Anorectal malformations and Hirschsprung disease
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Qualitative analysis of studies concerning quality of life in children and adults with Anorectal Malformations

Marieke J Witvliet  
Annelie Slaar  
Hugo A Heij  
Alida FW van der Steeg

Abstract

Introduction
Anorectal malformations are relatively common congenital anomalies in pediatric surgery. After definitive surgery constipation, soiling and fecal incontinence are frequently seen problems. QoL can be influenced by these problems. In the last decades QoL has become an important aspect in the treatment and follow-up of patients with anorectal malformations. This has resulted in various reports concerning QoL. In order to deduce whether the drawn conclusions in the different studies are correct and can be used to adjust standard care for patients with an anorectal malformation, a qualitative analysis of the studies was performed.

Material and methods
A literature study was performed in Pubmed, Psychinfo, Web of Science and the Cochrane Library (240 hits). Thirty articles were used, following application of our inclusion criteria and in-depth analysis of the articles. A methodological qualitative analysis was also performed and QoL outcome assessed.

Results
Six authors (20.0%) used validated QoL questionnaires. Four articles were longitudinal and had more than one measure moment. Eleven studies (36.7%) used only non-validated questionnaires, and 8 studies (26.6%) used only validated questionnaires. Nineteen studies correlated fecal continence to QoL, and seven studies established no correlation. Three of these seven studies used validated QoL questionnaires. All twelve studies, which did establish a correlation, used non-validated QoL questionnaires.

Conclusions
Approximately 83% of the studies had not used validated QoL questionnaires. Further, conclusions concerning QoL were often based on functional outcomes, for example fecal incontinence. So far, longitudinal high quality research on QoL in this group has not been achieved.
Introduction

Anorectal malformations are relatively common congenital anomalies in pediatric surgery. The reported incidence across the world varies between 1 in 1000 and 1 in 5000 live births, which means that in The Netherlands about 40 to 50 children are born with this anomaly each year.\(^1\)

Many classifications and surgical treatments have been proposed over the years. Some are based on embryological concepts (e.g. Stephens and Smith)\(^1\) and others are based more on anatomical principles (e.g. Wingspread).\(^2\) In 2005 an international classification system (Krickenberg) was reached by consensus, allowing everyone to talk the same language and to enable comparable follow-up of patients with anorectal malformations.\(^1\)

Fecal incontinence, constipation and soiling are frequent problems following reconstructive surgery for anorectal malformations. These, of course, may affect the QoL of this group of patients.

QoL is a multidimensional concept and in literature many different definitions can be found. The WHO Quality of Life Group has defined QoL as “an individual’s perception of his/her position in life in the context of the culture and value systems in which he/she lives and in relation to his/her goals, expectations, standards, and concerns”.\(^3\)

In other words, QoL is one’s evaluation of his/her functioning in a wide range of areas. Thus, QoL is subjective and refers to satisfaction. In the literature a distinction is sometimes made between QoL and health-related QoL with the latter only concerned with those aspects of life that are directly influenced by one’s health.

A typical questionnaire measuring all aspects of QoL includes six domains: the physical domain, the psychological domain, the social domain, environment, level of independence and spirituality.

Since QoL refers to (dis)satisfaction with functioning it can vary over time depending on life-events or due to coping strategies. Coping strategies may change when growing up with a chronic disease such as an anorectal malformation, and lead to changes in QoL. This may explain why patients who suffer from incontinence and/or soiling, can experience a good QoL.

QoL has become an important outcome measurement in medicine over the past decade and specifically an important aspect in the follow-up of patients with an anorectal malformation. This has resulted in various reports concerning QoL, although such studies often show many different outcomes and have used a wide variation of questionnaires. A qualitative analysis of such studies was performed in order to find out whether the conclusions in the different studies are correct and can be used to adjust for standard care for patients with an anorectal malformation.
Material and Methods

Search Strategy
A search of literature was performed in Pubmed (118 hits), Psychinfo (22 hits), Cochrane Library (0 hits) and Web of science (100 hits). The term “Quality of Life” was used in combination with “anal atresia”, “anorectal malformation” and “imperforate anus”. Cross references were searched and did not identify further relevant citations. After applying the selection criteria (see below) to these articles, 45 articles remained.

Selection criteria
Inclusion criteria were: (1) studies concerning patients with anorectal malformations; (2) QoL is measured with questionnaires; (3) the article was published in peer-reviewed journals, and (4) it was published in English or Dutch. The primary objective of the selected studies was QoL in patients treated for anorectal malformations.

The described inclusion criteria were applied to the initial 240 hits. Forty-five articles met the criteria based on the abstracts, and were included in this review. Hard copies were obtained for all studies.

Quality assessment
The methodological quality of the studies was independently reviewed by two reviewers (MW and AS). A checklist of 20 criteria was used (Table 1). This checklist was based on previous systematic reviews assessing QoL. When an item was scored positive it was assigned one point. The highest possible score was 20 points. After reviewing all articles 15 articles were excluded from further analysis. Some of these articles were only about developing a questionnaire describing fecal incontinence, while others did not measure QoL or described only an operative technique. Official validated QoL questionnaires were TACQoL, PedQL, Kidscreen, CHQ/ITQoL, DUX, SF-36 and WHOQOL. All other questionnaires were scored as validated if a publication concerning validation could be found.

Studies scoring 70% or more of the maximum attainable score (≥ 14 points) were considered to be of “high quality”. Studies scoring between 50% and 70% (10-13 points) were rated as “moderate quality”. Studies scoring lower than 50% (≤ 9 points) were considered as “low quality studies”.
Table 1: List of criteria for assessing the methodological quality of studies on QoL in children and adults with an ano-rectal malformation

Positive if:
QoL assessment:
1. Definition of QoL is provided
2. QoL is assessed with a validated and reliable questionnaire
Study population:
3. A description is given of the initial anomaly
4. A description is given of the surgical treatment
5. A description is given of associated anomalies (VACTERL-association)
6. A description is given of current treatment (bowel-management, medication)
7. Inclusion and/or exclusion criteria are provided
8. Participation rate is >75%
9. METC approval and/or informed consent was given
Study design:
10. A power analysis is provided to account for the number needed to include
11. The collection of data is prospective
12. The design is longitudinal and includes more than one measurement moment
13. Multivariate analysis is done
14. The follow-up period is at least 2 years
15. The loss to follow-up is <20%
16. A comparison of participants and non-participants is done
17. A comparison of dropouts and non-dropouts is done
18. In case of a subgroup, the characteristics of the subgroup are clearly described
Results:
19. The results are compared between 2 groups or more (e.g. healthy population, groups with different intervention therapy, groups with different type of anorectal malformation) and/or results are compared with at least two time points (e.g. longitudinal)
20. Psychometrically sound questionnaires are used to measure QoL and/or functional outcomes

Results

Methodological Quality
The reviewers disagreed on the methodological scoring in 4 articles (13.3%). Scoring difference was 1 point in all 4 articles. These disagreements were solved through discussion in a consensus meeting with all authors. The quality scores ranged from 2 points to 13 points. Mean quality score was 6.8 points. Twenty-six studies were of “low quality” and 4 of “moderate quality”. None of the studies were of “high quality” according to the methodological assessment.
Study characteristics

The main findings of the 30 rated articles are summarized in Table 2. All articles were cohort studies. The cohort size ranged between 20 and 341 patients. Twelve studies had a sample size of 50 or less \(^{21-32}\), 8 studies between 50-100 \(^{33-40}\), 3 studies between 100-200 \(^{41-43}\), and 7 studies had a sample size over 200 \(^{44-50}\). Four of these studies had a longitudinal design and more than one measure moment. \(^{26;32;47;48}\) Six articles used validated QoL questionnaires (20%) \(^{36;46-50}\).

A total of 49 different questionnaires were used in the 30 articles. Twenty-six questionnaires had never been validated in a published validation study. Eight studies (26.7%) used only validated questionnaires. \(^{21;22;41;45-49}\) Only non-validated questionnaires were used in 36.7% of the studies \((N=11)\) \(^{24-27;30;32;34;37-40}\), and in the other 36.6% both validated and non-validated questionnaires were used \((N=11)\). \(^{23;28;29;31;33;35;36;42-44;50}\) An average of 3.2 questionnaires per study was used, with a range of 1 to 7.

The original anomaly was described in 93.3% of the studies, and surgical treatment was described in 43.3%. Five studies gave a definition of QoL in the introduction of their article. \(^{45-49}\)

Findings of the studies

Of the 7 largest studies in this review, 5 studies used validated QoL questionnaires. Hartman et al. concluded that psychosocial functioning had the most important effect on QoL, while fecal incontinence and constipation had almost no effect. \(^{46}\) They also concluded that health care providers should be alert to children and adolescents who were female, with a severe form of disease, or had additional congenital disease, since they reported low levels of perceived self-competence and global disease-specific functioning. \(^{49}\) Children and adolescents did better with increasing age, both on disease-specific functioning and QoL. \(^{48}\) Poley et al found that patients aged 1-4 years old showed a significantly lower health related QoL compared with their healthy peers. This difference was not seen in other age groups. \(^{50}\)

Nine studies were interested in the QoL in adults with an anorectal malformation. In three studies QoL was affected by fecal incontinence \(^{30;38;39}\), while two other studies did not establish a correlation. \(^{25;46}\) Female adults, older patients and patients with other congenital disease or with a stoma reported lower QoL. \(^{47}\)

There is no consensus when comparing children and adults, one study stated that QoL decreased over time whereas the other study groups stated that adults showed better health related QoL compared with children. Self-efficacy is correlated to body image and sexual functioning in adults. \(^{44;45;50}\)

The remaining 21 articles considered either children and/or adolescents. Fourteen of these articles compared the level of QoL, psychosocial problems and depression to the
level of fecal continence. QoL, psychosocial functioning and depression were influenced by fecal incontinence, soiling or constipation in 64.3% (n=9) of the articles. However, none of these studies used a validated QoL questionnaire. The other 5 articles stated that there was no relation, of which 2 used validated QoL questionnaires.

Nineteen studies correlated fecal continence to QoL. Twelve studies did find a correlation whereas 7 studies established no correlation. Three of these 7 studies used validated QoL questionnaires. All 12 studies, which did establish a correlation, used non-validated QoL questionnaires.

**Discussion**

QoL is an entity that is dependent on many changing factors in a person’s life and environment and hence research on QoL is challenging. Definition of QoL is essential and without a clear definition of what is investigated, interpretation of the conclusions is almost impossible.

In this qualitative analysis only 16.7% of the included studies provided a definition of QoL. However, in order to deduct whether the used questionnaires assess QoL, as intended by the authors, and thus test the hypothesis of the study precise definition is necessary.

The construct of the questionnaires used has a profound influence on the outcome of the study. Van der Steeg et al. showed that a questionnaire assessing satisfaction with function (WHOQOL-100) reveals different areas of concern compared with a questionnaire that merely inquires about function in the different domains (SF-36). The first reflects the (dis)satisfaction of a patient and this does not necessarily mirror the functional limitations.

In this review only 6 authors used validated QoL questionnaires. Most other “QoL questionnaires” used concern functional outcome after surgical correction of anorectal malformations, or measure psychosocial aspects and do not contain questions about satisfaction. So, the authors say that they have investigated QoL, but at best they have assessed health related QoL.

Almost all studies, with the exception of 4, measured QoL on one moment in time. QoL, however, when looking at its definition, is not a static concept. It may fluctuate due to certain life events such as changing schools and changes in functioning. In addition coping strategies change during life and this also influences QoL. So in order to assess the development of QoL it is essential to assess a patient at different times in life. Some studies compared different age-groups with each other. Defining QoL as a longitudinal changing concept, which is different for each person, it is technically not correct to compare different age-groups with each other in order to come to conclusions about the development of QoL.
During the last decades the operation techniques have changed. Twelve of the 30 articles mention the operation technique, but the remaining 60% do not. Clearly, different operative techniques might influence outcome in later life. For instance De Vos et al. compared laparoscopic-assisted ano-rectoplasty (LAARP) with the gold standard posterior sagittal ano-rectoplasty (PSARP). They concluded that both techniques can successfully treat patients with an anorectal malformation but that they both have their own specific associated problems. These associated problems and functional outcome can influence health status and (health related) QoL.

In the conclusion of all included articles it is stated that longitudinal research on QoL is essential in order to define the problems in life after treatment of an anorectal malformation. If we have identified the problems and the moments in life in which they occur we can use this to intensify the follow-up of this patient group. Therefore, we plan to start a multicentre, longitudinal and prospective study in the Netherlands using validated questionnaires on the QoL in children with an anorectal malformation.
Table 2: Study characteristics

<table>
<thead>
<tr>
<th>Author (year)</th>
<th>Study size</th>
<th>ARM or ARM and HD</th>
<th>Initial anomaly / surgical treatment</th>
<th>Validated QoL questionnaire</th>
<th>Other questionnaires validated, not validated or both</th>
<th>Definition QoL given</th>
<th>Outcome</th>
</tr>
</thead>
</table>
| Aasland\(^2\)\(1999\) | 20 | ARM + / - | No | Validated | No | - Self-esteem comparable with normal population  
- Global self-worth associated to social acceptance and physical appearance |
| Amae\(^3\)\(2008\) | 66 | ARM + / - | No | Both | No | - Adolescents: bowel dysfunction influences psychology the most  
- Primary school children: psychology of mother influences psychology the most |
| Diseth\(^2\)\(1996\) | 33 | ARM + / + | No | Validated | No | QoL correlates negative with:  
- anal dilatation  
- resting pressure  
- fecal and flatus continence |
| Diseth\(^3\)\(1998\) | 36 | ARM and HD | + / + | No | Both | No | Psychosocial outcome is associated with:  
- continence dysfunction  
- family/parental functioning |
| Ditesheim\(^4\)\(1987\) | 61 | ARM + / + | No | Not validated | No | Early surgical intervention:  
- improved fecal continence  
- improvement in self-esteem and QoL |
| Funakosi\(^5\)\(2005\) | 50 | ARM and HD | + / - | No | Both | No | - Frequency of depression and anxiety among children and their mothers was associated with age of the child |
| Goyal\(^6\)\(2006\) | 62 | ARM + / + | Yes | Not validated | No | - Functional outcome is worse with increasing severity of ARM  
- No difference in QoL between patients and controls |
<table>
<thead>
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<th>Author (year)</th>
<th>Study size</th>
<th>ARM or ARM and HD</th>
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<th>Definition QoL given</th>
<th>Outcome</th>
</tr>
</thead>
</table>
| Grano\(^{44}\) (2008) | 237 | ARM / - | No | Both | No | Parents’ and patients self-efficacy gives:  
- More parental satisfaction on post-operative care  
- Better QoL in adults. |
| Grano\(^{45}\) (2010) | 237 | ARM + / - | No | Validated | Yes | Adult patients reported lower emotional functioning, worse body image and more physical symptoms compared with children |
| Hamid\(^{37}\) (2007) | 84 | ARM + / + | No | Not validated | No | - Children with ARM have ongoing physical and social morbidity  
- Need for continuous multidisciplinary follow-up |
| Hartman\(^{46}\) (2004) | 320 | ARM and HD + / - | Yes | Validated | Yes | Psychosocial functioning had the most important effect on QoL  
- Fecal incontinence and constipation: almost no effect on QoL |
| Hartman\(^{47}\) (2006) | 341 | ARM and HD + / - | Yes | Validated | Yes | - Female and older patients report poorer QoL over time |
| Hartman\(^{48}\) (2007) | 316 | ARM and HD + / - | Yes | Validated | Yes | Disease-specific functioning and mental QoL are better with increasing age |
Qualitative analysis of studies concerning quality of life in children and adults

Table 2: Study characteristics (continued)

<table>
<thead>
<tr>
<th>Author (year)</th>
<th>Study size</th>
<th>ARM or ARM and HD</th>
<th>Initial anomaly / surgical treatment</th>
<th>Validated QoL questionnaire</th>
<th>Other questionnaires, validated, not validated or both</th>
<th>Definition QoL given</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hartman49 (2008)</td>
<td>316 ARM and HD</td>
<td>+ / -</td>
<td>Yes</td>
<td>Validated</td>
<td>Yes</td>
<td>- Female children and adolescents with a severe form of disease, or additional congenital disease: low levels of perceived self-competence and global disease-specific functioning.</td>
<td>- QoL highly dependent on perceived self-competence</td>
</tr>
<tr>
<td>Hashish24 (2010)</td>
<td>48 ARM</td>
<td>+ / +</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>- Stooling score declined with increasing age</td>
<td>- A direct correlation between QoL and stooling score.</td>
</tr>
<tr>
<td>Hassink38 (1994)</td>
<td>58 ARM</td>
<td>+ / -</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>- Education level and relationships are affected by fecal incontinence.</td>
<td></td>
</tr>
<tr>
<td>Hassink41 (1998)</td>
<td>109 ARM</td>
<td>+ / -</td>
<td>No</td>
<td>Validated</td>
<td>No</td>
<td>- Parents have difficulties in coping with implications of the disorder of their child</td>
<td>- Parents of older, male, incontinent children: a higher risk of experiencing child-rearing problems</td>
</tr>
<tr>
<td>Iwai25 (2007)</td>
<td>29 ARM</td>
<td>+ / +</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>- 1/3 of the patients: fecal soiling disturbed their occupational life</td>
<td>- 1/5 of the male reported sexual problems</td>
</tr>
<tr>
<td>John26 (2010)</td>
<td>42 ARM</td>
<td>+ / +</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>- Bowel management in children showed a significant increase in QoL</td>
<td></td>
</tr>
<tr>
<td>Author (year)</td>
<td>Study size</td>
<td>ARM or ARM and HD</td>
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<tr>
<td>Ludman42 (1994)</td>
<td>160</td>
<td>ARM + / +</td>
<td>No</td>
<td>Both</td>
<td>No</td>
<td>- Incontinent children and adolescents not more emotionally disturbed than those with bowel control</td>
<td>- Good family variables predicted a lower prevalence of behavioral problems</td>
</tr>
<tr>
<td>Ludman43 (1995)</td>
<td>160</td>
<td>ARM + / -</td>
<td>No</td>
<td>Both</td>
<td>No</td>
<td>- Children are at risk, regardless of the level of incontinence, for difficulties with psychosocial adjustment.</td>
<td></td>
</tr>
<tr>
<td>Nisell27 (2008)</td>
<td>25</td>
<td>ARM + / -</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>Children and their mothers have a different response on questions about:</td>
<td></td>
</tr>
<tr>
<td>Nisell28 (2009)</td>
<td>25</td>
<td>ARM + / -</td>
<td>No</td>
<td>Both</td>
<td>No</td>
<td>- Psychosocial issues are challenging for children with ARM</td>
<td></td>
</tr>
<tr>
<td>Poley50 (2004)</td>
<td>286</td>
<td>ARM - / - / Yes</td>
<td>Both</td>
<td>No</td>
<td>- Patients aged 1-4 yr: significant lower health related QoL compared with controls</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Rintala39 (1992)</td>
<td>83</td>
<td>ARM + / +</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>Low ARM:</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>- Social disability relating to anal function in 39% of patients</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>- 13% had sexual problems</td>
<td></td>
</tr>
</tbody>
</table>
Qualitative analysis of studies concerning quality of life in children and adults

Table 2: Study characteristics (continued)

<table>
<thead>
<tr>
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<th>ARM or ARM and HD</th>
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<th>Validated QoL questionnaire</th>
<th>Other questionnaires validated, not validated or both</th>
<th>Definition QoL given</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rintala(^{30}) (1994)</td>
<td>33</td>
<td>ARM + / +</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>High or intermediate ARM:</td>
<td></td>
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<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>- Social disability relating to anal function in 85% of patients</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>- 30% had sexual problems</td>
<td></td>
</tr>
<tr>
<td>Tarnowski(^{31}) (1991)</td>
<td>34</td>
<td>ARM + / -</td>
<td>No</td>
<td>Both</td>
<td>No</td>
<td>- Children not at risk for learning problems</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>- Children at risk for behavioral maladjustment</td>
<td></td>
</tr>
<tr>
<td>Yuzuo(^{40}) (2000)</td>
<td>71</td>
<td>ARM + / -</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>- QoL is depressed in children with poor fecal continence.</td>
<td></td>
</tr>
<tr>
<td>Zhengwei(^{32}) (2005)</td>
<td>31</td>
<td>ARM + / +</td>
<td>No</td>
<td>Not validated</td>
<td>No</td>
<td>- Individual biofeedback training: improvement clinical outcome and QoL on short-term and long-term follow-up</td>
<td></td>
</tr>
</tbody>
</table>

\(ARM = \text{Anorectal malformation}; \ HD = \text{Hirschsprung disease}\)
Reference list

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