Journal of Pediatric Nursing 68 (2023) e16–e26

Contents lists available at ScienceDirect

Journal of Pediatric Nursing

journal homepage: www.pediatricnursing.org

Children and young people's experiences of living with rare diseases: An integrative review

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ARTICLE INFO

Article history:
Received 10 July 2021
Revised 24 October 2022
Accepted 24 October 2022

Keywords:
Children
Experience
Rare diseases
Young people
Integrative review

ABSTRACT

Problem: Rare diseases are any disease affecting fewer than five people in 10,000. More than 8000 rare diseases and 50–75% of all rare diseases affect children. The purpose of this review was to critically appraise and synthesize existing literature relating to the impact of rare diseases on children's day-to-day lives.
Eligibility criteria: An integrative literature review was conducted using the CINAHL Plus, PsycINFO, and PubMed databases. Studies were included if they were a primary source was published between the years 2005 and 2019 and written in the English language.
Sample: Eight primary sources met the inclusion criteria.
Results: Seven main themes emerged from the review as follows: (i) the experience of stigmatisations, (ii) self-consciousness, (iii) restrictions in independent living, (iv) developing resilience/coping strategies, (v) psychological and emotional impact, (vi) social impact vs social connectedness and (vii) transition challenges.
Conclusions: The experience of having a rare illness differed across different age groups. Children (typically aged 3–10) with rare diseases generally view themselves and their lives the same way like their healthy peers. They were more likely to report being adaptive and resilient than those aged 12 or older. Young people reported being different compared to young children, and they faced numerous challenges related to their illness.
Implications for practice: To provide the best possible level of care for children and families with rare disorders, health services must be informed and equipped to provide the necessary supports specific to the unique needs of children and young people living with rare diseases.

Background

Rare diseases are characterized by their relatively low prevalence, defined in Europe as affecting less than 1 in 2000 individuals (European Commission(EU), 2020) or less than 200,000 individuals in the USA (Orphan Drug Act of 1983, 2022; The United States Rare diseases act of 2002, 2022). Although small, this translates into approximately 350 million people living with a rare disease globally. About 8000 distinct rare diseases affect 6–8% of the EU population, i.e., between 27 and 36 million people, signifying rare diseases as a public health issue in Europe and globally (de Vrueh et al., 2013). Rare diseases are significantly associated with mortality, morbidity, and disability and are far more likely to be found in children and young people (Gunne et al., 2020). Approximately 70% of all rare diseases affect children, and 30% of these die before their fifth birthday, out of which 81.9% of rare diseases are ultra-rare (less than 1 per 100,000 persons) (Rare Disease Taskforce, 2020; Rare Disease UK, 2018; Department of Health (DOH),

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https://doi.org/10.1016/j.pedn.2022.10.014
0882-5963/

2014). Since most rare diseases affect children, it has been reported in the recent study by (McMullan et al., 2021) to understand carers' reported experiences caring for someone with rare diseases and 35.7% caring for children living with rare conditions. Children living with rare illnesses face several difficulties, including their school attendance, frequency of medical appointments, inaccessibility of facilities, and non-adapted teaching methods (Kole et al., 2021). One of the recommendations by the 2030 RD report (Kole et al., 2021) was to increase the quality of education by spreading knowledge on rare diseases to teachers, educators, and the next generation.

Most importantly, rare diseases affect those diagnosed and their families, friends, carers, and society as a whole (McMullan et al., 2020; Rare Disease Taskforce, 2020; Somanadhan et al., 2020; Spencer-Tansley, 2018). Individual rare diseases receive limited attention because of their rarity, which means that the total number of people with a specific condition can be very small. Many paediatric rare disorders are complex and chronically debilitating, which often cause intellectual and physical disabilities that place substantial demands requiring multidisciplinary input and regular hospitalizations (Elliott & Zurynski, 2015; Gunne et al., 2020; Somanadhan & Larkin, 2016). Unfortunately, most rare diseases (estimated 95% of rare diseases) still do not have an approved treatment even though rare disorders pose significant health, psychological, social and economic burdens to children and their families due to the inherent complexity and heterogeneity of these diseases (Eurordis, 2017; Kole et al., 2021).

Living with a rare disease is an ongoing learning experience for children and their families (Eurordis, 2017). Limited published literature conclusively addresses the context of the rare disease patients and carers 'patient experts' and their experiences of living with rare diseases. Due to the complexity and heterogenicity of rare diseases, there are challenges to all dimensions of children's lives. Child and family-focused experiences of living with a rare illness must be explored and targeted. This involves looking at how caregivers experience the specific burdens associated with their son or daughter's rare disease. We must deepen our understanding of children's perspective of living with rare diseases. No review has addressed the full range of studies that have examined the experiences and challenges among children living with a rare disease. This review is a part of a more extensive project called SAMPI: Giving voice to the children and their families with Rare Diseases through Sand Play, Arts Therapy, Music Therapy, Photovoice, Interviews and Survey to answer a simple research question "what is it like for a child to live with a rare disease and how can children be best supported to express this?"

Purpose

To critically appraise and synthesize research that examines children and young people's experience and their day-to-day challenges of living with a rare condition. The objectives are to (i) identify literature focusing on the children and young people's experience of living with rare diseases; (ii) determine the methodological quality of evidence; (iii) describe children and young people's experience of living with a rare disease, and (iv) make recommendations for future research and clinical practice.

Methods

An integrative review of the literature was utilised to better summarize past empirical or theoretical literature to understand a particular phenomenon (Broome, 1993). The integrative review plays a significant role in developing evidence-based practices and permits a narrative approach to evidence synthesis (Hopia et al., 2016). Further, it allows diverse methodologies to gain insight into the topic of concern (Whittemore & Knafl, 2005). This review was conducted and reported following (Whittemore & Knafl's, 2005) problem identification, literature search, data evaluation, data analysis, and presentation.

Review question

The population, concept and context (PCC) framework (ref: Joanna Briggs Institute (JBI), Peters et al., 2020) was utilised to identify/construct the main concepts of the review question, which is: *"What is known about children and young people's (P) experiences of living with (C) a rare disease (C)?"*

Literature search strategy

Systematic searches were conducted using the databases CINAHL Plus, Pubmed and PsychINFO to ensure that we focus on nursing and allied health-related journals to capture children and young people's experience and their day-to-day challenges of living with a rare condition. A comprehensive search strategy (Table 1) was developed following key phrases using the PCC framework, which was modified according to the databases. The Boolean operators "OR" and "AND" were used to combine terms within and across the constructs. A librarian was involved with the development of the search strategy.

After initial searches, it was agreed to add the names of specific diseases classed as rare to the search terms. Further, the term 'chronic illnesses' and related words were also used in the search to yield more content. These additions were included as few studies may explicitly focus on children's experiences of having rare diseases. Additional studies could be useful to draw comparisons between the experience of patients/children with chronic illnesses and patients/children with a rare disease and 'English' as a first language. Since this review is a part of a more extensive project, careful consideration was given to the search timeframe; as one objective of the study was to make recommendations for policy and practice, more up-to-date research was considered most appropriate. Therefore, searches were limited to evidence sources published over the past 15 years, between 2005 and 2020. However, in 2020, COVID-19 is an unprecedented public health emergency that emerged globally, and as a result, we decided to limit the search to 2019.

Inclusion criteria

Inclusion criteria were limited by (a) publication year between 2005 and 2019, (b) English language, and (c) age of participant from birth to 24 years old (d) Full Text Only (e) Peer-reviewed primary studies from academic journals and grey literature. The age range (0–24 years) was a pragmatic decision based on the literature on pubertal development in children and young people with rare diseases.

Exclusion criteria

Exclusion criteria were limited by (a) adults (24+ Years) (b) Language (other than English) (c) any articles published pre 2005 (d) any review articles (d) no full text available.

Search

Numerous tools, such as COVIDENCE, have been designed to help with one or more steps in the review process (Kellermeyer et al., 2018). However, this software platform license wasn't available at the moment of the review process. Studies returned from the search

Table 1
Review search terms.

PCC Tool	Search terms
POPULATION	Child* (children, childhood), Young people, Young person Adolescen* (adolescence, adolescents), Teenage* (teenager, teenagers)
CONTEXT	living with, lived experience*, life experience
CONCEPT	rare disease/s, OR rare condition AND 'chronic illnesses'

strategy were collated across the databases, and duplicates were removed. Two people then screened titles and abstracts for the suitability, followed by full-text screening to assess eligibility for inclusion. Reasons for the exclusion of studies were recorded and noted. The review team were aware of the breadth of potential sources of grey and unpublished literature. A number of steps were taken to ensure a systematic search of grey and unpublished literature relating to the phenomenon of interest. First, grey literature databases (e.g. Open Grey) were searched using keywords and phrases identified in published literature. Second, specific evidence sources were sought and screened, including abstracts submitted to conferences or workshops and dissertations (e.g. Electronic Theses Online Service). Finally, the review team contacted academic experts, professional societies and relevant organisations to ascertain the availability of any additional evidence sources not identified in previous searches of published, unpublished or grey literature. Additional six records were identified through other sources.

Quality appraisal

The methodological quality of included studies was appraised using the Hawker et al. (2002) quality assessment tool by two reviewers. This checklist is designed to be used in mixed reviews, and compared to other appraisal tools, Hawker et al. (2002) tool uses numeric scoring to assess the overall quality of studies. This tool estimates the overall quality of studies using nine appraisal dimensions, namely; (1) Abstract and title, (2) Introduction and aims, (3) Method and data, (4) Sampling, (5) Data analysis, (6) Ethics and bias, (7) Results, (8) Transferability or generalizability, and (9) Implications and usefulness. Each item can be answered and scored as 'good' (4 points), 'fair' (3 points), 'poor' (2 points) or 'very poor' (1 point). The numeric scoring allows one to identify a study's methodological strengths and limitations by producing a summary score. To create the overall quality grades, we used the following definitions: summary scores of 30 or more were defined as high quality (A), 24–29 points as medium quality (B), and scores of 23 or less as low quality (C).

Data extraction

A modified data-extraction form was utilised, based on The Cochrane Handbook of Systematic Reviews of Interventions Part 2: Checklist of items to consider in data extraction (Higgins & Deeks, 2011). Extracted data included the authors, year of publication, country, study tile, aim, number of participants in the study, data collection methods, quality appraisal score and a summary of findings.

Data synthesis

It is envisaged that studies would be diverse and largely qualitative, so a narrative synthesis of findings was proposed. Thematic analysis was used to identify patterns of themes within the included studies, i.e., patterns in the important or interesting data, and use these themes to address the review question (Clarke & Braun, 2013). This involved extracting the findings from each paper and identifying commonalities and patterns in the data related to the review aim (Braun & Clarke, 2006).

Results

A total of 1577 studies were identified via database searches (CINAHL Plus $n = 380$, PsychINFO $n = 500$, Pubmed $n = 697$) and 6 studies through other sources. After duplicates were removed, 1247 articles were screened by title and abstract. Thirteen full-text articles were assessed for eligibility, 5 articles were drawn to meet the inclusion criteria resulting in (review =3, wrong population = 2) final eight studies for the review. The PRISMA Flow diagram (Fig. 1)indicates the identification and selection of studies.

The studies' methodological quality assessment is shown in (Table 2) (Hawker et al., 2002). The eight studies were critically appraised and synthesized across four countries: Australia (2), United Kingdom (3), United States (2) and The Netherlands (1). Studies were published between 2011 and 2019 and comprised diverse methodologies such as Participatory Health Research (1), sequential mixed-methods approach (1), retrospective study (1) and a qualitative study (5). Four of these studies (Vines et al., 2018; Branch-Smith et al., 2018; Hanson et al., 2017; Adama et al., 2021) were rated as high quality (A) with a score between 30 and 36 points and 4 as medium quality (B) with a score between 24 and 29 points. No studies identified a low quality (C) score between 9 and23 points.

An overview of the data extracted from the included studies is presented in (Table 3). The conditions covered in this review of children's experiences had chronic kidney failure, cystic fibrosis, rare undiagnosed conditions, irritable bowel disorder, hemophilia, epilepsy, diabetes, juvenile idiopathic arthritis, sickle cell anaemia, nephritic syndrome and hematological malignancies, primary lymphoedema and Moebius syndrome.

Seven broad themes emerged from the data synthesis process (see Fig. 2): experiences of stigmatization, self-consciousness, restrictions in independent living, resilience/coping strategies, psychological and emotional impact, social impact versus social connectedness, and transitioning to adult healthcare. These shall now each be summarised in turn.

Experiences of stigmatization

Six studies reported children or young people's stigmatization experience (Currier & Zimmerman, 2019, Vines et al., 2018, Branch-Smith et al., 2018; Hanson et al., 2017; Bogart, 2014; Christie et al., 2011). Goffman (1963) defined stigma as "an attribute that is deeply discrediting" (p. 3). Self-consciousness and stigmatization due to their illness were prevalent themes across the literature, differentiated by age group. Bogart (2014) examined the social experiences of 10 adolescents aged 12–17 years with Moebius Syndrome, a rare condition involving congenital facial paralysis. Participants reported prejudice and bullying, including episodic and ongoing name-calling, harassment, and social exclusion. In another study, Branch-Smith et al. (2018) examined the peer bullying experiences of young people with cystic fibrosis and the associations between school bullying and these young people's psychological well-being. Cystic fibrosis is a rare disease with effects that are visible and potentially cause stigma. The overt symptoms of cystic fibrosis such as frequent coughing, shortness of breath, and medication use may lead peers to believe the person is contagious and underweight (for boys only), with flatulence symptoms also considered stigmatizing. Such stigma in cystic fibrosis (CF) was associated with school loneliness, anxiety, and depression (Branch-Smith et al., 2018). Low confidence was mentioned under the theme 'sensitivity/resilience' (Bogart, 2014), whereby sensitivity involved internalizing the stigma associated with having Moebius syndrome.

A project by Rare Disease UK, Spencer- Tansley (2018), was designed to explore and understand what it is like to be a child or young person affected by a rare disease. They reported that children with rare diseases are adaptive and resilient and do not see their illness as part of their own identity; however, as children grew older, they became more aware of feeling different compared to their healthy peers, wanted to be more actively involved in their care, and worried about the impact of living with a rare disease on their education. Abma and Schrijver *(2019)* study provided children with the opportunity to express their emotions or feelings, utilising photovoice as a child-friendly method to put children in a subject position to drive the research and social change. This was a way to provide space for children's voices and the problems they experienced and show audiences children's resilience, creativity, and capacity to see things in a new light. This approach is reflected in the Lundy (2007) model of participation with four key elements of space, voice,

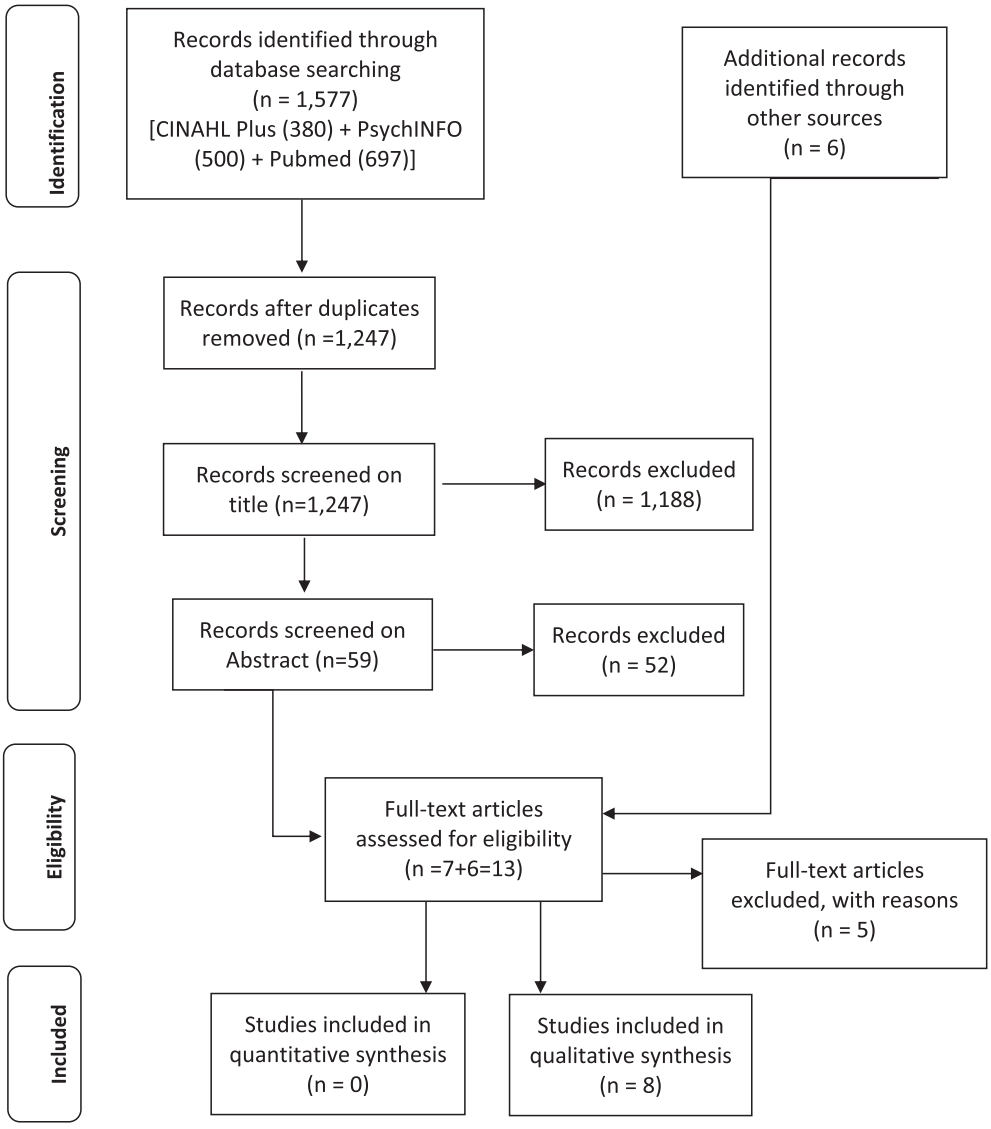

Fig. 1. PRISMA flowchart (2009).

audience, and influence, and these are essential for children's participation. This checklist also provides the young person or young people with an appropriate way to express their views and that those views will be given due weight.

Self-consciousness

Self-consciousness was a sub-theme in (Hanson et al., 2017, pp. 676) study of children and adolescents' experiences of primary lymphoedema. In this study, *"participants were 'exasperated' by the*

Table 2
Quality appraisal of included studies ($n = 8$).

Article	Abstract & Title	Introduction & Aims	Method and Data	Sampling	Data Analysis	Ethics and Bias	Findings/ Results	Transferability/ Generalizability	Implications & Usefulness	Total score	Overall Quality Grade
Abma & Schrijver, 2019	3	3	4	4	2	4	3	4	3	30	A
Bogart, 2014	3	3	4	3	4	2	3	3	4	29	B
Branch-Smith et al., 2018	4	2	3	4	3	4	3	4	4	31	A
Christie et al., 2011	3	3	2	3	3	3	2	3	2	24	B
Currier & Zimmerman, 2019	3	3	3	3	3	1	4	4	4	28	B
Hanson et al., 2018	4	3	4	4	4	4	4	4	4	35	A
Spencer- Tansley, 2018	3	2	4	3	1	1	3	3	4	24	B
Vines et al., 2018	4	3	4	3	4	4	4	3	4	33	A

Scoring across each dimension is from 1 (poor) to 4 (good), using the Hawker et al. (2002) quality assessment tool. Summary scores range from 9 to 36, and graded as high-quality A (30–36 points), medium-quality B (24–29 points) and low-quality C (9–23 points).

Table 3
Overview of the studies (n = 8) included in this review.

Author, year	Country	Rare disease/condition	Sample	Methodological approach	Focus	Key finding
Currier & Zimmerman, 2019	USA	Haemodialysis-dependent males and females.	N = 500, aged 5–25 years	Qualitative (directed content analysis)	Creative writing	Four metaphors from previous adult meta-synthesis were confirmed: (1) Having a physical shackle in life (2) feeling mental and emotional distress, (3) relying on haemodialysis machine, (4) dealing with problems.
Spencer-Tansley, 2018	UK	Rare or undiagnosed condition.	*N* = 60 aged 3–10 years 11–17 years	Participatory approach (Qualitative)	Write/draw	Children affected by rare diseases are adaptive and resilient and do not see their rare disease as a fundamental part of their identity. Young people felt they should be given more support around how to deal with the aftermath of receiving a diagnosis and spoke of a lack of support from healthcare professionals around the emotional aspects of living with a rare disease. They also stated that it was crucial for them to be involved in care but that this was seldom a reality.
Vines et al., 2018,	UK	Cystic Fibrosis	*N* = 9 Aged 12–19	Qualitative methodology	Semi-structured face-to-face interviews	Main themes were: (1) adjusting to living with isolation, (2) protection from a sense of threat, (3) noticing differences, (4) integrating the experience. Subthemes were: (1) a restrictive necessity, an imposed obligation, adjusting to my reality; (2) I could make others ill, others could make me ill, striving to stay safe day-to-day; (3) differences to others without CF, disruption to 'normal' self; (4) connecting to CF identity, distancing from CF identity.
Christie et al., 2011	UK	Chronic illnesses: irritable bowel disorder, hemophilia, epilepsy, diabetes, juvenile idiopathic arthritis, sickle cell anaemia, nephritic syndrome and hematological malignancies	*N* = 36 aged 3–11 years	Interpretative Phenomenological Analysis (IPA)	Board Game	Nine themes were identified: (1) family, (2) school, (3) peers, (4) physical status, (5) psychological status, (6) daily routine, (7) coping and control, (8) medical treatment, (9) understanding & disclosure of condition.
Branch-Smith et al., 2018	Australia	Cystic Fibrosis	For qualitative part of study: *N* = 11 young people in grades 6–10 (4 male, 7 female). Total sample (quantitative and qualitative parts of study): *N* = 26 young people aged 10–16 years.	Sequential mix-methods approach to data collection; quantitative and qualitative	Online Survey and Focus Groups	Qualitative findings: Three major themes (with related subthemes) emerged: (1) School connectedness, (2) Being "sick" are school, (3) Missing school. This research sought to describe experiences of school bullying victimization and related social experiences of young students with CF and to examine associations with psychological well-being. Overall, the young people with CF in our study were healthy and happy and, for the most part, described being at school as a positive experience. They expressed high levels of peer support and connection to school overall. Most felt comfortable disclosing their diagnosis to close friends, and they generally did not feel lonely at school.
Hanson et al., 2018	Australia	Primary lymphedema	*N* = 20 aged 8–21 years	Qualitative study.	Semi-structure interviews	Six themes (with associated subthemes) were identified: (1) Reinforcing Abnormality – (a) damaging self-esteem, (b) frustrating restrictions and isolation, (c) self-consciousness (2) Vulnerability and Caution – (a) avoiding pain and discomfort, (b) preventing severe and permanent consequences(c) depending on permission

Table 3 (*continued*)

Author, year	Country	Rare disease/condition	Sample	Methodological approach	Focus	Key finding
						(3) Negotiating Uncertainties – (a) fearing condition worsening, (b) (feeling) deprioritized and sidelined, (c) questioning cause and permanence, (d) widespread unawareness (4) Developing Resilience – (a) focusing on the positives, (b) embracing individuality, (c) recalibrating normality, (d) prioritizing coping (5) Taking Responsibility – (a) individualizing treatment, (b) needing support, (c) external pressure and motivation, (d) seeking independence, (e) sticking to a routine (6) Disruptive Transition – (a) resisting change, (b) losing progress and support, (c) avoiding treatment costs.
Abma & Schrijver, 2019	The Netherlands	Primary School Children were targeted. (Health Conditions such as arthritis)	For Photovoice workshops: $N = 12$ (8 female, 4 male). A total of $N = 73$ children were involved in the entire study	Participatory Health Research	Multiple methods Multiple methods were used. Participant observation, Document analysis, focus groups, interviews, Photovoice	Allowing children to take photos and then talk about them led to narratives about their subjective experiences which otherwise may not have been discussed. Experiences brought up by participants included: living in a disadvantaged area, living with a chronic disease, and what was important to them. Children wanted to share their experiences with the facilitators. Child with arthritis could not join sports in school, playing outside was forbidden by her mother because she felt playgrounds were unsafe. She felt swinging into the air gave her a feeling of freedom. As a result of adults coming to see their photos, the children felt "'seen' and acknowledged as human beings."
Bogart, 2014	USA	Moebius Syndrome	$N = 10$ Aged 12-17 years (7 female, 3 male).	Qualitative study.	Focus groups	Five themes were revealed: (1) Social engagement/disengagement (2) Resilience/sensitivity (3) Social support/stigma (4) Being understood/misunderstood (5) Public awareness/unawareness These replicated the themes derived during the adult study, however three additional codes emerged in the adolescent study: positive outlook, social strain, and aggression.

constant staring and intrusion into their privacy by strangers". This was a continual reminder of their condition 'lymphoedema', which challenged their attempts at normalization. Children and adolescents would wear clothing to conceal the garments they had to wear for their lymphoedema. Many participants in the (Christie et al., 2011) study expressed that being different was exacerbated by symptoms that prohibited them from engaging in social and physical activities, resulting in social isolation. Due to CF, isolation experiences increased teenagers' perception of differences between themselves and those without CF (Christie et al., 2011). This study also described feeling different, as was a disrupted view of participants' 'normal' sense of self. This suggests that despite having a rare illness, children and adolescents develop a sense of self that is normal to them, and the only things that make them feel 'different' are when something outside of their daily routine happens or because of a difference between them and someone with no illness.

Currier and Zimmerman (2019) study also reflected a 'feeling different' theme for children and young people with kidney failure. They felt peers didn't want to talk to them because of their short height, and they felt different because they needed dialysis for kidney problems, and these children and young people were aware that they did not look (their) age. This could be supported by the finding in Bogart (2014), whereby participants with Moebius syndrome emphasized that their challenges with their condition were primarily due to others'

Fig. 2. Emergent themes across included studies.

perceptions of them, highlighting how *"disability is primarily socially constructed"* (p.1586). Stigma reduction is the key to reducing this and the negative feelings of those living with a rare condition.

Restrictions in independent living

Multiple hospitalisations, daily treatment regimes, physical restrictions and isolation related to Cystic fibrosis were reported in three studies (Branch-Smith et al., 2018; Vines et al., 2018). Such lifestyle restraints and maintaining social relationships were seen as challenges (Abma & Schrijver, 2019; Jamieson et al., 2014). Unlike their peers without a rare disease, children and young people with CF took time to integrate back into school after extended periods of absence (Branch-Smith et al., 2018). The most common challenges were social isolation, as they could not spend time with their friends because they were in the hospital, or their parents did not want them to see their friends for fear of infection. In their own words, narratives about the children's experience were able to be expressed well through the use of photovoice in the Abma and Schrijver *(2019)* study. This provided children with the opportunity to express their emotions or feelings. For example, a child with rheumatic arthritis could not join sports in school and often felt alone. Playing outside was forbidden by her mother because she thought playgrounds were unsafe. The daughter reported that swinging into the air gave her a sense of freedom.

Currier and Zimmerman (2019) conducted a retrospective review of approximately 500 publicly available creative writing entries by haemodialysis-dependent males and females ranging in age from 5 to 25 years. They reported perceptions of strict diet and fluid intake among children with kidney failure undergoing dialysis; wherein one of their main themes identified – 'physical shackle' – one child commented how dialysis for their kidney failure meant they couldn't play games such as 'capture the flag', soccer or run. Children often wrote about their favorite foods they could not have due to kidney failure and dialysis. The concept of water was frequently mentioned in their entries (for example, rain, ocean, waves, river, flooded, swimming, drowning, tide, bath etc.). These children live with chronic kidney failure and fluid restrictions, and they often complain of thirst and demonstrate a persistent preoccupation with water. They reported that adherence to their fluid restriction is an ongoing struggle in their day-to-day life.

In the Hanson et al. (2017) study, people with lymphoedema spoke about causing swelling of the limbs or other body parts and wearing compression garments daily. They reported the impact of wearing garments to manage their condition, leading to low self-esteem, and many young people took a risk by taking a break from wearing their garments to fit in with their peers. Most participants refused to be considered disabled, insisting that they were as capable as their peers, and they were satisfied when they found suitable and enjoyable sports or activities. They felt frustrated by their diet and lifestyle restrictions and reduced sporting abilities.

Developing resilience/coping strategies

Children and young people described developing resilience and acceptance of their condition in all 6 out of 8 studies (Bogart, 2014; Christie et al., 2011; Currier & Zimmerman, 2019; Hanson et al., 2017; Spencer- Tansley, 2018; Vines et al., 2018). Children with a rare disease struggle with low self-esteem and fear and uncertainty about their condition (Hanson et al., 2017; Vines et al., 2018), but most studies highlighted the resilience of children and young people despite their challenging circumstances. In their research, Hanson et al. (2017) reported that adolescents focused on some of the positives about their condition, lymphoedema, as they compared it to more severe forms or terminal illnesses. This mindset helped motivate them to accept their body and wear their garments. (Bogart, 2014) study aggression and positivity were discussed in the context of being bullied. Despite all the challenges, several participants in the study reported having friends who would stand up to bullies; unfortunately, not all participants had friends like these. They would prefer the attitude of positivity, and they have been using this as a coping strategy in response to these challenges. The children with kidney failure undergoing dialysis treatments (Currier & Zimmerman, 2019, pp. 297) utilised creative writings to demonstrate their remarkable resilience with statements that generally focused on positive aspects of their life. For example, *"This is me, and this is my life. I will never let anyone, anything, any disease, any rejection, get the best of me. I am a survivor and a fighter"*.

The psychological and emotional impact

Feeling mental and emotional distress was reported as one of the central themes from (Currier et al., 2019) study, and Currier & Zimmerman (2019) study focused on paediatric patients on haemodialysis. All adolescents in Vines et al. (2018) study, aged 12 to 19 years, diagnosed with cystic fibrosis experienced a lack of available peer-to-peer support opportunities and experienced isolation as part of their routine treatment. They reported psychological difficulty resulting from adjusting to living in isolation. The lack of available peer-to-peer support opportunities and the risks of avoiding the realities of isolation may impede coping mechanisms in the context of CF in a way that is different from other conditions. Children in the Christie et al. (2011) study spoke about feeling isolated, anxious or worried and not wanting to be ill or different from others. The fear of one's condition worsening was an experience identified in the Hanson et al. (2017) study.

Branch-Smith et al. (2018) study examined associations between school bullying and the psychological well-being of young people with CF. They reported that bullying victimization and perpetration involvement for these young people was highly correlated with depression, anxiety, and loneliness at school. Hanson et al. (2017) reported that living with lymphoedema can profoundly impact children and adolescents' psychosocial well-being. They said that young people with lymphoedema felt frustrated by restrictions on their diet and physical activity, and they reported low self-esteem, fear and uncertainties about their condition's trajectory. CF is a rare life-limiting illness. It has multiple comorbidities and treatment burdens that impact their

identity, daily functioning, and life goals. Rare Disease UK project (Spencer- Tansley, 2018) reported that young people with a rare disease have different needs and challenges than other children. This report highlighted healthcare professionals' lack of understanding of the psychological and emotional impact on children and young people living with rare diseases and often unable to provide appropriate psychosocial support due to a lack of resources and fundings

Social impact vs social connectednessfunding

The difficulties involved in engaging in everyday activities and sport, social interaction with strangers and the effect that the disease and/or frequent hospitalizations have on relationships with friends and the feeling of school connectedness was highlighted in a number of studies (Branch-Smith et al., 2018; Christie et al., 2011; Hanson et al., 2017) and a research report (Spencer- Tansley, 2018). In the Bogart (2014) study, participants experienced social strain when parents and medical professionals offered unwanted help or treatment. Social support or social connectedness reports involved trusting and feeling valued by friends, family, and professionals. Participants felt that spreading public awareness can reduce social stigma, as their challenges with Moebius syndrome were primarily due to others' perceptions. However, one of the participants in this study was concerned that greater awareness would bring stereotyping and labelling. They emphasized that the Moebius Syndrome conferences were valuable for meeting new people and sharing each other's experiences.

Transition challenges

Wanting to take responsibility for one's care or to be more involved in their care and their transition from paediatric to adult healthcare services were all highlighted as ongoing challenges (Hanson et al., 2017; Spencer- Tansley, 2018), as was the social strain between them and parents or healthcare professionals that can arise from wanting to be more independent (Bogart, 2014; Spencer- Tansley, 2018). The disruptive transition between child and adult services was one of the core themes identified by the Hanson et al. (2017) study. Adolescents in this study were worried about moving to an adult hospital because they were concerned about being "*treated as an* adult" (Hanson et al., 2017, pp.678). They expressed concern about adult healthcare professionals being less aware of the challenges and priorities of patients with childhood-onset lymphoedema, compared to secondary lymphoedema, following cancer treatment in adult settings. They wanted simplified information and updates on ongoing research, peer support, more comfortable garments, specialised, flexible, and supportive care after transitioning to adult services. Treatment adherence was a problem due to the constant emotional demand of having a rare illness (Hanson et al., 2017), and the lack of explanation about certain aspects of treatment, such as isolation, was identified in (Vines et al., 2018) study. However, suggestions for improving this were described, such as enhancing shared decision-making processes. They suggested the need to individualize the transition approach, focusing on the individual's readiness and expectations for transition, facilitated by creating a care plan with the patient and their family.

In the Spencer- Tansley (2018) study, the young people in the UK felt they were in limbo as they felt too old for children's services but too young for adult services. These children and young people also commented on the lack of support at school. They also reported their teachers also lack understanding about their rare conditions and/or their individual needs. Adolescents living with lymphoedema reported feeling deprioritised and sidelined (Hanson et al., 2017), who thought they were not told about new research findings because of their age. However, (Hanson et al., 2017) study emphasize that transitioning to adult services means preparing them with simplified information and updates about ongoing research, peer support, specialised, flexible and supportive care after transitioning to adult services.

Discussion

The prevalence of children and young people living with rare diseases is increasing worldwide due to advancements in technology and medicine, enabling more accurate diagnostics. Approximately 70% of RDs affect children, and few are preventable or curable. Most of these are chronic, with 35% being life-limiting or life-threatening, often resulting in early death, and many causing intellectual and physical disabilities that place substantial demands requiring multidisciplinary input and regular hospitalizations (Elliott & Zurynski, 2015; Somanadhan & Larkin, 2016). Early detection, diagnosis, and intervention can prevent death or disability for children, but lack of health intelligence is a barrier to this, as most individual RDs receive limited attention due to their rarity (Slade et al., 2018). Often, patients are required to become experts in their own diseases due to a lack of specialist knowledge (Eurordis, 2017).

Despite the variability in the studies' quality in this review, there were similarities across findings illustrating the impact of rare diseases on children's day-to-day lives. There were interconnections and overlaps in the data across these nine themes. Research in rare diseases is developing, but there are many with little or no data available, particularly regarding the impact on the child and family. This review is consistent with evidence highlighting living with a rare disease is an ongoing learning experience for affected children, young people, and their families and is often associated with psychosocial impacts such as experiences of stigmatization, self-consciousness, restrictions in independent living, challenges in developing resilience and coping strategies, psychological and emotional impact, social impact versus social connectedness, and their transition experience (Abma and Schrijver *(2019)*; Bogart (2014); Branch-Smith et al. (2018); Christie et al., 2011; Currier and Zimmerman (2019); Hanson et al. (2017); Spencer-Tansley (2018); Vines et al., 2018.

Stigma is a universal phenomenon (Saylor et al., 2008) and has many facets, ranging from personal characteristics that deviate from societal norms to social stereotyping or categorization (Goffman, 1963). Reportedly, stigmatization attached to many illnesses and impairments, and evidence from the review is consistent with the literature (O'Donnell and Habenicht, 2022; Williams and Chapman, 2011) suggests that stigmatization can have devastating effects on the mental health and well-being of children and young people living with rare diseases (Bogart, 2014; Branch-Smith et al., 2018; Hanson et al., 2017). Abma and Schrijver *(2019)* urge researchers to offer children the autonomy to identify and express an important issue and offer them a 'playful' mindset and methods fitting children's needs. As a pragmatic approach, researchers and practitioners could utilize models of participation, such as the Lundy (2007) model of participation, which could be used as a guide to support children's involvement in research to be more effective, meaningful, and compliant with the participation of their rights. The analytical tool of Shier (2019) could be another example that could be utilised to aid the study in developing partnerships with children.

This review suggests that despite having a rare illness, children and young people developed a sense of self that is a new normal to them. Many participants in the (Christie et al., 2011) study expressed that being different was exacerbated by symptoms, while sometimes they felt like other 'normal' children, there were times when their differences from other children were highlighted and the only things that make them feel 'different' are when something outside of their daily routine happens or because of a difference between them and someone with no illness. Finding a new normal following this journey of living with rare diseases was identified in recent studies (Germeni et al. (2018); Belzer et al. (2022).

As specialist expertise is scarce, children, young people and their families may find it challenging to gain access to services to address day-to-day psychosocial caring needs, transition, self-management, and integrated care (care closer to home). This review indicated

children and young people with diseases experience negative social and psychosocial impacts which can lead to poor emotional and mental health well-being, low self-esteem, stigmatization, and feeling different compared to the normal population whether they have a physical disability or not (Adama et al., 2021; Currier & Zimmerman, 2019; Vines et al., 2018; Branch-Smith et al., 2018; Hanson et al., 2017; Bogart, 2014; Christie et al., 2011). Inconsistent with recent literature (Belzer et al., 2022) the review also highlighted children and young people feel isolated and may feel stigmas in settings of education with a lack of social support or understanding from their peers (Vines et al. (2018) and children and young people reported social isolation and feelings of difference from their peers (Christie et al. (2011), leading to lower self-esteem and resilience, which can result in poorer outcomes in adulthood (Anderson et al. 2013; Slade et al., 2018) This review findings emphasize that children and young people living with the rare disease also face discrimination at school, community, and in leisure(Christie et al., 2011; Branch-Smith et al. (2018). Very little is known about the experience of living with a rare disease with a focus on equality, diversity, and inclusion (EDI) as a method to promote transition, self-management, and social connectedness.

This review highlighted that the transition from paediatric to adult care is often an acute concern to the young people themselves. The majority of rare diseases are chronic or medically complex conditions. Specialty input is required from a specific discipline at the local/regional level by a clinician in the paediatric services with particular skills and experience to a clinician and team in the adult services. They will need to collaborate with the other members of the groups and prepare them with simplified information and care pathways. The transition plan should be collaborative, person-centred and holistic, focusing more on life transition than physiological and medical needs (Somanadhan et al., 2021). This involves young people taking responsibility, caring, advocating for service navigation and coordination, information gathering, and contributing to their disease management plan (Hanson et al., 2017; Kerr et al., 2018; Kerr et al., 2019; Spencer-Tansley, 2018).

Most of the literature on children's experience living with a rare disease is written by parents, caregivers, and health care professionals. There is very little understanding of the children's perspective dealing with its impact on their daily lives (Currier & Zimmerman, 2019; Spencer- Tansley, 2018). There was no clear definition of a child and young people within the studies cited in this review. The range of ages covered in this review was 3–25 years. Review findings highlighted that social isolation and feelings of difference from their peers, lead to lower self-esteem and resilience, which can result in poorer outcomes. Age definitions of 'child' and 'young people' differed across the included studies. The most widely accepted definition of 'child' was declared in 1989 by the United Nations Convention on the Rights of the Child (UNCRC), as 'every human below the age of eighteen years unless under the law applicable to the child, the majority is achieved earlier' (UNCRC, 1989). The UN Committee on the Rights of the Child (2003, para. 14c) reminds us that adults' responsibility is to create opportunities for young children to express their views, rather than expecting children to prove their capabilities. One of the four general principles of (Article 12) is that the "child's views must be considered and taken into account in all matters affecting him or her".

Four studies (Abma & Schrijver, 2019; Christie et al., 2011; Currier & Zimmerman, 2019; Spencer- Tansley, 2018) offered a creative arts-based approach to include children's voices to explore how children's voices can be genuinely taken into account to understand their lived experience. Most studies provided little or no information about obtaining informed assent/consent to preserve the child and adolescent participants' rights. Despite the growing recognition of children's rights, particularly about their involvement in decision-making and research that aims to learn about their experiences (Kirk, 2007), none of the reviewed studies adequately explained how they obtained informed assent/consent to preserve the rights of the child participants. With children's limited vocabulary and cognitive capacity to express their thoughts, emotions, and feelings, researchers and practitioners working with children need to acquire a mode of inquiry to enhance their communications to voice their needs and rights. A children's rights-informed approach to researching with children will offer safe, inclusive and engaging opportunities to express their views and deliberate strategies to help children form their opinions (Lundy & McEvoy, 2011).

Applying creative, arts-based participatory methods could offer children great potential to express themselves more easily through articulation (Lee et al., 2020). The resulting insights could inform a child-centred quality care plan for children with rare diseases. In addition to general quality of life (QoL) issues, such as physical, emotional, social, family and functional well-being, there are unique concerns among children with a rare progressive illness. The fear of one's conditioning worsening was an experience identified in this review; again, not all the conditions in this review fall under the category of rare life-limiting illness.

Implication for practice

The experience of living as a child or young person with a rare disease is inherently unique. There was also a difference like experiences between children aged below 10 and over 11 of years of age (Branch-Smith et al., 2018; Spencer- Tansley, 2018). Therefore, it is critical to acknowledge this and apply a strategy that could work for children and young people according to their developmental age. Children and young people living with rare diseases also appeared to have many commonalities in their day-to-day lived experience and the challenges they face, which was reflected in themes between studies. It has been highlighted from the review that living with a rare disease is an ongoing learning experience for affected children, young people, and their families. Despite living with day to day challenges of a rare disease, children and young people develop a sense of self that is a new normal. They may experience negative social and psychosocial impacts, leading to lower self-esteem and resilience. For these children and young people, equity means social opportunity, non-discrimination in education and work, and equitable access to health, social care, diagnosis, and treatment (Eurordis, 2022). This requires tackling the stigma, discrimination, and social marginalization faced by this population.These common themes have important implications for future research and clinical practice because policies and strategies developed for one type of rare disease could also apply to children and young people with other types of rare diseases. Table 4 outlines the implications for practice based on these common themes for this review.

Limitations

This review is subject to limitations within the available evidence base. The literature search was complex because many studies were identified using specific rare diseases as search terms in the electronic databases. As mentioned in the background sessions, there are approximately 8000 rare conditions; however, in this review, the children's experience was focused on some of these rare conditions; again, some studies involved children with the syndrome with no name (SWAN). It is possible that in some cases, these chronic illnesses may not have met the criteria for rare diseases. Three of the nine studies included are about the genetic disease cystic fibrosis (CF).

On the one hand, this gave a more consistent and solid insight into the experiences of children and young people living with it, but the potential patterns and findings from the review could be biased toward those living with CF. This review did not utilize a software platform like COVIDENCE for the multiphase review process, including data extraction. The selected review studies were limited to a few available databases, and only papers in English were analyzed dated between 2005 and 2019, which means there could be other relevant papers.

Table 4
Themes and corresponding implications for practice.

Theme	Implications for practice
Experiences of stigmatization	Promoting a child-friendly approach and involving children with rare diseases to lead and drive social and practice change.
Self-consciousness	Promoting public awareness campaigns for rare diseases to improve visibility, inclusivity, empowerment and positivity.
Developing resilience/coping strategies	Using creative methods to reflect children's lived experiences. Teaching interventions to empower the whole family by learning new strategies and interacting with each other.
Restrictions in independent living	Promote age-appropriate treatment plans to address children's and young people's needs, thereby improving their self-management and independent living. Involve children and young people in the decision-making process to co-create treatment and care needs either in the hospital or in the school or community settings.
Psychological and emotional impact	Co-create an educative component for peers of young people with rare diseases addressing ways to support each other in the hospital or the school or community settings. Addressing the knowledge gap in the primary care and school setting by ensuring information about rare diseases is readily available to them. Co-create interventions to promote mental health and school engagement among young people living with rare diseases.
Social Impact versus social connectedness	Encouraging social and academic networking opportunities within schools to mitigate the risk of social isolation. Using technology to stay connected with their friends and peers during their absence. Promote peer-to-peer health care, where patients and their families can connect and share practical health care tips.
Transitioning remaining a challenge	Co- design interventions through assessment and education to help chiildren and young people with rare disease to transition from the children's services to adults services and also from primary to high school. Ensuring there is a strategy in place to empower young patients to advocate for themselves during their transition to adult care (Hanson et al., 2018). Ensuring individualize the transition approach, focusing on the individual's readiness and expectations for transition can be facilitated by creating a care plan with the patient and their family. (Jamiessson et al. 2019) To appoint a transition coordinator to liaise between the paediatric and adult teams and provide education to families.

Conclusions

This integrative literature review has revealed limited research on children's experiences of living with rare diseases and how best they can be supported to express their experiences of living with a rare condition. The experience of certain aspects of having a rare illness differed across different age groups. Children (typically aged 3–10) with rare diseases generally view themselves and their lives in the same way as their healthy peers. They were more likely to report being adaptive and resilient than those aged 12 or older. Young people reported being different compared to young children, and they faced numerous challenges related to their illness. Considering the progressive nature of rare diseases such as CF and inclusion of an educative component for friends of young people with rare diseases addressing ways to support each other at school was recommended. This review confirms concerns that children with rare diseases experience stigma. This can have devastating effects on the mental health and wellbeing of children and young people living with rare diseases, so there is a pressing need to co-create interventions to promote mental health among children and young people living with rare diseases. To provide the best possible level of care for children and families with rare disorders, health services must be informed and equipped to provide the necessary supports specific to the unique needs of children and young people living with rare diseases.

Funding sources

This study is a part of the SAMPI research project, which received funding from the Children's Fund for Health at Children's Health Ireland at Temple Street (RPAC17–05).

Consent for publication

Not applicable.

CRediT authorship contribution statement

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Conflict of interest

None.

Acknowledgments

We would like to express our sincere gratitude to our funder Children's Fund for Health at Children's Health Ireland at Temple Street for the support.

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