or dissertation as a partial fulfillment of the requirements for an advanced degree from Emory University, I hereby grant to Emory University and its agents the non-exclusive license to archive, make accessible, and display my thesis or dissertation in whole or in part in all forms of media, now or hereafter known, including display on the world-wide web. I understand that I may select some access restrictions as part of the online submission of this thesis or dissertation. I retain all ownership rights to the copyright of the thesis or dissertation. I also retain the right to use in future works (such as articles or books) all or part of this thesis or dissertation.

Signature:

_____ _____

Mackenzie P. Taylor Date

Qualitative Perspectives on Stress and Coping from Parents of Children with Marshall-Smith Syndrome

By

Mackenzie P. Taylor
Master of Public Health

Behavioral, Social, and Health Education Sciences

_____

Robin McGee, PhD, MPH
Committee Chair

_____

Yue Guan, MB, PhD, ScM
Committee Member

_____

Briana Woods-Jaeger, PhD
Committee Member

_____

Colleen McBride, PhD
Department Chair

Qualitative Perspectives on Stress and Coping from Parents of Children with Marshall-Smith Syndrome

By

Mackenzie P. Taylor

Bachelor of Arts in Public Health
Taylor University
2018

Thesis Committee Chair: Robin McGee, PhD, MPH

An abstract of
A thesis submitted to the Faculty of the
Rollins School of Public Health of Emory University
in partial fulfillment of the requirements for the degree of
Master of Public Health
in Behavioral, Social, and Health Education Sciences
2020

Abstract

Qualitative Perspectives on Stress and Coping from Parents of Children with Marshall-Smith Syndrome

By Mackenzie P. Taylor

Up to 30 million people living in the United States are affected by a rare disease. Though the specific diseases are rare, the broader impacts of rare disease affect up to 10 percent of the United States population. Marshall-Smith Syndrome (MSS) is one rare disease that is infrequently described in the literature. The people with MSS require lifelong care which often makes their parents long-term caregivers. Previous research has demonstrated some of the challenges of caregiving for children with medically complex and rare diseases, but no research has specifically focused on stress and coping for parents of children with MSS. The purpose of this study was to understand more about the experiences of parents of children with MSS, the significance of being a part of a rare disease community, and major stressors and coping strategies. Guided by the Transactional Model of Stress and Coping (TMSC), qualitative data were collected from nine parents of children with MSS (from eight families) through individual semi-structured interviews in August and September of 2019. The parents were recruited from a Facebook support group for MSS families and the interviews focused on the usefulness of online social support for this population. Inductive and deductive codes were used for thematic analysis of the data and three key themes emerged: 1) Fear of the unknown, 2) Social connection, and 3) Information gathering. These themes illuminated experiences of stress and coping for parents of children with MSS and the ways that online social support impacts the rare disease community. The findings fit within the previous literature on rare diseases and inform the importance of disease-specific social support and care. Understand more about the experiences of disease-specific communities can inform practice of genetic counseling and online interventions for families with rare disease.

Qualitative Perspectives on Stress and Coping from Parents of Children with Marshall-Smith Syndrome

By

Mackenzie P. Taylor

Bachelor of Arts in Public Health
Taylor University
2018

Thesis Committee Chair: Robin McGee, PhD, MPH

A thesis submitted to the Faculty of the
Rollins School of Public Health of Emory University
in partial fulfillment of the requirements for the degree of
Master of Public Health
in Behavioral, Social, and Health Education Sciences
2020

Acknowledgements

A number of people supported me throughout the course of this study. I would like to start by thanking Dr. Robin McGee and my other committee members for all of the mentorship and support they have provided throughout this project. I greatly appreciate the conversations and feedback to help keep my project on track. All of the training and guidance provided by this committee prepared me to do my best work to honor the stories of the Marshall-Smith Syndrome families.

Next, I would like to thank the Marshall-Smith Syndrome community. I am continuously inspired by the joy and excitement that shines so brightly from each individual with Marshall-Smith Syndrome. I am incredibly thankful that the Marshall-Smith Syndrome families are so eager to share their experiences and support each other. I am honored that I had this opportunity to hear firsthand about their daily lives and emotional experiences. The parents of children with Marshall-Smith Syndrome, including my own parents, have all overcome tremendous obstacles and deserve recognition of their strength and resilience. I am proud to be a member of this special community.

Finally, I would like to acknowledge the ongoing love and support from my friends and family. I am so thankful for the friendship, advice, and encouragement from Rebekah, Isabelle, Amy, and Cristina. Thanks to my Mom and Dad for supporting my ambitions and teaching me what it means to advocate for vulnerable individuals. And thanks to my siblings: Zack, Savannah, and especially Jonah for always challenging me and shaping me into who I am today.

Table of Contents

Chapter 1: Introduction

Background

Up to 30 million people living in the United States are affected by one of the 7,000 known rare diseases (National Institutes of Health, 2017). Each distinct rare disease has unique challenges, but there are also known common experiences faced by many individuals living with rare diseases. In addition to the physical burden of living with disease, this population is likely to face complicated social stressors and difficulties within healthcare systems (Von der Lippe, Diesen, & Feragen, 2017). The known challenges faced by rare disease populations require attention, but many barriers limit effective research and support for people with rare diseases. Rare disease populations are often neglected due to the low count of individuals in each disease-specific community (Ladd, 2015). Although the specific diseases are rare, the broader impacts of rare diseases affect up to 10 percent of the United States population and their families. Research and the development of drugs for rare disease populations have been increasing public health priorities in the United States (Goldsmith, 2016). Still, more work is needed to increase awareness, develop treatments, and provide resources for rare disease communities.

Marshall-Smith Syndrome (MSS) is one rare genetic disease that is infrequently described in the literature. This syndrome is characterized by accelerated bone formation, physical and cognitive delays, nutritional challenges, respiratory concerns, and distinct facial features such as high forehead, shallow orbits, retracted chin, depressed nose bridge, and bluish eye-whites (Shaw et al., 2010). Fewer than 100 cases have been documented worldwide since MSS was first described in 1971 (MSS Research Foundation, 2018). Very little demographic information about MSS is available because it so infrequently detected in the general population, but it appears to impact males and females equally in all parts of the world (MSS Research Foundation, 2018).

Mortality due to respiratory complications and infections has been common in early childhood, but advances in respiratory care increasingly makes it possible for individuals with MSS to reach adulthood (Shaw et al., 2010). As medical treatments improve, the growing number of adult cases will present new opportunities and challenges for people with MSS and their families. More research is needed to increase the body of knowledge and continue caring for this aging population. Individuals living with MSS require supportive environments into adulthood, meaning that MSS families and caregivers are important members of this community (MSS Research Foundation, 2016). Individuals with MSS and their caregivers face unique challenges and more information is needed to improve supportive resources.

Very little information documents the stressors of parent caregivers to children with MSS. Among other groups of parent caregivers to children with chronic conditions, a significant amount of work has demonstrated unique stressors and emotional burdens (Cousino & Hazen, 2013). Parents of children with rare disease face additional stress due to the lack of medical information, indefinite disease identity, uncertain prognosis, and underdeveloped clinical treatment (Picci et al., 2015). In a review of the literature assessing the supportive care needs of parents with a child with a rare disease, this population most commonly reported social needs, information needs, and emotional needs (Pelentsov, Laws, & Esterman, 2015). In an attempt to meet these needs, many rare disease communities have turned to online support groups (Haik et al., 2019; Kauw et al., 2015; Meade, Buchanan, & Coulson, 2018; Rocha et al., 2018). A thematic analysis of one of these groups reported that the majority of the communication present falls into three categories: (1) information sharing; (2) emotional support, expression, and experience sharing; and (3) community building (Haik et al., 2019). These findings suggest that online support groups are useful in meeting the supportive care needs of parents caring for children with rare diseases.

Due to the scarcity of information regarding rare diseases available to healthcare providers, parents of a child with rare disease often take on the role of "expert" (Baumbusch, Mayer, & Sloan-Yip, 2019; Pelentsov et al., 2015). Online communities give these parents the platform to ask questions and share information with each other (Haik et al., 2019; Rocha et al., 2018). Despite the emerging literature surrounding online support groups for families with rare diseases, the qualitative data capturing the parents' perspectives on these types of groups are lacking. More investigation is needed to gather the qualitative perspectives of parents of children with MSS, especially regarding online communities. Learning from these parents' perspectives will help to clarify the experience of MSS and the perceived value of online support groups for parents. This information can be used to inform future studies and interventions that serve parent caregivers.

Problem

There is insufficient information documented in the literature about the experience of parenting a child with MSS and the usefulness of online social support for this population. The well-being of these parents should be a priority due to the known stressors related to caring for and parenting children with rare disease. More research is needed to better understand and support this population.

Purpose

The purpose of this study was to gather perspectives from parents who care for children with MSS. The study specifically focused on the perceptions of stress and coping, the experience of being a part of a rare disease community, and the ways that parents utilize social support. Because the literature highlights the importance of online social support for individuals with rare diseases, this study gathered perspectives on usefulness of online social support for MSS

families. Thematic findings may inform interventions and future directions to support MSS families.

Research Questions

The current study explores the following questions:

1. What is the experience of parents of children with MSS?
2. What do parents of children with MSS identify as major stressors and coping strategies?
3. What is the perceived significance of being a part of a rare disease community?
4. How have online social networks provided social support for these families?

Rationale

Due to a paucity of research on MSS, the current study fills multiple gaps in the literature related to unique experiences of families with MSS. First, this study investigates questions focusing on the parents who care for children with MSS. Second, this study provides qualitative data documenting the experience of MSS families. Third, this study evaluates the usefulness of social support for parents of children with MSS, especially as it relates to online communities. Working to fill these gaps will amplify the voices of parents of children with MSS and inform future directions for supporting this population.

Theoretical Framework

The current study was informed by Lazarus and Folkman's Transactional Model of Stress and Coping (TMSC) (Lazarus & Folkman, 1984). This model focuses on the person-environment relationship that guides the appraisal of life transitions (Devonport, 2011). Different people perceive a stressful event differently, and it is these perceptions, rather than the objective stressors that determine the effects on behaviors and health outcomes (Glanz, Rimer, &

Viswanath, 2008). Based upon the appraisal of a stressful event as a perceived harm, threat, or a challenge to well-being, an individual engages in coping strategies (Devonport, 2011). The TMSC provides a useful framework to inform the exploration of the experiences of parents of children with MSS.

Key constructs of the TMSC such as primary appraisals, secondary appraisals, coping efforts, social support seeking, and information seeking help to make meaning of the experience of stress for this population. Parents of children with MSS face unique and complicated stressors that come along with caring for a rare and medically complex child. Social support seeking was identified as a key coping effort within the study population based on the literature assessing online social support group for rare disease communities (Haik et al., 2019; Kauw et al., 2015; Meade, Buchanan, & Coulson, 2018; Rocha et al., 2018). Information seeking is another key coping effort for parents of children with rare diseases due to the limited information available within these populations (Picci et al., 2015; Pelentsov et al., 2015; Pelentsov, Fielder, Laws, & Esterman, 2016; Von der Lippe, Diesen, & Feragen, 2017; Baumbusch et al., 2019). These constructs provide useful parameters for the exploration of stress and coping for parents who care for children with MSS.

Chapter 2: Literature Review

Significance of Rare Disease

The National Institutes of Health (NIH) defines a rare disease as a condition affecting fewer than 200,000 people in the United States (approximately 1 in 16,00). Other countries have their own definitions of rare disease, for example: the European Union defines a rare disease as a condition that affects less than 1 in 20,000. It is reported that there are as many as 7,000 rare diseases, the majority of which are thought to have genetic causes. Though the individual diseases are rare, it is estimated that 25-30 million Americans are affected by rare disease. Despite increasing research efforts and prioritization of this population, there is still more information needed to understand, care for, and treat rare conditions (National Institutes of Health, 2017).

It is important to understand the diverse experiences of people living with rare disease in order to care for and treat these individuals. Von der Lippe, Diesen, and Feragen (2017) conducted a systematic review including 21 qualitative studies of adults living with various rare diseases. The purpose of this systematic review was to synthesize information about the shared experiences across rare diagnoses without focusing on any disease-specific challenges. The authors reported on three main themes: 1) Consequences of living with a rare disorder, 2) Social aspects of living with a rare disorder, and 3) Experiences with the health care system. The consequences of living with a rare disorder theme included information on constraints and physical limitations, physiological impacts, and coping strategies. The social aspects of living with a rare disorder theme involved discussion on telling others about the diagnosis, stigma, the experience of being different, and the importance of social support. The experiences with the healthcare system theme elaborated on a lack of knowledge in the health care system, interaction

with health providers, and patients as experts. Von der Lippe, Diesen, and Feragen concluded that the challenges faced by individuals with rare diseases go far beyond their medical conditions. They suggested that these challenges could be diminished by increasing knowledge and awareness about rare diseases, and prioritizing psychological health and coping strategies for individuals with rare disease (Von der Lippe, Diesen, & Feragen, 2017).

Individuals with rare diseases should be prioritized due to the complicated challenges related to rare disease. However, there are many barriers to supporting this vulnerable population. Specific rare diseases are often neglected due to the low number of patients affected by any one rare disease (Ladd, 2015). Moreover, small population sizes, disease heterogeneity, and geographic dispersion make it difficult to conduct robust research on specific rare diseases (Whicher, Philbin, & Aronson, 2018). To overcome these challenges, the United States passed the Rare Disease Act of 2002 (Public Law 107-280). The purposes of this law are to: 1) to establish the Office of Rare Disease at the NIH and 2) increase the national investment in the development of diagnostics and treatments for patients with rare diseases and disorders (Rare Disease Act, 2002). This policy reflects an increasing urgency to research and address the needs of people affected by rare disease. Expanding the understanding of specific rare diseases will promote favorable health outcomes for individuals with rare diseases and their families.

History of Marshall-Smith Syndrome

One specific rare condition, known as Marshall-Smith Syndrome (MSS), was first described in 1971 with a case report of two unrelated male infants with relative failure to thrive and skeletal abnormalities (Marshall, Graham, Scott, & Smith, 1971). At the time, these cases represented a newly recognized syndrome marked by an unprecedented acceleration of bone maturation. It was reported that both of these children had unusual physical features and

presumed developmental and cognitive delays. Not enough information was collected about these two children to understand the etiology of their syndrome because they both died infancy. The original authors suspected that this condition was difficult to recognize and observe due to death in infancy caused by breathing difficulties and respiratory complications (Marshall et al., 1971).

In 1980, Fitch reviewed the two original cases described by Marshall and colleagues with an additional nine cases. Fitch highlighted that the key characteristics seen in these 11 cases reaffirmed this syndrome as a distinct entity. This review found no sign of parental consanguinity or family reoccurrences. The 11 cases reviewed all included accelerated skeletal maturation and abnormal phalanges in the hands. Distinct facial features were noted including a prominent skull, small facial bones, protrusion of the eyes, blue sclera, flat nose bridge, upturned nostrils, and an undersized jaw. Poor weight gain, short stature, and psychomotor delays were observed in many of the MSS cases. Pneumonia and severe respiratory problems were noted in nearly every case often leading to early death (Fitch, 1980).

Additional case studies continued to highlight similar features with a focus on respiratory issues. Johnson et al. (1983) summarized the observed respiratory abnormalities of MSS in three categories: congenital defect, functional irregularities, and acquired complications. This report noted attempts to open the airway by tracheostomies and other physical facial modification but these procedures had been unsuccessful at prolonging life (Johnson, Carey, Glassy, Paglieroni, & Lipson, 1983). Yoder, Wiswell, Cornish, Cunningham, and Crumbaker (1988) described yet another case of a baby that had died at 91 days old due to aspiration pneumonia and pulmonary hypertension. At the time of this report 15 cases had been documented, and nearly all of them had died in early infancy with reported causes of death including pneumonia, airway obstruction, and "sudden, unexpected death." This report highlighted the early recognition of upper airway

obstruction and intensive treatment of these complications as priorities to prolong life for children with MSS (Yoder, Wiswell, Cornish, Cunningham, & Crumbaker, 1988).

Dying in early infancy was a major problem for MSS, but by 1990s sporadic case studies began emerging with documentation of children surviving into early childhood (Summers, Cooper, & Butler, 1997). Sperli et al. (1993) demonstrated more clinical variability with the report of a child with MSS at 5 years of age with no respiratory problems. This case study suggested the prospect of increased life expectancy for individuals with MSS free of respiratory complications (Sperli, Concolino, Barbato, Strisciuglio, & Andria, 1993). Hoyme et al. (1993) documented a 7-year-old and an 8-year-old with MSS who were successfully relieved of respiratory challenges. The first was treated in infancy with a tracheostomy and a partial glossectomy, and the second was treated with just a tracheostomy (Hoyme, Byers, & Guttmacher, 1993). Williams et al. (1997) presented another case living into childhood thanks to successful treatment. By three and a half years, this child was relieved from significant upper airway obstruction by a successful tonsillectomy and adenoidectomy (Williams, Carlton, Green, Pearman, & Cole, 1997). These cases demonstrated the possibility of longer-term survival when respiratory problems are detected early and aggressively treated.

Researchers continued to follow cases of MSS to expand the body of knowledge about this rare disease. Shaw et al. (2010) reviewed and summarized 39 cases of MSS described in the previous literature, followed up and reported on four previously documented cases, and described 15 new individuals with MSS, the oldest being 30 years old. Colleagues around the world and the Marshall-Smith Research Foundation referred all of the patients to this study. Each of the patients included were clinically examined by one of the authors and additional data were collected through an online wiki. The results highlighted the most common features including varying levels of developmental delays, severe respiratory problems, distinct facial features, abnormal

bone maturation, and failure to thrive. Additional common features included blue sclera, abnormal hair growth, and kyphoscoliosis later in childhood and into adolescence. These findings help to confirm and expand the phenotype and natural history of MSS. Additionally, the authors emphasize the importance of the online wiki to help collate information for an extremely rare condition such as MSS. The collection of this dataset from 11 different countries and various languages would not have been possible without this online tool. The authors recommend this innovative method to be used for future studies of extremely rare disease (Shaw et al., 2010). Advancements in technology improved the treatment and research of MSS, but there were still many unanswered questions about this unique population.

Genetics information on Marshall-Smith Syndrome

Though many rare diseases are thought to have genetic causes, the molecular etiology of MSS was unknown for many years. Mouse models have been used as an important tool to help detect candidate genes for human diseases, including MSS. Malan et al. (2010) first reported the *de novo* mutation in the NFIX gene as a cause for MSS based on observations from a mouse model. The NFIX deficient mice presented similar features to MSS, making NFIX a hopeful gene candidate for this disease. Malan et al. screened nine MSS patients for NFIX mutations and found seven had frameshift mutations and two had donor-splice mutations. The mutations identified among the nine patients provided evidence that supported NFIX as a cause of MSS (Malan et al., 2010). Illuminating the genetic etiology of this syndrome has helped to accurately detect and diagnosis children with MSS. The more that doctors and researchers understand about the etiology, the more they can support patients and their families.

Development and Behavior of Marshall-Smith Syndrome

The cognitive and developmental delays associated with MSS impact the behavior of the affected individuals. Shaw et al. (2010) briefly described the development and behavior of people with MSS. The authors noted moderate to severe cognitive delay along with severe motor delays. Additionally, speech milestones were significantly delayed, and some individuals remained non-verbal, which could be related to laryngeal and facial abnormalities. Shaw et al. reported a happy demeanor among individuals with MSS and a notable enjoyment of social activities with family or friends. A fixation on a favorite toy and repetitive, stereotypical play was described in this study sample (Shaw et al., 2010).

To further explore the behavioral findings of Shaw et al., Van Balkom et al. (2011) conducted an exploratory study focusing on autism symptomology and the behavioral features of MSS. This research team was able to assess six of the 19 individuals that participated in the previous study. The same child psychiatrist and neuropsychologist assessed all six of the study participants. Test instruments included: 1) the assessment of intellectual capabilities, 2) a child psychiatric examination and Autism Diagnostic Interview, 3) an adaptive functioning assessment, and 4) an assessment of behavioral characteristics. The key findings of this study demonstrated the moderate to severe delays in mental age, motor development, and adaptive function in MSS. The participants showed similarities in communication, social integrations, and behavior. Speech delays were present, yet participants generally enjoyed social interaction with a happy or friendly demeanor. Some behavioral characteristics resembled features of Autism Spectrum Disorder including repetitive behavior, limited imaginative play, and the fascination with a favorite toy, but the developmental progress over time is still unknown (Van Balkom et al., 2011). Although more investigation is needed to understand development over time, the reported developmental and behavioral delays have practical implications for the children with MSS and their families.

Information of Care for Individuals with Marshall-Smith Syndrome

Due to the complex features of MSS, affected individuals require specialized medical oversight and care. Families and healthcare providers need more information on caring for people with rare diseases. The paucity of information available about MSS makes it challenging for doctors to make decisions about treatment for individuals with MSS. In 2016, the Marshall-Smith Research Foundation (www.marshallsmith.org) published *Standards of care for Marshall-Smith Syndrome*. This resource provided critical information for healthcare providers on what to expect from a patient with MSS and how to manage care and treatment plans. The standard of care consisted of five key principles:

1. To report on all knowledge and information on MSS. This care standard describes the syndrome and highlights the problems arising in both acute and chronic settings.
2. To reinforce the interdependence between the patients with MSS, the symptoms and the treatment possibilities and to have a holistic view of the consequences of the syndrome and management.
3. To describe the minimum standards of good care for MSS patients, from the patient's perspective.
4. To support self-management with an active role for the parents.
5. To initiate comprehensive coordinated multidisciplinary care. (p. 5)

Alongside this document, the Marshall-Smith Research Foundation published a patient version to deliver accurate and accessible information to families caring for a person with MSS. This guide provided information on symptoms, diagnosis, prognosis, causes, and daily life impacts of living with MSS. Additionally, the patient information guide offered tips and resource guides to help families and caregivers design a care plan and manage the treatment of an individual with MSS. These documents highlighted the importance of active caregivers for people with MSS who are

unable to care for themselves (MSS Research Foundation, 2016). As children with MSS are living longer and healthier lives into adulthood, more responsibility is falling on parents to care for individuals with MSS later in life.

Parent Caregivers of Children with Chronic Disease

The physical and cognitive features of MSS create challenges for parents caring for children with MSS. While limited research focuses specifically on MSS parents, valuable lessons can be learned from other groups of parent caregivers. Parents of children with ongoing illnesses are often responsible for managing appointments and treatment plans, in addition to responsibilities of in-home caring. Cousino and Hazen (2013) conducted a systematic review of the stress of parenting children with chronic illness, including a meta-analysis of 13 studies and a qualitative analysis of 96 studies. The meta-analysis compared parenting stress among parents of children with chronic illness to parenting stress among parents of healthy children and found that parents of children with chronic illness reported greater general stress. The qualitative analysis compared parenting stress among different illness populations and found some disease specific stressors, but overall these parents were all burdened by the responsibility for treatment management (Cousino & Hazen, 2013).

Exceptionally complicated chronic diseases, such as MSS, require additional attention and care. Cohen et al. (2011) highlighted children with medical complexity (CMC) as an emerging priority population for research and intervention. This population is a sub-group of children with special health care needs that have the most intensive healthcare needs. Cohen et al. listed examples of CMC including “children who have a congenital or acquired multisystem disease, a severe neurologic condition with marked functional impairment, or patients with cancer/cancer survivors with ongoing disability in multiple areas” (p. 529). This report identified CMC as a

growing priority due the increasing long-term survival of prematurely born infants, infants with congenital anomalies, children with chronic conditions, and survivors of acute illnesses that required intensive care. Cohen et al. noted that the long-term survival of CMC has a growing impact on families and parental caregivers including increased stress, adverse health outcomes, marital discord, and financial hardship. The authors called for an increased family involvement in research and prioritization of family outcomes for CMC (Cohen et al., 2011).

Woodgate, Edwards, Ripat, Borton, and Rempel (2015) conducted a qualitative study of 68 parents from 40 distinct families to better understand the experience of parenting a CMC. The study found that parents of CMC were often assuming a variety of roles for their child in different settings including but not limited to the healthcare provider, the case manager, the student, the teacher, the detective, the guard, and the advocate. Woodgate et al. found that these complicated responsibilities required of parents to meet the needs of their CMC take a toll on the parents' physical and mental health. Parents of CMC are in need of adequate resources and supports to help them manage their intensive role of parenting (Woodgate, Edwards, Ripat, Borton, & Rempel, 2015).

Parents that care for children with ongoing and complex healthcare needs require special attention and support in order to cope with the demands of their daily lives. As more and more children with medical complexity are living into adulthood, parents are experiencing the ongoing stressors of caregiving into their later years. More research is needed to prioritize health outcomes for this population of parents.

Caring for a Child with a Rare Disease

In addition to the demands of managing complicated healthcare needs, parents who care for children with rare diseases face unique burdens. Picci et al. (2015) conducted a study to

compare the emotional burden and coping strategies of parents of children with rare disorders (CRD) and parents of children with chronic disease (CCD). This study found parents of CRD to be more anxious, confused and bewildered, and less active and rigorous than parents of CCD. However, both groups reported being slightly satisfied or satisfied with life one year after the diagnosis of their child. Regarding coping, Picci et al. found that parents of CRD less likely to engage in problem focused coping strategies, such as active coping or seeking social support, compared parents of CCD. Instead, parents of CRD more often rely more alternative coping strategies such as turning to religion for comfort. The study suggested that this difference in coping styles could be attributed to negative perceptions of children's rare disease, such as the perception of their disease as more severe. Picci et al. highlighted the importance of training health providers and providing more information to caregivers of CRD with hopes that this might improve the perception of the illness and lead to the use of more problem-focused coping strategies (Picci et al., 2015).

Pelentsov, Laws, and Esterman (2015) conducted a scoping review of the literature to identify the supportive care needs of parents of a child with a rare disease. After the systematic review of 29 studies, the authors found that many parents of children with various rare diseases face similar challenges. The most common supportive care needs for these parents reported in the literature were social needs, informational needs, and emotional needs. The authors noted the striking lack of literature documenting the burden of caregiving for parents of a child with rare disease (Pelentsov et al., 2015). To follow up on this scoping review, Pelentsov, Fieler, Laws, and Esterman (2016) conducted a survey to assess the supportive care needs of parents of a child with rare disease. The survey was distributed online to 301 parents of children with 132 distinct rare diseases. The results of this survey found that, regardless of the specific disease, parents of children with rare disease have common unmet needs. Parents indicated common challenges

including lack of knowledge from healthcare providers, social isolations and loneliness, lack of confidence in caring for complex health needs, symptoms of depression and anxiety, and financial burden. They concluded that healthcare providers should be aware of the frustrations and emotions associated with the challenges of caregiving for a child with a rare disease in order to best support these parents (Pelentsov, Fielder, Laws, & Esterman, 2016).

Baumbusch, Mayer, and Sloa-Yip (2019) conducted a qualitative study to investigate parents' experiences of navigating healthcare systems for their child with a rare disease. Sixteen parents were recruited to participate in semi-structured, in-depth interviews that were coded and analyzed for emerging themes. Important findings were documented about a parent's process of seeking and receiving a diagnosis, and subsequently seeking and accessing medical services for their child with a rare disease. Receiving a diagnosis was a difficult and emotional process for many of these parents. Even once they received a diagnosis for their child, parents were often left without much information about their child's prognosis. Healthcare providers were unable to satisfy the informational needs of parents due to the lack of knowledge about newly recognized and under-researched rare diseases. The authors highlighted peer support as a key resource identified by the participants. Many participants mentioned online peer support groups that connected families with rare diseases and allowed them to exchange information and support (Baumbusch et al., 2019). These types of support groups allow families of children with rare diseases to share valuable experiential knowledge. Learning from the lived experiences of other parents help families with rare diseases navigate healthcare systems and caregiving.

Family-Engaged Research and Care

Families caring for children with MSS have practical knowledge and valuable experiences with this disease. The lived experiences of parents should be considered a valuable asset for

doctors and researchers. Connecting researchers with parents can help inform relevant topics for future research and treatment. Cohen et al. (2011) recommended that researchers increase the involvement of families in the research of children with medical complexity in order to prioritize family health outcomes. Understanding the needs of parent caregivers is an essential part of preventing adverse health outcomes for this population. Additionally, Baumbusch and colleagues emphasized the importance of family-engaged healthcare for rare disease communities. When little is known about a disease, providers should view parents as "experts" of their experiences (Baumbusch, Mayer, and Sloa-Yip, 2019). Engaging parents in this way will increase collaborative knowledge and support for parents of children with rare disease. Doctors and researchers alike should take advantage of the valuable information available through the lived experiences of parent caregivers. Understanding the daily experiences of these families will enhance current knowledge about the disease and inform future directions for supporting families.

Online Social Support

Social support has been identified as an important coping mechanism for parents who care for children with rare disease. However, these parents may have a difficult time finding other people who understand the unique circumstances related to rare diseases. Online support may be a useful tool for overcoming isolation due to geographic dispersion and rare circumstances. A growing body of research is documenting the usefulness of online social support groups for rare disease communities (Haik et al., 2019; Kauw et al., 2015; Meade, Buchanan, & Coulson, 2018; Rocha et al., 2018). These papers focused on peer-to-peer support for patients in general, rather than just focusing on the parent needs.

Kauw et al. (2015) studied the purposes of online patient-to-patient forum conversations among people with Addison's disease and Cushing's syndrome, within the context of patient-centered care. They analyzed the content of 390 patient-driven questions to understand the purposes and usefulness of online forums for these rare populations. The results indicated that the majority of the questions were seeking knowledge and information about the disease (80.8%), and many of the other questions were used to call for emotional support (9.7%). Kauw et al. discussed the crucial importance of experience-based knowledge for patients with rare disease and the ways that health providers should incorporate this information to improve patient-centered care.

Meade, Buchanan, and Coulson (2017) conducted a thematic analysis of message posts in a support group for patients affected by rare neuromuscular disorders. The authors inductively analyzed 1,951 message posts from the first five months of newly formed neuromuscular disorder patient support group. They found this support group to be an important social community for patients to share information, raise awareness about their disorders, and connect with peers over similar experiences.

Rocha et al. (2018) administered a web-based survey to explore preferences for social media use and privacy among patients and families with rare genetic disorders. Of the 103 survey responses, 99% of people reported using Facebook, making it the most popular social media resource among the sample. Seeking information about test results and diagnoses were the most popular use of online social media (83%), followed by reading posts from rare disease organizations (73%), joining conversations about diagnoses (67%), and connecting with others to find support (58%). In regard to privacy, only 12% of particiapnts reported feeling uncomfortable with information sharing within an online family support group compared to the 60% who felt

uncomfortable sharing perosnal information within a public facebook group. This suggests that secure online environments for families may be an import feature of useful online social support.

Haik et al. (2019) conducted a thematic content analysis of an online community for a rare disease known as idiopathic subglottic stenosis. The majority of the content analyzed fit into three thematic catagories: 1) information sharing, 2) emotional support and experience sharing, and 3) community building. Additionally, the authors noted a high level of trust among group members and that the content within this community was overwhelming positive in tone. These findings add support to the exisiting literature demonstrating the usefullness of online social support for rare disease communities.

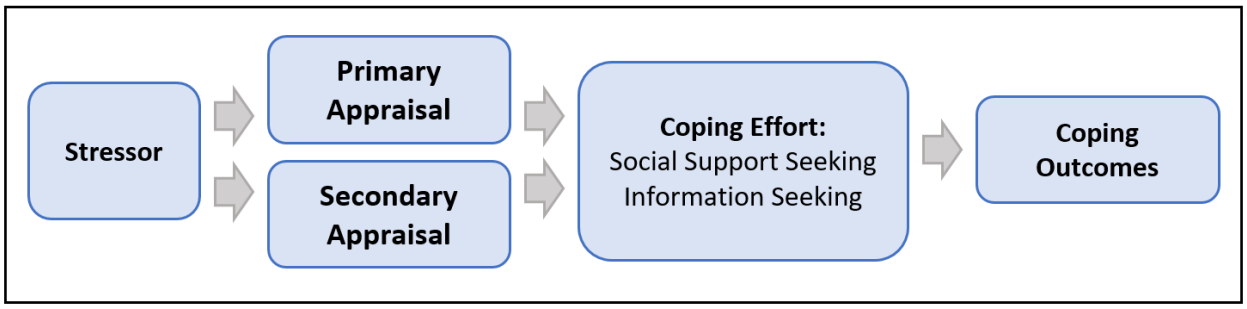
More research is needed to demonstrate and understand the usefulness of online peer support groups specifically for parent caregivers within rare disease communities. Baumbusch, Mayer, and Sloa-Yip (2019) suggested that peer support should be an important resource for parents of children with rare disease. Many participants in that study discussed their experiences with online support groups, but additional exploration is needed to understand online peer support among parents.

The Transactional Model of Stress and Coping

The experience of stress for parents of MSS is related to the ways they understand and respond to their circumstances. Lazarus and Folkman's (1984) Transactional Model of Stress and Coping (TMSC) was used to inform elements of the current study. Constructs of the TMSC were used to provide a framework to analyze the experience of stress and coping for parents who care for children with MSS. This model focuses on the person-environment relationship that guides the appraisal of life transitions (Devonport, 2011). An individual engages in coping strategies based upon the appraisal of a stressful event as a perceived harm, threat, or a challenge to well-

being (Devonport, 2011). Key constructs of this theory include primary appraisal, secondary appraisal, coping efforts, social support seeking, and information seeking (Lazarus & Folkman, 1984). The relationships between constructs of the TMSC can be seen in figure 1.

Figure 1. Transactional Model of Stress and Coping[a]



[a]Adapted from Glanz et al. (2008)

Primary appraisal refers to a person's judgments about the significance of an event as stressful, positive, controllable, challenging, benign, or irrelevant (Glanz et al., 2008). This construct can be seen through a parent's perspectives and reactions when their child with MSS is born and diagnosed. According to the TMSC, as a parent learns about MSS, they use primary appraisals to evaluate their personal risk and the severity of the situation. This primary appraisal process then prompts coping efforts or adaptations to deal with the given circumstances. Previous studies on parenting children with chronic illnesses and rare diseases found that negative perceptions of a child's condition relate to increased parenting stress (Cousino and Hazen, 2013, Picci et al., 2015). Assessing a parent's primary appraisals of caring for a child with MSS helps illuminate the experience of stress in this population.

Secondary appraisal refers to a person's judgments about how they can respond to a stressful situation. This may include perceived changeability of the situation, perceived ability to manage emotional reactions, and expectations about the effectiveness of coping resources (Glanz et al., 2008). The secondary appraisal relates to the MSS parent's evaluation of how they can control and respond to the challenges of caring for their child. According to the TMSC, a

person's perceived control over a situation can have important impacts on their coping efforts and adaptations to a stressful event (Glanz et al., 2008). The previous survey of the supportive care needs of parents of children with rare diseases found that these parents often lacked confidence in their ability to care for the complicated needs of their child (Pelentsov, Fielder, Laws, & Esterman, 2016). A parent's negative appraisal of their ability to respond to stressful circumstance is likely to increase their experience of stress. Exploring a parent's secondary appraisals of caring for a child with MSS clarifies the ways that personal perspectives impact stress.

Coping efforts are prompted based on the primary and secondary appraisals a stressful situation. The TMSC outlines two dimensions of coping efforts: 1) problem management, and 2) emotional regulation (Glanz et al., 2008). Problem management refers to behaviors that aim to change the stressful situation such as active coping, problem solving, and information seeking. Emotional regulation focuses on changing perceptions or feelings about the stressful situation through efforts such as social support, venting feelings, and avoidance. According to the TMSC, problem management efforts are most useful for changeable situations and emotional regulation efforts are most useful for situations that cannot be changed (Glanz et al., 2008). Understanding the various coping efforts of parents of children with MSS helps illustrate the daily experiences of these families.

Social support seeking is one type of emotional regulation highlighted by the TMSC. Emotional regulation coping is especially useful for individual to adapt to a stressor that is unchangeable (Glanz et al., 2008). Parents of children with MSS may rely heavily on emotional regulation strategies, such as social support seeking, because they face many circumstances that cannot be changed. Social support seeking may help these parents adapt by impacting the perceived severity of the situation (primary appraisal) or the perceived ability for parents to

respond to challenging circumstances (secondary appraisal) (Glanz et al., 2008). Previous studies have demonstrated the usefulness of online support groups within rare disease communities (Haik et al., 2019; Kauw et al., 2015; Meade, Buchanan, & Coulson, 2018; Rocha et al., 2018). These reported findings align with the constructs of the TMSC and provide rationale for the investigation of online social support for the parents of children with rare diseases.

Information seeking is an important problem focused coping mechanism to overcome uncertainty of circumstances. Uncertainty often hinders a person's ability to appraise a stressful situation and respond accordingly (Lazarus and Folkman, 1984). This may be especially important for rare disease populations that are constantly living in uncertainty (Picci et al., 2015; Pelentsov et al., 2015; Pelentsov, Fielder, Laws, & Esterman, 2016; Von der Lippe, Diesen, & Feragen, 2017; Baumbusch et al., 2019). With so few cases of MSS documented in the literature, MSS parents are left without a clear notion of what to expect for their child (MSS Research Foundation, 2016). The theory and literature suggest that this uncertainty may be a key stressor for parents of children with MSS. Information seeking may be an important coping strategy for these parents.

Using the constructs of primary appraisals, secondary appraisals, coping efforts, social support seeking, and information seeking from the TMSC provides a useful framework to understand the experiences of parents that care for children with MSS. More information is needed to comprehend the ways that parents perceive and cope with the stress of parenting a rare and medically complex child. Using this framework will help to delineate the ways that parents understand MSS at the time of birth, diagnosis, and throughout their daily lives. Learning about the coping efforts of these parents, will help to elaborate on the needs of this population and inform future directions for supporting parents of children with MSS.

Summary of the Current Problem and Study Relevance

With fewer than 100 cases documented in the literature, MSS is an extremely rare disease (Shaw et al., 2010). The literature describing the burden of parenting children with medical complexity and children with rare diseases suggests that parents that care for a child with MSS experience unique stressors possibly including social isolation, emotional instability, and the lack of information. Very little is documented about the personal experiences of the parents of children with MSS. Understanding more about these expert perspectives is a critical opportunity to increase the knowledge and information about MSS. Moreover, understanding the stressors and coping styles of parents of children with MSS will help to inform interventions and resources available for families.

Chapter 3: Methods

Project Conceptualization

This project was conceptualized and completed by the primary investigator (PI) throughout her Master of Public Health program at Rollins School of Public Health (first author). Many of her ideas were inspired from personal and professional experiences with children with disabilities and their families. The PI planned the study, conducted the literature review, collected the data, analyzed the data, and wrote the chapters of this thesis. The PI reached out to study team members as needed for guidance, discussion, and review of chapters.

The PI was supported by a team of three faculty members from the behavioral science and health education department at Rollins School of Public Health. Each team member has qualitative experience and content expertise to support this project. Each member of the study team provided proof of CITI certification for the protection of human subjects.

Reflexivity

The PI has a younger brother with MSS. This had an important impact on the study from conception to the analysis. Special care has been taken throughout the entire process to cultivate reflexivity and to anticipate ways to mitigate the potential biases of the PI. Memos were used to document and reflect upon personal reactions throughout the duration of the study. Despite potential bias, this personal connection to MSS has been a valuable asset for this project. The PI's status as an insider has helped to build rapport within the community and provided a unique point of access to the doctors, researchers, and many other families with MSS.

Research Design

This study employed qualitative methods to collect in-depth semi-structured interviews in order to understand the experiences of parents of children with MSS. Individual interviews were conducted with nine parents from eight distinct families. The study was informed by the previous literature on MSS, parents of children with rare diseases and medical complexity, and online social support, as well as constructs from the Transactional Model of Stress and Coping (TMSC). Thematic analysis informed by the TMSC was used to analyze the data. Emory University's Institutional Review Board (IRB) approved the study before data collection began to ensure the protection of study participants.

Population

The target population of this study was parents of children with MSS that were members of the MSS Facebook support group. At the time of recruitment, this Facebook group had 109 members, and this number included mostly parents, with some siblings or other family members. Approximately 30-40 families from a variety of countries were represented in this group. The MSS Facebook group is private, and the administrators carefully control who is allowed to join. As a family member of someone with MSS, the PI was already connected to this MSS Facebook group.

Stakeholder Engagement

Engaging with key stakeholders was a valuable preliminary step to learn about current research priorities within the MSS community. Initial contact was made through email with the MSS Facebook group administrator and several key doctors and researchers involved in MSS. The purpose of this step was to establish a relationship with the administrator as the gatekeeper, and to gather perspectives through stakeholder engagement. The PI introduced the conceptualized

project and asked for suggestions that would help to guide the study. Feedback provided from the key stakeholders helped the PI to formulate the final version of the data collection instrument. The MSS Facebook group administrator approved for the PI to engage the group for this study and other feedback provided from the key stakeholders helped the PI to formulate the recruitment plan the final version of the data collection instrument.

Eligibility

To enroll in the study, participants had to meet the following inclusion criteria: 1) participants had to be a parent of a child with MSS, 2) participants had to be a member of the MSS Facebook group, and 3) participants had to be comfortable speaking in English for the interview. There were no specific exclusion criteria, and the given requirements were based off the participants' self-report.

Recruitment

Both passive and purposive methods of samplings were used through the MSS Facebook group in order to make contact with as many parents as possible. The PI has been a member of the MSS Facebook group since her brother was diagnosed in 2016. A Facebook post on the private group was used to introduce families to the PI and the project. Interested parents were able to submit their email addresses on a Google Form so the PI could reach out to them with more details. To reach more families, the PI used purposive sampling and active recruitment to contact and invite specific parents from the MSS Facebook group. The PI recruited a total of nine participants representing eight families. Demographic information about the participants is

reported in table 1 and demographic information about their children with MSS is reported in table 2. The variables were presented in categories to protect the privacy of this small population.

Table 1. Demographic variables[a] of participants (N=9)

Variables	n	Percentage
Sex		
Female	8	89
Male	1	11
Age		
30-39 years	5	56
40-49 years	2	22
50 years and over	2	22
Education		
Some College	2	22
Bachelor's degree	4	44
Graduate Degree	3	33
Employment status		
Full-time	5	56
Part-time	2	22
Not employed	2	22

[a]Variables are reported in categories to protect privacy

Table 2. Demographic variables[a] of children with MSS[b] (N=8)

Variables	n	Percentage
Sex		
Female	5	63
Male	3	38
Age		
0-5 years	2	25
6-12 years	3	38
13 years and over	3	38
Timing of diagnosis		
0-1 month	2	25
2-12 months	3	38
>1 year	3	38

[a]Variables are reported in categories to protect privacy
[b]Marshall-Smith Syndrome

Eligibility was assessed through the following questions: 1) Do you have a child with a diagnosis of MSS? 2) Are you a member of the MSS group on Facebook? And 3) are you comfortable communicating in English for the interview? Those who answered yes to each of these questions were eligible for the study.

Interested and eligible individuals received an electronic informed consent form with an overview of the study from the PI. Before signing and returning the form through email, each potential participant had the opportunity to discuss any questions or concerns with the PI. Once the consent form was signed and returned, the participant was officially enrolled in the study and contacted by the PI to schedule an interview.

Instrument

Data were collected using a semi-structured qualitative interview guide designed to take 30-60 minutes. The interview guide questions were based upon the previous literature and constructs from the TMSC theoretical framework. The interview guide was broken into three distinct domains: 1) background, 2) daily stress and coping, and 3) online social support. The first section contained short-answer questions to gather demographics of the parent and some general background information about the family. The second section included questions to get a sense of the parent's daily stressors and coping efforts. Finally, the third section consisted of questions to understand the ways that parents use the MSS Facebook group for coping. Some questions included probes to encourage more discussion of key concepts. The interview guide can be found in Appendix A.

Data Collection

The PI scheduled individual time slots with all enrolled participants to conduct the interviews over the phone in August and September of 2019. Each interview was expected to take 30-60 minutes. Before each interview, the PI took the time to review the purpose of the study with the participant. The PI explained the three domains of the interview guide, and the participant's right to skip any question or stop the interview at any point. The participant was given another opportunity to ask questions before moving onto the interview questions. The PI

carefully followed the guide to facilitate each interview and used probing questions as needed. Notes were taken throughout the interviews and each participant consented for the audio to be recorded. Temi web-based automated transcription service was used to create verbatim transcripts of each recorded interview (Temi, 2020). The PI carefully reviewed the quality of each transcript by listening to each interview to manually correct any errors including typos and missing words.

Throughout the interview and transcription process the PI documented ideas and reactions with memos. This was a useful way to begin engaging the data and to keep personal biases in check. Memos were also used to record important concepts as they came up within individual interviews. Reflecting on these memos helped guide the subsequent interviews. Additional probes were used to explore emerging concepts over time. For example, participants often mentioned their other children while talking about their family backgrounds. The PI began probing more about these sibling relationships to understand the family dynamics surrounding MSS. The also PI used probes based on the topics highlighted by each participant. For example, if the participant mentioned having home nursing care, the PI would ask more about this experience for the family. No new content areas were added to the interview guide.

Data Management

Each study participant was assigned a unique ID number to ensure confidentiality. A list linking the ID numbers to participants was kept on a separate, password-protected document on a secure network. These strategies were used to protect the identities of the study participants. Any information that was potentially identifiable was redacted from the transcripts. All of the interview audio recordings and transcripts were stored on a password protected and secure network. At the end of the study, the audio recordings were permanently deleted.

Data Analysis

Thematic analysis was employed to analyze the verbatim transcripts. The PI utilized MaxQDA version 2018.2 throughout the coding and analysis process. A codebook was developed that included both deductive and inductive codes. To develop and refine the initial codebook, the PI engaged with the interview content by listening to the audio recordings. Memos were used to summarize and document initial ideas. Deductive codes were based upon the research questions, interview guide, and the TMSC framework. The inductive codes were added into the codebook as they emerged from the transcript content. For example, many parents mentioned the significance of physical similarities between the children with MSS, so an inductive code was added to track this pattern. The PI used the final codebook to carefully code each transcript line-by-line. A secondary coder assisted by coding every third transcript to compare for agreement. Reflecting on the code agreement and disagreement helped to ensure the consistent application of codes across the transcripts. The codebook is included in appendix b.

With the coded data, the PI engaged in a variety of analysis methods. Memos were used throughout the analysis process to keep track of emerging concepts and to document definitions, personal reflections, questions, and patterns found throughout the analysis. Each transcript was summarized individually to highlight key characteristics. To further develop the analysis, memos were also used to highlight important overarching concepts across transcripts. This memo process allowed the PI to engage the data deeply and begin drawing connections across participants. The PI reviewed reports of the memos to revisit important concepts and to direct the subsequent steps of the analysis.

The PI reviewed the frequencies and coverages of each code to get a sense of which codes are most prevalent in each transcript. A code matrix was used to start understanding how the codes were applied across the transcripts overall. This matrix allowed the PI to view coded

segments across each of the transcripts and summarize the content overall. The PI continued to document patterns, questions, and reflections through memos. Code query tools were used to understand code interactions and relationships. Intersecting and co-occurring codes were analyzed to better understand the connections between concepts. This analytical process was used to conceptualize a model to describe the relationships between themes and codes (see figure 2). After the development of the model, the PI went back through the data with code queries and read segments to assess the fit of the model across cases. The conceptual model and relationships are reported below with supporting quotations.

Chapter 4: Manuscript

Qualitative Perspectives on Stress and Coping from Parents of Children with Marshall Smith-Syndrome

Mackenzie P. Taylor[1], Robin E. McGee[1], Yue Guan[1], and Briana Woods-Jaeger[1]

[1]Behavioral, Social, and Health Education Sciences, Rollins School of Public Health, Emory University, Atlanta, GA, 30322 USA

Abstract

Up to 30 million people living in the United States are affected by a rare disease. Though the specific diseases are rare, the broader impacts of rare disease affect up to 10 percent of the United States population. Marshall-Smith Syndrome (MSS) is one rare disease that is infrequently described in the literature. The people with MSS require lifelong care which often makes their parents long-term caregivers. Previous research has demonstrated some of the challenges of caregiving for children with medically complex and rare diseases, but no research has specifically focused on stress and coping for parents of children with MSS. The purpose of this study was to understand more about the experiences of parents of children with MSS, the significance of being a part of a rare disease community, and major stressors and coping strategies. Guided by the Transactional Model of Stress and Coping (TMSC), qualitative data were collected from nine parents of children with MSS (from eight families) through individual semi-structured interviews in August and September of 2019. The parents were recruited from a Facebook support group for MSS families and the interviews focused on the usefulness of online social support for this population. Inductive and deductive codes were used for thematic analysis of the data and three key themes emerged: 1) Fear of the unknown, 2) Social connection, and 3) Information gathering. These themes illuminated experiences of stress and coping for parents of children with MSS and the ways that online social support impacts the rare disease community. The findings fit within the previous literature on rare diseases and inform the importance of disease-specific social support and care. Understand more about the experiences of disease-specific communities can inform practice of genetic counseling and online interventions for families with rare disease.

Keywords

Rare Disease ● Stress and Coping ● Social Media ● Parent Caregivers ● Qualitative

Introduction

Up to 30 million people living in the United States are affected by one of the 7,000 known rare diseases (National Institutes of Health, 2017). Each distinct rare disease has unique challenges, but there are also known common experiences faced by many individuals living with rare diseases. In addition to the physical burden of living with disease, this population is likely to face complicated social stressors and difficulties within healthcare systems (Von der Lippe, Diesen, & Feragen, 2017). The known challenges faced by rare disease populations require attention, but many barriers limit effective research and support for people with rare diseases. Small population sizes, disease heterogeneity, and geographic dispersion make it difficult to conduct robust research on specific rare diseases (Whicher, Philbin, & Aronson, 2018). Moreover, are disease populations are often neglected due to the low count of individuals in each disease-specific community (Ladd, 2015). Although the specific diseases are rare, the broader impacts of rare diseases affect up to 10 percent of the United States population and their families. Research and the development of drugs for rare disease populations have been increasing public health priorities in the United States (Goldsmith, 2016). Still, more work is needed to increase awareness, develop treatments, and provide resources for rare disease communities.

Marshall Smith Syndrome (MSS) is one rare genetic disease that is infrequently described in the literature. This syndrome is characterized by accelerated bone formation, physical and cognitive delays, nutritional challenges, respiratory concerns, and distinct facial features such as high forehead, shallow orbits, retracted chin, depressed nose bridge, and bluish eye-whites (Shaw et al., 2010). Fewer than 100 cases have been documented worldwide since MSS was first described in 1971 (MSS Research Foundation, 2018). Very little demographic information about MSS is available because it so infrequently detected in the general population, but it appears to impact males and females equally in all parts of the world (MSS Research Foundation, 2018).

Mortality due to respiratory complications and infections has been common in early childhood, but advances in respiratory care increasingly makes it possible for individuals with MSS to reach adulthood (Shaw et al., 2010). As medical treatments improve, the growing number of adult cases will present new opportunities and challenges for people with MSS and their families. More research is needed to increase the body of knowledge and continue caring for this unique population. Individuals living with MSS require supportive environments into adulthood, meaning that MSS families and caregivers are important members of this community (MSS Research Foundation, 2016). Individuals with MSS and their caregivers face unique challenges and more information is needed to improve supportive resources for this population.

Very little information documents the unique stressors of parents that care for children with MSS. Among other groups of parent caregivers to children with chronic conditions, a significant amount of work has demonstrated unique stressors and emotional burdens (Cousino & Hazen, 2013). Parents of children with rare diseases face additional stress due to the lack of medical information, indefinite disease identity, uncertain prognosis, and underdeveloped clinical treatment (Picci et al., 2015). In a review of the literature assessing the supportive care needs of parents with a child with a rare diseases, this population most commonly reported social needs, information needs, and emotional needs (Pelentsov, Laws, & Esterman, 2015). In an attempt to meet these needs, many rare disease communities have turned to online support groups (Haik et al., 2019; Kauw et al., 2015; Meade, Buchanan, & Coulson, 2018; Rocha et al., 2018). A thematic analysis of one of these groups reported that the majority of the communication present falls into three categories: (1) information sharing; (2) emotional support, expression, and experience sharing; and (3) community building (Haik et al., 2019). These findings suggest that online support groups are useful in meeting the supportive care needs of parents caring for children with rare diseases.

Due to the scarcity of information regarding rare diseases available to healthcare providers, parents of a child with rare disease often take on the role of "expert" (Baumbusch, Mayer, & Sloan-Yip, 2019; Pelentsov et al., 2015). Online communities give these parents the platform to ask questions and share information with each other (Haik et al., 2019; Rocha et al., 2018). Despite the emerging literature surrounding online support groups for families with rare diseases, qualitative data capturing the parents' perspectives on these types of groups are lacking. More investigation is needed to gather the qualitative perspectives of parents of children with MSS, especially regarding online communities. Learning from these parents' perspectives will help to clarify the experience of MSS and the perceived value of online support groups for parents. This information can be used to inform future directions for supporting this population of parent caregivers. The current study explores the following questions: (1) What is the experience of parents of children with MSS? (2) What do parents of children with MSS identify as major stressors and coping strategies? (3) What is the perceived significance of being a part of a rare disease community? and (4) How have online social networks provided social support for these families?

Theoretical Framework

The current study is informed by Lazarus and Folkman's Transactional Model of Stress and Coping (TMSC) (Lazarus & Folkman, 1984). This model focuses on the person-environment relationship that guides the appraisal of life transitions (Devonport, 2011). Different people perceive a stressful event differently, and it is these perceptions, rather than the objective stressors that determine the effects on behaviors and health outcomes (Glanz, Rimer, & Viswanath, 2008). Based upon the appraisal of a stressful event as a perceived harm, threat, or a challenge to well-being, an individual engages in coping strategies (Devonport, 2011). The

TMSC provides a useful framework to inform the exploration of the experiences of parents of children with MSS. The relationships between constructs of the TMSC can be seen in figure 1.

Key constructs of the TMSC such as primary appraisals, secondary appraisals, coping efforts, social support seeking, and information seeking help to make meaning of the experience of stress for this population. Parents of children with MSS face unique and complicated stressors that come along with caring for a rare and medically complex child. Social support seeking was identified as a key coping effort within the study population based on the literature assessing online social support group for rare disease communities (Haik et al., 2019; Kauw et al., 2015; Meade, Buchanan, & Coulson, 2018; Rocha et al., 2018). Information seeking is another as a key coping effort for parents of children with rare diseases due to the limited information available within these populations (Picci et al., 2015; Pelentsov et al., 2015; Pelentsov, Fielder, Laws, & Esterman, 2016; Von der Lippe, Diesen, & Feragen, 2017; Baumbusch et al., 2019). These constructs provide useful parameters for the exploration of stress and coping for parents who care for children with MSS.

Methods

This study employed qualitative methods to collect in-depth semi-structured interviews in order to understand the experiences of parents of children with MSS. Individual interviews were conducted with nine different parents from eight families. The study was informed by the previous literature on MSS, parents of children with rare diseases and medical complexity, and online social support, as well as constructs from the Transactional Model of Stress and Coping (TMSC). Thematic analysis informed by the TMSC was used to analyze the data. Emory University's Institutional Review Board (IRB) approved the study before data collection began to ensure the protection of study participants.

The target population of this study was parents of children with MSS that were members of the MSS Facebook support group. At the time of recruitment, this Facebook group had 109 members, and this number included mostly parents, with some siblings or other family members. Approximately 30-40 families from a variety of countries were represented in this group. The MSS Facebook group is private, and the administrators carefully control who is allowed to join. As a family member of someone with MSS, the PI was already connected to this MSS Facebook group.

Stakeholder Engagement

Engaging with key stakeholders was a valuable preliminary step to learn about current research priorities within the MSS community. Initial contact was made through email with the MSS Facebook group administrator and several key doctors and researchers involved in MSS. The purpose of this step was to establish a relationship with the administrator as the gatekeeper, and to gather perspectives through stakeholder engagement. The PI introduced the conceptualized project and asked for suggestions that would help to guide the study. The MSS Facebook group administrator approved for PI to engage the group for this study and other feedback provided from the key stakeholders helped the PI to formulate the recruitment plan and the final version of the data collection instrument.

Recruitment and Eligibility

Participants were recruited online through the MSS Facebook support group. To enroll in the study, participants had to meet the following inclusion criteria: 1) participants had to be a parent of a child with MSS, 2) participants had to be a member of the MSS Facebook group, and 3) participants had to be comfortable speaking in English for a qualitative interview. Passive recruitment was employed with a virtual flier inviting parents to participate. Purposive sampling

and active recruitment were utilized by reaching out to eligible parents to individually invite them. Interested and eligible individuals received an electronic informed consent form with an overview of the study from the PI. Once the consent form was signed and returned, the participant was officially enrolled in the study and contacted by the PI to schedule an interview.

Instrument

Data were collected using a semi-structured qualitative interview guide designed to take 30-60 minutes. The interview guide questions were based upon the previous literature and constructs from the TMSC theoretical framework. The interview guide was broken into three distinct domains: 1) background, 2) daily stress and coping, and 3) online social support. The first section contained short-answer questions to gather demographics of the parent and some general background information about the family. The second section included questions to get a sense of the parent's daily stressors and coping efforts. Finally, the third section consisted of questions to understand the ways that parents use the Facebook support group for coping. Some questions included probes to encourage more discussion of key concepts.

Data Collection and Management

The PI scheduled individual time with all enrolled participants to conduct the interviews over the phone in August and September of 2019. Before each interview, the PI reviewed the purpose of the study and explained the participant's right to skip questions or end the interview at any point. The participant was given another opportunity to ask questions and the PI asked for permission to record the audio of the interview. The PI carefully followed the interview guide to facilitate the data collection and used probing questions as needed to draw out more information. Memos were used to document the PI's reactions and ideas about each interview.

Temi web-based automated transcription service was used to create verbatim transcripts of each recorded interview (Temi, 2020). The PI carefully reviewed the quality of each transcript by listening to each interview to manually correct any errors including typos and missing words. Memos were used throughout the interview and transcription process to document ideas and reactions. Reflecting on these memos helped guide the subsequent interviews. Additional probes were used to explore emerging concepts over time. For example, participants often mentioned their other children while talking about their family backgrounds. The PI began probing more about these sibling relationships to understand the family dynamics surrounding MSS. The PI also used probes based on the topics highlighted by each participant. For example, if the participant mentioned having home nursing care, the PI would ask more about this experience for the family. No new content areas were added to the interview guide.

Each study participant was assigned a unique ID number to ensure confidentiality. All of the interview audio recordings and transcripts were stored on a password protected and secure network. At the end of the study, the audio recordings were permanently deleted.

Data Analysis

Thematic analysis was employed to analyze the verbatim transcripts. The PI utilized MaxQDA version 2018.2 throughout the coding and analysis process. A codebook was developed that included both deductive and inductive codes. To develop and refine the initial codebook, the PI engaged with the interview content by listening to the audio recordings. Memos were used to summarize and document initial ideas. Deductive codes were based upon the research questions, interview guide, and the TMSC framework. The inductive codes were added into the codebook as they emerged from the transcript content. For example, many parents mentioned the significance of physical similarities between the children with MSS, so an inductive code was added to track this pattern. The PI used the final codebook to carefully code

each transcript line-by-line. A secondary coder assisted by coding every third transcript to compare for agreement. Reflecting on the code agreement and disagreement helped to ensure the consistent application of codes across the transcripts.

With the coded data, the PI engaged in a variety of analysis methods. Memos were used throughout the analysis process to keep track of emerging concepts and to document definitions, personal reflections, questions, and patterns found throughout the analysis. Each transcript was summarized individually to highlight key characteristics. To further develop the analysis, memos were also used to highlight important overarching concepts across transcripts. This memo process allowed the PI to engage the data deeply and begin drawing connections across participants. The PI reviewed reports of the memos to revisit important concepts and to direct the subsequent steps of the analysis.

The PI reviewed the frequencies and coverages of each code to get a sense of which codes are most prevalent in each transcript. A code matrix was used to start understanding how the codes were applied across the transcripts overall. This matrix allowed the PI to view coded segments across each of the transcripts and summarize the content overall. The PI continued to document patterns, questions, and reflections through memos. Code query tools were used to understand code interactions and relationships. Intersecting and co-occurring codes were analyzed to better understand the connections between concepts. This analytical process was used to conceptualize a model to describe the relationships between themes and codes (see figure 2). After the development of the model, the PI went back through the data with code queries and read segments to assess the fit of the model across cases. The conceptual model and relationships are reported below with supporting quotations.

Results

The study sample consisted of eight mothers and one father from eight distinct families. The mean age of parents was 44 years old, and the ages of children ranged from early childhood to adulthood. Demographic information about the participants is reported in table 1 and demographic information about their children with MSS is reported in table 2. The variables were presented in categories to protect the privacy of this small population. The coding and analysis processes highlighted important content across the data known as themes. The PI has identified three key themes though the data analysis to address the research questions. The results illuminate the experience of experience of parents of children with MSS, the major stressors and coping strategies, the significance of being in community, and the usefulness of online social support. The themes have been named, described, and supported with quotations below. The dimensions of these themes have been explored to describe what is typical in the data and how much variation exists across cases.

The first theme addressed the research questions related to the experience of parents of children with MSS and major stressors for this population:

Theme 1: Fear of the Unknown

The unknown nature of rare diseases played an important role in the stress that parents experienced as caregivers for a child with MSS. Each participant described their experience navigating the unknown circumstances of caring for their child. Participants described many unknown factors that they worried about when their child was born with abnormalities. The timing of the diagnosis varied, but even when they received a diagnosis and got more information, there were still many unanswered questions. Parents wondered if their child would

ever walk, talk, or live a healthy life. All of the formal information available about MSS left families fearful about the unknown future.

All participants worried about the future at the time of birth when they realized that their child would need supportive care. The participants' experiences of the unknown varied depending on the timing of diagnosis. Some families had a diagnosis of MSS for their child within weeks, and other families waited years for this valuable information. The families that had longer time periods without knowing a diagnosis faced additional challenges because they had no roadmap for what to expect or what health problems to look out for. No matter the length of time between birth and diagnosis, all families faced the emotional burden of not knowing what was going on with their child for some amount of time. One participant described the time period before diagnosis this way:

> *They did all kinds of tests. They couldn't figure out any physical difficulties. You know, they checked her heart, they checked everything and sent us home with no answers…So our pediatrician tried to tell us, 'You know, every child is different. She's just a little different.' Well I knew that she was more than different…we just didn't know what exactly it was.*
>
> - *Mother 4*

This time period of the unknown caused stress and discomfort to parents. They worried about the health and future well-being of their child. Many parents explained that they were eager to find answers so that they could know what to be prepared for and how to care for their child. However, one parent who went years without a diagnosis explained that knowing some of the answers would also be difficult. This participant noted:

> *If someone would have told me this is what we were facing, it would have been harder for me to face it.*
>
> - *Mother 1*

This parent's assessment was mirrored throughout the experiences of others. Even though all parents were longing for more information, once they received a diagnosis and started getting

answers, many found it to be just as fearful. Especially because many parents felt that the medical literature on MSS was lacking and older articles portrayed children with MSS dying in infancy or early childhood. One parent explained:

> *We read about two cases from 1971, and horrible. It was just horrible. It was like the worst thing ever. It was like a death sentence and [the doctors] said, 'she's basically not going to make it.' And they did nothing.*
>
> - *Mother 3*

Many parents had similar experiences and had to wonder if their baby would make into childhood. Other parents did not even get this diagnosis and prognosis until they were already well beyond infancy and childhood. Some parents felt relief when they received a diagnosis because they had been looking for answers for so long. But the relief was followed by more uncertainty because most doctors had no previous knowledge of MSS. They did not have the answers that parents were looking for. This participant explained his feelings after receiving the diagnosis of MSS:

> *[When we received the diagnosis], many emotions were felt. There was the emotion of confusion, because obviously, Marshall-Smith was not very well known. There was a lot of confusion felt, but there was some relief felt that we actually had a diagnosis. I think the overall feeling for me was uncertainty for the future.*
>
> - *Father 1*

The only answers that the doctors could provide came from the limited medical literature about MSS. Participants explained that the research that was available to them depicted MSS in a disheartening way. This information was discouraging to parents and led to emotional stress. Many parents described situations where their children exceeded all expectations set by the doctors and medical literature. One parent reflected back on the time of her child's diagnosis:

> *[When we received the diagnosis], I think we were a little upset because they basically told us that she wasn't going to be much of anything. They said that she would be severely delayed mentally and physically. But as it turns out, she is very smart and is learning things. What they initially told us is far from the truth.*
>
> - *Mother 2*

The participants shared that the experience of receiving the diagnosis of MSS was emotional and overwhelming. Some felt relieved to have the answers but other felt more discouraged by the unknown prognosis. Many of the parents hoped and believed that their children could be more than what the medical literature predicted. Parents were often unsatisfied with the information available to them from the doctors, and they searched for more answers elsewhere.

The parents' appraisal of the unknown situation prompted them to find ways to adapt. The second and third themes addressed the research questions related to the experience of parents of children with MSS, major stressors and coping strategies for this population, the significance of being in community with other MSS families, and the value of online social support:

Theme 2: Information Gathering

All participating parents discussed searching for more information about MSS to some extent. Doctors often provided some publications to parents at the time of diagnosis, but parents looked to supplement this information with other sources. Before the internet was available on personal computers, parents had less access to research on their own. These parents often connected with families that had children with other syndromes and disabilities, but went years without hearing personal stories from other MSS families. One parent described her search for more information before the internet:

> *We had to do our own research. I contacted my brother-in-law who looked up some articles. We did have some doctors who were friends and we had them look things up.*
>
> - *Mother 3*

Parents often described creative ways that they used their networks to search for more information on MSS. It became even easier to connect and access information when the internet became popular. All of the parents who had children born after the year 2000 described their internet investigations to learn about MSS.

The internet gave parents more power to search on their own. In addition to giving parents access to more research, the internet also connected parents to other personal stories from families with MSS. Each parent described searching online and finding the Marshall-Smith Research Foundation website (www.marshallsmith.org). They explained that this resource provided more personal stories of people with MSS in addition to the medical information. The families that reached out to the MSS Research Foundation often got connected to the MSS Facebook group, but other families had a harder time finding the group because of its privacy settings. The MSS Research Foundation website provided medical journals as well as stories and pictures of families around the world with MSS. Many parents described the positive impact of finding these resources. Seeing other kids and hearing stories gave parents a much more optimistic outlook on their future. One parent described her experience on the MSS Research Foundation website:

> *We found the MSS website before we found the Facebook group page and we learned that [children with MSS] have a very happy disposition. And it was very comforting.*
>
> - *Mother 7*

Once parents got connected to the MSS Facebook group, they were able to ask questions and learn from the network of other families. All of the parents agreed that hearing personal stories was much more helpful than anything they read in the medical literature. This type of information gave parents the chance to learn about the positive experiences of children with MSS rather than just the medical complications. One parent shared her perspective on this:

> *The personal information is so much more helpful. All of the research is so old…back when they were largely dying in infancy. So, most of my information comes from other parents because I don't think the research can compare.*
>
> - *Mother 8*

Even with all of the online resources available, participants described a scarcity of information to be known about MSS. Some parents expressed that they spent time focusing on

the individual needs of their child rather than searching for answers online. One parent explained her approach:

> *You know, I don't remember looking for [information] all that much. We knew what it was, and we pretty much just looked at her and looked at what her issues were and just kind of went from there.*
>
> - *Mother 4*

This approach focused on the child's needs on a case-by-case basis. Many other parents described similar strategies after they did not find sufficient information online. Rather than trying to learn everything there was to know about MSS, parents focused on addressing individual problems as they emerged. This seemed to help parents take things one day at a time without becoming overwhelmed. This parent explained in her own words:

> *I feel like in the beginning everything was such a whirlwind and we were just putting one foot in front of the other and taking in information. I feel like my husband and I were realistic and kind of matter of fact about things at first. To make decisions, it was good to know what we were dealing with.*
>
> - *Mother 6*

Parents described that they did not know everything that they would have liked to know, so they made the best decisions based on the information that they had at the time.

According to the experiences of the participants, the MSS Facebook group opened an opportunity to learn in a new way. Parents could gather information from the lived experiences of others. This was a step further than just hearing about personal stories from other families, because the parents were able to ask questions to get feedback in real time. One participant explained the content of the MSS Facebook group:

> *I think there's a mix between people asking questions and looking for medical information. They look for what to expect and updates, and ask, 'did this happen?' or, 'what was this surgery about?' Everybody is just sharing their lives and what's happening.*
>
> - *Mother 6*

Many parents value the lived experiences of the other families and were eager to learn about the strategies and treatment plans that have worked for other children with MSS. The MSS Facebook group offered families a dynamic source of information that they can always revisit to learn more.

Theme 3: Social Connections

All nine participants discussed ways that social support has been important to them. Each parent mentioned the friends and family that they have felt supported by or that they could call when they have needed help. Having people to confide in has helped parents cope with the fears and stressors that comes with parenting a child with a rare disease. Fewer parents mentioned people in their lives that understand how to help specifically with their child with MSS. Socially confiding in other families with MSS was identified as a uniquely important component of coping for participants.

Many parents mentioned how appreciative they were of the supportive people in their lives. These parents had a sense that they could not have made it through difficult times without the people in their lives that have supported them over the years. This participant shared about her social support system:

> *We have tons of support in our church, our neighborhood, our jobs, and our family…We had a birthday party and invited everyone we know mostly as a 'thank you' to them for all of their kindness and support over the years. These people were willing to drop off food or watch the other kids if he's been in the hospital, and send cards.*
>
> - *Mother 6*

She explained that these people have been able to step in and help when things get difficult. However, this mother goes on to say:

> *So we have a lot of support, but when it comes to caring for our son…it's just myself, my husband, and my mother.*
>
> *-Mother 6*

Even parents that have a strong social support system often felt alone when it came to caregiving for their child with MSS. There was a sense that other people do not fully understand the situation or what the family is going through.

In addition to the MSS Facebook group, some parents described support groups or connections with other special needs parents. Even though these people do not know specifically about MSS, they still understand more as another parent of a child with special health needs. According to the experiences of participants, these types of support groups or connections have been a valuable resource for their families. This parent explained her experience with a support group:

> *I was part of a support group when my daughter was three years old. All of the people that were in physical therapy were part of this group and that was really good. Then I had a couple of friends who had children with different syndromes, and so it was good just for the mere fact of knowing two people that you could call at any time.*
>
> - *Mother 3*

This mother emphasized the importance of having someone to talk to about her daily challenges and stressors. These connections provided this mother with people who could relate before she ever met anyone else with MSS. Other participants mentioned other types of online support groups for families with special needs and they have some unique benefits. For example, if they had access to local groups, it gave the families opportunities to meet up in person. However, participants explained that these groups that are not disease-specific are much larger and do not have the same sense of closeness as the MSS community.

The MSS Facebook group provided families with easy access to other people who understand what they are going through. Parents described that connecting to the community gave them the opportunity to interact and engage with other families that are similar to them. One parent described his interaction with the MSS Facebook Group:

> *When my son had his surgery, we felt very supported. Everyone was saying that they're glad that the surgery went well and that he's doing better. And when we do post about us doing activities, we'll see the other families posting about how great it is to see my son out and about and doing fun things. And that is encouraging, because these are from the people that know the difficulty.*
>
> - *Father 1*

This father described the added layer of support that comes from people who understand what his family goes through. This sense of understanding contributed to a strong comradery among MSS family members. Other participants shared about how welcoming it was to become a member of the MSS Facebook group. One parent shared her experience when she first joined the group:

> *I did an introduction post, and everyone was really nice and introduced their children too. So that was welcoming.*
>
> - *Mother 5*

The participants explained that they felt comfortable sharing openly on the MSS Facebook group and consider each other like family. On the MSS Facebook group, parents described feeling supported by one-another because they all belong to a small and unique community. One parent even put it this way:

> *[Joining the Facebook group] was like a sense of coming home, I guess. It was very welcoming. Before, it was kind of isolating and I felt like we were trying to do everything on our own. And now, there are people who can help and understand.*
>
> - *Mother 7*

Relationship Between Stress and Coping

The relationships between the three themes mapped to the relationship between stress and coping. The core theme, *fear of the unknown,* was a major stressor that parents of children with MSS experience. The parents employed coping strategies such as *information gathering* and relying on *social connections.* These coping themes related to the core stressor theme via the MSS Facebook group. When parents participated with the MSS Facebook group, they engaged in active coping for the *fear of the unknown*, which helped them to adapt. Moreover, the connections between these two coping mechanisms happening simultaneously promoted even

more favorable outcomes. The relationships between these three themes is conceptualized in figure 2. This conceptual model is exemplified in the following quotation:

> *The medical articles can scare the crap out of you. But we already knew he didn't fit all of those pessimistic sort of stories. And you know, talking to families is 100 percent more helpful, but [the medical articles] gave me questions to ask. It gave me a point of reference to ask questions.*
>
> - *Mother 1*

This mother acknowledged her *fear of the unknown* based on what she found in the medical articles. She also highlighted that talking with families (*social connections*) provided much more helpful information (*information gathering*). With the help of the MSS Facebook group, she was able to better adapt to the challenges of parenting a child with MSS.

Each of the participants expressed that social connections to other families made it easier to face the unknown and gather information together. When these parents connected into the MSS Facebook group, they gained the opportunity to learn from real life experiences and interact with a group of people who understand some of the things they were going through. One participant described it this way:

> *You see pictures and you hear personal stories on Facebook. The ability to message one another, to not just see information and read it, but also to be able to interact and ask questions…not just kind of reading things that other people wrote without the ability to interact.*
>
> - *Mother 6*

Some families described relying on this social interaction more than others. For example, the families that already found helpful ways of adapting before they joined the MSS Facebook group did not rely as much on that community. One participant that had a child who received the diagnosis of MSS much earlier than the MSS Facebook group had been created explained:

> *So, at the point I came to [the MSS Facebook group], while it's a great resource, it didn't really help us. And I'm glad it can help other people. But we had to figure it out on our own.*
>
> - *Mother 3*

However, this mother went on to describe what it would have been like for her family if she had access the MSS Facebook group earlier in her journey with MSS:

> *Oh, I think it would have been a tremendous asset. Because you could turn to people who actually could relate to your situation. You could talk to them about doctors, feelings, fears, gratitude, and all of it. We couldn't talk to anybody like that. I could talk to the couple people who had kids with a disability, but no one else.*
>
> - *Mother 3*

This mother reflected on the importance of the MSS Facebook group and expressed that she is glad that parents now have such useful resources.

Even the participants that did not rely as heavily on the social components of the MSS Facebook group for coping still felt encouraged by the content posted. In addition to information gathering on the MSS Facebook group, parents mentioned sharing accomplishments, milestones, and photos of their children. This helped parents build perceptions about MSS on the positive attributes rather than just the medical challenges. For many participants this was a positive reminder of the sweet disposition of children with MSS. One participant shared:

> *The main thing that was very comforting was to see these kids, and to see how happy they were. And to see all of the posts from the families of kids smiling.*
>
> - *Father 1*

Each of the parents shared similar sentiments regarding their own children and the other children on the MSS Facebook group. This sharing of positivity may be an important source of hope for parents of children with MSS.

Assessment of Model Fit in the Data

After taking this model back to the data, it is clear that all nine cases fit the model to some extent. Some people appraise the future to be more severe than others and therefore experience greater *fear of the future*. Some brought up other coping mechanisms, however, all participants have experienced the coping benefits of *information gathering* and *social connections* on the

MSS Facebook group in some way. More data should be collected to test this model with a larger sample.

Discussion

The results outlined above illuminate the experience of caring for a child with MSS. Being a part of a rare disease community, especially through online social media is an important way of coping for families with extremely rare diseases. The MSS Facebook group gives parents an environment to connect socially and gain information from people who know the specifics of their child's condition. Parents of children with MSS face intense stressors especially due to a *fear of the unknown.* When children are diagnosed with MSS, the family does not know what to expect and there is little information available for them to learn from. The MSS Facebook group provides families with a platform to cope with the challenges of caring for a child with MSS through *information gathering* and fostering *social connections.*

These results for MSS are consistent with the stress and coping findings from studies of parents of children with other rare diseases (Pelentsov, Laws, & Esterman, 2015; Pelentsov, Fielder, Laws, & Esterman, 2016; Baumbusch, Mayer, & Sloa-Yip, 2019). The participants discussed their experiences of anxiety and confusion about MSS, which is not unusual among rare disease populations (Picci et al., 2015). The participants also shared about the disheartening information found in the medical literature that informed their perceptions of the severity of MSS. The experiences of MSS parents align with the literature describing social, emotional, and informational needs of parents of children with rare diseases (Pelentsov, Laws, & Esterman, 2015). Baumbusch, Mayer, and Sloa-Yip (2019) highlighted the importance of social support to meet the needs of these parent populations. The results of the current study suggested that online social support met social and informational needs of MSS parents. The benefits of using online

social support highlighted in this analysis are consistent with previous research, but these studies focus on peer-to-peer support for patients rather than parents (Haik et al., 2019; Kauw et al., 2015; Meade, Buchanan, & Coulson, 2018; Rocha et al., 2018). The MSS parents of the current study described using social media in ways that align with the findings of previous studies such as discussing diagnosis and connecting to find social and emotional support. Demonstrating these findings within a new disease population reinforces past rare disease findings and adds to the body of knowledge about MSS.

Implications for Theory and Practice

The TMSC served as a framework for understanding the experiences of stress and coping for parents within the MSS community. Constructs from the TMSC including primary appraisals, secondary appraisals, coping efforts, social support seeking, and information seeking guided this research from the development of the interview guide through to the analysis of the data. The relationships between the three themes are informed by the constructs from the TMSC (Lazarus & Folkman, 1984). Appraisals of stressors inform the coping efforts that a person employs (Glanz et al., 2008). The core theme, *fear of the unknown,* is a major stressor that parents of children with MSS experience. Parents appraise their futures by 1) judging the severity of circumstances (primary), and 2) judging their ability to respond to the given circumstances (secondary). As a result, the parents employ coping strategies such as *information gathering* and relying on *social connections.* These coping themes related to the core stressor theme via the MSS Facebook group. When parents participated with the MSS Facebook group, they engaged in active coping for the *fear of the unknown*, which helped them to adapt.

The results from the current study align with the TMSC and build upon the current understanding of these constructs. For example, social support is an important part of coping in this framework, but the capacity to engage in social support online brings in new elements. In

some ways, online social support may be more feasible when communities are limited by geographic dispersion or language differences, like the MSS community. Facebook can connect people across the world and translate text instantly. These technologies present new opportunities for social support that were not possible before time of personal computers and social media.

Health providers and genetic counselors should be aware of the psychological stressors related to caring for a child with a rare diagnosis and help connect parents to resources, such as online social support groups. It is important for these providers and counselors to understand the common experiences for families with rare disease while emphasizing the unique value of being connected to others within a disease specific community. Preparing a parent with realistic expectations and equipping them with tools to cope may increase the belief that they are capable of dealing with challenging circumstances. Working to make parents feel more prepared will improve secondary appraisals and decrease the impact of stress on this population.

Strengths and Limitations

A key strength of the current study is that it collected qualitative perspectives from a population of parents whose experiences have not been documented. Though the results are consistent with other rare disease populations, each specific group has unique experiences and stories to tell. Lifting up these voices increases the body of information available for parents of children who are newly diagnosed with MSS. Though the number of families may be few, each rare disease population deserves to have their stories documented in the literature. Additionally, these findings support the value of online social support groups for rare disease communities. Online social support groups may have additional value for extremely rare diseases such as MSS because the population is geographically dispersed. Care providers should work to connect families into rare disease-specific support groups upon diagnosis.

These results should be interpreted with the following limitations in mind. A small sample size limits the generalizability of the findings. The small sample of the current study lacks variety in sex, race, and ethnicity. The homogenous sample may not represent all of the variety of experiences within the MSS parent community. The sample also may be limited by selection bias. If the parents who agreed to participate are the most involved in the MSS Facebook support group, the results may be missing other perspective from families who do not care as much about being a part of the MSS community. It is important not to over-generalize and to keep in mind that every family has a unique experience. Finally, community members should also be involved in reviewing the results so that they can provide feedback on the interpretation of the data. This step would increase community accountability to produce the strongest results.

Future Directions

Future studies should continue assessing the experience of parents of children with rare diseases through the constructs of the TMSC framework and the conceptual model described above. Additional data from parents of children with MSS will help refine and clarify the current findings. A more diverse sample including various races and ethnicities will articulate more variety and nuance of experiences. Future studies should also investigate the impacts of connecting researchers and doctors into these online communities. If parents are comfortable sharing openly amidst doctors and researchers, this may be a useful way to diffuse information both directions. However, the presence of doctors and researchers may change the dynamics of these online support groups. Special care should be taken to preserve the safe and open environment to support parents of children with rare diseases.

Conclusions

In conclusion, the experience of parenting a child with MSS is stressful and many parents fear the unknown course of a rare disease. Parents of children with MSS face constant challenges but there are opportunities for these parents to adapt by engaging in coping practices. Finding online community with other MSS families promotes key coping strategies such as information gathering and social connection for parents. The MSS Facebook group is a unique tool to help parents of children with MSS cope with the stressors of rare disease. The MSS Facebook support group gives parents the opportunity to engage in disease-specific community and collective learning about MSS. Innovative disease-specific support groups should be prioritized to connect families with rare diseases and promote health to this population.

References

Baumbusch, J., Mayer, S., & Sloan-Yip, I. (2019). Alone in a Crowd? Parents of Children with Rare Diseases' Experiences of Navigating the Healthcare System. *Journal of Genetic Counseling*, *28*(1), 80–90. https://doi.org/10.1007/s10897-018-0294-9

Cohen, E., Kuo, D. Z., Agrawal, R., Berry, J. G., Bhagat, S. K. M., Simon, T. D., & Srivastava, R. (2011). Children with medical complexity: An emerging population for clinical and research initiatives. *Pediatrics*, *127*(3), 529–538. https://doi.org/10.1542/peds.2010-0910

Cousino, M. K., & Hazen, R. A. (2013). Parenting stress among caregivers of children with chronic illness: a systematic review. *Journal of Pediatric Psychology*, *38*(8), 809–828. https://doi.org/10.1093/jpepsy/jst049

Devonport, T. (2011). A brief review of commonly used theories and the importance of applying theory in practice. In T. J. Devonport (Ed.), *Managing stress: From theory to application.* (pp. 1–12). Retrieved from https://login.proxy.library.emory.edu/login?url=http://search.ebscohost.com/login.aspx?direct=true&db=psyh&AN=2013-06435-001&site=ehost-live&scope=site

Fitch, N. (1980). The syndromes of Marshall and Weaver. *Journal of Medical Genetics*, *17*(3), 174–178.

Glanz, K., Rimer, B. K., & Viswanath, K. (2008). *Health Behavior and Health Education : Theory, Research, and Practice*. Retrieved from http://ebookcentral.proquest.com/lib/emory/detail.action?docID=353367

Goldsmith, J. (2016). From our perspective : Encouraging drug development for rare diseases. Retrieved from United States Food and Drug Administration website: https://www.fda.gov/drugs/news-events-human-drugs/our-perspective-encouraging-drug-development-rare-diseases

Haik, D., Kashanchi, K., Tajran, S., Heilbronn, C., Anderson, C., Francis, D. O., … Verma, S. P. (2019). The Online Support Group as a Community: A Thematic Content Analysis of an Online Support Group for Idiopathic Subglottic Stenosis. *Annals of Otology, Rhinology and Laryngology*, *128*(4), 293–299. https://doi.org/10.1177/0003489418820348

Hoyme, H. E., Byers, P. H., & Guttmacher, A. K. (1993). Marshall-Smith syndrome: further evidence of an osteochondroplasia in long-term survivors. *Proceedings of the Greenwood Genetics Center*, *12*(70), 418–419.

Johnson, J. P., Carey, J. c., Glassy, F. J., Paglieroni, T., & Lipson, M. H. (1983). Marshall-Smith syndrome: Two case reports and a review of pulmonary manifestations. *Pediatrics*, *71*(2), 219–223.

Kauw, D., Repping-Wuts, H., Noordzij, A., Stikkelbroeck, N., Hermus, A., & Faber, M. (2015). The contribution of online peer-to-peer communication among patients with adrenal disease to patient-centered care. *Journal of Medical Internet Research*, *17*(2), 1–7. https://doi.org/10.2196/jmir.3869

Ladd, D. (2015). General and Health Information Challenges of Patients With Rare Diseases: The Importance of Health Information Provision and Web Sites for Locating Rare Disease Resources. *Journal of Hospital Librarianship*, *15*(2), 189–197. https://doi.org/10.1080/15323269.2015.1014762

Lazarus, R. S., & Folkman, S. (1984). *Stress, Appraisal, and Coping* (1 edition). Retrieved from https://books.google.com/books?hl=en&lr=&id=i-ySQQuUpr8C&oi=fnd&pg=PR5&dq=Lazarus,+R.S.,+%26+Folkman,+S.+(1984).+Stress,+Appraisal,+and+Coping.+New+York:+Springer+Publishing+Company.&ots=DfHVnshiSi&sig=N83GXocWJG1Pnv9vb_z6PhsM7Sk#v=onepage&q=Lazarus%2C R.

Malan, V., Rajan, D., Thomas, S., Shaw, A. C., Louis Dit Picard, H., Layet, V., … Cormier-

Daire, V. (2010). Distinct effects of allelic NFIX mutations on nonsense-mediated mRNA decay engender either a sotos-like or a Marshall-Smith Syndrome. *American Journal of Human Genetics*, *87*(2), 189–198. https://doi.org/10.1016/j.ajhg.2010.07.001

Marshall, R. E., Graham, C. B., Scott, C. R., & Smith, D. W. (1971). Syndrome of accelerated skeletal maturation and relative failure to thrive: A newly recognized clinical growth disorder. *The Journal of Pediatrics*, *78*(1), 95–101. https://doi.org/10.1016/S0022-3476(71)80269-X

Meade, O., Buchanan, H., & Coulson, N. (2018). The use of an online support group for neuromuscular disorders: a thematic analysis of message postings. *Disability and Rehabilitation*, *40*(19), 2300–2310. https://doi.org/10.1080/09638288.2017.1334239

MSS Research Foundation. (2016). Standards of Care for Marshall-Smith Syndrome. Retrieved from MSS Research Foundation website: www.marshallsmith.org

MSS Research Foundation. (2018). WHAT IS MSS? Retrieved from MSS Research Foundation website: https://www.marshallsmith.org/en/what-is-mss

National Institutes of Health. (2017). FAQs About Rare Diseases. Retrieved from National Institute of Health website: https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases

Pelentsov, L. J., Fielder, A. L., Laws, T. A., & Esterman, A. J. (2016). The supportive care needs of parents with a child with a rare disease: Results of an online survey. *BMC Family Practice*, *17*(1), 1–13. https://doi.org/10.1186/s12875-016-0488-x

Pelentsov, L. J., Laws, T. A., & Esterman, A. J. (2015). The supportive care needs of parents caring for a child with a rare disease: A scoping review. *Disability and Health Journal*, *8*(4), 475–491. https://doi.org/10.1016/j.dhjo.2015.03.009

Picci, R. L., Oliva, F., Trivelli, F., Carezana, C., Zuffranieri, M., Ostacoli, L., … Lala, R. (2015).

Emotional Burden and Coping Strategies of Parents of Children with Rare Diseases. *Journal of Child and Family Studies*, *24*(2), 514–522. https://doi.org/10.1007/s10826-013-9864-5

Rare Disease Act of 2002, Pub. L. No. 107–280, 116 Stat. 1988 (2002).

Rocha, H. M., Savatt, J. M., Riggs, E. R., Wagner, J. K., Faucett, W. A., & Martin, C. L. (2018). Incorporating Social Media into your Support Tool Box: Points to Consider from Genetics-Based Communities. *Journal of Genetic Counseling*, *27*(2), 470–480. https://doi.org/10.1007/s10897-017-0170-z

Shaw, A. C., Van Balkom, I. D. C., Bauer, M., Cole, T. R. P., Delrue, M. A., Van Haeringen, A., … Hennekam, R. C. M. (2010). Phenotype and natural history in Marshall-Smith syndrome. *American Journal of Medical Genetics, Part A*, *152*(11), 2714–2726. https://doi.org/10.1002/ajmg.a.33709

Sperli, D., Concolino, D., Barbato, C., Strisciuglio, P., & Andria, G. (1993). Long survival of a patient with Marshall-Smith syndrome without respiratory complications. *Journal of Medical Genetics*, *30*, 877–879. https://doi.org/10.1136/jmg.30.10.877

Summers, D. A., Cooper, H. A., & Butler, M. G. (1997). Marshall-Smith syndrome: case report of a newborn male and review of the literature. *Clin Dysmorphol.*, *8*(3), 207–210.

Temi. (2020). Retrieved from https://www.temi.com/

Van Balkom, I. D. C., Shaw, A., Vuijk, P. J., Franssens, M., Hoek, H. W., & Hennekam, R. C. M. (2011). Development and behaviour in Marshall-Smith syndrome: An exploratory study of cognition, phenotype and autism. *Journal of Intellectual Disability Research*, *55*(10), 973–987. https://doi.org/10.1111/j.1365-2788.2011.01451.x

Von der Lippe, C., Diesen, P. S., & Feragen, K. B. (2017). Living with a rare disorder: a systematic review of the qualitative literature. *Molecular Genetics and Genomic Medicine*, *5*(6), 758–773. https://doi.org/10.1002/mgg3.315

Whicher, D., Philbin, S., & Aronson, N. (2018). An overview of the impact of rare disease characteristics on research methodology. *Orphanet Journal of Rare Diseases*, *13*(1), 1–12. https://doi.org/10.1186/s13023-017-0755-5

Williams, D. K., Carlton, D. R., Green, S. H., Pearman, K., & Cole, T. R. P. (1997). Marshall-Smith syndrome: the expanding phenotype. *Journal of Medical Genetics*, *34*, 842–845. https://doi.org/10.1136/jmg.34.10.842

Wilson, B. T., & Hughes, J. (2019). Joining the hidden revolution in rare diseases: working with family support groups. *Archives of Disease in Childhood*, archdischild-2019-317227. https://doi.org/10.1136/archdischild-2019-317227

Woodgate, R. L., Edwards, M., Ripat, J. D., Borton, B., & Rempel, G. (2015). Intense parenting: A qualitative study detailing the experiences of parenting children with complex care needs. *BMC Pediatrics*, *15*(1), 1–15. https://doi.org/10.1186/s12887-015-0514-5

Yoder, C. C., Wiswell, T., Cornish, J. D., Cunningham, B. E., & Crumbaker, D. H. (1988). Marshall Smith Syndrome: Further Delineation. *Southern Medical Journal*, *81*(10), 1297–1300.

Tables

Table 1. Demographic variables[a] of participants (N=9)

Variables	n	Percentage
Sex		
Female	8	89
Male	1	11
Age		
30-39 years	5	56
40-49 years	2	22
50 years and over	2	22
Education		
Some College	2	22
Bachelor's degree	4	44
Graduate Degree	3	33
Employment status		
Full-time	5	56
Part-time	2	22
Not employed	2	22

[a]Variables are reported in categories to protect privacy

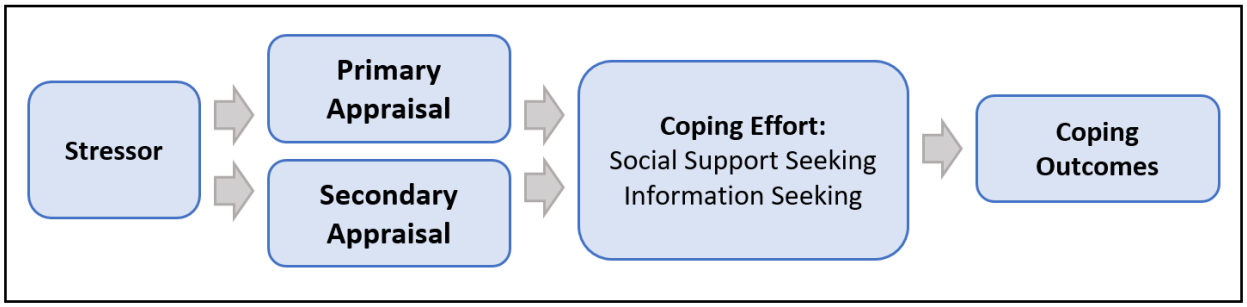
Table 2. Demographic variables[a] of children with MSS[b] (N=8)

Variables	n	Percentage
Sex		
Female	5	63
Male	3	38
Age		
0-5 years	2	25
6-12 years	3	38
13 years and over	3	38
Timing of diagnosis		
0-1 month	2	25
2-12 months	3	38
>1 year	3	38

[a]Variables are reported in categories to protect privacy
[b]Marshall-Smith Syndrome

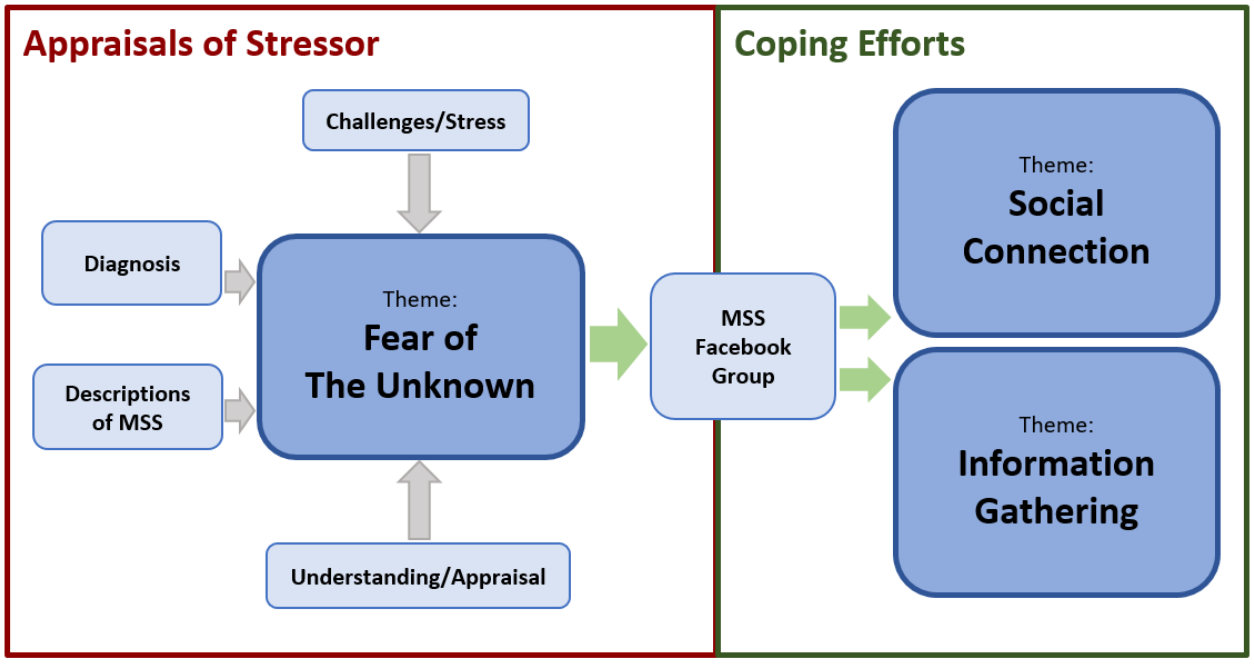
Figures

Figure 1. Transactional Model of Stress and Coping[a]



[a]Adapted from Glanz et al. (2008)

Figure 2. Conceptual model demonstrates relationships between codes and themes



Chapter 5: Public Health Implications

The results outlined above illuminate the experience of caring for a child with MSS. Being a part of a rare disease community, especially through online social media is an important way of coping for families with extremely rare diseases. The MSS Facebook group gives parents an environment to connect socially and gain information from people who know the specifics of their child's condition. Parents of children with MSS experience intense stressors especially due to a *fear of the unknown.* When children are diagnosed with MSS, the family does not know what to expect and there is little information available for them to learn from. Navigating every step of that journey can be isolating and stressful for these parents. The MSS Facebook group provides families with a platform to cope with the challenges of caring for a child with MSS through *information gathering* and fostering *social connections.* Parents have the opportunity to ask specific questions and learn from the experiences of other families with MSS in real time. The social connections online build feelings of community membership and families can focus on positive stories rather than the problems associated with MSS.

These results for MSS are consistent with the stress and coping findings from studies of parents of children with other rare diseases (Pelentsov, Laws, & Esterman, 2015; Pelentsov, Fielder, Laws, & Esterman, 2016; Baumbusch, Mayer, & Sloa-Yip, 2019). The participants discussed their experiences of anxiety and confusion about MSS, which is not unusual among rare disease populations (Picci et al., 2015). The participants also shared about the disheartening information found in the medical literature that informed their perceptions of the severity of MSS. The experiences of MSS parents align with the literature describing social, emotional, and informational needs of parents of children with rare diseases (Pelentsov, Laws, & Esterman, 2015). Baumbusch, Mayer, and Sloa-Yip (2019) highlighted the importance of social support to

meet the needs of these parent populations. The results of the current study suggested that online social support met social and informational needs of MSS parents. The benefits of using online social support highlighted in this analysis are consistent with previous research, but these studies focus on peer-to-peer support for patients rather than parents (Haik et al., 2019; Kauw et al., 2015; Meade, Buchanan, & Coulson, 2018; Rocha et al., 2018). The MSS parents of the current study described using social media in ways that align with the findings of previous studies such as discussing diagnosis and connecting to find social and emotional support. Demonstrating these findings within a new disease population reinforces past rare disease findings and adds to the body of knowledge about MSS.

Implications for Theory

The TMSC served as a framework for understanding the experiences of stress and coping for parents within the MSS community. Constructs from the TMSC including primary appraisals, secondary appraisals, coping efforts, social support seeking, and information seeking guided this research from the development of the interview guide through to the analysis of the data. The relationships between the three themes are informed by the constructs from the TMSC (Lazarus & Folkman, 1984). Appraisals of stressors inform the coping efforts that a person employs (Glanz et al., 2008). The core theme, *fear of the unknown,* is a major stressor that parents of children with MSS experience. Parents appraise their futures by 1) judging the severity of circumstances (primary), and 2) judging their ability to respond to the given circumstances (secondary). As a result, the parents employ coping strategies such as *information gathering* and relying on *social connections.* These coping themes related to the core stressor theme via the MSS Facebook group. When parents participated with the MSS Facebook group, they engaged in active coping for the *fear of the unknown*, which helped them to adapt.

The results from the current study align with the TMSC and build upon the current understanding of these constructs. For example, social support is an important part of coping in this framework, but the capacity to engage in social support online brings in new elements. In some ways, online social support may be more feasible when communities are limited by geographic dispersion or language differences, like the MSS community. Facebook can connect people across the world and translate text instantly. These technologies present new opportunities for social support that were not possible before time of personal computers and social media.

Implications for Practice

Health providers and genetic counselors should be aware of the psychological stressors related to caring for a child with a rare diagnosis and help connect parents to resources, such as online social support groups. It is important for these providers and counselors to understand the common experiences for families with rare disease while emphasizing the unique value of being connected to others within a disease specific community. Preparing a parent with realistic expectations and equipping them with tools to cope may increase the belief that they are capable of dealing with challenging circumstances. Working to make parents feel more prepared will improve secondary appraisals and decrease the impact of stress on this population.

Additionally, health providers and genetic counselors should be aware of the feelings of fear related to the unknown future for families with rare diseases. Even the resources that are available may be overwhelming or create even more fear for families. Providers and counselors should help equip parents with as much knowledge as possible along with the self-belief that they are capable of overcoming challenges. Disease-specific online social support may be a useful tool for accomplishing these goals. Connecting with other parents to ask questions and learn from the experiential knowledge may provide information that cannot be found in the academic research.

Health providers and genetic counselors should value and promote this way of learning along with information from previous research.

Strengths and Limitations

A key strength of the current study is that it collected qualitative perspectives from a population of parents whose experiences have not been documented. Though the results are consistent with other rare disease populations, each specific group has unique experiences and stories to tell. Lifting up these voices increases the body of information available for parents of children who are newly diagnosed with MSS. Though the number of families may be few, each rare disease population deserves to have their stories documented in the literature. Additionally, these findings support the value of online social support groups for rare disease communities. Online social support groups may have additional value for extremely rare diseases such as MSS because the population is geographically dispersed. Care providers should work to connect families into rare disease-specific support groups upon diagnosis.

Another strength of the current study is that it was conducted by an insider of the MSS community. Being a part of the MSS Facebook group gave the PI access to research an incredibly rare community. Additionally, the PI already had rapport built with this community and invested interest in supporting this population. Participants trusted the PI with their stories and were willing to share openly throughout the interviews. Finally, the PI had opportunities to consult with community members including parents, doctors, and researchers throughout the study. All of these factors contributed to the results of this study that will be given back to the MSS family community.

These results should be interpreted with the following limitations in mind. A small sample size limits the generalizability of the findings. The small sample of the current study lacks variety in sex, race, and ethnicity. The homogenous sample may not represent all of the variety of experiences within the MSS parent community. The experiences of fathers are often missing from the research on parents of children with disabilities, but their perspectives should not be forgotten. The sample also may be limited by selection bias. If the parents who agreed to participate are the most involved in the MSS Facebook group, the results may be missing other perspective from families who do not care as much about being a part of the MSS community. Though this study only recruited through the MSS Facebook group, it would have been valuable to gather perspectives from people who are not on Facebook at all. That sub population may have different experiences as parents and beliefs about online social support. It is important not to over-generalize and to keep in mind that every family has a unique experience. Finally, community members should also be involved in reviewing the results so that they can provide feedback on the interpretation of the data. This step would increase community accountability to produce the strongest results.

Future Directions

Future studies should continue assessing the experience of parents of children with rare diseases through the constructs of the TMSC framework and the conceptual model described above. Additional data from parents of children with MSS will help refine and clarify the current findings. A more diverse sample including various races and ethnicities will articulate more variety and nuance of experiences. Future studies should also investigate the impacts of connecting researchers and doctors into these online communities. If parents are comfortable sharing openly amidst doctors and researchers, this may be a useful way to diffuse information

both directions. However, the presence of doctors and researchers may change the dynamics of these online support groups. Special care should be taken to preserve the safe and open environment to support parents of children with rare diseases.

Conclusions

In conclusion, the experience of parenting a child with MSS is stressful and many parents fear the unknown course of a rare disease. Parents of children with MSS face constant challenges but there are opportunities for these parents to adapt by engaging in coping practices. Finding online community with other MSS families promotes key coping strategies such as information gathering and social connection for parents. The MSS Facebook group is a unique tool to help parents of children with MSS cope with the stressors of rare disease. The MSS Facebook support group gives parents the opportunity to engage in disease-specific community and collective learning about MSS. Innovative disease-specific support groups should be prioritized to connect families with rare diseases and promote health to this population.

References

Baumbusch, J., Mayer, S., & Sloan-Yip, I. (2019). Alone in a Crowd? Parents of Children with Rare Diseases' Experiences of Navigating the Healthcare System. *Journal of Genetic Counseling*, *28*(1), 80–90. https://doi.org/10.1007/s10897-018-0294-9

Cohen, E., Kuo, D. Z., Agrawal, R., Berry, J. G., Bhagat, S. K. M., Simon, T. D., & Srivastava, R. (2011). Children with medical complexity: An emerging population for clinical and research initiatives. *Pediatrics*, *127*(3), 529–538. https://doi.org/10.1542/peds.2010-0910

Cousino, M. K., & Hazen, R. A. (2013). Parenting stress among caregivers of children with chronic illness: a systematic review. *Journal of Pediatric Psychology*, *38*(8), 809–828. https://doi.org/10.1093/jpepsy/jst049

Devonport, T. (2011). A brief review of commonly used theories and the importance of applying theory in practice. In T. J. Devonport (Ed.), *Managing stress: From theory to application.* (pp. 1–12). Retrieved from https://login.proxy.library.emory.edu/login?url=http://search.ebscohost.com/login.aspx?direct=true&db=psyh&AN=2013-06435-001&site=ehost-live&scope=site

Fitch, N. (1980). The syndromes of Marshall and Weaver. *Journal of Medical Genetics*, *17*(3), 174–178.

Glanz, K., Rimer, B. K., & Viswanath, K. (2008). *Health Behavior and Health Education : Theory, Research, and Practice*. Retrieved from http://ebookcentral.proquest.com/lib/emory/detail.action?docID=353367

Goldsmith, J. (2016). From our perspective : Encouraging drug development for rare diseases. Retrieved from United States Food and Drug Administration website: https://www.fda.gov/drugs/news-events-human-drugs/our-perspective-encouraging-drug-development-rare-diseases

Haik, D., Kashanchi, K., Tajran, S., Heilbronn, C., Anderson, C., Francis, D. O., … Verma, S. P. (2019). The Online Support Group as a Community: A Thematic Content Analysis of an Online Support Group for Idiopathic Subglottic Stenosis. *Annals of Otology, Rhinology and Laryngology*, *128*(4), 293–299. https://doi.org/10.1177/0003489418820348

Hoyme, H. E., Byers, P. H., & Guttmacher, A. K. (1993). Marshall-Smith syndrome: further evidence of an osteochondroplasia in long-term survivors. *Proceedings of the Greenwood Genetics Center*, *12*(70), 418–419.

Johnson, J. P., Carey, J. c., Glassy, F. J., Paglieroni, T., & Lipson, M. H. (1983). Marshall-Smith syndrome: Two case reports and a review of pulmonary manifestations. *Pediatrics*, *71*(2), 219–223.

Kauw, D., Repping-Wuts, H., Noordzij, A., Stikkelbroeck, N., Hermus, A., & Faber, M. (2015). The contribution of online peer-to-peer communication among patients with adrenal disease to patient-centered care. *Journal of Medical Internet Research*, *17*(2), 1–7. https://doi.org/10.2196/jmir.3869

Ladd, D. (2015). General and Health Information Challenges of Patients With Rare Diseases: The Importance of Health Information Provision and Web Sites for Locating Rare Disease Resources. *Journal of Hospital Librarianship*, *15*(2), 189–197. https://doi.org/10.1080/15323269.2015.1014762

Lazarus, R. S., & Folkman, S. (1984). *Stress, Appraisal, and Coping* (1 edition). Retrieved from https://books.google.com/books?hl=en&lr=&id=i-ySQQuUpr8C&oi=fnd&pg=PR5&dq=Lazarus,+R.S.,+%26+Folkman,+S.+(1984).+Stress,+Appraisal,+and+Coping.+New+York:+Springer+Publishing+Company.&ots=DfHVnshiSi&sig=N83GXocWJG1Pnv9vb_z6PhsM7Sk#v=onepage&q=Lazarus%2C R.

Malan, V., Rajan, D., Thomas, S., Shaw, A. C., Louis Dit Picard, H., Layet, V., … Cormier-

Daire, V. (2010). Distinct effects of allelic NFIX mutations on nonsense-mediated mRNA decay engender either a sotos-like or a Marshall-Smith Syndrome. *American Journal of Human Genetics*, *87*(2), 189–198. https://doi.org/10.1016/j.ajhg.2010.07.001

Marshall, R. E., Graham, C. B., Scott, C. R., & Smith, D. W. (1971). Syndrome of accelerated skeletal maturation and relative failure to thrive: A newly recognized clinical growth disorder. *The Journal of Pediatrics*, *78*(1), 95–101. https://doi.org/10.1016/S0022-3476(71)80269-X

Meade, O., Buchanan, H., & Coulson, N. (2018). The use of an online support group for neuromuscular disorders: a thematic analysis of message postings. *Disability and Rehabilitation*, *40*(19), 2300–2310. https://doi.org/10.1080/09638288.2017.1334239

MSS Research Foundation. (2016). Standards of Care for Marshall-Smith Syndrome. Retrieved from MSS Research Foundation website: www.marshallsmith.org

MSS Research Foundation. (2018). WHAT IS MSS? Retrieved from MSS Research Foundation website: https://www.marshallsmith.org/en/what-is-mss

National Institutes of Health. (2017). FAQs About Rare Diseases. Retrieved from National Institute of Health website: https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases

Pelentsov, L. J., Fielder, A. L., Laws, T. A., & Esterman, A. J. (2016). The supportive care needs of parents with a child with a rare disease: Results of an online survey. *BMC Family Practice*, *17*(1), 1–13. https://doi.org/10.1186/s12875-016-0488-x

Pelentsov, L. J., Laws, T. A., & Esterman, A. J. (2015). The supportive care needs of parents caring for a child with a rare disease: A scoping review. *Disability and Health Journal*, *8*(4), 475–491. https://doi.org/10.1016/j.dhjo.2015.03.009

Picci, R. L., Oliva, F., Trivelli, F., Carezana, C., Zuffranieri, M., Ostacoli, L., … Lala, R. (2015).

Emotional Burden and Coping Strategies of Parents of Children with Rare Diseases. *Journal of Child and Family Studies*, *24*(2), 514–522. https://doi.org/10.1007/s10826-013-9864-5

Rare Disease Act of 2002, Pub. L. No. 107–280, 116 Stat. 1988 (2002).

Rocha, H. M., Savatt, J. M., Riggs, E. R., Wagner, J. K., Faucett, W. A., & Martin, C. L. (2018). Incorporating Social Media into your Support Tool Box: Points to Consider from Genetics-Based Communities. *Journal of Genetic Counseling*, *27*(2), 470–480. https://doi.org/10.1007/s10897-017-0170-z

Shaw, A. C., Van Balkom, I. D. C., Bauer, M., Cole, T. R. P., Delrue, M. A., Van Haeringen, A., … Hennekam, R. C. M. (2010). Phenotype and natural history in Marshall-Smith syndrome. *American Journal of Medical Genetics, Part A*, *152*(11), 2714–2726. https://doi.org/10.1002/ajmg.a.33709

Sperli, D., Concolino, D., Barbato, C., Strisciuglio, P., & Andria, G. (1993). Long survival of a patient with Marshall-Smith syndrome without respiratory complications. *Journal of Medical Genetics*, *30*, 877–879. https://doi.org/10.1136/jmg.30.10.877

Summers, D. A., Cooper, H. A., & Butler, M. G. (1997). Marshall-Smith syndrome: case report of a newborn male and review of the literature. *Clin Dysmorphol.*, *8*(3), 207–210.

Temi. (2020). Retrieved from https://www.temi.com/

Van Balkom, I. D. C., Shaw, A., Vuijk, P. J., Franssens, M., Hoek, H. W., & Hennekam, R. C. M. (2011). Development and behaviour in Marshall-Smith syndrome: An exploratory study of cognition, phenotype and autism. *Journal of Intellectual Disability Research*, *55*(10), 973–987. https://doi.org/10.1111/j.1365-2788.2011.01451.x

Von der Lippe, C., Diesen, P. S., & Feragen, K. B. (2017). Living with a rare disorder: a systematic review of the qualitative literature. *Molecular Genetics and Genomic Medicine*, *5*(6), 758–773. https://doi.org/10.1002/mgg3.315

Whicher, D., Philbin, S., & Aronson, N. (2018). An overview of the impact of rare disease characteristics on research methodology. *Orphanet Journal of Rare Diseases*, *13*(1), 1–12. https://doi.org/10.1186/s13023-017-0755-5

Williams, D. K., Carlton, D. R., Green, S. H., Pearman, K., & Cole, T. R. P. (1997). Marshall-Smith syndrome: the expanding phenotype. *Journal of Medical Genetics*, *34*, 842–845. https://doi.org/10.1136/jmg.34.10.842

Wilson, B. T., & Hughes, J. (2019). Joining the hidden revolution in rare diseases: working with family support groups. *Archives of Disease in Childhood*, archdischild-2019-317227. https://doi.org/10.1136/archdischild-2019-317227

Woodgate, R. L., Edwards, M., Ripat, J. D., Borton, B., & Rempel, G. (2015). Intense parenting: A qualitative study detailing the experiences of parenting children with complex care needs. *BMC Pediatrics*, *15*(1), 1–15. https://doi.org/10.1186/s12887-015-0514-5

Yoder, C. C., Wiswell, T., Cornish, J. D., Cunningham, B. E., & Crumbaker, D. H. (1988). Marshall Smith Syndrome: Further Delineation. *Southern Medical Journal*, *81*(10), 1297–1300.

Appendix A: Interview Guide

Introduction:

Thank you so much for taking the time to speak with me today! I am collecting these interviews for a study a qualitative research study I am working on with families that have a child with Marshall-Smith Syndrome (MSS). The purpose of my study is to get an inside look into the experiences of stress and coping for parents that care for MSS. I hope to get a glimpse of the creative ways that you manage daily stressors and how your family utilizes the MSS Facebook page. This interview shouldn't take more than 1 hour, and I appreciate all of ideas that you have to offer me. As a reminder, your participation is completely voluntary. You should feel free to skip any questions that you don't feel like discussing for any reason. Your answers will remain fully confidential and will be used strictly for the purpose of this study. A transcript of this interview will be shared within the research team, but by that time your name will not be attached to your answers. Do you mind if I record our conversation, so I don't forget any important details? Do you have any other questions? After getting all of the details, are you still willing to participate in my project?

Part 1: *Demographics & Survey*

1. Which gender do you most identify with?
2. How old are you?
3. What is the highest level of school you have completed or the highest degree you have received?
    a. Response options: (1) less than high school degree, (2) high school degree or equivalent (e.g., GED), (3) some college but no degree, (4) associate degree, (5) bachelors degree, (6) graduate degree
4. Do you work outside of the home?
    a. If yes, do you work fulltime or part time?
5. How old is your child with MSS?
6. How old was your child when they received their MSS diagnosis?
7. Would you consider yourself the primary caretaker of your child?
    a. Who else helps care for your child on a daily basis?

8. How often do you talk or message with someone that you met on through the MSS group?
    a. Response options: (1) Everyday, (2) Once per week, (3) Once per month, (4) A few times per year, (5) Rarely/never
9. How often do you go on the MSS Facebook page?
    a. Response options: (1) Everyday, (2) Once per week, (3) Once per month, (4) A few times per year, (5) Rarely/never
10. How often do you post or respond on the Facebook page?
    a. Response options: (1) Everyday, (2) Once per week, (3) Once per month, (4) A few times per year, (5) Rarely/never
11. How long did it take you to find the Facebook page after your child was diagnosed with MSS?

Part 2: *Daily life – stress, appraisal, and coping*

Intro

1. Can you tell me a little bit of background about your family? And specifically, your child with Marshall-Smith Syndrome.
2. How do you normally explain what Marshall-Smith Syndrome is to others?

The next few questions will help me to understand your personal experiences with caring for a child with Marshall-Smith Syndrome.

3. What was the experience of receiving this diagnosis like for you and your family?
    a. How has life changed after receiving this diagnosis?
4. What was your initial perception of Marshall-Smith Syndrome?
5. What were your initial fears about Marshall-Smith Syndrome?
    a. What were your initial fears about caring for your child?

Now we are going to move on to a few questions about your experience of stress and the ways that you cope with daily stressors.

6. What is stressful in your daily life as a caregiver?
7. What kind of things do you do to manage stress?
    a. Who do you turn to when you need help?
    b. What are some ways that you try to solve problems?
    c. What are some things in your life that you do to care just for yourself?
8. What are some ways that you are socially supported as a caregiver?

Part 3: *Facebook and coping*

The following questions focus on your personal use and preferences of the MSS Facebook support group.

1. Where did you look for more information about Marshall-Smith Syndrome when your child was first diagnosed?
    a. What did you find most helpful?
2. What was your experience like when you first connected to this Facebook page dedicated to Marshall-Smith Syndrome?
    a. What was it like to see photos and hear stories about people with Marshall-Smith Syndrome?
    b. How did you learn about the page?
3. What kind of information is shared in the MSS group?
    a. Do you mostly see posts, photos, videos, or questions?

4. How has this information played a role in your journey with Marshall-Smith Syndrome?
5. Can you tell me some of the ways that you feel supported or encouraged by being a part of this MSS community, if any?
    a. Can you tell me about times when you have not felt supported or felt discouraged by being a part of this community, if any?
6. What does it mean to you to be connected to other people who have experience with Marshall-Smith Syndrome?
    a. In what ways do you feel understood by the other members of this Facebook group?
7. How would you describe the role of trust between community members on this type of online group?
    a. How much personally information are you comfortable sharing on the page?
    b. In what ways do the group members' opinions impact your medical choices?
8. How does this community impact the way that you see yourself and your child?
9. What do you think your life would be like without this connection to others with Marshall-Smith Syndrome?
10. What are some of the important or unique features of the MSS group that differ from other Facebook groups that you have been involved in?
11. What other Facebook groups do you follow?
    a. How do they compare to the MSS group?

Closing: Thank you so much for participating in this study. I don't have any more formal questions, but is there any last thoughts that you would like to include in your interview?

Appendix B: Codebook

Overview of Codes:

1 Challenges/Stressors
  1.1 Physical
  1.2 Emotional
  1.3 Medical
  1.4 Life Expectancy/Death
2 Understanding/Appraisal
  2.1 Personal
  2.2 From Others
  2.3 From Doctors
3 Coping Effort
  3.1 Social Support
  3.2 Information Seeking
  3.3 Getting Away
  3.4 Religious
  3.5 Normal
  3.6 Respond one day at a time
  3.7 Fun
  3.8 Lack of Selfcare
  3.9 Cutting Work Hours
4 Family Background
  4.1 Other Children
5 Description of MSS
  5.1 Similarities
  5.2 Variety
  5.3 Child's Attributes
6 Diagnosis
7 MSS Facebook Group
  7.1 Learning
  7.2 Questions
  7.3 Perceptions
8 Openness/privacy

Code Memos:

1 Challenges/Stressors

Deductive
Description: Any time the participant is talking about something stressful or challenging that they face. Only use if the code does not fit into one of the subcategories.

1.1 Physical

Deductive
Description: Any time the participant mentions a physical challenge related to caring for their child (such as carrying their child or physical disabilities).
Clarifying note: this is sometimes hard to distinguish from medical challenges. In this case, double code.

1.2 Emotional

Deductive
Description: Social, emotional, or psychological stress of caring for their child (such as fear, isolation, etc).

1.3 Medical

Deductive
Description: Challenges working with medical professionals, nursing staff, or medical treatments.

1.4 Life Expectancy/Death

Inductive
Description: Stress of worrying about the life expectancy of their child or hearing about other children dying.
Clarifying note: I often double coded this with emotional stress.

2 Understanding/Appraisal

Deductive
Description: Any time the participant describes perspectives or appraisals of a situation. Typically, they all fall in the subcategories of "personal" (participant's perspective), "from others", "from doctors." Only use this code if it does not fit into the sub codes.

2.1 Personal

Deductive
Description: Any time the participant describes their own personal perspectives or appraisals of a situation.

2.2 From Others

Inductive

Description: Any time the participant describes perspectives or appraisals of a situation from others (friends, family, strangers).

2.3 From Doctors

Deductive
Description: Any time the participant describes perspectives or appraisals of a situation from a doctor.

3 Coping Effort

Deductive
Description: Any time the participant describes something they do that helps them deal with difficulty/stress. Only use this code if it does not fit into one of the subcategories.

3.1 Social Support

Deductive
Description: Any time the participant describes social connections that help them deal with difficulty/stress.

3.2 Information Seeking

Deductive
Description: Any time the participant describes searching for answers or more information to help them deal with difficulty/stress.

3.3 Getting Away

Inductive
Description: Any time the participant describes getting away (going on vacation, spending time alone, etc.) to help them deal with difficulty/stress.

3.4 Religious

Deductive
Description: Any time the participant describes religious practice or community to help them deal with difficulty/stress.

3.5 Normal

Inductive
Description: Any time the participant describes a challenge as "normal" to help them deal with difficulty/stress.
Clarifying note: Saying they are used to it, it is just a day in their normal life, it is "our normal", etc.

3.6 Respond one day at a time

Inductive
Description: Any time the participant describes coping with difficulty/stress by "taking things one day at a time". Or just taking on challenges as they come up.

3.7 Fun
Inductive
Description: Any time the participant describes doing fun or adventurous things to help them deal with difficulty/stress.

3.8 Lack of Selfcare
Inductive
Description: Any time the participant describes self-care or doing something just for them self to help them deal with difficulty/stress.

3.9 Cutting Work Hours
Inductive
Description: Any time the participant describes changing work schedule (or quitting work) to help them deal with difficulty/stress.

4 Family Background
Deductive
Description: Any time the participant describes their family. (In their household or extended family)
Clarifying note: I know background makes it seem like the past, but I also used it to tag current information about their household or extended families.

4.1 Other Children
Inductive
Description: Any time the participant describes their things about their other children or how their other children relate to their child with MSS.

5 Description of MSS
Deductive
Description: Any time the participant describes their understanding or attributes of MSS. Only use if this does not fit into one of the sub codes.
Clarifying note: I sometimes double coded this one with the sub code "child's attributes." Because sometimes the participant talks generally about MSS and uses examples from their own child at the same time.

5.1 Similarities
Inductive, "Like family"
Description: Any time the participant describes similarities between their experiences and other MSS family experiences. Or whenever the participant describes similarities between attributes or the way the MSS children look.

5.2 Variety
Inductive
Description: Any time the participant describes variation and differences across cases of MSS or experiences with MSS.

5.3 Child's Attributes

Inductive
Description: Any time the participant describes attributes of their child with MSS.
Clarifying note: This was sometimes hard to distinguish between the general main code "descriptions of MSS."

6 Diagnosis

Deductive
Description: Any time the participant describes the experience seeking, receiving, or processing the MSS diagnosis.

7 MSS Facebook Group

Deductive
Description: Any time the participant describes the MSS Facebook group. Only use this main code if it does not fit into one of the sub codes.

7.1 Learning

Inductive
Description: Any time the participant describes learning from the MSS Facebook group.
Clarifying note: this was often closely linked to "questions\MSS Facebook group." I often double coded these two.

7.2 Questions

Inductive
Description: Any time the participant describes asking questions on the MSS Facebook group.
Clarifying note: this was often closely linked to "learning\MSS Facebook group." I often double coded these two.

7.3 Perceptions

Deductive
Description: Any time the participant describes their perceptions of the MSS Facebook group.
Clarifying note: this was hard to distinguish from "personal\understanding/appraisal." I usually double coded this.

8 Openness/privacy

Inductive
Description: Any time the participant describes their feelings about openness or privacy online or on the MSS Facebook page.