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The Best Practices in Managing Rare Diseases: Insights from Nursing Perspectives on the Psychosocial and Healthcare Challenges Faced by Affected Children and their Families

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Abstract

Background: Rare diseases (RD) present significant challenges for children, their families, and healthcare providers. With varying definitions across regions, these conditions often lead to a myriad of psychosocial issues, including social isolation, stress, and inadequate healthcare access. Understanding the complexities surrounding RD is crucial for improving care strategies and outcomes.

Methods: This study conducted a comprehensive literature review utilizing databases such as Google Scholar and PubMed to analyze peer-reviewed articles related to the psychosocial aspects of managing rare diseases, particularly focusing on the experiences of children with reading disorders (RD) and their families. The search included articles published in English up to 2023.

Results: The findings indicate that children with RD face significant challenges in their family, educational, and social environments, often resulting in mental health issues and diminished quality of life. The review highlighted the necessity for coordinated, multidisciplinary care approaches to address the unique needs of these children and their families. Moreover, it identified gaps in research concerning the perspectives of children themselves and the impact of social determinants of health on their care experiences.

Conclusion: Enhancing the understanding of the psychosocial factors affecting children with rare diseases is essential for developing evidence-based interventions that improve quality of life. There is an urgent need for ongoing research and advocacy to ensure equitable access to resources and support

systems for these families, ultimately fostering better health outcomes and experiences for children with RD.

Keywords: Rare diseases, psychosocial factors, children, healthcare access, multidisciplinary care.

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1. Introduction

Rare diseases (RD) seldom afflict young people, teens, and their families, although they have a substantial influence. The US National Institutes of Health classifies such illnesses as occurring with a prevalence of 1 in 200,000 or less, however definitions differ elsewhere [1]. The US concept is associated with the 1983 Orphan Drug Act, a legislation designed to incentivize pharmaceutical companies to investigate and develop therapies for rare illnesses. In the European Union (EU), rare diseases (RDs) are classified as conditions affecting fewer than 1 in 2000 individuals. Similar to the orphan drug legislation in the United States, there is a focus on orphan illnesses and medications in the European Union and Japan [2-4]. Nonetheless, the lack of governmental focus on rare diseases and their effects remains a global concern [5]. This is indeed comprehensible, given 40 out of every 100,000 persons are affected with RD [1]. In this context, it is probable that globally, hundreds of millions of individuals are impacted by rare diseases, with estimations of over 25 million in North America as well as up to 36 million across Europe [1].

Genetic diseases constitute a significant proportion of RD [6]. Identifying a genetic foundation for RD may facilitate improved definitions and methodologies in treatment research and development; yet it complicates the detection and focus on particular disease characteristics owing to the uniqueness of gene-mediated processes. Features may present as a phenotypic manifestation identifiable by the physician, including seizures, but may have poor responsiveness to standard treatment drugs, necessitating symptom-driven genomic inquiry or specialized diagnostics. In instances when RD manifests as seizures, specialized examination of the CSF fluid may be required. Additional problems may include metabolic mechanisms affecting crucial organ function or notable developmental behavioral variations [7]. The diagnostic journey associated with raising an infant with RD is extensive and encompasses efficient, profound, interpersonal, and contextual challenges that are not fully understood by caregivers or those affected by the child's RD.

Children with RD frequently have persistent, intricate medical issues necessitating a multifaceted array of treatment from several professional providers. Individuals may experience isolation, anxiety, or sadness, and may encounter stigma in educational, employment, and workplace environments, resulting in a deficiency of self-sufficiency and a lack of social assistance or empathy from others [8]. Families of kids with reading difficulties have other hurdles as well. Parents of kids with neurological disorders have reported experiencing social isolation and feelings of being overwhelmed [9]. Other parents indicate a lack of understanding from friends with usually developing children and experience a significant treatment of their homes and family life [10-13].

This study analyzes RDs and their emotional implications for families, kids, and medical professionals. The realms of family, education, society and healthcare are examined, along with the ramifications of RD administration as kids progress into adulthood, necessitating that families get and battle for their healthcare requirements. Consequently, pertinent healthcare and governance issues, along with advanced translational research that examines the interplay of identities amongst RD, are suggested. Ultimately, suggestions for evidence-based treatments along with supplementary care in the specified areas are provided. Calls for intervention for families, physicians, researchers, and advocacy representatives are articulated to encourage ongoing efforts in providing evidence-based treatment for these children.

2. Methods

A literature search was performed in the Google Scholar and PubMed libraries for peer-reviewed works published in English until 2023.

3. The Child with Reading Disorder

People with RD are thriving and leading lifestyles formerly deemed unattainable, but remain little understood [14,15]. Current understanding of children with reading difficulties (RD) outlines the psychosocial circumstances of their relatives and caregivers; nonetheless, the kid with RD has to stay the central focus. Children with reading difficulties are more prone to face considerable problems in their family, school, and community environments [16-18]. Nevertheless, there is little study delineating the child's own experiences. Knowledge on RD is derived from the convergence of cohorts of kids with persistent illnesses. Children and youth with particular medical requirements possess, or are at heightened risk of possessing, a chronic physical, growing, or psychological condition that necessitates more healthcare services than those required by most typically developing children [19]. Children having healthcare complication (CMC) are a subset of children with special health care needs (CSHCN) and those with rare diseases (RD), often exhibiting functional impairments and reliance on medical equipment and technology, such as gastrostomy tubes.

The medical and emotional requirements of children with medical complexities (CMC) along with their caregivers are inadequately addressed by several current healthcare models, with CMC being almost twice as likely as normally growing kids to have inadequate medical care [20]. They constitute a minor segment of the pediatric population but use almost one third of the yearly healthcare expenditures allocated to children [21]. Children with reading disabilities need coordinated multidisciplinary treatment across several sectors, encounter a greater incidence of inpatient admissions compared to their peers, and have considerable challenges in getting their perspectives acknowledged [22,23]. Research indicates inconsistent attempts by care teams to engage children with a rare disease in their own care, mostly depending on reports from parent caregivers [24,25]. Although not all Children with Special Health Care Needs (CSHCN) and Children with Medical Complexity (CMC) experience Rare Diseases (RD), a significant number do. The severity and complexity of their clinical and psychological requirements render this demographic crucial for evaluating the psychosocial requirements of kids with RD.

The overlapping dimensions of the child's identity as well as observations are overlooked in research about RD. Research on rare diseases (RD) may be adversely affected by the limited population of persons with such diagnoses. However, neglecting intersectionality and failing to comprehend our care models, including their target populations and effective conditions, may restrict the value and applicability of the insights obtained [26]. Social aspects of health (SDH) are recognized as factors contributing to disparities in outcomes, however their influence on children with reading disorders (RD) remains little defined. Diagnostic genetic testing is a potent resource, however it may not be accessible to everyone who need it [27]. In the United States, Children with Special Health Care Needs (CSHCN) are mostly of Black, non-Hispanic/Latinx descent and are thought to face disparities in wellness and medical access owing to past marginalization [28].

Upon analyzing discrepancies related to race and ethnicity, main home language, insurance type, and socioeconomic status, it is shown that kids with medical complication are twice as likely to experience at least one unmet need compared to their counterparts without medical complexity. In one research, kids with medical complication had significantly more unmet needs compared to their counterparts without medical complication across all racial and ethnic groups [20]. Racial, regional, and socioeconomic disparities significantly affect healthcare worldwide, and these social determinants of health are projected to have a considerable influence on children with reading disorders.

Health-related quality of life (HRQOL) in kids with chronic illnesses is associated with self-management and self-efficacy [29]. Children with RD have obstacles that affect their quality of life (QOL) as well as psychosocial functioning, as seen by elevated mental health demands [30,31]. Recent cross-sectional research in Western Australia revealed that 43.9 percent of caregivers of children with reading issues stated their kid had mental health challenges [32]. Lum and associates discovered that parents of kids with chronic diseases were two times more inclined to indicate that their kid suffered mental discomfort and diminished self-confidence [33].

Children with reading disorders who experience emotional distress or other developmental behavioral issues are at risk of encountering communication difficulties. These children may have communication obstacles, some of which adversely affect their parental care, or they may possess a concomitant speech or language disability. The psychosocial consequences of possessing a speech or linguistic problem have been recorded and involve harassment, impairments in adaptability, and challenges in emotion control [34,35]. Research indicates that kids with language difficulties have a higher propensity for anxiety, sadness, ADHD, and externalizing emotions than their counterparts without such issues [36]. Lewis and colleagues discovered that teenagers with a young-onset speech sound problem had inferior psychosocial outcomes when accompanied by language impairment [37]. Consequently, youngsters with reading difficulties and communication impairments may want distinct care to foster favorable psychosocial outcomes.

4. Research and Development in the Family

Rare diseases affect the whole family. Hoover and colleagues have recognized that the current COVID-19 epidemic has highlighted the challenging experiences often faced by relatives of CSHCN [38]. Instances of such familial impact encompass compulsory homeschooling, confinement at home, the necessity to balance the social and academic demands of the child with their health needs, disparities in healthcare quality, and the inequities in outcomes associated with ethnicity, race, or economic status. The authors assert that the previously unacknowledged importance of family caregiving—encompassing nurture, duties, resources, and services to fulfill daily needs—is still insufficiently recognized [38]. Families must consistently address both ordinary day-to-night care and emergent emergencies, regardless of the hurdles they encounter or the availability of past knowledge and support resources. It is particularly crucial to recognize families' strength in confronting hardship [39].

5. Determinants of Health in Society

Social determinants of health (SDHs) significantly affect children with reading disabilities (RD) and their families [40]. Families with children with severe chronic disorders, especially rare diseases, are more prone to encounter medical financial difficulties. Medical financial strain is associated with worse child health outcomes, irrespective of a family's socioeconomic level or additional financial assets [41]. A significant correlation exists between relinquished family labor and family-supplied medical care [42-45]. Children with special health care needs have an increased risk of inadequate nutrition and malnutrition, significantly affecting their daily life and long-term performance [46,47]. Limited access to home resources correlates with heightened acute healthcare use and unfulfilled healthcare requirements among children with special healthcare needs [48,49]. This pertains not just to housing stability but also to the availability of accessible housing modifications for children with impairments. Families have challenging trade-offs in selecting feasible housing alternatives. High-acuity medical incidents, such extended critical care admissions or the emergence of new technological dependencies, may result in increased susceptibility [50,51]. The percentage of families experiencing unmet basic requirements rises after chemotherapy for newly diagnosed children’s malignancies; a similar trend may occur with acute health status changes in other illnesses such as RD [52].

Certain children with reading disorders have intricate medical requirements necessitating ongoing home health treatment. Private duty nurse (PDN), commonly referred to as "home nursing care," is a crucial component of treatment for certain children [53]. PDN activities involve tracheostomy/ventilator support, airway and lung care, tube feeding, medication administration, execution of prescribed home rehabilitation activities, and other vital health services. In the absence of PDN, some youngsters may not reside securely at home. Limited accessibility to home medical care and personnel in Europe, North America, and worldwide has been recognized as a catastrophe for families. PDN is fundamentally associated with the survival of kids and family stability, since family life is interconnected with home healthcare scheduling, staffing, and services [54]. Indeed, deficiencies in PDN personnel jeopardize the physical, emotional, and financial well of families. Families must persistently contest payors and government entities for their designated services, with out-of-pocket expenses being prevalent despite

their inequity. The persistent and extensive deficiency of home nurses, coupled with the regional variability in the quality and amount of nursing services, results in many children not receiving the hours to which they are entitled [43,55]. This leads to family caregivers resorting to improvised nursing care, potentially jeopardizing the child's health, causing parents to forgo job and money, and adversely affecting marital and familial relations [56].

Families have several psychological difficulties in delivering affectionate care to their kid with RD. The first diagnostic journey to uncover the genetic foundations of a rare illness might provide several significant challenges for families [57,58]. Mendelian genetic diseases are mostly attributable to modifications in a single gene or genomic anomalies and may manifest at birth or be evident in familial history. Mendelian genetic diseases, although individually uncommon, are together prevalent and significantly contribute to juvenile morbidity and death [27]. Genetic testing facilitates personalized treatment regimens and concludes the diagnostic odyssey, so preventing future unneeded testing and perhaps providing significant psychological benefits, ultimately enhancing quality of life [59]. Genetic testing may indicate that additional family members possess the same gene or condition, presenting challenges for families to manage. Moreover, guaranteeing the equal use of these advancements in genetic technology has proven to be difficult. Technology has limitations as well. Despite the availability of wider genome sequencing, it may not provide a comprehensible response, and after many years, the insights gained may only contribute little to the understanding or treatment of rare diseases [60].

Although genetic testing may facilitate prompt medical action, informed decision-making, access to clinical trials, and participation in disease-specific care for some individuals with rare diseases, substantial obstacles to well-being also arise. This encompasses isolation and solitude. Occasionally, being among the few individuals to get a certain diagnosis, frequently with little information on its progression, prognosis, or available treatments, results in a sense of isolation. In most situations outside of rare diseases (RD), a diagnosis provides clarity, treatment options, and realistic prognosis expectations; nonetheless, a prognosis of an RD is fraught with ambiguity [61]. This may heighten concern for the future, induce instability, and result in various repercussions for family members. Establishing a new normal after this trip may be challenging for many. Parents of children with complex medical conditions (CMC) report a diminished health-related quality of life (HRQOL) relative to parents of non-CMC children, with mental health quality of life evaluations being inferior to those of physical health [61]. Nonetheless, families may have inadequate access to appropriate mental health treatments or feel a "lack of fit" within group settings [62]. Family caregivers mostly have behavioral health difficulties, notably caregiver stress [63,64]. Family caregivers often believe they lack the time to attend to their own mental health requirements [60,61]. When they attempt to address their own requirements, they may encounter caregiving-related obstacles that hinder access to suitable resources [65-67].

Cardinali and colleagues observed that the problems encountered in caring for a kid with RD often differed between moms and dads [68]. Both appreciated details regarding the diagnosis, recognized the absence of a structured medical system, and exhibited several emotions and behaviors as a pair. Fathers identified difficulties related to economics, education, emotions, and conduct. Mothers identified challenges related to their careers, adapting to their child's demands, their educational roles, their own emotions, and the family's systemic functioning. Identifying others who really resonate with the distinctive characteristics of the person with RD is beneficial for the family; yet they may discover other people with RD exhibit shared themes of similar experiences [69]. Effective family life has been shown to enhance the quality of life for children with reading difficulties. Family cohesion, favorable intrafamily interactions, and acceptance correlate with healthy family and child performance; indeed, several families have derived beneficial significance from their interactions [70,71].

Parents report feeling misapprehended by relatives and acquaintances about the reality of their everyday caregiving expertise, and many express challenges in establishing connections with supportive individuals [9]. Social assistance and temporary assistance are recognized for their ability to enhance caregiver health and alleviate stress and load [72]. Nonetheless, the mechanics of obtaining these beneficial resources are fraught with obstacles [73]. Providing educational and interactive peer assistance for parent providers of

children with reading difficulties is a significant service to families [13,74]. These may manifest as in-person events, group offers, virtual live meeting spaces, or asynchronous communication platforms including social media or chat rooms [57]. Although virtual social support alternatives are readily available, parent caregivers may encounter considerable behavioral health issues that may be intensified by the stress of caregiving and the associated responsibilities of caring for RD [75].

Accessing the coordination of essential services is a considerable barrier. Registered Dietitians often need the involvement of several experts, resulting in numerous visits that require coordination and attendance by families, many of whom may lack expertise in intricate care environments. Although a patient- and family-centered care (PFCC) strategy for pediatric chronic disorders is often highlighted, care programming still has much progress to make in meeting the requirements of families with a child diagnosed with rare diseases (RD) [11,76]. Children with reading difficulties (RD) often present in clinics without defined treatment routes, resulting in families experiencing a continuous challenge in advocating for their child's needs [77]. Moreover, healthcare teams may lack familiarity with the RD.

Alongside difficulties in obtaining coordinated treatment, acquiring precise and beneficial information relevant to the RD might also be problematic. Insufficient or restricted access to precise evidence-based information on their child's health might exacerbate caregiver stress. Managing uncertainty and confronting circumstances devoid of answers or data is a significant difficulty for families of a kid with reading difficulties. Families often use the internet to seek answers to inquiries, accessing information that may be correct or inaccurate with expedience. The risks of disinformation are significant with this method. Even when information on the illness is accessible, it may not be provided in the caregiver's native language, exacerbating disparities in access and increasing stress [78]. Families indicate that they inquire with healthcare practitioners for information, only to be informed that it is unknown or not yet available. Care staff strive to get the most current information on patient care, but families may be required to endure uncertainty.

Providing care for kids with RD requires sufficient personal health literacy, including the ability to locate, comprehend, and use knowledge to guide health-related choices. These competencies include reading, listening, speaking, numeracy, and the capacity to seek and retrieve knowledge from credible sources [79]. An individual's health literacy might fluctuate and may diminish during periods of stress. A kid with RD may have many caregivers tasked with adhering to complex instructions on medical care, including medicines, nourishment, and equipment.

Notwithstanding the difficulties reported by families, the experience they acquire over time might be substantial. They get the status of 'professional medical caretakers' via expertise, occasionally about RD, but mostly concerning their own kid and their treatment [38]. Parents serve as intermediaries among the caregivers and their kid and may also have a teaching role in elucidating their child's RD to the care staff.

6. The Medical Team

Caring for a child with a rare disease (RD) necessitates a collaborative effort and community support. Due to the frequent use of healthcare services and the dispersion of healthcare facilities, people with RD along with their relatives often find themselves repeatedly informing new practitioners about their child's intricate medical history and care requirements, assuming the role of an expert rather than the professionals themselves. The turnover of healthcare workers has been recognized as a worry for individuals with rare diseases (RD). Families have recognized the need of consistently updating current or new "continuity" providers as a source of stress and dissatisfaction. Healthcare organizations and teams may enhance care coordination via many methods. Practices should strive to reduce wait times for both chronic and acute care while enhancing access to services. Children with reading disorders (RD) and their parents may depend on a coordinated multidisciplinary care team, since children with chronic diseases often consume social work services more than their healthy peers [73].

Healthcare teams include several specialists as well as trainees. Trainees must be informed about the distinct requirements of persons with reading disabilities. Clinicians must exemplify the ideals of family

and patient-centered treatment to their trainees. Teams must possess common objectives, defined responsibilities, reciprocal trust, good communication, and quantifiable results [80]. Healthcare teams have to exemplify and impart these ideas to medical trainees and within interprofessional education. Healthcare teams may get insights from one other in addition to from patients and their families. Project ECHO exemplifies an excellent methodology across several disciplines, using a "spoke and hub" framework for reciprocal learning and case-based instruction via tele-mentoring [81]. This model transfers knowledge rather than individuals. The ECHO concept may provide healthcare providers, educators, or people with RD a means to swiftly disseminate efficient procedures and knowledge [81].

Medical Homes exemplify a system that addresses patient requirements, enhances the patient expertise, and increases provider effectiveness and support. A Medical Home is an advantageous aspect of treatment for all children, particularly those with developmental disorders, impairments, or other medical complexities [9]. In the United States, there have been shown decreases in healthcare costs for families and insurance providers, along with a decrease in the usage of emergency services. Children with reading disorders sometimes have several healthcare providers, making it challenging to identify the principal individual or team responsible for overseeing comprehensive care coordination [13]. The Medical Home practitioner may be a specialist, such as a geneticist or neurologist with a care team knowledgeable in the rare disease (RD), or a primary care practitioner serving as the focal hub for general medical and collaboration for the kid with RD.

Medical treatment is but one facet of the existence of a kid with RD; collaboration with local organizations and school resources is equally vital. An integrated care approach is advised, where feasible, since it may enhance access to behavioral healthcare by including emotional health and psychological support within the medical environment and team. Care teams may engage with a patient, their parents, and the child's school, as well as home- as well as community-based care providers, to build a comprehensive care plan that encompasses the many environments influencing a child's care. Collaborate with educational institutions and relevant organizations involved with family members to extract data points that inform decision-making about treatments, including medication dose and behavioral strategies.

7. Conclusions

This study has aimed to provide a comprehensive analysis of psychosocial factors for the kid with reading difficulties. Consequently, several deficiencies in the scientific research have been found to enhance evidence-based therapy, psychological support, and societal access to resources for children with RD. Consequently, the current knowledge on the psychosocial aspects of children having chronic diseases, including Children with Special Health Care Needs (CSHCN), Children with Medical Complexity (CMC), and the little information available on Rare Diseases (RD) especially, is analyzed. This literature summary utilizes a targeted sampling of scientific studies, offering recommendations to incorporate recent advances, access optimal resources, and present proposals as 'calls to action' to enhance the quality of life and advocate for evidence-based care for youngsters with RD, their relatives, and healthcare providers. Comprehending the psychosocial factors affecting children with reading difficulties would ideally stimulate future efforts to enhance the understanding and support of these exceptional kids as well as their families.

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أفضل الممارسات في إدارة الأمراض النادرة: رؤى من منظور التمريض حول التحديات النفسية والاجتماعية والرعاية الصحية التي يواجهها الأطفال المصابون وأسرهم

الملخص

الخلفية: تشكل الأمراض النادرة (RD) تحديات كبيرة للأطفال وأسرهم ومقدمي الرعاية الصحية. تختلف تعريفات هذه الحالات عبر المناطق، وغالبًا ما تؤدي إلى مجموعة متنوعة من القضايا النفسية والاجتماعية، بما في ذلك العزلة الاجتماعية، الضغط النفسي، وعدم كفاية الوصول إلى الرعاية الصحية. يعد فهم التعقيدات المحيطة بالأمراض النادرة أمرًا ضروريًا لتحسين استراتيجيات الرعاية والنتائج الصحية.

الطرق:أجرت هذه الدراسة مراجعة شاملة للأدبيات باستخدام قواعد بيانات مثل Google Scholar و PubMedلتحليل المقالات المُحكّمة المتعلقة بالجوانب النفسية والاجتماعية لإدارة الأمراض النادرة، مع التركيز بشكل خاص على تجارب الأطفال المصابين باضطرابات القراءة (RD) وأسرهم. شمل البحث المقالات المنشورة باللغة الإنجليزية حتى عام 2023.

النتائج:تشير النتائج إلى أن الأطفال المصابين بالأمراض النادرة يواجهون تحديات كبيرة في بيئاتهم الأسرية والتعليمية والاجتماعية، مما يؤدي غالبًا إلى مشاكل صحية نفسية وتدهور جودة حياتهم. وأكدت المراجعة على ضرورة اتباع نهج رعاية منسق متعدد التخصصات لتلبية الاحتياجات الفريدة لهؤلاء الأطفال وأسرهم. كما حددت فجوات في البحث تتعلق بمنظور الأطفال أنفسهم وتأثير المحددات الاجتماعية للصحة على تجارب الرعاية التي يتلقونها.

الاستنتاج:يعد تعزيز الفهم للعوامل النفسية والاجتماعية التي تؤثر على الأطفال المصابين بالأمراض النادرة أمرًا ضروريًا لتطوير تدخلات قائمة على الأدلة لتحسين جودة الحياة. هناك حاجة ماسة إلى استمرار البحث والدعوة لضمان الوصول العادل إلى الموارد وأنظمة الدعم لهذه الأسر، مما يؤدي في النهاية إلى تعزيز النتائج الصحية وتجارب الرعاية للأطفال المصابين بالأمراض النادرة.

الكلمات المفتاحية:الأمراض النادرة، العوامل النفسية والاجتماعية، الأطفال، الوصول إلى الرعاية الصحية، الرعاية متعددة التخصصات.