Pediatr Surg Int (2010) 26:151–155
DOI 10.1007/s00383-009-2541-5

ORIGINAL ARTICLE

Disease-specific quality of life in children and adults with anorectal malformations

Caterina Grano · Dalia Aminoff · Fabio Lucidi · Cristiano Violani

Accepted: 29 October 2009 / Published online: 17 November 2009

Abstract

Purpose The aim of the present study was to analyze disease-specific quality of life, as assessed by the Hirschsprung Disease/Anorectal Malformation Quality of Life (HAQL) questionnaire, in children and adults with anorectal malformations (ARM).

Methods As much as 175 children and 62 adults who were members of the Italian Association for Anorectal Malformations were asked to complete the Italian version of the HAQL questionnaire developed for this study. For children under 16 years of age, mothers were asked to fill up the questionnaires. Patients were also asked to indentify their type of malformation from a list of eight choices.

Results Most subscales of the Italian HAQL had acceptable reliability. Compared to children, adults reported significantly lower levels of QL on subscales measuring emotional functioning, body image, and physical symptoms.

Conclusions Longitudinal studies are needed to clarify whether these results can be attributed to improvements in surgical techniques that have contributed to improved QL in younger cohorts, or if, instead, quality of life in patients with ARM decreases over time. Intervention efforts should focus on bowel management and psychological treatment for ARM-related emotional and body image distress.

Keywords Disease-specific quality of life · ARM · HAQL · Social functioning · Emotional functioning · Urinary continence · Fecal continence

Introduction

A recent focus of outcome research in chronic pediatric disease is the measurement of quality of life (QL) [1]. There are two main types of QL instruments: generic and disease-specific. Generic instruments assess aspects of health and well-being relevant to the general population, whereas disease-specific QL instruments assess those aspects of health and well-being that are associated with specific medical conditions or physical symptoms (e.g. fecal and urinary incontinence) and associated psychosocial functioning [2, 3]. There are advantages and disadvantages in the use of either type of instruments [4], but disease-specific types appear to be more appropriate for populations that share specific conditions [4].

Anorectal malformations are congenital anomalies of the rectum, which require surgical intervention in the neonatal period. They vary in severity from a relatively "simple" covered anus with a rectoperineal fistula to more complex defects, such as cloaca. The post-operative sequelae include medical follow-ups and therapies to control fecal and urinary incontinence.

Several studies have been conducted on QL in patients with anorectal malformations, but few have focused on disease-specific QL [2–4] and none of them have been carried out with Italian patients. Therefore, the first aim of

C. Grano (✉) · F. Lucidi · C. Violani
Department of Psychology, Sapienza University of Rome,
Via dei Marsi, 78, 00185 Rome, Italy
e-mail: caterina.grano@uniroma1.it

F. Lucidi
e-mail: fabio.lucidi@uniroma1.it

C. Violani
e-mail: cristiano.violani@uniroma1.it

D. Aminoff
AIMAR, Italian Parents and Patients Organization for Anorectal Malformations, Via Tripolitania, 211, 00199 Rome, Italy
e-mail: aimar@aimar.eu

the present study was to assess the frequencies of anorectal malformations, using the Krickenbeck classification system, as reported by adult ARM patients and by the mothers of children with ARM and compare them to the frequencies reported in the medical literature. A second aim was to assess the reliability of the subscales of the Italian version of the Hirschsprung Disease/Anorectal Malformation Quality of Life Questionnaire (HAQL). Finally, the third and most important aim of the study was to analyze differences in ARM-related quality of life in children and adults with ARM.

Materials and methods

All of the members ($N = 540$) of the Italian Association for Anorectal Malformations (A.I.M.A.R.) received a letter inviting them to participate in a questionnaire study approved by the ethical committee of the Department of Psychology of the University of Rome Sapienza which was designed to assess quality of life among patients with ARM. Patients present in the AIMAR database include children and adults residing in Italy who were born with anorectal malformations and who underwent surgical procedures for these malformations in surgical centers across the country. Members who returned the questionnaire and signed the informed consent form were included in the study. Informed consent was signed by the patient if he/she was at least 18 years old, and by guardians (most typically mothers) if the patient was under this age. Adult patients completed the adult version of the HAQL questionnaire, while guardians completed the appropriate child version for patients under the age of 16 years.

The HAQL is a 39–42 item, disease-specific QL questionnaire developed by Hanneman et al. [4] in the Netherlands. It assesses 10–11 areas of QL including dietary modification to reach a laxative diet (e.g. eating special food on purpose to get thin stools) or a constipating diet (e.g. eating special food on purpose to get thick stools), the presence of diarrhea (e.g. thin stools more than four times a day), presence of constipation (e.g. thick stools), fecal incontinence (e.g. soiling during the day or night), urinary incontinence (e.g. loss of urine before reaching the bathroom), social functioning (e.g. impaired social activities), emotional functioning (e.g. being ashamed of leaving the classroom to go to the bathroom/feeling embarrassed), physical symptoms (e.g. having abdominal pain), body image (e.g. feeling less attractive), and, for adult patients only, sexual functioning (e.g. less interest in sex). For each item, respondents were asked to indicate the frequency of occurrence in the past week using a 5-point scale raging from 1 (never) to 5 (always).

The different versions of the HAQL questionnaire (i.e. for various age groups under 16 years and for adults) are essentially identical with the following age-related modifications: the social functioning subscale consists of different items for children (e.g. not going to school due to his/her condition) than for adults (e.g. perceived restrictions in social activities); the emotional functioning subscale consists of 6 items in the children's version and 8 items in the adult version, with 5 of these items being identical in the two versions; and the adult version of the HAQL also includes a sexual functioning subscale.

Patients were also asked to report some demographic information and to identify, to the best of their knowledge, the type of their anorectal malformation. To assist with the latter task, patients were asked to choose one alternative from a list of anomalies based on the Krickenbeck classification system on fistulas. This list was taken from the AIMAR questionnaire [5].

Results

Of the 540 questionnaires sent to families, 237 were complete and returned with informed consent, for a response rate of 43.8%. Fifteen questionnaires were missing informed consent and 17 were incomplete, thus, an additional 32 questionnaires were returned that could not be included in the analyses. It should also be noted that the questionnaires were sent to families listed in the AIMAR database by the AIMAR association without confirmation of current mailing addresses and status in the association. Thus, while the response rate was <50%, the authors considered this an acceptable response rate under the circumstances and, more generally, for a mail-based questionnaire survey on such a delicate topic. Moreover, refusals to take part in surveys seem to be rising all over the developed world [6, 7]. Possible causes for this decrease in response rates are reduced amount of free time, increased concerns about privacy and confidentiality, and more survey requests than ever before [8].

Of the completed questionnaires, 62 were completed by patients (59.7% were males) and 175 by parents (61% were parents of sons with malformations, and 38.9% of daughters with malformations). The mean age of the children in the parent sample was 7.3 years ($SD = 4.26$), while the mean age in the adult sample was 24.0 years ($SD = 8.68$).

Classification of malformations

Table 1 summarizes the number of patients who, by self-report, have each type of fistula (using the Krickenbeck classification system). It should be noted that in some cases, patients considered their malformation to be

Table 1 Classification on the type of fistula

	Patients < 16 years old *N* (%)	Adult patients *N* (%)
Imperforate anus	29 (18.4)	5 (9.6)
Perineal fistula	20 (12.7)	8 (15.4)
Rectobulbar urethral fistula	27 (17.1)	12 (23.1)
Rectoprostatic urethral fistula	11 (7.0)	2 (3.8)
Rectobladderneck fistula	17 (10.8)	2 (3.8)
Vestibular fistula	22 (13.9)	2 (3.8)
Vaginal fistula	6 (3.8)	7 (13.5)
Cloaca	18 (11.4)	5 (9.6)
Other	6 (3.8)	6 (11.5)
I don't know	2 (1.3)	3 (5.8)
Total	158 (100)	52 (100)

different from the ones listed (6 parent respondents and 6 adult patient respondents), were not sure how to classify the type of fistula they were born with (2 parent respondents and 3 adult patient respondents), or did not respond to this question (17 parent respondents and 10 patient respondents).

Reliability of the Italian version of the HAQL

Reliability analyses for each of the HAQL subscales were calculated separately for the adult and child samples. Results are reported in Table 2. All the HAQL subscales had acceptable reliability (Chronbach's $\alpha \geq 0.70$), with the exception of three subscales: "laxative diet" (both for adults and children), "constipating diet" (both for adults and children), and "presence of diarrhea" (for the adults). Since the scale "presence of constipation" consists of only one item, it is not reported in Table 2.

Table 2 Reliability for the subscales of the HAQL

	Patients < 16 years old	Adult patients
Laxative diet	*0.56*	*0.21*
Constipating diet	*0.66*	*0.47*
Presence of diarrhea	0.70	*0.57*
Fecal continence	**0.88**	**0.88**
Urinary continence	**0.95**	**0.93**
Social functioning	**0.88**	**0.86**
Emotional functioning	**0.91**	**0.92**
Body image	0.76	**0.87**
Physical symptoms	0.79	0.73
Sexual functioning	–	**0.96**

Values in bold and italic indicate good and poor reliability, respectively, the rest of the values indicate acceptable reliability

Quality of life assessment

Responses to all of the items of the HAQL were first recoded and linearly transformed to a 0 to 100 scale, with higher scores indicating higher, more positive levels of functioning. For example, high scores on the physical symptoms, use of laxative, or constipating diet and presence of diarrhea subscales, would indicate fewer physical symptoms, less need to go on a laxative or constipating diet, and less frequent diarrhea, respectively. Then, for each subscale of the HAQL, the mean of the items on that subscale was calculated separately for the child and adult samples, with the exception of the presence of constipation subscale which consists of only one item and was, therefore, not considered in this analysis.

To test if the differences in the mean for the child and adult samples were statistically significant, a series of analyses of variance were carried out in which the HAQL subscales were considered as dependent variables and the two samples were considered as a between-groups factor (adults and children). These analyses were run for all the HAQL subscales with the exception of "sexual functioning" which is not included in the child version of the HAQL and "social functioning" which has different items in each version and is, therefore, not comparable.

As the results shown in Table 3 indicate, adult patients had significantly lower quality of life scores on three of the HAQL subscales: emotional functioning [$F(1,214) = 40.98$, $p < 0.001$], body image [$F(1,214) = 36.45$, $p < 0.001$], and physical symptoms [$F(1,214) = 7.4$, $p < 0.005$]. Considering the other areas of the HAQL, no other statistically significant differences were found between the two samples, although the mean of the adult sample were lower than the mean of the child sample in all

Table 3 Mean and standard deviations in the ANOVAs for the HAQL subscales in adults and children

HAQL subscales[a]	Patients < 16 years old	Adults patients	*P* value
Laxative diet	73.02 (26.92)	72.08 (28.22)	
Constipating diet	81.98 (24.21)	76.04 (27.16)	
Presence of diarrhea	77.28 (25.39)	76.97 (21.62)	
Fecal continence	79.87 (20.49)	80.55 (18.50)	
Urinary continence	82.56 (29.51)	88.42 (24.01)	
Social functioning	–	–	–
Emotional functioning	85.68 (21.69)	61.80 (30.82)	**
Body image	87.03 (21.80)	63.75 (33.40)	**
Physical symptoms	76.16 (17.67)	68.90 (16.09)	*
Sexual functioning	–	–	

* $p < 0.01$; ** $p < 0.001$

[a] Scores can range from 0 to 100 with higher scores indicating higher levels of functioning

areas except urinary and fecal incontinence, where children had lower scores compared to adults.

Discussion

The first aim of the present study was to assess the frequencies of anorectal malformations, using the Krickenbeck classification system on the type of fistula, as reported by adult patients and by the mothers of children with ARM and to compare them to the frequencies reported in the medical literature. In the present study, a large number of parents reported that their children were born with an imperforate anus (18.4%). This is a much higher number with respect to percentages reported by other authors [9]. Similarly, the percentages of vaginal fistulas reported in our sample, both for children (3.8%) and for adults (13.5%), are very high compared to what is reported in the literature [9]. Some authors have observed that some of the cases diagnosed as vaginal fistulas might be either a vestibular fistula or a cloaca [10], but we do not know whether patients and parents are aware of this possible misinterpretation. In our sample, perineal fistulas were reported in 12.7% of the young patients and in 15.4% of the adult patients. These percentages are lower than the ones recently reported by Hassett et al. [11] in their English sample.

Overall, the distribution of malformations reported by patients and parents in this study was considerably different from those of large groups of unselected patients with anorectal malformations reported elsewhere [9, 11, 12]. It is possible that patients and parents in our sample are not fully aware of the exact type of anomaly that they or their children have and so, may have difficulties in indicating the type of fistula on self-reported questionnaires. This uncertainty might reflect problems in communication between surgeons and families. A low level of awareness about the anomalies might have negative consequences on the expectations parents and patients have concerning the quality of life that can be achieved. To avoid these sorts of misinterpretations, it may be useful to give more precise information about the type of malformation to patients and their families.

The second aim of the present study was to analyze the reliability of the subscales of the Italian version of the HAQL. Most subscales had good or acceptable reliability (Chronbach's $\alpha \geq 0.70$), with the exception of the following three subscales: laxative diet (for both samples), constipating diet (for both samples), and presence of diarrhea (adult sample). Low reliabilities in these subscales have already been reported in the original Dutch version of the HAQL by Hanneman et al. [4] and are partially due to the fact that these scales include only two items.

Finally, the third and most important aim of the study was to analyze differences in ARM-specific quality of life in Italian ARM patients, comparing, in particular, QL scores for children with ARM and adult patients with ARM. With respect to the differences between the child and adult samples, adult patients reported significantly lower emotional functioning, as well as more problems in the areas of body image and physical symptoms. Compared to what parents reported about their children in the area of emotional functioning, adult patients more frequently felt embarrassed or ashamed about their condition, felt different and less appreciated by others or were afraid that others might smell that they were soiling.

Considering the area of body image, adult patients more frequently felt less attractive and dissatisfied with their bodies. Most likely, this result is due to the fact that awareness about the physical self, and the value one attributes to it, develops over time. Adult patients are likely to be more aware of the way their condition affects their body image, and as a consequence, they might be less satisfied with their bodies. However, this finding may also be due to the difficulty mothers might have in commenting on their children's body image, which is, by definition, a subjective concept. As a consequence, they might underestimate possible body image distress in their children.

Considering the physical symptoms subscale, adult patients reported significantly more physical symptoms compared to children. They more frequently felt a swollen abdomen and reported difficulties in discriminating between air and feces or in losing feces. Furthermore, they also reported more frequent air in the abdomen, abdominal pain, and bowel movements. The greater frequency with which adult patients reported these symptoms compared to the children may be related to the fact that they had their surgical interventions many years ago. While the children in this sample may have benefitted from recent advances in surgical techniques, the adults did not.

However, as previously noted, while the data from adults are self-reported, the data from the children are proxy reported by their mothers. We cannot exclude, then, the possibility that in self-reports, people judge their quality of life worse than mothers do for their children.

Future longitudinal studies might clarify whether the results we found can indeed be attributed to cohort effects that reflect changes in surgical procedures, or if, instead, ARM-related quality of life simply decreases as children get older.

A limitation of this study was the lack of access to the medical records of the patients. This prevented us from considering whether differences exist between the severity of the anomalies and various aspects of quality of life in children and adults. Future studies are needed to further analyze this issue.

Finally, from a practical point of view, some intervention suggestions might be proposed. In addition to offering bowel management programs [13] to decrease the frequency of physical symptoms, psychological intervention may be offered to adult patients to help them better cope with ARM-related emotional concerns and their body image [14, 15].

Acknowledgments The authors and AIMAR would like to thank the Johnson & Johnson Foundation for the funds received for this project as well as the patients and parents who participated in this study.

References

1. Mills LA, Konkin DE, Milner R, Penner JC, Langer M, Webber EM (2008) Long-term bowel function and quality of life in children with Hirschprung's disease. J Pediatr Surg 43:899–905
2. Hartman E, Oort F, Mechteld V et al (2005) Explaining change over time in quality of life of adult patients with anorectal malformations or Hirchsprung's disease. Dis Colon Rectum 49:96–103
3. Trajanovska M, Catto-Smith AG (2005) Quality of life measure for fecal incontinence and their use in children. J Gastroenterol Hepatol 20:919–928
4. Hanneman M, Sprangers ME, De Mik EL et al (2001) Quality of life in patients with anorectal malformations or Hirschsprung's disease: the development of a disease-specific questionnaire. Dis Colon Rectum 44:1650–1660
5. Aminoff D, La Sala E, Zaccara A (2006) Follow-up of anorectal anomalies: the Italian parents' perspective. J Pediatr Surg 41:837–841
6. Groves RM, Couper MP (1998) Nonresponse in household surveys. Wiley, New York
7. Groves RM, Dillman DA, Eltinge JL, Little RJA (2002) Survey nonresponse. Wiley, New York
8. Singer E, Mathiowetz N, Couper M (1993) The impact of privacy and confidentiality concerns on survey participation: the case of the 1990 U.S. Census. Public Opin Q 57:465–482
9. Peña A, Hong A (2000) Advances in the management of anorectal malformations. Am J Surg 180:370–376
10. Levitt MA, Peña A (2005) Outcomes from the correction of anorectal malformations. Curr Opin Pediatr 17:394–401
11. Hassett S, Snell S, Hughes-Thomas A, Holmes K (2009) 10-year outcome of children born with anorectal malformation, treated by posterior sagittal anorectoplasty, assessed according to Krinkenbeck classification. J Pediatr Surg 44:399–403
12. Cushieri A (2001) Descriptive epidemiology of isolated anal anomalies: a survey of 4.6 million births in Europe. Am J Med Genet 103:207–215
13. Bischoff A, Levitt MA, Bauer C, Jackson L, Holder M, Peña A (2009) Treatment of fecal incontinence with a comprehensive bowel management program. J Pediatr Surg 44:1278–1283
14. Helman CG (1995) The body image in health and disease: exploring patients' maps of body and self. Patient Educ Couns 26:169–175
15. Drewett RF, Corbett SS, Wright CM (2006) Physical and emotional development. appetite and body image in adolescents who failed to thrive as infants. J Child Psychol Psychiatry 47:524–531