Journal of Pediatric Surgery 60 (2025) 162418

Contents lists available at ScienceDirect

Journal of Pediatric Surgery

journal homepage: www.sciencedirect.com/journal/journal-of-pediatric-surgery

Review Article

Systematic Review on Questionnaires Reporting Bowel Function and Health-Related Quality of Life in Patients With Hirschsprung Disease

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ARTICLE INFO

Article history:
Received 13 January 2025
Received in revised form
14 April 2025
Accepted 10 June 2025

Keywords:
Systematic review
Bowel function
Health-related quality of life
Hirschsprung disease
Long-term

ABSTRACT

Aim: Impaired bowel function and reduced health-related quality of life (HR-QoL) are common after surgery for Hirschsprung disease (HD). There is yet no agreement on how to report these outcomes. This systematic review aims to describe which questionnaires, their properties, and how frequently they are used to report outcomes. Based on this, we recommend questionnaires to be used in future studies.
Methods: Predetermined searches were conducted in Pubmed, Embase, Cochrane Library and Trials, Ovid Medline, and Cinahl with no lower time limit until March 2024. The Covidence software was used in data collection, eligibility criteria and PRISMA guidelines were applied, and the review was PROSPERO-registered.
Results: Of 2925 identified studies, 357 (12 %) met inclusion criteria. Bowel function was reported in 350 studies with 232 different questionnaires. HR-QoL was assessed with 32 different questionnaires in 80 studies. Of the questionnaires, nine (4 %) reporting bowel function and 13 (41 %) reporting HR-QoL questionnaires were validated. The most commonly used bowel function questionnaires were non-validated, developed for patients with anorectal malformations, and without a control population. Most HR-QoL questionnaires were validated and developed for the general population.
Conclusion: Questionnaires to record bowel function in HD patients are mostly non-validated, whereas use of validated questionnaires is the norm when reporting HR-QoL. We recommend either HAQL, BFS or PICS to report bowel function. PedsQL Generic Core Scale and PedsQL Gastrointestinal Symptom Scale or HAQL, and either SF-36 or QLI in combination with GIQLI or HAQL is recommended to report HR-QoL in HD children and adults, respectively.
Level of Evidence: IV.

Abbreviations: AHRQ, The Agency for Healthcare Research Quality; ARM, Anorectal malformation; BCS, Baylor Continence Scale; BFS, Bowel Function Score; CCIS, Cleveland Clinic Incontinence Score; CHQ-CF87, Child Health Questionnaire Child Form 87; DeFeC, Defecation and Fecal Continence questionnaire; ERNs, European Reference Network; ERNICA, European Reference Network for Inherent and Congenital (digestive and gastrointestinal) Anomalies; FIQLS, Fecal Incontinence Quality of Life Scale; FIQOL, Fecal incontinence and constipation quality of life; GIQLI, Gastrointestinal quality of life index; HAQL, Quality of life in patients with anorectal malformations and Hirschsprung disease; HD, Hirschsprung disease; HR-QoL, Health-related quality of life; JSGA, Japanese Study Group of ARM function score; NOS, Newcastle–Ottawa Scale; OASIS, The Holistic Care in Hirschsprung Disease Network Group; PACCT, Paris consensus on Childhood Constipation; PedsQL, The Pediatric Quality of Life Inventory; PICS, Paediatric Incontinence and Constipation Score; POFC, Postoperative fecal continence score; PROSPERO, International prospective register of systematic reviews; PRISMA, Preferred Reporting Items for Systematic Reviews and Meta-Analysis; QLI, Quality of life index; SF-36, Short Form Health Survey 36; TACQOL, The Netherlands Organization for Applied Scientific Research Academic Medical Center (TNO-AZL) Child Quality of Life Questionnaire; VDESS, Vancouver Dysfunctional Elimination Symptom Survey; VSP-A, Adolescents' health and perceived health; WHOHRQOL-BREF, Brief version of The World Health Organization Quality of life Assessment; WHOHRQOL-100, The World Health Organization Quality of Life Assessment 100-item questionnaire.

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https://doi.org/10.1016/j.jpedsurg.2025.162418

1. Introduction

Hirschsprung disease (HD) is a rare congenital colorectal disease affecting around 1 in 5000 births. HD is characterized by the lack of ganglion cells in the distal bowel resulting in bowel obstruction [1]. Most patients are operated on during the first year of life with resection of aganglionic bowel and restoration of bowel continuity. Impaired bowel function is common in HD children as rates of constipation and fecal incontinence are reported in 8–20 % and 3–63 %, respectively [2–5]. Although bowel function may improve as the patients get older, constipation (0–38 %) and fecal incontinence (15–45 %) are still frequent in HD adolescents and adults [5–8]. In addition to impaired bowel function, HD patients may experience reduced health-related quality of life (HR-QoL). Up to 50 % of HD children have reduced HR-QoL, and fecal incontinence is thought to be a major contributor [2,9]. In HD adolescents and adults several studies show that generic HR-QoL is comparable to controls, while gastrointestinal QoL is reduced [10,11].

Frequency and extent of impairment in bowel function and HR-QoL in HD patients vary a lot between studies and patient populations [12–14]. There may be genuine differences, or the different results may be partially explained by different ways of assessing outcomes, for instance by the use of different questionnaires. The inconsistent ways of assessing postoperative outcomes make it difficult to compare surgical techniques, centers, and patient populations as well as inform patients and families about expected outcomes. At present, there is no general agreement on how bowel function and HR-QoL should be evaluated and reported in HD patients. Hence, as a start to agree on which questionnaires to use in HD patients, we performed this systematic review to record which questionnaires that are used for evaluating bowel function and HR-QoL in HD patients. We also aimed to report how frequently the various questionnaires were used and trends over time. Based on this, we recommend questionnaires to be used in future studies.

2. Methods

We conducted a systematic review registered in PROSPERO (Registration number: CRD42020213561) applying the PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analysis) guidelines [15]. This systematic review is generated within the European Reference Network for rare Inherited and Congenital (digestive and gastrointestinal) Anomalies (ERNICA) as the majority of authors are members of the organization.

2.1. Search strategy

PubMed, Ovid Medline, Cochrane Library and Trials, Cinahl, and Embase databases were searched, with assistance from a medical librarian, with no lower time limit on March 22nd 2024. No language or study design filters were used in the searches. References in included studies were also searched. The following search terms were run in PubMed: 1) (Hirschsprung disease OR Hirschsprung's disease OR colonic aganglionosis OR congenital megacolon) AND (bowel function OR functional outcome OR fecal incontinence OR soiling OR constipation) AND (long-term OR long term) and 2) (Hirschsprung disease OR Hirschpsrung's disease OR colonic aganglionosis OR congenital megacolon) AND (health-related quality of life OR HRQoL OR gastrointestinal quality of life) AND (long-term OR long term). For the other databases, similar search terms were applied in line with their indexing systems.

2.2. Eligibility criteria

The inclusion criteria were studies reporting postoperative bowel function and/or HR-QoL in HD patients of all ages and overall sample size $\geq$10 patients. Studies reporting bowel function and/or HR-QoL in selected HD patients, for instance patients following a bowel management program or patients treated for postoperative obstructive symptoms with botulinum toxin injection were included. The exclusion criteria were non-English language, non-human studies, single case reports, opinion articles, reviews, systematical reviews, and meta-analyses.

2.3. Data collection process

The Covidence software was used to simplify and ensure quality in all parts of the data collection process. Covidence enabled the whole review team, consisting of eight authors (ATH, SO, JRD, AM, ALG, CW, LB, and KB), to collaborate seamlessly through an online comprehensive standardized database. The data collection process was divided into four separate steps: Study identification, study screening, assessment for study eligibility, and study inclusion with data extraction. Two reviewers independently screened the titles and abstracts of the retrieved papers. After the initial screening, the full texts were screened in the same fashion as the first step. During full text screening for eligibility, included studies were labeled with sample size and applied questionnaire(s). If there were conflicts during the screening or the eligibility assessment, a third reviewer screened the paper independently to reach consensus. From each study publication year, study type, level of evidence, age and/or follow-up time of patients, and sample size were recorded as well as which bowel function and/or HR-QoL questionnaire(s) that had been used. The bowel function questionnaires were classified according to which symptoms they reported; fecal incontinence, constipation, fecal incontinence and constipation, dysfunctional defecation, and if bowel function impact social function. The HR-QoL questionnaires were classified as reporting generic, gastrointestinal or disease specific QoL. Data from included studies were extracted independently by two reviewers and compared by a third reviewer to ensure the highest standard of accuracy prior to final inclusion.

2.4. Definitions

To be classified as a validated questionnaire, the validation process including testing of psychometric parameters such as reliability and validity had to be available. A description of the validation process should be included in the original paper, preferably with indication of internal consistency with Chronbach's alpha and test-retest reliability with Pearson's correlation coefficient [16].

2.5. Assessment of study quality, design and origin

In systematic reviews, data on patients are collected and study quality commonly assessed by the Newcastle–Ottawa Scale (NOS) and/or The Agency for Healthcare Research Quality (AHRQ) checklist [17,18]. This was not applicable to this study, because the aim was to review questionnaires used to assess outcomes rather than patients' outcomes. Thus, the quality of the included studies

was rated with a modification of the level of evidence used by The Journal of Pediatric Surgery [19]. Level of evidence A includes level 1 studies (randomized control trials or high-quality prospective studies), level of evidence B includes level 2 studies (poor quality prospective or high-quality retrospective studies) and Level C includes level 3 and 4 studies (retrospective cohort study and case series). In addition, we recorded whether the study was prospective or retrospective. Country and continent of origin of the study were determined by the first author's affiliation.

2.6. Statistical analysis

Continuous variables are presented as median with range without decimals. IBM SPSS software for Windows version 25 (Armonk, NY: IBM Corp.) was used.

3. Results

3.1. Study characteristics

The database search identified 2925 potentially relevant studies of which 357 (12 %) met the inclusion criteria and were included in the analysis (Fig. 1). Of the included studies, 277 (78 %) reported only bowel function, seven (2 %) reported only HR-QoL and 73 (20 %) reported both bowel function and HR-QoL (Appendix 1A). In total, 23234 HD patients had answered questionnaires on bowel function and 5750 patients on HR-QoL. Studies reporting bowel function had median 43 (5–814) versus 53 (5–310) patients per study reporting HR-QoL. Validated questionnaires were applied in 40 (11 %) studies on bowel function versus 57 (71 %) studies reporting HR-QoL. A combination of a generic and a gastrointestinal or disease specific HR-QoL questionnaire was found in 16 (20 %) of studies. The majority of high-quality studies and studies using validated questionnaires, were European (Appendix 1B and 1C).

3.2. Bowel function questionnaires

A total of 232 different questionnaires assessing bowel function were identified, and nine (4 %) of these were validated (Tables 1 and 2A). The majority (205; 88 %) of the bowel function questionnaires were study specific. The three most commonly used questionnaires were “Bowel function score” (BFS), “Wingspread” and “Krickenbeck” classification, and of these, only BFS has results from a normal control population. The use of validated bowel function questionnaires has slowly increased during the last three decades (Appendix 2A). The only bowel function questionnaire specifically developed and validated for HD patients was the “Quality of life in patients with anorectal malformation or Hirschsprung disease” (HAQL) questionnaire.

3.3. Health-related quality of life questionnaires

A total of 32 different HR-QoL questionnaires were identified, and 13 (41 %) were validated (Tables 2B and 3). About half of the HR-QoL questionnaires (15; 47 %) were study specific. The use of validated HR-QoL questionnaires has steadily increased during the

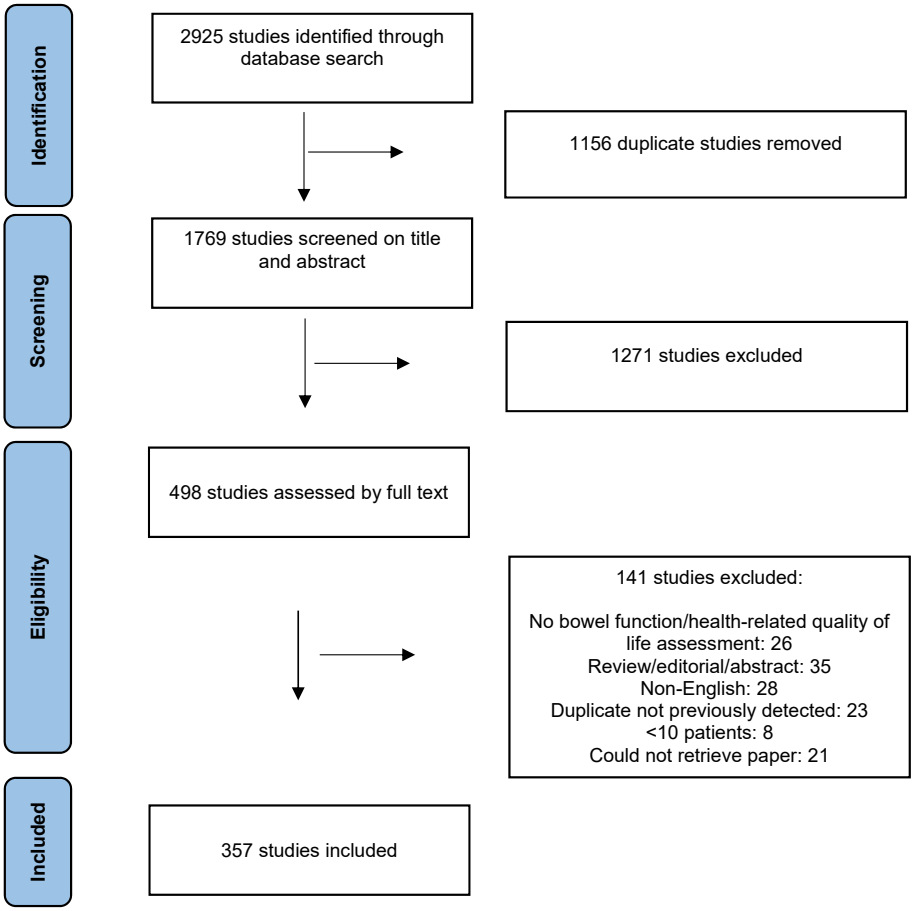

Fig. 1. Flow diagram demonstrating the four steps of identification, screening, eligibility assessment, and inclusion in this systematic review of studies reporting bowel function and/or health-related quality of life in patients with Hirschsprung disease.

Table 1
List of questionnaires used to report bowel function in patients with Hirschsprung disease. The number of studies that have applied the various questionnaires and the number of patients that were assessed are listed.

Questionnare	Validated	Studies	Patients	Age*
	Y/N	(n)	(n)	(Range)
Pediatric population				
Krickenbeck classification[c]	N	23	1127	0–18
Baylor continence scale[a]	Y	4	288	0–18
Heikkinen continence scoring system[a]	N	4	263	0–14
Vancouver dysfunctional elimination symptom survey (VDESS)[d]	Y	2	118	2–18
Teitelbaum continence scoring system[a]	N	2	83	3–10
Paris consensus on childhood constipation (PACCT)[b]	N	3	74	3–18
Wildhaber continence score[a]	N	3	69	2–17
Fecal incontinence and constipation quality of life (FICQOL)[c]	Y	1	58	2–18
Pediatric and adult population				
Bowel function score[c]	N	54	3624	2–51
Wingspread classification[a]	N	22	1720	0–20
Rome constipation criteria[b]	N	16	1599	1–24
Paediatric incontinence and constipation score (PICS)[c]	Y	9	1053	0–20
Holschneider continence score[a]	N	11	719	1–36
Quality of life in patients with anorectal malformations or Hirschsprung disease (HAQL)[c]	Y	10	634	1–77
El-Sawaf bowel function questionnaire[c]	N	5	593	3–41
Gastrointestinal quality of life index (GIQLI)[c]	Y	6	482	4–51
Defecation and fecal continence questionnaire (DeFeC)[a]	Y	3	463	8–45
Wexner constipation score (Cleveland Clinic constipation Score)[b]	Y	10	455	2–38
Kelly continence score[a]	N	8	375	1–36
Rome incontinence criteria[a]	N	1	346	8–45
Templeton and Ditesheim fecal continence score[a]	N	5	215	1–22
Japanese study group of ARM function score (JSGA)[c]	N	4	194	3–37
Postoperative fecal continence (POFC) score[a]	N	2	162	–
Miller incontinence score[a]	N	3	158	2–59
Quality of life score by Bai[a]	N	4	139	5–33
Wexner continence score (Cleveland Clinic incontinence score (CCIS))[a]	N	4	118	1–50
205 individual study specific questionnaires	N	205	12737	0–64
Adult population				
Österberg bowel function questionnaire[c]	Y	2	106	20–59

The superscripts describe which bowel symptom(s) the individual questionnaire report.
[a] Fecal continence.
[b] Constipation.
[c] Fecal continence and constipation.
[d] Other.
* Age-range of patients answering the questionnaire.

last two decades (Appendix 2B). Only the HAQL questionnaire is specifically developed and validated for HD patients.

4. Discussion

This systematic review shows that a huge number of different questionnaires have been used to report bowel function and HR-QoL in HD patients. For assessment of bowel function, study specific questionnaires without a control population were by far most used. In contrast, HR-QoL was usually evaluated by validated questionnaires.

Compared to findings in two recent systematic reviews presenting questionnaires used in pediatric surgery, non-validated questionnaires were applied more often in this review. In one of these reviews, including 38 studies on HD, only 28 % of the questionnaires were study specific [54]. The most likely reason for the discrepancy between this and our review is that we have included far more studies. The other review had excluded questionnaires not demonstrating validity and reliability in the pediatric target population, thus that study did not reflect current practice [55]. Therefore, the present systematic review better presents the actual practice for assessment of postoperative outcomes in HD patients.

Only one bowel function questionnaire, the HAQL questionnaire, is specifically designed and validated for HD patients [41]. Surprisingly, the HAQL questionnaire was used in only 3 % of studies reporting bowel function. We do not know why this questionnaire is so infrequently used, but the large number of questions (39–42 items) could be one explanation. Other plausible reasons are that the HAQL questionnaire has not been available very long or that it is not translated into many languages. The validated "Paediatric Incontinence and Constipation Score" (PICS) questionnaire is the only bowel function questionnaire that reports incontinence, constipation and obstructive symptoms, and is included in the "Hirschsprung disease core outcome set" proposed by the NETS[1HD] study [43,56]. The three most commonly used bowel function questionnaires were the BFS, Wingspread, and Krickenbeck classification [20,38,39]. None of these are validated, and they are designed for patients with an anorectal malformation (ARM). ARM is a different disease than HD, even though ARM patients have many of the same problems as HD patients. Obstructive symptoms due to internal sphincter achalasia are specific to HD, and none of the three abovementioned questionnaires records this symptom. An important advantage of the BFS questionnaire is results from a large control population [57]. Furthermore, the BFS questionnaire is short, and it can be used in both pediatric and adult populations. Studies have shown that easy and short questionnaires are very important for compliance [58,59]. The Holistic Care in Hirschsprung Disease Network Group (OASIS) proposes BFS as their chosen bowel function questionnaire [60].

Most studies reporting HR-QoL used either a validated or a commonly used non-validated questionnaire. Reports on HR-QoL started about two decades later than reports on bowel function, probably reflecting the raised awareness of HR-QoL as a vital outcome parameter. It is well known that generic HR-QoL

Table 2
Description of questionnaires reporting bowel function (Fig. 2A) and health-related quality of life (Fig. 2B) answered by ≥ 100 patients with Hirschsprung disease in this review.

2A	Population[a]	Validated	Validation population	Data from controls	Questions in total	Bowel movement frequency	Obstructive symptoms and/or incomplete emptying	Fecal incontinence[b]	Bowel medication	Social functioning	Mode of answering
		(Y/N)		(Y/N)	(n)	(Y/N)	(Y/N)	(n)	(Y/N)	(Y/N)	Interview (I) Self-administration (S) Not specified (NS)
Fecal incontinence											
Wingspread classification [20]	ARM	N	–	N	4	N	N	4	Y	N	NS
Kelly score [21,22]	ARM	N	–	N	3	N	N	2	N	N	I
Baylor continence scale (BCS) [23]	ARM	Y	ARM children	Y	23	Y	N	6	Y	Y	S
Templeton and Ditesheim fecal continence score [24]	ARM	N	–	N	6	N	N	3	N	Y	NS
Heikkinen clinical continence score [25]	ARM	N	–	N	7	Y	N	3	Y	N	NS
Holschneider continence score [26]	ARM	N	–	N	1	N	N	1	Y	N	I
Quality of life score by Bai [27]	Children with ARM 8–16 years	N	–	Y	6	N	N	2	N	Y	S
Postoperative fecal continence (POFC) score [28]	HD	N	–	N	7	Y	N	1	Y	N	NS
Miller incontinence score [29]	Adults with FI	N	–	Y	3	N	N	3	N	N	I
Wexner continence score (Cleveland Clinic incontinence score) [30]	Adults with FI	N	–	N	5	N	N	4	N	Y	NS
Defecation and fecal continence questionnaire (DeFeC) [31]	General	Y	Healthy adults	Y	88/79*	Y	Y	16	Y	Y	S
Rome incontinence criteria [32]	General	N	–	N	1	N	N	1	N	N	NS
Constipation											
Rome constipation criteria (II-IV) [33–36]	General	N	–	Y	8	Y	Y	0	Y	N	I
Wexner constipation score (Cleveland Clinic constipation score) [37]	Adults with constipation	Y	Adults with constipation	Y	8	Y	Y	0	Y	N	I
Fecal incontinence and constipation											
Krickenbech classification [38]	ARM	N	–	N	3	N	N	1	Y	N	NS
Bowel function score [39]	ARM	N	–	Y	7	Y	N	2	Y	Y	S
Japanese study group of ARM function score [40]	ARM	N	–	N	4	Y	N	2	N	N	NS
Quality of life in patients with anorectal malformations or Hirschsprung disease (HAQL) [41]	ARM and HD	Y	Children >6 years and adults with ARM/HD	N	39-42*	Y	Y	8	Y	Y	S
El-Sawaf questionnaire [42]	HD	N	–	N	15	Y	Y	5	Y	N	I
Paediatric incontinence and constipation score (PICS) [43]	Pediatric	Y	Healthy children	Y	13	Y	Y	3	N	N	NS
Gastrointestinal quality of life index (GIQLI) [44]	Adults with gastrointestinal diseases	Y	Healthy adults and adults with benign or malignant disorders of the gastrointestinal tract	Y	36	Y	N	1	N	Y	S
Dysfunctional elimination											
Vancouver dysfunctional elimination symptom survey (VDESS) [45]	Children with DE	Y	Children with DE	Y	14	Y	N	1	N	N	S

2B	Population[a]	Validated (Y/N)	Validation population	Data from controls (Y/N)	Proxy report (Y/N)	Items in total (n)	Domains (n)	Dimensions	Mode of answering Interview (I) Self-administration (S) Not specified (NS)
Generic									
The Netherlands organization for applied scientific research academic medical center (TNO-AZL) Child quality of life questionnaire (TACQOL) [46]	Pediatric	Y	Dutch children 8–16 years	Y	Y	56-87*	5	Pain and symptoms, basic motor functioning, autonomy, cognitive functioning, interaction with parents and peers	S
The paediatric quality of life inventory (PedsQL) [47]	Pediatric	Y	Pediatric cancer patients	Y	Y	23	4	Physical-, psychological-, social, and school functioning	S
Child health questionnaire Child form 87 (CHQ-CF87, 8–17 years) [48]	Pediatric	Y	Children >10 years	Y	Y	87	10	Physical functioning, role functioning – emotional, role functioning – behavioral, role functioning – physical, bodily pain, behavior, mental health, self-esteem, general health, family cohesion	S
Short-form health survey (SF-36) [49]	Adult	Y	Adults with chronical medical and/or psychiatric conditions	Y	–	36	8	Physical functioning, role-physical, bodily pain, mental health, role-emotional, social functioning, vitality, general health perceptions	S
The world health organization quality of life assessment 100-item questionnaire (WHOHRQOL-100, adult) [50]	Adult	Y	Healthy adults and adults with a variety of diagnoses and varying degrees of severity of disease or disability	Y	–	100	4	Physical capacity, psychological, social relationship, environment	S
The quality of life index (QLI) [51]	Adult	Y	Patients on dialysis	Y	–	64	18	Health care, physical health and functioning, marriage, family, friends, stress, standard of living, occupation, education, leisure, future retirement, peace of mind, personal faith, life goals, personal appearance, self-acceptance, general happiness, general satisfaction	S
Visick score [52]	Gastrectomy in adult men	N	–	N	–	1	–	(Item: Symptom severity)	I
Gastrointestinal									
Fecal incontinence and constipation quality of life (FICQOL) [53]	Children with bifid spine	Y	Children with bifid spine	Y	Y	51	7	Bowel program, dietary management, symptoms, travel and socialization, family relationship, caregiver emotional impact, financial impact	S
Quality of life score by Bai [27]	Children with ARM 8–16 years	N	–	Y	N	6	–	(Items: Soiling, incontinence, school absence, unhappy or anxious, food restriction, peer rejection)	S
Gastrointestinal quality of life index (GIQLI) [44]	Adults with gastrointestinal diseases	Y	Healthy adults and adults with benign or malignant disorders of the esophagus, stomach, gallbladder, pancreas, small intestine, colon, and rectum	Y	-	36	5	Core symptoms, disease-specific items, psychological items, physical items, social items	S
Disease specific									
Quality of life in patients with anorectal malformations or Hirschsprung disease (HAQL) [41]	ARM and HD	Y	Adults and children with ARM or HD	N	Y	39-42*	10–11	Laxative diet, constipating diet, presence of diarrhea, presence of constipation, fecal continence, urinary continence, social functioning, emotional functioning, body image, physical symptoms, sexual function (≥17 years)	S

[a] The population the questionnaire was originally designed for.
[b] Number of questions exploring the listed item.
* Age-specific versions. Numbers in brackets refer to the original paper of the individual questionnaire and can be found in the reference list. ARM: Anorectal malformations. HD: Hirschsprung disease. FI: Fecal incontinence. DE: Dysfunctional elimination.

Table 3
List of questionnaires used to report health-related quality of life in patients with Hirschsprung disease. The number of studies that have applied the various questionnaires and the number of patients that have answered are listed.

Questionnaire	Validated (Y/N)	Studies (n)	Patients (n)	Age* (Range)
Pediatric population				
Visick score[a]	N	5	487	1–18
The Netherlands organization for applied scientific research academic medical center (TNO-AZL) child quality of life questionnaire (TACQOL)[a]	Y	4	402	8–18
Fecal incontinence and constipation quality of life (FICQOL)[b]	Y	2	118	2–18
KIDSCREEN 10 and 52[a]	Y	2	45	4–13
Quality of life score by Barrena[a]	N	2	40	2–17
Adolescents' health and perceived health (VSP-A)[a]	Y	1	15	7–13
Pediatric and adult population				
The pediatric quality of life inventory (PedsQL)[a]	Y	25	1725	0–43
Quality of life in patients with anorectal malformation or Hirschsprung disease (HAQL)[c]	Y	14	1064	1–77
Gastrointestinal quality of life index (GIQLI)[b]	Y	10	635	4–51
Child health questionnaire Child form 87 (CHQ-CF87, 8–17 years)[a]	Y	3	200	8–45
The world health organization quality of life assessment 100-item questionnaire (WHOHRQOL-100, adult)[a]	Y	2	170	8–64
Quality of life score by Bai[b]	N	6	155	5–33
Fecal incontinence quality of life scale (FIQLS)[b]	Y	3	63	7–50
15 individual study specific questionnaires	N	15	976	1–55
Adult population				
Short-form health survey (SF-36)[a]	Y	9	638	16–52
Quality of life index (QLI)[a]	Y	1	310	18–50
Brief version of the world health organization quality of life assessment (WHOHRQOL-BREF, adult)[a]	Y	1	92	23–32

The footnotes describe what the individual questionnaire report:
[a] Generic.
[b] Gastrointestinal.
[c] Disease specific.
* Age-range of the patients answering the questionnaire.

questionnaires may not detect disease specific challenges related to chronic conditions [61]. Therefore, disease specific questionnaires have been developed for many conditions and this trend is also reflected in the HD literature. Generic HR-QoL questionnaires, such as the most frequently used “Pediatric Quality of Life Inventory” (PedsQL Generic Core Scale), dominated the early era when HR-QoL started to be published [47]. The PedsQL Generic Core Scale is validated, has a control population and is relatively short. In later years, an increasing number of publications has included disease specific questionnaires to get a comprehensive evaluation of patients' HR-QoL. However, there is a potential for improvement as only 20 % of studies combined a generic with a gastrointestinal or disease specific HR-QoL questionnaire. This may be the reason why the gastrointestinal HR-QoL questionnaire the “PedsQL Gastrointestinal Symptom Scale”, published already in 2014, could not be identified in this review [62]. Of the gastrointestinal or disease specific HR-QoL questionnaires for children, both the HAQL and PedsQL Gastrointestinal Symptom Scale are validated and have age-specific versions. As expected, the “Short Form Health Survey” (SF-36) was the most commonly used questionnaire for reporting generic HR-QoL in adult HD patients [49]. The advantage of using a commonly used questionnaire is that one can compare results from different patient populations. However, SF-36 may be less valid in younger HD adults, as this questionnaire was validated on adults with chronic medical and/or psychiatric conditions. An advantage of the HAQL questionnaire is that it measures disease specific HR-QoL in addition to bowel function. Thus, by using this particular questionnaire there is no need for two separate questionnaires to evaluate bowel function and HR-QoL. One should be aware of possible limitations if using HAQL, as there are conflicting reports about the questionnaire's validity in children and adolescents [63–65]. GIQLI is used in many publications on adults, and it is therefore possible to compare results from HD patients with that of other patient groups undergoing surgery for benign colorectal diseases [44,66].

We found a considerable variation concerning geographic origin for studies presenting HR-QoL as most of these were European. These findings are in line with a recent review showing that studies on HR-QoL were predominantly from Europe [67]. European studies used validated questionnaires more often than studies from other continents. We can only speculate why studies from Europe generally have a higher tendency to use validated questionnaires and focus on HR-QoL. Influence from the European Reference Network (ERN) for rare Inherent and Congenital (digestive and gastrointestinal) Anomalies (ERNICA) may have been important. It is also possible that the health system in Europe is organized in a way that facilities studies on HR-QoL.

There are several strengths of this systematic review: PROSPERO registration, no lower time limit allowing a large historical search, exhaustive search by a medical librarian, robust data collection through an online database, and many included studies even with strict inclusion criteria. It is a limitation that we did not perform a detailed psychometric evaluation of the identified questionnaires.

The study design did not allow us to decide on which questionnaires are the best at evaluating outcomes in HD patients. Even so, we would like to recommend some questionnaires based on known criteria. The ideal questionnaire should 1) have gone through a validation process, 2) include a validated translation and cultural adaptation to the country it is used in, 3) have a control population in the same country, 4) be user-friendly, and 5) be suitable for different age groups. However, just the fact that a questionnaire is validated does not necessarily mean it is applicable to every study. Studies using questionnaires that have not been validated on the population of interest or in the language they are used may be subject to measurement error [16]. Even though none of the identified questionnaires in this review satisfies all the abovementioned criteria, we would still like to recommend one of the following questionnaires for assessment of bowel function; HAQL, BFS or PICS. HAQL is recommended because it is validated, designed for HD patients, can be used in different age groups, and records both bowel function and HR-QoL. Advantages of the BFS questionnaire include its user-friendliness, that it can be used by both pediatric and adult patients, and that there are results from a

healthy control group. PICS is a validated questionnaire which have questions for different age groups of pediatric patients and it also has questions regarding obstructive symptoms. By using one of these questionnaires and not a study specific questionnaire, comparisons to other studies are possible. Concerning selection of questionnaires to evaluate HR-QoL, we would recommend using both generic and gastrointestinal or disease specific questionnaires to enable a comprehensive evaluation of HR-QoL. We recommend PedsQL Generic Core Scale in HD children and either SF-36 or "Quality of life Index" (QLI) in HD adults to report generic HR-QoL [51]. In combination with a generic HR-QoL questionnaire, we recommend PedsQL Gastrointestinal Symptom Scale in HD children and GIQLI in HD adults to report gastrointestinal HR-QoL. HAQL is applicable to both HD children and adults to report disease specific HR-QoL. These HR-QoL questionnaires are good choices due to all being validated, having control populations, translated into many languages, in addition to being well-known.

Previous communication

Preliminary results were presented at EUPSA 2021.

Category of manuscript

Systematic review.

Funding

The study was funded by the Norwegian Dam Foundation.

Conflict of interest

The authors report no conflict of interest.

Acknowledgments

We would like to thank medical librarian Tarjei Fiskergård Werner for the literature search.

Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.jpedsurg.2025.162418.

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