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ABSTRACT

The objective of this review was to examine disease-specific functioning (ie, faecal functioning and disease-related psychosocial problems) and quality of life (QoL)in patients with anorectal malformations (ARMs) or Hirschsprung's disease across different developmental stages. A search on PubMed yielded 22 eligible studies which were analysed. Patients with ARM or Hirschsprung's disease report slightly more QoL problems than comparison groups. As compared with adolescents, children reported better QoL but worse disease-specific functioning. Relationships between disease-specific functioning and QoL remain unclear. Therefore, to disentangle the complex relationship between the faecal functioning of these patients and their QoL through childhood, adolescence and adulthood, longitudinal studies should examine disease-specific functioning with validated QoL questionnaires that include age-specific versions.

INTRODUCTION

Due to surgical correction in early childhood, the survival rate of patients with anorectal malformations (ARMs) or Hirschsprung's disease has increased to nearly 100%. Surgery is used to remove the non-functioning part of the bowel in order to restore bowel function. For babies with ARM, the nature and severity as well as the presence of associated conditions will determine surgical treatment. Some newborns may need a single operation, while others will require several. The long-term functional outcome of patients with ARM or Hirschsprung's disease depends on the exact anatomy of the birth defect and on the function of the defaecation mechanism after reconstruction.[1 2] Generally, for ARM patients with mild defects, the outcome is usually good and they develop adequate bowel control, although some need medical treatment to avoid constipation. Other patients, mostly those with more complex defects, may never develop adequate bowel control, and have long-lasting functional problems varying from persistent, severe constipation to soiling to complete faecal incontinence.[3 4] Chronic difficulty with long-lasting functional defaecation problems may affect the quality of life (QoL) of patients with ARM or Hirschsprung's disease in all developmental stages.[5–14] It is a challenge for healthcare providers to decrease physical symptoms and improve the QoL of these patients during childhood, adolescence and adulthood.

QoL has become clearly established as an important endpoint in medical care.[15] This is especially true for chronic diseases for which complete recovery is unlikely. Although definitions of QoL vary widely, there is consensus about two central aspects. First, QoL should be regarded as a multidimensional construct incorporating at least three broad domains that can be affected by one's disease or treatment, including physical, mental and social functioning.[16 17] Second, QoL should be assessed from the patient's perspective wherever possible.[17–20] However, as recommendations regarding the minimum age of children administered QoL instruments vary from 7 to 9 years,[20] parents often serve as proxies for younger children.[21]

ARM and Hirschsprung's disease are diseases with different aetiologies that require different surgical and other treatments. However, patients with ARM and Hirschsprung's disease are both born with a chronic disease, both need surgical correction in early childhood and both have problems with faecal functioning (constipation and faecal incontinence). Consequently, the impacts of these diseases and their treatment on patients' QoL and disease-specific (faecal) problems are very similar. We therefore decided to include studies on both patients with ARM and Hirschsprung's disease in this review.

Clearly, problems with faecal continence may impair the QoL of patients with ARM or Hirschsprung's disease. However, only Engum and Grosfeld[22] have reviewed functional outcome and QoL in Hirschsprung's disease patients, but not systematically. An overview of disease-specific functioning in combination with the QoL of patients with ARM or Hirschsprung's disease is lacking, indicating that the overall impact of the disease on QoL through the different developmental stages is unknown. Ideally, clinicians should have a clear picture of the impact of ARM or Hirschsprung's disease on QoL so that they can inform, treat and refer their patients to the care they require.

With this review we have attempted to synthesise the findings of the limited studies available to maximise the information concerning disease-specific functioning (ie, symptoms and disease-related problems) and QoL domains (eg, social problems) across different developmental stages. As relatively few studies employed the above definition of QoL, we also included studies that focused at least on one of the three aspects of QoL (ie, physical, mental or social functioning) in patients with ARM or Hirschsprung's disease.

Our review is guided by three objectives:

(A) to compare QoL domains of groups of patients with ARM or Hirschsprung's

disease with reference groups, and to examine the extent to which disease-specific functioning and QoL differed across age groups

(B) to examine the prevalence of faecal continence problems across different developmental stages and

(C) to examine relationships between disease-specific functioning and QoL.

METHODS

Search strategy

A series of literature searches were conducted on the Medline (PubMed) database for the years 1990 through August 2009. The terms 'anorectal malformation' or 'anorectal malformations' or 'anorectal anomalies' or 'anal atresia' or 'imperforate anus' or 'Hirschsprung's disease' or 'Hirschsprung' were used in combination with 'quality of life' or 'mental' or 'social' or 'psychological' or 'psychosocial' or 'health status' as keywords in title and/or abstract. The first search resulted in 222 abstracts, from which 31 studies were selected using the following inclusion criteria: (A) empiric quantitative studies which (B) focus on at least one of the three generic QoL domains and disease-specific functioning of patients with ARM or Hirschsprung's disease (children, adolescents, adults), (C) were published in or after 1990 and (D) were available in English.[3 8 10 11 13 14 23–47] The search and selection of studies is shown in the flow chart in figure 1. Two of these studies were omitted from this review to prevent similar samples being used. Hence, the 31 identified research reports yielded 29 eligible studies. Additionally, a manual search of the references of the selected articles was carried out, which resulted in one more article.[44] After thorough inspection of these 30 studies, we decided to exclude those[25 30 33 35–37 41 43] that did not include sufficient data to calculate (A) an effect size of the generic QoL between the experimental and comparison groups (objective 1) or (B) a correlation coefficient between QoL and disease-specific functioning (objective 3C), leaving 22 eligible studies [3 8 10–14 23 24 26–29 31 32 34 38–40 42 44 45](see appendix A). We additionally searched in PsycINFO (EBSCOhost interface), Web of Science and ScienceDirect, none of which yielded eligible articles.

Statistics

To correct for small sample sizes, Hedges' d was used to calculate effect sizes. First, to compare the QoL domains in groups of patients with ARM or Hirschsprung's disease with reference groups, we calculated Hedges' d based on standardised differences between mean scores, t statistics or correlations.[48 49] Separate weighted means d+ were calculated for physical QoL, psychosocial QoL and total QoL.[48] According to Cohen, effect sizes d of 0.20, 0.50 and 0.80 can be considered small, medium and large, respectively.[50] To examine the extent to which disease-specific functioning and QoL differed across age groups, effect sizes were calculated for differences between age groups. Second, to examine the prevalence of faecal continence problems across different developmental stages, we presented frequencies of faecal incontinence, constipation and disease-specific psychosocial functioning. Third, to examine relationships between disease-specific functioning and QoL, we presented correlation coefficients. When correlation coefficients were not available, they were derived from means and SD or from t statistics. Correlation coefficients between 0.10 and 0.25 were considered small, between 0.25 and 0.40 medium, and over 0.40 large.[50]

RESULTS

Characteristics and evaluation of the included studies

An overview of the studies, their characteristics and their evaluation are presented in appendix A. Variability and quality differences across the studies were large. Fourteen studies included only patients with ARM, four only patients with Hirschsprung's disease and four patients from both disease groups. Sample sizes ranged from 10 to 341, the male:female ratio varied from 1:1.8 to 4.7:1, mean age ranged from 6 to 16.3 years (range 5 months to 52 years) and age groups varied from infants to adults (of which half included statistics by age group). It was difficult to group the patients according to mutually exclusive age groups because age ranges varied across the included studies. For example, in the study of Ludman *et al*,[38] patients aged between 6 and 11 years were designated as children and patients aged between 12 and 17 as adolescents, whereas in the study of Poley *et al*,[10] patients aged between 5 and 10 years were designated as children and between aged 11 and 15 as adolescents. We therefore employed the following age ranges: infants/toddlers 0–7 years, children 4–11 years, adolescents 10–16 years, adults 15+. Twelve studies included comparison groups and 18 studies included relationships between disease-specific functioning and QoL. In eight studies, QoL

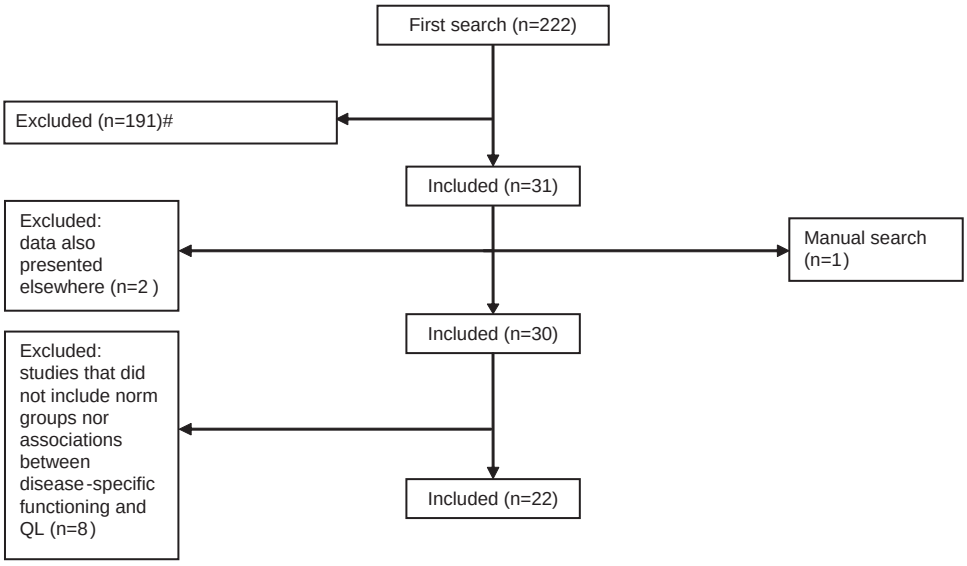

Figure 1 Flow chart of literature search and selection. #Due to the exclusion criteria, studies with the following topics were excluded: studies examining aetiology (n=53), reviews or other types of literature studies (n=31), evaluations of treatment (eg, surgical technique, evaluation of stoma use, evaluation of healthcare) (n=36), studies that mainly examined another disease/syndrome, or where ARM and HD were analysed together with other diseases (n=34), studies examining functional outcome without generic QoL domains (n=13) or other topics (n=24).

was measured by means of six different validated questionnaires (Child Health Questionnaire, Pediatric Quality of Life Inventory, Medical Outcome Study 36-Item, TNO/AZL Child Quality of Life Questionnaire/TNO/AZL Infant Quality of Life Questionnaire, Medical Outcome Study, European Quality of Life). However, usually only one QoL domain was assessed, such as behavioural problems or depression. Disease-specific functioning was assessed with study-specific questionnaires, except for two studies that used the Hirschsprung's Disease/Anorectal Malformation Quality of Life Questionnaires,[14 34] and one study that used the Baylor Social Continence Scale.[27]

Comparisons of QoL between patients with ARM or Hirschsprung's disease and reference groups

The effect sizes at the outcome and domain levels (Hedges' d) of the 12 studies that included comparisons between experimental and reference groups are given in online table 1. As shown in the table, also included were studies which used questionnaires that measured only aspects of QoL, such as self-esteem (as measured with the Self-Perception Profile for Children/Adolescents, SPP-C/A), depression (as measured with the Children's Depression Inventory) or behavioural problems (as measured with the Child Behaviour Checklist). Mean effect sizes of the three QoL domains were calculated from the effect sizes d with the separate effect sizes of the subscales of the questionnaires. For example, in the study of Aasland and Diseth,[23] the mean effects size (Md=0.08) of psychosocial QoL included the effect sizes of the subscales of the SPP-C: scholastic competence (d=0.00), social acceptance (d=0.00), physical appearance (d=0.12), romantic appeal (d=0.00), close friends (d=0.24) and global self-worth (d=0.14). Mean effect size of physical QoL (Md=−0.12) only included the effects size of athletic competence (d=−0.12). Finally, the weighted effect size d+ across these all 12 studies was −0.19 for physical QoL, −0.12 for psychosocial QoL and −0.25 for overall QoL (data not shown). Accordingly, patients with ARM or Hirschsprung's disease reported a lower level of physical, psychosocial and overall QoL than comparison groups, although effects sizes were small.

Differences in QoL and disease-specific functioning across age groups

As there were only two studies that included comparisons between infants/toddlers versus children, and adolescents versus adults, we omitted these comparisons from this review.[10 44] We included effect sizes at outcome and domain levels (Hedges' d) of the six studies that included comparisons between children and adolescents. However, the small study sample did not allow us to calculate the weighted effect size d+.

As shown in online table 2, effect sizes of the three studies that examined differences in physical QoL between children and adolescents[34 38 44] showed that children did better than adolescents (d=0.27,[34] d=0.14[44]) or showed no differences (d=0.03,[34] d=0.04[38]). The studies that examined differences in psychosocial QoL[24 29 34 38 44] revealed similar patterns of results. Children reported better psychosocial QoL than adolescents (d=1.15,[29] d=0.18 and d=0.14,[14] d=0.66[24]) or no differences (d=0.04,[38] d=−0.06[44]). In addition, only one study examined the overall QoL of patients with ARM,[44] which results indicated that adolescents did better than children. The three studies that examined disease-specific functioning were unambiguous: adolescents reported less faecal problems than children (d=−0.10,[10] d=−0.11,[44] d=−0.19[24]).

Prevalence of faecal continence problems

Table 1 shows the prevalence of faecal incontinence, constipation and disease-specific psychosocial functioning. The studies which included multiple age groups, but did not report frequencies by group, showed that frequencies varied between studies from 10% to 78% for faecal incontinence,[3 10–13 32] from 16% to 72% for constipation[12 13 32 42] and from 13% to 80% for disease-specific psychosocial problems.[12 26 32] Results of the studies that examined the prevalence of faecal functioning by age group revealed no clear patterns.[3 8 11 28 38 40]

Relationship between disease-specific functioning and QoL domains

Table 2 shows the associations between disease-specific functioning and QoL domains. Most studies showed that patients with impaired faecal functioning also reported impaired QoL, with correlations varying from small[10 11 14 24 26 34 44] to medium[14 24 29 32 34 44] to large.[10–13 24 26 44] Seven studies reported positive associations without presenting actual data.[3 8 28 32 39 40 45] However, zero or negative relationships were found in four studies,[14 24 38 44] indicating that patients with more faecal continence problems do not report more, or report less, QoL problems. Remarkably, only three studies examined constipation problems in relation to QoL,[14 24 32] which precludes us from presenting any results on possible differences in relationships between constipation and QoL versus faecal incontinence and QoL. Only one study examined disease-specific psychosocial problems in relation to QoL.[14]

Results of the studies that examined relationships between disease-specific functioning and QoL revealed no clear differences in relationships between faecal functioning problems and QoL by age group.[3 8 10 14 24 38 44]

DISCUSSION

On average, patients with ARM or Hirschsprung's disease reported more physical, psychosocial and overall QoL problems than comparison groups, although the effects were small. These results are averaged across all age groups (age ranged from 0 to 52 years) and across both ARM and Hirschsprung's disease patients. We also examined whether disease-specific functioning and QoL differed across age groups. The effect sizes from comparisons between children and adolescents showed that, when differences were found, children with ARM or Hirschsprung's disease reported better QoL than adolescents. In contrast, adolescents reported less faecal problems than children. Apparently, despite fewer faecal symptoms, adolescents experienced more QoL problems than children. It should be noted that Hartman *et al*[34] showed that psychosocial QoL was also poorer for healthy adolescents. To conclude, continence problems seem less in adolescence, however, adolescents with ARM or Hirschsprung's disease seem at more risk for QoL problems than children with ARM or Hirschsprung's disease. Not enough studies were available to allow us to compare infants/toddlers or adults with other age groups.

Patients with ARM or Hirschsprung's disease encountered diverse problems such as faecal incontinence, problematic flatus control, use of diapers, problems with toilet training, increased stool frequency (>5 times a day), muddy foul smelling stools, use of enemas, need for medication (eg, laxatives), abdominal pain, gynaecological and sexual difficulties and marked limitations in social life (peer rejection, difficulties making friends). We sought an insight into disease-specific functioning across the different developmental stages of these

Table 1 Prevalence of disease-specific functioning and QoL of patients with ARM or HD in different age groups

Disease-specific functioning

	ARM				HD			
	Infants/toddlers*	**Children***	**Adolescents***	**Adults***	**Infants/toddlers***	**Children***	**Adolescents***	**Adults***
Faecal incontinence†	10–66%‡[32]	57%[38] ‡[32] 33–68%‡[10]	37–77%§[11] 20–30%[28] 59–65%§[3] ‡[32] 70%[38] ‡[10]	§[11] §[3] ‡[32] ‡[10]	38%[40]	20–35%[40] 13–78%¶[12]	16–32%§[3] 7%[40] ¶[12] 16–32%§[13]	§[3] §[13]
Constipation†	49–62%‡[32]	‡[32] 72%¶[42]	‡[32] 48% 47% ¶[42]	‡[32] §[11] §[3]	2–4%[40]	16% 36%¶[12]	¶[12] 16%§[13] 2%[40]	§[13] §[3]
Problems with psychosocial disease-specific functioning†	80%‡[32]	‡[32] 17–24%¶[26]	70%[28] ‡[32] 50–86%[8] ¶[26]	‡[32] 5–50%[8]		13–66%¶[12]	¶[12]	

*Infants/toddlers: 0–7 years; children: 4–11 years; adolescents: 10–16 years; adults: 15+.
†Umbrella terms: faecal incontinence consists of the subscales impaired faecal control, staining, mucus leak, faecal urgency, difficulty holding back stool, faecal soiling, faecal incontinence, frequent flatus, problems with flatus control, diapers, need to have access to a toilet, toilet trained, increased stool frequency (>5 times a day), muddy foul smelling stools and urological problems. Constipation includes the subscales constipation and enemas, diapers, medication and abdominal pain.
‡Infants/toddlers, children, adolescents and adults were analysed together.
Disease-specific functioning includes the subscales gynaecological and sexual difficulties, marked limitations in social life (peer rejection, problems with making friends), food restrictions, school absence, disease-specific psychosocial problems, (extreme) anxiety and embarrassment, depression and low self-esteem, feelings of frustration, suicidal thoughts, poor appetite, difficulty sleeping, social life (routine school activities, school camps, sleepovers, participation in sports (particularly swimming)).
§Adolescents and adults were analysed together.
¶Children and adolescents were analysed together.

patients by examining the prevalence of faecal functioning problems in infancy/toddlerhood, childhood, adolescence and adulthood. However, most studies only showed the frequencies of multiple age groups which prevented us from drawing conclusions about the prevalence of disease-specific problems by age group. Furthermore, it appeared that these disease-specific problems fluctuated widely within age groups as well as between age groups. Likely, these fluctuations between studies result from the inclusion of study-specific questionnaires instead of validated disease-specific questionnaires. It should be noted, however, that the inclusion criteria meant we had to omit studies that only examined functional outcome. It might be that there are studies available with sound data on disease-specific functioning in patients with ARM or Hirschsprung's disease across different age groups.

Most studies found positive associations between disease-specific functioning and QoL. However, one third of the studies reported only small associations. Moreover, some studies even found negative associations (ie, more symptoms were related to better QoL).[14 24 38 44] Apparently, fewer symptoms do not automatically imply better QoL. From studies by our own research group, it appeared that psychosocial functioning was the most consistent predictor of QoL and changes in QoL of patients with ARM or Hirschsprung's disease in all age groups, whereas disease-specific functioning was not.[14 33–35]

As mentioned in the Methods section, two studies[34 46] used samples that had been published previously.[11 13 47] Although we omitted two studies to prevent using the same samples,[46 47] there were still several reports from different research groups within the same country (eg, three from the Netherlands and five from Norway) that might have collected their data from within the same pool of patients. However, it appeared that the hospitals from which the data were collected only slightly overlapped between studies within the same country, indicating sufficient variability in results. Moreover, as the studies discussed in this review included patients from nine different countries, we conclude that the results are valid internationally. Furthermore, due to the smaller number of studies with patients with Hirschsprung's disease (k=8), the results of the present review might be more applicable for patients with ARM (k=18). Whether QoL differed for ARM and Hirschsprung's disease patients could not be examined in this review as not enough studies were available that included type of disease (ARM vs Hirschsprung's disease) as a variable. Also, other possible moderators such as age, severity of disease, type of surgery or gender, could not be included due to lack of studies that examined these variables in relation to the QoL of patients.

Another problem with reviewing the disease-specific functioning and QoL of these patients arises from the heterogeneity of study methodology across studies, thus making comparison difficult. Only eight of the 22 studies included in this review that claimed to examine the QoL of patients with ARM or Hirschsprung's disease, employed the above consensus definition and used validated generic QoL questionnaires. Most studies focused only on single aspects, such as physical outcome, or addressed related problems, such as psychiatric functioning. Moreover, included studies employed selective samples (eg, limited by age) or small samples. Other limitations were that statistics were often not calculated by age group but were averaged across age groups, sometimes no comparison groups were available, or insufficient data were included to calculate effect sizes or correlations. Finally, different questionnaires were used.

Consequently, future studies should compare QoL and disease-specific functioning between different age groups by using validated generic and disease-specific questionnaires that are currently available and include age-specific versions to

Table 2 Relationship between disease-specific functioning and aspects of QoL: Pearson correlation coefficient, r*

Disease-specific functioning	Quality of life					
	Physical			Psychosocial		
	Children†	Adolescents†	Adults†	Children†	Adolescents†	Adults†
Faecal incontinence	NA(+)¶40 NA(+)**45	¶40 **45	–0.02‡14 ¶40 0.41[10] NA(+)[8]	–0.13 to 0.01[24] 0.82§** to 0.19**[26] 0.48**[12] 0.31**[29] NA(+)¶32 –0.01[38] NA(+)**39 NA(+)**45	NA(+)[28] 0.30–0.40[24] **26 **12 0.58–0.89††[13] NA(+)[3] **29 ¶32 0.09[38] **39 0.20–0.52††11 **45	††11 ††3 ¶32 0.00, 0.02‡14 NA(+)[8] 0.20[10] ††13
Constipation			–0.01‡14	0.15–0.36[24] NA(+)**32	0.31–0.47[24] ¶32	¶32 0.01, 0.15‡14
Disease-specific psychosocial functioning			0.14†‡14			0.14, 0.33‡14
Defaecation total	0.10–0.55[44] 0.26†‡**34	0.15–0.55[44] **34	0.34–0.22[44]	–0.11 to 0.35[44] 0.17[24] 0.16‡**34	0.05–0.35[44] **34 0.44[24]	0.26[44]

*Correlation-coefficients between 0.10 and 0.25 are considered small, between 0.25 and 0.40 medium, and over 0.40 large.[50]
†Children: 4–11 years; adolescents: 10–16 years; adults: 15+.
‡Standardised regression coefficients.
§Correlation coefficients were derived from effects sizes (d).
¶Children, adolescents and adults were analysed together.
**Children and adolescents were analysed together.
††Adolescents and adults were analysed together.
NA(+): r value not available.
Umbrella terms: faecal incontinence consists of the subscales incontinence, soiling, flatus, defaecation desire, and urological symptoms, presence of diarrhoea and constipating diet.
Constipation includes the subscales symptoms, constipation, abdominal pain and laxative diet.
Disease-specific psychosocial functioning includes the subscales gynaecological and sexual functions, emotional functioning and body image.
To facilitate interpretation of the direction of correlations, studies were equalised so that positive correlations indicated that better QoL was associated with better disease-specific functioning (less symptoms) and the other way around: more symptoms were associated with worse QoL. Negative correlations indicated that low disease-specific functioning (more symptoms) was associated with better QoL.

allow comparisons between different age groups. Also comparison groups should be included. Finally, future studies that examine the QoL of patients with ARM or Hirschsprung's disease should examine the effects of possible moderator variables on the QoL of these patients, such as age, gender, severity of disease or type of surgery, to increase the clinical significance of the results.

In general, the results of this review indicated in particular that little is known about the functioning of the youngest patients (infants/toddlers/preschoolers). To provide tailored care for these young patients and their parents, knowledge is needed about faecal functioning and QoL, and about factors that affect young patients' QoL. Only two studies used a longitudinal study design,[33 35] and showed that QoL and disease-specific functioning improve with age and stabilise in adulthood. Clearly, definite conclusions about possible changes in disease-specific functioning and QoL can only be provided with longitudinal studies.

Clinical implications

First, paediatric surgeons who treat patients with ARM or Hirschsprung's disease should be aware of deficits in psychosocial functioning and therefore not only enquire about their patients' physical well-being but also about their patients' emotional and social well-being. This is especial import in adolescents, as they reported lower levels of psychosocial functioning than children. Since our results showed varying prevalence of physical and psychosocial problems, healthcare providers should provide treatments specifically tailored to individual patients. Finally, as most studies found positive associations between disease-specific functioning and QoL, attention should be directed towards alleviating symptoms (eg, reducing faecal continence problems) and improving psychosocial functioning (eg, feelings of embarrassment, feelings of unattractiveness, worries about the future). When clinicians become aware of such and other psychosocial problems, they should refer their patients for further support and treatment.

CONCLUSION

Patients with ARM or Hirschsprung's disease have to learn to live with a variety of symptoms. The prevalence of faecal functioning problems varies across studies and within age groups. In addition, studies comparing faecal functioning and QoL across age groups, or using a longitudinal design, are limited. Therefore, it remains unclear whether these problems remain consistent over time. From the few available studies, it might be concluded that faecal functioning is better in adolescence than in childhood, whereas adolescents tend to report more psychosocial QoL problems. Furthermore, correlations between faecal functioning and QoL are conflicting. To disentangle the complex relationship between faecal functioning

Appendix A Characteristics of the included studies: objective, sample characteristics, outcome measures and evaluation score

Study	Objective	1. Disease 2. Sample size (n) 3. Mean age years (range) 4. Male:female ratio 5. Mild (M); Severe (S): n (%)	Outcome measures Disease-specific functioning, including symptom scores	Outcome measures QoL, including related measures	Evaluation score*
Aasland and Diseth[23]	To explore the SPP-A as an indicator of psychosocial outcome in adolescents with chronic physical disorders	1. ARM 2. n=20 3. 13.6 (12–17) 4. NA 5. M: n=13 (65%); S: n=7 (35%)	–	SPP-A	6
Amae *et al*[24]	To clarify factors that influence the level of depression in Japanese children with ARM	1. ARM 2. n=33 3. NA (6–16) 4. NA 5. M: n=6 (18%); S: n=27 (82%)	Study-specific defaecation score (Japanese Study Group for ARM)	CDI *CGAS*† *CAS*† *CBCL*†/*YSR*†	7
Bai *et al*[26]	To investigate the influence of faecal incontinence on children,s QoL after surgically corrected ARM	1. ARM 2. n=71 3. M=NA(8–16) 4. 2:1 5. M: n=37 (52%); S: n=34 (48%)	Quantitative scoring method (Weilin, 1991[51]) Study-specific disease impact questionnaires Study-specific Quality of Life Questionnaire		4
Bai *et al*[12]	To investigate long-term outcome and QoL after the Swenson's operation for rectosigmoid HD	1. HD 2. n=45 3. M=10.9 (8–16) 4. 4.6:1 5. M: n=45 (100%)	Study-specific stool-functioning questionnaire Clinical scoring system Study-specific Quality of Life Questionnaire		4
Brandt *et al*[27]	To develop and evaluate the Baylor Continence Scale to measure social continence in children after surgical correction of ARMs	1. ARM 2. n=8–34 3. M=6 (5 months to 20 years) 4. 1.25:1 5. M: n=7 (20%); S: n=24 (71%) Unknown: n=3 (8%)	BCS	*PHSCS*† *CHQ*†	6
Diseth *et al*[28]	To evaluate the somatic status, mental health and psychosocial status of adolescents with mild ARM	1. ARM 2. n=10 3. M=14.5 (12–16) 4. 1.5:1 5. M: n=10 (100%)	Wingspread classification Study-specific semi-structured interview	CAS CGAS CBCL/*YSR*	5
Diseth and Emblem[11]	To examine somatic status, mental health and psychosocial adjustment among adolescents with mild and severe ARM, and to study relationships between somatic state and psychosocial functioning	1. ARM 2. n=33 3. Median=15 (12–20) 4. 1.4:1 5. M: n=17 (52%); S: n=16 (48%)	Wingspread classification Flatus continence: VAS	CAS YSR/*CBCL* CGAS	5
Diseth *et al*[13]	To examine whether adolescents with HD have more psychosocial problems than their healthy peers, and to explore possible relationships between somatic and psychosocial variables	1. HD 2. n=19 3. M=15.7 (10–20) 4. 2.2:1 5. M: n=12 (64%); S: n=7 (36%)	Wingspread classification Study-specific constipation items	CAS *CBCL*/YSR CGAS PACS	6
Diseth *et al*[3]	To compare adolescents with HD and mild ARM with regard to bowel function, medical procedures and family/parental factors, and to evaluate the relationships between these variables and mental and psychosocial outcome	1. HD or low ARM 2. n=36 (HD: n=19; ARM: n=17) 3. HD: median 16 (10–20); ARM: median 15 (12–20) 4. HD: 2.2:1; ARM: 1.8:1 5. HD: NA; M ARM: n=17 (100%)	Continence according to Wingspread *Flatus continence: VAS*	CAS *CBCL/YSR*† *CGAS*† *PACS*†	5
Funakosi *et al*[29]	To investigate the psychological status of children with ARM or HD and their mothers so as to develop appropriate psychiatric interventions	1. ARM and HD 2. n=29 (ARM: n=19; HD: n=10) 3. NA (6–16 years) 4. 2.6:1 5. M ARM: n=5 (26%); S ARM: n=14 (64%); HD: NA	Study specific: Japanese Rectum Anal Malformed Meeting	CDI	8
Goyal *et al*[31]	To assess functional outcome and QoL in children with ARM	1. ARM 2. n=62 3. Median=6.7 (4.1–10.3) 4. 1.6:1 5. NA	Study-specific functional outcome scale	PedsQL	7

(continued)

Appendix A Continued

Study	Objective	1. Disease 2. Sample size (n) 3. Mean age years (range) 4. Male:female ratio 5. Mild (M); Severe (S): n (%)	Outcome measures Disease-specific functioning, including symptom scores	Outcome measures QoL, including related measures	Evaluation score*
Hamid *et al*[32]	To determine the long-term outcome of surgery for ARM from the patient's perspective	1. ARM 2. n=84 3. M=10 (3–20) 4. 1.6:1 5. M: n=35 (42%); S: n=49 (58%)	Study-specific Hirschsprung's disease family impact questionnaire	Study-specific psychosocial functioning questionnaire, including HOPES	5
Hartman *et al*[14]	To compare generic QoL between adult patients with ARM versus HD (and within these respective diseases, between those with mild and severe forms, and to compare the QoL of both disease groups with healthy subjects). The second objective was to test the extent to which objective background characteristics of patients with ARM or HD explain generic QoL via mediating disease-specific functioning and psychosocial functioning	1. ARM and HD 2. n=341 (ARM: n=195; HD: n=146) 3. Median ARM=24 (17–52); median HD=23 (16–52) 4. ARM: 1.2:1; HD: 2.9:1 5. M ARM: n=88 (49%); S ARM: n=61 (34%); missing: n=29 (16%); M HD: n=95 (67%); S HD: n=29 (20%); missing: n=18 (13%)	HAQL	SF-36 +† *SMS*† *SSLI*† *ICQ*†	11
Hartman *et al*[34]	To compare QoL and perceived competence between children and adolescents with ARM or HD with those of reference groups, and to test whether patients' QoL is explained by clinical and demographic characteristics via effects of disease-specific functioning and perceived self-competence	1. ARM and HD 2. n=316 (ARM: n=164; HD: n=152) 3. M ARM=12 (8–16); M HD=12 (8–17) 4. ARM, 2:1; HD, 3.9:1 5. M ARM: n=68 (41%); S ARM: n=78 (48%); missing: n=18 (11%); M HD: n=103 (68%); S HD: n= 43 (28%); missing: n=6 (4%)	HAQL	TACQOL SPP	11
Hassink *et al*[8]	To perform a detailed study of patients with severe ARM, reviewing the support they had been given in handling their difficulties, their QoL, and their general and mental health	1. ARM 2. n=58 3. Median=26 (18.1–56.9) 4. 2.1:1 5. S: n=58 (100%)	Holschneider Templeton and Ditesheim Study-specific QoL questionnaire	MOS	7
Ludman *et al*[38]	To assess the emotional and social adjustment of children with ARM in relation to the level of continence achieved, and to assess whether the prevalence of behavioural and emotional disturbance increases during adolescence. To examine whether children with faecal continence fare less well emotionally than those who achieve continence	1. ARM 2. n=160 3. M=11.3 (6–17) 4. 1.3:1 5. M: n=62 (42%); S: n=86 (54%)	Kelly-score Study-specific disease-specific functioning	CBCL *CAS*† *DSRS*† *GHQ*†	9
Ludman *et al*[39]	To compare long-term functional and psychosocial outcomes following surgical treatment for TCA with those in age and gender matched groups of patients with RSA	1. HD 2. n=30 3. Median=12 (7–18) 4. 2.8:1 5. M: n=15 (50%); S: n=15 (50%)	Kelly score Study-specific interview	CBCL DSRS† SPP†	6
Mills *et al*[40]	To investigate the long-term bowel function and QoL of children with HD as they mature	1. HD 2. n=51 3. M=9.8 (3–21) 4. 4.7:1 5. M: n=34 (66%); S: n=17 (33%)	Constipation scoring system Templeton and Ditesheim	PedsQL	8
Ojmyr-Joelsson *et al*[42]	To evaluate the psychosocial effects of severe and intermediate ARM on school-aged children	1. ARM 2. n=25 3. M=10.5 (8–13.9) 4. 1:1.8 5. M: n=13 (52%); S: n=12 (48%)	VAS: study-specific disease-specific questionnaire	CBCL	5
Poley *et al*[10]	To clarify the QoL of survivors of ARM and CDH in a lifetime setting and examine how it compares with that of the general population	1. ARM 2. n=286 3. M=15.1 (1–51) 4. NA 5. NA	Study-specific symptom checklist	TAIQOL TACQOL SF-36	10

(continued)

Appendix A Continued

Study	Objective	1. Disease 2. Sample size (n) 3. Mean age years (range) 4. Male:female ratio 5. Mild (M); Severe (S): n (%)	Outcome measures: Disease-specific functioning, including symptom scores	Outcome measures: QoL, including related measures	Evaluation score*
Stolk *et al*[44]	To investigate the feasibility and validity of a proxy version of the EuroQol in children treated for imperforate anus	1. ARM 2. n=232 3. M=16.3 (5–51) 4. 1.5–1 5. NA	LSQ	EuroQoL TACQOL	10
Tarnowski *et al*[45]	To evaluate cognitive and adaptive behaviour competencies, and to examine the utility of a multivariate model that included child, maternal and family utilitarian and psychological resources as predictors of children's behavioural adjustment	1. ARM 2. n=34 3. M=10.54 (6–16) 4. NA 5. NA	Templeton and Ditesheim	CBCL *WISC*† *WJPB*† *VABS*†	6

Mild ARM/HD: low/short segment (M); severe ARM/HD: high or intermediate/long segment (S).
*The following criteria were developed to evaluate the quality of the study. Questionnaires assessing all three QoL domains were given two points, while questionnaires that assessed only one or two QoL domains were given one point. Non-validated questionnaires scored zero points (criterion A). If both generic and disease-specific types of instruments were included, one point was scored, if only one type was included, zero points were assigned (criterion B). Given the second part of the definition of QoL which states that quality of life "should be assessed from the patient's perspective whenever possible", studies that included only self-reports were allocated two points, studies that partly used self-reports were given one point and studies that only included proxy reports were given zero points, except when patients were too young for self-reports (criterion C). Samples with more than 100 patients, between 51 and 99, between 10 and 50, or less than 10 were given three, two, one, or zero points, respectively (criterion D). If data on infants/toddlers, children, adolescents or adults were presented separately, one point was allocated, if not, zero points were allocated (criterion E). If comparisons with norm groups were included, one point was allocated, if not, zero points were allocated (criterion F). If associations between disease-specific functioning and QoL were included, one point was allocated, if not, zero points were allocated (criterion G). Hence, scores for evaluating the included studies vary from 0 to 11.
†Outcome measures in italic were included in the study concerned but appeared not to be useful for the objectives of this review.
ARM, anorectal malformations; BCS, Baylor Social Continence Scale; CAS, Child Assessment Schedule; CBCL/YSR, Child Behaviour Checklist/Youth Self-Report; CDH, congenital diaphragmatic hernia; CDI, Children's Depression Inventory; CHQ, Child Health Questionnaire; CGAS, Children's Global Assessment Scale, self-description questionnaire; DSRS, Depression Self-Rating Scale; EuroQol, European Quality of Life Questionnaire; GHQ, General Health Questionnaire; HAQL, Hirschsprung's disease/anorectal malformation Quality of Life Questionnaire; HD, Hirschsprung's disease; HOPES, Hunter Opinions Personal Expectations Scale; ICQ, Illness Cognition Questionnaire; LSQ, Langemeijer Stool Questionnaire; MOS, Medical Outcome Study; NA, not available; PACS, Parental Account of Children's Symptoms; PedsQL, Pediatric Quality of Life Inventory; PHSCS, Piers Harris Self-Concept Scale; QoL, quality of life; RSA, rectosigmoid aganglionosis; SES, Self-Esteem Scale; SF-36, Medical Outcome Study 36-Item Short-Form Health Survey; SMS, Self-Mastery Scale; SPP-(C/A), Self-Perception Profile for Children/Adolescents; SSLI, Social Support List Interactions; TCA, total colonic aganglionosis; TACQOL, TNO/AZL Child Quality of Life Questionnaire; TAIQOL, TNO/AZL Infant Quality of Life Questionnaire; VAS, visual analogue scale; VABS, Vineland Adaptive Behaviour Scale; WISC, Wechsler Intelligence Scale for Children Revised; WJPB, Woodstock Johnson Psychoeducation Battery.

and QoL through childhood, adolescence and adulthood, longitudinal studies are needed to examine disease-specific functioning with standardised self-report disease-specific and generic QoL questionnaires that include age-specific versions.

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