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Understanding the Psychosocial Challenges of K-12 Students with Rare Diseases: A Systematic Literature Review

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ABSTRACT

Background: Students with rare diseases in the United States face significant psychosocial challenges within the K-12 education system that extend beyond the physical dimensions of their conditions and deeply affect their emotional, social, and academic lives.

Methods: A systematic literature review was conducted following Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines to identify and synthesize peer-reviewed literature on the psychosocial challenges faced by K-12 students with rare diseases in the United States.

Results: Three overarching themes emerged from the evidence. First, a pervasive lack of awareness among educators, school administrators, and peers regarding rare diseases and their impact on students. Second, significant adverse psychosocial challenges, including stigma, bullying, depression, social isolation, and anxiety, are experienced by students and their families. Third, a critical shortage of accessible resources and structured support systems for students with rare diseases navigating the K-12 education environment.

Conclusions: The findings confirm an urgent need for sustained long-term research, comparative studies, structured investment in stakeholder education, policy prioritization, and enhanced comprehensive collaboration. Addressing these gaps is essential to ensuring equitable educational access and improved quality of life for K-12 students with rare diseases in the United States.

Introduction

Rare diseases collectively affect a significant portion of the population, yet individually, each condition remains largely unfamiliar to the general public, healthcare providers, and educators alike. In the United States, approximately 30 million individuals are affected by rare diseases, and rare disorders disproportionately impact children. There are approximately 7,000 rare diseases identified in the United States, and only 10% have a cure or treatment available due to the heterogeneity of each condition[1].

For students navigating a rare disease, the school environment presents a distinct and often overlooked set of challenges. K-12 students living with rare diseases may face unique psychosocial challenges that can lead to emotional and social difficulties. Psychosocial health can be defined as the intersection and interaction of social, cultural, and environmental influences on the mind and behavior[2]. The psychosocial well-being of a child encompasses emotional, psychological, and social dimensions of health[3], and is connected to mental balance, social functioning, and overall wellness[4]. There is a documented gap in the literature between

medical and psychosocial health care for children with rare diseases and their families, to improve their mental health and quality of life in the long term[5]. It is often challenging to acquire information about uncommon disorders that would make it possible for families and students to manage the disease, because they are unknown or understudied conditions, and many do not have a specific treatment[6].

Several factors may impact the psychosocial well-being of students with rare diseases and their families, including the diagnostic odyssey, feelings of isolation, financial burden, emotional toll, academic challenges, parent and caregiver stress, stigma, discrimination, and communication obstacles. The quality of life and everyday experiences of students with rare diseases may decline significantly. Poor quality of life can be compounded by maladaptive health behaviors that may lead to depression, self-harm and suicidal ideation, low self-esteem, impaired physical functioning, and psychological distress[7]. United States schools may find it challenging to accommodate students with rare diseases due to the increasing number of children with complex medical conditions, which may result in poor socialization, excessive stress, and academic stagnation[8].

Unmet healthcare needs for individuals with chronic medical conditions are linked to a decline in quality of life and an increased probability of negative behaviors and poor health outcomes[9]. Despite the documented scope of this challenge, there is limited understanding of rare conditions, a lack of comprehensive research focus, and a significant gap in the literature. The purpose of this systematic literature review was to examine peer-reviewed literature to identify the most urgent psychosocial challenges facing K-12 students with rare diseases and their families, and to recommend evidence-based directions for action.

Theoretical Framework

This study was grounded in Vygotsky's theory of child development, also known as sociocultural theory. Vygotsky's theory stresses the importance of social interaction and the development of cognition in students[10]. From Vygotsky's perspective, emotion is recognized as a pervasive phenomenon that influences behavior, decisions, and thoughts[11]. Vygotsky believed the human mind to be a product of dynamic adjustment to new and ever-changing conditions in the external environment[10].

Vygotsky's developmental theories emphasize that integrating behavior and consciousness leads to the unification of the mind, with social interaction playing a significant role in this process[12]. The development of the mind is influenced by two key principles: a systemic approach that encompasses both bio-social and bio-psychological aspects of development, and the role of language in the development of human cognition[13]. This framework provided a lens through which the psychosocial challenges of K-12 students with rare diseases could be examined within the context of their social and educational environments.

The findings of this review connect directly to Vygotsky's sociocultural theory in several important ways. First, the pervasive lack of awareness among educators and peers represents a failure of the social environment to support the cognitive and emotional development of students with rare diseases. Vygotsky emphasized that learning and development occur through social interaction, and when the social environment lacks understanding and awareness, students with rare diseases are denied the scaffolding necessary for their full development[10]. Second, the adverse psychosocial challenges identified, including stigma, isolation, and bullying, directly impair the social interactions that Vygotsky identified as essential to healthy cognitive and emotional growth[11]. Third, the diagnostic odyssey disrupts the continuity of social and educational experiences foundational to development within Vygotsky's framework, thereby representing a significant barrier to the zone of proximal development for these students.

Methods

Study Design

A systematic literature review was conducted to examine peer-reviewed literature on the psychosocial challenges facing K-12 students with rare diseases and their families. Systematic literature reviews are a distinct form of research that seeks to identify, appraise, and synthesize all empirical evidence that meets pre-specified eligibility criteria to answer a given research question. The review followed the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines to ensure comprehensive and transparent reporting of the review process and findings[14].

Research Questions

Three research questions guided this systematic literature review:

RQ1: What are common psychosocial challenges faced by students in K-12 with rare diseases in the United States?

RQ2: How do K-12 students with rare diseases cope with the psychosocial challenges of their disease?

RQ3: What support systems are available for K-12 students with rare diseases to address their psychosocial challenges?

While the three research questions center on the experiences of K-12 students with rare diseases, the inclusion criteria for this review also encompassed studies examining the psychosocial experiences of

parents, caregivers, and siblings, given the documented interconnection between family well-being and student outcomes confirmed across the included literature.

Literature Search Strategy

A comprehensive and systematic search strategy was used to locate relevant studies. The following databases were searched: St. Thomas University Library, ProQuest Central, ERIC, Open Access Journals, Orphanet Journal of Rare Diseases, Frontiers, family medicine journals, research journals, international databases, government resources, and Google Scholar. The Orphanet Journal of Rare Diseases was the primary database used, given its emphasis on all facets of rare diseases and orphan medications. The literature was searched from 2019 through 2024 to ensure comprehensive coverage. Keywords included but were not limited to psychosocial challenges, rare diseases, rare disorders, orphan diseases, complex medical conditions, chronic diseases, uncommon medical conditions, rare diseases in children, and rare disorders in education. Specific rare diseases were also searched.

Inclusion and Exclusion Criteria

Studies were included if they examined psychosocial challenges, defined as the intersection and interaction among social, cultural, and environmental influences on the minds and behaviors of K-12 students with rare diseases and their families[2]. Additional inclusion criteria required that studies were published between 2019 and 2024, focused on children under 18 years of age diagnosed with rare diseases, addressed psychosocial challenges in the K-12 educational setting, were published in peer-reviewed journals, and were available in English. This review also included studies examining the experiences of parents, caregivers, and siblings of children with rare diseases, as well as relevant government resources.

Studies were excluded if they were published before 2018, were not specific to rare chronic diseases, focused on individuals older than 18, were published in languages other than English, or were not peer-reviewed.

While the primary focus of this review was the United States K-12 educational context, international studies were included to provide a more robust and comprehensive review of the literature, given the limited body of United States-specific literature on this topic. This decision was intentional and consistent with the heterogeneous nature of rare diseases and the limited number of children affected, necessitating reliance on a broader international body of evidence.

Quality of Life Instrument Selection

The review did not require the use of standardized or validated quality-of-life instruments as an inclusion criterion. Studies were selected for relevance to the psychosocial challenges experienced by K-12 students with rare diseases and their families, regardless of the specific measurement tools used. While several included studies used validated instruments to assess quality of life and psychosocial well-being, the diversity of measurement tools across studies reflects the heterogeneity of rare diseases and the varied populations examined.

PRISMA Flow

Searches using the keywords and phrases for this review yielded 379 peer-reviewed articles and 3 additional records from other sources, for a total of 382 records identified. After removing duplicates, 274 articles remained. Following title and abstract screening, 111 articles were excluded because their titles or abstracts did not meet the inclusion criteria. A total of 134 full-text articles were assessed for eligibility. Nine articles were excluded with reasons. Fourteen studies were eligible and included in the final analysis.

Records identified: 382

Duplicates removed: 108

Records screened: 274

Records excluded after screening: 111

Full-text articles assessed: 134

Full-text articles excluded with reasons: 9

Studies included in final review: 14

Data Analysis

The data analysis phase involved synthesizing and extracting meaningful information from selected articles, analyzing literature for leading authors, journals, locations, and organizations, identifying correlations among topics, and summarizing large amounts of data to identify emerging trends[15]. A literature matrix was created in Microsoft Excel that outlined the objectives, methodologies, and outcomes of each study, focused on studies relevant to the research questions[16]. The systematic literature review process included developing and validating the research questions; establishing criteria for database searches; conducting title and abstract screening, full-text screening, and manual searching; extracting data and assessing its quality; and conducting statistical analysis[17].

Ethical Considerations

This study is a systematic literature review that uses secondary data analysis of peer-reviewed scientific literature. No ethical approval was required. No human participants were involved in this study; therefore, no participant protection protocols were required. This systematic review followed the PRISMA guidelines for data collection and reporting.

Results

Five overarching themes were identified from the systematic review of peer-reviewed literature on the psychosocial challenges facing K-12 students with rare diseases and their families.

Theme 1: Lack of Awareness of Rare Diseases

The need to understand and heighten awareness of rare diseases among the biotech and pharmaceutical industries, healthcare providers, schools, teachers, and peers is critical. More research on rare diseases is needed to ensure the

Evidence Summary Table

Author (Year)	Design	Sample	Disease	Measures	Key Findings	Theme	RQ
Adama et al. (2021)	Cross-Sectional Survey	41 parents of children ages 6-18	Various rare conditions including cystic fibrosis	Stigma, mental health, bullying measures	75.6% experienced stigma. Nearly half reported bullying. Negative school experience.	Themes 1, 2	RQ1, RQ2
Angural et al. (2020)	Review Article	17 individuals in 8 families	Multiple rare diseases	Psychosocial challenges assessment	Psychosocial stress, lack of resources, social stigma, diagnostic delay, financial strain.	Themes 1, 5	RQ1, RQ3
Ashtari & Taylor (2022)	Qualitative Interviews	30 participants with genetic diseases	Ehlers-Danlos Syndrome	Framework interview analysis	Knowledge gap causing delayed diagnoses. Significant pain and isolation.	Themes 1, 5	RQ1
Berkelbach van der Sprenkel et al. (2022)	Qualitative Survey	7,168 students from 78 schools	Chronic and rare heart conditions	Quality of life assessment	Poor life satisfaction, mental health, and school performance.	Theme 2	RQ1, RQ2
Boettcher et al. (2021)	Systematic Review	31 articles included	Various rare conditions	Parental quality of life	Lower parental quality of life, depression, anxiety, limited support, financial burden.	Themes 3, 4	RQ2, RQ3
Brandt et al. (2022)	Systematic Review	24 articles, 12-962 participants	Spinal Muscular Dystrophy	Psychosocial well-being	Reduced quality of life, caregiver stress, financial burden, limited social support.	Theme 3	RQ2, RQ3
Chung et al. (2022)	Case Study	Undiagnosed rare hereditary conditions	Various rare conditions	Psychosocial well-being and quality of life	Psychosocial impairment, lack of mental health resources, increased financial burden.	Themes 1, 3, 5	RQ1, RQ3
Depping et al. (2021)	Cross-Sectional Mixed Methods	300 patients with rare disease	Various rare conditions	Quality of life and psychosocial support	Lack of psychosocial support across well-being, physical, and daily activities.	Themes 1, 4	RQ1, RQ3
Kolemen et al. (2021)	Qualitative Questionnaire	40 randomly selected parents	Various rare conditions	Parents' psychosocial well-being	Increased anxiety and depression while awaiting diagnosis. Delayed diagnosis a key challenge.	Themes 3, 5	RQ1, RQ2
Paz-Lourido et al. (2020)	Qualitative Interviews	28 participants including students, parents, school staff	Various rare conditions	Psychosocial well-being, educator perceptions	Decreased quality of life. Perceived discrimination. Health-related quality of life impacted by schooling.	Themes 1, 2, 4	RQ1, RQ2, RQ3
Rojvik et al. (2023)	Systematic Review	14 articles included	Various rare conditions	Impact on education	Academic achievements compromised. Lack of research and understanding among educators.	Themes 1, 2, 4	RQ1, RQ3
Saggu et al. (2021)	Review Article	Education and health policies reviewed	Various rare conditions	School experience evaluation	Multi-disciplinary, multi-sectoral approach required to address K-12 rare disease needs.	Theme 4	RQ3
Sandilands et al. (2022)	Literature Review	31 articles included	70 rare diseases	Caregiver quality of life	Heavy caregiver burden. Emotional, social, mental burden. Scarce resources. Limited empathy.	Themes 3, 4	RQ1, RQ2
Ward et al. (2022)	Pilot Study	Literature review past 10 years	29 rare conditions	Care pathway models	Lack of multi-disciplinary care model. More interventions and collaborations needed.	Theme 4	RQ3

availability of adequate evidence to implement protocols in clinical practice, health, and social policy[18]. Individuals diagnosed with rare conditions consistently expressed feeling regularly misunderstood due to a widespread lack of awareness[19]. There is a limited number of experts with comprehensive knowledge and experience in rare diseases, and standardized guidelines and specific infrastructure for these conditions are needed[20].

Subtheme 1a: Lack of Awareness Among Educators and School Staff

There is a deficiency in knowledge and attitudes among school administrators, teachers, and staff members, who fail to recognize a student's condition as a genuine health concern[18]. Classmates and teachers failing to take the student's illness seriously can result in discriminatory behavior, including isolation, stigma, and reduced educational opportunities[18]. The confidence and competence of educators play a significant role in influencing students' learning development, particularly in inclusive education settings where there is a growing demand for teachers to address the needs of students with rare diseases[21]. Intersectoral partnerships through policies supporting initiatives like the rare disease teacher resource modules can help educators identify red flags indicating potential disease in students, and address the academic needs of students with rare diseases[22].

Subtheme 1b: Lack of Awareness Among Peers

Social stigma and discrimination among peers represent a significant dimension of the awareness gap identified in the literature. When classmates fail to understand or acknowledge a student's rare disease, the consequences extend beyond individual interactions, affecting the overall school experience. Students with rare diseases consistently reported experiences of being misunderstood by their peers, leading to social isolation and exclusion[19]. Adama et al.[23] found that nearly half of parents reported their child with a rare disease was bullied by peers, highlighting the serious social consequences of peer unawareness.

Subtheme 1c: Lack of Awareness in Healthcare and Biopharma

Beyond the educational setting, the lack of awareness among healthcare providers and the biopharmaceutical industry further compounds the challenges faced by students with rare diseases and their families. Ashtari and Taylor[24] found that limited scientific knowledge and information about rare disorders result in delays in diagnosis, causing significant stress on students with genetic disorders, and prompting their parents or caregivers to educate themselves about the condition and explore symptom management strategies. The limited number of experts with comprehensive knowledge and experience in rare diseases, and the absence of standardized guidelines, underscore the systemic nature of this awareness gap across sectors[20].

Theme 2: Adverse Psychosocial Challenges Experienced by Children with Rare Diseases

Children with rare disorders often experience psychosocial stress, which manifests in forms such as stigma, bullying, depression, isolation, discrimination, and anxiety. Growing up with a chronic disease in adolescence is associated with impaired psychosocial functioning[25]. Psychosocial impairment can lead to school difficulties, affecting these students' quality of life and academic outcomes.

Subtheme 2a: Emotional and Mental Health Challenges

The quality of life and everyday experiences of students with rare diseases may decline significantly. Poor quality of life can be compounded by maladaptive health behaviors, leading to depression, self-harm, and suicidal ideation, low self-esteem, impaired physical functioning, and psychological distress[7]. In a cross-sectional survey of 41 parents of school-aged children and adolescents diagnosed with a rare disease, the findings revealed that 75.6% of participants experienced stigma, and almost half reported their child was bullied[23]. Adolescents with rare chronic disorders characterized by physical symptoms frequently face stressful situations and emotional distress, leading to a higher risk of mental health problems, impaired social functioning, and stigmatization[25].

Subtheme 2b: Social Challenges and Isolation

Classmates and teachers failing to take the student's illness seriously can result in discriminatory behavior, including isolation, stigma, and reduced educational opportunities[18]. These actions can lead to a feeling of loneliness, and can affect the mental health of children with rare diseases. Students with rare diseases consistently reported feeling regularly misunderstood due to a widespread lack of awareness among their peers and educators[19]. Students should experience a sense of belonging and inclusion, and enhancing the knowledge of teachers and administrators is essential in establishing adequate support systems[21].

Subtheme 2c: Academic Impact and School Performance

United States schools may find it challenging to accommodate students with rare diseases due to the increasing number of children with complex medical conditions, which may result in poor socialization, excessive stress, and academic stagnation[8]. Overall academic achievements are compromised for students with rare diseases, and there is a documented lack of research and

understanding on how rare diseases specifically affect educational outcomes[21]. Thongseiratch and Chandeying[26] confirmed that chronic illness directly impacts student academic performance, creating a cycle of educational disadvantage that further affects the psychosocial well-being of students with rare diseases.

Theme 3: Adverse Psychosocial Challenges Experienced by Families and Caregivers

Rare diseases can have a consequential effect on families. These families may face significant challenges and a range of emotions, leading to feelings of helplessness. Depending on the severity of the disease, parents may experience grief over their child's health, which is a normal reaction to loss, and the stages of grief include denial, anger, bargaining, depression, and acceptance[27]. Carers of children living with rare diseases face a significant burden and a multitude of unmet needs[28].

Subtheme 3a: Caregiver Burden and Mental Health

Parents caring for children with rare diseases are concerned about the long-term progression of their child's condition, and encounter numerous challenges that significantly affect their psychosocial well-being and overall quality of life[29]. Caregivers voiced concerns about the adverse effects on their quality of life, the presence of stigma, and the strain on their mental health, all attributed to the lack of external support in terms of psychological, social, emotional, and financial assistance[28]. Rihm et al.[30] found that the psychosocial situation of family caregivers of children with rare diseases is severely strained, and clinical distress, including physical, social, and family, cognitive, emotional, and parenting issues, was reported by almost 90% of caregivers.

Subtheme 3b: Family Dynamics and Relationships

Parenting a child with a rare disease can be stressful for parents and other family members, including healthy siblings, and can negatively influence marital relationships, parent-child interactions, and family cohesion[5]. Parents of children with a rare genetic disease often assume new roles and responsibilities that can become a significant source of stress, while the waiting process to receive a diagnosis can present challenges for the child and the parents[27]. Furthermore, in a case study that evaluated a family's unique story of how a rare disease affected their dynamics, their daughter grappling with the grief of the premature death of her siblings and the lifelong implications of her diagnosis created a mental whirlwind, encompassing a strange mix of relief, guilt, anger, shock, confusion, psychological trauma, and survivor's guilt[31]. Currie and Szabo[32] found that mothers mainly provide caregiving for children with rare diseases, and taking care of children outside of the traditional discourses of motherhood makes them more likely to experience stress and burnout, as well as oppression and marginalization.

Subtheme 3c: Financial Burden

The financial burden associated with rare diseases represents a significant and often overlooked dimension of the psychosocial challenges experienced by families. The financial burden associated with prolonged diagnostic journeys compounds the existing psychosocial challenges, and further limits access to resources and support systems[33]. Brandt et al.[34] found increased financial burden among families of children with spinal muscular dystrophy, with caregivers experiencing reduced paid employment and working hours to meet the care demands of their children.

Theme 4: Limited Resources and Support Systems

Limited resources and inadequate support systems impede efforts to address psychological needs, thereby hindering improvements in the overall school experience for students and families, enhancing their well-being, and ensuring a better quality of life. Every child should have equitable access to education, regardless of their economic background or health status. Paz-Lourido et al.[18] highlighted the importance of establishing an equitable educational system that ensures equal opportunities and non-discriminatory practices, focusing on accommodating students' individual needs arising from disabilities and challenges relating to chronic illness.

Subtheme 4a: Limited Educational Support Systems

There are limited resources and interventions on rare diseases to accommodate students with complex medical needs and their families. The confidence and competence of educators play a significant role in influencing students' learning development, especially in inclusive education settings where there is a growing demand for teachers to address the needs of children with rare diseases[21]. Teachers need to ensure that their students in K-12 with rare diseases feel included in the classroom and social settings, as this is pivotal for laying the foundation for their academic success and overall school experience.

Subtheme 4b: Limited Mental Health Resources

Mental health is a growing area of concern in the school system, and the needs of students with rare diseases intersect directly with this concern in ways that remain underaddressed. Unmet healthcare needs for individuals with chronic medical conditions are linked to a decline in quality of life, and an increased probability of negative behaviors and poor health outcomes[9]. The absence of adequate mental health resources and support systems leaves both students and families without the infrastructure necessary to navigate the psychosocial challenges associated with rare diseases[19].

Subtheme 4c: Need for Interdisciplinary Collaboration

Given the unique needs of students with rare diseases, it is imperative to establish an interdisciplinary and intersectoral approach between health services and educational environments, to develop a comprehensive strategy centered on children with rare diseases, addressing their specific needs, and implementing effective policies and actions to support their educational requirements[18]. The lack of a multidisciplinary care model to address the needs of individuals with rare diseases remains a significant gap, with additional interventions and collaborations needed to improve outcomes[35].

Theme 5: The Diagnostic Odyssey and Its Impact on Students and Families

The diagnostic odyssey represents one of the most distinctive and challenging aspects of the rare disease experience for students and their families. There is no universal definition of rare diseases, and the prevalence of these complex conditions varies significantly across regions of the world[33]. This heterogeneity, combined with limited awareness among healthcare providers, contributes to prolonged, often distressing diagnostic journeys with profound psychosocial consequences for students and families.

Subtheme 5a: Delayed and Missed Diagnoses

Ashtari and Taylor[24] found that limited scientific knowledge and information about rare disorders result in delays in diagnosis, causing significant stress on students with genetic disorders, and prompting their parents or caregivers to educate themselves about the condition and explore symptom management strategies. According to a survey on rare diseases, families from the United Kingdom and the United States usually consult with approximately eight physicians and receive two to three misdiagnoses, leading to a diagnostic journey lasting 5.6 to 7.6 years, resulting in unnecessary medical follow-ups and financial burden[33]. The prolonged nature of the diagnostic odyssey means that students with rare diseases may spend significant portions of their K-12 educational experience without a clear diagnosis, further complicating efforts to obtain appropriate educational accommodations and support.

Subtheme 5b: Psychosocial Impact of the Diagnostic Journey

The diagnostic odyssey carries significant psychosocial consequences for students with rare diseases. The uncertainty and prolonged waiting associated with seeking a diagnosis can exacerbate existing emotional and mental health challenges, creating a compounding effect on the overall psychosocial burden experienced by these students. Ashtari and Taylor[24] documented that individuals with rare diseases experienced significant pain and felt isolated during the diagnostic process. The lack of a clear diagnosis can paradoxically increase mental well-being concerns, as students and families navigate an uncertain and often isolating journey without the clarity and direction that a confirmed diagnosis can provide[18].

Subtheme 5c: Impact on Families During the Diagnostic Process

The diagnostic odyssey does not affect students in isolation, but has profound consequences for the entire family unit. Parents of children with a rare genetic disease often assume new roles and responsibilities that can become a significant source of stress, while the waiting process to receive a diagnosis can present challenges for the child and the parents[27]. Kolemen et al.[27] found that anxiety levels and depression among parents increased significantly while awaiting the diagnosis, confirming that the diagnostic process itself represents a distinct psychosocial stressor for families. Furthermore, Zhang[31] documented in a case study that the lifelong implications of a rare disease diagnosis created a mental whirlwind for families, encompassing grief, trauma, and survivor's guilt, illustrating the profound and lasting psychosocial impact of the diagnostic journey on family dynamics and well-being.

Discussion

The findings of this systematic literature review confirm that K-12 students with rare diseases and their families face a distinct and largely overlooked set of psychosocial challenges within the educational system and beyond. The five themes that emerged from the reviewed literature are consistent with and reinforce one another, revealing a system that lacks the awareness, resources, and coordinated infrastructure necessary to adequately serve this population. A rare disease can have a significant effect on a child's quality of life and the quality of their family life, and the diagnosis itself can trigger a range of emotions that profoundly affect mental health across the entire family system[23]. When examined through the lens of Vygotsky's sociocultural theory, these findings reveal not simply a collection of individual challenges, but a systemic failure of the social environment to support the development, well-being, and educational success of one of the most vulnerable and underserved student populations in the United States.

Theme 1: Lack of Awareness of Rare Diseases

The lack of awareness documented in this review is perhaps the most consequential finding of all, as it lies at the root of every other challenge identified. Stigma cannot be addressed without awareness. Educational accommodations cannot be implemented without awareness. Peer inclusion cannot be fostered without

awareness. In this sense, awareness is not one challenge among many. It is the precondition for all meaningful progress.

Subtheme 1a: Lack of Awareness Among Educators and School Staff

Vygotsky's sociocultural theory holds that the adults in a child's learning environment are the architects of that child's developmental opportunity. Teachers and administrators who do not understand a student's rare disease cannot scaffold learning effectively, recognize when a student is struggling for medical rather than academic reasons, or create the inclusive environment that Vygotsky identified as essential to healthy cognitive and emotional growth[10]. The implications for practice are direct and urgent. Rare disease education must become a component of teacher training and ongoing professional development, not an afterthought. The rare disease teacher resource modules referenced in the literature represent exactly the kind of intersectoral solution this crisis demands[22], and research-grounded tools designed to support educator awareness represent a critical step toward closing this gap at the school level.

Subtheme 1b: Lack of Awareness Among Peers

The social consequences of peer unawareness are not simply uncomfortable. They are developmentally damaging. Vygotsky understood that children learn who they are through their interactions with others[11]. When a student with a rare disease is consistently met with confusion, dismissal, or cruelty from peers, the message they internalize is not merely that others do not understand their condition. It is that they do not belong. This internalized sense of exclusion has lasting consequences for identity, self-worth, and mental health that extend well beyond the K-12 years. Peer education and awareness initiatives are not supplemental programs. They are developmental necessities.

Subtheme 1c: Lack of Awareness in Healthcare and Biopharma

The awareness gap does not begin at the school door. It begins in the healthcare system, where diagnostic delays of up to 7.6 years mean that many students arrive at school without a confirmed diagnosis, and therefore without any framework for understanding their own experience[33]. When the healthcare system fails to identify and name a child's condition in a timely manner, it denies that child and their family the language, the resources, and the legitimacy that a diagnosis provides. The biopharmaceutical industry's historical underinvestment in rare disease research, particularly for conditions disproportionately affecting communities of color, such as sickle cell disease, has compounded this problem for decades. Addressing the awareness gap requires action across all three sectors simultaneously.

Theme 2: Adverse Psychosocial Challenges Experienced by Children with Rare Diseases

The psychosocial challenges documented in this review do not exist in isolation from one another. They form a compounding cycle in which each challenge amplifies the next. Stigma leads to isolation. Isolation leads to anxiety and depression. Depression impairs academic performance. Academic failure deepens the sense of difference and inadequacy. Inadequacy makes a student less likely to advocate for themselves or seek support. Breaking this cycle requires intervention at multiple points simultaneously, not a single programmatic response.

Subtheme 2a: Emotional and Mental Health Challenges

The finding that 75.6% of students with rare diseases experience stigma, and that almost half are bullied, is not simply a statistic[23]. It is an indictment of the school environments these children navigate every day. From a Vygotskian perspective, the emotional environment of the classroom is not separate from learning. It is the foundation of learning. A child who fears ridicule, who carries the weight of an invisible condition, and who lacks a trusted adult who understands their experience, cannot fully engage in the zone of proximal development that Vygotsky identified as the engine of cognitive growth[10]. Mental health support for students with rare diseases is therefore not a supplemental service. It is an educational imperative.

Subtheme 2b: Social Challenges and Isolation

Social isolation is one of the most consistently documented experiences of students with rare diseases across the included literature, and it is also one of the most developmentally damaging. Vygotsky's entire theoretical framework rests on the premise that human development is fundamentally social[12]. A student who eats alone, who is excluded from recess, and who has no peers who understand their experience is being denied the very conditions that Vygotsky identified as necessary for healthy development. The coping strategies identified in the literature, including problem-solving, cognitive reappraisal, and positive thinking[36], are important, but they place the burden of adaptation entirely on the child. The social environment itself must change.

Subtheme 2c: Academic Impact and School Performance

The academic consequences of rare disease are both direct and indirect. Directly, symptoms such as fatigue, pain, and cognitive challenges interfere with concentration, attendance, and performance. Indirectly, the psychosocial burden of stigma, isolation, and anxiety consumes cognitive and emotional resources that would otherwise be available for learning. United States schools are currently

ill-equipped to address either dimension of this challenge[8]. The implications for policy are clear. IEPs, 504 plans, and flexible attendance policies are not accommodations that give students with rare diseases an unfair advantage. They are tools for creating the level playing field that every student deserves.

Theme 3: Adverse Psychosocial Challenges Experienced by Families and Caregivers

One of the most significant contributions of this review is its documentation of the psychosocial burden experienced by families and caregivers as a distinct and equally urgent concern. Vygotsky recognized that the family is the child's first and most fundamental social environment[10]. When that environment is under severe strain, the developmental consequences for the child are profound. The 90% rate of caregiver distress documented by Rihm et al.[30] is not a peripheral finding. It is a central indicator of the systemic inadequacy of current support structures.

Subtheme 3a: Caregiver Burden and Mental Health

A caregiver who is experiencing depression, anxiety, financial strain, and social isolation is not positioned to provide the consistent, emotionally available parenting that Vygotsky identified as foundational to healthy child development. The research is unambiguous: when caregivers suffer, children suffer. Yet the support systems available to caregivers of children with rare diseases remain critically inadequate. Addressing caregiver mental health is not separate from addressing student outcomes. It is inseparable from them. Policy interventions that support caregiver well-being, including respite care, mental health resources, and financial assistance, are investments in the developmental outcomes of the children they serve.

Subtheme 3b: Family Dynamics and Relationships

The rare disease diagnosis does not affect one person in a family. It reorganizes the entire family system. Marital relationships are strained, siblings feel overlooked, and parents are forced into roles they were never prepared for[5]. The case study documented by Zhang[31], in which a family described their experience as a mental whirlwind of grief, guilt, and trauma, illustrates the profound and lasting nature of this reorganization. From a Vygotskian perspective, the disruption of healthy family dynamics is a disruption of the child's most immediate social scaffold. Rebuilding and supporting that scaffold requires family-centered interventions that go beyond the individual student.

Subtheme 3c: Financial Burden

The financial burden associated with rare disease is both a cause and a consequence of psychosocial distress. Families who spend years and significant financial resources pursuing a diagnosis while managing the daily costs of complex care are families under compounding and relentless pressure[33,34]. This financial strain directly limits access to the very resources, including mental health support, educational advocacy, and specialist care, that could alleviate the psychosocial burden. Policy solutions must address the financial dimensions of rare disease alongside the clinical and educational ones.

Theme 4: Limited Resources and Support Systems

The shortage of resources and support systems documented in this review is both a cause and a reflection of the systemic invisibility of students with rare diseases in educational policy and practice. When a population is too small, too heterogeneous, and too poorly understood to generate consistent policy attention, the result is predictable. Children fall through the cracks of systems that were not designed with them in mind. Addressing this requires deliberate, coordinated, and sustained effort across multiple sectors.

Subtheme 4a: Limited Educational Support Systems

The current educational support landscape for students with rare diseases is reactive rather than proactive. Schools respond when crises arise rather than building the structures and knowledge necessary to prevent them. The interdisciplinary approach called for consistently across the literature[18,22] represents a fundamentally different model, one in which healthcare providers, educators, families, and policymakers collaborate continuously rather than episodically. This model is consistent with Vygotsky's vision of development as a collaborative social process, one that requires all members of a child's community to work in concert.

Subtheme 4b: Limited Mental Health Resources

The mental health needs of students with rare diseases are significant, well-documented, and chronically unmet. School counselors and psychologists who are not trained in the specific psychosocial dimensions of rare disease are not equipped to provide meaningful support. The development and dissemination of rare-disease-specific mental health resources must become a priority for school systems, healthcare organizations, and the biopharmaceutical industry alike. Effective coping strategies exist[36], but they must be taught, supported, and reinforced within a system that understands the unique context in which these students are applying them.

Subtheme 4c: Need for Interdisciplinary Collaboration

The evidence reviewed in this study consistently points to one conclusion: no single sector can solve this problem alone. The diagnostic odyssey requires action from healthcare. The educational gaps require action from schools and policymakers.

The financial burden requires action from the government and industry. The psychosocial needs require action from mental health professionals and community organizations. Implementing a coordinated care model has been shown to reduce adverse outcomes among students with complex medical conditions[37], and this model must become the standard rather than the exception.

Theme 5: The Diagnostic Odyssey and Its Impact on Students and Families

The diagnostic odyssey is perhaps the most unique dimension of the psychosocial burden documented in this review. Unlike the challenges of stigma or resource scarcity, which affect many student populations, the experience of spending years without a name for what is wrong is specific to rare diseases. And its consequences ripple through every other theme. Without a diagnosis, educational accommodations cannot be secured. Without a diagnosis, peers and educators have no framework for understanding. Without a diagnosis, families cannot access the rare disease community, which is often the most meaningful source of support available to them.

Subtheme 5a: Delayed and Missed Diagnoses

A diagnostic journey lasting 5.6 to 7.6 years means that a student may enter kindergarten, progress through elementary school, and reach middle school before anyone can tell them and their family what is wrong[33]. The educational, psychosocial, and developmental consequences of this prolonged uncertainty are compounding and immense. From a Vygotskian perspective, the absence of a diagnosis is also the absence of a shared language, and Vygotsky understood language as the primary vehicle through which social understanding and cognitive development are built[13]. Reducing diagnostic delay requires investment in rare disease medical education, expanded access to genetic testing, and stronger coordination between healthcare and educational systems.

Subtheme 5b: Psychosocial Impact of the Diagnostic Journey

The psychological experience of the diagnostic journey is not simply stressful. It is disorienting in a way that is difficult to convey to those who have not lived it. Students and families navigate a world in which something is clearly wrong, but no one can confirm what it is. They are met with skepticism, misdiagnosis, and repeated disappointment. The isolation this creates is compounded by the fact that without a diagnosis, these families cannot easily find others who share their experience. The psychosocial impact of this isolation is consistently documented across the included studies and represents a distinct area of need, separate from but related to the challenges associated with the disease itself.

Subtheme 5c: Impact on Families During the Diagnostic Process

The diagnostic process does not pause family life. Parents continue working, siblings continue growing up, and relationships continue evolving, all while the family carries the invisible weight of an unresolved medical crisis. The grief, guilt, and trauma documented by Zhang[31] are not responses to the diagnosis itself. They are responses to the journey. Policy and clinical interventions that focus exclusively on the post-diagnosis period miss a critical window of intervention. Families navigating the diagnostic odyssey need psychosocial support, financial resources, and community connection from the moment they begin seeking answers, not only after they find them.

Synthesis and Implications

Taken together, the five themes identified in this review reveal a deeply interconnected set of challenges that cannot be addressed in isolation. The lack of awareness creates the conditions for stigma and isolation. Stigma and isolation compound the psychosocial burden. The psychosocial burden strains the family system. The strained family system further compromises the child's developmental environment. And through all of it, the diagnostic odyssey extends and deepens every dimension of suffering. This is not a collection of separate problems. It is a system of interlocking failures, and it requires a systemic response. Vygotsky's sociocultural theory provides a unifying framework for understanding why, because healthy development requires a healthy social environment, and these students do not have one. Building that environment requires the sustained, coordinated effort of every sector that touches the lives of children with rare diseases, and it requires the kind of evidence-based advocacy that this review is intended to support.

Limitations

This review is subject to several limitations. The reviewed literature was limited to English-language peer-reviewed publications, which may have excluded relevant studies published in other languages. Additionally, the existing body of literature frequently captures only a single moment in time, rather than longitudinal outcomes across the full K-12 experience. The inclusion of international studies, while intentional to address the limited United States-specific literature base, may have introduced variability in educational and healthcare system contexts. The heterogeneity of rare diseases across the 14 included studies limits the ability to draw conclusions specific to any single condition. Future research should consider conducting comparative studies between specific categories of rare diseases to examine whether the psychosocial impact varies by condition type, severity, or diagnostic timeline. These limitations underscore the need

for sustained, long-term, and methodologically diverse research, as identified in this review's recommendations.

Conclusions

This systematic literature review confirms that K-12 students with rare diseases and their families face significant and largely overlooked psychosocial challenges within the educational system. Five overarching themes emerged from the reviewed literature: lack of awareness of rare diseases among educators and key stakeholders; adverse psychosocial challenges experienced by children with rare diseases; adverse psychosocial challenges experienced by families and caregivers; limited resources and support systems; and the diagnostic odyssey and its impact on students and families. These findings confirm that 90% of rare diseases have no cure, that rare diseases are complex and affect a limited number of individuals, making them difficult to research, and that there is a vast unmet need for these students and their families.

Based on the findings of this review, the following recommendations are offered:

First, it may be helpful to conduct more longitudinal mixed-method studies using surveys and interviews as data sources to better understand the challenges that students and families with rare diseases face and the long-term consequences regarding academic experience, quality of life, and mental health.

Second, there may be a need for more comparative studies between students with rare chronic diseases and those with more common diseases, to increase awareness and understanding of uncommon conditions.

Third, it is essential to educate and raise awareness among key stakeholders, including teachers, school administrators, and peers, about the unique psychosocial challenges that students with rare diseases face. This awareness may help reduce stigma and instill confidence in students with complex medical conditions.

Fourth, policies that support the academic success of students with rare diseases should be prioritized and expanded to provide them with special education services, accommodations, and resources.

Fifth, there is a need for enhanced, comprehensive support systems to promote collaboration and partnerships among researchers, healthcare providers, educators, biopharma companies, and policymakers. Improving support systems and resources to enhance the quality of life and mental well-being will foster a positive school experience for these students and their families.

Declarations

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Conflict of Interest

The author declares no conflicts of interest. The research design and reporting were not influenced by academic, financial, or other personal interests. The author is the founder of Rare Advocacy Institute, LLC, a research translation platform grounded in this doctoral research.

Data Availability

All data supporting the findings of this review are available within the published peer-reviewed literature cited in the references section of this article.

Prior Dissemination

This systematic literature review was conducted as part of a doctoral dissertation published in 2024, and is currently indexed in ProQuest and Google Scholar. The manuscript submitted here represents an original journal article derived from that doctoral research, and has not been previously submitted to or published in any other peer-reviewed journal.

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