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Quality of life outcomes and predictors for youth with intellectual disability and rare diseases

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ABSTRACT

The concept of quality of Life (QoL) has become particularly relevant in the field of intellectual disability (ID). However, the existing scientific literature on QoL of people with ID and rare diseases (RDs) is very limited. Therefore, this study aimed to analyze individual QoL of children and youth with RDs and to evaluate the impact of personal and contextual factors on their QoL. The sample was composed of 114 Spanish participants with ID and RDs, aged between 4 and 21 years. The KidsLife Scale was applied. This instrument includes 96 items, third-person formulated, organized across eight QOL domains: physical wellbeing, material wellbeing, emotional wellbeing, interpersonal relationships, social inclusion, rights and personal development. Descriptive statistics were calculated and correlation coefficients and multiple linear regression were computed. The most elevated scores were achieved in physical well-being and material wellbeing, whereas the lowest scores were shown in social inclusion and self-determination domains. Sex, level of support needs, percentage of disability and size of the organization were predictive factors of QoL scores. These outcomes shed light into priority areas and predictors that should be considered in the development of individualized supports, organizational approaches and policy initiatives intended to promote the QoL of children and youth with RDs and ID.

What this paper adds?

This study contributes to the understanding of the quality of life (QoL) of children and youth with rare disorders (RDs) and intellectual disability (ID) by addressing several critical gaps in the existing literature. First, it expands the scope of QoL research beyond the commonly focused health-related aspects, analyzing individual QoL across eight diverse. This broader approach

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allows for a more comprehensive view of the lived experiences of this population.

The findings indicate that while the overall QoL scores seem generally positive, with strengths in material and physical well-being domains. However, significant challenges remain in the social inclusion and self-determination. These lower scores highlight the need for targeted interventions and support services to enhance social participation and personal agency among them.

Moreover, the study identifies personal and contextual variables that significantly impact QoL, such as sex, level of support needs, percentage of disability, and the size of the organization providing support. The study underscores the necessity of integrating a gender perspective in developing strategies and programs to ensure equitable opportunities, as well as the importance of equipping organizations with sufficient resources so that they can offer inclusive, person-centered services.

Overall, it provides a foundation for future research and emphasizes the need for inclusive policies and practices that improve QoL outcomes for this population.

1. Introduction

Rare diseases (RDs) are characterized by their low prevalence, defined in Europe as affecting fewer than one in 2000 individuals (European Commission, 2020). The World Health Organization (WHO) recognizes more than 7000 diseases as rare, impacting approximately 7 % of the global population in their physical abilities, mental skills, and sensory and behavioral functions (González et al., 2016). Despite their etiological differences, most RDs are chronic, genetic, and affect multiple body systems and medical specialties, with few having effective treatments or cures (Bogart et al., 2022). Most of these diseases are present in childhood (Nguengang et al., 2020), but due to their low prevalence, the accurate diagnosis of RDs is complex and subject to significant delays (López-Bastida et al., 2016a).

RDs are often severely debilitating or even life-threatening, leading to reduced quality of life (QoL) for those affected (Chen et al., 2022; Gao et al., 2020). Despite their impact, RDs remain under-researched, primarily due to their low prevalence, which makes it difficult to recruit sufficient study populations, as well as limited resources allocated for research. Consequently, there is insufficient evidence for effective translation into clinical practice and health and social policy (López-Bastida et al., 2016a; Paz-Lourido et al., 2020). Given that many challenges are experienced by people with RDs, over the past two decades, researchers have highlighted the importance of measuring the QoL of children with RDs to better understand the effects these diseases have on their lives (Gao et al., 2020; González et al., 2016; Wiegand-Grefe et al., 2022), and considering as a collective for improving policy planning (Ng et al., 2022).

As far as we know, the limited studies addressing quality of life in people with RDs focus on the narrower construct of health-related QoL (HrQoL; WHOQOL, 1995). These few studies have found that the population with RDs scores lower on HrQoL compared to other common chronic disease groups and the general population (Bogart et al., 2017, 2022; Efthymiadou et al., 2018; Johansen et al., 2013; Ng et al., 2022; Wiegand-Grefe et al., 2022). In this context, the most used tool in the literature assessing HrQoL of people with RDs is the EQ-5D scale, a five-item generic scale developed by the EuroQol group and recommended by the NICE guidelines for clinical and economic assessment. This instrument has demonstrated moderate responsiveness but has limited validity evidence based on test content when applied to this population (Dumbuya et al., 2024; Rao et al., 2024).

Some studies on people with RDs have examined the associations between HrQoL and influencing factors. Regarding sex, findings are somewhat mixed. Indeed, Gao et al. (2020) did not find significant differences, while Xu et al. (2023) reported lower scores in men, and Bogart & Irvin, 2017 found lower scores in women. Similarly, the connection between age and QoL remains ambiguous: some studies indicate lower scores among older participants (Bogart & Irvin, 2017; Xu et al., 2023), whereas Ng et al. (2022) found no relations with mean participant age. Additionally, the association between the type of schooling and QoL is inconclusive, as no link haven been observed (Linertová et al., 2019).

Variables associated with lower health-related QoL in people with ID include a long duration of the disease, having multiple RDs, being unemployed, having a low income, having children, and not using assistive devices (Bogart & Irvin, 2017; Xu et al., 2023). Additionally, experiencing pain or discomfort (Efthymiadou et al., 2018), having neurological diseases (Ng et al., 2022; Xu et al., 2023), lower disease prevalence (Rao et al., 2024), and living in public housing or subdivided units (Ng et al., 2022) are also linked to reduced HrQoL.

Our research focuses on people who have both RDs and intellectual disability (ID) due to the lack of specific research focused on the QoL of this population. Despite the high percentage of people with RDs who also present with ID and severe impairments in language and cognition (Valdez et al., 2016), the existing scientific literature is very limited. Notably, studies specifically examining HrQoL and rare disorders co-occurring with ID, such as fragile X syndrome, Prader-Willi syndrome, and Cri du Chat syndrome, have primarily focused on social and economic costs. These studies report lower QoL outcomes in comparison to the general population but do not explore the influencing variables (Chevreul et al., 2016; Chevreul etal., 2015; Kodra et al., 2020; López-Bastida et al., 2016b).

Additionally, much of the existing literature regarding the QoL of people with RDs examines HrQoL, focusing on how the disease impacts individual functioning (De Maeyer et al., 2011) and understanding QoL as the absence of pathology (Cummins et al., 2004), our research adopts a different perspective. We move away from the narrow focus of medical models to emphasize a more holistic understanding of QoL, transcending health-related aspects and considering all significant facets of people's life (Bacherini et al., 2024). This approach aligns with the QoL supports model, which highlights the importance of strengths, preferences, rights, and supports systems in enhancing QoL (Gómez et al., 2021a; Morán et al., 2023; Verdugo et al., 2023; Verdugo & Schalock, 2024).

Within the QOL supports model, the eight-domain QoL model proposed by (Schalock & Verdugo, 2002) is particularly significant in the field of ID. According to this model, individual QoL is understood as a desired state of personal well-being that encompasses all important facets of life for all human beings, including physical wellbeing, material wellbeing, emotional wellbeing, interpersonal relationships, social inclusion, rights and personal development. This notion includes universal properties while others are determined by contextual or cultural variables; it includes both objective and subjective components; and it is affected by personal and contextual variables (Schalock et al., 2011). This model is the most recognized and extensively employed framework by professionals providing support to people with ID in many countries, including Spain (Navas et al., 2024; Verdugo et al., 2023). Furthermore, it is backed by robust empirical evidence supporting its applicability not only for people with ID but also for other groups in vulnerable situations (e.g., Amor et al., 2023; Amor et al., 2025; Arias et al., 2018; Gómez et al., 2016, 2020a, 2020b, 2021b, 2025; Fernández et al., 2018; Morán et al., 2019a; Morán & Gómez, 2025; Swerts et al., 2022, 2023).

This broader and contextual perspective on QoL, encompassing aspects beyond those strictly related to health, has been largely overlooked in people with RDs. Notably, within this socioecological framework, there is only one exploratory study focusing on assessing QoL-related outcomes and predictors in people with RDs (González et al., 2016). In this study, González et al. (2016) employed the field-test version of the KidsLife Scale (Gómez et al., 2016), an instrument specifically designed to address the QoL of children and adolescents with ID and significant support needs. Their findings indicated that they scored higher in physical wellbeing, while the lowest scores were found in social inclusion. The level of ID and support needs resulted in significant differences for the total score of the scale. Analyses by domains showed differences by sex, level of ID, and type of schooling. In contrast, no significant differences were found for age, type of disease, or region of residence.

Although these findings provide an important foundation for further research, this study has several limitations: (a) it used the field-test version of the KidsLife Scale rather than a validated version, rendering its results preliminary and lacking of evidence of reliability and validity; (b) the analyses conducted were univariate, preventing the evaluation of potential interactions between independent variables or the influence of potential confounding variables on QoL; and (c) other factors that may affect QoL and that were examined in other populations, such as comorbidities or the size of the organization providing support, were not explored. In this sense, for instance, attending small organizations was associated with lower outcomes in the rights domain in populations of youth with other disabilities (Morán et al., 2022, 2024).

To address this gap and advance research in this area, the objectives of the current study were: (a) to analyze individual QoL across the eight domains for children and youth with RDs and ID; and (2) to evaluate the impact of various personal (i.e., sex, age, level of ID, level of support needs, degree of dependency, percentage of disability, and comorbidities) and contextual variables (i.e., type of schooling and size of the organization providing support) on their QoL scores. Drawing on findings from earlier studies involving this group and others with disabilities, we hypothesize that participants with comorbidities, greater age, increased support needs, a higher degree of dependency, percentage of disability, or level of ID, and receiving support by smaller organizations will score lower in QoL (e.g., Bogart & Irvin, 2017; González et al., 2016; Morán et al., 2022, 2024; Morán & Gómez, 2025; Xu et al., 2023). Conversely, we anticipate that no significant differences in QoL will be found based on sex and type of schooling (Gao et al., 2020; Linertová et al., 2019), except in the physical wellbeing and social inclusion domains, respectively (González et al., 2016).

2. Methods

2.1. Participants

Participants were 114 people with RDs and ID that were attending educational, social, or health services that were delivered through mainstream schools, special education centers, day services, residential centers and clinics in Spain. The sole exclusion criterion was if the person was not within educational settings (as the questionnaire encompass items pertaining to educational but not employment settings). Their ages were between 4 and 21 years ($M = 13$; $SD = 4.8$) and the majority ($n = 64$; 56 %) were male.

According to official records at participating organizations, most participants had a severe (47 %) or a moderate (31 %) level of ID, and a generalized level of support needs (52 %). In regard to the legally recognized degree of dependency, most people (61 %) were recognized as having great dependency. The most prevalent RD, as shown in appendix 1, was Fragile X Syndrome (18 %) and seven participants had two RDs at the same time. In addition, the majority of participants (76 %) presented associated conditions (appendix 2) such as physical disability (51 %), or epilepsy (25 %). The vast majority (78 %) attended special schools. Most participants were living in their family home (82 %). Sociodemographic characteristics of people being evaluated are presented in more detail in appendix 2.

The questionnaires were completed by 97 respondents, mostly women (82 %) whose average age was 44.5 years old ($SD = 12.1$). Most of them were professionals (71 %), mainly teachers (48 %). The rest were parents (29 %), primarily mothers (25 %). Each of these respondents evaluated between 1 and 5 people. The frequency of contact with the person being evaluated was daily or several times per week in most cases (93 %) and the mean length of relationship with the person was 6.6 years ($SD = 5.7$). A total of 59 % of the main respondents needed to consult other people in order to complete the scale, mostly professionals (43 %) and family members (33 %).

Participants received support from 54 organizations that spread across Spain, primarily located in urban settings (82 %). These organizations provided subsidized (72 %), private (24 %), or public (4 %) services in the fields of health care (1 %), social care (12 %), and education (87 %). Most organizations (69 %) were serving fewer than 100 users.

2.2. Instrument

The KidsLife Scale (Gómez et al., 2016) was applied. This instrument assesses the individual QoL of people with ID, aged from 4 to 21 years old who are users of health, educational or social services. The scale is administered by third-party raters (e.g., relatives, caregivers, teachers), who know the person well (at least 6 months) and who have opportunities to observe them for long periods in different contexts. The scale includes 96 items, third-person formulated, organized across the eight QOL domains proposed by (Schalock & Verdugo, 2002): physical wellbeing, material wellbeing, emotional wellbeing, interpersonal relationships, social inclusion, rights and personal development. Each domain has a total of 12 items with four frequency options (never sometimes, often, always). The scale also includes a section to collect sociodemographic data about the person with ID, the main informant, and the center providing support.

The KidsLife Scale yields a general QoL score and total scores for the eight domains. Standardized scores and percentiles for the eight domains along with total QoL index are also included, which allows obtaining a QoL profile. They can be utilized for reference comparisons or to assess improvements through longitudinal or cross-sectional studies. The scale is freely available in English, Korean, Portuguese and Spanish at https://inico.usal.es/instrumentos-de-evaluacion/

The KidsLife Scale has excellent evidence related to its reliability and validity. In the validation study carried out with participants with ID, the internal consistency coefficients for each of the eight domains were suitable, ranging from .78 to .90, and the Cronbach's alpha for the entire scale was .96 (Gómez et al., 2016). Furthermore, the scale presented content validity evidence, along with evidence based on its internal structure, through confirmatory factor analysis that supports the intercorrelated eight-domain model (Gómez et al., 2014, 2016).

2.3. Procedure

Data collection was carried out through contact with organizations that support people with ID in Spain. A thorough web search was conducted and a mass-email was sent to all eligible participant organizations outlining the nature of the study and asking for their participation. Once centers indicated their interest in participating in the research, they were sent a link to an online survey requesting further details about their organization, the number of people to be evaluated, and the person who would coordinate the study.

Each coordinator was provided with a protocol needed to complete the scales with the aim of uniformizing the assessment procedure and enhancing the study's rigor. The protocol included information about the research, the instruction manual, access to the scale, and the informed consent to be completed by participants or their legal guardians. The research team was available to respond to questions and issues related to assessment administration, as well as arrange deadlines to deliver the questionnaires.

The study was also announced in scientific conferences, on social networks and on the websites of INICO and Plena inclusion (Full inclusion), the associative movement that advocates in Spain for the rights of people with intellectual and developmental disabilities comprising almost 900 member organizations.

The Ethics Committee of the University of Oviedo granted ethical approval and the principles for conducting research contained in the Declaration of Helsinki were followed. Alphanumeric codes were used to ensure data confidentiality. These codes permitted us to supply organizations with outcomes obtained by their users, so that they could apply them to implement evidence-based practices aimed to improve their QoL.

2.4. Data analyses

Data were analyzed with SPSS 25.0. Descriptive statistics of the total and domain-specific raw scores were collected (i.e., kurtosis, skewness, mean, standard deviation, median, mode, minimum and maximum). The existence of univariate outliers (i.e., participants with a z-value exceeding |3.29|) and multivariate outliers (i.e., participants with a Mahalanobis distance probability lower than.001) in the KidsLife total and domain-specific normalized scores, as well as in the two continuous normalized variables percentage of disability and age has been checked. The normality of the univariate and multivariate distributions of normalized continuous variables was analyzed by computing the skewness and kurtosis indices and Mardia's test as appropriate. It should be noted that the number of participants in some specific categories of level of support needs, level of ID, degree of dependency and size of the organization was very low. To address this, categories with low frequencies were grouped based on conceptual similarity and statistical justification, allowing us to retain interpretability while ensuring sufficient cell sizes for regression analysis. In this sense, categories with very few cases (e.g., fewer than 15) may produce unstable coefficients or inflated standard errors, potentially compromising model validity (Harrell, 2015; Peduzzi et al., 1996). Thus, we set a minimum threshold of approximately 15 cases per category to ensure the stability and reliability of estimates in the multiple regression models. In addition, the difference between the frequencies in the different categories of each ordinal variable were minimized but trying to maintain the highest number of categories as much as possible. For this purpose, the category with the lowest frequency was merged with the consecutive one. Thus, we retained three out four categories for three variables (level of ID, level of support needs, size of the organization) while only for degree of dependency the three original categories were merged into two ones.

Correlation coefficients were obtained to evaluate the relation among each individual variable (i.e., age, sex, level of support needs, level of ID, degree of dependency, percentage of disability) and contextual variable (i.e., size of the organization) and the QoL total score and each QoL domain. All the continuous variables were normalized before performing correlation analyses, according to (Tabachnick & Fidell, 2013) recommendation. Spearman's correlation coefficients were performed for the ordinal variables (i.e., level of support needs, level of ID, degree of dependency, and size of the organization), Pearson's correlation coefficients for the continuous

variables (i.e., age, percentage of disability), and point-biserial correlation coefficient for the nominal variable (i.e., sex). Following the guidelines proposed by Cicchetti et al. (2011), the correlation coefficients were classified as trivial (< .10), small (.10 – .29), medium (.30 – .49), large (.50 – .69), and very large (≥ .70).

Subsequently, multiple regression analyses were computed to examine the impact of several variables on the total and each QoL domain score. Specifically, nine multiple regressions were performed, one for each QoL domain or QoL total score, separately inserted as the dependent variable. All the variables identified as statistically significant in relation to the considered QoL domain or the QoL total score ($p < .05$) were entered simultaneously as independent variables. The squared semipartial correlation coefficient (sr^2) was calculated for each statistically significant independent variable to determine its unique contribution to the total explained variance of QoL in children and youth with RD. According to (Tabachnick & Fidell, 2013) recommendations, the absence of multicollinearity between independent variables was examined for each regression by checking the variance inflation factor (VIF) and tolerance index. Furthermore, normality, linearity, and homoscedasticity of residuals, independence of errors, and absence of outliers was ascertained. The adequacy of the number of participants was checked a posteriori, considering Green's recommendations (1991) based on the number of IVs inserted in the regression models.

Even though other associated conditions (i.e., physical disability, epilepsy, visual impairments, autism spectrum disorder, behavioral problems, hearing impairments, cerebral palsy, mental health problems, serious health problems, and attention deficit hyperactivity disorder), type of schooling and informant's sociodemographic characteristics (i.e., type of respondent, age, sex, frequency of contact and length of relation) were initially contemplated, these variables were ultimately excluded from the data analysis due to the small number of participants with these concurrent diagnoses or in mainstream education (appendix 2), and the presence of several informants for the majority (59 %) of the questionnaires. In addition, to assess whether consulting other informants influenced the results, a series of analysis of covariance tests (ANCOVA) were performed. Each analysis included the normalized total or domain scores as the dependent variable, the consultation status as the fixed factor (yes/no), and the same covariates used in the corresponding regression models.

3. Results

Neither univariate nor multivariate outliers were found in the KidsLife Scale scores or in the variables percentage of disability and age. Table 1 reports the descriptive statistics of the KidsLife Scale's domain and total scores. All skewness and kurtosis values were included within −1 and 1, indicating univariate normality. Similarly, multivariate normality was satisfied, given that the critical value calculated with the formula $k(k + 2)$, where k is the maximum number of the independent variables inserted in the regression model ($k = 6$), was higher than the mean of the Mahalanobis distances for the total QoL and its eight domains ($M = 7.20$). The observed scores of KidsLife Scale's total ranged from 177 to 360. The mean 303.59 ($SD = 35.15$), the median 309.5 and the mode 315 exceeded the theoretical midpoint of the scale (equal to 192). The theoretical midpoint score for all the KidsLife Scale's domains is 24. The most elevated scores were obtained in physical well-being ($M = 41.85$, $SD = 4.40$), and material wellbeing domains ($M = 41.13$, $SD = 5.81$), while the lowest scores were shown in social inclusion ($M = 31.68$, $SD = 6.10$) and self-determination domains ($M = 30.85$, $SD = 6.26$).

3.1. Relationships between factors and QoL: correlation analyses

Table 2 shows the correlation scores among variables and KidsLife Scale's domains and total score. Regarding individual factors, sex was statistically significantly positively associated with the total score, rights and interpersonal relationships domains. All these correlation coefficients had a small magnitude. Age did not show a significant association with any QoL domain or the total score.

Level of ID exhibited statistically significant negative correlations with the total score, and all domains except emotional wellbeing and physical wellbeing. The magnitude of the correlation coefficients was small for social inclusion and material wellbeing and medium for the total score and the other domains.

Level of support needs was statistically significantly negatively associated with the total score and all domains except emotional wellbeing. The magnitude of the correlation coefficient was medium in all cases except for social inclusion and physical wellbeing which had a small magnitude.

Table 1
Descriptive statistics for raw and composite score in QoL domains ($N = 114$).

	SI	SD	EW	PW	MW	RI	IR	PD	Total
n items	12	12	12	12	12	12	12	12	96
Mean	31.68	30.85	40.36	41.85	41.13	40.31	38.00	39.41	303.59
Median	31	31	41	43	42.5	40	38.5	40	309.5
Mode	31	35	45	44	48	45	45	36	315
SD	6.10	6.26	5.20	4.40	5.81	4.90	6.37	6.62	35.15
Skewness	−.00	−.11	−.42	−.59	−1.01	−.21	−.59	−.70	−.68
Kurtosis	.18	−.43	−.64	−.46	1.10	−.71	.14	.28	.39
Min.	14	16	26	30	18	29	17	16	177
Max.	45	45	48	48	48	48	48	48	360

Note. SI= social inclusion; SD= self-determination; EW= emotional wellbeing; PW= physical wellbeing; MW= material wellbeing; RI= rights; IR= interpersonal relationships; PD= personal development.

Table 2
Spearman's, Pearson's or Point-Biserial correlation coefficients among variables and QoL domains and total score ($N = 114$).

Variables	SI	SD	EW	PW	MW	RI	IR	PD	Total
Sex	.10	.16	.15	.13	.17	.23*	.24*	.16	.22*
Age	−.03	.14	.05	−.01	.11	.09	.02	.01	.04
Level of ID	−.21*	−.33***	−.06	−.13	−.27**	−.40***	−.46***	−.30***	−.37***
Level of support needs	−.27**	−.35***	−.09	−.19*	−.34***	−.38***	−.49***	−.34***	−.42***
Degree of dependency	−.21*	−.25**	−.01	−.13	−.26**	−.23*	−.28**	−.20*	−.26**
Percentage of disability[a]	−.23*	−.31***	−.09	−.10	−.21*	−.30**	−.37***	−.27**	−.33***
Size of the organization	.04	−.03	.01	.11	.17	.23*	.08	.04	.09

Note. SI= social inclusion; SD= self-determination; EW= emotional wellbeing; PW= physical wellbeing; MW= material wellbeing; RI= rights; IR= interpersonal relationships; PD= personal development.
[a] $n = 109$
*** $p \leq .001$; ** $p \leq .01$; * $p \leq .05$

Degree of dependency and percentage of disability exhibited significant negative correlations with both the total score and each of the domains except emotional wellbeing and physical wellbeing. The magnitudes were small except for the medium magnitude found for the correlations between percentage of disability and total score, self-determination, rights and interpersonal relationships.

Lasty, the environmental factor size of the organization showed a significant positive correlation with rights, with a small correlation coefficient. No individual or environmental factors were associated with emotional well-being.

3.2. Factors affecting QoL: multiple regression analysis

Statistically significant correlations with children's QoL were found for five individual factors (i.e., sex, percentage of disability, level of support needs, level of ID, degree of dependency) and one environmental factor (i.e., size of the organization); these variables were subsequently included as independent variables in the regression analyses. A maximum of six predictors were included simultaneously in the multiple regression analysis. Therefore, following Green (1991), the required minimum number of 47 participants for each regression model was fulfilled. No issues regarding variance regression prerequisite (i.e., tolerance index, normality, linearity, and homoscedasticity of residuals, independence of errors, and absence of outliers) were identified, except for VIF for level of support needs (i.e., values ranging from 2.19 to 2.24) and level of ID (i.e., values ranging from 2.49 to 2.55; thus slightly higher than the VIF cutoff value equal to 2) in all domains except emotional and physical wellbeing.

Table 3 shows the outcomes of the multiple regression analyses performed to examine factors affecting proxy perspectives on QoL. Regarding individual factors, sex was positively associated with the total score, rights and interpersonal relationships domains. More specifically, being male was associated with higher scores. Level of support needs was negatively associated with the total score and all domains except social inclusion. The percentage of disability was negatively associated with the total score, interpersonal relationships and personal development domains. Even though both level of ID and degree of dependency showed significant negative correlations with the total score and all the specific domains (except for physical and emotional wellbeing), they did not influence QoL when analyzed alongside with the other individual and environmental factors. Concerning the environmental factor size of the organization, it was positively associated only with the rights domain. Finally, as shown by the squared semi-partial correlation coefficients (sr^2), level of support needs was the independent variable that had the strongest effect on the total score and almost all QoL domains, with values between .03 and .10. Sex, percentage of disability, and size of the organization affected specific QoL domains, with squared semi-partial correlation coefficients ranging from .03 to .06.

3.3. Potential influence of informant assistance on QoL scores: ANCOVA analysis

Table 4 shows the outcomes of ANCOVA analyses conducted to examine the potential influence of consulting other informants on the total and domain scores. Across all analyses, the effect of requiring assistance was not statistically significant ($p > .05$).

4. Discussion

Despite the impact of RDs, QoL of children and youth with RDs remains under-researched. The scarce studies that exist on the QoL of people with RDs focus primarily on the narrower construct of health-related QoL. To address this gap, the goals of this study were to analyze individual QoL across the eight domains for children and youth with RDs and ID according to proxy perspectives and to examine the impact of several personal and contextual variables on their QoL scores.

The first step was to examine the overall QoL scores. The mean, median and mode of the total individual QoL score were above the theoretical midpoint of the scale. These positive outcomes are in consonance with the only study focusing on assessing individual QoL in people with RDs (González et al., 2016). Nevertheless, HrQoL studies had found that the population with RDs scores lower on HrQoL compared to other common chronic disease groups and the general population (Bogart et al., 2022; Chevreul et al., 2016; Chevreul et al., 2015; Efthymiadou et al., 2018; Johansen et al., 2013; Kodra et al., 2020; López-Bastida et al., 2016b; Ng et al., 2022; Wiegand-Grefe et al., 2022). Therefore, without a comparison group, we cannot determine whether, despite these favorable results, the individual QoL of this group is lower than that of their peers with other chronic conditions or without disabilities.

Table 3
Standard multiple regressions of factors on third-party perspectives on QOL domains and total score.

	SI (n = 109)		SD (n = 109)		PW (n = 114)		MW (n = 109)		RI (n = 109)		IR (n = 109)		PD (n = 109)		Total score (n = 109)	
Factors	β	sr^2	β	sr^2	β	sr^2	β	sr^2	β	sr^2	β	sr^2	β	sr^2	β	sr^2
Sex									.23**	.05	.23**	.05			.25**	.06
Level of ID	.08		−.01				.09		−.06		.05		.07		.15	
Level of support needs	−.27		−.27*	.03	−.20*	.04	−.29*	.04	−.30*	.04	−.48***	.10	−.28*	.04	−.45***	.09
Degree of dependency	−.04		−.01				−.06		.12		.15		.04		.10	
Percentage of disability	−.16		−.20				−.12		−.15		−.28**	.05	−.23*	.03	−.26*	.04
Size of the organization									.21*	.04						
Adjusted R^2	.07		.13		.03		.07		.24		.30		.09		.23	
F	2.97*		5.08***		4.66*		3.09*		6.55***		10.29***		3.62**		7.33***	

Note. SI= social inclusion; SD= self-determination; PW= physical wellbeing; MW= material wellbeing; RI= rights; IR= interpersonal relationships; PD= personal development.
*** $p \leq .001$; ** $p \leq .01$; * $p \leq .05$.

Table 4
Effect of consulting other informants on the total and domain scores (ANCOVA).

		SI (n = 109)	SD (n = 109)	PW (n = 114)	MW (n = 109)	RI (n = 109)	IR (n = 109)	PD (n = 109)	Total score (n = 109)
M (SD)	Yes	32.6 (6.24)	30.86 (5.76)	42.11(4.41)	40.63 (5.79)	40.15 (5.23)	38.28 (5.91)	39.68 (5.90)	305.01 (34.30)
	No	30.40 (5.72)	30.83 (6.97)	41.47(4.40)	41.83 (5.84)	40.53 (4.43)	37.59 (7.01)	39.02 (7.58)	301.55 (36.60)
F		3.35	.15	.857	2.55	.02	.27	.21	.28
Partial η^2		.03	.00	.01	.02	.00	.00	.00	.00

Note. SI= social inclusion; SD= self-determination; PW= physical wellbeing; MW= material wellbeing; RI= rights; IR= interpersonal relationships; PD= personal development.
Each ANCOVA tested the effect of consulting other informant to complete the scale (Yes/No) on the total and domain scores while statistically controlling for covariates previously included in the corresponding regression model.
Partial η^2 = partial eta squared.
*** $p \leq .001$; ** $p \leq .01$; * $p \leq .05$.

A closer examination of the results by QoL domain reveals that, similar to other studies conducted with this and other groups with disabilities (González et al., 2016; L. Morán et al., 2022; M.L. Morán et al., 2019a; Verdugo et al., 2019), the highest scores were obtained for material well-being and physical well-being. The nature of the RDs, in which much of the effort is dedicated to the treatment of physical health problems and the predominance of the welfare model (Jaeger et al., 2015), which has focused on reducing the impact of impairments and providing material resources, may account for these results.

Furthermore, in line with earlier studies with this and other groups with disabilities (e.g., González et al., 2016; Leonard et al., 2022; Morán et al., 2022, 2024; Morán & Gómez, 2025; Verdugo et al., 2019; Wiegand-Grefe et al., 2022), the lowest scores were found in social inclusion and self-determination. A possible reason for this phenomenon is that the physical needs derived from the disability of children with RDs may take better care of than their socioeducational needs (Johansen et al., 2013), despite the fact that social participation is recognized as a protective factor for the QoL of people with RDs (Chen et al., 2022). Besides, social exclusion and a lack of social and emotional support are commonly described among children with RDs and their parents (e.g., Rao et al., 2024; Von der Lippe et al., 2017; Witt et al., 2023), and particularly for more visible RDs (Ng et al., 2022). Additionally, children with RDs and ID find it difficult to negotiate the expectation of being independent and capable of their own decision making (Belzer et al., 2022), even though there is plenty of evidence demonstrating the capacity of people with disability to develop self-determination skills with the adequate supports and the positive impact of these skills on their QoL and adult life (e.g., Hagiwara et al., 2020; Morán et al., 2021; Mumbardó-Adam et al., 2024; Wehmeyer, 2020). These low results in social inclusion and self-determination highlight the crucial domains for the planning of supports and services aimed at promoting the QoL of children and young people with RDs as well as the need of strengthening social awareness regarding RD to reduce stigma and enable inclusion (Witt et al., 2023).

The second step was to examine the impact of several variables on the QoL scores of children and youth with RDs and ID. Regarding individual variables, being male was associated with higher levels of rights, interpersonal relationships and QoL, in consonance with Bogart and Irvin (2017) and contrary to several previous studies (Gao et al., 2020; González et al., 2016; Xu et al., 2023). Different roles by gender might be part of the explanation for these results (Morán et al., 2019b). In this sense, women with disabilities are affected by double discrimination, which arises from the intersection between the stigma of being a woman and the stigma of disability (Senaldi et al., 2022). In any case, the lower scores obtained by girls in our study highlight the need to incorporate a gender perspective in the strategies designed to improve the rights, interpersonal relationships and QoL of children and youth with RDs and ID with the aim of ensuring equal opportunities for boys and girls. Accordingly, programs and policies should not only provide specific training for support providers, but also actively involve girls and remain mindful of the gender stereotypes that can impact their development and well-being. Furthermore, age did not have a significant impact on the total QoL and domain scores, in accord with González et al. (2016) and Ng et al. (2022), but contrary to other studies that indicate lower scores among older participants (Bogart & Irvin, 2017; Xu et al., 2023).

As far as we know, no previous study has examined the relation between individual QoL in youth with RDs and percentage of disability. More specifically, percentage of disability is a legally determined measure of the degree of disability in Spain. It is an assessment conducted by an interdisciplinary team. Disability is officially recognized once it reaches a minimum of 33 %. In this sample, the percentage of disability showed a negative association with the total score, interpersonal relationships and personal development domains. Additionally, level of support needs was negatively associated with the total score and all domains except social inclusion in consonance with González et al. (2016). In addition, the strongest association was that of the level of support needs in interpersonal relationships. These results were expected taking into account that the personal outcomes of people with most significant disabilities rely on opportunities that are often not offered to this sector of the population. To address this limitation, organizations need to implement strategies that promote the transformation of services toward inclusive models centered on all people with ID, especially those with greater support needs, by means of incorporating assistive devices and approaches such as active support, person-centered planning, supported decision-making or supported employment (Beadle-Brown et al., 2016; Shogren et al., 2017; Verdugo & Navas, 2017).

Regarding environmental variables, to the best of our knowledge, no previous study had examined the relationship among individual QoL in youth with RDs and size of the organization either. Specifically, the size of the organization was positively associated with rights in consonance with prior literature conducted with youth with other disabilities (Morán et al., 2022, 2024). A possible reason for this phenomenon is that larger organizations may have more resources enabling them to expand their services and therefore,

foster greater improvements in their users' rights. In this sense, it is crucial to consider that resources should not solely depend on the size of organizations or the number of users, but rather on the specific support needs of individuals. Encouraging collaborations between organizations and leveraging local resources could also facilitate for the smaller ones the access to specialized resources they might not have on their own as well as the chance to receive technical support. However, it is worth noting that the size of the organization in our study corresponds to the total number of participants who receive support from the organization, but they may be distributed in smaller divisions or centers within the organization, and our study did not incorporate further detail.

The potential influence of consulting other informants on the total and domain scores was also examined. Across all analyses, the effect of requiring assistance was not statistically significant indicating that the need to consult someone else did not significantly affect the scores, even when controlling for other relevant variables.

Some limitations must be noted. First, this study is based on a small convenience sample since recruitment of this group is particularly challenging due to the rarity of the diseases and the lack of an official census in Spain to allow for random sampling. Therefore, results must be interpreted with caution. Second, the scales were filled in by third parties. Despite the importance of including the perspectives of this group on their own QoL (Balboni et al., 2013; Paz-Lourido et al., 2020; Schalock et al., 2021), children who are very young or have severe conditions may not be able to complete the assessment directly (Gao et al., 2020). Future studies could benefit from investigating ways to reliably obtain the views of children with RDs and higher severity levels, incorporating both proxy measures and the child's perspective (Benedetto et al., 2023). Third, this study was cross-sectional, so association between predictors variables and QoL scores cannot be interpreted as inferring causality. A longitudinal design approach could also be considered for future studies. Neither it was exhaustive in the inclusion of other possible predictors of QoL, such as the type of schooling, the presence of other associated conditions or respondent's sociodemographic data (Bacherini et al., 2021; Balboni et al., 2021; Linertová et al., 2019; Menardo et al., 2017), which may have led to higher values of explained variance. Hence, there is a need for increasing the range of variables studied, focusing particularly on those factors that can be modified to enhance QoL (Balboni et al., 2020). Lastly, this study was conducted in one single country, further research should expand to the international context to allow a more comprehensive understanding of the individual QoL of the RDs population (Rao et al., 2024).

5. Conclusion

This study has shed light into the strengths and challenges faced by children and youth with RDs and ID across the different QoL domains. Additionally, it offers insights into factors that influence QoL. Our results show that the overall QoL scores of children and youth with RDs and ID were generally positive. However, self-determination and social inclusion obtained the weakest scores and therefore are relevant for future research and priority areas in provision of support aimed at the QoL enhancement of this sector of the population. Despite the role of other variables that need to be further researched, sex, level of support needs, percentage of disability, and size of the organization had a significant impact on their QoL. It is important, therefore, that these factors are considered in the provision of supports and for the development of professional practices organizational, approaches and policy initiatives intended to promote the QoL of children and youth with RDs and ID.

CRediT authorship contribution statement

M. Lucía Morán: Writing – review & editing, Writing – original draft, Validation, Methodology, Investigation, Formal analysis, Data curation, Conceptualization. **Asunción Monsalve:** Writing – review & editing, Resources, Funding acquisition, Investigation. **Yolanda Fontanil:** Writing – review & editing, Resources, Funding acquisition, Investigation. **Alice Bacherini:** Writing – review & editing, Methodology, Formal analysis. **Giulia Balboni:** Writing – review & editing, Supervision, Methodology, Formal analysis. **Laura E. Gómez:** Writing – review & editing, Writing – original draft, Supervision, Resources, Project administration, Funding acquisition, Methodology, Conceptualization, Investigation.

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Consent to participate

Informed consent was obtained from all individual participants included in the study.

Declaration of Competing Interest

The authors report no conflict of interest.

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Appendix 1. Frequency and percentage of RD ($N = 114$)

Disorders	Frequency (%)
Fragile X syndrome	21 (18 %)
West syndrome	14 (12 %)
Rett syndrome	9 (8 %)
Angelman syndrome	8 (7 %)
Tuberous sclerosis	6 (5 %)
Cri du Chat syndrome	4 (4 %)
Williams syndrome	4 (4 %)
Dandy-Walker syndrome	3 (3 %)
Idic (15) syndrome	3 (3 %)
Lennox-Gastaut syndrome	3 (3 %)
Opitz syndrome	3 (3 %)
Agenesis of the Corpus Callosum	2 (2 %)
Arnold-Chiari syndrome	2 (2 %)
DiGeorge syndrome	2 (2 %)
Dravet syndrome	2 (2 %)
Mosaicism	2 (2 %)
Smith-Magenis syndrome	2 (2 %)
Steinert syndrome (myotonic dystrophy)	2 (2 %)
Turner syndrome	2 (2 %)
Cardio-facio-cutaneous syndrome	1 (1 %)
Coffin-Lowry syndrome	1 (1 %)
Coffin-Siris syndrome	1 (1 %)
Chromosopathy 22q13	1 (1 %)
Chromosomal alteration of chromosome 8	1 (1 %)
Chromosomal alteration of chromosome 10	1 (1 %)
Holoprosencephaly	1 (1 %)
Kabuki syndrome	1 (1 %)
Lissencephaly	1 (1 %)
Maroteaux-Lamy syndrome	1 (1 %)
Microcephalic osteodysplastic primordial dwarfism type 1	1 (1 %)
Mowat-Wilson syndrome	1 (1 %)
Pallister-Killian syndrome	1 (1 %)
Partial monosomy of chromosome 13	1 (1 %)
Prader-Willi syndrome	1 (1 %)
Pediatric Autoimmune Neuropsychiatric Disorders Associated with Streptococcal Infections (PANDAS)	1 (1 %)
Trisomy 2	1 (1 %)
Trisomy 8	1 (1 %)
Unspecified autosomal chromosopathy	6 (5 %)
Unspecified chromosopathy	3 (3 %)

Appendix 2. Sociodemographic characteristics of participants with RD ($N = 114$)

	Frequency (%)
Level of ID	
Mild	6 (5 %)
Moderate	35 (31 %)
Severe	54 (47 %)
Profound	19 (17 %)
Level of support needs	
Limited	4 (4 %)
Intermittent	21 (18 %)
Extensive	30 (26 %)
Generalized	59 (52 %)
Percentage of disability	
Mean (SD)	69.21 (16.56)
Range	33–99
Degree of dependency	
Grade I (moderate)	13 (11 %)
Grade II (severe)	32 (28 %)
Grade III (maximum)	69 (61 %)
Other associated conditions	
Physical disability in the lower extremities	35 (31 %)
Epilepsy	29 (25 %)
Physical disability in the upper extremities	23 (20 %)
Visual impairments	20 (18 %)

(*continued on next page*)

(*continued*)

	Frequency (%)
Autism spectrum disorders	19 (17 %)
Behavioral problems	17 (15 %)
Hearing impairments	11 (10 %)
Cerebral palsy	9 (8 %)
Mental health/emotional problems	8 (7 %)
Serious health problems	7 (6 %)
Attention deficit hyperactivity disorder	5 (4 %)
Type of school	
Special	89 (78 %)
Mainstream	15 (13 %)
Both	10 (9 %)
Size of the organization	
Less than 51 users	44 (39 %)
51–100	34 (30 %)
101–200	14 (12 %)
More than 200 users	22 (19 %)

Data availability

Data will be available upon reasonable request.

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