

Predictors of Caregiver Burden for Caregivers of Children with DDx3x Syndrome

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Abstract

Rare diseases pose significant challenges to afflicted patients and caregivers [1]. Many rare diseases are diagnosed during childhood, turning parents into lifelong caregivers and significantly increasing parental responsibilities and effort [2]. Families of children with rare diseases often experience a profound impact, especially if the diagnoses are difficult and treatment is complex or not well-developed [3]. This study examined caregiver burden and factors that impact caregiver burden among parents of children with DDx3x syndrome. The significance of this study is to bring awareness to the factors influencing caregiver burden in families with a rare disease. This information can be used for families and those within the health science community to better understand these factors and influences of anxiety, depression, and social support upon overall caregiver burden. A glimpse into the results indicated that depression is negatively related to anxiety ($r = -0.372$, $p = 0.008$). Depression is positively related to care burden ($r = 0.285$, $p = 0.048$). Another interesting aspect of the depression variable is that it positively relates to social support ($r = 0.334$, $p = 0.016$). Income and depression were both associated with caregiver burden. This paper examined the relationship between caregiver burden and caregivers of individuals with rare diseases, contributing to future research in the health science community to understand the dynamics of these variables.

Keywords

Caregiver, Burden, Genetics, Mental Health, Rare Disease

1. Introduction

Approximately 25 million individuals in the United States and 400 million worldwide are afflicted by a rare disease [4]. As of 2022, more than 7000 rare diseases

have been identified, although the genetic causes for many remain unknown [4]. According to Khoury [4], a rare disease affects fewer than 200,000 people. Rare diseases pose significant challenges to afflicted patients and caregivers [1]. Many rare diseases are diagnosed during childhood, turning parents into lifelong caregivers and significantly increasing parental responsibilities and effort [2].

Families of children with rare diseases often experience a profound impact, especially if diagnoses are delayed, and treatment is complex or not well-developed [3]. The Centers for Disease Control and Prevention (CDC) reported that families with children afflicted by a rare disease commonly experience reduced quality of life, difficult life circumstances, and frequent barriers compared to families with neurotypical children [5].

The research presented in this paper analyzed the burden experienced by caregivers of children living with the rare disease DDx3x, which is caused by genetic mutation(s). This study used recent data on caregiver burden and considerations such as caregiver sociodemographic factors, social support, and mental health.

2. Literature Review

Prior research has documented the caregiver burden experienced by family members who provide long-term care for a family member [6]. Caregiver burden is defined as a strain experienced by a person caring for a disabled, chronically ill, or elderly family member and is considered a multidimensional response to psychological, physical, emotional, social, and financial stressors associated with caregiving [6].

Caregiver burden refers to the responsibilities and limitations placed on caregivers and their families [7]. It is a mental, physical, and social reaction to being a caregiver [8], arising from a lack of balance between caregiving duties and personal needs [8]. It includes both objective and subjective dimensions. Objective burden includes time and effort required to meet the patient's needs, including family relationships, financial costs, and social functioning. Subjective burden encompasses the emotional stress imposed on caregivers when facing objective demands such as mental, emotional, and behavioral strain [7].

Several factors contribute to stress, including caregiver characteristics, care-receiver characteristics, social factors, economic factors, and cultural background [8]. Unique challenges arise for family caregivers of individuals with rare diseases [1]. Generally, caregivers are relatives of the patient and take on most caregiving burdens without financial compensation [9].

The caregiver's role in managing the disease includes both physical and psychological care of the patient, impacting the care that caregivers can provide [10]. Those caring for individuals with rare diseases face additional challenges compared to those caring for individuals with more common conditions, including a lack of disease information, delays in diagnosis, inadequate treatment options, and limited access to healthcare [10].

3. Methodology

3.1. Population of Sample

The sample consisted of a group of caregivers of children diagnosed with the rare genetic disorder DDx3x and was a purposive sample. Participants were recruited from a Facebook research group linked to the DDx3x Foundation. The population sample size consisted of roughly 1074 individuals who met the inclusion criteria.

3.2. Instrumentation

The four validated research instruments used in this study were the:

- Burden Scale for Family Caregivers-Short Version (BSFC-s) [11]
- Multidimensional Scale of Perceived Social Support (MSPSS) [12]
- Zung Self-Rating Depression Scale [13]
- Zung Self-Rating Anxiety Scale [14]

These instruments were selected for their strong psychometric foundations and extensive use in caregiver-related research.

4. Results

4.1. Sample Characteristics

Of the 65 individuals who began the survey, 49 (75%) completed it. The majority were female (96%), with only 4% identifying as male. Eighty percent of participants were married, while 20% were unmarried. Ages ranged from 21 to 61 years old, with an average age of 40.5. Length of caregiving ranged from 7 months to 201 months. Income ranged from \$0 USD to \$1.2 million USD.

4.2. Quantitative Findings

Depression was positively related to caregiver burden. It may be that depressed individuals are more likely to perceive caregiving responsibilities as more burdensome than those who are not depressed. Alternatively, caregiving demands themselves may intensify depression.

The results showed relationships between caregiver burden, income, depression, anxiety, and social support. Low levels of depression, severe levels of anxiety, and moderate levels of social support were identified among caregivers.

5. Discussion

The results of this study offer beneficial insight into how caregivers experience and interpret social support, emotional distress, and burden. An unexpected finding in this study was that there was a positive relationship between depression and perceived social support. Although it can be found that social support is usually viewed as a protective factor, there are several explanations that may help clarify this result. Caregivers who feel more depressed may reach out for support, which can increase their perception of support. Another possibility is that friends and family step up when they notice signs of depression in their loved ones, showing

that there is a response to the distress that can help prevent it. Another explanation may be that caregivers place a higher value on the support that they receive when they are having emotional struggles, which can lead to higher support when depression is present. The negative correlation between depression and anxiety also requires a more detailed interpretation. While these two conditions often occur together, stress may be expressed in different ways. Some responses to stress may include exhaustion, sadness, and withdrawal, which align more with depression. Other responses are worry and restlessness that align with anxiety. Since there are different response patterns, it can create an inverse relationship in the data.

The link between caregiver burden and depression was consistent with existing research. This shows how increased caregiving demands can lead to emotional exhaustion. Higher levels of burden show that caregivers are at higher risk for depressive symptoms. This emphasizes the need for early support in caregivers as well as interventions and resources.

Overall, the findings show that there are complexities in caregiver well-being. Social support may increase in response to distress rather than prevent it; caregivers may show emotional strain in different ways, and heavy caregiving responsibilities strongly contribute to depression. These patterns highlight the importance of assessing caregivers thoroughly and providing targeted support that matches their unique needs.

6. Interpretation of the Findings

Depression is associated with caregiver burden, according to the current analysis. Weitzner *et al.* [15] found that caregiving can adversely affect psychological health and increase caregiver burden. Caregiver well-being is influenced by factors related to the patient's condition as well as the caregiver's relational, psychological, and demographic characteristics.

Moderate caregiver burden was identified in this study. Research indicates that individuals living with a rare disease face daily challenges that extend beyond the patient, affecting the entire family [10]. Additionally, depression was positively related to both social support and caregiver burden. Social support plays an essential role in buffering caregivers from stress [16]-[18].

7. Limitations

This study has limitations. Only one genetic syndrome, DDX3X, was evaluated. Expanding research to include multiple syndromes would provide a more comprehensive view of caregiver burden. Additionally, 96% of participants were female, limiting generalizability. The small sample size also constrained the study's breadth and statistical power.

8. Implications and Future Research

High rates of anxiety were identified among caregivers, suggesting the need for stress-management interventions. Income was negatively related to anxiety ($r =$

-0.363, $p = 0.010$), indicating that individuals with lower income experience more anxiety than those with higher income. Income was positively related to caregiver burden ($r = 0.304$, $p = 0.034$), possibly due to pressures of maintaining a career while caregiving.

Future research should focus on increasing participation among caregivers of children with rare diseases, expanding to additional syndromes, and including a more gender-diverse sample. Interventions targeting caregiver support and mental health should be further explored.

9. Conclusion

Individuals living with a rare disease have their own complex experiences; however, the suffering associated with a rare disease impacts entire families [10]. Although limitations were present, this research provides valuable insights for caregivers and health science communities to better understand caregiver burden. Caregiver burden will continue to affect parents of children with rare genetic diseases, but increased knowledge and research can help reduce this burden over time.

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Conflicts of Interest

The author declares no conflicts of interest regarding the publication of this paper.

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