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The Barriers and Quality of Care Issues for Individuals Diagnosed with 22q11.2 Deletion Syndrome

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Abstract

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Barbara Bavette Miller

M.S., Oklahoma State University, 2006

B.S., Oklahoma Wesleyan University, 2004

Dissertation Submitted in Partial Fulfillment

of the Requirements for the Degree of

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Health Services

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Abstract

Individuals diagnosed with 22q11.2 deletion syndrome (22q) have many barriers and quality of life issues associated with the condition. Without an obvious anomaly, affected individuals may live years without a proper diagnosis. The purpose of this qualitative case study research was to explore perceptions of caregivers of individuals with 22q. Uncertainty in illness theory was used as it describes the inability to find answers with unpredictable medical issues. The central research question focused on at how challenges associated with a 22q diagnosis are addressed. Interviews were conducted with 10 caregivers. Guided by the uncertainty in illness theory, data analysis was conducted by coding through NVivo to find themes. Themes identified included, but were not limited to: (a) age and symptoms at diagnosis; (b) usage of the internet for answers; (c) future transition uncertainties; and (d) lack of provider knowledge. Five out of 10 participants had a child with a known heart condition at birth, which led to a 22q diagnosis. Five out ten individuals with 22q were diagnosed at a later age following a manifestation of other serious conditions. A greater index of suspicion could have led to a more timely diagnosis of 22q. All individuals expressed the desire for a more prompt and thorough diagnosis. The positive implications for social change include influencing physicians and policy makers through education and implemented policies that can lead to more timely diagnoses and treatment for better health outcomes. This social change can influence the target population through scholarly publications in medical journals. It may also be influenced through policy proposals influencing early detection screenings at birth. The addition of caregiver advocates may also bring about change within the 22q community.

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Dedication

I would like to dedicate this dissertation to my husband and best friend, Ryan, who has continuously loved and supported me throughout my education and career aspirations. Since the beginning of our relationship, you have continued to encourage me to keep going in order to fulfill the goals of our family. I love you more than you can possibly imagine. This dissertation is also dedicated to my children, E.J. and Teagan. You two are the reason I strive to be a better person. E.J.—your life is the reason I chose this dissertation topic. I am so happy you are my inspiration for research. Teagan—I started working on this when you were four years old. I am so excited to make you proud of me! I love you both.

To Poppa (my dad). Although aphasia has taken away your ability to brag to everybody you know about your little girl being a doctor, I know you are proud of me. I can hear you telling everybody that they need to call me “Dr”. I love you and thank you for the pride you have always had in me. Mom, Pam, and Mike. Thank you for your amazing trust and support, and for understanding the limited time I have been able to be with my family. I am forever grateful for your love. Finally, Jim. Without you coming into my office three years ago and telling me I needed to go back to school, I would not be here today. You have been a great mentor and colleague over the last several years. Thank you for always believing in me. Thank you to all of my family, friends, and CHS students for playing a significant role in helping me achieve my goal of earning a Ph.D.

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Finally, to all of the caregivers of 22q who participated across the United States, thank you! To the participants in Oklahoma whom I have met on this journey we share, your support and dedication to 22q research is incredible. Thank you all for your trust in me and this research project.

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Chapter 1: Introduction to the Study

Introduction

Meeting the needs of individuals with physical, developmental, and emotional deficiencies due to a rare genetic disorder can be demanding on caregivers (Christian, 2016). Initiating the necessary educational and physical accommodations for these individuals can take a toll on the family. To successfully manage a genetic disorder and the educational and clinical treatments necessary, the individual must be diagnosed properly and promptly. To obtain an appropriate and timely diagnosis, healthcare providers must have a familiarity with and suspicion of clinical presentations that could be the result of something more severe (Friedman, Rienstein, Yeshayahu, Gothelf, & Somech, 2016; Vogels et al., 2014).

Access to appropriate and timely healthcare treatment is necessary for individuals with chronic health conditions associated with genetic disorders. Healthcare providers can only deliver the quality of care demanded when individuals have access to the services needed (Rodrigues, Tatsch Neves, Bigolin Jantsche, & daSilveira, 2016). In order for individuals affected with 22q to have access to required care, healthcare providers must be knowledgeable about common issues associated with different genetic disorders.

Ware and Jeffries (2012) suggested the prevalence of those diagnosed with 22q11.2 deletion syndrome (22q) could be 1:4,000 while Shprintzen (2008) reported a number of findings citing the incidence from 1:2,000 to 1:7,000. However, a possibility of 1:1,600 could be more accurate, due to misdiagnoses of those affected (Shprintzen,

2008). With this prevalence, obtaining a timely and accurate diagnosis is imperative in the facilitation and treatment of this syndrome. In this research, I explored barriers and quality of care issues of individuals with 22q from the caregiver's perspective in order to potentially offer a greater opportunity for better education, leading to a more timely and accurate diagnosis. Barriers included any type of circumstance that impacts the medical, educational, and general wellbeing of an individual. My aim was to better educate the public, healthcare providers, and educational specialists in order to heighten awareness of the syndrome. With this heightened awareness, future implications could include a more timely and accurate diagnosis and treatment, leading to better health outcomes and life transitions.

This chapter presents the background of 22q, including current research and the gap in knowledge presenting the need for additional research. Additionally, I explore the current and relevant research problem and discuss the intent of the study. I present research questions and introduce the appropriate theoretical framework. I provide the methodology and define key terms related to the research. I also explain assumptions, scope, delimitations, and limitations of the study. Lastly, I illustrate the significance of this study's contribution to the discipline.

Background

Clinical features vary with 22q, but there is the possibility of having any number of more than 180 known anomalies (Shprintzen, 2008). The most known clinical feature found in 22q patients is congenital heart disease (McDonald-McGinn & Sullivan, 2011). Other prevailing clinical indicators include palate abnormalities, developmental delays

and disorders, facial dysmorphism, immunodeficiency, and psychiatric disorders (Evers et al., 2014; Furuya, Sasaki, Takeuchi, & Urita, 2015). Developmental deficiencies may be represented throughout all ages in childhood and teenage years. An increased risk of psychiatric disorders, such as schizophrenia, anxiety, depression, and bipolar disorder, manifests later in life (Fabbro, Rizzi, Schneider, Debbane, & Eliez, 2012).

Specialty multidisciplinary clinics are significantly influential in properly and efficiently treating and monitoring a patient diagnosed with 22q (Bassett et. al., 2011). Children or adults with multifaceted demands necessitate effectual partnerships with and cooperation among healthcare providers in order to be provided timely and accurate care (Nicholl & Begley, 2012). The availability of clinics experienced with 22q patients may be geographically variable, leading to a lack of access to care for families (Bassett et al., 2011).

Guidelines for Physicians

Guidelines to medically care for patients with 22q were developed to assist primary care physicians (Bassett et al., 2011). The guidelines were developed through focus groups that included clinicians and researchers, as well as through a thorough scientific-based systematic literature review (Bassett et al., 2011). Experts are central in leading this process, but healthcare providers have to be open-minded and consider individual cases (Nicholl et al., 2014).

Guidelines for Caregivers

Parents can be placed in the unfortunate role of being the expert on their child's needs. This can lead to recapping symptoms and reiterating needs through multiple visits

and to numerous professionals (Nicholl & Begley, 2012). An information leaflet was developed for parents to use when communicating with a diverse group of education and healthcare providers (Nicholl et al., 2014). In the study conducted to create the leaflet, information important to caregivers related to communication and addressing significant issues of 22q was included (Nicholl et al., 2014). Three wide-ranging topics were recognized in the qualitative data derived from the parents' feedback. This included the individuality of the diagnosed, general information related to the syndrome, and the frustration and experience in seeking medical care (Nicholl et al., 2014). This study concluded that the information presented was relevant to the support group researched and could be relevant if studied with a different population. The group resided in Ireland so this study may not have represented other cultures, communities, or countries (Nicholl et al., 2014).

Guidelines for Clinical Management

Researchers suggested that guidelines were necessary for publicly funded community and health services entities in the United Kingdom. Guidelines for the management of 22q were created to promote good practices within these health services entities (Habel et al., 2014). Parents, therapists, and clinicians participated in the study (Habel et al., 2014). This research consisted of an all-encompassing standard of care plan for those affected by 22q. (Habel et al., 2014).

Effort in Addressing the Issue

Although the research shows great strides in attempting to address the lack of knowledge and information available to patients, caregivers, education specialists, and

health providers, someone has to possess the knowledge or suspicion to initiate action. Medical treatment and educational accommodations are most important in addressing the issue. The cost of treating individual symptoms and not focusing on the overall condition is financially draining.

Without a proper diagnosis, a later onset of relevant treatment may lead to unknown and more challenging chronic medical conditions, including transmission through reproduction, for the patient and possibly children born to the individual (Fung et al., 2015; McDonald-McGinn & Sullivan, 2011). Untreated chronic medical conditions may lead to a lower life expectancy (Bassett et al., 2011). A timely and accurate diagnosis may provide significant value to diagnosed individuals, their caregivers, treating physicians, and education professionals (Costain, Chow, Ray, & Bassett, 2012). The most common determinant of 22q is through genetic testing, which may be justified if a high index of suspicion is present, for proper and timely medical treatment (McDonald-McGinn & Zackai, 2008; Vogels et al., 2014). This study potentially contributes to researcher and physician education by explaining some barriers to diagnosis and health care management of those diagnosed with 22q as currently perceived by caregivers.

Problem Statement

Once 22q is considered a possibility, the diagnosis process is straightforward (McDonald-McGinn & Sullivan, 2011). Unfortunately, many syndromes have likenesses and can overlap without a high index of suspicion that would lead to appropriate testing (McDonald-McGinn & Sullivan, 2011; Shprintzen, 2008). A precise genetic diagnosis is

crucial due to the numerous and various symptoms and possible inheritance patterns that may be at hand (McDonald-McGinn & Sullivan, 2011). Distinction between 22q and other conditions is important because diagnostic and treatment implications may be substantial (Shprintzen, 2008). A multidisciplinary attitude in identifying and treating the potential irregularities that any one individual could have is vital (Bassett et al., 2011; Costain et al., 2012; Nicholl et al., 2014).

Recent research examined the lived experiences of parenting a child under the age of three with 22q (Goodwin, McCormack, & Campbell, 2017a). Additional research explored the positive and negative experiences of parenting an adult child with 22q (Goodwin, McCormack, & Campbell, 2017b). This research suggested future research into specific outcomes and resolutions be explored. Past studies suggested additional qualitative research needs to be conducted in order to better focus on patient perspectives through interviewing patients and caregivers (Costain et al., 2012). Other researchers proposed the exploratory experience in finding a diagnosis may be distressing for adult patients, parents, and caregivers, meriting additional research to help address this need (Faux, Schoch, Eubanks, Hooper, & Shashi, 2012; Goodwin et al., 2015). The research in this study filled the gap in understanding the caregivers' perceptions and perspectives in the diagnosis and treatment process that could positively affect a more timely and accurate diagnosis of 22q. This information should further provide education to healthcare providers in order to help them better understand, diagnose, treat, and support those diagnosed with 22q in a more timely fashion.

Purpose of the Study

The purpose of this qualitative case study approach was to understand the barriers to quality of care for individuals diagnosed with 22q in order to possibly influence a more timely and accurate diagnosis. Understanding these issues from the perspective of a caregiver is imperative. This case study inquired by means of interviews and documentation of field notes in order to better identify the opinions of these caregivers. This understanding will lead to further education opportunities for affected individuals, their caregivers, healthcare providers, and education specialists, which may lead to diagnoses that are more accurate.

Research Questions

The central research question for this study was as follows:

CRQ1: How are the challenges associated with a 22q diagnosis addressed?

The subquestions for this study were:

RSQ1: How was a suspicion of 22q initially considered?

RSQ2: How did caregivers cope with the news once a diagnosis was made?

RSQ3: How do caregivers perceive the patients coped with the diagnosis?

RSQ4: How have caregivers addressed the barriers for patients diagnosed with 22q?

RSQ5: How do caregivers plan to address these barriers and quality of care issues in the future?

RSQ6: How did healthcare providers' education and experiences impact diagnosis and treatment of 22q?

RSQ7: Why is 22q often under- or misdiagnosed despite being relatively common?

RSQ8: How can increased education of healthcare providers and the public help better identify and address these barriers related to this common yet sometimes unidentified disorder?

Theoretical Framework

Mishel's (1981) reconceptualization of the uncertainty in illness theory was used in this study. This construct resides within a theoretical model of uncertainty evaluation, coping, and adaptation (Mast, 1995). This theory explains the inability to find a meaning in or answers of an illness due to uncertainty in ambiguous and unpredictable symptoms, treatments, explanations, information, and feedback (Mishel, 1981). Recent research used this theory in a similar study (Goodwin et al., 2017a). The diagnosis and treatment of 22q is unpredictable and often multifaceted. This theory directly relates to the issue being studied.

Nature of the Study

The nature of this study was a qualitative approach. Qualitative research involves an inductive approach with the purpose of creating or supporting theory and meaning through analyzing individuals' understandings and interpretations (Gog, 2015). Qualitative research was consistent with the goal of understanding the perceptions of caregivers of individuals diagnosed with 22q, which was the primary focus of this dissertation. I used a case study approach for this study.

A case study is defined by Yin (2014) as “an empirical inquiry that investigates a contemporary phenomenon (the ‘case’) in depth and within its real-world context, especially when the boundaries between phenomenon and context may not be clearly evident” (p.16). This suggests the goal of case study research is to see subjects’ interaction with the environment and the influence that environment brings (Göttfert, 2015). Case studies deal with unique and characteristically specific circumstances (Gog, 2015).

Case studies justify the use of a specific and particular population in order to study the goals of the case (Maxwell, 2013). Case studies are used to contribute to knowledge of a specific individual, group, or phenomenon (Yin, 2014). The goal is not to generalize but to offer an accurate representation of the case (Maxwell, 2013). Case studies allow a deep and complex understanding of a social phenomenon in the natural environment with a real-world perspective (Yin, 2014).

I collected data from caregivers of individuals diagnosed with 22q. Data was collected through interviews, documents, artifacts, and documentation of field notes. I analyzed the data through NVivo software, focusing on categorization and coding that led to relevant themes.

Definitions

22q deletion syndrome (22q): A disorder caused by the deletion of a small piece of chromosome 22. This specific deletion occurs near the middle of the chromosome at a location designated as q11.2 (National Institutes of Health, 2017).

Barrier: An obstacle, obstruction, or circumstance that prevents individuals from living their lives to the fullest. Barriers in this research may include medical, educational, or basic quality of life influences that impact the wellbeing of an individual.

Caregiver: Any family member who regularly provides direct care to an individual, specifically assisting with day-to-day activities.

Clinical presentation: The typical signs and symptoms associated with a medical condition.

Emotional deficiencies: A significant limitation on a person's emotional functioning that may impair or limit the individual's quality of life.

Index of suspicion: Having an awareness or concern that a potentially underlying illness may be present.

Mental deficiencies: A significant limitation on a person's mental functioning that may impair or limit the individual's quality of life.

Physical deficiencies: A significant limitation on a person's physical functioning that may impair or limit the individual's quality of life.

Assumptions

The assumption prior to beginning this research was that there is limited knowledge and index of suspicion of 22q among healthcare providers. This can lead to confusion, misguidance, fear, and anxiety for the patient and caregiver. The extent to which those with 22q access appropriate, timely, and relevant care may be directly linked to a timely diagnosis and appropriate education. It is expected that through applied research, the lack of education can be solved through knowledge communicated and

understood (Patton, 2002). This assumption was necessary in understanding the barriers and quality of care issues for those diagnosed with 22q.

Scope and Delimitations

Understanding the barriers and quality of care issues for persons diagnosed with 22q allows a focus to be placed on areas of educational need. The population in this study included English-speaking caregivers who were 18 and older, male and female, who cared for anybody at any age diagnosed with 22q. Non-English speaking individuals were not included due to the small size of this study. This population provided the appropriate sample to assess the multiple experiences that led to a diagnosis. This information can be used as an educational opportunity by healthcare providers and families to assist with proper and timely diagnosis in future cases. Theoretical frameworks that were considered and not used included the health belief model, cognitive appraisal theory, and extended parallel process model. While these were all considered, recent research conducted used the uncertainty in illness theory. This theory proved to be a better fit for the study. This study had high transferability as the need is significant and the design would work for additional 22q caregivers or caregivers to those with other physical and mental disabilities.

Limitations

This research study was a qualitative case study. This study was intentionally set up as a case study of caregivers of individuals diagnosed with 22q, specifically utilizing interviews to obtain data. I used purposeful sampling to specifically target a population that has experienced the particular phenomenon. A limitation of this study was that the

results were derived from a small group of 10 interviewees. Due to geographical differences, this study may not fully represent the perspectives of all caregivers or those diagnosed with 22q. As a caregiver, I have personal experience with this phenomenon. This could create a bias in data collection and assessment. However, this proved to be a benefit, as the participants felt comfortable and better understood. Data collection procedures negated the possibility of researcher bias in this study. This is discussed later in Chapter 3. Due to the specific nature of the study and small sample size, this study may not be generalizable. However, replication could allow researchers in other geographical areas to address this limitation.

Significance of the Study

Practical implications for this study include a more strategic and purposeful management of chronic medical conditions by physicians, patients, and caregivers. This can be attained through increased knowledge with a higher index of suspicion of 22q. Early detection can only arise from healthcare providers being educated on the prevalent clinical presentations of this syndrome, increasing the index of suspicion. This study empowers future caregivers with the ability to better understand the importance of the diagnosis and treatment plan from their healthcare provider.

Implications for Social Change

The long-term study implications are important for caregivers in knowing the best treatment options, best institutions for primary and secondary education, and most important and appropriate life skills for their loved ones affected by 22q. The benefit for those around the globe is that this small qualitative case study offers a template that can

be replicated; reaching other diagnosed individuals, their caregivers, and healthcare providers. This study shares the experiences of those closest to the patient affected by 22q. It communicates their frustrations, concerns, fears, and emotions in obtaining a diagnosis and creating and maintaining an on-going treatment plan. Enhancing awareness through appropriate research, scholarly publication, and physician education will lead to better clinical outcomes through a proper diagnosis.

Summary

Despite being the second most common genetic disorder, 22q underdiagnosed (Bassett et al., 2011; Shprintzen, 2008). The anomalies are many with varied manifestations. I conducted this qualitative case study research to explore and understand the perspectives and perceptions of caregivers of those diagnosed with 22q. The barriers and quality of life issues associated with the disorder were considered and future recommendations explored. As the most common of all genetic deletion syndromes, more education and awareness is needed in order to increase timely and accurate diagnoses. With a prevalence that is generally estimated at 1:4,000 but could be as high as 1:1,600 (Shprintzen, 2008), research is necessary to increase awareness and the index of suspicion.

In Chapter 1, I presented an overview of the study and the significance of the problem being addressed. The chapter consisted of the history of 22q and the many clinical manifestations that can be found in someone who has 22q. I described the clinical issues that may arise without a proper diagnosis. Chapter 2 presents a review of the

current literature on 22q, caregivers' perspectives of living with genetic disorders, and implications for future research.

Chapter 2: Literature Review

Introduction

With the association of many chronic medical conditions with 22q unsuspected, some individuals have a late or no diagnosis. This can cause serious problems with addressing chronic health issues. This may also leave serious medical conditions untreated due to an unknown association. The purpose of this qualitative multiple case study research was to understand barriers and quality of life issues related to the diagnosis and medical treatment of individuals diagnosed with 22q.

A comprehensive and iterative process was used in conducting a health services literature search that identified a need for further exploration to understand a caregiver's perspective of perceived barriers and quality of care issues for individuals diagnosed with 22q. Multiple studies have identified the importance of early diagnosis, which begins with healthcare providers having a high index of suspicion (Bajaj, Thombare, Tullu, & Agrawal, 2016). Minimal research has been conducted on perspectives on the importance of a 22q diagnosis. One study found was a mixed methods approach conducted in 2012 in Canada, which studied caregiver and adult patient perspectives through a 28-item survey (Costain et al., 2012). Two additional recent studies focused on perspectives of caregivers of children under three and adult children (Goodwin et al., 2017a; Goodwin et al., 2017b).

In the literature review I expound on the search criteria in health services, education, and psychology subjects for 22q; the conceptual framework, uncertainty in illness theory; and methodology to support a qualitative case study. The search criteria

were narrowed to explore clinical anomalies, genetic disorder diagnosis processes, and caregiver perspectives.

Literature Search Strategy

The inquiry specifically focused on peer-reviewed journals. There was minimal research published on 22q compared to other genetic disorders. Many searches were done looking generically at genetic disorders and caregivers. The Walden University databases accessed included CINAHL, MEDLINE, ERIC, Science Direct, SAGE Premier, and Thoreau Multi-Database Search. Search terms included *22q, DiGeorge, VCFS, caregiver, genetic disorders, healthcare barriers,* and *education*. Current research studies including 195 specific to 22q, which provided sound science and a variety of findings, were selected for review. Additional searches were used for appropriate methodology, framework, and research traditions. Journals identified as appropriate and relevant to this study were used in this research.

Theoretical Framework

Malterud (2016) proposed that qualitative studies need stronger theoretical awareness and commitment than what is currently recognized, which includes more than just summarizing the data. Maxwell (2013) described theory as a simple set of concepts and ideas and the proposed connection or relationship among these. Malterud (2016) found that health scholars see theory as a way to solve problems in the world while academic scientists see problems as an opportunity for theorizing.

Uncertainty in Illness

The theory of uncertainty in illness was used in this study. Caregivers who have children with unknown or unsuspected illnesses, especially without a timely diagnosis, have an elevated amount of stress and anxiety in dealing with medical conditions. The blend of the perceived severity and susceptibility associated with loss of health status and fear and anxiety increase risk perception (So, 2013). Individuals affected by 22q often have severe to sometimes fatal chronic conditions that are only perceived or apparent with diagnosis. This may present as something parents have little to no control over, leading to the inability to determine the meaning of these illness-related events (Mishel, 1981).

Research has found that emotion and problem-focused coping strategies are often used in uncertain situations but do not associate with health-related quality of life issues (Padilla, Mishel, & Grant, 1992). Uncertainty in illness theory was also used in research conducted by Goodwin et al. (2017a). Individuals and their caregivers desire to properly evaluate and take action with their health care as appropriate. Without a proper diagnosis, this cannot occur. This research was conducted to better understand perceptions of a previous diagnosis and quality of medical treatment. With this understanding, greater knowledge will be shared with healthcare providers and the public.

Literature Review Related to Key Concepts

Qualitative Research Approach

The nature of this case study was a qualitative approach. The qualitative approach is used to explore and understand the beliefs individuals or groups possess in certain

experiences or situations (Creswell, 2014; Maxwell, 2013). Qualitative research allows those with personal lived experiences the opportunity to share, bond, and come to a general understanding through similar or shared experiences (Cleary, Escott, Horsfall, Walter, & Jackson, 2014). Cleary et al. (2014) contended that a qualitative approach allows the views of participants to be communicated via a sensitive and nonjudgmental technique. This qualitative research was specifically designed to understand and capture the perceptions of caregivers of individuals diagnosed with 22q.

Qualitative research allows the individuals directly affected to communicate the how and why related to their experiences in their own voices (Carroll & Rothe, 2010). Qualitative research approaches are vastly beneficial in gathering a range of responses, based on feedback from participants that may support or develop a specific theory and specify potential medical needs (Alderfer & Sood, 2016). The goal of qualitative research is to rely as heavily as possible on the findings of participants' views of a situation (Creswell, 2013). In this research I sought to find the barriers and quality of care issues for individuals diagnosed with 22q from the perspective of their caregivers. Finally, the most recent research in this discipline used a qualitative approach, which provided a true understanding of caregivers' perspectives of individuals with 22q.

Case Study

This qualitative study used a case study approach. Case studies are used to contribute to knowledge of a specific individual, group or phenomenon (Yin, 2014). Case studies are influential in increasing medical knowledge and sharing clinical findings (Kienle, 2012). They can be a significant communication device for nonspecialists by

raising awareness, providing insight, offering suggestions, and providing solutions (Yin, 2014). They provide timely and pertinent information that may lead to improved patient outcomes and advances in research (Cohen, 2006). Case studies allow a deep and complex understanding of a social phenomenon in the natural environment with a real-world perspective (Yin, 2014). A case study approach allows participants to share experiences in an interview format. In other Walden dissertations, this process proved to be comprehensive for gaining an in-depth knowledge that can come from ideas being shared and communicated in a group setting, as well as perspectives being shared that may not otherwise be communicated one-on-one. Using this data collection process along with field notes, documents, and artifacts provided deep and rich information about the perceptions and experiences of caregivers for those affected with 22q.

Prevalence of 22q

22q is currently known to be prevalent in approximately 1:3,000 to 1:6,000 births (McDonald-McGinn et al., 2015; Shrprintzen, 2008; Ware & Jeffries, 2012) but may be as common as 1:1,600 (Shrprintzen, 2008). This is the most common and frequent occurring microdeletion syndrome (Bajaj et al., 2016). Approximately 5%-15% of those diagnosed with 22q have inherited it from a parent (Wu et al., 2013). Due to 22q being a developmental disorder with clinical findings sometimes not apparent until later in life, birth records data collection would likely underestimate the prevalence of 22q in the population (Pretto, Maar, Yrigollen, Regan, & Tassone, 2015).

Importance of Diagnosis

Increased survival into adulthood of individuals diagnosed with 22q has been found among much of the recent research conducted (Fung et al., 2015). Adults with moderate to severe intellectual disabilities can be diagnosed at an older age and tend to have less 22q typical craniofacial features (Vogels et al., 2014). The concern of later-onset chronic medical conditions and the importance of addressing these issues are prevalent. Due to varied and numerous manifestations, 22q is most likely under recognized (Bajaj et al., 2016; Pretto et al., 2015). There is an increased need of suspicion on behalf of general practitioners in order to address those being misdiagnosed or not diagnosed (Bajaj et al., 2016; Costain et al., 2012; Vogels et al., 2014).

The blend of the perceived severity and susceptibility associated with loss of health status and fear and anxiety related to that loss of status may form a risk perception that could lead to an extended amount of stress and perceived threat (So, 2013). Extreme stress and anxiety can negatively impact health (Drew et al., 2016). A diagnosis can provide relief for caregivers who have felt helpless (Goodwin et al., 2015). Patients and caregivers who know the risks and conditions of this disorder tend to seek treatment and become more efficient in their healthcare treatment.

Medical Anomalies

Congenital heart disease is the most familiar anomaly discovered in patients with 22q. Heart disease appears in approximately 77% of 22q cases (McDonald-McGinn & Sullivan, 2011). Additional clinical features may include a compromised immune system, facial malformations, palate insufficiencies, psychiatric conditions, and significant

developmental delays (Evers et al., 2014; Furuya et al., 2015; Mariano, Tang, Kurtz, & Kates, 2015). More than 180 clinical anomalies could be present in 22q, but no single diagnosed individual has all anomalies and no individual's clinical features are alike (Robin & Shprintzen, 2005; Shprintzen, 2008).

Infancy and Childhood Diagnosis

For newborns, congenital cardiac defects associated with neonatal hypocalcemia are the most frequent clinical features leading to a 22q diagnosis in the first months of life, suggesting that newborns with typical cardiac defects associated with facial features should be examined for 22q (Cancrini et al., 2014). In 29% of patients without classic 22q presentation, developmental delay, learning difficulties, minor cardiac issues, and facial features led to a diagnosis later in life (Cancrini et al., 2014). In childhood years, learning disabilities and feeding difficulties may manifest (Furuya et al., 2015). Speech delays and facial dysmorphisms become more apparent during childhood (Friedman et al., 2016; Wu et al., 2013). Severe hypocalcemia leading to seizures has been present at older ages, without seizures surfacing in infancy or early childhood (Friedman et al., 2016).

Educational Concerns

One of the earliest noticeable 22q anomalies is difficulty with speech and language, with many children requiring therapy for multiple years (Reilly & Stedman, 2013). Academic difficulties may become more noticeable in upper primary years (3rd through 5th grades) as cognitive skills, such as working memory and more abstract areas, specifically in mathematics and literature, become the focus (Cutler-Landsman, 2007;

Reilly & Stedman, 2013; Wong, Riggins, Harvey, Cabaral, & Simon, 2014). While these issues may be present, education in the mainstream classroom is common with necessary additional adult classroom support.(Cutler-Landsman, 2007).

Postchildhood Diagnosis

Individuals with 22q who live past childhood are at an increased risk of premature death when compared to their unaffected siblings (Bassett et al., 2009). In order to properly diagnose adult patients, physicians should have a high indication of suspicion in the presented clinical features, as diagnosis could be difficult (Friedman et al., 2016; Vogels et al., 2014). Research conducted presented multiple clinical presentations, including convulsions, palate insufficiency, developmental delay, and psychiatric issues that may be more dominant in later years (Friedman et al., 2016). The 22q associated premature adult deaths are likely multifactorial, including cardiac and noncardiac associations (Bassett et al., 2009). Surprisingly, the most common premature death in one study was found to be unexplained sudden death (Bassett et al., 2009).

Social and Cognitive Concerns

Difficulties from 22q that may affect any age include social cognition and executive functioning (Wray, Shashi, Schoch, Curtiss, & Hooper, 2013). Social engagement between mother and affected 22q child showed low interaction and engagement on behalf of the child, compared to other groups studied (Weisman et al., 2015). Campbell, McCabe, Melville, Strutt, and Schall (2015) concluded a general dysfunction in 22q included the executive ability to understand cause and effect, to logically reason about social scenarios, specifically misunderstanding and responding

inaccurately to social cues. Social problems may be associated with speech difficulties, anxiety, and/or bullying (Shashi et al., 2012) but may also be caused by social deficiencies. Significant difficulties in multi-tasking and real-world functioning were observed in adolescents, which are related to adaptive functioning (Schneider et al., 2016). The family social environment and the approaches of parenting have been shown to be related to functional outcomes of children diagnosed with 22q (Allen et al., 2014).

Future Medical Concerns

Diagnosis at any age is important in order to provide a unifying explanation for a complex history, obtaining more in-depth medical care, and for future personal healthcare forecasting (Costain et al., 2012). Screening for 22q has recently been suggested in individuals with Parkinson's disease and a history of congenital palatal or cardiac defects, developmental delay, psychiatric disorders, immune deficiencies, or endocrine dysfunction (Pollard, Hannan, Tanabe, & Berman, 2016). In this example, not having a diagnosis of 22q when the individual is affected could have led to additional medical complications. Diagnosis can better help manage the multi-symptom 22q condition while offering the opportunity for a prenatal diagnosis in additional pregnancies (Bajaj et al., 2016).

Psychiatric concerns. Psychiatric issues are a significant concern for those diagnosed and not yet diagnosed with 22q, specifically the prevalence of intellectual and behavior deficits, along with the risk of schizophrenia (Mariano et al., 2015). These intellectual and behavior deficits include Autism Spectrum Disorder (ASD) (14%-50%), attention-deficit/hyperactivity disorder (ADHD) (3%-46%), specific and social phobias

(23%-61%), generalized anxiety disorder (17%-29%), separation anxiety disorder (16%-21%), oppositional defiant disorder (16%-21%), obsessive-compulsive disorder (OCD) (4%-33%), major depressive disorder and dysthymia (10%-20%) and bipolar disorder (0-64%) (McDonald-McGinn & Sullivan, 2011). Due to cognitive deficiencies, patients with 22q may have difficulties reflecting on their own behavior and feelings (Klaassen et al., 2015).

Up to 30% of patients with 22q develop psychotic disorders, such as schizophrenia (Bassett et al., 2011). Evers et al. (2014) found that those who have a moderate to severe intellectual disabilities are more likely to develop psychiatric issues than those with a higher IQ while Niarchou et al. (2014) found no association between cognitive impairment and psychopathology. Individuals with 22q have 30 times increased risk of developing psychosis compared to typically developing individuals with other neurodevelopmental disabilities, at a 10 times risk of those without neurodevelopmental disabilities (Schneider et al., 2014). Clinicians must often rely on parents and educators to communicate the affected individual's behavior (Klaassen et al., 2015). Negative subthreshold psychotic symptoms should be evaluated and monitored carefully in individuals diagnosed with 22q (Mekori-Domachevsky et al., 2016).

Parent-Child Communication

While knowing about a genetic disorder can significantly decrease stress and anxiety while increasing health outcomes, the decision of the caregiver to communicate the diagnosis may exhibit an additional amount of stress (Goodwin et al., 2015). Individuals diagnosed with 22q have borderline intellectual functioning but the capacity

to understand the implications of their disorder (Faux et al., 2012). While getting a diagnosis may provide relief and explanation, receiving a diagnosis may also cause distress (Goodwin et al., 2015).

Life Transition

Individuals diagnosed with 22q struggle with transition from childhood to adulthood. Children's ongoing health issues interfere with normal life, leading some parents to focus on safety and basic needs while others focus on planning for the future (Rossetti, Lehr, Lederer, Pelerin, & Huang, 2015). Caregivers' needs are complex and varied, depending on the needs of the diagnosed individual (Wereiamson & Perkins, 2014). Understanding caregivers' experiences can identify barriers and issues to timely and quality care while supporting the improvement of services (Jones et al., 2016).

Lived Experiences of Parents with Children Diagnosed with 22q

The only research found to date that addresses the specific issue of exploring experiences and perceptions of caregivers of individuals affected with 22q was two studies recently published in 2017. In the first study, participants were caregivers of children under three years old with 22q. Goodwin et al. (2017a) presented the ongoing uncertainty that was experienced by caregivers of young children diagnosed with 22q. This research suggested future research into the experiences and families throughout a child's life including ongoing chronic medical conditions and associated features that may be present at different stages of development. Recent research conducted also focused on the qualitative nature of the lived experiences of parenting an adult child with 22q (Goodwin et al., 2017b). These two studies found the many cumulative stressors of

parents of individuals diagnosed with 22q. Those caregivers of children under the age of three expressed an uncertainty that presents grief, wonder, stigma, pain, and coming to terms with the diagnosis (Goodwin et al, 2017a). For caregivers of adults, the concern was a progression of lost friendships for adult children, stigma, life redefinition, and a general lack of awareness on behalf of healthcare and educational professionals (Goodwin et al., 2017b).

Caregiver Perspectives in Diagnosis

Caregiver perspectives in previous research suggested that a diagnosis experience was negative, due to low-quality information and poor understanding from the healthcare professional, leading to extreme stress and worry (Goodwin et al., 2015). The importance of a diagnosis, including better health outcomes, must be acknowledged (Costain et al., 2012). Healthcare providers must also understand how the needs of these children are met when they are home and assist parents to meet the changing demands and often-unknown trajectory (Nicholl & Begley, 2012). Healthcare providers must meet the informational and emotional needs of the caregivers at diagnosis, as well as throughout the stages of the syndrome (Goodwin et al., 2015).

Characteristics to Consider

The clinical findings of 22q may overlap with other syndromes. A clinical diagnosis of 22q depends on the experience of clinicians, which can be biased or inaccurate (Wu et al., 2013). An accurate diagnosis is imperative for treatment (McDonald-McGinn & Sullivan, 2011). Some characteristics not present at birth may

have a delayed onset, which supports that an early suspicion and diagnosis can lead to better health outcomes and prognosis (Looman, Thurmes, & O'Conner-Von, 2010).

Clinical focus in 22q patients changes and requires constant evaluation, consideration, and adjustments. Early diagnosis is imperative for appropriate intervention and multi-disciplinary care (Cancrini et al., 2014). By accurately diagnosing, physicians can offer appropriate treatment to the disorders, clinical conditions, and behavioral problems that were previously untreated or mistreated. Caregivers are currently given a better explanation of current and future difficulties, which were unexplained in the past. Patients with 22q need long-term treatment and follow-up by a multidisciplinary team with care tailored to their specific clinical findings (Friedman et al., 2016; Fullman & Boyer, 2012). Diagnosing patients over 10 years old with 22q is common and should raise suspicion and awareness among physicians to the late presentation and delayed diagnosis (Cancrini et al., 2014; Friedman et al., 2016).

Adult Guidelines for Managing 22q

Fung et al. (2015) suggested practical strategies for recognizing, evaluating, observing, and managing the illnesses associated with 22q. Evidence-based guidelines and protocols are imperative for individuals managing multiple chronic health conditions with complex needs (Habel et al., 2014). Areas of concern have been described by expert clinicians and researchers who are highly involved in the management of 22q conditions prominent in adults (Fung et al., 2015). Adults may have social complications and employment difficulties (Habel et al., 2014).

Habel et al. (2014) created guidelines in the United Kingdom for the management of 22q, specifically focused on promoting good practice across publicly funded community and health services through a multidisciplinary team approach. Fung et al. (2015) created the first set of guidelines specifically for adults to manage areas of morbidity found within 22q. These guidelines focused on managing cardiovascular, psychosocial, endocrine, neuropsychiatric, genetic counseling, reproductive, and other adult issues that may be presented with a 22q diagnosis (Fung et al., 2015). Fung et al.'s research suggests the need to assess the knowledge provided to caregivers and patients at diagnosis, which were addressed through this qualitative research.

Need for Suspicion

Despite 22q being the most common genetic deletion disorder, diagnosis is often delayed as the level of knowledge and experience with the syndrome is low among healthcare providers (Goodwin et al., 2015). Just over half of parents studied reported they perceived an apparent lack of knowledge and experience among professionals about the characteristics of 22q and how these characteristics affected their child (Nicholl et al., 2014). Bajaj et al. (2016) described the necessity of a high index of suspicion to diagnose 22q. This syndrome is the most common microdeletion syndrome. Recognizing the changes of predominate clinical features with different ages and stages in life is important in identifying 22q (Friedman et al., 2016; Fuyura et al., 2015). Prevalence may be underestimated, due to missed associations.

Facial characteristics are significant in many individuals diagnosed with 22q. Out of post childhood individuals assessed, 88% had characteristic facial features of patients

diagnosed with 22q (Friedman et al., 2016). These features include up slanting palpebral fissures, hooded eyelids, low set posteriorly rotated ears, widened area below nasal bridge, ocular hypertelorism, bulbous nose tip, high arched palate, tapered fingers, micrognatia, short filtrum, and bifid uvala. Out of a group of 55 suspected 22q patients, 43 patients were positively diagnosed with the disorder (Wu et al., 2013). Out of the 43 diagnosed, 100% displayed characteristic face and palate abnormalities, indicating that these features may strongly associate with 22q (Wu et al., 2013). In the same 43 patients, 86% were found to have cognitive/behavioral disorders while 53.5% had immune deficiencies while only 23.3% had a congenital heart disease (Wu et al., 2013). Although heart disease is a dominate anomaly, those with a heart condition are typically diagnosed at an early age. As patients' age without a diagnosis, the presenting features of cellular immunodeficiency, intestinal malrotation, and hypocalcaemia can lead to severe illness (Friedman et al., 2016; Furuya et al., 2015).

Vieira et al. (2015) examined clinical features of patients diagnosed with palate abnormalities and those with 22q. The frequency of selected clinical features including speech delay, learning disabilities, hooded eyelids, hearing loss, and facial dysmorphisms along with a palate abnormality showed a statistical association (35%) with 22q (Vieira et al., 2015). Recognition of common features would help during the diagnostic process. Increasing pediatricians and specialists' knowledge of these features could contribute to an earlier identification of 22q (Cancrini et al., 2014).

Testing

Currently, the proposed diagnostic criteria for 22q are based on reduced number of CD3+ T cells (Friedman et al., 2016). This is appropriate for full 22q but the majority of patients have partial 22q. Partial 22q results in various immune functions, which does not allow for this diagnostic criterion. Due to variable clinical features and presentations, the lack of a diagnostic criterion for partial 22q creates a challenge, specifically in post-childhood patients (Friedman et al., 2016). Traditional testing for 22q, fluorescent in situ hybridization (FISH), requires special equipment, is labor-intensive, and is extremely expensive (Pretto et al., 2015). This expense, expertise needed, and time commitment leads to fewer individuals being screened. Researchers found that droplet digital PCR (ddPCR) blood spot card testing is approximately $5-6 per reaction, with 100% accuracy when sufficient DNA concentrations were used (Hwang et al., 2014; Pretto et al., 2015). This procedure enables large-scale population screens in order to precisely determine the prevalence of 22q. A true incidence rate will only be found through screening the population, based on clinical presentation (Pretto et al., 2015).

Parent Participation in Healthcare Process

Parents are often placed in the role of informing and educating the health professional about their child's specific needs (Nicholl et al., 2014). Parents must identify the individuality of their child's signs and symptoms while recognizing the need for a unique approach to the management and care of the child's health (Nicholl et al., 2014). This has the potential to increase suspension on behalf of the medical provider.

Summary and Conclusions

This chapter reviewed the scholarly literature that substantiates the critical need for continued research and practical implications for those diagnosed, misdiagnosed, or not yet diagnosed with 22q. The needs of individuals diagnosed change throughout ages and stages in life. The literature provided results through multiple findings suggesting a greater necessity of a higher level of suspense on behalf of medical providers.

The primary theoretical framework guiding the study was uncertainty in illness theory. This theory suggested that an elevated level of stress is apparent, which is specifically true with the lack of or misunderstood diagnosis. The goal of using this theory was to better understand the uncertainty, stress, and anxiety that are associated in dealing with medical conditions. This includes, specifically, the time at diagnosis and the depth of medical treatment needed.

Qualitative methods allowed the opportunity to understand the perspectives of caregivers of those diagnosed with 22q. Case studies provide practical knowledge to those in the medical field. Interviews, documents, artifacts, and documentation of field notes provided a greater opportunity to expand, compare, and contrast previous experiences and perspectives of caregivers through developing relative themes. Interviews offered an opportunity for participants to share information that may not normally be shared in a group setting.

A high index of suspicion is important for medical providers while a comprehensive medical history and physical examination may reveal the hidden 22q symptoms an individual has (Friedman et al., 2016). Previous research was conducted

assessing the perspectives of caregivers and adults who obtained a late diagnosis (Costain et al., 2012). A limitation of the study was that the conclusions of the study were dependent on the diagnosis being accompanied by genetic counseling, which is not always true in practice (Costain et al., 2012; Starke & Moller, 2002). Overall, the majority of the population in this dissertation perceived they did not have appropriate genetic counseling, which negatively affected the knowledge of the disorder. This was determined in the study. Chapter 3 discusses the methodology used to understand the perspective of caregivers and the barriers and challenges in diagnosis and treatment associated with individuals diagnosed with 22q. It describes the research design and rationale for that design, the role of the researcher, and trustworthiness of the study.

Chapter 3: Research Method

Introduction

The purpose of this qualitative explanatory case study was to understand the perceptions, opinions, perspectives, and experiences of caregivers on how the diagnosis of 22q of their loved one has affected their lives. The goal was to understand how the caregivers developed coping skills, addressed barriers, and managed quality of care issues. This research also identified how caregivers plan to address 22q issues in the future, caregivers' thoughts on how physician education and experience influenced the diagnosis and treatment, and their beliefs on why 22q is under- or misdiagnosed. This study focused on the lived experiences of the study participants in order to present an in-depth understanding of the phenomenon of diagnosing and treating 22q. This chapter includes a detailed description of the research design and rationale, role of the researcher, methodology, and trustworthiness.

Research Design and Rationale

Research Questions

The central research question in this study asked how caregivers addressed the barriers and quality of care issues of individuals diagnosed with 22q. Caregivers were asked what their perspectives, opinions, and perceptions of these areas are based their personal experience. Uncertainty in illness theory was used in order to evaluate and assess how any part of the diagnosis or treatment encounter affected the caregiver and/or individual. The perceived severity, exposure, and vulnerability of a loss of health and the

fear of that loss can be detrimental (So, 2013). These emotions were understood in this study.

The central research question for this study was as follows:

CRQ: How are the challenges associated with a 22q diagnosis addressed?

The subquestions for this study were:

RSQ1: How was a suspicion of 22q initially considered?

RSQ2: How did participants cope with the news once a diagnosis was made?

RSQ3: How do caregivers perceive the patients coped with the diagnosis?

RSQ4: How have caregivers addressed the barriers for patients diagnosed with 22q?

RSQ5: How do caregivers plan to address these barriers and quality of care issues in the future?

RSQ6: How did healthcare providers' education and experiences impact the diagnosis and treatment of 22q?

RSQ7: Why is 22q often under- or misdiagnosed despite being relatively common?

RSQ8: How can increased education of healthcare providers and the public help better identify and address these barriers related to this common yet sometimes unidentified disorder in the future?

Central Phenomenon

The central purpose of this study was to describe the lived experiences of caregivers of those diagnosed with 22q. Understanding these experiences help to better

educate individuals, including healthcare providers, about the disorder. With this education, those affected can be diagnosed more accurately and in a timelier manner.

Research Tradition

A qualitative research method using an explanatory case study design provided detailed caregivers' experiences of how 22q has affected individuals diagnosed with the syndrome. Qualitative research is being increasingly recognized as useful within health sciences research (Rosenthal, 2016). The case study is a distinctive form of empirical inquiry and must be pursued with the appropriate rigor of systematic procedures (Yin, 2014). A case study investigates a contemporary phenomenon in-depth and in the real-world context when the boundaries may not be evident (Yin, 2014). This case study did not ask the question *what*, but instead asked *how* and *why*. I used this qualitative case study design to explore caregivers' perceptions of how and why decisions were made and how these decisions affected their lives. This research tradition allowed me to take a retrospective approach for the personal histories while allowing for recommendations on how to improve the knowledge base to develop.

Rationale for Chosen Tradition

A clear understanding of the goal of the research being conducted motivates the choice of research design (Maxwell, 2013). Due to the desire of understanding the perceptions of the process of diagnosis and treatment of caregivers of those affected with 22q, I chose a qualitative case study design. Understanding the perceptions of caregivers in this research could not be attained via a quantitative study. Furthermore, knowing how and why the process occurred is best represented in an explanatory case study design.

Explanatory case studies are often chosen in order to answer a question that explains the assumed underlying connections in real-life interventions that are too multifaceted for survey or experimental research (Baxter & Jack, 2008). I chose the qualitative research method with a case study design utilizing one-on-one interviews to analyze the experiences of caregivers regarding the process of 22q diagnosis while offering the opportunity for them to make recommendations on how to improve the knowledge of the general public, healthcare providers, and educational specialists.

Role of the Researcher

In qualitative research, the researcher is the instrument used to gather information (Creswell, 2013; Patton, 2002). In this study, I collected data through studying documents, artifacts, documentation of notes, observing behaviors, and inquiring through interviews (Patton, 2002). Researchers may be distracted by responses in in-depth interviews, leading to losing control of the research focus (Raheim et al., 2016). It was important that I remained focused on extracting data that was specific to the study while not disregarding the full experience and my observations. Researchers are dependent on the study participants' cooperation in taking part in the interview and sharing their experiences (Karnieli-Miller, Strier, & Pessach, 2009). In conducting research, researchers aim for trustworthiness and authenticity. (Patton, 2002). As the research tool, researchers should be willing to listen to all stories and feedback in order to show respect and gain the trust of participants (Karnieli-Miller et al., 2009).

I have experience with 22q in the role of a caregiver. My now-adult son received a 22q diagnosis at the age of 15. This diagnosis was prompted by parental online research

following a first-time seizure at 14 years old, resulting in a diagnosis of epilepsy and an autoimmune disorder known as Hashimoto's syndrome. Although I was a person interested and experienced with 22q, each 22q-affected individual and caregiver have differences in timelines and anomalies that led to diagnosis.

The participants were recruited through social media with which I was personally affiliated due to being a caregiver of a child diagnosed with 22q. There was no professional or financial relationship among any of the entities or participants. The only personal relationships that existed were those that came to fruition through a common diagnosis of 22q.

My role was to recruit study participants, conduct interviews and make observations of study participants, collect field notes, documents, and artifacts, and transcribe, code, and analyze data. I considered personal experiences along with participant perceptions collected through research in order to better represent participant perspectives. There was no personal gain dependent on the outcome of this study as the goal of this study was to explain the perceptions of caregivers with loved ones diagnosed with 22q.

Methodology

Population

The population of this qualitative research study were male or female caregivers who were 18 years of age and older who cared for individuals diagnosed with 22q. These English-speaking individuals could have been located anywhere throughout the United States. This participant criterion properly aligned with the research questions. Participants

were delimited to caregivers of an individual diagnosed with 22q. English-speaking male and female participants who were 18 years and older of any ethnicity and who self-identified as being a caregiver of an individual diagnosed with 22q were eligible to participate in the study.

Sampling Strategy

Qualitative studies typically recognize purposeful selection (Creswell, 2014; Maxwell, 2013). Purposeful selection consists of studying an intentional setting, a target population, and specific activities in order to help better understand that population (Creswell, 2014; Maxwell, 2013; Patton, 2002). The recruitment strategy chosen for this research study was purposeful recruitment and sampling.

Participants Meeting Participation Criteria

Potential study participants who responded to the recruitment must have met the participation criteria for this research study in order to be included. This criterion included:

- The participants identified themselves as being an English-speaking caregiver of an individual diagnosed with 22q, and
- The participants were men or women who were 18 years of age or older.

Conducting research for all relevant populations was necessarily an ethical duty. *The Belmont Report* requires fair and just research procedures and outcomes, which includes appropriate inclusion criteria (National Commission, 1978). Limiting individuals with limited English proficiency can exclude people of specific backgrounds, including

ethnic and cultural characteristics that may otherwise lead to new outcomes or generalizations (Frayne, Burns, Hardt, Rosen, & Moskowitz, 1996).

Resnik and Jones (2006) suggested excluding individuals without a scientific or ethical reason is unfair. One scientific reason that may apply is the process of determining what the anticipated enrollment could be in a study (Resnik & Jones, 2006). Considering the anticipated overall enrollment allowed me to answer what the anticipated total number of participants with limited English proficiency are likely to enroll (Resnik & Jones, 2006). An illustration of this shows that 10 participants are expected to enroll in this qualitative study. The U.S. Census Bureau (2011) found that approximately 3.2% of those five years of age and over living in the United States speak English “not well” or “not at all”. Finally, the most recent research conducted assessing the population of 22q found that only 4% of a population of 370 studied was diagnosed with 22q. Based on these statistics, less than one person who is not English speaking would have enrolled in the study. This leads to a scientific reason in which English-speaking only is allowed in research.

Resnik and Jones (2006) suggested it is unreasonable to require a translation of consent forms and other materials when it is expected that none in that particular category will enroll. Due to the small sample size chosen for this qualitative inquiry, these numbers provide rationale that suggest less than one person who does not speak English well was anticipated to enroll in this research study. Therefore, with the limitation of research resources, this study was limited to English-speaking participants, only.

Sample Size and Rationale for Size

There are no definite rules regarding the sample size in qualitative research (Patton, 2002). Purposeful sampling is an intentional focus on information-rich, small sample cases that allow insight and in-depth understanding of a phenomenon (Maxwell, 2013; Patton, 2002). Determining the appropriate sample size for case study research is imperative in the research process (Rosenthal, 2016). A sample can always increase during fieldwork (Patton, 2002). The sample size for this research was 10 participants, or until saturation occurred. One-on-one interviews were conducted. Field notes were documented. The combination of interviews, documents, artifacts, and field notes helped better triangulate the data and results. Qualitative research is not used for generalization but for obtaining the experiential depiction from interviewees, keeping equal representation of experiences in mind (Patton, 2002).

Participants were identified and recruited through public 22q Facebook sites. Specific details, deadlines, and contact information were shared on the recruitment flyer. Saturation is apparent when no newfound information is being produced (Patton, 2002). Saturation did not take place.

Instrumentation

As the researcher, I was the main research instrument in qualitative inquiry. I was the only person collecting data in this study. Qualitative research design uses interviews that consist of open-ended questions (Creswell, 2013; Maxwell, 2013; Patton, 2002). These questions allow the researcher to ask follow-up questions for better clarification (Patton, 2002). Qualitative research is inductive and a less structured approach allows

researchers to focus on the phenomena being studied, allowing for an individualized or tailored method (Maxwell, 2013). However, as an inexperienced researcher, this study consisted of semi-structured interviews.

The basis for this researcher-developed instrument was so that the questions were specific to 22q and the design of a qualitative case study approach. Content validity was measured through obtaining the perspectives, based on the open-ended questions. In this inquiry, the data collection instrument appropriately answered the research question, focusing on the barriers and quality of care issues for individuals diagnosed with 22q.

Procedures for Data Collection

Data collection for this case study research included gathering data through one-on-one interviews, documents, artifacts, and documentation of field notes (Creswell, 2013; Miles, Huberman, & Saldana, 2014). A neutral location at a public university was utilized for interviews. Pertinent information was retrieved from the interview guide, field notes, partial drafts, audio recordings, and coded data, for reflection, as appropriate. I produced all data collection instruments. The interview guide assists in assuring the type of data being collected is appropriate and directly related to the research question (Creswell, 2013). The interview guide was used during interviews to collect data provided by the caregiver of the individual with 22q.

In this qualitative case study approach, there were 10 interviews. Interviews were conducted using an interview guide, audio recorder, and video conferencing, as needed. Data was also collected through documents, artifacts, and field notes. One-on-one interviews utilized audio recorders and Skype. Interviews were conducted for

approximately 45 minutes. Informed consent, including the voluntary participation of the study, was given to each participant.

Due to the size and focus of this case study approach, there should be adequate participants to successfully complete the project. The research questions proposed, along with the sub-set of interview questions, were appropriate and sufficient for this study. Participants were given the opportunity to obtain the results of the study by providing their e-mail address on the consent form. Follow-up e-mails were sent for member checking procedures.

Data Management and Analysis Plan

Gathering multiple forms of data, including interviews, documents, artifacts, field notes, and audio recordings can be overwhelming, even with the smallest of studies. While data collection is important in qualitative research, the management of data collected is more important. Researchers should have a sound process for the collection and storage of data. This allows a more effective and efficient data analyzing process. Creswell (2013) suggested that computer software programs are the best option for large databases of research data. In small research studies, data may still be overwhelming. Having an electronic process of sorting and coding data is important. Qualitative data analysis software, NVivo, was used in the storage and data analysis of this study.

Beginning researchers often develop too many theme codes, which can affect the ability to publish results (Creswell, 2013). Maxwell (2013) implied that researchers should connect strategies found within the data in order to address deficiencies of separating the original contextual relationship within the data collected. Miles et al.

(2014) proposed that working with raw text and field notes shows recurring patterns and themes that blend data that would otherwise be independent. Linking the data offers various ways to identify the original context while creating a well-rounded approach that shows relationships among interviews and field notes (Maxwell, 2013). Creswell (2013) recommended starting with a small number of themes and gradually emerging is a better tactic than developing too many codes. Using NVivo offered a structured way to find themes while keeping the data organized. A discrepant case is presented with the results.

Issues of Trustworthiness

Credibility

Qualitative research has grown significantly over the years not only in social sciences, but also more recently in health sciences (Houghton, Casey, Shaw, & Murphy, 2013; Santiago-Delefosse, Gavin, Bruchez, Roux, & Stephen, 2016). Validity establishes if the results are credible and authentic to the population and those who read the research (Miles et al., 2014). Credibility involves the process of conducting the research in a believable manner and being able to demonstrate that (Houghton et al., 2013). Credibility is determined by the consistency that is present between the research question, theoretical framework, data collection procedures, and data analysis (Santiago-Delefosse et al., 2016). Credibility can be verified through multiple methods, including triangulation of data and analyses, member checking and feedback to participants, and the search for negative cases (Santiago-Delefosse et al., 2016). Within this study, triangulation, member checking, feedback to the participants, and the search for negative cases were conducted. In order to conduct member checks, e-mail addresses were requested.

Transferability

Qualitative research studies are most often designed to study a specific issue within a certain population, focusing on a particular phenomenon within a certain context (Leung, 2015). The transferability of qualitative research lies in the eyes of the reader. The original research must be presented and adequately described with an emphasis on original text, raw data, and direct quotes in order for readers to make informed decisions and interpretations about the applicability of the findings within context (Houghton et al., 2013).

Dependability and Confirmability

Dependability is comparable to the concept of reliability in quantitative research, referring to the stability of the data (Houghton et al., 2013). Dependability was established through audit trails and triangulation. Confirmability addresses the neutrality and accuracy of the data, closely linked to dependability (Houghton et al., 2013). Consultation with mentors, advisors, committee members, and colleagues through the data analysis process assisted in greater reflexivity in the study. As the research instrument, I provided all perspectives of data from various sources in order to maximize the potential for in-depth understanding, insight, and completion of each case and their context Houghton et al., 2013).

Ethical Procedures

Permission from Walden University's Institutional Review Board (IRB) was requested. Walden University requires an IRB research proposal application. This application was submitted with the appropriate and supporting documents prior to any

recruitment or data collection. Walden University's IRB reviewed all processes within the research project including the confidentiality, privacy, informed consent, and study design. Once Walden University's IRB granted permission to proceed (Walden IRB approval number 05-05-17-0530552), recruitment materials were shared via social media pages within appropriate 22q groups. Copies of the recruitment materials, informed consent process, and form were submitted with the IRB research proposal application.

Informed consent consists of more than just requesting the participant complete a consent form. It requires the researcher to communicate and explain the participant's role and rights in the research process, outlining the opportunity and process for research study withdrawal. An identification alphabetical letter identified each participant. The population in this study represented caregivers of those who have chronic medical conditions. These medical conditions may lead to any number of life-long accommodations. It was important to consider any potential questions or concerns they may have in study participation. Personal relationships may exist between me and some study participants, due to personally being a caregiver of a child with 22q. These relationships came to fruition through having a family member diagnosed with 22q. However, with the use of an interview guide, the questions and inquiry were the same for all participants. No professional relationships exist between the study participants and me.

The data collected were protected appropriately. Data were stored on a password-protected computer within a password-protected document. In order to prevent data loss, this password-protected document was backed up daily on a flash drive. The flash drive,

a hardcopy of all data collected, and the audio recordings were placed in a locked safe in order to prevent possible loss of data. Consent forms with participants' signatures and names were stored separate from my notes and audio recordings. Only I had access to the consent forms and research data.

Summary

This qualitative explanatory case study approach helped to better understand the perceptions, opinions, perspectives, and experiences of caregivers on how the diagnosis of 22q has affected their lives. The theoretical framework that guided this study was Mishel's (1981) uncertainty of illness theory. This theory agrees with the study as parents who perceive a condition as misunderstood or incomplete may experience greater uncertainty in obtaining the appropriate health care for their child's chronic condition. This chapter focused on the participants in the study, data collection and analysis processes, and participant and research protections. Chapter 4 outlines the results of the analyzed data.

Chapter 4: Results

Introduction

This purpose of this qualitative case study was to understand the barriers and quality of care issues for caregivers of those diagnosed with 22q. I attempted to understand their perceived concerns, coping skills, communication techniques, understanding of relevant issues, opinions of physician experience and knowledge, plans for the future, and understanding and preparation for addressing future issues. Several studies have identified the importance of an increased need of suspicion on behalf of undiagnosed or misdiagnosed cases of 22q. Much of the current literature was focused specifically on the early childhood years. This study specifically addressed issues that are relative throughout childhood, adolescence, and adult transitioning. Since there was no specific age requirement of the affected individual, this study provided a well-rounded sample with relevant and appropriate feedback. The CRQ answered in this study was "How are the challenges associated with a 22q diagnosis addressed?" Mishel's (1981) reconceptualization for the uncertainty in illness theory directed the specific research subquestions used in this study.

RSQ1: How was a suspicion of 22q initially considered?

RSQ2: How did caregivers cope with the news, once diagnosed?

RSQ3: How do caregivers perceive the patients coped with the diagnosis?

RSQ4: How have caregivers addressed the barriers for patients diagnosed with 22q?

RSQ5: How do caregivers plan to address these barriers and quality of care issues in the future?

RSQ6: How did healthcare providers' education and experiences impact the diagnosis and treatment of 22q?

RSQ7: Why is 22q often under- or misdiagnosed despite being relatively common?

RSQ8: How can increased education of healthcare providers and the public help better identify and address these barriers related to this common yet sometimes unidentified disorder?

Additionally, in this chapter I describe the research instrument, setting, participant sample, interview process, data collection process, trustworthiness, qualitative analysis, and provide a summary. Chapter 5 focuses on the interpretation of the data and findings, limitations of the study, future recommendations, and implications of the study.

Research Tools

I developed an interview guide (see Appendix B) that consisted of 15 open-ended questions for the interviews conducted. The first section included questions related specifically to the details of diagnosis, such as age at diagnosis and how diagnosis occurred. The second section focused on the biggest concerns with the diagnosis, followed by the ways the diagnosis was addressed. The third section looked at the communication of the diagnosis to others, including the affected individual. The next section inquired as to the current mental and physical condition of the affected individual, based on the perception of the caregiver. Finally, the last section examined the healthcare

providers' knowledge and education of 22q from the caregiver's perspective. This was based on resources provided, past and future plans for transitions, and additional issues foreseen. Each of the 15 questions in the interview guide was directly related to the research questions in the study.

Setting

The study was conducted either in-person or through video conferencing in a face-to-face setting. For in-person interviews, I chose to conduct them at a public university that was open to the public where I could reserve a private room. The room temperature was controlled and set at a comfortable level. I chose this setting due to the public location and to ensure privacy of the individuals being interviewed. For video conferencing, I chose a private setting to conduct the virtual interviews. Some of the study participants online were alone while others had their children present during some of the interview. The two in-person interviews were conducted including only the study participant and me. All in-person study participants arrived at the interview on time. A few of the virtual study participants were a couple of minutes late, due to technology difficulties. The interviews were scheduled for 45-minutes. Interviews spanned between 23 minutes and 52 minutes in length.

Demographics

Ten caregivers agreed to participate in the study. All study participants identified as English-speaking caregivers of a loved one diagnosed with 22q and who were 18 years or older. Within the group were one single mother and nine married mothers who had children diagnosed with 22q anywhere from 3 months to 23 years old.

Data Collection

After obtaining approval and authorization to conduct the study from the Walden University IRB, I posted recruitment flyers on three social media Facebook pages. Thirty-five interested individuals reached out to me for more information. I provided an electronic standard blurb of the intent of the study. One interested individual was not a caregiver but was affected herself. Another interested individual had 22q duplication and not 22q deletion. These two individuals did not qualify for the study. Nine of the thirty-three qualified individuals were interested in participating. Surprisingly, a friend who had a mutual acquaintance contacted me and asked if I would be willing to share my personal story with someone who had a newborn who was newly diagnosed. I agreed and shared my personal story with this person. She asked if she could be in my study. She met the qualifications, so she was one of the participants. All 10 participants were screened to ensure they were English speaking, a caregiver of an individual diagnosed with 22q, and at least 18 years old. I scheduled interviews online or in-person.

Each individual participating online was provided a consent form through e-mail. Upon the scheduled virtual interview, a standard blurb was read reminding them of the nature of the study and the opportunity to quit the study at any time. Each individual participating in-person was provided the same consent form with the explanation of the voluntary nature of the study reiterated prior to the interview. All participants were asked if they had any questions prior to the study. I gave each in-person participant a $20 Visa gift card when each one arrived to the interview. I mailed each virtual participant the gift card at the conclusion of the interview. Walden University's IRB had approved

compensating each participant for their time by giving them a $20 Visa gift card. Nobody stopped participation prior to the end of the interview.

In both the in-person and virtual face-to-face interviews, I thanked the participants for their cooperation, restated the voluntary nature of the study, and reminded them the interview would be audio-recorded. Upon completion of the interview, I communicated the process and importance of member checking, as stated in the consent form. I had three days to transcribe and forward the transcripts for member checking. The participant had five days to return any feedback. I asked each individual if there were any other questions or comments at the conclusion of each interview. I also collected data through field notes and observations during each in-person and virtual interview.

The interviews were scheduled in 45-minute sessions. The shortest interview was 23 minutes, which was with the caregiver of a 3-month old with a recent diagnosis. The longest interview was 52 minutes. I had two interviews that spanned longer than 45 minutes. During these two interviews, I paused at an appropriate time and notified the participant we had interviewed for 45 minutes and told them how many questions were left. I asked if they wanted to continue or would like to stop. They both chose to continue. All transcripts were e-mailed for member checking. I had four responses for minor changes. On one, I had specifically asked an acronym question. On the second, I had used “she” instead of “he” when referring to the affected individual. The third made some grammatical revisions and updated one proposed surgery name and location. The last one requested I leave some specific feedback out, as it was more in conversation.

A variation in data collection was requested to Walden University's IRB in order to allow individuals who are unable to meet in person or who do not have access to a webcam the opportunity to participate in the study. Although this request was made and approved by the IRB, the ten individuals who participated had means to attend in-person or via video conference so this approval was not needed. There were not any unusual circumstances encountered during the data collection process.

Data Analysis

Data analysis started during the participant interviews when I began noticing and identifying recurring themes. I translated the audio files into word documents. By transcribing each interview myself within three days, I was able to become more familiar with the data. In order to maintain confidentiality, I used a participant letter for reference to that individual. I utilized NVivo 11 for PC by QSR International for data storage and further analysis. I read each transcript at least three times for coding and identifying common themes, starting with participant responses to each interview question. They were then coded within the areas of the overall research questions in this study (see Table 1).

By using NVivo after my initial finding of meanings, emergent themes began to be identified. Coding categories were changed and reidentified in order to present the most relevant and appropriate data. Relating and correlating the data within the constructs of the uncertainty in illness theory facilitated in specifically answering the study's research questions. The themes identified contributed to the development of a rich, thick

understanding of the perceptions of barriers and quality of care issues of individuals diagnosed with 22q.

Table 1

Themes

Research subquestion	Themes
1. How was a suspicion of 22q initially considered?	age and symptoms at diagnosis
2. How did caregivers cope with the news, once diagnosed?	internet searching, research
3. How do you perceive the patient coped with the diagnosis?	doesn't understand
4. How have caregivers addressed the barriers for patients diagnosed with 22q?	enrolled in research studies, communicated the diagnosis with family, pediatrician, teachers, other caregivers, addressed medical care follow-ups
5. How do caregivers plan to address these barriers and quality of care issues in the future?	continued medical care and follow-ups on possible conditions, unsure of future transitioning and independency
6. How did healthcare providers' education and experiences impact the understanding in diagnosing and treating 22q?	very confident in specialists and primary doctors, not confident at all in their experience/education,
7. Why is 22q so common yet under- or misdiagnosed?	22q was only considered at birth for those with a heart condition; lack of provider knowledge
8. How can healthcare providers' and the publics' increased education and knowledge better identify and address these barriers related to this common yet sometimes unidentified disorder?	more provider collaboration, genetics testing at birth, mandatory annual check-ups.

Themes

RSQ1: How was a suspicion of 22q initially considered?

RSQ1 was answered by determining the age and symptoms at diagnosis.

RSQ2: How did caregivers cope with the news once a diagnosis was made?

RSQ2 had the following themes emerge during data analysis: (a) internet searching, and (b) research. Major concerns included current or potential heart conditions, the possibility for psychiatric disorders (specifically schizophrenia), and the unknown. Participant D said, “Immediately, we started researching it.”

RSQ3: How do caregivers perceive the patients coped with the diagnosis?

RSQ3 had the following themes emerge during data analysis: (a) they did not believe the diagnosed individual understands, and (b) the caregiver communicated the diagnosed individual did not like to be different from peers but wanted to be and feel normal physically and do things their friends did socially. Participant I said,

> It’s not 22q that bothers her. She doesn’t care. It’s the heart. She really doesn’t care. With the heart, she can’t run. She doesn’t get to participate in track, basketball, and that’s what bothers her. Because of her ejection fraction ratio being out of line and her aorta stenosis is narrow; it is kind of like an adult with a 65% blockage. It isn’t her heart but her aorta that keeps her from participating.

RSQ4: How have caregivers addressed the barriers for patients diagnosed with 22q?

RSQ4 had the following themes emerge during data analysis: (a) enrolled in research studies; (b) communicated and searched for answers from family members who

were physicians; (c) communicated the diagnosis with their pediatrician, teachers, and others who shared in caregiving roles; (d) proposed Individualized Education Plans (IEPs) in school; and (e) addressed medical care follow-ups as suggested/needed. Participant F stated,

> I've used DiGeorge syndrome to open doors for her as far as what specialists she can see because I do know specialists are interested. They want to know what she's like. So if, say, there's a specialist that I want her to see who is difficult to get into or at a well-known university or whatever, I tend to say, "Look, she has DiGeorge syndrome," and people, thank goodness, they are interested in learning more about her because of that. I have found that it has opened doors for her to see medical care that I don't think otherwise she would be able to get.

RSQ5: How do caregivers plan to address these barriers and quality of care issues in the future?

RSQ5 had the following themes emerge during data analysis: (a) continued medical care and follow-ups on possible conditions; (b) proposed additional options for later school years; (c) unsure of future transitioning; (d) social interactions; and (e) what happens when the caregivers are gone. Issues handling money, the possibility of not being able to drive, and the need to push to independency were all concerns with adulthood transitioning. Participant C shared,

> I worry about his learning in school. He will be in special education classes to begin with but I worry about how he seems to learn things or process things. He does have a lot of social anxiety so I worry about that.

RSQ6: How did healthcare providers' education and experiences impact the understanding in diagnosing and treating 22q?

RSQ6 had the following themes emerge during data analysis: (a) very confident in specialists and primary doctors; (b) not confident at all in health care providers' experience/education, parents have to be the educators; (c) did not receive any resources upon diagnosis; (d) received handouts, referrals to a genetics counselor; (e) genetics counselor did not tell us anything; (f) suggested the best places for care; (g) did not feel prepared for additional conditions, somewhat prepared after researching; and (h) no academic preparation was provided. Participant G said, "Every now and then, I go back to the 180 list of anomalies and wonder."

RSQ7: Why is 22q often under- or misdiagnosed despite being relatively common?

RSQ7 had the following themes emerge during data analysis: (a) six had a heart condition but nobody without a heart condition was diagnosed at birth; (b) genetics counselors suggested parents know more than they do about 22q; and (c) some never visited with a genetics counselor. Participant I answered, "None at all. The genetics counselors didn't tell us anything. They didn't seem to know anything."

RSQ8: How can increased education of healthcare providers and the public help better identify and address these barriers related to this common yet sometimes unidentified disorder?

RSQ8 had the following themes emerge during data analysis: (a) no resources other than a brochure or referral were provided; (b) the best 22q clinics and hospitals

were suggested; (c) originally prepared for death of child; (d) no tools were provided; (e) a possible genetics diagnosis was undercommunicated; (f) better to discuss the unknowns; (g) other parents on social media help with knowledge; (h) nobody talks about the future; (i) more parent/physician/educator collaboration; (j) genetics testing at birth when they test for everything else; and (k) mandatory complete annual check-ins for those diagnosed with 22q.

While collecting and analyzing the interview data, I developed a greater suspicion of what 22q caregiving and proactive treatment should look like. I decided I wanted to analyze additional documents and artifacts found on public websites and social media pages that were relevant to this study. A second variation in data collection requested to Walden University's IRB was to allow data analysis of public documents, videos, and other documents/artifacts that may be posted on public Facebook pages and websites. The request did not include private social media groups and was approved.

Artifact

The first analyzation was of an artifact from a documentary named *Just Like Us* (ANAID Entertainment, 2017). This documentary was a short video of a caregiver (non-parent) and an adult client who tackle daily living together while battling the ongoing issues related to 22q. The caregiver suggested it is very important for everyone to have control of his or her own life (ANAID Entertainment, 2017). Although this seven-minute documentary was brief, it focused on two basic needs of anybody, including those with developmental disorders. These needs are meaningful relationships including both friendships and lovers, and a purpose in life where individuals can work to accomplish

something (ANAID Entertainment, 2017). The caregiver stressed the importance of supporting individuals in getting through their anxiety and emotional dysregulation. Anxiety is normal and emotions are valid.

Documents

The documents analyzed during data analysis included tables of guidelines pulled from scholarly articles related to managing 22q. Both of these guidelines were created for healthcare providers in order to better communicate and initiate current and future plans for medical care. The first set of guidelines was pulled from an article focused on general guidelines (Bassett et al., 2011). These guidelines show multisystem features of 22q, with researched frequencies of occurrence. Many of the symptoms found in the caregiver interviews could have been diagnosed prenatal, with the accurate knowledge and education of this syndrome. For instance, cardiovascular, palatal, gastroenterological, genitourinary, skeletal, and growth and development are all areas that can be found prior to birth (Bassett et al., 2011). Once diagnosis occurs, there are general recommendations for assessments and health monitoring that follow children throughout infancy, preschool age, school age, adolescence, and adulthood (Bassett et al., 2011; Fung et al., 2015). Fung et al. (2015) provided signs and symptoms that represent a change from baseline that could be indicative of a treatable psychiatric illness. Bassett et al. (2011) offered important cautions and considerations for patients with 22q while Fung et al. gave general recommendations for prenatal and perinatal care of adults with 22q. Lastly, Fung et al. provided genetic counseling strategies specifically for adults with 22q. Based on the data collected in the interviews and the practical guidelines explored, caregivers of

individuals diagnosed with 22q would be better suited to address barriers and quality of care issues if healthcare providers were knowledgeable and comfortable with these guidelines.

Discrepant Case

While the diagnosis of one caregiver's child was undiagnosed until age 2 ½, the caregiver felt confident in the ability of her pediatrician and the hospital's resources to address any upcoming issues that may be presented, post diagnosis. This caregiver was only 6 months post-diagnosis so there are several things that have not yet been tested for or addressed. However, there was complete confidence in anything that may present itself and the ability, knowledge, and experience of the pediatrician to help treat the issues, leading to full confidence in addressing the future.

Evidence of Trustworthiness

Validity in qualitative research refers to the way researchers can confirm their findings presented are accurately reflecting the participants' perceptions and experience (Ravitch & Carl, 2016). The words trustworthiness and validity are often used interchangeably in order to show the importance of ensuring rigor, authenticity, and credibility in qualitative research (Miles et al., 2014

Credibility

Credibility was verified through triangulation of data and analyses, member checking, and searching for discrepant cases (Santiago-Delefosse et al., 2016). In this study, interviews were conducted via video conference and in-person. Data were

transcribed and returned for member checking. Analyses conducted included data collected through interviews, documentation, artifacts, and field notes.

Transferability

Due to the specific nature of this research being conducted to understand the barriers and quality of care issues of those diagnosed with 22q, emphasis on original text and raw data, including direct quotes, was placed. This allows readers the opportunity to make an informed decision on the applicability and interpretation of the findings within the context of the phenomenon (Houghton et al., 2013). The goal of this research was not to produce findings but to accurately and descriptively communicate the context within the relevant parameters (Guba, 1981).

Dependability and Confirmability

Dependability was established through triangulation and audit trails. With the use of an audio recorder, researcher notes, and member checks, I was able to audit and report the data accurately. Confirmability was ensured through coding verbatim recordings and consultation with interviewees, mentors, advisors, committee members, and colleagues, when appropriate.

Results

The following section presents the research findings in this study. The findings presented were based on interview questions directly associated with the study's research questions. The themes chosen were identified from reviewing transcripts. They are linked throughout the findings to provide more in-depth detail. I applied the framework of

uncertainty in illness theory to the analysis of the interview data. All responses written are direct quotes from the participant's perspectives.

Research Subquestion 1

The interview questions for RSQ1 were:

- At what age did you find out your loved one has 22q11.2 deletion syndrome? and
- How did you find out about the diagnosis?

Initial suspicion of 22q. Five out of 10 caregivers stated their child was diagnosed within the first month of life, due to a heart condition prior to, at, or immediately after birth (see Table 2). One participant whose son was only three months old currently provided this recollection of diagnosis:

> The head doctor in the NICU, I believe it was while we were waiting for his heart surgery because we had to wait about 6 days before they could offer it, she casually said "Yeah, do you know what DiGeorge syndrome is?" and we said "Yes" because we knew that there was a risk he could have it and she said "Yeah, he has that". So it was very casual, we weren't really prepared for that.

Table 2

Demographic Characteristics of Diagnosis

Caregiver	Current age	Age at diagnosis	Diagnosing symptom
Participant A	18 years	8 years	Lack of growth
Participant B	21 years	4 years	Palate
Participant C	5 years	3 ½ weeks	Heart
Participant D	3 months	1 week	Heart
Participant E	23 years	5 years	Palate
Participant F	7 years	1 week	Heart

Participant G	16 years	8 months	High fever
Participant H	13 years	1 week	Heart
Participant I	14 years	1 week 6 days	Heart
Participant J	3 years	2 ½ years	Palate

The remaining five participants found out anywhere between eight months and eight years old. All five of the remaining had a palate deficiency, recognized at birth or shortly after. Participants B found the diagnosis through their own research. “I was doing research about palate issues and I came across an article about 22q and it fit so I had the doctor run the test.”

Participant A’s child had a palate deficiency and heart condition at birth. This participant’s daughter was 8 years old when diagnosed. Participant A said, “She had stopped growing for two years and our pediatrician sent us to an endocrinologist for growth hormones. He is the one who took a look at her and found out.”

Research Subquestion 2

The interview questions for RSQ2 were:

- What were the biggest concerns you had when your loved one was diagnosed with 22q11.2 deletion syndrome? and
- In what ways did you address the diagnosis?

Caregivers coping with diagnosis. All of the parents had significant concerns. Two immediately brought up the fact that they did not know if their child would live to leave the hospital. Participant H shared,

> I really didn't even know if I was taking him home from the hospital. He lost a roommate while we were there. He's my first baby. I never really left home. I was by myself and I didn't know if he would live. It was his overall health.

Three participants were immediately concerned with the heart, upon diagnosis. Participant E described the immediate relief of knowing, due to serious academic issues in need of school services, after her 5-year-old son's diagnosis.

> Initially, we were happy because we felt at that time that he would get the services. That's what we felt was important. We wanted him to progress and he was slow with everything. It was joy, which was terrible, but it was joy. And then after that, came the concerns with the heart so we started checking up on the other things

Ten out of 10 participants conducted their own research through multiple means. Six out of 10 specifically stated they used the internet to research. Three out of 10 were told to not Google it. Participant D said, "She [the doctor] said, "It's probably not best for you to research it on your own. It's scary getting on the internet." Although, my husband and I are intelligent enough to figure out what is accurate and what is not, so we did it anyway.

> She [physician] said, "Try to hold off and we will get a genetics counselor because at the hospital they have all of that there." So she said, "I'll get a genetics counselor to talk to you". It had probably been over a week when we reminded her we needed the genetics counselor to talk to us

> and eventually she did. By that time, she was not telling us anything we hadn't already found out. So, we just tried to make sure whatever we were reading was credible. Published research and other credible sources.

Participant F said, "They gave me a brochure and they told me not to look things up on Google."

Research Subquestion 3

The interview question for RSQ3 was:

- RSQ3: How did your loved one cope with the news, once diagnosed?

Diagnosed individual's coping skills. Two of the caregivers' diagnosed children were too young of age to be told or understand. The caregivers for an additional two believe the diagnosed individuals do not understand. Participant C shared, "I don't think he (5-year old diagnosed) understands. His four-year old sister understands. He isn't quite to the point he understands."

Others do not like being "different" by not being able to physically participate in activities, by always being sick, having learning differences, and by not being able to do things their friends do. Participant H answered,

> He says I wish I didn't have this and they didn't think I'm weird or I wish I didn't have a bad heart because now he realizes he has other things that impair his future. Like he can't go into the military. I'll say something like it's cool and he says it's not cool to be different.

Some, however, did not seem bothered by the differences but embraces them. Participant F shared,

> She knows that she is different. The way we explain it culturally is that she has a medicine and she was made in this way. She has cultural responsibilities that she has to carry out. This is part of her medicine. The way you are made and what you need to do, this is part of your responsibility.

Research Subquestion 4

The interview questions for RSQ4 were:

- In what ways did you address the diagnosis?
- To which healthcare, educational, or other professionals did you communicate the diagnosis?
- What is the nature of your loved one's physical medical condition?
- What is the nature of your loved one's mental cognitive ability? and
- What accommodations have been made for the affected individual?

Caregivers' self-efficacy in addressing barriers. Caregivers enrolled in research studies and other opportunities available, due to the condition. Two caregivers turned to family members who were physicians for answers. Participant I shared,

> I asked for information about DiGeorge syndrome because that's what they were calling it. They did not give me anything. They told me that they would have a genetic counselor come and talk to us. I had a cousin who was in medical school. I asked my aunt to get some information out of my cousin's medical books, if there was anything. At this point, when she was in the hospital, all I was

> researching was her heart condition and I feel like I read everything I possibly could read about a heart. But I didn't know anything about 22q. My aunt wouldn't bring it to me because there was only one page in the medical book 14 years ago that the students were learning from and basically it was gloom and doom and she was going to die. She told me years later that she didn't feel comfortable bringing it to me because it was very depressing.

Nine out of ten told their pediatricians after a hospital diagnosis. The diagnosed had many physical and medical conditions including heart conditions, cognitive delays, palate deficiencies, failure to thrive, scoliosis, social delays, joint pain, artery and vein misplacements, digestive and colon issues, and immune issues. Participant G said,

> He's (16-year old) had multiple surgeries on the colon. He had anal stenosis. We fixed that. We had the colostomy bag for four years. We went through probiotics, antibiotics regimens. Two years ago, they added a port in his belly button. He flushes every night. He does 1,000 ML of liquid so he is able to go before bed and has had very few accidents at school this year.

Participant B shared,

> Right now, she (21-year old) has schizoaffective bipolar disorder. Sometimes she suffers from a bit of depression but it is controlled pretty well right now. Anxiety issues. She complains about joint pain. She gets cysts under her arms that we are having to treat quite frequently. She suffers from severe allergy issues. She also has lichen sclerosis. She suffers with the issues as far as she is completely flatfooted. She has typical facial characteristics, tapered fingers, low IQ. Her IQ is

74. She is delayed. She still considered failure to thrive. She's still tiny and weight is a big issue as far as putting anything on. And I think that is about it.

Many communicated with teachers and school officials, leading to 7 out of 10 having an IEP with the last one having a medical 504 accommodation. Two diagnosed individuals are not at school age. Participant G explained what part of the IEP allows. "The school lets him use notes for most tests. He can leave and test in a separate area so he doesn't get distracted. He gets out for speech therapy."

Research Subquestion 5

The interview questions for RSQ5 were:

- What is the nature of your loved one's physical medical condition?
- What is the nature of your loved one's mental cognitive ability?
- What accommodations have been made for the affected individual?
- In the future, what additional issues do you foresee, and
- How do you expect your loved one to transition into adulthood?

Caregivers' plan for the future. All caregivers that were interviewed shared they would need to follow-up on previous and potential new anomalies that may present themselves during childhood and throughout the transition to adulthood. As previously mentioned, many physical and mental medical conditions can last long-term or present themselves later. The two caregivers with children not yet in the school system understand the necessity in making appropriate plans, depending on individual needs. Caregivers worry what happens when they are gone. Participant B questioned, "What happens after we are gone?" Participant H answered,

> We always joke that he is going to be living in my basement my whole life. I don't know how I spent so many years of my life planning his funeral because I didn't think I would have him that long so I don't see how he's going to succeed and be productive. I know people get married but they still struggle. I don't expect him to be a Harvard graduate but eventually we die. It's not like they build a 22q community and live like smurfs. I hope he can achieve and he can fit into this world better.

Research Subquestion 6

The interview questions for research question 6 were

- How sure are you that your loved one's healthcare providers have enough knowledge and experience to properly treat you/your child?
- What resources did your loved one's healthcare providers provide upon diagnosis?
- How prepared did you feel in addressing possible additional conditions related to 22q? and
- In what ways, if any, could your loved one's healthcare providers have better prepare you for academic and adulthood transitions?

Perceived impact of healthcare providers' experience and education. Overall, the perceived experience and education of the specialists and pediatricians the caregivers currently work with is positive. Participant C said, "As far as the specialists that we see . . . I have full confidence in them." Participant J added, "I'm very confident that they have enough [experience and knowledge]."

However, there were many negative perspectives of the diagnosis process. Participant A said, “It is always about educating the doctors.” Participant E stated, “I don’t think they are very knowledgeable. “

When identifying resources provided to them upon diagnosis, most of the participants said “none” while a couple said “handouts.” Some were referred to geneticists. Participant B said, “The geneticist looked at me and said, “Honestly, you know more than I do.”

The majority of caregivers felt unprepared for future issues. Participant F stated, “I was preparing for her death, and how I going to care for her remains and where she was going to be buried. That’s what I was preparing for. They didn’t give me any tools. They really didn’t.” Participant J offered, “I feel more prepared now that I know the information I have read, talked with parents and within Facebook groups, chatting with other parents.”

Although some caregivers felt prepared for the issues that may present themselves, they would like to have had more information on transitions. Participant A stated, “Nobody really talks about the future.” Participant E added, “Any help with what kind of accommodations throughout school that he should have had would have been nice. They pretty much gave us no preparation.” Participant G offered, “We have had several teachers who have been willing to read the *Faces of Sunshine* book and get to know stuff. I wish the physicians would have just said ‘I don’t know . . . why don’t we find something out’ instead of me being my own research partner and everything.” Participant H said, “I wish we had a person or annual studies and that you could do

annual check-ins. I wish we could go to a pediatrician and it was a mandatory part of all ages."

Research Subquestion 7

The interview questions for RSQ7 were:

- How did you find out about the diagnosis?
- How sure are you that your loved one's healthcare providers have enough knowledge and experience to properly treat you/your child? and
- What resources did your loved one's healthcare providers provide upon diagnosis?

Perceived contributions to under- and misdiagnoses. Six individuals had a heart condition. Five of these six were diagnosed at birth. Out of the ten caregivers interviewed, nobody without a heart condition was diagnosed at birth. Genetics counselors were often late on the scene (after parents had already conducted their own research) or unhelpful. When I asked what resources were provided, Participant I said,

> The genetics docs didn't help at all. They treated her like a science experiment and wasted our time, literally hours at a time just sitting waiting for them. We stopped going because, in my eyes, there was no point. They weren't giving me any helpful information; actually, they weren't giving me any information.

Once diagnosed, the specialists and primary physicians seemed to be able to satisfactorily treat the issues of those diagnosed with 22q. Participant G said, "I think right now we are good. For the longest time we were not good which put me in a really bad place."

Research Subquestion 8

The interview questions for RSQ8 were:

- How sure are you that your loved one's healthcare providers have enough knowledge and experience to properly treat you/your child?
- What resources did your loved one's healthcare providers provide upon diagnosis?
- How prepared did you feel in addressing possible additional conditions related to 22q? and
- In what ways, if any, could your loved one's healthcare providers have better prepare you for academic and adulthood transitions?

Increased education and knowledge of healthcare providers and the public. Based on the analysis of this data, healthcare providers, once aware, are capable of treating or referring on issues related to 22q. Unfortunately, there was a lack of sufficient support during the diagnosis process and immediately following. Resources provided were limited to brochures, referrals, and, in many cases, nothing. This leads caregivers to not have self-confidence leading to a lack of self-efficacy in treatment options during the early and adult years. Feedback suggests healthcare providers could be helpful in providing resources for academic and adulthood transitions.

Discrepant case. The discrepant case was a new diagnosis of only six months for a 3 year old. This case was in direct contrast with the other nine individuals who suggested the health care professionals did not provided adequate or any resources for further health care treatment, academics, or adulthood transitions. This contrast, however,

did not influence the results as the uncertainty in illness theory still applies to the pre-diagnosis concerns that may lead to inadequate or delayed treatment.

Summary

Chapter 4 provided an overview of the processes used to recruit participants and collect, manage, and analyze data collected from caregivers of individuals diagnosed with 22q. Participants were selected based on purposeful sampling. Responses from in-depth interviews communicated the perceptions of the barriers and quality of care issues of caregivers of those diagnosed with 22q. All participants in this study had one of the two most predominate anomalies noted in the research of 22q. These two anomalies are the heart condition and palate abnormalities. These two anomalies caused a significant amount of stress for the caregiver. Often, appropriate care for other possible conditions was nonexistent until a diagnosis of 22q. The themes indicated caregivers were often afraid of the unknown and always conducted their own research while also relying on that provided by healthcare providers.

Chapter 5: Discussion, Conclusions, and Recommendations

Introduction

The purpose of this qualitative case study was to understand the barriers and quality of care issues of caregivers for those diagnosed with 22q. The aim was to understand the perspectives, thoughts, and experiences of the process from prediagnosis through treatment and into the adulthood. I explored the caregivers' perceptions of healthcare providers' experience and knowledge, specifically when it came to diagnosing the individual. I further explored their opinions of the current level of treatment their loved ones were receiving. Interview data were stored, organized, and coded through NVivo in order to better identify the common themes amongst the data collected through the construct of Mishel's (1981) uncertainty of illness theory.

Caregivers communicated the concern of missed anomalies or late diagnosis of additional anomalies. A recent case study found a 54-year old man who was referred for follow-up treatment for primary hypoparathyroidism (Hoshino, Machida, Shimano, and Taya, 2017). No abnormal echocardiogram findings were found but an aortic arch anomaly was identified and the adult tested positive for 22q. This was one example of the concerns of caregivers interviewed in this study. Current research demonstrated that more than 50% of individuals diagnosed with hypocalcemia episodes occurred after the age of 17 in individuals diagnosed with 22q (Cheung, et al., 2014). The most recent research focused primarily on 22q and psychiatric disorders. Psychosis research shows that early intervention is necessary in order to develop timely and effective treatment strategies, as well as to reduce the long-term effects associated with this condition (Armando et al.,

2017). Psychosis was another concern of the caregivers interviewed. These most recent studies provided the rationale for a greater need of early suspicion and a timely diagnosis.

Interpretation of the Findings

This study was based on the following research questions:

The central question for this study was as follows:

CRQ: How are the challenges associated with a 22q diagnosis addressed?

The subquestions for this study were:

RSQ1: How was a suspicion of 22q initially considered?

RSQ2: How did caregivers cope with the news, once diagnosed?

RSQ3: How do the caregivers perceive the patients coped with the diagnosis?

RSQ4: How have caregivers addressed the barriers for patients diagnosed with 22q?

RSQ5: How do caregivers plan to address these barriers and quality of care issues in the future?

RSQ6: How did healthcare providers' education and experiences impact the diagnosis and treatment of 22q?

RSQ7: Why is 22q often under- or misdiagnosed despite being relatively common?

RSQ8: How can increased education of healthcare providers and the public help better identify and address these barriers related to this common yet sometimes unidentified disorder?

Case studies use multiple sources of evidence in order to triangulate to determine the consistency of the findings (Yin, 2014). To answer the research questions, I used a qualitative case study approach with 10 in-depth face-to-face interviews in-person or on videoconference. These interviews were conducted with caregivers to individuals diagnosed with 22q. Individuals interviewed self-disclosed their locations, which ranged from the east coast to the west coast of the United States. In this study, I also examined and analyzed public documents used for diagnosis and treatment. Finally, I reviewed public artifacts that communicated the concerns of caregivers of those diagnosed with 22q.

Initial Suspicion of 22q

For newborns, congenital cardiac defects associated with neonatal hypocalcemia are the most frequent clinical features leading to a 22q diagnosis (Cancrini et al., 2014). RSQ1 explored the ways and timelines in which these individuals initially received a diagnosis. The age of diagnosis ranged from one week to eight years old. Consistently, the only at-birth medical condition that triggered a diagnosis was a heart defect. Heart disease was found in approximately 77% of individuals diagnosed with 22q (McDonald-McGinn & Sullivan, 2011). Based on the data in this study, the heart condition was found first in five out of ten cases I interviewed. The 22q diagnosis was found after an apparent heart disorder.

Those affected with a palate abnormality as the primary condition were diagnosed later. Recent studies including a large population showed that 71% had palate abnormalities, including velopharyngeal insufficiency, submucous cleft palate, overt cleft

palate, and cleft lip and palate (McDonald-McGinn & Sullivan, 2011). Additional clinical features found in the literature may include a compromised immune system, including an absent or small thymus; characteristic facial features, including the eyes, nose, and ears; palate deficiencies, including cleft lip and palates; psychiatric disorders, including schizophrenia; and significant developmental delays (Evers et al., 2014; Furuya et al., 2015; Mariano et al., 2015). Each of these features was found in at least one of the participants' loved ones in this study.

Caregivers' Coping with Diagnosis

RSQ2 explored the caregivers' coping methods once diagnosis had been made. While caregivers of those diagnosed at birth were scared, a majority of those with a later diagnosis were initially relieved to finally have answers. All caregivers researched the syndrome, conditions, and treatment options associated with 22q. Goodwin et al. (2015) found most caregivers rated the experience of being told of the diagnosis as negative. While this was the situation in this study with those whose loved ones were diagnosed early, as noted, a later diagnosis led to the ability to address previously unexplained medical conditions and concerns.

Healthcare providers should focus on reducing the impact of the stressful news for the caregiver. The unknown implications of the diagnosis can bring distress and grief to caregivers as they may focus on the worst-case scenarios (Goodwin et al., 2017a). In my research, caregivers sometimes spent days and even weeks following the diagnosis waiting to be talked to by a geneticist or genetics counselor. Caregivers shared that the geneticists' and genetics counselors' knowledge regarding 22q was extremely limited.

Fung et al. (2015) shared that genetic counseling may provide up-to-date information but also explained that the extent of genetic counseling could vary, depending on the background of the clinician providing counseling.

Diagnosed Individual's Coping Skills

RSQ3 inquired as to how the diagnosed individual has coped with the diagnosis, from the perspective of the caregiver. Some of those with 22q are not aware of the difference while others struggle, specifically in teen and adult years, to fit in. Social and medical conditions accentuate the differences. Goodwin et al. (2015) found a relationship between the diagnosis experience and parental disclosure of 22q. In recent literature, caregivers typically disclosed the diagnosis to affected individuals between 5-10 years of age (Goodwin, et al., 2015). Goodwin et al. found that 86% of caregivers of individuals with Down syndrome felt prepared to disclose to their child sufficient information while 32.6% of caregivers of those diagnosed with 22q felt adequately prepared. This may be due to the high uncertainty associated with the latter condition (Goodwin et al., 2017a; Mishel, 1981).

Caregivers' Self-Efficacy in Addressing Barriers

RSQ4 asked about the ways caregivers have addressed the barriers for patient treatment. These include seeking additional medical treatment, seeking academic accommodations, conducting research on their own, and finding relevant and experienced specialists to better address serious matters. Caregivers struggle with the stigma and perception of professionals who think their loved one appears to be fully functioning (Goodwin et al., 2017b). In younger children, Goodwin et al. (2017a) found that

caregivers consciously looked at uncertainty as an opportunity, known as reorganizing an aversive experience (Mishel, 1990).

Caregivers' Plan for the Future

RSQ5 involved the ways caregivers plan for the future. This included making appropriate follow-up medical appointments and planning for the unknown. Goodwin et al. (2017b) suggested 22q adults may have hopes for their futures that include goals they lack the physical or mental capability to achieve. Caregivers have the responsibility of bringing the child back to reality without crushing their dreams (Goodwin et al., 2017b). Two participants interviewed in this study had children who wanted to be a veterinarian or veterinarian assistant. One child wanted to be a physician. Caregivers have to balance the dreams of the child with the reality of the child's capabilities.

Perceived Impact of Healthcare Providers' Experience and Education

RSQ6 looked at the caregivers' perspectives of the healthcare providers' education and experience based on the caregivers' personal experiences with diagnosis and treatment. Previous research found the diagnosis experience negative due to low-quality information and poor understanding from the healthcare professional, leading to extreme anxiety, stress, and concern (Goodwin et al., 2015). While caregivers interviewed in this research felt the diagnosis process was less than it should have been, the treatment process, once the diagnosis was made, seemed to be satisfactory to the majority of caregivers interviewed. Caregivers lose themselves in their child's needs as they become the expert due to professionals lacking knowledge about the syndrome.

(Goodwin et al., 2017b). It is important that professionals educate themselves on the appropriate treatment of these patients.

Perceived Contributions to Under and Misdiagnoses

RSQ7 represented an attempt to understand why a condition as common as 22q is often underdiagnosed or misdiagnosed. Due to a lack of follow-through by physicians on palate abnormalities and a lack of public knowledge of 22q, early detection is less than appropriate for this most common chromosomal deletion syndrome. In contrast to Down syndrome, awareness of 22q is minimal. Caregivers in recent research suggested there was lost time due to medical professionals' lack of understanding, delaying when they could get help for their children (Goodwin et al., 2017b).

Increased Education and Knowledge of Healthcare Providers and the Public

RSQ8 attempted to address the lack of education of healthcare providers and the public. Medical professionals have contributed to feelings of sorrow and loss and the sense of unpreparedness of caregivers, due to their ignorance of 22q or dismissive attitude toward the concerns of these caregivers (Goodwin et al., 2017b). In order to properly diagnose adult individuals, physicians need a high index of suspicion in the presented clinical features, as diagnosis can be difficult (Friedman et al., 2016; Vogels et al., 2014). Health care providers should recognize and rectify any knowledge gaps in order to increase suspicion (Goodwin et al., 2017b). Recent research suggested new innovations including facial analysis technology that can assist in diagnosis (Kruszka et al., 2017). Healthcare providers and parents can lead the way in better communicating the prevalence of this syndrome (Goodwin et al., 2017b). Medical professionals should

acknowledge any gaps in their own knowledge when presented with medical conditions that are unfamiliar (Goodwin et al., 2017b). Clinicians can access recently published research, specifically guidelines for managing adults with 22q (Fung et al., 2015). Through knowledge and technology, clinicians can potentially make an earlier diagnosis in order to address 22q comorbidities on an individual basis (Kruszka et al., 2017). This would not only help their patients who are currently diagnosed with 22q, but could increase the index of suspicion with future patients.

Limitations of the Study

This study was a qualitative case study. The purposeful sampling was specifically used to target a population that has experience with this phenomenon. A limitation was that this study was focused on a small sample of 10 interviews. However, utilizing multiple data sources allowed triangulation to occur, strengthening the study. Due to geographical and cultural differences, this study may not fully represent the perspectives of all caregivers of those diagnosed with 22q. As a caregiver of someone diagnosed with 22q, I have personal experience with this phenomenon. While this could have been a bias in my data collection and analysis, I feel it benefited the research as the participants felt comfortable and understood as I conducted the interviews. Audio recording and member checking negated the possibility of researcher bias in this study. Due to the specific nature of the study and small sample size, this study may not be generalizable to other groups of individuals. This study may not represent the perceptions and beliefs of caregivers of 22q individuals in the larger population. However, the benefit for those

around the globe is that this small qualitative case study offers a template that can be replicated, reaching other diagnosed individuals, caregivers, and healthcare providers.

Recommendations

This study was an introductory look at the barriers and quality of care issues for those diagnosed with 22q, from the perspective of the caregiver. The findings of this study, based on the constructs of the uncertainty in illness theory, contribute to the increasing need of future research that assesses the current knowledge of 22q with healthcare and educational providers. It is recommended that additional research be conducted to drill further into the anomalies found in those with a delayed 22q diagnosis. Research should be conducted to assess the role of educational providers when areas, specifically speech and learning disabilities, are prominent. Future quantitative, qualitative, and mixed methods research should be conducted to provide the diagnosis and quality of care issues from the perspective of the healthcare provider. Additional research is recommended for different ethnic groups, based on differences found in cultures regarding diagnosis and health care treatment. Finally, geographical research could be conducted, in relation to individuals in close proximity to 22q.

Continuing medical educational opportunities should communicate these research results. Another recommendation is that learning sessions be conducted at medical and educational conferences. A virtual learning network guided practice model that focuses on medical education and care delivery may be appropriate to further educate providers. Extension for Community Healthcare Outcomes (Project ECHO), should present education through shared networks to health care and educational providers in order to

offer a collaborative approach to provide better education and care, increasing positive healthcare outcomes. This initiative could propose future policy changes that will lead to positive social change.

Implications

The findings of this study have the potential to create positive social change for a population that often feels neglected, misunderstood, and insecure, due to the nature of medical conditions that are under- or misdiagnosed. Findings contribute to the existing literature that suggests healthcare providers need a greater suspicion of the predominate anomalies that are directly related to a 22q diagnosis. Goodwin et al. (2017b) found that participants' knowledge grows and these caregivers wish to pass on their knowledge and expertise through volunteering.

Caregivers in this area of diagnosis were excited to share their perceptions with me, as a researcher. They continuously stated that they were eager to make a difference for other caregivers and patients who deal with this often-unfamiliar genetic disorder. The results of this study will be disseminated to the participants and the two individuals who did not meet the criteria. The goal is to provide the results not only for their own records, but to also show what the end perceptions of all participants were. This may encourage additional opportunities for further exploration of related symptoms and/or treatment. Caregivers can make a difference by spreading awareness and continuing to participate in developing research projects, bringing positive outcomes to influence areas that lead to positive social change. Future caregiver social change opportunities include the establishment of a caregiver advocacy program within the 22q community.

The largest contribution to positively affect social change is that from providing the opportunity to influence a more timely and accurate diagnosis. In order to specifically influence this, it is important that physicians have the knowledge necessary to create a suspicion. The knowledge gained in this study can be used to influence not only more education for health care providers, but also a policy and best practices standard of care for individuals who possess the most frequently diagnosed conditions associated with 22q. The policy that will ultimately be pursued is one that recommends 22q deletion syndrome screenings at birth, with a best practices standard of care disseminated to caregivers. This policy proposal will start within a local healthcare clinic system and hospital. The next step will move into the tribal clinic systems, followed by a larger effort across the United States.

Scholarly writing can be a form of social change when it could potentially positively affect an outcome. This study will be submitted to a peer-reviewed medical journal, specifically directed to the healthcare professional population where greater knowledge is needed. A presentation will be held at the next family practice physician conference in my local area. This not only serves as an educational opportunity, but the chance to spread awareness to those who initiate the diagnosis. Finally, a virtual educational program that trains rural physicians will host a seminar featuring the findings of this study.

The long-term study implications for social change are key for caregivers in knowing the best treatment options, best institutions for primary and secondary education, and most significant and applicable life skills for their loved ones affected by

22q. This study shares the experiences of those closest to the patient affected by 22q. It communicates their frustrations, concerns, fears, and emotions in obtaining a diagnosis and creating and maintaining an ongoing treatment plan. Enhancing awareness through appropriate research, scholarly publication, and physician education will lead to better clinical outcomes through a proper diagnosis.

Conclusion

In conclusion, based on the data analysis conducted in this study, I was able to understand the fear, anxiety, sadness, and anger that are present for those caregivers who not only received a late diagnosis, such as me, but those who found out early on. Mishel (1981) described the inability to find a meaning or answers of an illness, due to uncertainty and unpredictable symptoms, explanations, information, and unclear feedback. This theory seemed to be founded on the feelings of caregivers of individuals with 22q. With the initial diagnosis being so misunderstood by healthcare providers and caregivers, the treatment and future forecasting are stressful and full of uncertainties. Positive social change includes the opportunity to increase awareness through scholarly publications, healthcare professional training, caregiver advocacy groups, and policy changes.

Although there are many anomalies that were studied and each individual caregiver had different urgencies to deal with, there are many similarities with these cases. All caregivers provided their loved one had a developmental delay, with the exception of the newborn that was three months old. All caregivers specified their loved ones had facial and/or other physical characteristics. All caregivers had a specific medical

emergency that led to the diagnosis of 22q. The emotional road traveled by all 10 was very similar. Finally, all caregivers interviewed were extremely willing to share their knowledge with others. Although the differences with this syndrome are significant, the likenesses that bind these individuals are strong.

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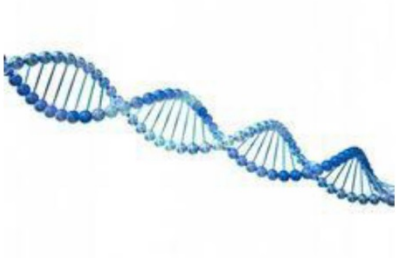
Appendix A: Recruitment Flyer

Are you a caregiver of someone diagnosed with 22q?

You are invited to participate in a research study conducted to understand the barriers and quality of care issues for individuals diagnosed with 22q. This Walden University student research is being conducted in partial fulfillment of Ph.D. requirements.

If you are at least 18 years old and a caregiver to someone diagnosed with 22q, you could qualify to participate in this study.

Contact Bavette Miller at
918-230-3412 or email at
barbara.miller6@waldenu.edu



Appendix B: Interview Guide

1. At what age did you find out your loved one has 22q11.2 deletion syndrome?
2. How did you find out about the diagnosis?
3. What were the biggest concerns you had when your loved one was diagnosed with 22q11.2 deletion syndrome?
4. In what ways did you address the diagnosis?
5. How did your loved one cope with the news, once diagnosed?
6. To which healthcare, educational, or other professionals did you communicate the diagnosis?
7. What is the nature of your loved one's physical medical condition?
8. What is the nature of your loved one's mental cognitive ability?
9. What accommodations have been made for the affected individual?
10. In the future, what additional issues do you foresee?
11. How do you expect your loved one to transition into adulthood?
12. How sure are you that your loved one's healthcare providers have enough knowledge and experience to properly treat you/your child?
13. What resources did your loved one's healthcare providers provide upon diagnosis?
14. How prepared did you feel in addressing possible additional conditions related to 22q?
15. In what ways, if any, could your loved one's healthcare providers have better prepare you for academic and adulthood transitions?