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Instruments for the assessment of quality of life in children and adolescents with Down syndrome: a scoping review

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Abstract

The construct of quality of life (QoL) includes aspects of health and well-being of people. Down syndrome [DS] or trisomy 21 is one of the most common congenital anomalies. DS is characterized by motor and cognitive alterations that affect health and QOL of both the child and caregiver.

In pediatrics, there are various instruments to assess Health-Related Quality of Life (HRQoL) and QoL. The advantage of these instruments is that they can be implemented in any type of disease and population in general. However, they may have certain disadvantages, such as the difficulty in evaluating specific aspects of each disease or condition related to Down syndrome. The aim of this study was to identify 1: instruments used to assess quality of life in children with Down syndrome. 2: psychometrics properties of instruments validated in children with Down syndrome to assess quality of life.

Methods A Scoping review was conducted to identify instruments used in children and adolescents with Down syndrome, and a second systematic searched psychometric properties of these instruments. The electronic databases PubMed, Embase, Epistemonikos and other sources were explored with a search strategy that included keywords such as "Down syndrome," "Quality of life" or "Life Quality," "Health-Related Quality of Life" and psychometrics properties. The quality of the included studies was evaluated using the COSMIN (Based Standards for the Selection of Health Measurement Instruments) methodology.

Results Twenty-seven studies were selected that used twelve instruments to evaluate quality of life in children or adolescents with Down syndrome. Two of the twelve evaluated quality of life and ten health-related quality of life. In the second search, ten studies reported the psychometric properties of six instruments evaluated in minors with Down Syndrome.

Conclusion There is limited information available regarding the psychometric properties of instruments used to assess quality of life, particularly health-related quality of life. Commonly employed instruments in this area include the PedsQL 4.0 and KIDSCREEN. Notably, while the PedsQL 4.0 lacks specific evaluation in children with DS, data from KIDSCREEN assessments are inconsistently reported. Rigorous evaluation of the performance of Kidslife and

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Kidslife Down in clinical settings is necessary, or the development of new instruments tailored for children with DS is warranted to comprehensively assess quality of life in clinical settings.

Keywords Quality of life, Health-related quality of life, Down syndrome, Psychometric properties, Children, Adolescents

Introduction

The concept of quality of life (QoL) is becoming increasingly relevant given its potential for evaluating outcomes of interventions and service delivery, as well as the possibility of finding a common language across disciplines. According to the World Health Organization, QoL is considered as "the perception of an individual of his or her place in life within the cultural context and value system in which he or she lives and with respect to his or her goals, expectations, norms, and concerns [1].

The construct of QoL is composed of objective and subjective dimensions, and includes aspects related to health, feelings of satisfaction and well-being that occur in relation to life experiences and circumstances [2]. Quality of life is influenced positively or negatively by internal and external factors such as each person's perceptions regarding their life, cultural contexts, previous experiences, personal values, and aspirations, aspects that are difficult to measure [3].

Down syndrome (DS) is caused by a trisomy 21 (partial or total), this additional genetic material alters the course of development. DS has a higher risk of associated diseases and challenges in different areas of development and cognition that affect the QoL of both the child and the caregiver [3–7]. The estimated incidence of DS worldwide is 10 in every 10,000 newborns [8]. In Colombia the incidence is estimated between 16 and 18 per 10,000 live births and ranks third among congenital disorders [9].

Children with DS have the same physical, psychological, social, and learning needs as other children; however, due to the alterations accompanying this condition, they also do have some special needs, which must be addressed with effective health care and education to impact their QoL. Some studies show that in general, the QoL of children with DS is lower than those without this condition, although there are variations in some dimensions, showing low levels of physical well-being but high levels of emotional well-being [10, 11].

Currently, instruments are available to assess the QoL of children and adolescents, mainly those with chronic diseases. Children with DS exhibit different characteristics that could make them prone to have a reduced QoL when compared with a child having other chronic disease. Additionally, they present a higher risk of developing multiple comorbidities and impairments in the dimensions of physical health, social functions, and problem-solving [4]. They present various degrees of disability and have communication and comprehension difficulties [10], as well as emotional and behavioral affectations [11] that can alter interpersonal relationships and their functional performance in school activities, among others [12].

It is very important to assess QoL in this population since it allows the identification of how different areas of their lives may be affected by the condition and by the effect of the therapeutic interventions they receive throughout their life. Considering that DS is a genetic condition that requests long term care, outcomes such as QoL become important since interventions are aimed at improving the way they live and not at curing the disease [13]. However, there is no consensus on the construct with which this outcome has been assessed in this population and which instruments have been used in pediatrics considering the specific characteristics of the DS population [4].

Quality of life can be assessed as a multidimensional construct that includes aspects like emotional, economic, and physical well-being, as well as interpersonal relationships, social inclusion, personal development, self-determination, and rights, integrated by Schalock and Verdugo in their QoL model [14–17]. Its assessment provides useful information to professionals working in health, social and educational organizations for the well-being of people with DS in different service and support delivery contexts [18]. When evaluating QoL in people with DS it is necessary to consider the use of subjective and objective measures in order to provide a holistic assessment. In addition, consider the use of proxy or self-reported data due to difficulties related to communication and cognitive problems, among others [18, 19].

In pediatrics, there are various instruments to assess health-related quality of life (HRQoL), such as the Pediatric Quality of Life Inventory (PedsQL), TNO-AZL Children's Quality of Life (TACQOL), the Child Health Questionnaire (CHQ-PF50), and the European Quality of Life-5 Dimensions-YOUTH (EQ-5D-Y). The advantage of these instruments is that they can be implemented in people with any type of disease and population in general since they allow the comparison of life perceptions before and after any health intervention. However, they may have certain disadvantages, such as the difficulty in evaluating the specific aspects of each disease or condition, which are important for the patient or his/her caregivers [20]. The health care of the DS population benefits with information from QoL assessment with specific valid and reliable instruments to guide clinical decisions

and interventions according to their needs. The aim of this study, therefore, was to identify 1: instruments used to assess quality of life in children with Down Syndrome. 2: psychometrics properties of instruments validated in children with Down syndrome to assess quality of life.

Methods

The objectives, inclusion criteria and methods for this scoping review were prespecified and published in a protocol with Open Science Framework [21] and it is available in https://doi.org/10.17605/OSF.IO/V2YX8. We used previously established scoping review methodology to guide our study methods and applied the Preferred Reporting Items for Systematic Reviews and Meta-Analyses for Scoping Reviews (PRISMA-Scr) [22, 23].

Design An exploratory literature search for the present work showed that there are few valid instruments in people with DS to assess their quality of life. Contradictorily, that same search also showed that quality of life is a very important outcome in this population due to the number of publications that evaluate quality of life [24, 25].

Eligibility criteria

Population: Population with DS under 21 yo.

Outcome: Quality of life or health-related quality of life.

Design: For inclusion, studies identified in the literature search that described the evaluation of the QoL in children and adolescents with DS to extract the names of the instruments used. In addition, all the studies of creation, validation, and evaluation of the psychometric properties of instruments validated in children under 21 yo with DS were included.

Search and identification of studies

Two researchers independently conducted a literature search in PubMed, Embase and Epistemonikos during the month of February 2024 to identify QoL instruments used in children or adolescents with DS. We employed several combinations of keywords and MeSH [26] search terms in each electronic search engine (Appendix 1) The first group of search terms consisted of synonyms for DS. The second group of search terms included quality of life.

A second systematic search was carried out in the same database including google academic, where we reviewed the first 100 results to identify the psychometric properties of the instruments found in the first systematic search. In this systematic search, we used terms related to the names and abbreviations of the identified instruments and terms related to psychometrics properties according to the Consensus Based Standards for the Selection of Health Measurement Instruments (COSMIN) filter proposed to identify studies evaluating psychometric properties (Appendix 2).

Two authors (PM and MD) independently screened article abstracts to identify potentially relevant articles. Full texts of these articles were obtained, and the authors independently reviewed the texts. The authors discussed disagreements to reach a consensus on the final sample. The Rayyan platform [www.rayyan.ai], an online tool specifically designed to enhance the efficiency and thoroughness of article screening and review, was used to screen reports and identify disagreements [27].

Extraction of information

Two researchers extracted information from the studies identified in the first search. The information extracted included the names of the instruments used to assess the quality of life of children with DS. From the second search, the psychometric properties of the identified instruments were extracted using a structured template to extract information on the characteristics of each study, including the study's purpose, participants, and the QoL dimensions among others.

Evaluation of the psychometric properties

In this study, the COSMIN methodology [28] criteria were applied to identify the psychometric properties of the QoL instruments used in DS. This method identifies the quality conditions of the instruments that report patient outcome measurements. The method is used for evaluation purposes based on the quality criteria of the evaluated psychometric properties and establishes quality standards from the design and statistical methods.

The COSMIN instrument has nine dimensions: structural, cross-cultural, measurement invariance, criterion, construct, reliability, internal consistency, measurement error, and sensitivity to change [28]. The evaluation of each psychometric property is contrasted with the quality criteria established in COSMIN on a three-level ordinal scale: sufficient [+], insufficient [-], and undetermined [?], as shown in Table 1 [28, 29].

Two researchers independently applied the COSMIN criteria to the psychometric properties of the QoL instruments in DS and performed data extraction. The evaluation was performed based on consensus between the two evaluators, if consensus was not reached; a third evaluator defined the rating of the quality of the instrument.

Results

Literature search and selection

In the first search, 759 articles were identified, 125 duplicate articles were eliminated, and 592 articles were excluded from the review based on their title and abstract. Finally, twenty-seven studies, including twelve instruments, were selected for the review (Fig. 1).

Table 1 COSMIN criteria

Properties	Grade	Criteria
Structural validity	+	Classical theory: Confirmatory factor analysis: comparative fit index or Tucker–Lewis index > 0.95
	?	Classical theory: no information to report "+" Item response theory/Rasch: Unidentified fit model
	-	Criteria for "+" not known
Cross-cultural validity and measurement invariance	+	No significant differences among the group factors [such as age, gender, and language] were found in the group multiple factor analysis, or no differential item functioning was found across the group factors.
	?	Group factor analysis or differential item functioning analysis was not performed.
	-	Significant differences were found between factor group or item differential functioning.
Criterion validity	+	Gold Standard correlation ≥ 0.70 or area under the curve ≥ 0.7
	?	The information to state that it is positive is not reported.
	-	Gold Standard correlation < 0.70 or area under the curve < 0.70
Construct validity [hypothesis testing]	+	The result is consistent with the hypothesis.
	?	There are no defined hypotheses.
	-	The result is not in accordance with the hypothesis.
Reliability	+	ICC or Kappa ≥ 0.70
	?	ICC or Kappa are not reported.
	-	ICC or Kappa < 0.70.
Internal consistency	+	At least low evidence for sufficient structural validity, and Cronbach's alpha ≥ 0.70.
	?	It does not meet the criteria for determining at least low evidence for structural validity.
	-	At least low evidence for sufficient structural validity, and Cronbach's alpha < 0.70.
Measuring error	+	The minimum detectable change or limits of agreement < minimum significant change
	?	The minimum major change is not defined
	-	The minimum detectable change or limits of agreement > minimum significant change
Sensitivity to change	+	The result agrees with the hypothesis, or the area under the curve is ≥ 0.70
	?	The hypothesis is not defined.
	-	The result agrees with the hypothesis, or the area under the curve is ≥ 0.70.

Taken from: Prinsen C et al. Qual Life Res Int J Qual Life Asp Treat Care Rehabil. 2018;27 [5]:1147-57

In the second search, 117 articles were identified, three duplicate articles were removed, and finally ten studies were included in the review (Fig. 2).

Instruments used to assess quality of life in children and adolescents with Down syndrome

Twelve instruments were found: PedsQL [30–38]; TACQoL [39, 40]; Preschool Quality of Life (TAPQoL) [40]; Child Health Questionnarie PF 50 (CHQ-PF 50) [41]; Quality of Life Inventory-Disability (QI-Disability) [42]; Kidscreen-52 [43]; Kidscreen-27 [44–46]; Health Utilities Index (HUI) [47]; Personal Outcomes Scale [48]; The 5-level EQ-5D version (EQ-5D-5 L) [49, 50]; Kidslife-Down [51]; and Kidslife [52–55].

Six studies were self-reports from children and their parents [35, 39, 45, 48, 50, 51] and twenty one were from parents only (proxy) [30–34, 36–38, 40–44, 46, 47, 49, 52–56]. The age range assessed by the instruments was 2–21 yo. The temporary framework used was either the week before or the last month [29, 33–37] (Table 2).

Of the twelve instruments identified, Kidslife and Kidslife-Down evaluated quality of life in a general concept, the other instruments evaluated HRQoL. The Kidslife-Down questionnaire is the only one, which assess QoL in children and adolescents with DS, specifically (Table 2).

Characteristics of the identified instruments

The Pediatric Quality of Life Inventory (PedsQL) comprises four versions tailored to children or adolescents across different age groups. Specifically designed for individuals aged 5 to 18 years old, the inventory includes separate versions to be completed by the child and their parent. Participants are instructed to assess the extent to which each item has been problematic over the past month. Responses are recorded on a 5-point scale ranging from 0 (never a problem) to 4 (almost always a problem). Notably, items undergo reverse-scoring and subsequent linear transformation to a 0–100 scale (0=100, 1=75, 2=50, 3=25, 4=0) ensuring that higher scores correspond to better QoL. The instrument evaluates four primary dimensions of quality of life in children and adolescents: physical, emotional, social, and school functioning [30–38].

The TACQoL, originating from the Netherlands, is designed for children between the ages of six and fifteen.

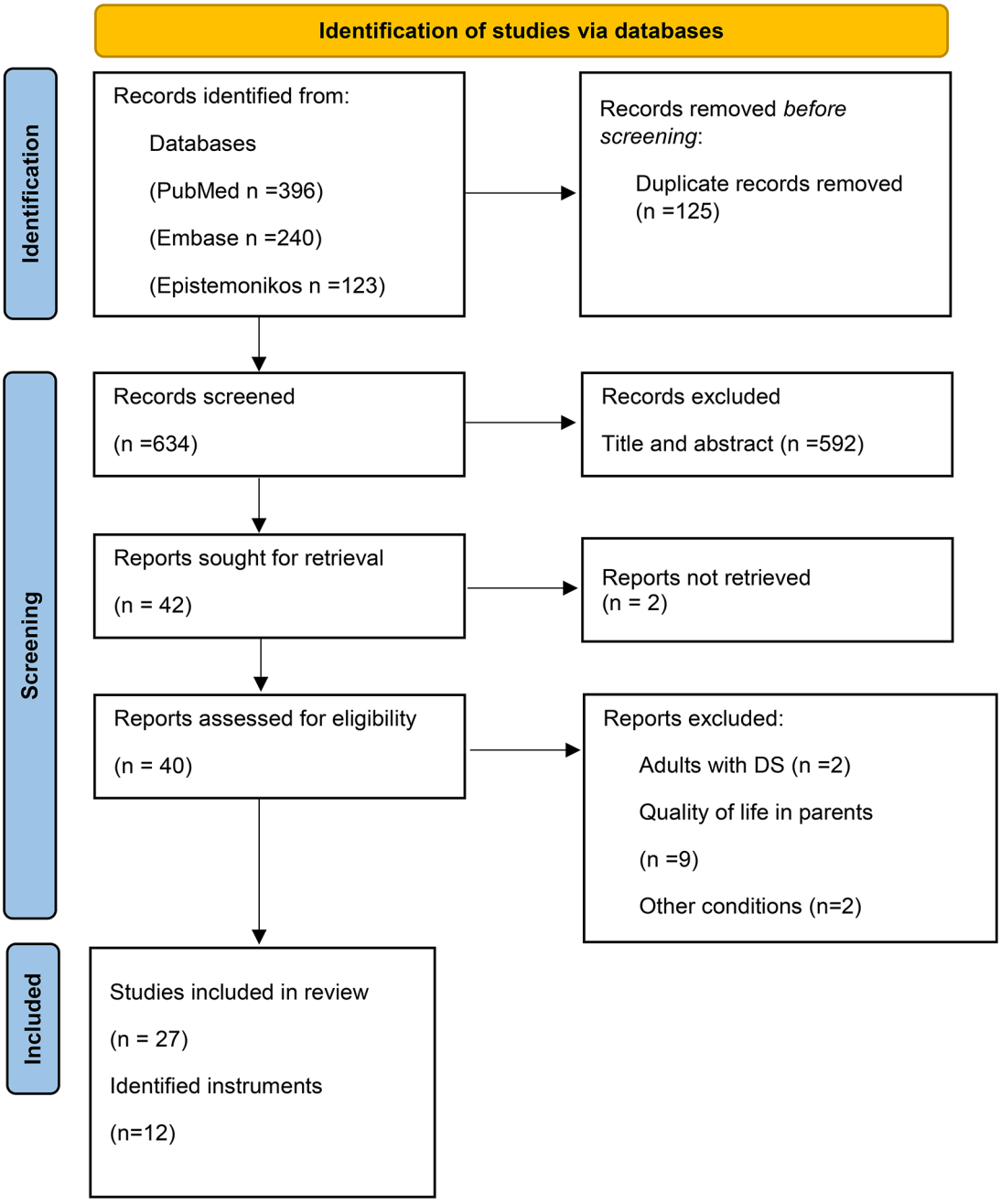

Fig. 1 First search flow Diagram. From: Page MJ, McKenzie JE, Bossuyt PM, Boutron I, Hoffmann TC, Mulrow CD, et al. The PRISMA 2020 statement: an updated guideline for reporting systematic reviews. BMJ 2021;372:n71. doi: https://doi.org/10.1136/bmj.n71

This instrument evaluates functional difficulties, considering the child's emotional responses to these challenges. Comprising 56 items, the questionnaire encompasses seven scales: physical complaints, gross motor skills, autonomy, cognitive functioning, social functioning, positive emotions, and negative emotions [39, 40].

Preschool Quality of Life (TAPQoL) questionnaire is a comprehensive, multidimensional tool consisting of 43 items designed to assess Health-Related Quality of Life (HRQoL) across four primary domains, further divided into 12 subdomains. These domains encompass physical functioning (including sleeping patterns, appetite, and various physiological issues) social functioning (addressing problem behaviors) cognitive functioning (evaluating communication skills) and emotional functioning (assessing anxiety levels, positive mood, and liveliness) The number of items per scale varies from three to seven. Rather than providing an overall summary score, the questionnaire yields domain-specific scores, each ranging

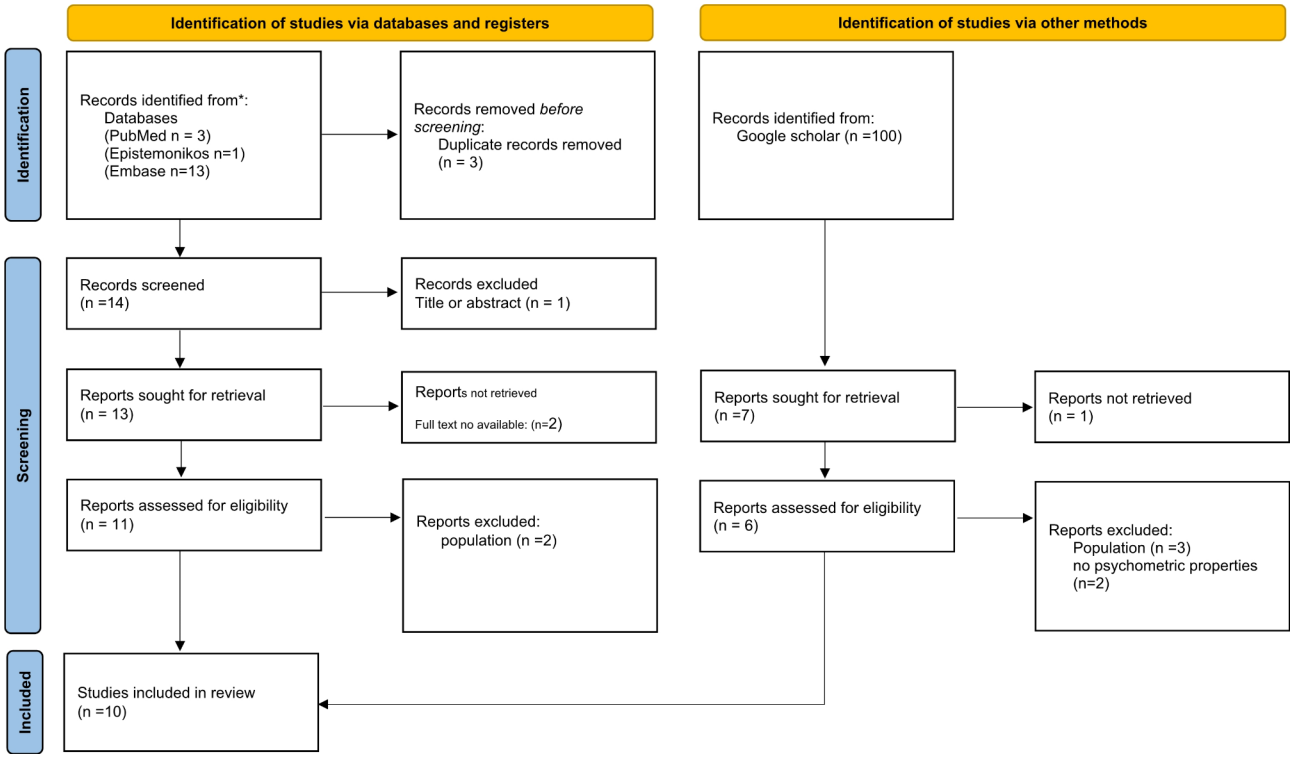

Fig. 2 Second search flow Diagram. From: Page MJ, McKenzie JE, Bossuyt PM, Boutron I, Hoffmann TC, Mulrow CD, et al. The PRISMA 2020 statement: an updated guideline for reporting systematic reviews. BMJ 2021;372:n71. doi: https://doi.org/10.1136/bmj.n71. For more information, visit: http://www.prisma-statement.org/

from 0 to 100, where higher scores denote enhanced QoL [40].

Child Health Questionnarie PF 50 (CHQ-PF 50) is a widely utilized 50-item parent-report survey intended for evaluating the physical and psychosocial well-being of children. It has been extensively employed in assessing QoL across various patient populations, including individuals with cancer, psychiatric disorders, and severe developmental disabilities. The questionnaire assesses 14 domains encompassing both physical and psychosocial aspects, including general health perceptions, physical functioning, role/social physical functioning, bodily pain, role/social emotional functioning, role/social behavioral functioning, parent impact-time, parent impact-emotional, self-esteem, psychosocial health, behavior, family activities, family cohesion, and change in health. Scores from these scales are transformed onto a 0-100 scale, where 0 represents the poorest possible health state and 100 signifies the optimal health state. Additionally, the individual scale scores are amalgamated to generate two summary component scores: the physical functioning and psychosocial health summary scores. These summary scores are then converted into norm-referenced T-scores, with a mean of 50 and a standard deviation of 10, facilitating comparison across different populations [41].

Quality of Life Inventory-Disability (QI-Disability) The parent-report QI-Disability questionnaire is a 32-item measure designed to assess the QoL of children with intellectual disabilities. The questionnaire encompasses six domains: Social Interaction [7 items], Positive Emotions [4 items], Negative Emotions [7 items], Physical Health [4 items], Leisure and the Outdoors [5 items], and Independence [5 items]. Caregivers rate items on a 5-point Likert scale, reflecting their observations of the child's well-being and enjoyment of life over the preceding month. Responses are linearly transformed to a scale ranging from 0 to 100, with higher scores indicating better QoL. Domain scores are derived by averaging item scores, while total scores are calculated by averaging domain scores [42].

Kidscreen-52 has ten domains measures self-perception of the subject. The KIDSCREEN-52 HRQoL questionnaire is a comprehensive instrument designed to assess HRQoL in children and adolescents. Consisting of 52 items, this parent-reported survey evaluates various aspects of well-being across multiple domains. These domains include physical well-being, psychological well-being, autonomy and parent relation, peers and social support, and school environment. Additionally, the questionnaire covers aspects such as financial resources and health and overall satisfaction with life. Responses are

Table 2 Description of the instruments evaluated

Instruments	Domains	Report	Age of the target population [years]	Number of Items	Temporary frame
TNO-AZL Questionnaire for Children's Health-related Quality Of Life [TACQOL] [39, 40, 56]	Physical complaints motor functioning [physical] Autonomous functioning [daily life] Social functioning [social] Cognitive functioning Positive and negative moods	Parents Children	5–15 8–15	56	Last month
PedsQL 4.0 [30–38]	Physical Emotional Social College	Children Parents	2–18	23	Last month
TAPQoL [40]	**Physical functioning** Seeping patterns Appetite Various physiological issues **Social functioning** Addressing problem behaviors **Cognitive functioning** Evaluating communication skills, **Emotional functioning** Assessing anxiety levels Positive mood Liveliness	Parents or caregivers	1.5 to 6	43	Not mentioned
Kidscreen-52 [43]	Physical wellbeing Psychological Wellbeing Mood Self-Perception Autonomy Relationship with parents and family life Friends and social support School environment Social acceptance [Bullying] Financial resources	Children Parents	8–18	52	Last week
Kidscreen-27 [44–46]	Physical wellbeing Psychological wellbeing Autonomy and parent relationship Friends and social support School environment	Children	8–18	27	Last week
Health Utilities Index Mark 2 [HUI2] and HUI3 [47]	**HUI2** Sensation Mobility Emotion Cognition Self-Care Pain Fertility **HUI3** Vision Hearing Speech Ambulation Dexterity Emotion Cognition Pain	Children or parents or caregivers	5 and above	15 and 16 respectively	Not mentioned

Table 2 (continued)

Instruments	Domains	Report	Age of the target population [years]	Number of Items	Temporary frame
CHQ-PF50 [41]	Physical functioning role/social constraints- physical General health perceptions Pain/body discomfort Family activities Role/social limitations: emotional/behavioral [two domains] Impact of parents-time Impact of parents-emotion Self-esteem Mental health Behavior Family cohesion Health changes	Parents	5–18	50	Last month
QI-Disability [42]	Social Interaction Positive Emotions Negative Emotions Physical Health Leisure and the Outdoors Independence	Parents or caregivers	5–18	32	Past month
Personal Outcomes Scale [48]	Independence Social participation Wellbeing	Adults or caregivers	> 18	20 to 30 items	Not mentioned
EQ-5D-5 L [49, 50]	Mobility Self-care Usual activities Pain/discomfort Anxiety/depression	Caregivers	8 and 22	5	Not mentioned
Kidslife-Down [51]	Social inclusion Self-determination Emotional wellbeing Physical wellbeing Material wellbeing Rights Personal Development Interpersonal relationships	Parents or caregivers	4–21	96	Not mentioned
Kidslife [52–55]	Emotional Well-being Physical Well-being Material Well-being Personal Development Self-Determination Interpersonal Relations Social Inclusion Rights	Parents or caregivers	4–21	96	Not mentioned

collected on a Likert scale, with higher scores indicating better HRQoL. The KIDSCREEN-52 provides a valuable tool for capturing the multidimensional nature of children's quality of life and has been widely used in research and clinical settings [43].

Kidscreen-27 contains 27 statements that respond to the five domains, including physical and psychological well-being, autonomy and parental relations, social well-being and their peers, the school, and the learning environment [44–46].

The Health Utilities Index Mark 2 (HUI2) and Health Utilities Index Mark 3 (HUI3) are non-disease-specific indices applicable to individuals aged 5 and above, serving as independent yet complementary systems for measuring HRQoL. Each system comprises a comprehensive health status classification and a utility scoring component. The 15-item multiple-choice questionnaire allows for scoring subjects according to both HUI2 and HUI3. HUI2 characterizes an individual's functional health status based on 7 health dimensions (Sensation, Mobility, Emotion, Cognition, Self-Care, Pain, and Fertility), each distinguished by 3–5 descriptive levels. Conversely, HUI3 assesses health using 8 single dimensions (Vision, Hearing, Speech, Ambulation, Dexterity, Emotion, Cognition,

and Pain) with each attribute having 5–6 descriptive levels. Multi-attribute utility scores, which serve as numerical measurements for HRQoL, are obtained through a multiplicative scoring algorithm based on the individual health attributes of both HUI2 and HUI3. The HRQoL score possesses an interval-scale property, ranging from 0.00, representing conventional death, to 1.00, indicative of perfect health [47].

Personal Outcomes Scale employs a 3-point Likert scale to assess the patient's quality of life, both self-reported and through direct observation, across three dimensions (independence, social participation, and wellbeing) which are further divided over eight domains (personal development, self-determination, interpersonal relations, social inclusion, rights, and emotional, physical, and material wellbeing). The scores from these domains are summed to calculate the quality-of-life self-report index and quality of life observation index [48].

The 5-level EQ-5D version (EQ-5D-5 L) essentially consists of 2 pages: the EQ-5D descriptive system and the EQ visual analogue scale (EQ VAS). The descriptive system comprises five dimensions: mobility, self-care, usual activities, pain/discomfort and anxiety/depression. Each dimension has 5 levels: no problems, slight problems, moderate problems, severe problems and extreme problems [49, 50].

The KidsLife is a 96-item assessment tool specifically designed to evaluate the eight core domains of QoL in individuals with Intellectual Disabilities. It is offered in both Spanish and English versions. Responses are recorded on a 4-point Likert scale, where 1 represents 'totally disagree', 2 indicates 'disagree', 3 signifies 'agree', and 4 corresponds to 'totally agree' or 'always' [52–55].

The KidsLife-Down assessment tool comprises eight domains that include the individual's self-determination within their daily life. With an estimated completion time of 30 min and freely accessible. The KidsLife-Down Scale is a specific adaptation for the DS population of the Kidslife Scale. Its primary objectives include guiding evidence-based interventions and tailoring individualized support plans. The scale furnishes standardized scores and percentiles across eight fundamental dimensions of QoL: emotional well-being, physical well-being, material well-being, personal development, interpersonal relationships, social inclusion, self-determination, and rights. Furthermore, it facilitates the visualization of acquired data through a QoL profile format. This assessment tool targets populations encompassing childhood, adolescence, and youth [51].

These eight dimensions encompass emotional well-being, physical well-being, material well-being, personal development, interpersonal relationships, social inclusion, self-determination, and rights. Depending on the child's age, the total raw score for each domain is converted to the corresponding standard score. Higher scores reflect higher levels of QoL. An overall QoL score is derived by summing the standard scores of the eight domains. Additionally, the scale provides percentiles based on the standard scores [52–55].

Psychometrics properties of instruments validated in children and adolescents with Down syndrome

Ten studies evaluated the psychometric properties of six instruments validated in children and adolescents with DS [45, 47, 49, 50, 53, 54, 57–60] (Table 3). Regarding the psychometric properties, a better report and performance was identified for the Kidslife and Kidslife-Down instruments. Among HRQoL evaluation instruments, there was a better report for QI-disability. For the HUI, the psychometric properties evaluated in the population with DS are scarce, for the Kidscreen-27, only reproducibility is reported, and this is low for this population (Table 4). None of the reported studies evaluated criterion validity or sensitivity.

Discussion

Measurement of QoL in children and adolescents is an aspect that has gained great clinical importance in recent years and is a field of research interest because of the increase in the number of children and adolescents with chronic diseases and disorders [61–63]. These measures should have validated psychometric properties of reliability and validity, suitability of measures for specific age ranges, and measures that do not exhibit large practice, ceiling, or floor effects [64, 65]. This scoping review identifies instruments used to evaluate quality of life; however, it does not aim to recommend the use of any specific instruments described herein.

Children and adolescents with DS have special healthcare and service provision needs, and QoL assessment can give useful information to professionals working in health organizations for the well-being of people with DS in different service and support delivery contexts [38]. It is a way of evaluating the effectiveness of the interventions [45], as it measures how health status, and treatments affect QoL.

Quality of life measurement in people with neurodevelopmental disabilities considers person-centered and family-centered planning. Quality of life is a social construct about the ongoing and lasting changes in people's lives. Quality of life assessment needs to be interpreted through the lens of the lived experience of people with disabilities or families that include disability. This is essential, as human beings characteristically find and express somewhat positive levels of satisfaction, happiness, and quality even in conditions that others might judge to lack quality [66].

Table 3 Characteristics of the studies evaluating the psychometric properties of the instruments included in the review

Instrument	Authors	Aim of the study	Country of application	Responder population	Dimensions	Scale or punctuation	Comparison instrument	Psychometrics properties reported and results
Health Utilities Index Mark 2 [HUI2] and Health Utilities Index Mark 3 [HUI3]	Mok W et al. 2014 [47]	[1] To validate the Chinese version of Health Utilities Index [HUI-Ch]; [2] To examine the Health-related Quality of Life [HRQoL] of Chinese subjects with Down syndrome	China	Parents or caregivers of people with DS between 5 and > 18 years old.	**HUI2**: Sensation Mobility Emotion CognitionSelf-care Pain and Fertility **HUI3**: Vision Hearing Speech Ambulation Dexterity Emotion Cognition Pain	Score of 0.00 being conventional dead to 1.00 being in perfect health	NR	**Cross-cultural validation**: the chinese version was validated according to standard guidelines.
KIDSCREEN-27	Rofail D et al. 2017 [45]	To report the HRQoL of adolescents and adults with DS from a non-interventional, longitudinal, multi-national study that used the KIDSCREEN-27 parent proxy-report form and to assess test-retest reliability of the KIDSCREEN-27	Argentina, France, Italy, Spain, Canada, United Kingdom and United States of America	Caregivers of individuals with DS between 12 and 30 years old.	Physical Well-being Psychological Well-being Autonomy and Parent Relations Social Support and Peers School Environment	A severity scale of 5 points: 'not at all,' 'slightly,' 'moderately,' 'very' and 'extremely.' A frequency scale of 5 points: 'never,' 'seldom,' 'quite often,' 'very often' and 'always.'	NR	**Reliability**: Demonstrated poor-to-moderate test-retest [ICC] reliability across domains and age groups, with ICCs ranging from 0.31 to 0.66. The strongest ICC value was recorded for the School Environment domain in the adolescent group [0.66].

Table 3 (continued)

Instrument	Authors	Aim of the study	Country of application	Responder population	Dimensions	Scale or punctuation	Comparison instrument	Psychometrics properties reported and results
EQ-5D-Y-5 L	Blackmore AM et al. 2023 [49]	To determine how closely descriptions by parents of children with intellectual disability aligned with their EQ-5D responses.	Australia	Caregivers	Mobility Self-care Usual activities Pain/discomfort Anxiety/depression	5 questions, each with a 5-point rating scale ranging from ´no problems´ to ´cannot´ in the first 3 questions [mobility/walking around, looking after self, usual activities] and ´no´ / ´not´ to ´extreme´ in the last 2 questions [pain or discomfort, feeling worried, sad or unhappy]	Instrument was compared with descriptions provided by parents of children	**Content validity**: Was considered clear, concise, and largely relevant, but insufficiently comprehensive for this population.
EQ-5D-Y	Burström K et al. 2014 [50]	To test the feasibility and validity of the EQ-5D-Y in a Swedish patient sample of children and adolescents with functional motor, orthopaedic, and medical disabilities	Sweden	Children and adolescents with functional disability	Mobility [walking about] Looking after myself Doing usual activities Feeling worried, sad or unhappy Having pain or discomfort.	´no problems´, ´some problems´, and ´a lot of problems´ in each of the mentioned dimensions	KIDSCREEN-10, KIDSCREEN-27	**Divergent validity**: ´mobility [walking about]´ dimension of the EQ-5D-Y did not correlate with the psychological well-being dimension of the KIDSCREEN-27, indicating divergent validity between these dimensions **Convergent validity**: Moderate significant correlations between the dimensions of the EQ-5D-Y and the KIDSCREEN-27, Specifically, significant correlations were found between the ´having pain or discomfort´ dimension of the EQ-5D-Y and the KIDSCREEN-10 in the general population.
Quality of Life Inventory -Disability [QI-Disability]	Downs J. et al. 2018 [57]	To evaluated a new parent-report measure for children with intellectual disability.	Australia	Families with a child with Down syndrome, or Rett syndrome, or cerebral palsy or with ASD.	Social interaction Positive emotions Physical health Negative emotions Leisure and the outdoors Independence	0-100 points	NR	**Construct validity**: the exploratory factor analysis show factor loadings greater than 0.5 for all items except 2. **Reliability**: values ranged from 0.75 for "physical health" to 0.91 for "positive motions", **Internal consistency**: Between 07-0.9.

Table 3 (continued)

Instrument	Authors	Aim of the study	Country of application	Responder population	Dimensions	Scale or punctuation	Comparison instrument	Psychometrics properties reported and results
Quality of Life Inventory-Disability [QI-Disability]	Epstein A et al. 2019 [58]	To provide a deeper description of the content validity of QI-Disability including item generation, cognitive debriefing, refinement prior to administration, and its conceptual foundation.	Australia	Parent caregivers of children with intellectual disability [Down syndrome, Rett syndrome, cerebral palsy, or autism spectrum disorder]	Health Comfort Behaviour and emotions Communication Movement Routines Family and friends Leisure and Recreation Nature and the outdoors	NR	NR	**Content validity**: Satisfactory content validity is reported
KidsLife	Gómez L et al. 2016 [59]	To evaluate thepsychometric properties of the field-test version of the KidsLife Scale.	Spain	Professionals, parents or caregivers of people with intellectual disabilities between the ages of 4 and 21.	Social inclusion self-determination, emotional wellbeing, physical wellbeing, material wellbeing, rights, personal development. interpersonal relationships	Frequency scale with four options: never, sometimes, often, always.	NR	**Internal consistency**: 0.812 [rights] and 0.949 [personal development]. **Construct validity**: demonstrated adequate indexes of fit for the eight-domain model.
Kidslife	Mora A et al. 2020 [60]	to analyze the dimensional structure of the KidsLife scale.	Colombia	Caregivers of people, aged between 4 and 21 years, diagnosed with intellectual disability	Social inclusion self-determination, emotional wellbeing, physical wellbeing, material wellbeing, rights, personal development. interpersonal relationships	Frequency scale with four options: never, sometimes, often, always.	NR	**Construct validity**: All the eigenvalues exceeded the value of 1, the lowest was 1.70 and the maximum value was 2.52. The first factor explained more than 40% of the total variance. **Fiability**: was in a range of 0.81 y 0.87 for all dimensions. **Internal consistency**: was in a range of 0.79 y 0.86 for all dimensions.

Table 3 (continued)

Instrument	Authors	Aim of the study	Country of application	Responder population	Dimensions	Scale or punctuation	Comparison instrument	Psychometrics properties reported and results
Kidslife-Down	Gómez L et al. 2020 [53]	To adapt the KidsLife scale by selecting the most reliable and discriminant items for children and youth with DS	Spain	An informant who knew the child or young person with DS between the ages of 4 and 21	Social inclusion self-determination, emotional wellbeing, physical wellbeing, material wellbeing, rights, personal development. interpersonal relationships	Frequency scale with four options: never, sometimes, often, always.	NR	**Internal consistency**: was in a range of 0.81 y 0.90 for all dimensions. **Construct validity**: the domains achieved a high degree of convergent validity because the majority of the average variance extracted values were close to 0.5
Kidslife-Down	Jofré Urrutia et al. 2023 [54]	To adapt and validate the KidsLife-Down scale for children and young people with Down syndrome in Chile	Chile	Families of boys, girls, and young people with DS between the ages of 4 and 21 years. The scale was answered by relatives or caregivers.	emotional well-being, physical well-being, material well-being, personal development, rights, self-determination, social inclusion, and interpersonal relationships	a Likert scale [never, sometimes, often, always]. Each item in the scale was scored on a scale of 1 to 4 points, and the scores for each dimension were added to calculate a "QoL Index" correlated with a percentile of quality of life.	NR	**Internal consistency**: was greater than 0.7 for all items **Construct validity**: factor loadings above 0.40, with correlations between the domains ranging from 0.39 to 0.86. The average variance extracted values were close to 0.50, and omegas ranged from 0.85 to 0.90

ASD: Autism spectrum disorder; NR: Not reported; DS: Down Syndrome; HRQoL: Health-related quality of life

Table 4 Evaluation of psychometrical properties with COSMIN

	Kidslife			Kidslife-Down			QI-Disability			Health Utilities Index			Kidscreen-27			EQ-5D-Y-5 L		
	Grade +/-/?			Grade +/-/?			Grade +/-/?			Grade +/-/?			Grade +/-/?			Grade +/-/?		
	RV 1	RV 2	C	RV 1	RV 2	RV 1	RV 2	C	RV 1	RV 2	C	RV 1	RV 2	C	RV 1	RV 2	C	
Structural validity	+	+	+	+	+	+	+	+	+	+	+	?	?	?	?	?	?	?
Internal consistency	+	+	+	+	+	+	+	+	?	?	?	?	?	?	?	?	?	?
Cross-cultural validity	+	+	+	+	+	+	+	+	?	?	?	?	?	?	?	?	?	?
Measurement invariance	+	+	+	+	+	+	+	+	?	?	?	?	?	?	?	?	?	?
Reliability	?	?	?	?	?	?	+	+	?	?	?	-	-	-	?	?	?	?
Measuring error	?	?	?	?	?	?	-	-	?	?	?	?	?	?	?	?	?	?
Criterion validity	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?
Construct validity	+	+	+	+	+	+	+	+	+	+	+	?	?	?	?	?	?	?
Sensitivity	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?	?

RV: Reviewer; C: consensus. Table adjusted from: Prinsen CAC et al. Quality of Life Research. 2018;27 [5]:1147–57

Disability impacts the whole family and the determination of appropriate conceptualization of family outcomes requires an understanding of the impact of members with a disability on family QoL. Instruments that measure health-related QoL with focus on the individual person, they still support a theoretical perspective of QoL near from the traditional medical approach. QoL assessment should not represent a classification of individuals, services or systems, but it should help provide, within health service systems and organizations, a value system coherent with those values held by people with DS [67].

Brown and Faragher considers QoL as a value system which needs to permeate both formally and informally the life of each child, specially the concepts and education´s principles. So, a better approach in the provision of health system services could be include education, social and cultural contexts in order to understand whole life of people [66].

The search conducted in this review found one specific instrument for assessing this outcome in children and adolescents with DS [63]. In the first search of the instruments identified for evaluating quality of life in children with DS, only six of these have studies that report some of the psychometric properties in children and adolescents with DS. This review found two instruments to assess QoL in children and adolescents with DS and ten to assess HRQoL in the general pediatric population.

Although, QI-Disability was designed specifically for children with intellectual disability such as Down syndrome, Rett syndrome, cerebral palsy or autism spectrum disorder, psychometric properties are not reported differentially for each of these conditions [58].

Instruments such as Kidscreen-24, Kidscreen-52, PedsQL 4.0 and TACQoL include physical, psychological/emotional, and social dimensions, these could be used in clinical settings. The correlation between these dimensions and the special characteristics of children with DS has not been established [46].

We find generic and specific instruments, which have features. On the one hand, generic instruments allow comparisons of health status between individuals in the general population or patients with different conditions. Moreover, they provide an initial idea of the impact of that disease on the patient's HRQoL. However, one of their major limitations is that they are usually not sufficiently sensitive to significant clinical changes in dimensions that would be included in specific instruments. On the other hand, specific instruments include only the important aspects of a given health problem in each population (e.g., children or the elderly) to assess certain functions (e.g., sexual function) or a given clinical symptom (e.g., pain) They have the advantage of being more sensitive to changes in HRQoL than generic ones to the specific health problem being assessed [65–69].

The instruments identified dimensions such as physical activity and health, mood and feelings, family life and free time, social support and friends, and their school environment. Parents or caregivers completed the instruments, i.e., they were proxy instruments. Proxy instruments in the case of people with DS seem to facilitate the assessment of QoL of patients since they reduce the methodological difficulties involved in measuring it directly in patients. Such instruments consider the fact that people with DS may have different levels of intellectual disability, which would hinder their ability to understand the test, the scale, and their competence to assent. This is a limitation in the exploration of QoL of the child or adolescent with DS, because it is assessed from the caregiver perspective and not from the perspective of the patient living with the condition.

Low concordance between self-report and proxy-report has been consistently demonstrated in the measurement of QoL of chronically ill and healthy children, mainly for items related to feelings such as sadness, school rejection, pain, and symptoms that are not observable by the caregiver, such as gastrointestinal symptoms [47–49]. These discrepancies arise, among other reasons, because it is impossible for the caregiver to separate his/her own QoL from that of the child or adolescent. Otherwise, caregivers are conditioned by the emotions they have for the subject and the time they have shared with them to provide a score [69]. Nevertheless, there is a consensus that an individual's self-assessment of their own HRQoL is usually more reliable and accurate than proxy assessment, and therefore self-report should be used wherever possible because the age might condition the questionnaire development time, since the younger age, the more detailed the explanation of the questionnaire will be necessary [70]. Some instruments recognize as informants social service professionals, family members or close relatives who have known the person with Down syndrome for at least six months [20].

As stated by Gómez et al. [18, 62] it is important to consider the perspective of people with DS, because although the proxy reports are adequate, future instrument development and studies that consider the experiences and point of view of people with this condition are needed.

The sociocultural context of the parents, caregivers, and the family in general also has an impact on the QoL assessment performed with the children. For example, it has been documented that parents of children with low height tend to rate them as having lower social functioning, worse self-esteem, and more behavioral and cognitive problems than those with average height, while this opinion is rarely shared by their children [50, 51]. Therefore, this limited parent–child agreement meant that children and their parents reported data as complementary sources of information [71–73] for the assessment of QoL in children and adolescents.

Psychometric property studies report at least one method and one statistician for each property evaluated. This method allows the clinicians and researchers to know how the instrument measures what it is intended to measure and yields consistent results even in case of variability in the conditions of the populations participating in the studies [52–54]. Furthermore, it is possible to exchange information between the national and international scientific community and build a database to study the clinical and functional behaviors of people with DS. Studies suggest that measures for young children should be developed based on a strong conceptual model and dimensions that inform observable behavior. In this way, observer- or proxy-reported outcome measures allow the observer to report behaviors they have seen, rather than having to infer the QoL experienced by the child, based on their own subjective assessment [74, 75].

We identified two specific instruments for the intellectually disabled population, as well as for children and adolescents with Down syndrome, which specifically assess the general construct of quality of life: Kidslife and Kidslife Down. These instruments have demonstrated several important psychometric properties. However, other crucial properties, such as criterion validity and the instrument's performance in assessing the effectiveness of therapeutic interventions in health contexts, have not yet been reported. This is significant because dimensions like Social Inclusion, Material Well-being, and Rights may not be directly impacted by pharmacological or therapeutic interventions assessed in controlled clinical trials [51–55].

Efforts should prioritize assessing QoL from the perspective of individuals with DS, with proxy instruments serving as supplementary information. Future research should focus on evaluating kidslife and kidslife Down in various clinical contexts to gauge therapeutic intervention effectiveness, or consider developing new, culturally sensitive instruments. Such approaches can foster a holistic understanding of QoL, enhancing patient-centered healthcare services and aligning clinical and academic communities with the population's needs.

This scoping review has several limitations that should be considered when interpreting its findings. Firstly, the literature search was focused on a specific set of databases and sources, which may have excluded relevant studies published on other platforms or as gray literature. Additionally, the inclusion of studies with varying methodologies and approaches to assessing quality of life may have introduced variability in the results, complicating the comparison, synthesis, and understanding of the data. These limitations underscore the need for further research to address these aspects and provide additional

elements that could facilitate the selection of the most appropriate instrument for assessing quality of life in this population, according to the context and the specific needs of the evaluation.

Conclusions

Quality of life serves as a pivotal outcome measure in evaluating interventions for children with Down Syndrome, highlighting the necessity of employing suitable instruments. The psychometric properties of these tools are paramount, mitigating measurement bias and potentially influencing sample sizes in clinical studies.

Commonly utilized instruments in this field include the PedsQL 4.0 and KIDSCREEN. Notably, while the PedsQL 4.0 lacks specific evaluation in minors with DS, KIDSCREEN data is inconsistently reported.

Emerging instruments like Kidslife and Kidslife Down are increasingly employed, particularly in assessing social and community intervention program efficacy. Rigorous evaluation of their performance in clinical contexts is imperative, or the development of tailored instruments for children with DS to comprehensively assess QoL in clinical settings, considering their unique needs.

Abbreviations

QoL	Quality of Life
DS	Down Syndrome
HRQoL	Health-Related Quality of Life
COSMIN	Consensus Based Standards for the Selection of Health Measurement Instruments

Supplementary Information

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Supplementary Material 1

Supplementary Material 2

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EIRG, MLDG, PCMP, OCVP, MRTN, NRM performed the research, screening, selection, and analysis of the studies and data, performed the meta-analyses. All authors read and approved the final manuscript.

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Data availability

All data generated or analyzed during this study are included in this published article and its supplementary information files.

Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

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Competing interests

The authors declare no competing interests.

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References

1. Botero de Mejía BE, Pico Merchán ME. Calidad De Vida Relacionada Con La Salud (Cvrs) En Adultos Mayores De 60 Años: Una Aproximación Teórica. Hacia Promoc Salud Noviembre De. 2007;12(1):11–24.
2. Felce D, Perry J. Quality of life: its definition and measurement. Res Dev Disabil febrero de. 1995;16(1):51–74.
3. Crawford D, Dearmun A. Down's syndrome. Nurs Child Young People 8 De Noviembre De. 2016;28(9):17–17.
4. Rojnueangnit K, Khaosamlee P, Chunsuwan I, Vorravanpreecha N, Lertboonnum T, Rodjanadit R, et al. Quality of life and comprehensive health supervision for children with Down syndrome in Thailand. J Community Genet 1 de Julio De. 2020;11(3):351–8.
5. Departamento de Salud del Estado de Nueva York. Normas de práctica clínica guía rápida de consulta para padres y profesionales síndrome de down evaluación e intervención para niños pequeños (desde El Nacimiento Hasta Los 3 años de edad). En Nueva York: Departamento de Salud del Estado de Nueva York; 2002. pp. 7–11. https://www.health.ny.gov/publications/5304.pdf.
6. Bower C, Leonard H, Petterson B. Intellectual disability in Western Australia. J Paediatr Child Health. 2000;36(3):213–5.
7. Bittles AH, Bower C, Hussain R, Glasson EJ. The four ages of Down syndrome. Eur J Public Health Abril De. 2007;17(2):221–5.
8. Nations U, United Nations. United Nations; [citado 18 de agosto de 2021]. Día Mundial del Síndrome de Down | Naciones Unidas. Disponible en: https://www.un.org/es/observances/down-syndrome-day
9. Emotional Burden in Parents of Children with Trisomy 21. Descriptive Study in a Colombian Population [Internet]. [citado 18 de agosto de 2021]. Disponible en: http://www.scielo.org.co/scielo.php?script=sci_arttext&pid=S1657-92672016000100003&lng=e&nrm=iso
10. Leonard S, Msall M, Bower C, Tremont M, Leonard H. Functional status of school-aged children with Down syndrome. J Paediatr Child Health Abril De. 2002;38(2):160–5.
11. de Winter CF, Jansen Aa, Evenhuis C. Physical conditions and challenging behaviour in people with intellectual disability: a systematic review. J Intellect Disabil Res JIDR Julio De. 2011;55(7):675–98.
12. Foley KR, Jacoby P, Girdler S, Bourke J, Pikora T, Lennox N, et al. Functioning and post-school transition outcomes for young people with Down syndrome. Child Care Health Dev Noviembre De. 2013;39(6):789–800.
13. Organización Mundial de la Salud [Internet]. OMS. Enfermedades crónicas. Disponible en: https://www.who.int/topics/chronic_diseases/es/
14. Cáceres-Manrique F, de Parra-Prada M, Pico-Espinosa LM. OJ. Calidad de vida relacionada con la salud en población general de Bucaramanga, Colombia. Rev Salud Pública. 1 de marzo de 2018;20(2):147 – 54.
15. Martín JCM. Modelo de calidad de vida de Schalock y Verdugo (2002/2003): análisis multivariante [Internet] [http://purl.org/dc/dcmitype/Text]. Universidad de Salamanca; 2006 [citado 18 de agosto de 2021]. Disponible en: http://dialnet.unirioja.es/servlet/tesis?codigo=144464
16. Vega V, Jenaro C, Flores N, Cruz M, Artaza C. Quality of life of adults with Intellectual Disabilities Institutionalized in Chile from the perspective of Service Providers. Univ Psychol 1 de Julio De. 2013;12:923–32.
17. Kober R, editor. Enhancing the Quality of Life of People with Intellectual Disabilities: From Theory to Practice [Internet]. Dordrecht: Springer Netherlands; 2011 [citado 22 de febrero de 2024]. (Social Indicators

Research Series; vol. 41). Disponible en: http://link.springer.com/https://doi.org/10.1007/978-90-481-9650-0
18. Escala KidsLife-Down. Evaluación de la calidad de vida de niños y adolescentes con síndrome de Down – SID [Internet]. [citado 22 de febrero de 2024]. Disponible en: https://sid-inico.usal.es/documentacion/escala-kidslife-down-evaluacion-de-la-calidad-de-vida-de-ninos-y-adolescentes-con-sindrome-de-down/
19. Ijezie OA, Healy J, Davies P, Balaguer-Ballester E, Heaslip V. Quality of life in adults with Down syndrome: a mixed methods systematic review. PLOS ONE 1 de mayo de. 2023;18(5):e0280014.
20. Carvajal A, Centeno C, Watson R, Martínez M, Sanz Rubiales Á. ¿Cómo validar un instrumento de medida de la salud? An Sist Sanit Navar. abril de. 2011;34(1):63–72.
21. OSF [Internet]. [citado 23 de octubre de 2022]. Disponible en: https://osf.io/.
22. Levac D, Colquhoun H, O'Brien KK. Scoping studies: advancing the methodology. Implement Sci 20 de Septiembre De. 2010;5(1):69.
23. Tricco AC, Lillie E, Zarin W, O'Brien KK, Colquhoun H, Levac D et al. PRISMA Extension for Scoping Reviews (PRISMA-ScR): Checklist and Explanation. Ann Intern Med. 2 de octubre de. 2018;169(7):467 – 73.
24. Peters MDJ, Godfrey CM, Khalil H, McInerney P, Parker D, Soares CB. Guidance for conducting systematic scoping reviews. Int J Evid Based Healthc Septiembre De. 2015;13(3):141–6.
25. Arksey H, O'Malley L. Scoping studies: towards a methodological framework. Int J Soc Res Methodol 1 de febrero de. 2005;8(1):19–32.
26. Home - MeSH. - NCBI [Internet]. [citado 17 de julio de 2022]. Disponible en: https://www.ncbi.nlm.nih.gov/mesh/
27. Rayyan. – Intelligent Systematic Review [Internet]. [citado 1 de junio de 2021]. Disponible en: https://www.rayyan.ai/
28. Prinsen Ca, Mokkink C, Bouter LB, Alonso LM, Patrick J, de Vet DL. COSMIN guideline for systematic reviews of patient-reported outcome measures. Qual Life Res Int J Qual Life Asp Treat Care Rehabil mayo de. 2018;27(5):1147–57.
29. COSMIN-syst-review. -for-PROMs-manual_version-1_feb-2018-1.pdf [Internet]. [citado 19 de agosto de 2021]. Disponible en: https://www.cosmin.nl/wp-content/uploads/COSMIN-syst-review-for-PROMs-manual_version-1_feb-2018-1.pdf
30. Xanthopoulos MS, Walega R, Xiao R, Prasad D, Pipan MM, Zemel BS, et al. Caregiver-reported quality of life in Youth with Down Syndrome. J Pediatr Octubre De. 2017;189:98–e1041.
31. Abd El-Hady SS, Abd El-Azim FH, El-Talawy HAEAM. Correlation between cognitive function, gross motor skills and health – related quality of life in children with Down syndrome. Egypt J Med Hum Genet. 2018;19(2):77–81.
32. Garcia Guerra G, Joffe AR, Robertson CMT, Atallah J, Alton G, Sauve RS, et al. Health-related quality of life experienced by children with chromosomal abnormalities and congenital heart defects. Pediatr Cardiol. 2014;35(3):536–41.
33. Katsiana A, Strimpakos N, Ioannis V, Kapreli E, Sofologi M, Bonti E, et al. Health-related quality of life in children with Autism Spectrum Disorder and Children with Down Syndrome. Mater Socio-Medica. 2020;32(2):93–8.
34. Alqahtani AS, Algabbani MF, Alhammad SA, Alwadeai KS, Alhusaini A. Physical activity status and its association with quality of life among children with down syndrome in Saudi Arabia: a comparative cross-sectional study. PLoS ONE. 2024;19(2):e0297111.
35. Alhammad SA, Alqahtani AS, Alwadeai KS, Algabbani MF, Alhusaini AA. Walking capacity and its association with quality of life among children with down syndrome in Saudi Arabia. BMC Pediatr. 2024;24(1):55.
36. Supramaniam N, Zanudin A, Azmi NA, Body Mass Index P, Activity. Cardiorespiratory Endurance and quality of life among children with physical disabilities. Child [Internet]. 2023;10(9). Disponible en: https://www.embase.com/search/results?subaction=viewrecord&id=L2025666301&from=export U2 - L2025666301
37. Fucà E, Galassi P, Costanzo F, Vicari S. Parental perspectives on the quality of life of children with Down syndrome. Front Psychiatry. 2022;13:957876.
38. Fernández Scotto E, Eymann A. Health-related quality of life in children with Down syndrome. Arch Argent Pediatr. 2023;121(4):e202202756.
39. van Gameren-Oosterom HBM, Fekkes M, Buitendijk SE, Mohangoo AD, Bruil J, Van Wouwe JP. Development, problem behavior, and quality of life in a population based sample of eight-year-old children with Down syndrome. PLoS ONE. 2011;6(7):e21879.
40. Alhaddad FA, Alkhushi NA, Alharbi AM, Al Talib SA, Sultan SM, Bahawi YO. Quality of Life among Down Syndrome patients with and without congenital heart disease at King Abdulaziz University Hospital, Jeddah, Saudi Arabia. Cureus. 2023;15(1):e33553.
41. Schultz KAP, Chen L, Kunin-Batson A, Chen Z, Woods WG, Gamis A, et al. Health-related quality of life (HR-QOL) and Chronic Health conditions in survivors of Childhood Acute myeloid leukemia (AML) with Down Syndrome (DS): a Report from the children's Oncology Group. J Pediatr Hematol Oncol enero de. 2017;39(1):20–5.
42. Leonard H, Whitehouse A, Jacoby P, Benke T, Demarest S, Saldaris J et al. Quality of life beyond diagnosis in intellectual disability – latent profiling. Res Dev Disabil [Internet]. 2022;129. Disponible en: https://www.embase.com/search/results?subaction=viewrecord&id=L2019603478&from=export U2 - L2019603478
43. Jung HK, Chung E, Lee BH. A comparison of the function, activity and participation and quality of life between down syndrome children and typically developing children. J Phys Ther Sci agosto de. 2017;29(8):1377–80.
44. Shields N, Leonard H, Munteanu S, Bourke J, Lim P, Taylor NF, et al. Parent-reported health-related quality of life of children with Down syndrome: a descriptive study. Dev Med Child Neurol Abril De. 2018;60(4):402–8.
45. Rofail D, Froggatt D, de la Torre R, Edgin J, Kishnani P, Touraine R, et al. Health-Related Quality of Life in individuals with Down Syndrome: results from a non-interventional longitudinal multi-national study. Adv Ther agosto de. 2017;34(8):2058–69.
46. Alrayes N, Issa NM, Alghubayshi OY, Al-Amaa JY, Alsabban AH, Al Shaer DS, et al. Quality of life in children with Down syndrome and its association with parent and child demographic characteristics: parent-reported measures. Mol Genet Genomic Med. 2024;12(1):e2337.
47. Mok WKY, Wong WH, Mok GTK, Chu YWY, Ho FKW, Chow CB et al. Validation and application of health utilities index in Chinese subjects with down syndrome. Health Qual Life Outcomes [Internet]. 2014;12(1). Disponible en: https://www.embase.com/search/results?subaction=viewrecord&id=L612220551&from=export
48. Bermudez BEBV, Franklin GL, Oliveira CM, Coutinho L, Crippa ACS. Quality of life in Down syndrome in Brazil: a cross-sectional study. Arq Neuropsiquiatr. 2023;81(11):943–8.
49. Blackmore AM, Mulhern B, Norman R, Reddihough D, Choong CS, Jacoby P, et al. How well does the EQ-5D-Y-5L describe children with intellectual disability? «There's a Lot more to my child than that she can't wash or Dress Herself». Value Health J Int Soc Pharmacoeconomics Outcomes Res. 2024;27(2):190–8.
50. Burström K, Bartonek Å, Broström EW, Sun S, Egmar AC. EQ-5D-Y as a health-related quality of life measure in children and adolescents with functional disability in Sweden: testing feasibility and validity. Acta Paediatr Oslo nor 1992 Abril De. 2014;103(4):426–35.
51. Camacho R, Castejón-Riber C, Requena F, Camacho J, Escribano BM, Gallego A et al. Quality of Life: Changes in Self-Perception in People with down Syndrome as a Result of Being Part of a Football/Soccer Team. Self-Reports and External Reports. Brain Sci [Internet]. 2021 [citado 1 de enero de 2d. C.];11(2). Disponible en: https://pubmed.ncbi.nlm.nih.gov/33673165/
52. Lee A, Knafl G, Knafl K, Van Riper M. Quality of life in individuals with Down syndrome aged 4 to 21 years. Child Care Health Dev. 2021;47(1):85–93.
53. Gómez LE, Verdugo MA, Rodríguez M, Morán L, Arias VB, Monsalve A. Adapting a measure of quality of life to children with Down Syndrome for the development of evidence-based interventions. Psychosoc Interv. 2020;29(1):39–48.
54. Jofré Urrutia M, Bedregal P, Lizama M. Adapting a quality of life scale for children and young people with Down syndrome in Chile. Am J Med Genet febrero de. 2024;194(2):233–42.
55. Morán L, Gómez LE, Balboni G, Monsalve A, Verdugo MÁ, Rodríguez M. Predictors of individual quality of life in young people with Down syndrome. Rehabil Psychol mayo de. 2022;67(2):205–14.
56. Verstegen RHJ, van Gameren-Oosterom HBM, Fekkes M, Dusseldorp E, de Vries E, van Wouwe JP. Significant impact of recurrent respiratory tract infections in children with Down syndrome. Child Care Health Dev Noviembre De. 2013;39(6):801–9.
57. Downs J, Jacoby P, Leonard H, Epstein A, Murphy N, Davis E, et al. Psychometric properties of the quality of life inventory-disability (QI-Disability) measure. Qual Life Res Int J Qual Life Asp Treat Care Rehabil. 2019;28(3):783–94.
58. Epstein A, Williams K, Reddihough D, Murphy N, Leonard H, Whitehouse A, et al. Content validation of the quality of Life Inventory-Disability. Child Care Health Dev. 2019;45(5):654–9.
59. Gómez LE, Alcedo MT, Arias B, Fontanil Y, Arias VB, Monsalve A, et al. A new scale for the measurement of quality of life in children with intellectual disability. Res Dev Disabil. 2016;53:399–410.
60. Mora-Antó A, Salamanca-Duque LM, Córdoba-Andrade L, Gómez-Sánchez LE. Estructura Dimensional De La Escala KidsLife, versión Colombia, para la

Evaluación De Calidad De Vida En Discapacidad Intelectual. Psykhe Santiago mayo de. 2020;29(1):1–21.
61. Esbensen AJ, Hooper SR, Fidler D, Hartley SL, Edgin J, d'Ardhuy XL, et al. Outcome measures for clinical trials in Down Syndrome. Am J Intellect Dev Disabil mayo de. 2017;122(3):247–81.
62. Hart SJ, Visootsak J, Tamburri P, Phuong P, Baumer N, Hernandez MC, et al. Pharmacological interventions to improve cognition and adaptive functioning in Down syndrome: strides to date. Am J Med Genet Noviembre De. 2017;173(11):3029–41.
63. Keeling LA, Spiridigliozzi GA, Hart SJ, Baker JA, Jones HN, Kishnani PS. Challenges in measuring the effects of pharmacological interventions on cognitive and adaptive functioning in individuals with Down syndrome: a systematic review. Am J Med Genet Noviembre De. 2017;173(11):3058–66.
64. Ravens-Sieberer U, Karow A, Barthel D, Klasen F. How to assess quality of life in child and adolescent psychiatry. Dialogues Clin Neurosci junio de. 2014;16(2):147–58.
65. Gómez LE, Schalock RL, Verdugo MÁ. A quality of life supports model: six research-focused steps to evaluate the model and enhance research practices in the field of IDD. Res Dev Disabil. 1 de diciembre de 2021;119:104112.
66. Faragher R, Broadbent C, Brown R, Burgess J. Applying the principles of quality of life to education. En 2014. pp. 37–54.
67. Bertelli M, Francescutti C, Brown I. Reframing QoL assessment in persons with neurodevelopmental disorders. Ann Ist Super Sanita. 2020;56(2):180–92.
68. Goodman MJ, Brixner DI. New therapies for treating Down syndrome require quality of life measurement. Am J Med Genet Abril De. 2013;161A(4):639–41.
69. Kenzik KM, Tuli SY, Revicki DA, Shenkman EA, Huang IC. Comparison of 4 Pediatric Health-Related Quality-of-Life instruments: a study on a Medicaid Population. Med Decis Mak 1 de Julio De. 2014;34(5):590–602.
70. Velarde-Jurado E, Avila-Figueroa C. Consideraciones metodológicas para evaluar la calidad de vida. Salud Pública México Septiembre De. 2002;44(5):448–63.
71. Badia Llach X. Qué es y cómo se mide la calidad de vida relacionada con la salud. Gastroenterol Hepatol 2 de marzo de. 2004;27:2–6.
72. Eiser C, Varni JW. Health-related quality of life and symptom reporting: similarities and differences between children and their parents. Eur J Pediatr Octubre De. 2013;172(10):1299–304.
73. Karande S. Health-related quality of life in children with specific learning disability: children's self-reports versus parents' proxy reports. Eur J Pediatr. 30 de noviembre de 2013;173.
74. Quitmann J, Rohenkohl A, Sommer R, Bullinger M, Silva N. Explaining parent-child (dis)agreement in generic and short stature-specific health-related quality of life reports: do family and social relationships matter? Health Qual Life Outcomes. 21 de octubre de. 2016;14(1):150.
75. Montgomery KE, Vos K, Raybin JL, Ward J, Balian C, Gilger EA, et al. Comparison of child self-report and parent proxy-report of symptoms: results from a longitudinal symptom assessment study of children with advanced cancer. J Spec Pediatr Nurs JSPN Julio De. 2021;26(3):e12316.

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