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Anorectal Malformations in a Tertiary Pediatric Surgery Center from Romania: 20 Years of Experience

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Abstract

Introduction: The main goal of surgery and multidisciplinary behavioral therapy in anorectal malformations (ARMs) is to achieve good anatomical and functional outcomes and a better quality of life.

Material and Methods: A retrospective analysis of 146 patients with ARMs was conducted over a 20 years period in our pediatric surgery department. The clinical and demographical data, operative protocols, short and long term outcomes were subject to assessment.

Results: The patient series included 85 boys and 61 girls aged between one day and two years, of which 82.19% were diagnosed in the first few days of life by means of clinical examination and current imaging tests. In 26.02% of cases we found associated congenital abnormalities. A total of 142 (97.25%) cases of all our series underwent surgical treatment; four patients died before surgery. For 131 children the definitive treatment involved posterior sagittal anorectoplasty (PSARP) respectively 11 posterior sagittal anorectovaginourethroplasty (PSARVUP). Long-term follow-up in 115 patients showed good clinical continence in 82 cases (71.30%) but more than half of the teenagers had body dissatisfaction and social difficulties.

Conclusions: Through this review of 20 years of experience our findings are consistent with other reports regarding male predominance, low birth weight, clinical and imaging diagnosis. We found however a lower incidence of associated abnormalities. The most frequent surgical intervention was posterior sagittal anorectoplasty (PSARP) or posterior sagittal anorectovaginourethroplasty (PSARVUP), both followed by satisfactory functional outcome reporting nevertheless moderate social insertion. Our experience in ARMs proved the effectiveness of the consecrated surgical strategy; long-term assistance for patients and their families should be improved.

Keywords: Rectum; Anorectal malformations; Anorectoplasty; Anorectovaginourethroplasty

Introduction

Anorectal malformations (ARMs) are a group of commonly encountered congenital anomalies in pediatric surgery practice, with an incidence of 2 in 1000 to 1 in 5000 live births, but with significant variations between different regions throughout the world, as reported in the literature [1,2]. According to Peña and Bechit there is a wide spectrum of presentation, ranging from low perineal fistula to high anomalies with complex surgical management [3,4]. As Stoll and Moore discussed, ARMs are often associated with a complex range of another anomalies i.e. esophageal atresia, and central nervous system, cardiovascular, urogenital, gastrointestinal and skeletal defects [5,6]. In his report, Holschneider stated that the advances in imaging techniques and improvements in the knowledge of embryology, anatomy and physiology of ARM cases have refined both diagnosis and surgical therapy [7]. Other authors found that the classical approach to this pathology have moved towards PSARP and minimally invasive techniques with a clear goal of improving anatomical and functional outcomes and quality of life [8]. However constipation and fecal incontinence are still the main concerns regarding the long-term outcomes in these patients, as was discussed by de Blaauw et al. [9]

Material and Methods

We performed a retrospective study by analyzing the clinical records of all the ARMs patients managed in the Pediatric Surgery Department of "St. Mary" Children Emergency Hospital of Iasi, Romania over a 20 years period (1996-2015). The collected data included demographic and social characteristics of the mother and child, birth weight, type of ARMs, clinical features including associated malformations, imaging tests, initial and definitive surgical approach and postoperative outcomes, comprising a long term follow-up with respect to functional aspects, psychological modifications associated with chronic disabilities, social insertion, and quality of life.

Results

The patient series included 146 children, 85 boys and 61 girls, with ages between 1 day and 2 years old, with a mean age of 46.4 ± 5.7 days. Our report showed male predominance (58.21% vs 41.78%) along with a higher frequency in neonates with low birth weight (68.4% of all the patients, N = 100). A significant percentage of these children came from rural areas (64.4%, N = 94) and from families with low incomes (75.34%, N = 110). Mothers' age ranged from 16 to 42 years old, with a mean age of 21.3 ± 2.3 years old. The initial diagnosis was made

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in the first four days of life by means of a full physical examination in 120 patients (82.19%), within the first year of life in 14 cases (9.58%), and after one year in 12 children (8.21%) respectively. In the cases with delayed diagnosis, the main presenting complaint was chronic constipation. Although most of the patients admitted in our department were usually diagnosed at birth, some of them the anomaly correctly diagnosed beyond the early newborn period. All these cases were patients with perineal fistula, both males and females, or girls with vestibular fistula, in which a superficial examination of the perineal area and the presence of feces evacuation through the fistula in the first days of life led to a delay in establishing the diagnosis. These children subsequently developed constipation that consistently coincided with a change in their diet from breast milk to formula, or at the time of switching to solid food, so they were admitted for a pediatric surgical examination which identified the malformation. In the neonatal period, the anomaly presented most frequently with abdominal distension, vomiting and failure to pass meconium, associated with an abnormal aspect of the perineum (with or without the presence of an orifice). Upon perineal inspection, the most frequent clinical sign that we identified was the absence of a normal anal opening. Other frequently encountered findings upon physical examination included a bulge in the perineum or an anterior anal opening, meconium in urine and stool passing through the vestibule or, in the case of girls, through the vagina. Older patients presented with constipation and/or narrow anal opening (Table I).

Clinical diagnosis of so called "low" ARMs was made in almost all of the male and female patients,while the more complex cases required further explorations. However, all the patients underwent plain thoracic and abdominal X-Ray, as well as cardiac and abdominal ultrasound to assess cardiac, genitourinary, vertebral and other associated anomalies. Invertography was performed in 27 cases (18.49%) to determine the location of the rectal pouch in patients without visible fistula (Figure 1).

Other imaging contrast studies such as rectal pouch examination, fistulography and urethrography were performed in 40 cases (27.39%), while voiding cystography was conducted in 15 cases (10.27%) in order to detect the rectal urethral fistula or associated vesicoureteral reflux. Endoscopic evaluation of the genitourinary tract was performed in 62 patients (42.46%), including 11 cases of cloaca, in order to assess the complexity of the malformation. In 8 of these cases the length of the common channel was less than 3 cm. Other endoscopic findings included the presence of a septate vagina (2 cases) and the identification of recto-urethral fistula (3 cases). In two cases perineal ultrasound measured the distance between the rectal pouch and perineal skin. None of the patients underwent CT or MRI explorations. As far as the type of ARMs according to Peña's classification is concerned, there were 87 perineal fistulas (59.58%), 26 cases (17.80%) of imperforate anus without fistula, 14 vestibular fistulas (9.58%), 11 cases (7.53%) of cloaca, 6 rectourethral fistulas (4.10%), and two patients with rectal atresia (1.36%) (Table II and Figure 2).

Associated abnormalities were found in 38 patients (26.02%), some of them having two or even three congenital defects. The most common were genitourinary lesions in 14 patients (9.58%), including congenital hydronephrosis, hypospadias, renal agenesis, undescended testis, and Mayer-Rokitansky syndrome. Cardiovascular anomalies were reported in 11 cases (7.53%), including tetralogy of Fallot (TOF), ventricular or atrial septal defect, coarctation of the aorta or left ventricle hypoplasia. We noticed a significant incidence of Down syndrome (12 cases; 8.21%), esophageal atresia (8 cases; 5.47%), vertebral anomalies (myelomeningoceles) (6 cases; 4.10%) and omphalocele, cloacal exstrophy, imperforate anus and spinal defects complex (OESI) (2 cases; 1.36%).

Surgical treatment was performed in 142 out of 146 patients; four patients died before surgery due to multiple associated congenital anomalies. The surgical approach and timing were dictated by the type and anatomy of the malformation, general condition at the time of presentation, and additional pathology (Table III). A total of 33 patients (23.23%) underwent colostomy; in 30 children, the colostomy was placed on the transverse colon, while the other three were fitted with a divided colostomy placed at the end of the descending colon. All the female patients with cloaca and vestibular fistula received an ostomy, and all the rectourethral fistulas in male patients were managed in the same way. Also, the colostomy as the first surgical procedure was performed on the two patients with rectal atresia. The preliminary colostomy was part of three stage repair, as all cases subsequently underwent posterior sagittal anorectoplasty (PSARP) or posterior sagittal anorectovaginourethroplasty (PSARVUP), and finally colostomy closure. All the patients with imperforate anus or perineal fistula (109 patients) underwent a mini PSARP or PSARP procedure, without requiring a colostomy, so in 76.76% of the cases the original and preferred surgical protocol was a definitive anorectal reconstruction (Figure 3). The definitive treatment in 131 children (92.25%) was PSARP, with an additional abdominal access in 4 cases due to a high placed rectal pouch. All cloacal cases underwent a PSARPVUP procedure (7.74%) with abdominal access in 4 cases. Early postoperative complications were minor, including perineal wound infection (4 patients), and colostomy related complications – prolapse (4 cases) and stenosis (1 case); all such complications were treated conservatively. We also reported three postoperative obstructions which required surgical therapy. Patient follow-up was carried out on a regular basis for all the patients every 1, 3, 6 and 12 months, and then annually. In patients over the age of three, periodic monitoring included clinical continence, social insertion and their quality of life. The functional outcome was assessed in 115 patients over the age of three, and the results were as follows: good in 82 cases (71.30%) – presence of voluntary defecation, no or occasionally soiling, absent or mild constipation; acceptable in 25 cases (21.73%) – presence of bowel movement, rare soiling, moderate constipation); and poor in 8 cases (6.95%) – absence of voluntary defecation, moderate to severe soiling and any degree of constipation. Three of these cases required additional surgery for continence purposes. In the follow-up group, 39 children (33.91%) presented with constipation, which was more prevalent in the group with perineal, vestibular fistula and imperforate anus, but it

Table I: Associations of clinical findings in children with ARMs.

Clinical Findings	N (%)
Vomiting, abdominal distension	69 patients (47.26%)
Failure to pass meconium	63 patients (43.15%)
Bulge in perineum with absent anus	42 patients (28.76%)
Anterior anal opening	33 patients (22.60%)
Constipation/narrow anal opening	26 patients (17.80%)
Meconium in urine	21 patients (14.38%)
Passing meconium through the vagina / vestibule	20 patients (13.69%)

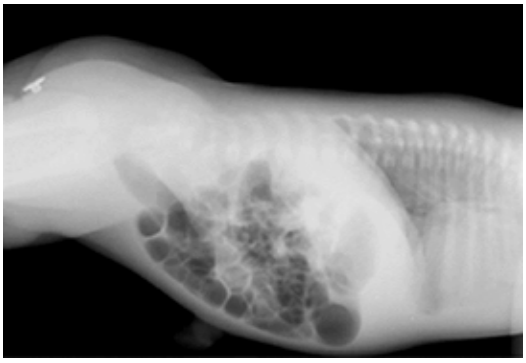

Figure 1: Invertography: Holding the baby upside down, a lateral radiography is performed to observe the distance between the level of gas in the distal rectum and a metal piece placed over the expected anus

Table II: ARM type; case distribution (according to Peña's classification).

Males	N/%	Females	N/%
Perineal fistula	57/39.04	Perineal fistula	30/20.54
Imperforate anus without fistula	21/14.38	Imperforate anus without fistula	5/3.42
Rectourethral fistula	6/4.1	Vestibular fistula	14/9.58
Rectal atresia	1/0.68	Cloaca	11/7.53
		Rectal atresia	1/0.68

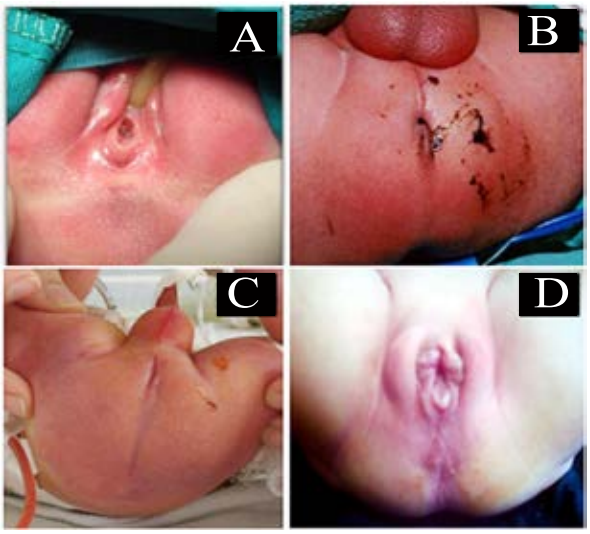

Figure 2: Anorectal malformation–clinical aspects. A: Perineal fistula; B: Vestibular fistula; C: Rectourethral fistula; D: Cloaca.

was associated with good bowel control. On the other hand, cases with severe malformations (cloaca, recto-urethral fistula, rectal agenesis) were found to have a high prevalence of soiling (8 cases, 6.95 %). A small group of 44 children (24 males and 20 females) remained in our database until the age of 18; psychological assistance was provided in all cases, along with short psychotherapy meetings. Standardized interviews were used to assess the patients' school performances and behavior, while validated tests and questionnaires were used to evaluate their intelligence, attention, self-esteem and quality of life. The majority of the patients (35 cases, 79.54%) have normal intelligence (for IQ, M = 92) with no statistically significant difference between boys and girls. All patients were enrolled in normal schools. Psychological distress and emotional difficulties were identified in teenagers (32 cases, 72.72%) together with a low level of body-consciousness and self-confidence, as well as a relatively reduced autonomy and social contacts (28 cases, 63.63%). Over half of these children (26 patients, 59.09%) avoided participating in activities that are specific for their age.

Discussion

Malformations of the anus, anal canal and rectum are relatively common congenital defects, often coexisting in a significant percentage of cases with other congenital anomalies, but as different authors reporting their incidence has major differences in terms of worldwide geographic distribution [1-3,7]. Researchers such as de Blaauw and Wijers found that the causes of ARMs are unknown, but some studies demonstrated a genetic predisposition to these conditions [9,10]. ARMs include a wide spectrum of inborn defects ranging from perineal fistulas that require a relatively simple modality of treatment to major defects that require complex management. The multiple taxonomy of these lesions (Wingspread, Peña, Kirkenbeck) proposed in the literature is rather confusing, most practitioners currently favoring a classification based on the anatomical criteria depending on the position of the rectal pouch relative to the puborectal sling, associated abnormalities ("syndromic" forms) and the presence or absence of fistulas [2,10,11]. Advances in imaging techniques along with improvements in embryology, morphology and pathogenesis of these lesions have refined both diagnosis and management, moving from classical procedures to PSARP (VUP) and even minimally invasive methods, as discussed by different authors [12-18]. In our report we noticed a male predominance and a higher frequency in male neonates with low birth weight. The median age of the mothers was 21.3+/-2.3 years, lower than the national average reported by the National Statistics Institute for that period, i.e. 22.5-26.0 years [19]. These findings are consistent with Vermes' report, which suggested that the male gender and intrauterine growth restriction, together with mother's age and low socioeconomic conditions may have an impact on the risk of ARMs [20]. In our series, the majority of ARMs were diagnosed at or soon after birth upon routine postnatal examination. Evident features of these anomalies included the absence of the anus or an abnormal appearance of the perineum, i.e. lack of or minimal passage of meconium, but evidence of perineal, vestibular, recto-urethral fistula or persistent cloaca (as a frequent deformity in female patients). In addition to this, clinical signs in the neonatal period included vomiting, abdominal distension, and failure to pass meconium. We performed VACTERL (Vertebral anomalies, Anal atresia, Cardiac defects, Tracheoesophageal and/or Esophageal atresia, Renal and Radial anomalies, and Limb defects) screening in all newborn with a diagnosis of ARMs upon the first perineal inspection. However, we have identified a number of 26 cases (17.80%) with a late diagnosis in children with perineal fistula or vestibular fistula misdiagnosed in the neonatal period and referred to us due to severe constipation or narrow anal opening. We consider that a correct physical examination of the perineal area is crucial in the neonatal period in order to avoid a delay in the diagnosis of these cases. In a quarter of the cases, ARMs presented with associated abnormalities, the most common of which were urinary tract and cardiovascular defects. Our findings are not consistent with other authors' research that finds an association between anorectal malformations and other inborn defects ranging from 68% to 78%. As far as the types of associated congenital defects are concerned, the literature reports a variety of multisystemic conditions associated with ARMs including MURCS (Müllerian duct aplasia, Renal aplasia, Cervicothoracic Somite dysplasia), trisomy 13, 18, 21 or cat-eye, caudal regression, facio-auriculo-vertebral and fetal alcohol syndromes [21]. Authors like Bălănescu and Nah found an increased incidence of gastrointestinal and urogenital defects, while others found more genitourinary and spinal anomalies [14,22]. The most frequent associations in our study were genitourinary malformations and Down syndrome, followed by cardiovascular anomalies and esophageal atresia. The diagnosis of ARMs in our series was mainly clinical, including routine imaging (thoracic, abdominal, pelvic and spinal X-rays) and invertography, abdominal ultrasound and contrast studies, along with endoscopic evaluation that aimed to evaluate the topography and extent of the lesion. The recent ARM-Net meeting brought minor changes to the preoperative management of ARMs regarding the 24 hours re-evaluation and cross-table lateral X-ray, compared to Levitt and Peña's flow chart [23,24]. It was suggested that in a newborn male with a perineal fistula, a cross-table lateral X-ray is unnecessary, this procedure being recommended only in cases of normal buttocks, normal spine, normal sacrum, and negative urinalysis on meconium; this protocol was extended to female neonates with visible fistula. Early diagnosis was essential for short and long term prognosis. Almost all our patients underwent surgery, except for a few patients (4 cases, 2.73%) with delayed admission, major comorbidities and impaired general condition, who died shortly after presentation. Neonates with low-type ARMs benefit from minimal PSARP, with good short and long term results. PSARP was performed in male patients with cases of high-type ARMs, preceded by a protective colostomy. All cloaca cases were treated in the neonatal period by

Table III: Types of surgical procedures in patients with ARM.

Arm Type	First Surgical Procedure	Definitive Surgery
Perineal fistula	(mini)-PSARP – 85 cases	
Rectourethral fistula	Colostomy - 6 cases	PSARP – 4 cases PSARP + abdominal approach – 2 cases
Vestibular fistula	Colostomy – 14 cases	PSARP – 14 cases
Imperforate anus without fistula	(mini)-PSARP – 24 cases	
Rectal atresia	Colostomy – 2 cases	PSARP + abdominal approach – 2 cases
Cloacal malformation	Colostomy – 11 cases	PSARVUP – 7 cases PSARVUP + abdominal approach – 4 cases

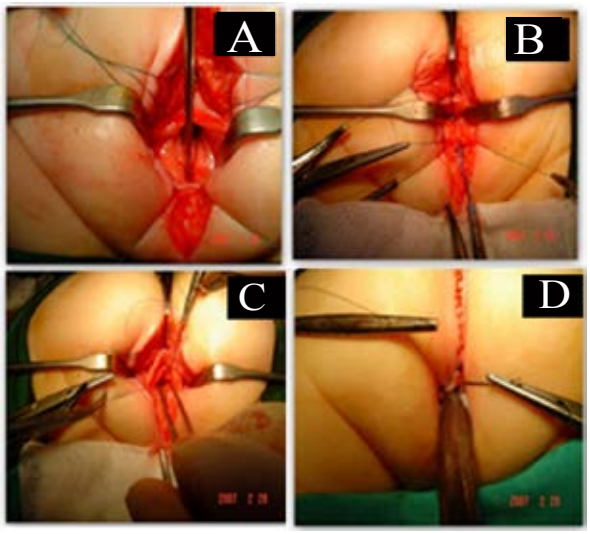

Figure 3: Rectourethral fistula: PSARP procedure; A: Identification of the rectourethral fistula (with metal probe inside); B: Isolation of the fistula with multiple stitches; C: Closure of the fistula and complete separation of the rectum; D: Final aspect of the anoplasty.

placing a colostomy, and a PSARVUP procedure was later used as final surgery, sometimes requiring an associated abdominal approach. As far as the treatment alternatives for female patients with vestibular fistula are concerned, the literature argues for either a one or a three stages surgery. The supporters of perineal surgery in the neonatal period champion the idea of one stage surgery for the purpose of reducing possible anesthesia and morbidities-related colostomy complications. However, this type of surgery requires good bowel preparation, as there is a great risk of infection. We preferred to do a three stages surgery (colostomy, followed by PSARP and then colostomy closure) in a more cautious approach, although there are studies like Adeniran's and Liu's showing no significant differences in functional outcomes between a one stage and three stage operation [25,26]. The final goal of the surgical treatment in ARMs is to obtain an anus placed within the muscular complex, by separating the rectum from the genital or urinary tract and also to gain continence without any social impairment. The long term functional outcome is essential for these patients' quality of life. During our follow up period, we noted that 71.30% of the cases acquired voluntary bowel movements, these results being similar to those previously reported by Sohyun Nam (84.8%) or Peña (75%) [8,27]. Almost half (42.10%) of the children with high anomalies like cloaca, recto-urethral fistula and rectal atresia presented with moderate and severe soiling (i.e.6.95 % of patients in the follow-up group), a result that also seemed similar to other series, as Rintala reported a rate of 30% soiling in these patients [28]. The majority of patients that remained in our database until the age of 18 had a normal intelligence, but their sustained attention was below the norm. This data is consistent with Hondel's studies, which also suggest that these patients should benefit from special education or remedial teaching [29,30]. All the children from our sample were enrolled in normal schools. Due to educational policies in Romania of the last two decades, we have no cases of children integrated in a special institution. We found that 63.63% of teenagers were dissatisfied with their body and 59.09% were having difficulties in social activities; our results are consistent with Grano's reports on self-perception and social insertion in children with ARMs [31]. Schmiedeke considers that multidisciplinary behavioral treatment (MBT) should offer psychological assistance both for the parents and the child, as well as physical therapy, thus reducing psycho-social distress. There is still an open discussion regarding the possibilities of accurate quantification of the short and long term outcomes. Despite our sometimes limited possibilities to investigate these children, we succeeded in taking a correct approach to our cases, with promising short and long term results of definitive surgery; reduced morbidity and lack of postoperative mortality come to support our approach to surgical treatment of this pathology. This study has several limitations as it is a single center report and it only provides limited data regarding the long-term monitoring of the patients. We intend to focus our future research on vestibular fistula dilation in selected patients. (Low birth weight <2,000 g or severe associated congenital anomalies).

Conclusion

A better understanding of pathogenesis with more accurate pre- and intraoperative diagnosis together with continuous perfectible practice of established operative techniques appear to be the keys to success in ARMs management, aligning our team experience and expertise toward reputed centers. We support a multidisciplinary approach to these cases, using complex teams that include neonatologists, pediatrics, surgeons, pediatricians, psychologist and family doctors, together with a monitored transition towards adult gastroenterology centers.

Conflict of interest

Authors have no conflict of interests to disclose. All the authors had a significant contribution to this paperwork.

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