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PARENTAL CHALLENGES AND PERCEPTIONS WITH A GENETIC DISORDER CHILD: A REVIEW PAPER

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Abstract

Objective: *This review aims to analyze existing literature on the challenges faced by parents of children with genetic disorders, focusing on their perceptions, coping mechanisms, and access to healthcare. It seeks to identify gaps in current research and provide insights for improving support systems and policy interventions.* ***Method and Material:*** *A structured search was conducted in Google Scholar, CINAHL, and PubMed, adapting search terms to each database's syntax and indexing. Boolean operators (AND/OR) combined keywords, with MeSH terms for PubMed and database-specific headings for CINAHL. Google Scholar required a flexible approach. The initial search yielded 54,348 articles, refined by filters to 6,320. After reviewing titles, abstracts, and backgrounds, 15 studies were selected. Most were qualitative or mixed-method, aligning with the research topic, excluding editorials and letters*

Results: *Parents face multifaceted challenges, including significant emotional distress, financial strain, and societal stigma. Many parents experience heightened anxiety and depression, compounded by a lack of structured support systems. However, coping strategies such as resilience, religious beliefs, and social support networks help mitigate these challenges.*

Conclusion: *Parents face complex challenges regarding their child care that demand comprehensive healthcare, psychosocial support, and policy reforms. Strengthening healthcare infrastructure, increasing community awareness, and establishing structured support systems can enhance parental well-being. Future research should focus on improving access to specialized care, mental health interventions, and policy-driven solutions to support families affected by genetic disorders.*

INTRODUCTION

Genetic disorders pose substantial challenges for affected individuals and their families, particularly in low- and middle-income countries (LMICs) where healthcare resources and specialized support services are often limited (World Health Organization [WHO], 2020). Parents who had child with a genetic disorder faces profound emotional, and financial, as they navigate the complexities of medical care, developmental needs, and societal perceptions (Dixon et al., 2019). These challenges are further exacerbated in communities with limited awareness and persistent cultural stigmas (Ahmed et al., 2021).

In Pakistan, where healthcare infrastructure is still developing, parents of children with genetic disorders frequently struggle with late diagnoses, and financial constraints, making it difficult to provide optimal support for their children (Khan et al., 2022). Additionally, the lack of structured support systems often leaves families feeling isolated and overwhelmed, increasing their vulnerability to mental health issues such as stress, anxiety, and depression (Jabeen & Malik, 2020). While existing research has primarily focused on the clinical and genetic aspects of these conditions, there is a gap in literature regarding the parental challenges and their coping mechanisms (Raza et al., 2023). Understanding these parental challenges and perceptions is essential for informing healthcare policies, developing community-based interventions, and enhancing the quality of life for both children with genetic disorders and their families.

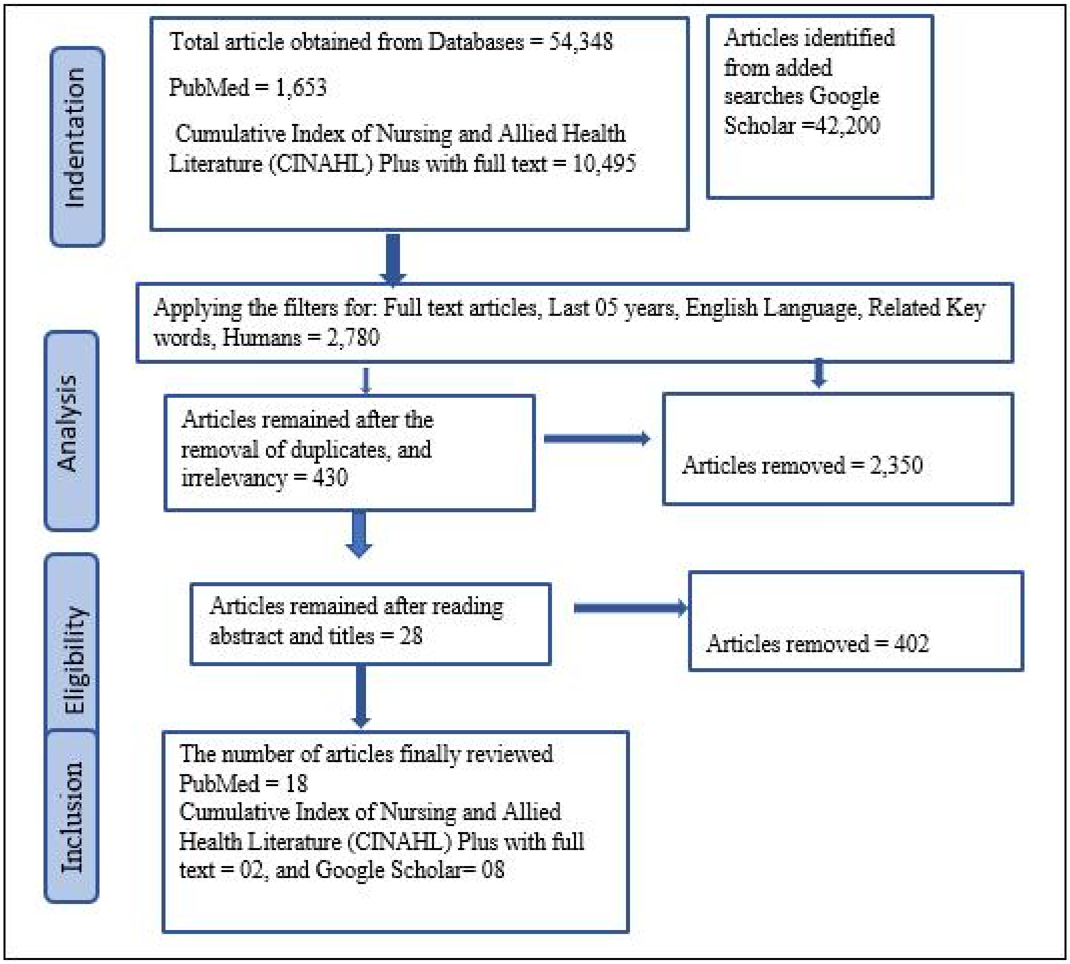
This review paper aims to understand the perceptions and challenges of parents raising a child with a genetic disorder, shedding light on their emotional, financial, and social struggles. By examining existing research, this study seeks to highlight the coping mechanisms employed by parents and propose strategies for enhancing caregiver support systems. Ultimately, this review aims to contribute valuable insights for healthcare professionals, policymakers, and social support organizations in addressing the complex needs of these families.

Method and Materials:

A systematic search strategy was meticulously designed to identify relevant literature on parental challenges and perceptions related to genetic disorders. The search was conducted across three major databases: **Google Scholar, CINAHL, and PubMed**, each requiring tailored search strategies due to variations in syntax, indexing systems, and MeSH terms.

To ensure comprehensive coverage, **Boolean operators** (AND/OR) were used to refine search results. **MeSH terms** were applied in **PubMed** to align with its indexing system, while **CINAHL-specific subject headings** were used to capture nursing and allied health literature. Given the broad indexing scope of **Google Scholar**, a more flexible approach was employed, modifying search terms accordingly. The search strategy included terms such as **"parental lived experiences"** OR **"genetic disorders"** OR **"caregiving challenges"** AND **"under five years of children"** AND **"Pakistan"**, strategically combined using Boolean operators. The initial search across these databases yielded **54,348 articles**. To enhance relevance, multiple filters were applied, including **full-text availability, studies from the last five years, relevant keywords, and human studies**, which narrowed the selection to **6,320 articles**. These shortlisted studies were further screened based on **title, abstract, and background content** to remove duplicates and irrelevant articles. After this rigorous selection process, **28 studies** were finalized for in-depth review. Notably, most of the selected studies employed **qualitative or mixed-method approaches**, aligning well with the research focus on parental perceptions and experiences. **Editorials and letters to editors** were excluded, whereas **systematic reviews and meta-analyses** were considered where relevant. This methodological approach ensures that the findings presented in this review are drawn from credible, high-quality sources, providing valuable insights into the multifaceted challenges faced by parents of children with genetic disorders.

Figure 1. PRISMA Flow Chart



Literature Review on the parental challenges and perceptions with a genetic disorder child

Over the past few decades, research on genetic disorders (GDs) has expanded significantly, shedding light on the experiences of parents navigating the challenges of raising a child with such conditions. Genetic disorders, including Down syndrome, cystic fibrosis, and thalassemia, impose considerable emotional, financial, and psychological burdens on families worldwide (Turner & Biesecker, 2019). The prevalence of genetic disorders varies across different regions, with low- and middle-income countries facing significant healthcare challenges due to limited resources and awareness (World Health Organization, 2021). The journey of parents raising a child with a genetic disorder is marked by various stages, including diagnosis, adaptation, coping, and long-term caregiving, each carrying its own set of challenges and emotional tolls (Kearney & Griffin, 2020).

Emotional and Psychological Impact on Parents

A descriptive phenomenological study revealed that parents often experience intense emotional distress upon receiving their child's diagnosis of a genetic disorder. Feelings of shock, denial, guilt, and sadness are commonly reported, particularly in cultures where genetic conditions are stigmatized (Pang et al., 2018). A qualitative study conducted in Pakistan highlighted that mother face significant mental health challenges due to societal pressures and expectations surrounding child-rearing (Ali et al., 2020). Additionally, fathers often experience emotional distress but may suppress their feelings due to traditional gender roles that emphasize strength and resilience (Gholami et al., 2019).

An Iranian study demonstrated that parents of children with genetic disorders have a higher prevalence of anxiety and depression compared to parents of neurotypical children (Behjati et al., 2017). Another cross-sectional study conducted in Turkey found that stress levels in parents were directly correlated with the severity of the child's condition, financial constraints, and lack of support systems (Kaya & Isik, 2021). The burden of caregiving often leads to emotional exhaustion, further impacting the mental health of parents (Smit et al., 2020).

Financial and Social Challenges

The economic burden of raising a child with a genetic disorder is substantial. A retrospective cross-sectional study conducted in India found that families with a child diagnosed with a genetic disorder often struggle with increased medical expenses, loss of employment, and reduced financial stability (Gupta et al., 2018). Parents frequently report difficulties in accessing affordable healthcare, special education services, and therapeutic interventions, especially in low-income countries (Mahmoudi & Mehmood, 2021).

Social stigma and discrimination further exacerbate parental stress. In many cultures, genetic disorders are misunderstood, leading to social exclusion and strained family relationships (Baum & Fisher, 2020). A cross-sectional study in Nigeria found that parents often encounter societal rejection, which limits their access to community support and resources (Ademola et al., 2019). The lack of awareness and education about genetic conditions contributes to misconceptions and prejudices, intensifying the social burden on affected families (Chandran et al., 2021).

Coping Strategies and Resilience

Despite the numerous challenges, many parents develop resilience and adopt coping mechanisms to navigate the complexities of raising a child with a genetic disorder. A longitudinal study conducted in Sweden found that social support, including family, friends, and healthcare professionals, plays a crucial role in reducing parental stress and improving coping abilities (Lundqvist & Ahlström, 2019). Similarly, a meta-analysis of qualitative studies on parental resilience indicated that religious beliefs, personal acceptance, and participation in support groups contribute significantly to emotional well-being (Hall et al., 2022).

A comparative study between families in the United States and Brazil found that parental education and awareness about genetic disorders positively influence their ability to advocate for their child's needs (Rodrigues et al., 2020). Additionally, cognitive-behavioral therapy (CBT) and mindfulness-based interventions have shown promising results in alleviating stress and promoting adaptive coping strategies among parents (Perez et al., 2021).

Healthcare System and Parental Experiences

Parents frequently report challenges in navigating the healthcare system while seeking appropriate medical care for their child. A systematic review of parental experiences highlighted common issues such as lack of access to specialized care, inadequate communication from healthcare providers, and fragmented healthcare services (Nelson et al., 2022). A qualitative study in Egypt emphasized the importance of healthcare professionals adopting a family-centered approach, which includes clear communication, emotional support, and collaborative decision-making with parents (El-Sayed et al., 2020).

A prospective study in the United Kingdom found that early interventions, including genetic counseling and multidisciplinary care models, significantly improve parental satisfaction and overall well-being (Robinson et al., 2019). Parents who received proper guidance from genetic counsellors were more likely to engage in proactive healthcare planning and advocacy for their child (Barlow et al., 2021).

Conclusion

Parents face challenges such as presents profound emotional, and financial, for parents, particularly in resource-limited settings. This review highlights the significant burden faced by caregivers, including psychological distress, financial strain, societal stigma, and inadequate healthcare support. Despite these hardships, many parents demonstrate resilience by relying on coping strategies such as social support, religious beliefs, and advocacy efforts. However, the lack of structured support systems and accessible healthcare further exacerbates their struggles.

To improve the well-being of parents and children affected by genetic disorders, there is a pressing need for targeted interventions. Future research should focus on exploring comprehensive support mechanisms, developing culturally appropriate counseling programs, and enhancing healthcare accessibility. Additionally, policymakers and healthcare providers must collaborate to establish inclusive environments that offer both medical and psychosocial support, ensuring a better quality of life for affected families.

Recommendations

Based on the findings of this review, the following recommendations are proposed to better support parents raising a child with a genetic disorder:

Enhanced Parental Education and Psychosocial Support

- Healthcare professionals should implement structured educational programs to improve parental understanding of genetic disorders, treatment options, and caregiving strategies.
- Psychosocial support services, including mental health counseling and peer support groups, should be made widely available to help parents cope with emotional stress.

Strengthening Community and Family Support Systems

- Community awareness campaigns should be launched to reduce societal stigma and promote acceptance of children with genetic disorders.
- Family-centered interventions should be encouraged to foster emotional and financial support within extended families.

Culturally Sensitive Mental Health Interventions

- Counseling programs tailored to cultural beliefs and social dynamics should be developed to address the psychological distress faced by parents.
- Religious and spiritual coping mechanisms should be incorporated into support programs where culturally appropriate.

Policy Reforms for Improved Healthcare Access

- Governments should prioritize policies that ensure early diagnosis, affordable treatments, and access to specialized healthcare services.
- Financial assistance programs should be introduced to ease the economic burden on families managing chronic genetic conditions.

Early Intervention and Long-Term Support Strategies

- Screening programs for early detection of genetic disorders should be implemented to improve treatment outcomes.
- Long-term care plans should be developed, integrating medical, educational, and social support services to improve the child's overall well-being.

By implementing these recommendations, healthcare providers, policymakers, and community organizations can work together to create a more inclusive and supportive environment for families affected by genetic disorders. Strengthening these support systems will not only enhance parental coping mechanisms but also contribute to improved long-term outcomes for children living with genetic conditions.

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