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Review Article

Patient-reported outcome measures in Spina Bifida from adolescence to adulthood — A systematic review

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Summary

Aims

Spina bifida (SB) is a complex condition, predisposing individuals to lifelong challenges in mobility, continence, and overall health. Care modalities are multimodal, and advancements have improved survival, shifting clinical focus towards optimizing long-term function and quality of life.

The aim of this systematic review is to outline patient-reported outcome measures (PROMs) used to assess health-related quality of life (HRQOL) and other conditions in transitional SB patients, and to analyze factors influencing the results they provide.

Methods

We conducted a systematic review according to PRISMA guidelines, including sixteen studies with 2009 participants.

Results

Participants' ages ranged from 12 to 74 years, with a gender distribution of 59 % female. 72 % used CIC, 48 % had urinary incontinence and 32 % had bowel incontinence. The Pediatric Global Health 7 (PGH-7) and Neurogenic Bladder Symptom Score (NBSS) were the most frequently used tools, capturing general health and bladder-specific outcomes, respectively. The Quality of Life Assessment in Spina bifida (QUALAS) measures emerged as a concise, SB-specific tool addressing physical and psychosocial domains. CIC was linked to improved bladder-specific outcomes, though its effect on overall HRQOL was inconsistent. Mental health, literacy, and transition readiness significantly influenced HRQOL.

Conclusions

The use of PROMs in the treatment and research of individuals with SB proved to be surprisingly variable and fragmented. Disease-specific PROMs, such as QUALAS, are an obvious and necessary response to the problem emphasizing the continuous development of these tools to be more tailored to the target group. When complemented by targeted measures like NBSS, they offer a practical and comprehensive approach in understanding and improving HRQOL in SB patients. Optimizing bladder and bowel management, mental health, and social support remains critical for improving outcomes.

Level of evidence

III.

Introduction

SB is a complex congenital condition that affects multiple organ systems, leading to lifelong challenges in mobility, continence, and overall health. Individuals with SB often require multidisciplinary care to address complications ranging from orthopedic and musculoskeletal disorders to neurosurgical, gastrointestinal, and urological dysfunctions [1,2]. Historically, hydrocephalus, urinary sepsis, and renal failure were among the leading causes of morbidity and mortality in this population. However, advancements in medical management, surgical interventions, and structured follow-up care have significantly improved survival, shifting the clinical focus towards optimizing long-term function and quality of life [3–5].

As individuals with SB transition into adulthood, their medical and psychosocial priorities evolve. While mobility limitations, chronic pain, and gastrointestinal issues remain concerns, urological dysfunction, especially neurogenic bladder, continues to be a major source of morbidity and determinant of quality of life. Neurogenic bladder affects 24–98 % of individuals with SB, frequently resulting in overactivity, underactivity, or acontractility of the detrusor and sphincter mechanisms [6–11]. These dysfunctions impact both the filling and emptying phases of bladder function and can lead to complications related to pressure concerns and upper tract sequelae [4,5]. When leak point pressures are elevated, there is an increased risk of urinary tract infections and hydronephrosis.

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Over time, a notable shift in urological morbidity has been observed in this population [12]. Incontinence has emerged as a primary challenge, particularly among adolescents transitioning to independence [13,14].

Most individuals with SB use clean intermittent catheterization (CIC) for bladder emptying and pressure control, with a significant proportion requiring additional interventions to address reduced bladder capacity [15]. The initial treatment usually consists of oral anticholinergics or beta 3 agonists followed by injections of botulinum toxin to manage bladder dysfunction [16]. When injection therapy is insufficient, surgical procedures, including bladder augmentation and bladder neck surgery, are commonly employed to preserve renal function and improve continence outcomes [17]. However, despite these interventions, many continue to experience daily episodes of incontinence, often due to incompetent bladder outlet, even in those who have undergone bladder neck surgery. Notably, there is no clear association between patient characteristics, treatment approaches, and continence outcomes, underscoring the need for individualized management and diligent follow-up as well as better therapies since the response to treatments mentioned above often remain limited [18].

Beyond clinical challenges, individuals with SB face the burden of repeated investigations and numerous interventions. Many symptoms may remain undetected unless specifically addressed. Patient-reported outcome measures (PROMs) allow individuals to convey their symptom burden and HRQOL, which represent distinct but interrelated concepts [19]. Symptom burden refers to the objective presence and severity of functional impairments such as incontinence or difficulty with catheterization. HRQOL reflects the subjective impact of these symptoms on an individual's well-being. While symptom-focused PROMs assess functional status and treatment outcomes, HRQOL tools provide insight into how these impairments affect daily life and overall satisfaction with care. Distinguishing between these domains is important in selecting appropriate PROMs for individuals with SB [20]. While some systematic reviews have examined these topics, clear guidance on optimal use of PROMs is still lacking [21—24].

This systematic review aims to outline the methods for assessing HRQOL through PROMs in individuals with SB and analyze factors influencing these outcomes as reported in the current literature. The primary objective is to detail the approaches for identifying patient-reported outcomes in SB, while the secondary objective is to examine the factors that impact these outcomes according to published evidence. We hypothesize that while multiple validated PROMs exist, their combined use may pose a significant response burden to patients. By systematically mapping these tools and their domains, we seek to identify an approach that balances comprehensiveness with practicality, ensuring that PROMs remain both informative and accessible for patients with SB.

Materials and methods

We conducted a systematic literature review pertaining to the guidelines of the Preferred Reporting Items for Systematic Reviews and Meta-analysis statement (PRISMA) [25]. This systematic review was registered April 17th, 2024 in the International prospective register of systematic reviews (PROSPERO) as CRD42024532303; Patient-reported outcomes of orthopedic, surgical, and urological concerns in Spina Bifida. We used an AI-based tool (Grammarly) to assist with grammar and spelling during the writing process of this systematic review.

Research strategy and data sources

We used PubMed, Scopus and Web of Science (WoS) databases for searches on July 31st, 2024. Search results were restricted to English articles published between 1946 and 2024. The search string used the following keywords:("spinal dysraphism"[MeSH Terms] OR ("spinal"[All Fields] AND "dysraphism"[All Fields]) OR "spinal dysraphism"[All Fields] OR ("spina"[All Fields] AND "bifida"[All Fields]) OR "spina bifida"[All Fields]) AND ("patient reported outcome measures"[MeSH Terms] OR ("patient"[All Fields] AND "reported"[All Fields] AND "outcome"[All Fields] AND "measures"[All Fields]) OR "patient reported outcome measures"[All Fields] OR ("patient"[All Fields] AND "reported"[All Fields] AND "outcome"[All Fields]) OR "patient reported outcome"[All Fields]). The search string yielded 546 articles (135 PubMed, 202 Scopus and 219 WoS).

Inclusion criteria and study selection

The following inclusion criteria were applied according to the PICO systematic review framework (Patient, Intervention, Comparison, Outcome): (P) SB patients over the age of 12; (I) Methods and management to evaluate and improve HRQOL and other outcomes in SB patients; (C) No direct control group, comparisons were made across different management techniques and their impact on outcomes; (O) Improvements in HRQOL and other outcomes focusing on bladder and bowel functioning, mental health, transition readiness, and sexual health. PROMs were to be previously published tools with outcome data provided. Data were extracted manually in increments. Two authors (P.J. & N.P.) independently screened the abstracts of the studies identified through the search string and selected potential articles for full text retrieval. After screening of abstracts and removal of duplicates, 40 articles were approved for full text scrutiny from which two of them were not available in full text. The remaining 38 articles were then sought for retrieval, of from which 17 studies were excluded because an actual PROM was not used, or the study design was a review. From the 21 eligible articles, another five were excluded because of the reasons listed in Fig. 1, yielding a total of 16 articles for the final analysis. Although these excluded studies did not meet the eligibility criteria for the actual patient-reported outcome analysis, they were reviewed to identify additional PROMs. A qualitative analysis was then conducted by listing the measures and assessing their usability and suitability for the target group. As a result, potential PROMs were included in the analysis. A single round of reference checking (snowballing) was conducted by reviewing the reference lists of the included

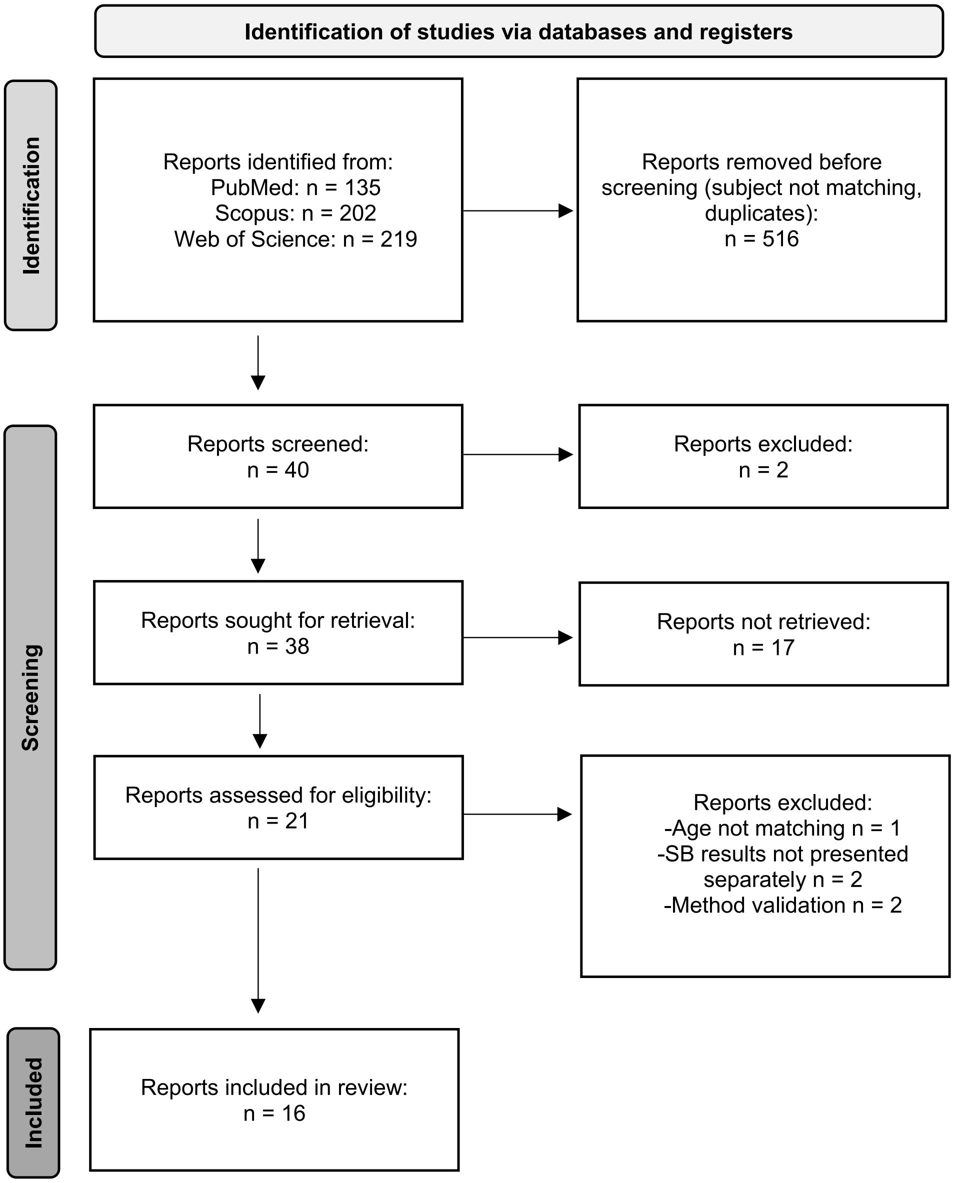

Fig. 1 Flow diagram.

studies to identify additional relevant articles. No further includable studies were found. Disagreements between authors were minimal and no study required resolution by the third author (S.T.).

Data extraction

Two authors independently extracted and documented data from the 16 included studies, focusing on different PROMs and factors influencing the outcomes. The data extraction process involved gathering detailed information on the study design, population characteristics, assessment tools used (e.g., PGH-7, NBSS, SF-36), and the outcomes reported for each tool. Extracted data were recorded in a spreadsheet file, categorizing PROMs into distinct domains such as physical health, mental health, bladder and bowel management, and psychosocial factors. This allowed for a systematic analysis of the tools used and outcomes provided. Discrepancies between the authors' extractions were resolved through discussion and consensus to ensure accuracy and consistency in the data analysis.

Quality assessment

All authors evaluated the quality of the included studies using a set of predetermined criteria focusing mainly on urological outcomes relevant to SB management. These criteria assessed the comprehensiveness of urological reporting, including bladder and bowel management, and their impact on patient-reported outcomes. Additionally, two authors independently assessed the methodological quality of each study using the Newcastle—Ottawa scale (N—O) [26], focusing on selection, comparability, and outcome assessment. Agreement between the authors on the N—O scoring was high, with any discrepancies resolved through discussion and consensus.

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Statistical analysis

Descriptive statistics were used to summarize outcomes across studies. Medians and interquartile ranges (IQRs) were calculated for the percentages of participants exhibiting categorial variables, such as ambulatory status, CIC usage, urinary incontinence, and bowel incontinence. Group comparisons between ambulatory and non-ambulatory participants were performed using the Mann—Whitney U test with statistical significance set at $p < 0.05$. All analyses were performed using available data from the included studies, with any missing data addressed through exclusion from specific statistical tests.

Results

We identified sixteen eligible studies comprising 2009 participants with SB (Table 1) [27—42]. The participants' ages ranged from 12 to 74 years across the studies, with a gender distribution of 59 % female. Most studies distinguished between myelomeningocele (MMC) and non-MMC types, with MMC being the most prevalent, reported in 62 %—80 % of participants. 42 % of the participants had a ventriculoperitoneal (VP) shunt. Functional outcomes were inconsistently reported. For the available data, the median percentage of ambulatory participants was 71 % (IQR: 41 %—78 %). CIC was used by a median of 72 % of participants (IQR: 70 %—100 %). Urinary incontinence affected a median of 48 % of participants (IQR: 45 %—57 %), and bowel incontinence impacted a median of 32 % (IQR: 29 %—54 %), with no significant differences observed between ambulatory and non-ambulatory groups ($p = 1.0$ for both). All participants who reported their level of education had completed at least primary education, with 30 % reporting education beyond secondary school. Most participants responded independently to PROMs, with independence rates ranging from 60 % to 100 %.

Primary outcome measures

In the 16 identified studies, 22 different standardized tools were used to assess patient-reported outcomes in individuals with SB (Table 2). These tools were categorized into two groups (functioning vs subjective experience) as they captured various aspects of health, including general well-being, neurogenic bladder and bowel dysfunction, and readiness for healthcare transitions for example. While no single tool comprehensively addresses all aspects of health in SB, the most used were the Pediatric Global Health 7 (PGH-7) and Neurogenic Bladder Symptom Score (NBSS), each appearing in multiple studies.

The PGH-7 measures general health across physical, mental, and social domains and was used in children, adolescents, and young adults up to 31 years of age. While educational level did not significantly impact PGH-7 scores, higher health literacy, measured by the Brief Health Literacy Screening Tool (BRIEF) and Transition Readiness Assessment Questionnaire (TRAQ) in the two studies by Rague et al. [31,32], was associated with better outcomes, including higher readiness for healthcare transitions. The NBSS specifically evaluates bladder symptoms and their impact on HRQOL, often used to track improvements following interventions like bladder augmentation. The SF-36, employed in three studies, offers a broader physical and mental health perspective, facilitating comparisons between individuals with SB and the general population. Other tools such as TRAQ and BRIEF together with International Index of Erectile Function (IIEF) were used in individual studies, focusing on transition readiness, health literacy, and sexual function, respectively. These tools highlighted the importance of cognitive, behavioral, and functional capabilities in managing healthcare needs, particularly during the transition to adult care. Bladder and bowel management emerged as crucial domains in several studies, with tools like the Neurogenic Bowel Dysfunction score (NBD) and Short Form (SF)-Qualiveen addressing specific challenges in maintaining continence among individuals with SB. Data collection methods varied: two studies used annual hospital visits, two used Facebook advertisements, and others relied on a combination of clinical visits, email, and telephone methods. Some studies did not fully report their recruitment methods.

To better guide clinical use, we eventually analyzed a total of 25 PROMs identified through our review strategy. 14 of the 25 measures were validated for SB and the last three tools, Quality of Life Assessment in Spina bifida (QUALAS) -family of questionnaires, were added as a result of a qualitative analysis performed on the excluded articles. They were not applied in any of the 16 articles included in the patient-reported outcome analysis due to unmatching patient criteria or research setup.

From all the 25 PROMs, target domains, estimated completion times, and validation statuses were collected in Table 3. The number of questions used varied between 4 and 63 with a median of 10 items. We estimated that the time required to complete the measure varies from less than 5 min to 30 min, with most questionnaires being completed in 10 min. Two of the 25 measures were not available for evaluation.

Secondary outcome measures

Several factors influencing HRQOL and the other outcomes among individuals with SB were identified across the included studies. Bladder and bowel management remained the most frequently studied factor, with seven studies highlighting the impact of CIC on patient outcomes. CIC use was associated with fewer bladder dysfunction symptoms and higher bladder-specific quality of life (QoL) compared to other management techniques, although it did not correlate with overall HRQOL [27,29,31,32,34,37,39]. Neurogenic bowel dysfunction was another significant determinant of bowel-specific QoL, with six studies noting a greater impact on bowel-related outcomes than on general HRQOL [28,29,34,38—40].

Transition readiness from pediatric to adult care was examined in three studies, demonstrating that better-prepared patients exhibited improved health outcomes, greater independence, and smoother transitions to adult healthcare [31,32,42]. Higher health literacy was strongly associated with increased readiness for transition, particularly in those performing CIC and those still in school [42]. Mental health was highlighted as a significant factor in five

Table 1 Demographics.

Study	Participants (n)	Mean age (y), [range]	Sex, female	Type of Spina Bifida MMC/non-MMC	V. Shunt/ hydrocephalus	Ambulatory, any degree	CIC	C. channel	Urinary incontinence, any degree	Bowel incontinence, any degree	Level of education, beyond secondary school
Hirsch, J. et al. [27]	146	18 [14–23]	59 %	80/20 %	62 %	77 %	100 %	27 %	64 %	N/A	31 %
Emrich Accioly, J.P. et al. [28]	20	38 [23–50]	55 %	N/A	N/A	25 %	100 %	50 %	N/A	N/A	N/A
Valeska Halstead, N. et al. [29]	173	18 [12–31]	56 %	65/35 %	N/A	67 %	69 %	17 %	49 %	26 %	31 %
Rague, J.T. et al. [30]	20	23 [19–28]	0 %	80/20 %	75 %	75 %	100 %	25 %	N/A	N/A	N/A
Rague, J.T. et al. [31]	226	17 [12–31]	54 %	62/38 %	50 %	81 %	70 %	28 %	47 %	28 %	27 %
Rague, J.T. et al. [32]	200	17 [12–31]	52 %	63/37 %	50 %	83 %	71 %	26 %	47 %	29 %	29 %
Showen, A. et al. [33]	195	40 [18–72]	75 %	N/A	45 %	N/A	N/A	N/A	N/A	N/A	N/A
Ausili, E. et al. [34]	24	N/A [12–17]	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
Davis, M.C. et al. [35]	188	31 [19–74]	63 %	87/13 %	72 %	39 %	N/A	N/A	56 %	57 %	42 %
Cardenas, D.D. et al. [36]	121	25 [15–35]	57 %	N/A	50 %	N/A	38 %	N/A	59 %	53 %	77 %
Mackay, A. et al. [37]	24	25 [18–36]	67 %	83/17 %	N/A	42 %	88 %	N/A	N/A	N/A	N/A
Wagner, R. et al. [38]	72	33 [18–68]	65 %	N/A	65 %	N/A	N/A	28 %	36 %	54 %	25 %
Hacker, E.C. et al. [39]	261	39 [N/A]	76 %	N/A	48 %	N/A	72 %	N/A	N/A	N/A	N/A
Dicianno, B.E. et al. [40]	190	34 [18–77]	54 %	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
Chan, W.M. and B.E. Dicianno [41]	59	34 [N/A]	39 %	100/0 %	100 %	N/A	N/A	N/A	N/A	N/A	24 %
Wood, D. et al. [42]	90	12–25	50 %	79/21 %	N/A	N/A	N/A	N/A	40 %	32 %	N/A

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Table 2 Results.

Study	Design	PROM(s) used	Recruitment	Answered independently/ with assistance	Outcomes
Hirsch, J. et al. [27]	Catheterization via channel vs urethra	[a]1.Pediatric Global Health 7 (PGH-7) [b]2.Neurogenic Bladder Symptom Score (NBSS)	Annual hospital visits	60 %/40 %	Clean intermittent catheterization per channel associated with fewer bladder symptoms but had no significant association with overall health-related quality of life nor bladder-related quality of life.
Emrich Accioly, J.P. et al. [28]	Acquired vs congenital spinal cord injury with neurogenic bladder	[a]1.Decisional Regret Scale (DRS) [c]2.Short-Form Qualiveen (SF-Qualiveen)	E-mail, telephone and through electronic medical record	N/A	Neurognic bladder has worse impact on urologic quality of life for acquired than for congenital spinal cord injury. No differences in decisional regret were seen between the groups.
Valeska Halstead, N. et al. [29]	Differences between bowel management programs	[b]1.Modified Peristeen Neurogenic Bowel Dysfunction questionnaire (NBD) [a]2.Pediatric Global Health 7 (PGH-7)	Routine clinical care visits	62 %/38 %	No significant association between bowel-specific quality of life nor overall quality of life across bowel management programs.
Rague, J.T. et al. [30]	Applicability of the IIEF -questionnaire in young men with spina bifida	[b]1.International Index of Erectile Function (IIEF)	Telephone, $30 incentive for participation	100 %/0 %	Participants reported of severe erectile dysfunction, low orgasmic function and intercourse satisfaction, but high sexual desire.
Rague, J.T. et al. [31]	Association between health literacy and health-related quality of life	[a]1.Pediatric Global Health 7 (PGH-7) [b]2.Brief Health Literacy Screening Tool (BRIEF)	Annual hospital visits	60 %/40 %	Health literacy was associated with health-related quality of life after adjusting for demographic and clinical factors.
Rague, J.T. et al. [32]	Association between health literacy and transition readiness	[b]1.Transition Readiness Assessment Questionnaire (TRAQ) score [b]2.Brief Health Literacy Screening Tool (BRIEF)	As part of standard clinical practice	64 %/36 %	Transition readiness associated with health literacy, even after adjustment for education level and other demographic and clinical factors. Those with low health literacy reported being less ready to transition.
Showen, A. et al. [33]	Depression, anxiety, and social isolation among adults with spina bifida	[b]1.Neurogenic Bladder Symptom Score (NBSS) [a]2.PROMIS Depression 8a + Anxiety 8a + Social isolation Short Forms	Facebook advertisement	N/A	High rates of depression, anxiety, and social isolation was reported. Association between each of these psychosocial phenomena and bladder dysfunction existed.
Ausili, E. et al. [34]	Clinical efficacy of transanal irrigation in children with spina bifida	[c]1.Medical Outcomes Study 36-Item Short-Form Health Survey (SF-36)	N/A	N/A	Sustained improvement in bowel management and quality of life in children with spina bifida or anorectal malformation after transanal irrigation, more significantly in short term than in mid-long term.
Davis, M.C. et al. [35]	Risk factors for permanent disability	[d]1.National Spina Bifida Patient Registry survey	During routine care	N/A	Level of education and degree of stool incontinence are the strongest predictors of "permanent

					disability" among adults with spina bifida.
Cardenas, D.D. et al. [36]	Sexual education and functioning in adolescents and young adults	[a]1.Perceived Quality of Life Scale (PQOL) [c]2.Medical Outcomes Study 36-Item Short-Form Health Survey (SF-36) [a]3.Satisfaction With Life Scale (SWLS)	E-mail	N/A	Individuals with spina bifida do receive sexual education primarily at school rather than from family or physicians. Men were less satisfied with their level of sexual activity.
Mackay, A. et al. [37]	Effectiveness of botulinum toxin type A on neurogenic bladder	[a]1.Incontinence- Quality of Life (I-QOL) score [b]2.Neurogenic Bladder Symptom Score (NBSS)	As part of standard clinical practice	N/A	Botulinum toxin type A significantly improved quality of life as well as end filling pressures and bladder compliance.
Wagner, R. et al. [38]	Presence and impact of secondary conditions on daily life	[a]1.The Spina Bifida Secondary Conditions (SBSC) survey tool	Letter	100 %/0 %	Pain, musculoskeletal conditions, constipation, incontinence, and skin lesions were particularly prevalent and impacting daily life.
Hacker, E.C. et al. [39]	Factors associated with sexual interest and activity	[c]1.PROMIS Sexual Function and Satisfaction Measures Brief Profile	Facebook advertisement	100 %/0 %	The majority of respondents reported interest in sexual activity, though less than half were sexually active recently. Over 80 % of participants who were sexually active, reported high levels of sexual satisfaction.
Dicianno, B.E. et al. [40]	Depressive symptoms in adults with spina bifida	[a]1.Beck Depression Inventory-II (BDI-II) [b]2.Craig Handicap Assessment Reporting Technique SF (CHART-SF)	Annual clinical care visits	N/A	High rate (26 %)of adults with spina bifida considered to have depressive symptomatology. Low mobility scores were significantly associated with these symptoms.
Chan, W.M. and B.E. Dicianno [41]	Virtual socialization and quality of life	[a]1.WHO's Medical Outcomes Study 26-item SF (WHOQOL-BREF) [b]2.Craig Handicap Assessment Reporting Technique SF	N/A	N/A	The effect of virtual socialization on quality of life was positive but not statistically significant. Increased degree of virtual socialization was associated with a greater number of friends.
Wood, D. et al. [42]	Self management skills and applicability of the TRAQ-SB	[b]1.Transition Readiness Assessment Questionnaire Spina Bifida (TRAQ-SB)	N/A	N/A	TRAQ-SB scores negatively associated with bladder incontinence in youth with spina bifida. Stool continence and skin breakdown were not associated with the scores.

PROM assessing.

[a] subjective experience.

[b] Functional outcomes.

[c] Both subjective experience and functional outcomes.

[d] Both subjective experience and functional outcomes, the survey also includes non-PROM data.

Table 3 Tools.

Tool	Number of Questions	Estimated Completion Time (min)	Validated in SB	PROM category	Notes
Pediatric Global Health 7 (PGH-7)	7	<5	Yes	Subjective Experience, Generic	General health domains: Physical, mental, social
Neurogenic Bladder Symptom Score (NBSS)	24	15	Yes	Functional Status, Generic	Bladder-specific; SB-validated
Decisional Regret Scale (DRS)	5	<5	No	Subjective Experience, Generic	Evaluates regret after medical decisions
Short-Form Qualiveen (SF-Qualiveen)	8	<5	Yes	Subjective Experience and functional status, Generic	Targets bladder-specific QoL
Peristeen Neurogenic Bowel Dysfunction Score (NBD)	10	5	Yes	Functional Status, Generic	Focus on bowel-specific outcomes
International Index of Erectile Function (IIEF)	15	<10	No	Functional Status, Generic	Measures male sexual function
Brief Health Literacy Screening Tool (BRIEF)	4	<5	No	Functional Status, Generic	Quick assessment of health literacy
Transition Readiness Assessment Questionnaire (TRAQ)	20	<10	Yes	Functional Status, Generic	Assesses readiness for transition to adult care
PROMIS Depression 8a SF	8	<5	No	Subjective Experience, Generic	Depression-specific scale
PROMIS Anxiety 8a SF	8	<5	No	Subjective Experience, Generic	Anxiety-specific scale
PROMIS Social Isolation SF	8	<5	No	Subjective Experience, Generic	Assesses perceived social isolation
SF-36 (Short Form Health Survey)	36	<20	Yes	Subjective Experience and functional status, Generic	Comprehensive general QoL
National Spina Bifida Patient Registry Survey	N/A	N/A	No	Subjective Experience and functional status (also includes non-PROM data)	Comprehensive SB-specific data collection
Perceived Quality of Life Scale (PQOL)	20	10	No	Subjective Experience, Generic	Quick measure of perceived QoL
Satisfaction With Life Scale (SWLS)	5	<5	No	Subjective Experience, Generic	Assesses general satisfaction with life
Incontinence-Quality of Life (I-QOL)	22	<10	Yes	Subjective Experience, Generic	Targets QoL impacts of incontinence
Spina Bifida Secondary Conditions (SBSC)	63	30	Yes	Functional Status, Ccondition specific	Evaluates secondary conditions in SB patients
PROMIS Sexual Function and Satisfaction Measures Brief Profile	N/A	N/A	No	Subjective Experience and functional status, Generic	Measures satisfaction and function
Beck Depression Inventory-II (BDI-II)	21	10	No	Subjective Experience, Generic	Widely used for depression assessment
Craig Handicap Assessment Reporting Technique SF (CHART-SF)	19	<15	Yes	Functional Status, Generic	Measures social and physical functioning
WHOQOL-BREF (26-item)	26	10	Yes	Subjective Experience, Generic	Covers multiple QoL domains
Transition Readiness Assessment Questionnaire Spina Bifida (TRAQ-SB)	32	15	Yes	Functional Status, Condition specific	Adapted for spina bifida; transition readiness
The QUAlity of Life Assessment in Spina bifida (QUALAS-C)	10	5	Yes	Subjective Experience, Condition specific	Child-specific PROM focusing on physical health and psychosocal well-being
The QUAlity of Life Assessment in Spina bifida (QUALAS-T)	10	5	Yes	Subjective Experience, Condition specific	Teen-specific PROM focusing on physical health and psychosocial well-being
The QUAlity of Life Assessment in Spina bifida (QUALAS-A)	15	>10	Yes	Subjective Experience, Condition specific	Adult-specific PROM focusing on physical health and psychosocial well-being

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studies, showing that depressive symptoms, anxiety, and social isolation negatively impacted HRQOL. Patients with higher levels of depressive symptoms consistently reported lower HRQOL scores, underscoring the importance of addressing mental health in this population [31,33,38–40].

In addition, virtual socialization was associated with improved psychological QoL in one study, although its impact on HRQOL was not statistically significant [41]. Two studies identified educational attainment and social support as significant contributors to HRQOL, with higher education levels and robust social support networks correlating with better HRQOL [35,36].

Sexual health also emerged as a factor influencing HRQOL, with one study reporting severe erectile dysfunction as a significant contributor to poorer sexual health-related outcomes [30]. Finally, surgical interventions, including bladder surgeries and shunt placements for hydrocephalus, were referenced in three studies as positively influencing PROMs and functional outcomes, contributing to enhanced HRQOL [31,37,39].

Quality assessment

The included studies were evaluated using the Newcastle–Ottawa Scale across three domains: selection, comparability, and outcome. Most had well-represented participants, but none included a control group (i.e., a group without SB for comparison), limiting comparability. While many studies controlled for confounders such as age, gender, and socioeconomic factors, some relied on self-reported data (e.g., engagement in virtual socialization platforms), which may introduce reporting bias. Newcastle–Ottawa scores ranged from 4 to 7, with most studies being cross-sectional, limiting the ability to assess long-term outcomes. Overall, methodological quality was moderate (Table 4).

Discussion

Our systematic review of 16 studies with 2009 participants identified a diverse range of PROMs used to evaluate both the functioning outcomes and subjective experience in individuals with SB [27–42]. Tools like the PGH-7 and NBSS were most frequently used, capturing general health and bladder-specific outcomes, respectively. The SF-36, employed in three studies, provided broader insights into physical and mental health, while tools like the TRAQ and BRIEF emphasized transition readiness and health literacy. While these PROMs assess multiple aspects of HRQOL, urinary and bowel incontinence were consistently reported as ongoing challenges across studies, indicating that management strategies may require further optimization. Other factors, including mental health, social support, and sexual health, also played crucial roles in influencing HRQOL.

Selecting appropriate PROMs

Selecting appropriate PROMs is critical to address SB patients' multifaceted needs. While tools like the NBSS, NBD, and PROMIS Depression and Anxiety scales provide valuable insights into specific domains, they often require multiple instruments to achieve a comprehensive assessment. Many of the questionnaires also pose a significant burden to patients due to the extensive number of questions and time required to complete them. This highlights the challenge of balancing comprehensiveness with feasibility, particularly in clinical and research settings. The QUALAS family of PROMs (QUALAS-C, QUALAS-T, and QUALAS-A) is possible solution for primary assessment of SB patients as these tools are specifically designed for SB, encompassing physical and psychosocial domains in a concise, 10–15 question format requiring minimal time to complete [43–45].

Table 4 Quality Assessment (Newcastle–Ottawa scale).

Study	Selection	Comparability	Outcomes	Total
Hirsch, J. et al. [27]	****	**	**	8
Emrich Accioly, J.P. et al. [28]	***	**	**	7
Valeska Halstead, N. et al. [29]	****	**	**	8
Rague, J.T. et al. [30]	***	*	**	6
Rague, J.T. et al. [31]	**	**	**	6
Rague, J.T. et al. [32]	***	*	**	6
Showen, A. et al. [33]	**	**	**	6
Ausili, E. et al. [34]	***	*	***	7
Davis, M.C. et al. [35]	**	**	**	6
Cardenas, D.D. et al. [36]	**	**	**	6
Mackay, A. et al. [37]	***		**	5
Wagner, R. et al. [38]	**		**	4
Hacker, E.C. et al. [39]	**	**	**	6
Dicianno, B.E. et al. [40]	**	**	**	6
Chan, W.M. and B.E. Dicianno [41]	**	**	**	6
Wood, D. et al. [42]	**	**	**	6

One score = *
Maximum score = 9

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By integrating QUALAS with targeted tools like the NBSS for bladder-specific symptoms or the TRAQ for transition readiness, clinicians and researchers can achieve an optimal balance between breadth and feasibility. This approach minimizes patient burden while ensuring meaningful and robust data collection, particularly for longitudinal assessments.

Factors influencing HRQOL

Bladder and bowel management, transition readiness, mental health, and sexual health were consistently linked to HRQOL across the included studies [27,29,32,36]. CIC use, while associated with fewer bladder dysfunction symptoms, did not consistently improve overall HRQOL, highlighting the complexity of bladder management [27]. Similarly, robust bowel management programs reduced neurogenic bowel dysfunction severity but showed variable effects on HRQOL [34]. While factors such as urinary incontinence, catheterization methods, and surgical interventions like bladder augmentation and bladder neck reconstruction influence HRQOL, their effects appear to vary between individuals. For instance, urinary incontinence is consistently associated with poorer HRQOL, whereas catheterization route often does not significantly affect it when other variables are controlled for [27]. This emphasizes the importance of individualized bladder management programs, which may need to extend beyond urological care to address psychosocial and mental health considerations.

Mental health conditions, such as depression and anxiety, were significant contributors to reduced HRQOL, particularly during transitions to adulthood [40]. Higher health literacy and education levels were associated with better outcomes, emphasizing the need for educational support and interventions tailored to individual patient needs [32].

With our review strategy, sexual health remained an underexplored domain. Challenges in sexual function, especially among men, underscore the need for tailored interventions and the development of SB-specific tools to assess this aspect comprehensively [30].

Comparisons to previous work

Previous systematic reviews, such as those by Patel et al. and Bakaniene et al., have highlighted the heterogeneity in PROM use and the underutilization of validated tools like Qualiveen and QoL-BM [23,46]. While these reviews focused on bladder and bowel-specific tools or pediatric populations, our review encompasses a broader age range, including adolescents and adults with MMC and non-MMC SB. This expanded scope provides a more comprehensive perspective, particularly with the inclusion of PROMs addressing transition readiness and mental health. The application of PROMs in SB has evolved over the past decades, reflecting broader advancements in patient-centered outcome research. While earlier studies often employed generic QoL measures, more recent research has increasingly incorporated condition-specific instruments [14,47,48].

Limitations and future directions

Heterogeneity in patient characteristics, recruitment methods, and PROMs across studies posed challenges for direct comparisons. Most studies were cross-sectional, limiting the assessment of long-term outcomes. A notable limitation was the assessment of urinary incontinence as a binary variable (present/absent) in many studies, which does not fully capture its severity, frequency, and impact on QoL. Although our predefined outcomes included sexual function, our search strategy did not explicitly target studies assessing PROMs specific to sexual health. This likely resulted in an incomplete representation of this domain, as only one included study addressed sexual function. The sexual function of individuals with SB has previously been assessed in its own separate review, which concluded that SB -specific instruments are necessary in the future due to the high prevalence and incomprehensive nature of these problems [49]. Additionally, despite using broad search terms, our strategy may have missed relevant studies that were indexed under highly specific keywords. Future research using refined search strategies with targeted terminology may provide a more comprehensive overview. Future research should also incorporate more nuanced measures of incontinence severity and its effect on daily life to improve clinical applicability and patient-centered outcome assessment. Future research should prioritize standardizing PROMs to enhance consistency, incorporate longitudinal designs to capture outcome trajectories and explore the integration of PROMs with patient-reported experience measures (PREMs) [50]. A multidisciplinary approach addressing bladder and bowel management, mental health, education, and sexual health is essential to optimize HRQOL as patients transition to adulthood.

Conclusion

This systematic review mapped the use of PROMs in individuals with SB and identified key factors influencing QoL. While multiple validated tools exist, their combined use may impose a significant response burden. The heterogeneity in PROM application limits comparability across studies, highlighting the need for a structured approach to selection. Standardization, refined incontinence assessment, and longitudinal studies are needed to ensure PROMs remain both comprehensive and practical for patient-centered care.

Ethics approval

This systematic review did not involve any human participants or experimental interventions; therefore, ethical approval was not required.

Consent to participate was not applicable due to the nature of the study which did not involve human participants or experimental interventions.

We adhered to **PRISMA guidelines** for methodology.

This systematic review was registered April 17th, 2024 in the International prospective register of systematic reviews (PROSPERO) as CRD42024532303; Patient-reported outcomes of orthopedic, surgical, and urological concerns in Spina Bifida.

Material from other sources was not used.

Availability of data and materials

The dataset supporting the conclusions of the current study is available from the corresponding author on reasonable request.

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Conflict of interest statement

None of the authors have conflicts of interest to declare.

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