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Follow-up of anorectal anomalies: the Italian parents' and patients' perspective

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Abstract

Background: Several studies addressed the long-term follow-up of anorectal anomalies (ARM) in relation to clinical issues (eg, continence) and quality of life.

However, most of these studies are based upon questionnaires designed by physicians and/or health-care professionals, which may be sources of bias.

Methods: To investigate whether parents of children (patients themselves or older children or adults) who were born with ARM had the perception that they received appropriate care and follow-up, a survey was carried out in Italy, in 2003, among families with children with ARM. A 20-item questionnaire was mailed to 425 patients and parents listed in the AIMAR (Italian association for anorectal malformation) database and was returned by 209 families.

The questionnaire covered different aspects of ARM: type of malformations and surgery, associated anomalies, fecal and urinary continence, as well as aspects about information given to the parents and satisfaction of care and follow-up received.

Result: The patients and parents demonstrated a good understanding of distribution of malformations and their anatomical classification; nevertheless, 67% of responders had to travel outside their living area for surgery. Bowel management (BM) was commonly used among subjects; however, a significant percentage of patients using regular enemas were still soiling (58 patients were clean and 116 soiled).

Urinary continence problems were mostly found in females with cloaca; nevertheless, 21 male patients reported occasional dribbling of difficult interpretation. Most subjects were provided with a good explanation about their or their child's malformation at time of reconstructive surgery, but the same level of information was missing about functional prognosis later in life when the need of an appropriate psychologic support was also felt.

Conclusions: Patients and parents born with ARM are generally satisfied with the information received and with the short-term postreconstructive follow-up care.

At longer follow-up, although more than a quarter of patients are completely clean, there is a significant percentage of subjects who still soil while following a BM program. This is explained by the small number of nurses and BM specialists who are involved in the rehabilitation process

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and by the lack of appropriate information about functional prognosis that are conveyed to the parents. In this respect, psychologic support in bridging the gap between cure and care may be critical.

Anorectal anomalies (ARMs) still are one of the most challenging problems in pediatric surgery; in a recent study, their prevalence was estimated at 4.05 per 10,000 births [1]. Not surprisingly, many studies have been done over the years regarding the long-term follow-up of different surgical techniques [2-4].

However, all these studies mostly invariably carry some limitations; first of all, they are all conducted by health-care professionals with questionnaires and personal interviews in which complacency effects may be sources of bias [5]; secondly, results vary just as much as in the very subjective classification into good, satisfactory, and sufficient continence, thereby making comparisons very difficult.

In the recent years, increased attention to the problems of families of children with ARM [6], along with a progressive implementation of Web educational materials for medical sciences, have led to the creation of many nonprofit support and resource organizations for families of children born with imperforate anus, cloaca, and any of the associated defects.

AIMAR (Italian association for ARM) was founded in 1994 and has now more than 500 members from all over Italy. AIMAR provides matching services for members who request networking with families in their geographic area and with a similar diagnosis and has a board of medical advisors including pediatric surgeons, pediatric surgical nurses, and bowel management (BM) specialists. AimarNews is the quarterly newsletter of the association. The articles range from medical information, personal stories, and helpful information to the various issues faced by families of a child born with an ARM. Moreover, a guide for parents as well as a BM program booklet is distributed free of charge to members and families. To investigate the perception regarding the various short and long-term aspects of surgical care and follow-up, a survey was conducted in 2003, by AIMAR, among parents of children or patients themselves (older children or adults) who were born with an ARM and associated defects.

1. Material and methods

A 20-item, multiple-choice questionnaire was sent to all parents and patients in the AIMAR database. This database includes patients residing in Italy who were operated on for different types of ARM in surgical centers across the country.

Questionnaire used was a simplified version of a specific questionnaire, which was used in the Netherlands [7] in cooperation with the Dutch association of patients with ARM. To comply with AIMAR Privacy Policy, members who agreed in writing to give out information were included in the study. The questionnaire was sent by mail. If patients did not respond, they were reminded with a telephone call. When the necessary language skills and cognitive abilities were present, patients themselves were asked to fill out the questionnaire; otherwise, the questionnaires were filled out by one or both parents (one questionnaire per family). Patients were also asked to answer all questions only on the basis of what they had perceived from the medical records and/or from caring physicians.

The questionnaire was divided into 4 sections:

1. Demographics and information about surgery (place, type of operations) (5 items);
2. Level of malformation and type of fistula according to anatomy (1 item for females and 1 item for males); associated defects (1 item), most relevant clinical variables such as control over defecation and/or micturition, use of therapeutic aids (enemas, laxatives, diapers) (6 items); degree of continence was subjectively assessed;
3. Quality of information given to the parents (both after reconstructive surgery and at long term) (5 items);
4. Satisfaction about care and follow-up received (2 items).

All questions were phrased in common language; questionnaires had to be filled out anonymously, and none of the medical advisors was involved in any step of the study; all responders were made aware of that when mailing the questionnaire. The entire questionnaire is present on the AIMAR Web site (http://www.romacivica.net/aimar/). Medical advice was sought only when preparing the manuscript.

2. Results

Questionnaires were sent to 425 patients and parents and were returned by 209 families for a response rate of 49%. The questionnaire was filled out by 123 mothers (59%), by 38 fathers (18%), and, in 13% (27) of cases, by both parents. In 10% (21) of cases, it was filled out by affected individuals.

Patients' sex distribution was as follows: females 37% and males 63%, with a mean age of 11 years (range, 1-45 years).

Initial operation consisted of posterior sagittal (95), perineal (26), abdominoperineal (71), or sacroabdominoperineal (11) approach. In 6 patients, the exact approach cannot

Table 1 Type of malformation, fistula, and associated defects in males and females

Type of malformation	Female		Male		Fistula	Female		Male		Associated malformations	Female		Male	
	n	%	n	%		n	%	n	%		n	%	n	%
Anal stenosis	4	5	4	3	Imperforate anus no fistula	14	18	24	18	None	25	33	42	32
Anal atresia	6	8	40	8	Perineal	11	14	18	14	VATER	7	9	13	10
Low ARM	23	30	17	13	Vestibular	23	30			Cranio/facial	1	1	3	2
Intermediate ARM	8	10	26	20	Vaginal	18	24			Cardiac	8	11	16	12
High ARM	9	12	71	53	Bulbar urethra			34	26	Genetic	6	8	7	5
Cloaca	22	29			Rectoprostatic			13	10	Orthopedic	28	36	36	27
Others	5	6	4	3	Bladder neck			16	12	Neurologic	14	18	36	27
					I don't know	6	8	26	19	Urogenital	45	59	73	55
					Others	5	6	1	1	Gastroesophageal	5	7	8	6

be traced. Level of malformations and type of fistula for males and females and associated defects are detailed in Table 1.

Sixty-seven percent of responders had to travel outside their area of residence for surgery, whereas 33% had surgery performed at a local medical center.

Regarding fecal continence and soiling, 50 patients (25%) considered themselves continent and 116 subjects (64%) reported soiling; of these, 82 patients (45%) soiled less than 3 times a week, whereas 34 (19%) soiled more than 3 times a week. Fifteen responders (8%) were totally incontinent, whereas 6 have a permanent stoma. Twenty-eight did not answer this question.

Enemas were used by 90 subjects (43%); 29 of these were in the continence group, whereas 61 were in the soiling group. Fifteen patients in the continence group were clean with medical or dietary treatment only, whereas the other 14 reported no treatment.

Of the 55 remaining patients who soiled, 43 were using some form of medical or dietary treatment, whereas 12 reported no therapeutic aid. Overall, 143 patients of 209 were on some form of treatment.

Patients were also asked about BM program [8]: 73 (35%) subjects reported they were currently using BM, 119 subjects (57%) reported they were not informed about BM, and 17 (8%) reported they did not understand the question. Bowel management program was started at an average age of 5 years and was reported to improve significantly the quality of life in 58% of users; another 26% of patients reported minor improvements, whereas 12% reported no changes. A further 4% responded "I don't know."

Complete urinary continence was reported in 146 patients (70%); occasional dribbling was reported in 38 (21%) of subjects; of these, 16 were females (13 cloaca and 3 vestibular fistula) and 21 were males (4 with recto–bladder neck [RBN] fistulae and 17 with less severe forms of ARM). Twelve patients reported that they were totally incontinent: 8 were females (all cloacas) and 4 were males (RBN fistulae). Thirteen patients did not respond.

Results of survey in relation to the information given to the parents and the satisfaction with care and follow-up are shown in Tables 1-6.

3. Discussion

Follow-up of ARM is long established, and this report, rather than being a comprehensive analysis of functional outcome after repair of ARMs, represents a view over what is going on in the patients' families.

As previously mentioned, this enables to eliminate some significant biases when interviewing parents who may feel uncomfortable reporting suboptimal outcomes to the pediatric surgeon.

Response rate was rather high (49%) for a mailed questionnaire in such a delicate matter; we speculate that this might be because of a high level of motivation of patients and their families; 209 responders represent one of the largest follow-up series on ARM.

Table 2 Results of questionnaire for follow-up: "Who offered postoperative assistance to you or your child?"

Surgeon who performed the primary operation	84	40%
Other surgeons	25	12%
Urologist	17	8%
Pediatrician	27	13%
Psychologist	8	4%
Stomatherapist/BM specialist	4	2%
Others	44	21%

Table 3 Results of questionnaire regarding perception of information given: "Were you given an explanation about your child's malformation prior to surgery?"

Yes	115	55%
Not completely	63	30%
No	27	13%
I don't know	4	2%

Table 4 Results about functional prognosis: "At the time of the surgical reconstruction were you informed about the functional prognosis?"

Yes	115	55%
Not completely	59	28%
No	31	15%
I don't know	4	2%

A remarkable percentage of responders (67%) complained of limited or no local surgical specialist services and had to rely on services provided only in metropolitan centers. This resulted in significant time lost from work and travel expenses.

However, only 29% of patients reported a type of malformation (cloaca or RBN fistulae) for which a large experience in the management, a high level of expertise concentrated only in few tertiary referral centers, is mostly invariably required [9]; this underscores the necessity of regional centers where most of the pediatric surgeons can be trained to repair satisfactorily less complex malformations, thereby reducing personal costs and difficulties incurred by remote residents.

Regarding anatomy and type of fistulae, their distribution in males and females reflects that of larger series and demonstrates a good understanding by patients and parents of the classification, which is currently considered the most appropriate [10].

Interestingly, there is a striking discrepancy between percentage of malformations perceived as "high," "intermediate," and "low" by patients and parents and anatomy and type of fistula; in particular, in males, 53% of anomalies were reported as high compared with only a 12% of RBN fistulae. It is likely that these conflicting information conveyed to the parents reflect the still persisting difficulties of assessment of these anomalies among physicians in different centers.

The exceedingly high number of vaginal fistulae (24%) confirms some authors' observations that some of the cases diagnosed as vaginal fistulae might actually be either a vestibular fistula or a cloaca [11]. Whether or not parents and patients are aware of this possible misinterpretations and of its repercussions deserves further investigation.

As far as continence is concerned, 25% of patients considered themselves as continent, whereas 64% of them reported soiling. As previously mentioned, the degree of continence was admittedly subjective; scoring systems were not used in the questionnaire, neither were continence results stratified according to type of surgical repairs.

Table 5 Results about prognostic accuracy: "Was the prognosis accurate?"

Yes	134	64%
Not completely	50	24%
No	17	8%
I don't know	8	4%

Table 6 Satisfaction after surgical reconstruction: "Were you satisfied with the care and follow-up you or your child received after the surgical reconstruction?"

Category offering care and follow-up	Very satisfied	Not satisfied	Care/follow-up not offered
Surgeon who performed the primary operation	165 (79)	33 (16)	10 (5)
Other surgeons	130 (62)	23 (11)	56 (27)
Urologist	71 (34)	23 (11)	115 (55)
Pediatrician	106 (51)	54 (26)	50 (24)
Psychologist	38 (18)	17 (8)	155 (74)
Stomatherapist/BM specialist	205 (98)	15 (7)	193 (92)
Social assistant	17 (8)	21 (10)	171 (82)
Others	23 (11)	4 (2)	194 (93)

Values are presented as n (%).

Nevertheless, these percentages are at variance with most of the observations in literature both for abdominoperineal pull-through operations [12] and for posterior sagittal anorectoplasty [13], where 30% to 90% of patients, depending on type of ARM, have good bowel control at age 3 years.

Such difference might be because of the fact that the largest published series are usually from the most experienced surgical centers; moreover, there may also be a tendency to ignore moderate continence defects in children who were much sicker at birth.

Therapeutic aids were widely used among patients; however, the use of enemas was not only limited to patients considering themselves (or their children) clean but also, to a greater extent, to patients who soiled where enemas were common practice in more than half (64%) of the patients.

These data are quite surprising because this is the first follow-up series in which data were compiled after the introduction of the BM program [8]; it is surprising that, of 143 patients having enemas or other forms of therapeutic aid, only 73 were doing so within a regular BM program, with only 29 achieving complete cleanliness; on the other hand, there is still a significant percentage of patients who are trying to keep themselves clean empirically.

As recently emphasized [14], it may be worth renouncing the assessment of fecal continence by means of scores in favor of newer classifications according to the treatment that the patient needs postoperatively.

Interestingly, there was also a wide use of enemas among patients who still soil. These data indicate that BM is probably not adequate and that, for parents of patients, soiling is not a major concern but, rather, a "normal" condition. These data significantly differ from other large experiences: in a series of 58 adult patients, Hassink et al [15] reported that soiling was the most disabling aspect of incontinence.

Such differences are partly explained by the small number of BM specialists who were called upon during postoperative assistance to children and their families (Table 2) and by the insufficient degree of satisfaction about their care (Table 6). Therefore, because most subjects reported a good explanation about their or their child's malformation and functional prognosis before surgery, it appears evident that parents are not provided with the same level of information later in life, where the importance of a multidisciplinary team cannot be underestimated [16].

These observations are much more important when considering that more than half (58%) of responders on BM declared that BM had improved their lives and that, nowadays, currently available surgical techniques such as Malone antegrade continence enema [17] enable the patient to decide when an enema should be performed.

Based on these findings, we believe that soiling after ARM repair is not to be considered inevitable and more efforts should be made to inform both families and health-care professionals about the possibilities to improve level of cleanliness.

Inadequacy of follow-up is also confirmed by results of urinary continence; as confirmed by larger series [8], of the 12 totally incontinent patients, 8 were females (all cloacas) and 4 were males with RBN fistulae; however, in 20 patients (17 males with perineal or rectobulbar fistulae and 3 females with rectovestibular fistulae), occasional incontinence with dribbling was reported with apparent no explanation. Again, this denotes lack of appropriate urologic follow-up; we speculate that such minor forms of incontinence might be either part of a spectrum of voiding dysfunction or reflect some degree of iatrogenic injury.

Interestingly, lack of psychologic support was of concern to 74% of responders. In this respect, many studies describe the effect of anal malformations on the lives of affected children and their parents; in 1996, a comparative study of a small sample of adolescents revealed a high frequency of mental (58%) and psychosocial problems (73%) [18].

On the other hand, Hassink et al [19] found that the whole family is affected when a child is born with imperforate anus and that, because of the huge impact of the malformation, the family needs support throughout the child's development.

Although nonspecific quality of life instruments were used in the questionnaire, it is surprising that, with a such wide amount of data available in the medical literature, a psychologic support is still not routinely offered to the patients and/or the parents throughout follow-up care.

There are some limitations to this study: first, the questionnaire was developed by nonphysicians, and therefore, some key issues may have been missed; second, educational background and, obviously, age of responders varied widely with repercussions on understanding; third, responses about continence were admittedly subjective, and results were not stratified by age; and fourth, a significant percentage of patients and/or parents did not return the questionnaire.

Nevertheless, the study provides significant insights into ARM from a perspective that is rarely addressed in the medical literature but that is probably the closest to parents' and families' needs.

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