

# Living with Rare Genetic Disorders: A Qualitative Study on Daily Challenges, Social Stigmas, and Coping Strategies

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## Abstract

**Background:** Individuals living with rare genetic disorders face unique physical, emotional, and social challenges. These conditions often lead to social stigmas and discrimination, further complicating the daily lives of those affected. This study explores the lived experiences of individuals with rare genetic disorders, focusing on the challenges they encounter, the social stigmas they face, and the coping strategies they develop.

**Methods:** A qualitative, phenomenological study was conducted at a tertiary hospital, using in-depth, semi-structured interviews with 20 individuals diagnosed with rare genetic disorders. Thematic analysis was employed to identify key themes regarding daily challenges, social stigmas, and coping mechanisms.

**Results:** Four key themes emerged: (1) Daily Challenges, (2) Social Stigmas and Discrimination, (3) Coping Mechanisms, and (4) The Role of Support Networks. Participants reported difficulties accessing medical care, physical limitations, and social isolation. Social stigma was prevalent, particularly in public interactions and workplaces. Coping mechanisms included acceptance, advocacy, and support from family, friends, and peer networks.

**Conclusion:** Individuals with rare genetic disorders face significant physical and social barriers, compounded by public misunderstanding and stigma. However, strong support networks and advocacy efforts play a vital role in improving their quality of life. These findings highlight the need for improved healthcare access, societal education, and emotional support.

**Keywords:** Rare genetic disorders, social stigma, coping strategies, qualitative study, patient experiences, support networks

## Introduction

Rare genetic disorders, defined as conditions affecting fewer than 1 in 2,000 individuals, encompass a broad spectrum of diseases that can significantly impact the lives of those affected. While each disorder may vary in its symptoms and severity, individuals living with rare genetic conditions often face unique challenges, not only medically but also socially and emotionally (Lanar et al., 2020). These challenges are often compounded by a lack of public awareness, limited treatment options, and a shortage of healthcare professionals with expertise in rare diseases (Stoller, 2018).

Living with a rare genetic disorder can be an isolating experience. Patients frequently encounter social stigmas, which stem from public misconceptions, visible differences, or the rarity of their condition. This stigmatization can lead to discrimination, social exclusion, and emotional distress, further complicating the already difficult reality of managing a lifelong condition (Somanadhan et al., 2023). In addition to dealing with the physical aspects of their disorders, individuals often grapple with feelings of alienation from society, as their conditions are poorly understood by both the public and healthcare providers (Currie and Szabo, 2020).

Despite these challenges, many individuals with rare genetic disorders develop coping strategies that help them navigate the complexities of their daily lives. These strategies may include building strong support networks, seeking professional counseling, or engaging in advocacy efforts to raise awareness and reduce stigmatization (Zhu et al., 2017). However, the emotional toll of coping with social stigmas while managing the physical and psychological demands of their condition remains a critical issue that warrants further exploration.

This study aims to explore the lived experiences of individuals with rare genetic disorders, focusing on the daily challenges they face, the social stigmas they encounter, and the coping strategies they employ. By understanding these experiences, healthcare providers and policymakers can better address the needs of this underserved population, promoting more effective support systems and interventions to improve their quality of life.

#### Research Objectives

- To investigate the daily challenges encountered by individuals with rare genetic disorders.
- To explore the social stigmas associated with these conditions.
- To examine the coping strategies employed by individuals to manage the emotional and psychological effects of living with a rare genetic disorder.

#### Literature Review

##### 1. Overview of Rare Genetic Disorders

Rare genetic disorders, though individually uncommon, collectively affect millions of people worldwide. These conditions are often chronic, complex, and debilitating, making diagnosis and treatment challenging (Lanar et al., 2020). A rare disease is typically defined as one that affects fewer than 1 in 2,000 people, though this threshold varies slightly by country (Stoller, 2018). Despite their rarity, the cumulative burden of rare diseases is substantial, impacting not only physical health but also psychological and social well-being. The medical community often lacks specific knowledge about these conditions, resulting in delayed diagnoses and misdiagnoses, which further exacerbates the difficulties faced by patients (Currie and Szabo, 2020).

##### 2. Social and Psychological Challenges

Living with a rare genetic disorder often comes with significant psychological challenges, including feelings of isolation, anxiety, and depression. The rarity of these conditions means that affected individuals frequently struggle to find others who understand their experiences, leading to social isolation (Zhu et al., 2017). Research suggests that this isolation is often compounded by inadequate emotional and psychological support from healthcare professionals, who may not be familiar with the complexities of rare diseases (Pelentsov et al., 2016).

Social stigmas are also a prominent issue for individuals with rare genetic disorders. Stigma can manifest in several ways, including public misconceptions about the condition, discrimination in employment or social settings, and pity or avoidance from others (Somanadhan et al., 2023). As Goffman (2009) outlined in his seminal work on stigma, individuals who are marked by visible or perceived differences are often treated as “other,” which can result in social exclusion. For people living with rare disorders, stigma may be exacerbated by the lack of public awareness, which leads to misunderstandings about the nature of their condition. A study by Fournier et al. (2023), revealed that patients with rare disorders reported higher levels of perceived social stigma than those with more common chronic conditions, indicating the unique burden they face.

Moreover, the stigma associated with rare genetic disorders can have a profound impact on an individual’s self-esteem and mental health. According to Link and Phelan (2001), stigma not only affects how individuals are treated by others but also how they perceive themselves. This internalized stigma can lead to self-isolation, lower quality of life, and increased psychological distress (Currie and Szabo, 2020). The interplay between stigma, isolation, and emotional well-being is an area that has received limited attention in the context of rare genetic disorders, despite its critical importance for patient care and support.

### 3. Coping Strategies in Rare and Chronic Disorders

In response to the challenges of living with rare genetic disorders, individuals often develop coping strategies that help them navigate both the physical demands of their condition and the emotional toll of social stigmas. Coping strategies can be broadly categorized into problem-focused strategies, which aim to manage or solve the challenges posed by the disorder, and emotion-focused strategies, which seek to regulate the emotional impact of these challenges (Lazarus & Folkman, 1984).

Problem-focused strategies include actively seeking medical treatment, joining support groups, and engaging in advocacy efforts to raise awareness and improve public understanding of the disorder (Zhu et al., 2017). For example, rare disease advocacy organizations play a vital role in providing patients and families with resources, emotional support, and a sense of community (Pelentsov et al., 2016). These organizations often serve as platforms for patients to connect with others facing similar challenges, mitigating the feelings of isolation that are so common among this population.

Emotion-focused strategies, on the other hand, include psychological resilience, acceptance, and the use of spiritual or mindfulness practices to cope with the ongoing stress of living with a rare disorder (Zhu et al., 2017). A study by Zurynski et al. (2017) found that patients with rare disorders often relied on emotional support from close family members and friends to manage the emotional and psychological impact of their condition. Additionally, some patients reported finding solace in advocacy, transforming their personal struggles into a mission to educate others and reduce the social stigmas surrounding their condition.

However, not all patients are able to access these coping mechanisms. Research by Somanadhan et al. (2023) suggests that individuals with lower socioeconomic status, limited access to healthcare, or fewer social supports may struggle to develop effective coping strategies. This underscores the need for targeted interventions to ensure that all individuals with rare genetic disorders have access to the resources and support they need to manage both the physical and emotional challenges of their condition.

#### 4. Gaps in the Literature

While the existing literature provides valuable insights into the social and psychological challenges faced by individuals with rare genetic disorders, there are several notable gaps. First, much of the research has focused on the medical aspects of rare diseases, with comparatively less attention paid to the social stigmas and emotional challenges these individuals face. Moreover, the coping strategies employed by patients to navigate these challenges have not been studied in-depth, particularly in relation to how they interact with societal stigmas (Currie and Szabo, 2020).

Furthermore, studies on rare genetic disorders often rely on quantitative methods, such as surveys and questionnaires, which can fail to capture the nuanced, lived experiences of patients. Qualitative research, which allows for a more in-depth exploration of personal experiences, is needed to better understand how individuals cope with the intersection of rare genetic disorders and social stigmas (Fournier et al., 2023). By focusing on these personal narratives, future research can provide healthcare professionals with the knowledge and tools they need to support patients both medically and emotionally.

In summary, the literature demonstrates that individuals with rare genetic disorders face significant social and psychological challenges, including isolation, social stigma, and emotional distress. While many patients develop coping strategies to navigate these difficulties, there is a clear need for more research into the social stigmas associated with rare disorders and how they shape patient experiences. This study seeks to fill this gap by exploring the lived experiences of individuals with rare genetic disorders, with a particular focus on their daily challenges, social stigmas, and coping strategies.

### Methodology

#### 1. Study Design

This qualitative study employed a phenomenological approach to explore the lived experiences of individuals with rare genetic disorders, with a focus on the daily challenges they face, the social stigmas they encounter, and the coping strategies they employ. The phenomenological approach was chosen to capture the rich, in-depth experiences of the participants, allowing for a deep understanding of how these individuals navigate their unique challenges.

#### 2. Study Setting

The study was conducted at a large tertiary care facility that provides comprehensive services for patients with rare genetic disorders. The hospital has a dedicated genetics clinic, which includes genetic counseling services, diagnostic facilities, and support for patients and families. The clinic serves a diverse population of individuals with a wide range of genetic conditions.

#### 3. Participants

A purposive sampling technique was used to recruit individuals who had been diagnosed with a rare genetic disorder and were receiving care at the genetics clinic in the tertiary hospital. The aim was to ensure that participants represented a variety of genetic conditions, ages, and social backgrounds to capture diverse experiences.

Inclusion Criteria:

- Patients aged 18 years and older.
- Diagnosed with a rare genetic disorder (affecting fewer than 1 in 2,000 individuals).
- Regularly attending follow-up appointments at the genetics clinic.
- Willing to participate in a 45-60 minute interview.

#### Exclusion Criteria:

- Patients with cognitive impairments that could interfere with their ability to provide informed consent or participate in interviews.
- Individuals who were not fluent in the primary language of the interview.

A total of 20 participants were recruited for the study, including 12 females and 8 males, ranging in age from 19 to 55 years. The sample included individuals diagnosed with a variety of rare genetic disorders, including cystic fibrosis, Marfan syndrome, Huntington's disease, and Ehlers-Danlos syndrome.

#### 4. Data Collection

Data were collected through semi-structured, in-depth interviews conducted. The interviews were carried out in private rooms within the genetics clinic to ensure a comfortable and confidential setting. Each interview lasted between 45 to 60 minutes, allowing participants the opportunity to share their experiences in detail.

The interview guide included open-ended questions designed to explore participants' daily challenges, experiences with social stigmas, and coping strategies. Sample questions included:

- "Can you describe some of the challenges you face on a daily basis due to your condition?"
- "Have you experienced any social stigma or discrimination as a result of your genetic disorder?"
- "What strategies do you use to cope with the difficulties associated with your condition?"

Interviews were audio-recorded with the consent of the participants and transcribed verbatim for analysis. Field notes were also taken to capture non-verbal cues and contextual details.

#### 5. Data Analysis

The data were analyzed using thematic analysis, following the six-step process outlined by Braun and Clarke (2006). The steps included familiarization with the data, coding, theme development, review of themes, defining and naming themes, and writing up the results.

##### - Step 1: Familiarization with the Data

The researchers read and re-read the interview transcripts to immerse themselves in the data, making initial observations and noting key ideas.

##### - Step 2: Generating Initial Codes

Initial codes were developed by systematically reviewing each transcript and highlighting significant statements and phrases related to the daily challenges, social stigmas, and coping strategies described by participants.

##### - Step 3: Searching for Themes

Codes were grouped into potential themes. For example, codes related to discrimination, isolation, and public misconceptions were combined under the theme "Social Stigma." Similarly, codes related to resilience and seeking social support were grouped under "Coping Strategies."

##### - Step 4: Reviewing Themes

The themes were reviewed and refined to ensure they accurately reflected the participants' experiences. At this stage, any redundant or overlapping themes were collapsed, and key themes were finalized.

#### - Step 5: Defining and Naming Themes

The final themes were clearly defined and named. Four key themes emerged: "Daily Challenges," "Social Stigmas and Discrimination," "Coping Mechanisms," and "The Role of Support Networks."

#### - Step 6: Writing Up the Findings

The themes were organized into a coherent narrative, supported by direct quotes from participants to illustrate their lived experiences.

### 6. Ethical Considerations

Ethical approval for this study was obtained from the ethics committee, ensuring that the research complied with ethical guidelines for conducting qualitative research with human participants.

#### Informed Consent

Participants were provided with detailed information about the study, including its objectives, procedures, and potential risks and benefits. Written informed consent was obtained from all participants prior to their involvement in the study.

#### Confidentiality

To ensure confidentiality, all participants were assigned pseudonyms, and any identifying information was removed from the interview transcripts. Audio recordings and transcripts were stored securely on password-protected devices, accessible only to the research team.

#### Emotional Support

Given the sensitive nature of discussing social stigmas and personal challenges, participants were informed that they could pause or withdraw from the interview at any time without consequence. Additionally, participants were provided with referrals to counseling services available through the hospital, should they feel the need for emotional support following the interview.

### 7. Trustworthiness and Rigor

To ensure the trustworthiness of the findings, the following strategies were employed:

- **Credibility:** Member checking was conducted by sharing the transcriptions with participants and asking them to verify the accuracy of their statements and provide additional insights if necessary.
- **Transferability:** Thick descriptions of participants' experiences were provided to allow other researchers to assess the applicability of the findings to other contexts or populations.
- **Dependability:** An audit trail was maintained throughout the research process, documenting all steps in data collection and analysis to ensure the study could be replicated in the future.
- **Confirmability:** Reflexivity was practiced, with the researchers maintaining a reflective journal to document any potential biases or preconceptions that could influence the interpretation of the data.

### Findings

The thematic analysis revealed four key themes that captured the experiences of individuals living with rare genetic disorders: (1) Daily Challenges, (2) Social Stigmas and Discrimination, (3) Coping Mechanisms, and (4) The Role of Support Networks. These themes, along with their respective sub-themes, illustrate the

complexity of living with a rare genetic condition and the ways in which individuals navigate their unique challenges.

### Theme 1: Daily Challenges

Participants consistently described the ongoing challenges they faced in managing their rare genetic conditions. These challenges were often multifaceted, involving physical, psychological, and logistical difficulties.

#### Sub-theme 1.1: Physical Limitations and Symptoms

Many participants reported struggling with the physical limitations imposed by their condition, including chronic pain, fatigue, and restricted mobility. These physical challenges affected their ability to carry out daily activities, work, and engage in social interactions.

- Participant 3 (Female, 29, Ehlers-Danlos syndrome):

“Some days I can’t even get out of bed because of the pain. I try to push through, but it’s exhausting, both physically and mentally.”

#### Sub-theme 1.2: Medical Care and Treatment Access

Participants also spoke about the difficulties they encountered in accessing appropriate medical care. The rarity of their conditions often meant that healthcare providers were unfamiliar with their specific needs, leading to misdiagnoses, delayed treatment, or inadequate care.

- Participant 7 (Male, 42, Marfan syndrome):

“I’ve had doctors tell me they don’t know enough about my condition to help me. It’s frustrating because it feels like I’m not getting the care I need, even though I’m going to specialists.”

### Theme 2: Social Stigmas and Discrimination

A pervasive theme across the interviews was the social stigma that participants experienced due to their rare genetic disorders. This stigma often manifested in negative social interactions, discrimination, and misconceptions about their condition.

#### Sub-theme 2.1: Public Misunderstanding and Judgment

Participants frequently reported that others misunderstood their condition or judged them based on visible symptoms or assumptions about their health. This lack of awareness often led to uncomfortable or hurtful encounters in public or social settings.

- Participant 10 (Female, 35, Huntington’s disease):

“People don’t understand my condition. They just see the symptoms and assume I’m drunk or on drugs. It’s hard to explain something so complicated when people already have their minds made up.”

#### Sub-theme 2.2: Workplace Discrimination

Several participants shared their experiences of workplace discrimination, where employers or colleagues treated them differently due to their condition. Some were denied accommodations or faced biases that made it difficult to maintain employment.

- Participant 12 (Male, 38, cystic fibrosis):

“I had to leave my job because my boss wouldn’t make any accommodations for my condition. They didn’t understand that I could still work if I had a flexible schedule for my treatments.”

### Theme 3: Coping Mechanisms

Despite the challenges they faced, participants described various coping mechanisms that helped them manage both the physical and emotional burdens of their condition. These coping strategies were essential for maintaining their mental health and quality of life.

#### Sub-theme 3.1: Acceptance and Adaptation

Many participants spoke about the process of accepting their condition and adapting their lifestyle accordingly. While acceptance took time, it ultimately helped them regain a sense of control and peace.

- Participant 6 (Female, 33, Marfan syndrome):

“I’ve learned to accept that this is my reality. It took years, but now I focus on what I can do instead of what I can’t. I’ve adapted my life, and that’s helped me stay positive.”

#### Sub-theme 3.2: Advocacy and Awareness Efforts

Some participants found strength in advocacy and raising awareness about their condition. By educating others, they felt empowered and more in control of their narrative, counteracting the stigma they faced.

- Participant 9 (Male, 41, Ehlers-Danlos syndrome):

“I became involved in advocacy groups, and it’s been a game changer for me. It helps me feel like I’m doing something productive and raising awareness about what people like me go through.”

### Theme 4: The Role of Support Networks

The importance of support networks was highlighted throughout the interviews. Participants described how family, friends, and support groups played a critical role in helping them manage their condition and overcome the challenges of daily life.

#### Sub-theme 4.1: Family and Friends as Emotional Anchors

Family members and close friends were often described as the primary sources of emotional support. Participants noted that having loved ones who understood their condition and provided both practical and emotional help made a significant difference in their ability to cope.

- Participant 2 (Female, 27, cystic fibrosis):

“My family has been amazing. They’ve learned everything they can about my condition, and they’re always there to help me when I’m feeling down or struggling.”

#### Sub-theme 4.2: Peer Support and Community

In addition to personal relationships, participants frequently mentioned the value of connecting with others who shared their experience. Online forums, support groups, and rare disease organizations provided them with a sense of community and understanding that they could not always find elsewhere.

- Participant 8 (Male, 24, Huntington’s disease):

“I found an online support group, and it’s been incredible to talk to people who actually understand what I’m going through. It’s comforting to know I’m not alone.”

### Discussion

The findings of this study shed light on the complex and multifaceted experiences of individuals living with rare genetic disorders. The key themes of daily challenges, social stigmas, coping mechanisms, and the role of support networks highlight the significant impact these conditions have on individuals' physical, emotional, and social well-being. By understanding these experiences, healthcare providers, policymakers,



and society at large can better address the needs of individuals with rare genetic disorders and improve their quality of life.

### 1. Daily Challenges

The daily challenges faced by participants were profound and often multifaceted, encompassing physical limitations, chronic pain, and difficulties accessing appropriate medical care. These findings align with previous research indicating that individuals with rare genetic disorders frequently encounter physical and logistical barriers in managing their condition (Lanar et al., 2020). Participants' reports of frustration over delayed or inadequate medical treatment due to the rarity of their condition mirror findings from Pelentsov et al. (2016), who emphasized the lack of specialized care and knowledge among healthcare providers for rare diseases.

Moreover, the physical limitations imposed by chronic symptoms such as pain, fatigue, and reduced mobility had a substantial impact on participants' ability to engage in everyday activities and maintain employment. This is consistent with studies highlighting the disabling nature of many rare genetic conditions, which can severely affect individuals' functional independence and economic stability (Zurynski et al., 2017). These findings emphasize the need for better access to specialized healthcare and rehabilitation services tailored to the needs of individuals with rare conditions.

### 2. Social Stigmas and Discrimination

Social stigma emerged as a central theme in the experiences of participants, with many reporting negative social interactions, discrimination, and public misunderstanding of their condition. This finding is consistent with Goffman's (2009) work on stigma, which describes how individuals with visible or perceived differences are often treated as outsiders. Participants in this study described feeling judged or misunderstood based on their symptoms, which resulted in social isolation and emotional distress.

The impact of stigma extended to the workplace, where several participants reported discrimination from employers or colleagues who were unwilling to accommodate their needs or misinterpreted their abilities. These experiences echo the findings of Currie and Szabo (2020), who documented the significant barriers that individuals with rare conditions face in maintaining employment due to stigmatization and lack of workplace support. Addressing workplace discrimination through education and legal protections is critical to improving the social and economic outcomes for individuals with rare genetic disorders.

Despite efforts by rare disease organizations and advocacy groups to raise awareness, the persistence of public misconceptions highlights the need for ongoing societal education. Increasing awareness and understanding of rare genetic disorders can help reduce stigma and foster a more inclusive and supportive environment for affected individuals (Somanadhan et al., 2023).

### 3. Coping Mechanisms

Despite the significant challenges they faced, participants demonstrated resilience by adopting various coping strategies. The two primary coping mechanisms identified were acceptance and advocacy. Many participants spoke of the importance of accepting their condition and adapting their lifestyle to manage their symptoms. This finding is in line with the work of Lazarus and Folkman (1984), who describe acceptance as a key emotion-focused coping strategy that helps individuals manage chronic stressors.

Advocacy emerged as another important coping mechanism, with some participants engaging in awareness-raising efforts and community-building activities. As noted by Zhu et al. (2017), becoming involved in advocacy can empower individuals by giving them a sense of agency and purpose, helping them combat the social stigma they face. Through advocacy, participants not only improved their own emotional well-being but also contributed to broader societal understanding of rare genetic disorders.

These findings suggest that healthcare providers should not only focus on the physical management of rare conditions but also encourage patients to engage in advocacy or support networks as part of a holistic approach to coping. Providing resources to connect patients with advocacy groups or online support communities could enhance their ability to cope with the emotional and social aspects of living with a rare disorder.

#### 4. The Role of Support Networks

Support networks, including family, friends, and peer communities, played a vital role in helping participants manage both the physical and emotional challenges of their condition. The findings of this study align with research by Zurynski et al. (2017), which highlights the importance of strong social support in maintaining psychological well-being among individuals with rare genetic disorders. Participants who had the support of their families and close friends reported feeling more emotionally resilient and better equipped to cope with their daily challenges.

Peer support from online forums and advocacy groups was also crucial in providing participants with a sense of belonging and understanding. As Currie and Szabo (2020) noted, individuals with rare genetic disorders often feel isolated due to the rarity of their condition, making peer support essential for emotional connection and validation. These findings underscore the importance of healthcare providers and support services facilitating access to peer networks, either through in-person groups or online platforms.

#### 5. Implications for Practice

The findings of this study have several important implications for healthcare providers, policymakers, and society. First, there is a clear need for improved access to specialized healthcare for individuals with rare genetic disorders. Healthcare professionals should be trained to recognize the specific needs of these patients, ensuring timely diagnosis, adequate treatment, and proper referral to specialists.

Additionally, efforts to reduce social stigma must be prioritized. Public education campaigns that raise awareness about rare genetic disorders can help mitigate the misconceptions and discrimination that patients experience. Workplaces should also implement policies that support individuals with chronic conditions, offering accommodations that enable them to remain productive and valued employees.

Finally, providing emotional and psychological support should be integral to the care of individuals with rare genetic disorders. Healthcare providers can encourage patients to engage in peer support groups and advocacy efforts, which have been shown to enhance coping and emotional well-being.

#### 6. Limitations of the Study

While this study provides valuable insights into the experiences of individuals living with rare genetic disorders, there are several limitations to consider. First, the sample size was small, which is typical for qualitative research but limits the generalizability of the findings. Additionally, the study was conducted at a

single tertiary hospital, which may not capture the full range of experiences across different geographic or socioeconomic contexts.

Second, self-reported data from interviews may be subject to recall bias, as participants may have difficulty accurately recalling all aspects of their experiences. Despite these limitations, the study provides an in-depth understanding of the lived experiences of individuals with rare genetic disorders and offers valuable recommendations for improving their care and support.

## 7. Future Research

Future research should expand on the findings of this study by including larger and more diverse populations of individuals with rare genetic disorders. Longitudinal studies could provide further insights into how patients' experiences and coping strategies evolve over time. Additionally, further research on the effectiveness of public awareness campaigns and workplace accommodations for individuals with rare conditions is needed to guide policy development.

## Conclusion

In conclusion, this study highlights the daily challenges, social stigmas, and coping strategies experienced by individuals living with rare genetic disorders. Despite the significant physical and emotional burdens, participants demonstrated resilience through acceptance, advocacy, and support from their social networks. However, the persistence of social stigma and discrimination underscores the need for continued societal education and policy reform. By addressing these challenges holistically, healthcare providers and policymakers can improve the quality of life for individuals with rare genetic disorders.

## References:

1. Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative research in psychology*, 3(2), 77-101.
2. Currie, G., & Szabo, J. (2020). Social isolation and exclusion: the parents' experience of caring for children with rare neurodevelopmental disorders. *International journal of qualitative studies on health and well-being*, 15(1), 1725362.
3. Fournier, H., Calcagni, N., Morice-Picard, F., & Quintard, B. (2023). Psychosocial implications of rare genetic skin diseases affecting appearance on daily life experiences, emotional state, self-perception and quality of life in adults: a systematic review. *Orphanet Journal of Rare Diseases*, 18(1), 39.
4. Goffman, E. (2009). *Stigma: Notes on the management of spoiled identity*. Simon and Schuster.
5. Lanar, S., Acquadro, C., Seaton, J., Savre, I., & Arnould, B. (2020). To what degree are orphan drugs patient-centered? A review of the current state of clinical research in rare diseases. *Orphanet Journal of Rare Diseases*, 15(1), 134.
6. Lazarus, R. S. (1984). *Stress, appraisal, and coping* (Vol. 464). Springer.
7. Link, B. G. (1986). Phelan, J. C. (2001). Conceptualizing stigma. *Annual review of Sociology*, 27(1), 363-385.
8. Pelentsov, L. J., Fielder, A. L., Laws, T. A., & Esterman, A. J. (2016). The supportive care needs of parents with a child with a rare disease: results of an online survey. *BMC family practice*, 17, 1-13.
9. Somanadhan, S., O'Donnell, R., Bracken, S., McNulty, S., Sweeney, A., O'Toole, D., ... & Kroll, T. (2023). Children and young people's experiences of living with rare diseases: An integrative review. *Journal of pediatric nursing*, 68, e16-e26.
10. Stoller, J. K. (2018). The challenge of rare diseases. *Chest*, 153(6), 1309-1314.

11. Zurynski, Y., Gonzalez, A., Deverell, M., Phu, A., Leonard, H., Christodoulou, J., & Elliott, E. (2017). Rare disease: a national survey of paediatricians' experiences and needs. *BMJ Paediatrics open*, 1(1).
12. Zhu, X., Smith, R. A., & Parrott, R. L. (2017). Living with a rare health condition: the influence of a support community and public stigma on communication, stress, and available support. *Journal of Applied Communication Research*, 45(2), 179-198.