**Clinical Barriers for Caregivers of those diagnosed with 22q Deletion Syndrome**

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## Abstract

A 22q Deletion Syndrome (22q) diagnosis at any age is life-changing for the affected individual as well as the caregiver. Short of a recognizable anomaly, caregivers may spend decades searching for medical solutions that go unanswered. The purpose of this qualitative research was to explore insights of caregivers of those affected with 22q. Interviews of caregivers of 22q individuals were conducted face-to-face in-person or virtually. In this research, caregivers were defined as any family members who regularly provide direct care to a 22q affected individual. These caregivers specifically assist with day-to-day living. All caregivers in this study were parents. This study was guided by the Uncertainty in Illness model, with data analyses conducted through NVivo coding. Based on the caregiver perceptions articulated in this research, the emotional pressure on caregivers was apparent. Five out of 10 participants had a child born with congenital heart disease, leading to an early 22q diagnosis. Five out of 10 were diagnosed with 22q post-birth, following an indication of other serious medical conditions. A greater inquisitiveness could have led to an earlier diagnosis, leading to improved clinical outcomes.

## Introduction

Caregivers have a great responsibility meeting the developmental, physical, and emotional deficiencies of their loved ones diagnosed with a rare genetic disorder.[1] In order to meet the high demands required for appropriate and timely treatment, a timely and accurate diagnosis is necessary. To acquire a timely and accurate diagnosis, healthcare providers must have an awareness of clinical presentations that could be suspect of a more serious underlying health condition.[2,3] An increase in physician awareness and knowledge has shown to increase the likelihood of a geneticist referral, which would lead to a more timely diagnosis.[4]

Some experts find the prevalence of 22q to be 1:4,000[5] while others report the incidence could be closer to 1:1,600, due to misdiagnoses.[6] With the varied prevalence, an accurate diagnosis is important for treatment. Clinical features vary with 22q but the possibility of more than 180 anomalies could be presented within any case.[6] The most common feature presented is congenital heart disease[7] but other prevalent clinical indicators include developmental delays, palate abnormalities, psychiatric disorders, facial dysmorphism, and immunodeficiency.[8] Developmental deficiencies, cognitive deficiencies, and psychiatric onsets may be represented throughout all ages in childhood, adolescence, and teenage years. An increased risk of psychiatric disorders including anxiety, depression, bipolar disorder, and schizophrenia manifests in adulthood.[9] Miller[4] found that 105 out of 107 healthcare providers surveyed gained some to extensive knowledge regarding 22q in a one-hour continuing medical education seminar offered at an annual conference held in Tulsa, Oklahoma. This significant number demonstrates the importance of basic education for providers, which can lead to earlier suspicion and diagnosis.

In researching the best framework for this study, Michel's[10] reconceptualization of the Uncertainty in Illness Theory was used. This theory construct resides within a theoretical model of uncertainty evaluation, coping, and adaptation.[11] This theory explores the inability to find a meaning or answers of an illness, due to uncertainty in ambiguous and unpredictable symptoms, treatments, explanations, information, and unclear feedback.[10] Recent research used this theory in a related study looking at the often misunderstood and complex health variances found in 22q

patients[12]. The diagnosis and treatment of 22q is unpredictable and often multifaceted. This theory directly relates to the issues related to uncertainty in complex diagnoses.

## Methods

*Population:* The population of this qualitative research study consisted of male or female caregivers who are 18 years of age and older who care for individuals diagnosed with 22q. These English-speaking individuals were located throughout the United States. Participants were delimited to caregivers of an individual diagnosed with 22q. English-speaking male and female participants who are 18 years and older, comprised of any ethnicities, and who self-identified as being a caregiver of an individual diagnosed with 22q. The recruitment strategy chosen for this research study was purposeful recruitment and sampling. The sample size for this research was 10 participants. 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[13] (Patton, 2002). Institutional Review Board approval was obtained through Walden University IRB. Participants were identified and recruited through public 22q Facebook sites. Specific details, deadlines, and contact information of the researcher were shared on the recruitment flyer.

*Instrument*: The researcher was the main research instrument in qualitative inquiry. The researcher was the only person collecting data in this study. Data collection for this case study research included gathering data through one-on-one interviews, documents, artifacts, and documentation of field notes.

## Results

Data analysis started during the participant interviews when recurring themes were noticed and identified. NVivo 11 for PC by QSR International for data storage and further analysis was used. Participant identification was removed and a unique alphabetical letter representing each participant was assigned. Themes were identified and then coded within the areas of the overall research questions in this study (see Table 1).

Table 1: Interview Questions and Themes

| **Interview Question** | **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<br><br>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. |

Table 1 presents the themes of concern for caregivers on the eight questions discussed in interviews.

**Discussion**

The purpose of this qualitative case study was to understand the barriers and quality of care issues of caregivers to those diagnosed with 22q. Within this purpose, the aim was to understand the perspectives, thoughts, and experiences of the process from pre-diagnosis through treatment

and into the adulthood. Caregivers' perceptions of healthcare providers' experience and knowledge, specifically when it came to diagnosing the individual, were explored. The opinions of the current level of treatment their loved one was receiving was further considered. Interview data were stored, organized, and coded through NVivo in order to better identify the common themes amongst the data collected. Caregivers communicated the concern of missed anomalies or late diagnoses of additional anomalies.

*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. Although research bias is possible, audio recording and member checking negated the likelihood of researcher bias in this study. Due to the specific nature and small sample size, this study may not be generalizable to any other groups of individuals. This study may not represent the perceptions and beliefs of caregivers of 22q individuals within 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 and Implications:* 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 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, as was done in previous research.[4] 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 healthcare and educational providers in order to offer a collaborative approach to provide better education and care, increasing appropriate diagnoses and positive healthcare outcomes. This initiative could propose future policy changes that will lead to positive future implications.

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