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Caregiving for Children and Youth with CHARGE Syndrome: Impact of Family Caregiver Quality of Life and Coping Strategies

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Abstract

Individuals with CHARGE syndrome often experience motor delays impacting their balance, flexibility, and hand–eye coordination. Due to the medical complications associated with CHARGE syndrome, 40% of children will not develop functional communication skills and many more will have difficulty with speech and oral communication. Family caregivers play a critical role in the care of children and youths with disabilities, especially CHARGE syndrome. However, there is minimal research on the parental experiences of children and youth with CHARGE syndrome. The purpose of this study was to understand the family caregiver experiences and needs of parents of children and youth with CHARGE syndrome and related disabilities. Six family caregivers of children and youth living with CHARGE syndrome in the United States were interviewed to understand their current quality of life, factors influencing their quality of life, and the coping strategies they use. The interviews were transcribed verbatim and then analyzed using qualitative content analysis. The themes generated from the qualitative analyses of interviews showed that family caregivers experienced significant challenges in their quality of life and faced barriers as they navigated a complex system of care. However, family caregivers described robust approaches such as social support for coping with some of the stresses.

Keywords: CHARGE syndrome; disabilities; child health; children; youth; family caregiving; quality of life; burden; coping strategies

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1. Introduction

Coloboma, heart defects, atresia choanae, retardation of growth, genital abnormalities, and ear abnormalities (CHARGE) Syndrome is a complex genetic disorder characterized by a variety of developmental and health issues, including hearing and vision impairments, cardiac anomalies, and cognitive delays [1]. Due to the unique characteristics experienced by children with CHARGE syndrome they may exhibit delays with balance, hand–eye coordination, and flexibility [2–4]. Currently, CHARGE syndrome affects one in every 10,000 births and happens when a gene mutation occurs during prenatal development [1–4]. Both hearing and vision are affected by CHARGE syndrome, and as a result, may be labeled Deafblind [5]. Forty percent of children with CHARGE Syndrome will have delays or will not develop functional communication and others may have complications with speech and oral communication due to medical complications [6].

Behaviors displayed by children with CHARGE are complex and inconstant. Hair pulling, biting, pinching, kicking, shoving, scratching, throwing objects, smearing feces, undressing, self-injury, and resistance are some physical behaviors that may be exhibited. Verbal behaviors seen may be yelling, complaining, agitation, pacing, invading personal space, repetitive statements or questions, and withdrawal. Some children may be labeled with Psychiatric diagnoses and are often assigned to individuals with CHARGE, with the most common being autism, as well as attention-deficit/hyperactivity disorder, and obsessive–compulsive disorder [6].

Painful experiences such as intensive medical procedures, acute complications, medications, and various therapies (e.g., physical therapy) are common. These individuals may encounter considerable occurrences of intense pain, mostly due to complications related to CHARGE Syndrome which include constipation, migraines, surgery-related pain, chronic otitis media, sinusitis, dental issues, gastroesophageal reflux, and breathing-related pain [7]. All 12 cranial nerves may be impacted in children with CHARGE, yet the extent of the relationship to this extreme discomfort is unknown [8]. Cranial nerve V functioning has been associated with migraine headaches [9]. Over half of these children with CHARGE experience cranial nerve V anomalies [8]. In the Stratton and Hartshorne (2010) investigation migraines were described by the parents as the most extreme and painful medical complication [7].

Individuals with CHARGE also have chronic pain concomitant to their intense acute pain experiences [7]. When the parents rated their child's occurrence of pain for a variety of identified painful experiences, difficulty swallowing, gastroesophageal reflux, breathing difficulty, abdominal migraines, hip/back pain, and muscle pain occurred 95 or more days annually, frequently happening along with one or more other uncomfortable and painful experiences. Pain from surgery is frequent along with persistent conditions that result in chronic pain. Between the ages of 1 and 18, the average number of surgeries reported was 12 (range of 1–47) [7].

Common concerns among family caregivers of children with CHARGE are anxiety, diagnosed anxiety disorders, and anxious behaviors. An investigation of 87 families with children with CHARGE discovered that anxiety disorders happened to be the most common mental health identity [10]. A survey conducted by Blake and colleagues (2005) reported that 43% of family caregivers of individuals with CHARGE shared that their child was diagnosed as having obsessive–compulsive disorder and 37% stated anxiety disorder as a diagnosis [8]. Relatedly, Hartshorne et al. (2016) [11] discovered that 50% of their children with CHARGE Syndrome reported complications related to anxiety and/or obsessive-compulsive behaviors. These studies indicated that anxiety is common in children with CHARGE Syndrome. Other family caregiver populations, including those with family members with dementia and visual impairments, also reported similar findings [12,13]. Undoubtedly, the abundance of research related to medical complications, anxiety, and issues with functional communication indicates a stressful experience for family caregivers of children with CHARGE syndrome.

Parents often develop unique coping strategies to manage the difficulties of having a child with CHARGE Syndrome. These include seeking social support, maintaining an optimistic outlook, and finding meaning in their experiences [14,15]. Resilience is often influenced by family dynamics, including the availability of resources, emotional support from family members, and effective communication within the family [15]. While parents may experience higher levels of stress, many demonstrate remarkable adaptability. This includes redefining their roles and expectations and finding joy and strengths in small achievements [16]. Support from healthcare professionals and access to resources play a crucial role in fostering resilience. Parents value factual information, emotional support, and recognition of their efforts [14].

The purpose of this study was to understand the family caregiver experiences and needs of parents of children with CHARGE syndrome and related disabilities. The research questions were as follows:

1. What is the current quality of life among family caregivers of children and youth with CHARGE syndrome and related disabilities?
2. What factors (e.g., severity of symptoms, age, urban/suburban/rural location, siblings, coping strategies, etc.) influence the quality of life of family caregivers of children and youth with CHARGE syndrome and related disabilities?
3. What type of coping strategies are used among family caregivers of children and youth with CHARGE syndrome and related disabilities?

Results from this study provide an understanding of the overall quality of life and coping strategies employed by family caregivers in managing the needs of their loved ones with CHARGE syndrome and related disabilities.

2. Materials and Methods

2.1. Study Context and Design

This study focused on family caregivers of children and youth living with CHARGE syndrome and related disabilities. Specifically, the study sought to understand their current quality of life, the factors influencing their quality of life, as well as the coping strategies they used in their role as family caregivers. A qualitative descriptive interview study was carried out. A qualitative design was chosen because the aim of the study was to understand the quality of life and coping strategies used by family caregivers of children and youth living with CHARGE. This research approach makes it possible to gain in-depth understanding and lay the foundation for future research on this topic.

2.2. Study Participants and Recruitment

To be eligible, participants must be unpaid family caregivers of a child or youth with CHARGE syndrome or related disability. Therefore, six family caregivers of children and youth living with CHARGE syndrome in the United States were interviewed to understand their current quality of life, factors influencing their quality of life, as well as the coping strategies they use. Participants were recruited through organizations (specifically the CHARGE Syndrome Foundation) that serve individuals living with CHARGE syndrome and related disabilities throughout the United States. These organizations helped send recruitment letters and e-mails to potential participants (family caregivers) to invite them and their loved ones to participate in the study. Once they contacted the research team, potential participants were screened and then enrolled in the study if found to be eligible. We also advertised the study through social media (Facebook/Meta, Twitter/X).

A total of six family caregivers of children living with CHARGE syndrome were interviewed for this study. These participants were located throughout the United States. The interviews took place virtually via Zoom at a time that was convenient for the family caregiver. At least two members of the research team attended each interview. Each qualitative interview lasted between 20 and 30 min. The study received Institutional Board Review approval and participants gave informed consent prior to being interviewed for the study.

2.3. Data Analysis

The qualitative interviews were recorded and transcribed verbatim. Transcripts from the interviews with the six family caregivers of children and youth living with CHARGE syndrome were cleaned and then analyzed by three authors using qualitative description methodology [17]. In the context of this study, qualitative description allowed the use of direct, rich descriptions of experiences and events as lived by the family caregivers of children and youth living with CHARGE syndrome. To begin the process, each author read the transcripts generated from the qualitative interviews independently and then grouped the key information into categories. These categories represented similar meanings in relation to the three research questions that the study sought to address. After their independent analysis of the six interview transcripts included in the study, the authors met to compare their analysis results. Any areas of disagreement were discussed by the three authors until a final consensus was reached on the final themes and sub-themes of the study. The approach utilized for this study enabled the generation of themes relating to the perception of the current quality of life of family caregivers of children with CHARGE syndrome, factors influencing the quality of life, as well as their coping strategies.

2.4. Study Rigor

Several steps were taken to ensure the study's rigor. This is important because the researchers have extensive experience in this field of research. One of the researchers has a track record in family caregiving research. In addition, two researchers have extensive experience in CHARGE syndrome. To ensure a rigorous study, the researchers noted their biases and discussed these as part of the data analysis process. In addition, a critical friend was asked to review the interview transcripts and the emergent themes. This helped in ensuring that the emergent themes and their descriptions were close to how the family caregivers of children and youth with CHARGE described them. Also, the researchers established positionality before each interview. In other words, they explained who they were in relation to the research such as educational background, experience with children with CHARGE Syndrome, and why they were completing the research. Specifically, one of the investigators conducts research in family caregiving. Another researcher conducts research in the field of physical education, while the other one is an expert in motor development for children with disability. The final one is a student research assistant, this disclosure ensured the participants knew who they were being interviewed and why.

Further, family caregivers of children and youth with CHARGE were recruited throughout the United States. Recruiting from such a wide geographic area helped to improve the transferability of the findings to family caregivers from diverse settings in the United States. Therefore, this helped to increase the transferability of the findings. In addition, an audit trail of all relevant decisions regarding family caregiver recruitment and participation in the study were kept and available for auditing. The audit was performed by the lead researcher. The audit trail includes the researchers' post-reflections, researchers' meetings, data analyses, and manuscript writing. Therefore, the study achieved confirmability since the findings are linkable to the perspectives of the family caregivers of children and youth with CHARGE syndrome and related disabilities who participated in the interviews [18–20]. Importantly, the interview questions were validated by experts on CHARGE syndrome and one parent of a child living with CHARGE syndrome.

3. Results

Table 1 shows the basic demographic characteristics of the study participants and their dependent children/youth. Further, Table 2 provides a summary of the themes, sub-themes, and supporting quotes from the analyses of the interviews with family caregivers of children and youth living with CHARGE syndrome. Additionally, Table 3 presents themes, sub-themes, and quotes concerning factors influencing quality of life. Finally, Table 4 highlights themes, sub-themes, and quotes concerning coping strategies. The themes and sub-themes are organized by research questions. Five themes emerged from research question #1 regarding the current QOL among family caregivers of youth with CHARGE syndrome including caregiving competence, high burden/stress of caregiving, grief and acceptance, lack of connection with others, and positive experiences and outlooks. Four themes emerged from research question #2 regarding the factors that influence QoL of family caregivers including communication with two subthemes: assertiveness and sign language, flexible jobs, support with two subthemes: paid support and spousal support, and managing medical systems. Two themes emerged from research question #3, social support and stress management with two subthemes wellness routine and walking.

Table 1. Demographic characteristics.

Characteristics	Number (%)
Family caregiver gender	
Male	1 (16.6)
Female	5 (83.3)
Child/youth gender	
Male	4 (66.6)
Female	2 (33.3)
Child/youth age in months (years)	
Average	97.6 (8.1)
Youngest	16 months (1.3)
Oldest	264 (22.0)
Family caregiver Region (U.S.A)	
Northeast	3 (50.0)
Midwest	
South	2 (33.3)
West	1 (16.6)

Table 2. Themes, sub-themes, and quotes concerning current quality of life.

Themes	Sub-Themes	Supporting Quotes
Family caregiver perception of their own caregiving competence influence their assessment of quality of life.		*"I'd say our daughter is on the mild side of a few of the things, heavy on some, and then none of them, and none of the symptoms or whatever the acronym letters are present. So, I'd say, you know, this, the stress is more related to like, are we doing things correctly? Are we getting the early intervention that we need to help set her up, for the best kind of possible future?"* (Interview #1) *"But everything we do, I have to sort. I have an underlying filter based on my kids' needs. . . .and how he might be able to access what we are doing. Would he even want to be there? Would it be stressful? Would it be appropriate for him? So, what we do as a family is often dictated by his needs but also my other kids' needs as well."* (Interview #5)
The high level of burden/stress associated with caregiving affects family caregiver quality of life.	Dealing with the medical system impacts quality of life.	*"The stress is always very high. . . .even my 4 yr old can feel it at times. . .and it's all just trying to minimize that stress level too. it came to a head again last week when we received an order and it was incorrect, these are monthly supplies there shouldn't be hiccup with these, but it's just like everything you should be able to rely on, it falls through, so you're just constantly having to re-do, re-do, re-do, and it's a very high level of stress and anxiety, and just trying to manage it and figure it out."* (Interview #2) *"We don't have to have a nurse go to school with her, so it was extremely stressful just a year ago where if the nurse called off, she wasn't able to go to school, so we were missing work. If we didn't have the overnight nurse, we were staying up with her. Now, if she gets sick, we have to sleep on her floor, she still has to be on monitors. It's definitely stressful."* (Interview #4) *"My threshold is different than maybe typical people and I'm just used to it. . .I don't feel like I really notice the stress but like he was in the hospital a year ago and it had been a long time since he'd been hospitalized. That experience was incredibly stressful for me. I get very anxious, I don't sleep you know?"* (Interview #5) *"It's worse when we're trying to deal with like, the cluster that is the medical system in terms of who's talking to who, how is insurance involved?"* (Interview #1)

Table 2. *Cont.*

Themes	Sub-Themes	Supporting Quotes
There is grief associated with caregiving; however, acceptance eventually sets in.	Coping strategies help ameliorate the impact of caregiving on quality of life.	*“I think a big factor for many families is they’re dealing with grief. The loss of what they thought they should have or could have with a child that doesn’t have disabilities. I feel like the part that I didn’t really have to experience because I work in the disability community but then I had to deal with it with my husband even though he works with disability… not as much with him but my mother who had lived with us and she was a caregiver to help us in the early years. But it’s really about managing other people’s feelings and that’s a big burden. But I think when parents are dealing with the grief of what they lost or what they don’t know or what to expect.”* (Interview #6) *“I mentioned like drinking more or just kind of like distracting myself from the world around me, not trying to be as connected to family and friends because you kind of feel othered in a sense even though you kind of have unbelievable and amazing support system from friends and family.”* (Interview #1) *“I’m the only one that learned any sign language and ___________ only signs. My husband knows very basic signs, but none of our other family has really learned sign language.”* (Interview #4)
Lack of connection with others affects family caregiver quality of life.	Isolation from other people affects family caregiver quality of life. People problems can worsen family caregiver quality of life.	*“It kind of makes me feel a little sad about things and it does impact how ______can interact with our friends that have kids the same age. They don’t understand what _____ is saying.”* (Interview #4) *“Being a caregiver takes a lot of effort and it’s expense on your mental health and a lot of times of not being able to, you know, connect with other friends who have kids around the same age, or even your spouse.”* (Interview #1) *“That people just aren’t comfortable or familiar with CHARGE syndrome and I’ve had people be nice to my face but at the end of it they ’re like we can’t help you so it’s a huge waste of time.”* (Interview #6)
Having positive experiences and outlooks helps improve family caregiver quality of life.		*“I guess I don’t really necessarily feel like it’s a burden and maybe that’s just my outlook so I feel, I’ve always felt like caregiving for ___________ is my greatest act of worship. It truly is a gift to be able to care for him.”* (Interview #5) *“These kids are scary, their medical history is like this long form their first month of birth, but they’re very smart and very resilient kids, they figure it out, they adapt, and that’s just what __________ has done, he can walk but it might not be safe, he can see and he can hear, but he’s still deaf, blind, there’s just all these dif-ferent things.”* (Interview #2)

Table 3. Themes, sub-themes, and quotes concerning factors influencing quality of life.

Themes	Sub-Themes	Supporting Quotes
Family caregiver communication skills influence their quality of life.	Knowledge of sign language influences family caregiver quality of life. Assertiveness helps family caregiver quality of life.	*“I know I was talking about family learning sign language, and they have had the opportunity, there are classes offered and things like that, but it would be nice if there was more of that within our family.”* (Interview #4) *“And I can train them in sign language but I’ve had people who can sign beautifully and there’s areas that I can’t train them in with care and compassion. So, I always say that to people, like if you’re interested in learning sign, I can train you in that but do you have these other qualities? Because that I can’t fix.”* (Interview #5) *“I’m just more assertive because I learned I just need to be upfront with so many things that way my time and their time isn’t wasted.”* (Interview #6)
Having a flexible job influences family caregiver quality of life.		*“Super lucky that I work from home. Super lucky that my wife works at home because we’re able to put box on our calendars and run to the hospital which is 10 min away versus if I had to go from my office, but if I was still living in New York, we moved about four years ago. There’s no way I’d be able to do any of the care stuff that we’re doing. No way, I’d be able to get out of the office to go to hospital to go to an appointment. Um, so Again, quality of life, in terms of that, it makes it a lot easier to do stuff as a caregiver, but if I had to be, you know, working in a store, there’s no way I’d be able to stop midday when people have appointments available to go do a lot of this stuff.”* (Interview #1)

Table 3. *Cont.*

Themes	Sub-Themes	Supporting Quotes
Availability of support influences family caregiver quality of life	Availability of paid support influences family caregiver quality of life. Availability of spousal support influences family caregiver quality of life.	*"We have been fortunate to have nurse support along the way. . .we actually have a Medicaid based program where it's called IHSS and there is another one called WPCS and I will say this has helped my stress level because it supports our family's financial well-being. The state has these programs available so now my husband, he's a stay-at-home dad now, he used to be a PE teacher but now he's a stay at home caregiver and so he gets paid through one program and then they just allowed the other parent who works who had employment outside to do the other program. So, we both get paid to do around the clock nursing-tech care for her at this point."* (Interview #6) *"We have a nanny. Fortunate enough to have someone who was really interested in kind of caring. . .on a day-to-day basis that, you know, she's cared for other kids or friends who've had disorders or similar type things."* (Interview #1) *"I take on a bulk of the medical aspect, my husband will do things like, maybe make his feed for his day, or we try to divvy up, we call them ______________ chore for the night or the day before to prepare for the next day, but if we were to have two people managing, scheduling, or any type of appt. for _____________, there's no way, I'm also the main "caller" of emergencies, I kind of feel it out, I figure out if we're going to hospital what our next step is, is we're moving to sick plan, if we're not moving to sick plan."* (Interview #2)
Managing medical systems brings challenges that affect family caregiver quality of life.		*"They all share notes, I know they share notes, I see the notes, but doctors still come in and asks us if _______ever had an eye exam, because they didn't read his chart. . .that says he's blind, or they ask us, or they start talking to ___________ as if he can fully hear him, and then I have to remind him "you're gonna have to talk louder, he's deaf". Its just A, B, C, its just things like that, if they could just take two seconds to look into his chart and read about medications or procedures he's had, it would be a lot less of a headache on us as parents to have to go through everything again, and then have to repeat ourselves for the next doctor coming on, and then the nurse didn't get that insight, and so we're repeating it to her, it's just things like that or G-tube care, I've pretty much, do all of ____________ feeds now and medications because when they do it at the hospital they're not maybe specially trained in G-tube care, or ton he basics of how to use a G-tube and then they're feeding the bed with pain meds or he's not getting the feed, or again I'm getting yelled at about nutrient when he has so many different teams looking at nutrition for him."* (Interview #2) *"I'm still doing a lot of the paperwork because we don't have people that should be doing it that are getting paid to do it but then there is also a super decrease in the workforce, so again systemic issues that I can't change in my little bubble."* (Interview #4)

Research Question 1: What is the current quality of life among family caregivers of children and youth with CHARGE syndrome and related disabilities?

Regarding this research question, the results show that family caregiver quality of life is affected by the family caregiver's perception of their own competence. Also, a high level of burden negatively affects a family caregiver's quality of life and a lack of social connections with others makes it worse. Despite the initial grief, acceptance eventually sets in with some family caregivers reporting positive experiences.

Research Question 2: What factors (e.g., severity of symptoms, age, urban/suburban/rural location, siblings, coping strategies) influence quality of life of family caregivers of children and youth with CHARGE syndrome and related disabilities?

Regarding this research question, the results show that family caregiver quality of life is influenced by several factors including family caregiver communication skills, job flexibility, availability of support, and the ease of managing medical systems.

Research Question 3: What type of coping strategies are used among family caregivers of children and youth with CHARGE syndrome and related disabilities?

Regarding this research question, the results show that family caregivers with strong social support and effective stress management techniques can cope better with the negative impact of caregiving.

Table 4. Themes, sub-themes, and quotes concerning coping strategies.

Themes	**Sub-Themes**	**Supporting Quotes**
Social support helps ameliorate some of the negative impact of family caregiving.		*"The charge syndrome network is fabulous, the Facebook group is great, but it's also the other disability stuff that gets really heavy and lost that isn't CHARGE specific. You're having to manage so many systems early on in figuring that out."* (Interview #6) *"We've made connections with others who have kids with CHARGE and, you know, see what they're going through and some of those things, and it's good to an extent to have those connections because you can relate to some of the stuff that's going on."* (Interview #1)
Effective stress management techniques help reduce the impact of caregiving on family caregiver quality of life.	Walking helps improve family caregiver quality of life. Checking out and taking short breaks helps family caregiver quality of life.	*"Feel like I have a pretty good quality of life considering. We travel, we travel as a family. I go to counseling but I'm not on medication. I'm not sure. It's not any worse than the next person"* (Interview #5) *"Getting outside as much as I can. I grew up in the Catskills so whenever I can get out, go for a walk. I think those are the things that bring me the most peace when I need it."* (Interview #4) *"You could cancel a month. Depends on, you know, where we're at mentally. That's typical sometimes. It's the, I just need to sit and watch TV endlessly and like be on my phone and not talk to anyone."* (Interview #1)

4. Discussion

The present study examined the experiences of family caregivers of youth with CHARGE syndrome to better understand the current QoL, the factors influencing QoL, and the coping strategies among the family caregivers and their children with CHARGE syndrome. Overall, the results of the first research question show that family caregivers' quality of life is affected by the family caregiver's perception of their own competence. Also, a high level of burden negatively affects a family caregiver's quality of life and a lack of social connections with others makes it worse. Despite the initial grief, acceptance eventually sets in with some family caregivers reporting positive experiences. For the second research question, the results show that family caregiver quality of life is influenced by several factors including family caregiver communication skills, job flexibility, availability of support, and the ease of managing medical systems. Finally, the results of the third research question show that family caregivers with strong social support and effective stress management techniques can cope better with the negative impact of caregiving.

Youth with CHARGE syndrome have a higher incidence of anxiety-related disorders [8,11] which is also prevalent in their family caregivers [10]. With the many medical and behavioral challenges as well as sensory impairments related to CHARGE syndrome, there is a lot for family caregivers to manage. High anxiety has also been reported for family caregivers of other populations including dementia and visual impairments [12,13].

The first research question examined the current QoL among family caregivers and their children with CHARGE syndrome with five themes emerging: caregiving competence, high burden/ stress of caregiving, grief and acceptance, lack of connection with others, and positive experiences and outlooks. Many of the family caregivers in the present study questioned their medical choices. Choices related to surgeries, medications, feeding tubes, cochlear implant surgery, growth hormones, and the many specialists for each medical issue are constant. The extreme stress of parents of children with CHARGE syndrome was also found in a study by Reda and Hartshorne (2008) [21]. These findings are somewhat different from the findings of Wulffaert et al., (2009) [22] who found that the main source of stress for parents of children with CHARGE Syndrome was behavioral issues. The decision fatigue was real, and the worry was constant for these parents in the current study. This is similar to the findings of Reda and Hartshorne (2008) [21] who determined that the caregiving responsibilities of a child with CHARGE syndrome are directly connected to higher perceived stress levels than caring for children without CHARGE.

Grief and acceptance were common themes for the family caregivers which is not uncommon for caregiving for other populations. Parents of children with severe disabilities can endure feelings of significant loss resulting in grief. This grief can significantly impact their QoL as well as their relationship with other family and friends [23]. Lond and Williamson (2020) [24] found themes of "moving through denial toward acceptance"; "confronting and managing ambiguous loss"; and "becoming an expert carer" for family caregivers of partners with acquired brain injuries. They recommended interventions to facilitate hope and acceptance while resolving feelings of grief and loss for family caregivers faced with permanent disability (ies) of their loved ones and to focus on developing caregiving practices that are sustainable. Similar findings are found with family caregivers for children with mental illness [23]. Unfortunately, parental loss and grief are not included in many support services.

Parents of children with severe disabilities often experience unique forms of grief and loss. This grief is not necessarily tied to the loss of a loved one but rather to the loss of expectations, dreams, or the envisioned future for their child. Parents may feel a mix of sorrow, guilt, and even shame for grieving while their child is alive and loved. This grief can resurface at different stages of the child's development. Parents often experience recurrent grief as their child grows, especially during developmental milestones or transitions. This grief is influenced by personal and societal factors, as well as the evolving understanding of their child's condition [25]. Healthcare professionals can address grief and loss with parents of children with severe disabilities through supportive interventions to mitigate anticipatory and illness-related grief [26].

While all family caregivers discussed the high stress that can be associated with caregiving, some expressed positive outlooks of caregiving for their child with CHARGE syndrome. One family caregiver asserted, "*I guess I don't really necessarily feel like it's a burden and maybe that's just my outlook so I feel, I've always felt like caregiving for is my greatest act of worship. It truly is a gift to be able to care for him*", and another focused upon their child's strengths '*they're very smart and very resilient kids, they figure it out, they adapt*'. Hastings and Taunt (2002) [27] focused on the positive perceptions of family caregivers of youth with developmental disabilities and proposed that positive perceptions can serve as coping strategies for the family caregivers.

Four themes emerged from research question #2 regarding the factors that influence QoL of family caregivers including communication with two subthemes: assertiveness and sign language, flexible jobs, support with two subthemes: paid support and spousal support, and managing medical systems. In this current study, it was found that children who were non-verbal or children and only used sign language experienced a whole different set of challenges. Some family caregivers asserted that it was challenging not only for family members to communicate with their child but also to find paid support who could communicate with their child. Wulffaert et al. (2009) [22] found that communication disturbances were a source of stress but not one of the major stress-inducing sources. While communication challenges were a concern for several of the family caregivers, one of the main challenges that family caregivers must manage was the medical systems due to the medical complications faced by children with CHARGE Syndrome. Managing the medical system for their child's medical care is a common journey for families with CHARGE Syndrome [6]. Along the same lines, most children with CHARGE Syndrome have many surgeries and procedures and are in and out of the hospital [7]. Having a large support network and having flexible jobs were keys to being able to cope with this very stressful aspect of caregiving. This fact illuminated another theme which was the paperwork and systemic issues related to insurance. Several of the families said it was literally like a part-time job navigating the medical and billing systems. A fact that complicated this issue was that many of the specialists did not communicate with each other and caregivers would have to go to separate specialists with separate co-pays and treat each part of the body individually. The most successful experience was when one parent had a hospital that looked at the whole child at once and the medical team worked together. These issues navigating the medical system are common and were also discussed in a manuscript by Hartshorne and Slavin (2023) [28]. One parent suggested specifically that a multidisciplinary approach be taken so all medical staff work together as a team. This is similar to the suggestion by Gabriel et al., (2017) [29] who suggested using a systematic approach with the medical professionals to treat children with CHARGE syndrome.

Two themes emerged from research question #3, social support and stress management with two subthemes wellness routine and walking. Family caregivers had several good ideas to alleviate some of the stresses and challenges of caregiving their child with CHARGE Syndrome. Many of the family caregivers joined the CHARGE Syndrome Facebook group as parents are very helpful and supportive of each other. Several of the parents also had a social network of other parents of children with CHARGE Syndrome and other complex disabilities. This proved invaluable to several of them as these friends could understand the barriers they faced, and they could have play dates with no judgment. Going to therapy regularly was helpful for several of the participants as well as walking and being outside.

5. Conclusions

5.1. Implications and Future Directions

These current findings add to the limited literature on the quality of life of family caregivers of children with CHARGE Syndrome [21–24]. The study highlights the various factors that influence the quality of life of family caregivers of children and youth living with CHARGE syndrome. The results have implications for research and practice since many family caregivers rely on support from institutions to meet the demands of their role. Further, the major issues and suggestions here should be further studied so a comprehensive approach can be developed to support these families. Future research should include more family caregivers. It should highlight the specific sources of stress and anxiety. It should also look at a family systems approach and determine the best practices to alleviate these very real factors that create such quality-of-life issues for family caregivers of children with CHARGE Syndrome.

5.2. Strengths

The strengths of this study are noteworthy and provide a robust foundation for understanding the experiences of family caregivers of children with CHARGE syndrome and related disabilities. Specifically, this study addressed critical aspects of family caregiver experiences and needs. The inclusion of detailed research questions allowed for a thorough exploration of various dimensions, including quality of life, influencing factors, and coping strategies. This holistic approach enhanced the depth of the study. By examining practical factors such as location (urban, suburban, rural), sibling dynamics, and coping strategies, this study directly addressed issues that impact family caregivers' daily lives. Furthermore, this study adds valuable insights to a relatively under-researched area, filling gaps in the existing literature on the quality of life and coping strategies of family caregivers of children with CHARGE syndrome. The findings have the potential to inform healthcare professionals and policymakers in developing targeted interventions and support systems to improve the well-being of these family caregivers.

5.3. Limitations

As with most qualitative studies, the sample size is smaller which can affect generalizability. Considering CHARGE syndrome is a small population, it is difficult to increase the sample size. Transferability may also be affected due to most of the participants being female, which might not fully represent male family caregivers' experiences. It is also important to note that differences in the ages of the children of the family caregivers or the severity of CHARGE syndrome can introduce variability, making it harder to draw consistent conclusions. Additionally, bias can occur in qualitative research as the accuracy of the findings relies on participants being open and honest, which can be influenced by social desirability or discomfort with sensitive topics. The presence and behavior of the interviewer can unintentionally shape participants' responses.

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