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The relationship between stigma and quality of life of children with rare diseases in Romania

Shir Grunebaum[a*], Noel Parnis[b] and Liana Nagy[a**]

[a]School of Occupational Therapy, Oxford Brookes University, Oxford, UK; [b]Academic Skills Development Department, Brunel University of London, Uxbridge, England

ABSTRACT

The current study examines the impact of parental perceived stigma on the quality of life (QoL) of children with rare diseases in Romania. As rare diseases habitually affect a small portion of the population, they are frequently under-researched, underfunded and patients are often unsupported. Additionally, people with rare diseases often experience high levels of stigma. The present study uses the Paediatric Quality of Life Measure (PedsQL) and the Parental Perceptions of Public Attitudes Scale (PPPAS) to ascertain the parent/primary caregiver reported QoL in children with rare diseases (CwRD) and explore the relationship between stigma and QoL. Using two validated measures, this study identified that stigma has a statistically significant negative impact on QoL whilst controlling for the age, gender, education and employment status of the parent. Every SD increase of stigma ($M = 3.35$, $SD = 1.12$) results in a decrease of 0.268 SD of QoL ($M = 36.04$, $SD =$ 18,13). Over 75% of responders reported physical QoL and low psychosocial health. Since parent/primary caregivers who experience higher levels of stigma are likely to report a lower (QoL) for their CwRD, coproduced interventions to address stigma should be considered by future research to support the physical and psychosocial QoL of CwRD in Romania.

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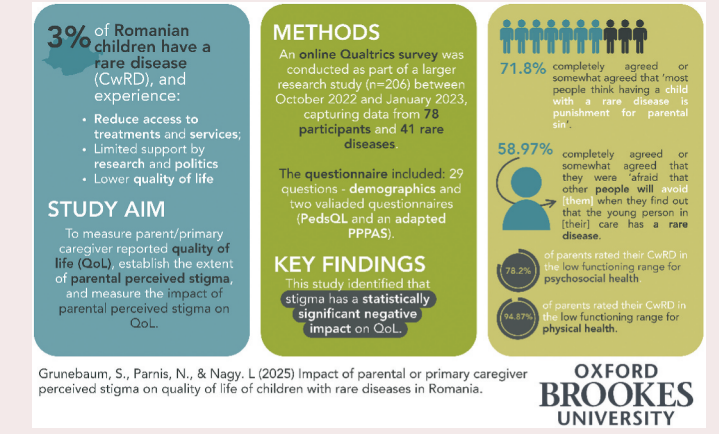
CONTACT Shir Grunebaum shir.grunebaum@nhs.net School of Occupational Therapy, Oxford Brookes University, Jack Straw's Ln, Marston, Oxford OX3 0FL , UK

*Shir Grunebaum is now affiliated with North Middlesex University Hospital, Royal Free 495 NHS Foundation Trust.
**Liana Nagy is now affiliated with Brunel University and Oxford University Hospitals NHS Foundation Trust.



Introduction

A rare disease is defined in the European Union as a condition that impacts less than one in 2000 people (European Commission, 2022). As of 2010, rare diseases affect approximately 1 million people in Romania, and nearly 3% of Romanian children have a rare disease (Puiu & Dan, 2010). However, Romania is one of the smallest European markets for rare disease treatments (Detiček et al., 2018). Therefore, minimal research is presently available on rare disease patients in this country, and more recent population predictions are not available.

Whilst rare diseases are largely heterogeneous in presentation and origin, the burden that these have on families and children has been identified, including vast physical, social, education and emotional difficulties (Wiegand-Grefe et al., 2022). Importantly, many of these rare diseases are accompanied by emotional, cognitive and physical impairments (von der Lippe et al., 2017). Currently, those with rare diseases face significant diagnosis delays (Boulanger et al., 2020) and cannot access treatments due to their inadequate availability, accessibility, affordability and acceptability (Douglas et al., 2022), thereby causing significant family difficulties (Chu et al., 2022; Zurynski et al., 2017).

Within daily life, those with diseases may experience structural, interpersonal and perceived stigma (Munro et al., 2022). Structural stigma refers to the systemic barriers and discriminatory policies that limit access to care and resources for individuals with diseases (Major et al., 2018). These institutional obstacles often perpetuate health disparities and hinder effective treatment (Major et al., 2018). Interpersonal stigma manifests through direct negative attitudes and behaviours encountered in social interactions with those affected (Major et al., 2018). Perceived stigma reflects the anticipation or fear of judgment and discrimination, which can profoundly impact the well-being of individuals living with illness (Major et al., 2018). This includes disbelief and invalidation from healthcare practitioners and community members (Bogart et al., 2022). Further, rare disease patients experience a lack of understanding and educational support from healthcare practitioners, increased isolation, avoidance, feelings of personal blame and

limited opportunities to participate in social roles (Bogart & Irvin, 2017). Additionally, people with rare diseases often encounter a deficient understanding from others and insufficient social support (Bogart et al., 2022). Some research has indicated that children with rare diseases may experience increased absenteeism from school, an inability to participate in physical education and academic activities (Verger et al., 2020), and increased bullying (Adama et al., 2020). This, thereby, heightens the child's and family's experiences of stigma (Adama et al., 2020), which is further associated with poorer QoL (Pierpont et al., 2021).

Emerging research suggests that parents' perception of stigma, particularly affiliate stigma, may significantly influence their children's quality of life (QoL), especially in families navigating disabilities (Sakiz & Kaçan, 2023). Affiliate stigma refers to the internalisation of public stigma by caregivers who are closely associated with someone who is stigmatised, often resulting in guilt, shame and social isolation (Hu et al., 2023). This internalised stigma can negatively affect caregiver mental health and parenting behaviours, contributing to a less supportive family environment and limiting the child's opportunities for emotional and social development (Rusu et al., 2024)). Moreover, stigma experienced by parents can restrict their social participation and roles within the community, which may reduce the family's overall social inclusion (Rusu et al., 2024). This diminished social engagement can contribute to the child's own experiences of exclusion and stigma, reinforcing a cycle of isolation for both parent and child (Rusu et al., 2024).

Several studies have elucidated mechanisms through which affiliate stigma affects children's QoL. Rusu et al. (2024) found that higher levels of parental stigma were significantly associated with increased caregiver stress and reduced family functioning, both of which mediated lower psychosocial QoL outcomes in children. A study by Mitter et al. (2019) further demonstrated that stigma-related distress in parents can lead to more critical or withdrawn parenting behaviours, impacting children's development and coping skills. The QoL and the impact of stigma on patients with rare diseases have been studied in some populations, including the United States (Bogart & Irvin, 2017) and China (Chen et al., 2022). However, current data does not exist on the relationship between these two factors in most other parts of the world, including Romania. Further, direct empirical evidence connecting parental stigma and children's QoL remains limited, particularly in CwRD. It is plausible that children with poorer QoL experience higher levels of stigma themselves, which in turn can exacerbate parental feelings of stigma. This bidirectional relationship highlights the complexity of stigma within families affected by rare diseases.

Uniquely, Romania was under communist rule from 1948 to 1989, with a recent liberalisation of socioeconomic structures in 1991 (Birau et al., 2019). Some researchers claim that the previously instilled communist values on disability and health remain embedded within Romanian culture and its institutional practices (Baciu & Lazar, 2017), thereby heightening experiences of stigma.

Therefore, this study aims to: (1) measure the parent/primary caregiver reported quality of life of children with rare diseases (CwRD) in Romania, (2) establish the extent of perceived stigma faced by parent/primary caregiver of CwRD in Romania and (3) test the hypothesis that stigma has an adverse impact on Qol whilst controlling for age, sex, education and employment of the parent or primary caregiver.

Drawing on prior research from other countries demonstrating the adverse impact of stigma on QoL among individuals with chronic and rare illnesses, this study hypothesised that parents/caregivers of CwRD in Romania would report low QoL. Furthermore, it was expected that these parents/caregivers would experience substantial perceived stigma associated with their child's condition. Finally, it was expected that higher levels of perceived stigma would be inversely correlated with caregiver-reported QoL, indicating that greater stigma is associated with poorer perceived quality of life in this population.

Materials and methods

Survey design and participants

A cross-sectional online survey of parent/primary caregivers of CwRD was conducted as part of a more extensive study. This study forms part of a broader research initiative examining the QoL and perceived stigma experienced by parents and primary caregivers of children with disabilities and rare diseases in Romania. Led by a team of researchers, the larger project, yet to be published, aims to illuminate the social and psychological challenges faced by these families. While the overarching study included data from caregivers of children with a range of disabilities, the present analysis focuses specifically on the experiences of those caring for CwRD, offering a more nuanced understanding of this often-overlooked population. The survey was conducted between October 2022 and January 2023 and disseminated through online social media platforms, including Facebook and Twitter/X. In addition, patient organisations for rare diseases in Romania were contacted directly to distribute the survey to community members.

The inclusion criteria mandated that participants be 18 years of age or older. Participants were also required to be the parents/primary caregivers of a child who is between 2 and 16 years old, due to previous questionnaire verification, and the child had to have been diagnosed with a rare disease. The study employed the EURORDIS criteria to define and verify rare disease classifications, ensuring consistency with internationally accepted standards (Eurordis, 2025). A single coder conducted a systematic review of all reported conditions to confirm their alignment with these criteria. Importantly, no participants were excluded from the sample on the basis of their reported diagnosis, as all conditions met the established threshold for inclusion as a rare disease.

Measures

QoL instrument

QoL was assessed using the Paediatric QoL Inventory (PedsQL). Research has indicated that this measure is valuable for determining responses of those with long-term health conditions (Verstraete & Scott, 2022), such as rare diseases. Evidence has also specified that patients with rare diseases find the PedsQL relevant to their lived experience (Lee Aiyegbusi et al., 2020).

The PedsQL questionnaire has previously been used to determine functioning levels in the literature by dividing participants into three categories: low, intermediate and high functioning (Beverung et al., 2015). The low functioning level includes participants who

answered 'never' or 'almost never' on one-third of the items or less for each subscale (Beverung et al., 2015). The intermediate functioning levels included participants who answered 'never' or 'almost never' for more than one-third but less than two-thirds of the items on the subscale (Beverung et al., 2015). The high functioning level included participants who answered 'never' or 'almost never' on the Likert scale for more than two-thirds of the items on the subscale (Beverung et al., 2015). As such, these classifications were used in analysing the participant data.

The PedsQL consisted of 23 items rated on a 5-point Likert scale, where higher values represent better quality of life. Internal consistency for this scale in the current sample was excellent ($\alpha = 0.90$)

Experiences of stigma instrument

Parent/primary caregivers experiences of stigma were assessed using the Parental Perceptions of Public Attitudes Scale (PPPAS) (Čolic & Milačić-Vidojević, 2021). This scale was developed to examine the perceived stigma among parent/primary caregivers of children with autism spectrum disorder. Importantly, this scale was developed in Serbia and therefore was identified as a culturally appropriate tool for assessing this population. While this tool has not yet been validated in other research studies, its culturally relevant nature made it valuable for assessing parent/primary caregiver perceived stigma in the Romanian context.

As per the validation study, this questionnaire was also analysed as two separate components, 'child characteristics and causes of the disorder' as well as 'parental blame'. As per the PPPAS, the scores were analysed in three categories: low (1.1–2.5), moderate (2.51–3.5) and high (3.51–5.0) degrees of perceived stigma. Minimal changes were made to the questionnaire to ensure its appropriateness for the population. These revisions were made following discussion and agreement with the original authors and included changing the questions to include 'children with disabilities or rare disease' rather than 'ASD'. Two items that were not conceptually aligned with the experiences of CwRD were excluded from the questionnaire. The first, which assessed a child's ability to speak, was removed due to its stronger relevance to neurodevelopmental conditions such as autism, rather than the diverse clinical profiles typically associated with rare diseases. It was also determined that the item concerning inappropriate behaviour was not suitable for this population, as children with rare diseases are not frequently reported to exhibit challenging behaviours (Adama et al., 2021) compared to other clinical groups.

The PPPAS consisted of eight items rated on a 5-point Likert scale, where higher scores represented higher rates of perceived stigma. Internal consistency for the scale in the current sample was excellent (Cronbach's $\alpha = 0.93$).

Procedure

Ethical approval for this study was obtained from the Oxford Brookes University Research Ethics Committee (HLS/2022/VI/73). A Romanian translation of the PedsQL was available for use. However, the other questionnaire components were translated by researcher LN, a native Romanian clinician and academic in

the UK. The complete survey was piloted and reviewed by two native Romanian academics with research and clinical experience with childhood disability in Romania and the UK. All data was anonymously collected using Qualtrics (Provo, UT).

Data analysis

All statistical analyses were performed using SPSS Statistics 25.0 (BM SPSS Statistics). p values of less than 0.05 were considered to be statistically significant. Descriptive statistics including mean, SD and range were selected to address the first two aims [(1) measure the parent/primary caregiver reported quality of life of children with rare diseases (CwRD) in Romania, 2) establish the extent of perceived stigma faced by parent/primary caregiver of CwRD in Romania] the hierarchical linear regression adopting an incremental approach was selected in

Table 1. Participant demographics.

Category	Sub-Category	Frequency (N)	Percent (%)
Relationship to child	Parent	69	88.5
	Step-Parent	5	6.4
	Grandparent	2	2.6
	Older Brother/Sister	1	1.3
	Aunt/Uncle	1	1.3
	Legal Guardian (Not Related)	0	0
Age (Years)	18–25 young	2	2.6
	26–35 young	21	29.5
	36–45 middle age	34	43.6
	46–55 mature	12	15.4
	56–65 mature	7	9
	>65 mature	2	2.6
Sex	Female	60	76.9
	Male	18	23.1
	Prefer Not to Say	0	0
Education Level	Primary School (Level 1)	1	1.3
	Secondary School (Level 1)	13	16.7
	High School Level 2	20	25.6
	Vocational School Level 2	6	7.7
	Undergraduate Degree Level 3	26	33.3
	Masters Degree Level 4	11	14.4
	Phd/Doctorate Level 4	1	1.3
Employment Status	Full Time Employment (> 20 Hrs/Week)	40	51.3
	Part Time Employment (≤20 Hrs/Week)	16	20.5
	Unemployed/Not Working	22	28.2
Participant Ethnicity	Romanian	69	88.5
	German	3	3.8
	Hungarian	3	3.8
	Polish	1	1.3
	Ukrainian	2	2.6
Child/Young Person's School Type	Mainstream Education	31	39.7
	Does not Attend Education	21	26.9
	Specialist Education	17	21.8
	Home Education	9	11.25
Child/Young Person's Age (Years)	2–4	20	25.6
	5–7	27	34.6
	8–12	22	28.2
	13–16	9	11.5

order to address the third aim of the study [(3) test the hypothesis that stigma has an adverse impact on Qol whilst controlling for age, sex, education and employment of the parent].

Results

Sample characteristics

Eighty-seven parents/primary caregivers of CwRD were enrolled in the study, and 78 completed the entire questionnaire.

Table 1 reports the demographic characteristics of the parents/caregiver participants who have CwRD. Notably, 88.46% ($n = 69$) of participants who completed the questionnaire were parents of CwRD. Furthermore, 90% ($n = 70$) identified as Romanian. The remaining were German, Hungarian, Polish or Ukrainian. Moreover, 77% ($n = 60$) were female, whilst 23% ($n = 18$) were male. Further, the data captures the experience of parent/primary caregivers of children with 41 different rare diseases in Romania, with the top conditions being Fabry Disease which affected 14% ($n = 11$) of CwRD in this study and Duchenne Muscular Dystrophy, affecting 11.53% ($n = 9$).

With a sample of 78 respondents from a population of approximately 110 762 children with rare diseases in Romania and using a margin of error of ±5%, the achieved confidence level is approximately 70%. While this is below the conventional 95% threshold, it reflects the practical and ethical challenges of engaging this population. The estimated population of children with rare diseases was derived from United Nations data indicating the total number of children under 18 in Romania (United Nations, 2023), combined with an estimated prevalence rate of 3% for rare diseases among Romanian children (Puiu & Dan, 2010). A post hoc power analysis indicated that with a sample of 78 participants, the study was powered at approximately 70% to detect medium-to-large effects (Cohen's $d \approx 0.5–0.6$) at an alpha level of 0.05. This effect size threshold was selected based on the expectation that differences in outcomes between CwRD and general populations would be clinically meaningful rather than subtle. The research was conducted over a limited four-month period, from October 2022 to January 2023, which constrained recruitment efforts. Additionally, families of children with rare diseases often face considerable barriers to participation, including high caregiving demands, medical fragility and geographic dispersion (Černe et al., 2024). Despite the modest sample size, the data collected provide meaningful preliminary insights and a foundation for future research in this underrepresented population.

Table 2. PedsQL scores.

Functioning Type	Mean	SD
Physical Functioning	38.18	27.45
Emotional Functioning	42.60	29.66
Social Functioning	34.93	22.78
School Functioning	33.17	22.78
Psychosocial Health	37.66	18.94
Overall QoL	36.04	18.12

Table 3. PPPAS levels of stigma.

Perceived Stigma Score ($M = 3.34$, SD = 1.11)	Frequency (N)	Percent (%)
High (3.51–5)	44	56.61
Medium (2.51–3.5)	16	20.51
Low (1.1–2.5)	18	23.08

Descriptive statistics

The questionnaire consisted of three parts, a demographic component, the PedsQL questionnaire and an adapted PPPAS.

The PedsQL was scored following the standardised method outlined by Varni et al. (1999), which involves reverse-scoring and linearly transforming item responses to a 0–100 scale. Scores are then averaged to yield three primary scores: the Physical Health Summary Score (from eight physical functioning items), the Psychosocial Health Summary Score (from 15 items covering emotional, social and school functioning) and the Total Scale Score (mean of all 23 items), see Table 2.

In the current study, the mean for the overall QoL is 36.04 (SD 18.13) ranging from 9 to 90 in a scale of 0–100. The mean score for the Physical Health Summary is 41.6 (SD = 20.66) whist the mean score for the Psychosocial Health Summary is 34.92 (SD = 17.18). Overall, 78.2% of parents ($n = 61$) rated their CwRD in the low functioning range (defined as ≤69.7) on the Physical Health Summary Score, and 94.87% ($n = 74$) did so for the Psychosocial Health Summary Score. Several individual items within the PedsQL revealed a high frequency of difficulties experienced by children with rare diseases, as reported by their parents. A notable proportion of participants indicated that their child experienced these issues 'often' or 'almost always'. Specifically, 65.4% ($n = 51$) reported problems with running, and 66.7% ($n = 52$) noted difficulties participating in sport or exercise. Additionally, 62.8% ($n = 39$) reported low energy levels, and 61.25% ($n = 49$) indicated trouble sleeping. Concerns about participation and social comparison were also prominent, with 83.3% ($n = 65$) reporting difficulties doing things that others their age can do, and 71.8% ($n = 58$) struggling to keep up with peers. Absences were also common, with 78% ($n = 49/59$ in education) indicating that their child missed school or events due to not feeling well, and 74.6% ($n = 44/59$ in education) due to medical appointments or hospital visits.

With respect to the PPPAS questionnaire, it is notable that the highest percentage to a question was 71.8% ($n = 56$), in which the participants indicated that they completely agreed or somewhat agreed that 'most people think having a child with a rare disease is punishment for parental sin'. In addition, 58.97% ($n = 46$) of the participants designated that they completely agreed or somewhat agreed that they were 'afraid that other people will avoid [them] when they find out that the young person in [their] care has a rare disease.

In the current study, 56.41% ($n = 44$) of participants scored as experiencing a high degree of perceived stigma, 20.51% ($n = 16$) recorded a moderate degree of perceived stigma and 23.08% ($n = 18$) attained a low degree of perceived stigma score (Table 3).

Inferential statistics

To test the hypothesis that perceived stigma negatively affects quality of life (QoL) in children with rare diseases (independent of parental age, sex, education and employment status), a hierarchical linear regression analysis was conducted. QoL was entered as the dependent variable, with stigma as the primary independent predictor. Control variables (age, sex, education and employment) were sequentially added in subsequent steps to examine their impact on the relationship.

Assumptions for linear regression, including linearity, independence of errors, homoscedasticity, normality of residuals and absence of multicollinearity, were checked and found to be satisfactorily met. In Step 1, stigma alone significantly predicted QoL, $F(1, 76) = 14.653$, $p < .001$, explaining 15.1% of the variance (adjusted $R^2 = .151$). Higher levels of perceived stigma were associated with lower QoL scores ($\beta = -6.516$, $p < .001$).

In Step 2, employment status was added as a categorical control variable. The model remained significant, $F(3, 74) = 6.144$, $p < .001$, though the change in explained variance was not statistically significant, $\Delta R^2 = .032$, $F(2, 74) = 1.643$, $p = .202$. Employment was not a significant predictor, and stigma continued to significantly predict lower QoL ($\beta = -6.516$, $p < .001$).

In Step 3, parent sex was introduced as a dichotomous variable. The model was significant, $F(4, 73) = 4.661$, $p = .002$, but again the increase in explained variance was non-significant, $\Delta R^2 = .004$, $F(1, 73) = 0.371$, $p = .545$. Stigma remained a robust and significant predictor ($\beta = -5.155$, $p = .008$).

In Step 4, parental age was added as a categorical variable. The model remained significant, $F(6, 71) = 3.468$, $p = .005$, with a non-significant increase in explained variance, $\Delta R^2 = .023$, $F(2, 71) = 1.091$, $p = .350$. Stigma continued to predict lower QoL ($\beta = -4.441$, $p = .030$).

Finally, in Step 5, education level was introduced. The full model was significant, $F(9, 68) = 2.883$, $p = .006$, with a modest and non-significant increase in variance explained, $\Delta R^2 = .050$, $F(3, 68) = 1.511$, $p = .209$. Stigma remained a statistically significant predictor of QoL ($\beta = -4.342$, $p = .035$). The final model accounted for 18% of the variance in QoL (adjusted $R^2 = .180$). Standardised results showed that a one standard deviation (SD) increase in perceived stigma was associated with a 0.268 SD decrease in QoL. Taken together, these findings indicate that perceived stigma has a consistent and significant negative impact on children's quality of life, even after adjusting for key demographic variables.

Discussion

This study sought to deepen understanding of the experiences of families caring for CwRD in Romania by focusing on three key objectives. First, it aimed to quantify the QoL of these children as reported by their parents/primary caregivers. Second, it endeavoured to elucidate the extent to which caregivers perceive stigma associated with raising a CwRD. Finally, the study explored the potential impact of perceived stigma on QoL, thereby shedding light on how social attitudes may influence the lived experiences of these families. Consistent with existing literature, it was expected that parents/caregivers would report diminished QoL and significant perceived stigma,

and that higher levels of stigma would be associated with lower reported QoL. Together, these aims and hypotheses provide a foundational framework for interpreting the nuanced interplay between societal perceptions and family wellbeing in an under researched context.

QoL

As indicated above, 78.2% of the participants in this study scored low functioning on the Physical Functioning Subscale, and 97.43% scored low functioning on the Psychosocial Health Subscale. Furthermore, the participants averaged a PedsQL score of 36.04. This finding aligns with previous studies on QoL in rare diseases, which have ascertained that individuals with rare diseases experience a lower QoL than the general population and patients with common diseases (Bogart & Irvin, 2017). To contextualise this score, recent literature indicates that the average Total Score in healthy paediatric samples is typically above 80 (Smyth & Jacobson, 2021). The difference observed in this study corresponds to a Cohen's d effect size of approximately 0.89, representing a large effect and reinforcing that CwRD experience significantly poorer QoL than their healthy peers. This is not only clinically relevant but also consistent with findings from other chronic illness cohorts, such as children undergoing induction chemotherapy or those with moderate-to-severe inflammatory bowel disease (Smyth & Jacobson, 2021).

There is currently a limited number of studies providing normative PedsQL scores specifically for children with rare diseases, which makes direct comparison challenging. In general, published data indicate that healthy children typically score between 80 and 85 on the Total Scale Score, with physical functioning often highest (85–90), and emotional or school functioning slightly lower (typically 75–85) (Varni et al., 2014). Children with more common chronic health conditions tend to score lower, usually in the range of 65–75, with some domains dropping to 50–60 depending on disease severity (Raghunandan et al., 2023). In the present study, the PedsQL scores reported for children with rare diseases in Romania were considerably lower than these established thresholds. Mean scores across key domains ranged between 34 and 40, indicating substantial impairment when compared to healthy and chronically ill paediatric populations in other countries. These findings underscore the pronounced vulnerability of this population and the critical importance of targeted support services for children with rare diseases and their families.

These comparisons support the interpretation that children with rare diseases are at substantial risk for diminished physical and psychosocial functioning. Moreover, the variability in scores (as reflected in the standard deviations) suggests considerable heterogeneity in QoL among CwRD, which may reflect differences in disease severity, access to care and family support systems. Taken together, these results highlight an urgent need for tailored interventions that address the complex needs of children with rare diseases and their families.

The above may relate to the physical and physiological experiences that accompanying rare diseases. People with rare diseases often encounter a range of physical symptoms impacting their mobility, cardiovascular function, sleep, experiences of pain and more (Marcus et al., 2022). Previous research has also revealed that people

with rare diseases experience lower physical QoL due to feelings of exhaustion (Marcus et al., 2022). In the present study, 62.8% of parent/primary caregivers indicated that their child almost always or often experiences low energy levels, aligning with previous research findings.

Nevertheless, the present and previous studies also identify other factors, external to physical aspects, influencing the QoL in patients with rare diseases. An earlier study observed that most patients with rare disease experience lower QoL due to their social functioning, chiefly being able to do usual activities and lead an ordinary life (Marcus et al., 2022). In the present study, 83.3% of parent/primary caregivers indicated that their children have problems with doing things other people their age can do, and 71.8% indicated that their children cannot keep up with their peers. Therefore, the findings of this study reverberate the findings of other research studies on the QoL of patients with rare diseases.

Some research has indicated that the experiences of misunderstanding within healthcare systems are a key determinant of QoL for patients with rare diseases (Caputo, 2014). Rare disease patients have previously reported that the healthcare they receive is largely disjointed, with limited congruency between treatments (Caputo, 2014; Grut & Kvam, 2012). Uncoordinated care is evident within the Romanian healthcare system, where patients with rare diseases often do not experience equitable access or continuity in healthcare (Severin & Dan, 2022).

Experiences of perceived stigma

Notably, it is widely accepted that people with rare diseases encounter stigma because of their health conditions. However, the present research study has identified that 56.41% of the participants experienced high degrees of perceived stigma and a further 20.51% indicated that they are confronted with moderate degree of perceived stigma, comprising 76.92% of the total participants. This finding aligns with research in the United States that has identified that people with rare diseases experience high levels of stigma which has a pervasive impact on their lives (Munro et al., 2022).

Further, many participants in previous studies have identified that they experience high levels of interpersonal stigma (Munro et al., 2022; von der Lippe et al., 2017), emerging due to a scarcity of perceived social support, invalidation of their health conditions or a limited understanding from others about the nature of these conditions (Munro et al., 2022). High levels of interpersonal stigma were evident in the responses to various questions in the present study through the PPPAS. For example, 58.97% of parent/primary caregivers identified that they were afraid that people would avoid them if they found out about the child's rare disease. This, again, may be because of inadequate education about these conditions provided to parent/primary caregivers and communities.

Another principal aspect of rare conditions is that many have genetic origins. As such, people with rare diseases in other research studies have identified that due to the genetic origin of their rare disease, they are more likely to keep these a secret from their communities for fear of being blamed for their conditions (Zhu et al., 2017). A similar finding was identified in the present study, with 69.23% of the respondents indicating

that they believe that others think that bad parental genres are the causes of their child's disability.

Impact of stigma on QoL

Previous research has identified a relationship between stigma and QoL in diverse populations (Chou et al., 2009; Ernst et al., 2017; Mendonca et al., 2023). In addition, some studies have also studied the relationship between these two measures in rare disease populations (Bogart & Irvin, 2017; Konradi, 2022).

In line with the two previous studies on QoL and stigma in patients with rare diseases, the present research designates a negative relationship between experiences of perceived stigma and QoL in CwRD in Romania. As parent/primary caregivers reported higher perceived stigma, the reported PedsQL score declined even when controlled for age, gender, education and employment.

The relationship between stigma and quality of life was explored and deemed important in this study because families of children with rare diseases often face a unique and compounded set of challenges that are underrepresented in research (Černe et al., 2024), particularly in Eastern European contexts like Romania. Rare diseases are frequently misunderstood by the public and medical professionals alike, which can result in parents encountering judgement, blame or social exclusion, phenomena that contribute to perceived stigma (Černe et al., 2024). Unlike more common chronic conditions, rare diseases are often invisible, complex and without clear treatment pathways, intensifying caregiver isolation and emotional strain (Bogart & Irvin, 2017).

By exploring this relationship, the current study aimed to uncover how stigma operates within the family system and impacts child outcomes beyond the direct effects of the medical condition itself. This is particularly relevant in countries with limited support infrastructures, where stigma may be more pervasive and formal resources scarcer (Birau et al., 2019). Investigating the link between parental stigma and child QoL provides a rationale for designing holistic interventions, ones that address not only medical needs but also social and psychological barriers to participation and wellbeing. This research contributes to the growing body of literature that calls for greater attention to the psychosocial dimensions of rare disease care, and the importance of family-focused support models.

Lasting impact of communism on stigma and QoL

Although not a primary focus of the study, participant responses highlighted the significance of broader sociocultural influences, particularly religious and political ideologies, on experiences of stigma and QoL in CwRD. Notably, 71.8% of the respondents identified that parent/primary caregivers felt that most people think that having a CwRD is a punishment for parental sin. The concept of 'sin', otherwise perceived as a 'violation of G-d's will', is deeply intertwined with stigma in religious countries (Rawls, 2009), such as Romania. While religion was not a demographic variable explicitly measured in this study, this finding echoes longstanding cultural narratives in Romania, a country where religious identity remains prevalent. The prominence of this theme emerged unexpectedly, particularly through responses to the PPPAS. Although demographic data on

religious affiliation and political orientation were not collected, the consistency with which respondents endorsed items referencing moral judgment (e.g. sin as a cause of a child's condition) warrants careful reflection. Future research would benefit from exploring these dimensions more explicitly to better understand their impact on stigma and QoL in families navigating life with a rare disease.

Strengths and limitations

This study was the first to measure the QoL of CwRD and the experiences of perceived stigma of parent/primary caregivers of CwRD in Romania, thereby contributing to the otherwise limited body of research in this population. Further, few studies globally have looked at either experiences of stigma or QoL in CwRD. As such, this study can guide future research on these measures within this population.

However, this study used convenience sampling techniques, and many patients with rare diseases do not receive a timely diagnosis and, therefore, would not have been eligible to complete this questionnaire (Boulanger et al., 2020). In addition, a cross-sectional design was used in this study. This is significant as the adjustment to chronic conditions, such as rare diseases, is an ongoing and dynamic process (Livneh, 2021) that cannot be adequately captured in this study design. Further, this may preclude the determination of causality and raises the possibility that children's quality of life may, in fact, influence parental perceptions of stigma. Additionally, QoL was assessed solely through caregiver reports, rather than directly from the CwRD themselves. As such, the findings reflect parental perceptions, which may not fully capture the children's lived experiences or accurately represent their own assessments of wellbeing.

It may be beneficial for future research to identify trends between demographic factors of both the children and their parent/primary caregivers to QoL and experiences of perceived stigma. Additional studies may also benefit from intervention-based methodologies to determine how these perceived experiences of stigma and lower QoL may be addressed through tangible interventions. Furthermore, this research identified that there was a relatively low variance in QoL, indicating that there must be other factors outside of stigma to explain the remaining 82% variance. As such, future research may benefit from identifying what other factors may be contributing to the low quality of life in CwRD in Romania.

Conclusions

Based on the findings of this study, it is evident that there is a negative correlation between high parent/primary caregivers perceived stigma and a low quality of life of their CwRD.

This study also identified that a significant majority of the population experienced a low-functioning physical QoL. As such, it may be beneficial for more physical functioning support to be provided to this population. Increasing physical functioning provision may be done by increased access to physiotherapy and occupational therapy. Further, through increasing the education and resources available to parents of CwRD, parent/primary caregivers will be better prepared to facilitate the physical needs of their children.

Grounded on the findings of this study, it is also palpable that CwRD experiences a low psychosocial functioning level. It is, thereby, imperative to identify interventions that may address these difficulties, including improved education within social and community settings to encourage the inclusion and development of programs for CwRD.

In addition, experiences of stigma have adverse effects on the QoL of this population. As such, interventions are needed to address the public perception of rare diseases, their origins and the impacts that these can have on people's lives. It may be valuable to target these messages through religious organisations due to the spiritual nature of the stigma experienced by parent/primary caregivers of CwRD in Romania. Further, it may also be advantageous to develop interventions that empower teachers to educate the next generation about rare diseases and the importance of inclusion for these populations.

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Data availability statement

The data that support the findings of this study are available on request from the corresponding author, SG. The data are not publicly available due to the data containing information that could compromise the privacy of research participants.

References

Adama, E. A., Arabiat, D., Foster, M. J., Afrifa-Yamoah, E., Runions, K., Vithiatharan, R., & Lin, A. (2020). The psychosocial impact of rare diseases among children and adolescents attending mainstream schools in Western Australia. *International Journal of Inclusive Education*, *2020*(9), 1–14. https://doi.org/10.1080/13603116.2021.1888323

Adama, E. A., Arabiat, D., Foster, M. J., Afrifa-Yamoah, E., Runions, K., Vithiatharan, R., & Lin, A. (2023). The psychosocial impact of rare diseases among children and adolescents attending mainstream schools in Western Australia. *International Journal of Inclusive Education*, *27*(12), 1273–1286. https://doi.org/10.1080/13603116.2021.1888323

Baciu, E.-L., & Lazar, T.-A. (2017). Between equality and discrimination: Disabled persons in Romania. *Transylvanian Review of Administrative Sciences*, *51*(1), 5–19. https://doi.org/10.24193/tras.51E.1

Beverung, L. M., Varni, J. W., & Panepinto, J. A. (2015). Clinically meaningful interpretation of paediatric health-related quality of life in sickle cell disease. *Journal of Paediatric Haematology and Oncology*, *37*(2), 128–133. https://doi.org/10.1097/MPH.0000000000000177

Birau, F. R., Dănăcică, D.-E., & Spulbar, C. M. (2019). Social exclusion and labour market integration of people with disabilities: A case study for Romania. *Sustainability*, *11*(5014), 1–15. https://doi.org/10.3390/su11185014

Bogart, K., Hemmesch, A., Barnes, E., Blissenbach, T., Beisang, A., Engel, P., Tolar, J., Schacker, T., Schimmenti, L., Brown, N., Morrison, K., Albright, T., Klein, M., Coleman, J., Nelsen, K., Blaylark, R., LaFond, K., Berkowitz, S.Schultz, K. A. & The Chloe Barnes Advisory Council on Rare Diseases. (2022). Healthcare access, satisfaction, and health-related quality of life among children and adults with rare diseases. *Orphanet Journal of Rare Diseases*, *17*(1), 1–18. https://doi.org/10.1186/s13023-022-02343-4

Bogart, K., & Irvin, V. L. (2017). Health-related quality of life among adults with diverse rare disorders. *Orphanet Journal of Rare Diseases*, *12*(1), 177. https://doi.org/10.1186/s13023-017-0730-1

Boulanger, V., Schlemmer, M., Rossov, S., Seebald, A., & Gavin, P. (2020). Establishing patient registries for rare diseases: Rationale and challenges. *Pharmaceutical Medicine*, *34*(1), 185–190. https://doi.org/10.1007/s40290-020-00332-1

Caputo, A. (2014). Exploring quality of life in Italian patients with rare disease: A computer-aided content analysis of illness stories. *Psychology, Health & Medicine*, *19*(2), 211–221. https://doi.org/10.1080/13548506.2013.793372

Černe, T., Kragelj, L. Z., Turk, E., & Pavlič, D. R. (2024). Experiences of quality of life and access to health services among rare disease caregivers: A scoping review. *Orphanet Journal of Rare Diseases*, *19*(1), 319. https://doi.org/10.1186/s13023-024-03327-2

Chen, S., Wang, Y., Zhu, L., Gan, Y., & Dong, D. (2022). Factors associated with the psychological quality of life among adolescents with rare diseases in China: A national repetitive cross-sectional study. *Social Psychiatry and Psychiatric Epidemiology*, *57*(1), 1723–1726. https://doi.org/10.1007/s00127-022-02286-y

Chou, Y. C., Pu, C. Y., Lee, Y. C., Lin, L. C., & Kröger, T. (2009). Effect of perceived stigmatisation on the quality of life among ageing female family carers: A comparison of carers of adults with intellectual disability and carers of adults with mental illness. *Journal of Intellectual Disability Research*, *53*(7), 654–664. https://doi.org/10.1111/j.1365-2788.2009.01173.x

Chu, S.-Y., Wen, C.-C., & Weng, C.-Y. (2022). Gender differences in caring for children with genetic or rare diseases: A mixed-methods study. *Children*, *9*(627), 1–13. https://doi.org/10.3390/children9050627

Čolic, M., & Milačić-Vidojević, I. (2021). Perceived stigma among Serbian parents of children with autism spectrum disorder and children with physical disabilities: Validation of a new instrument. *Journal of Autism and Developmental Disorders*, *51*(1), 501–513. https://doi.org/10.1007/s10803-020-04559-4

Detiček, A., Locatelli, I., & Kos, M. (2018). Patient access to medicines for rare diseases in European countries. *Value in Health*, *21*(1), 553–560. https://doi.org/10.1016/j.val.2018.01.007

Douglas, C. M. W., Aith, F., Boon, W., de Neiva Borba, M., Doganova, L., Grunebaum, S., Hagendijk, R., Lynd, L., Mallard, A., Ali Mohamed, F., Moors, E., Cordovil Oliveira, C., Paterson, F., Scanga, V., Soares, J., Raberharisoa, V., & Kleinhout-Vliek, T. (2022). Social pharmaceutical innovation and alternative forms of research, development, and deployment for drugs for rare diseases. *Orphanet Journal of Rare Diseases*, *17*(344), 1–13. https://doi.org/10.1186/s13023-022-02476-6

Ernst, J., Mehnert, A., Dietz, A., Hornemann, B., & Esser, P. (2017). Perceived stigmatisation and its impact on quality of life: Results from a large register-based study including breast, colon, prostate and lung cancer patients. *BMC Cancer*, *17*(1), 741. https://doi.org/10.1186/s12885-017-3742-2

European Commission. (2022, September 20). *Rare diseases*. Research and innovation. Retrieved November 15, 2022, from, https://research-and-innovation.ec.europa.eu/research-area/health/rare-diseases_en

EURORDIS. (2025). *What is a rare disease?* Eurordis rare disease Europe. Retrieved July 22, 2025, from, https://www.eurordis.org/information-support/what-is-a-rare-disease/

Grut, L., & Kvam, M. H. (2012). Facing ignorance: People with rare disorders and their experiences with public health and welfare services. *Scandinavian Journal of Disability Research*, *15*(1), 20–32. https://doi.org/10.1080/15017419.2011.645870

Hu, Y.-L., Chang, C.-C., Lee, C.-H., Liu, C.-H., Chen, Y.-J., Su, J.-A., Lin, C.-Y., & Griffiths, M. D. (2023). Associations between affiliate stigma and quality of life among caregivers of individuals with dementia: Mediated roles of caregiving burden and psychological distress. *Asian Journal of Social Health and Behaviour*, *6*(2), 64–71. https://doi.org/10.4103/shb.shb_67_23

Konradi, A. (2022). Fibrous dysplasia patients with and without craniofacial involvement report reduced quality of life inclusive of stigma, depression, and anxiety. *Chronic Illness*, *18*(4), 927–936. https://doi.org/10.1177/17423953211049436

Lee Aiyegbusi, O., Isa, F., Kyte, D., Pankurst, T., Kerecuk, L., Ferguson, J., Lipkin, G., & Calvert, M. (2020). Patient and clinician opinions of patient reported outcome measures (PROMs) in the management of patients with rare diseases: A qualitative study. *Health and Quality of Life Outcomes*, *18*(177), 1–12. https://doi.org/10.1186/s12955-020-01438-5

Livneh, H. (2021). Psychosocial adaptation to chronic illness and disability: An updated and expanded conceptual framework. *Rehabilitation Counseling Bulletin*, *65*(3), 171–184. https://doi.org/10.1177/00343552211034819

Major, B., Dovidio, J. F., Link, B. G., & Calabrese, S. K. (2018). *The Oxford handbook of stigma, discrimination, and health*. Oxford University Press.

Marcus, E., Stone, P., Thornburn, D., Walmsley, M., & Vivat, B. (2022). Quality of life for people with primary sclerosing cholangitis: A pragmatic strategy for identifying relevant quality of life issues for rare disease. *Journal of Patient-Reported Outcomes*, *6*(76), 1–12. https://doi.org/10.1186/s41687-022-00484-5

Mendonca, C. J., Newton-John, T. R. O., Aperstein, D. M., Begley, K., Hennessy, R. M., & Bulsara, S. M. (2023). Quality of life of people living with HIV in Australia: The role of stigma, social disconnection and mental health. *AIDS & Behaviour*, *27*(2), 545–557. https://doi.org/10.1007/s10461-022-03790-7

Mitter, N., Ali, A., & Scior, K. (2019). Stigma experienced by families of individuals with intellectual disabilities and autism: A systematic review. *Research in Developmental Disabilities*, *89*, 10–21. https://doi.org/10.1016/j.ridd.2019.03.001

Munro, M., Cook, A. M., & Bogart, K. R. (2022). An inductive qualitative content analysis of stigma experienced by people with rare diseases. *Psychological Health*, *37*(8), 948–963. https://doi.org/10.1080/08870446.2021.1912344

Pierpont, E. L., Simmons, J. H., Spurlock, K. J., Shanley, R., & Sarfoglou, K. M. (2021). Impact on paediatric hypophosphatasia on behavioural health and quality of life. *Orphanet Journal of Rare Disease*, *16*(80), 1–10. https://doi.org/10.1186/s13023-021-01722-7

Puiu, M., & Dan, D. (2010). Rare diseases, from European resolutions and recommendations to actual measures and strategies. *Medica*, *5*(2), 128–131.

Raghunandan, R., Howard, K., Smith, S., Killedar, A., Cvejic, E., Howell, M., Petrou, S., Lancsar, E., Wong, G., Craig, J., & Hayes, A. (2023). Psychometric evaluation of the PedsQL GCS and CHU9D in Australian children and adolescents with common chronic health conditions. *Applied Health Economics and Health Policy*, *21*(6), 949–965. https://doi.org/10.1007/s40258-023-00836-2

Rawls, J. (T. Nagel (Ed). (2009). *A brief inquiry into the meaning of sin and faith*. Harvard University Press.

Rusu, D. M., Stevanović, D., & Enea, V. (2024). Affiliate stigma and parental stress among parents of children with autism spectrum disorder: The mediating role of shame. *Focus on Autism and Other Developmental Disabilities*, *39*(2), 127–135. https://doi.org/10.1177/10883576231221751

Sakiz, H., & Kaçan, H. (2023). Resilience and mental health literacy mediate the effect of caregiver burden on internalised stigma among mothers of children with disabilities. *Children's Health Care*, *54*(4), 1–24. https://doi.org/10.1080/02739615.2023.2290264

Severin, E., & Dan, D. (2022). Lack of information on the effects of COVID-19 on rare pathologies has further hampered access to healthcare services. *Frontiers in Public Health*, *10*(852880), 1–3. https://doi.org/10.3389/fpubh.2022.852880

Smyth, M., & Jacobson, K. (2021). Pediatric quality of life inventory™ version 4.0 short form generic core scale across pediatric populations review data. *Data in Brief*, *39*, 107599. https://doi.org/10.1016/j.dib.2021.107599

United Nations. (2023). Department of Economic and Social Affairs, Population Division. World population prospects 2023. https://population.un.org/wpp/

Varni, J. W., Burwinkle, T. M., & Seid, M. (2014). The PedsQL as a paediatric patient-reported outcome: Reliability and validity of the PedsQL measurement model in 25, 000 children. *Expert Review of Pharmacoeconomics & Outcome Research*, *5*(6), 705–719. https://doi.org/10.1586/14737167.5.6.705

Varni, J. W., Seid, M., & Rode, C. A. (1999). The PedsQL: Measurement model for paediatric quality of life inventory. *Medical Care*, *37*(2), 126–139. https://doi.org/10.1097/00005650-199902000-00003

Verger, Sebastià, Negre, F., Rosselló, Paz-Lourido, B., & Rosselló, M. R. (2020). Inclusion and equity in educational services for children with rare diseases: Challenges and opportunities. *Children and Youth Services Review*, *119*(1), 105518. https://doi.org/10.1016/j.childyouth.2020.105518

Verstraete, J., & Scott, D. (2022). Comparison of the EQ-5D-Y-5L, EQ-5D-Y-3L and PedsQL in children and adolescents. *Journal of Patient-Reported Outcomes*, *6*(67), 1–12. https://doi.org/10.1186/s41687-022-00480-9

von der Lippe, C., Diesen, P. S., & Feragen, K. B. (2017). Living with a rare disorder: A systematic review of the qualitative literature. *Molecular Genetics & Genomic Medicine*, *5*(6), 758–773. https://doi.org/10.1002/mgg3.315

Wiegand-Grefe, S., Liedtke, A., Morgenstern, L., Hoff, A., Csengoe-Norris, A., Johannsen, J., Denecke, J., Barkmann, C., Grolle, B., Daubmann, A., Wegscheider, K., & Boettcher, J. (2022). Health-related quality of life and mental health of families with children and adolescents affected by rare diseases and high disease burden: The perspectives of affected children and their siblings. *BMC Paediatrics*, *22*(1), 1–9. https://doi.org/10.1186/s12887-022-03663-x

Zhu, X., Smith, R. A., & Parrott, R. L. (2017). Living with a rare health condition: The influence of a support community and public stigma on communication, stress, and available support. *Journal of Applied Communication Research*, *45*(2), 179–198. https://doi.org/10.1080/00909882.2017.1288292

Zurynski, Y., Deverell, M., Dalkeith, T., Johnson, S., Christodoulou, J., Leonard, H., Elliott, E. J., & ASPU Rare Disease Impacts on Families Study Group. (2017). Australian children living with rare diseases: Experiences of diagnosis and perceived consequences of diagnostic delays. *Orphanet Journal of Rare Diseases*, *12*(68), 1–9. https://doi.org/10.1186/s13023-017-0622-4