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Post-Operative and Long Term Prognosis of the Patients with Anorectal Malformations: A 15-Year Single-Center Experience at a Tertiary Neonatal Intensive Care Unit in Türkiye*

Ünal BAKAL [1, a]
Mustafa Tamer GÜRBAZ[2,b]
Tugay TARTAR [1, c]
Ahmet KAZEZ [1, d]

[1] Fırat University,
Faculty of Medicine,
Department of Pediatric Surgery,
Elazığ, TÜRKİYE

[2] Meydan Hospital,
Department of Pediatric Surgery,
Şanlıurfa, TÜRKİYE

[a] ORCID: 0000-0002-5140-8618
[b] ORCID: 0000-0002-1636-0938
[c] ORCID: 0000-0002-7755-4736
[d] ORCID: 0000-0003-0440-3555

Objective: To evaluate the post-operative and long-term outcomes of patients with anorectal malformations (ARMs).

Materials and Methods: The clinical and demographic data of patients who underwent surgery for ARM between June 1996 and December 2011 were retrieved from the hospital records. Anorectal functions of patients aged >4 years were examined according to the Krickenbeck criteria. Patients were examined according to gender, birth weight, the type of ARM as per the Wingspread classification, accompanying anomalies, surgical procedures and complications.

Results: Out of 96 patients, 67 were male, and the type of ARM as per the Wingspread classification and gender distribution (M/F) was as follows: high type, 41 (28/13) (43%); intermediate type, 10 (4/6) (10.5%) and low type, 45 (35/10) (46.5%). Distribution by birth weight was as follows: of the total, 19 (19.8%) patients were <1800 grams, 47 (49%) were between 1800 and 2500 grams and 30 (31.2%) were ≥2500 grams. Mortality was significantly higher with comorbid anomalies of the gastrointestinal tract (p=0.036). The most common complication was dermatitis. Statistically significant correlations were found between the types of ARM and faecal soiling (FS) and constipation (p=0.001). There was a statistically significant correlation between mortality and the presence of high-type ARM (p=0.012).

Conclusions: Comorbid anomalies that cause high mortality in high and intermediate type should be carefully examined, and these anomalies should be considered in the treatment and follow-up. Despite medical and surgical advances, stool incontinence, FS and constipation continue to be major problems in patients with high- and intermediate-type ARMs.

Key Words: *Anorectal malformation, surgical repair, complication, anorectal functions, long term prognosis*

Anorektal Malformasyonlu Hastaların Ameliyat Sonrası ve Uzun Dönem Prognozu: Türkiye\'deki Üçüncü Basamak Yenidoğan Yoğun Bakım Ünitesinde 15 Yıllık Tek Merkezli Deneyim

Amaç: Anorektal malformasyonlu (ARM) hastaların ameliyat sonrası ve uzun dönem sonuçlarını değerlendirmek.

Gereç ve Yöntem: Haziran 1996 ile Aralık 2011 arasında ARM nedeniyle ameliyat edilen hastaların klinik ve demografik verileri hastane kayıtlarından elde edildi. 4 yaş üstü hastaların anorektal fonksiyonları Krickenbeck kriterlerine göre incelendi. Hastalar cinsiyet, doğum ağırlığı, Wingspread sınıflamasına göre ARM tipi, eşlik eden anomaliler, cerrahi prosedürler ve komplikasyonlara göre incelendi.

Bulgular: Doksan altı hastanın 67'si erkekti. Wingspread sınıflamasına ve cinsiyet dağılımına (E/K) göre ARM tipleri; yüksek tip, 41 (28/13) (%43); orta tip, 10 (4/6) (%10.5) ve düşük tip, 45 (35/10) (%46.5) idi. Doğum ağırlığına göre dağılımı ise; tüm hastaların 19'u (%19.8) <1800 gram, 47'si (%49) 1800 ile 2500 gram arasında ve 30'u (%31.2) ≥2500 gramdı. Mortalite, gastrointestinal sistemin komorbid anomalileri ile istatistiksel olarak anlamlı şekilde daha yüksekti (p=0.036). En sık görülen komplikasyon dermatitti. ARM tipleri ile fekal kirlenme (FK) ve kabızlık arasında istatistiksel olarak anlamlı korelasyonlar bulundu (p=0.001). Mortalite ile yüksek tip ARM varlığı arasında istatistiksel olarak anlamlı bir korelasyon vardı (p=0.012).

Sonuç: Yüksek ve orta tipte yüksek mortaliteye neden olan komorbid anomaliler dikkatlice incelenmeli ve bu anomaliler tedavi ve takipte dikkate alınmalıdır. Tıbbi ve cerrahi gelişmelere rağmen, dışkı inkontinansı, FK ve kabızlık yüksek ve orta tip ARM'li hastalarda önemli sorunlar olmaya devam etmektedir.

Anahtar Kelimeler: *Anorektal malformasyon, cerrahi onarım, komplikasyon, anorektal fonksiyonlar, uzun dönem prognoz*

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Correspondence

Tugay TARTAR
Fırat University,
Faculty of Medicine,
Department of Pediatric Surgery,
Elazığ - TÜRKİYE

tugaytartar@gmail.com

Introduction

Congenital anorectal malformations (ARMs) occur in 1 in 4,000–5,000 live births (1). ARMs are classified as high, intermediate and low according to the Wingspread classification (2). Although there have been significant advances in the diagnosis and

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treatment of ARMs, it has been reported that significant problems, such as absence of voluntary bowel movements (VBMs) after surgery, anal incontinence and constipation, continue at a rate of 30%–70% (3).

The aim of this study was to evaluate the demographic and clinical features, comorbid congenital anomalies and late-period anorectal functions according to the Wingspread classification of patients who underwent surgical treatment for ARM at a tertiary university hospital having a paediatric surgery and neonatal intensive care unit.

Materials and Methods

Research and Publication Ethics: Ethical approval for this study was obtained from Fırat University School of Medicine Non-Interventional Clinical Researches Ethics Committee (date: 23/02/2012, decree number: 06).

Research Design: The patients who underwent surgery due to ARM between January 1996 and December 2011 in the Fırat University Hospital Paediatric Surgery and Neonatal Intensive Care Unit were included in the study. All surgical interventions were performed by two surgeons, where at least one surgeon was experienced in paediatric colorectal surgery. The records of these patients were retrospectively reviewed. The demographic data, gender, place of residence, birth weight of the patient, gestational age, type of ARM, comorbid anomalies, type of surgery and complications were recorded. Anorectal functions and defecation status of the patients who completed the age of 4 years were evaluated according to the Krickenbeck criteria (Table 1) (4). Patients who underwent surgical intervention at other centres were excluded from the study.

Surgical Technique and Follow-Up: The operations were performed in accordance with the posterior sagittal anorectoplasty (PSARP) procedure developed by Pena (5). After the anal sphincter complex was found using an electromyostimulation device, it was dissected, and the rectum was placed in the centre of the sphincter complex. Colostomy was performed in all patients except in those with low-type ARM. Y-V anoplasty and cut-back anoplasty were performed in patients with low-type ARM. Ostomy was performed in 5 patients with low-type ARM due to the presence of other anomalies of the gastrointestinal tract (GIT). Anal calibration was performed in all patients during the examination at 14 days after surgery. Regular dilatation was performed, if necessary.

All patients were screened for VACTERL association. X-ray, ultrasonography and voiding cystourethrography were performed for urinary tract anomalies. Echocardiography was performed to screen for congenital heart anomalies. Magnetic resonance imaging was performed if there was an evidence of tethered cord and neurogenic bladder.

Statistical Analysis: For all statistical analysis, Statistical Package for the Social Sciences 21 for Windows (IBM SPSS Statistics, Armonk, NY, USA) was used. Numerical data were expressed as mean ± standard deviation, and categorical data were expressed as percentage (%). The Mann–Whitney U test was used to compare nonparametric data, and the Chi-square or Fisher's Exact Test was used to compare categorical data. A p value <0.05 was considered statistically significant.

Table 1. International classification Krickenbeck postoperative results for anorectal malformation

Voluntary Bowel Movements Feeling of Urge, Capacity to Verbalize, Hold the Bowel Movements	Yes/No
Soiling	No
Grade 1	Occasionally (1–2/week)
Grade 2	Every day, no social problem
Grade 3	Constant, social problem
Constipation	No
Grade 1	Manageable by changes in diet
Grade 2	Requires laxative
Grade 3	Resistant to laxative and diet

Table 2. Distribution of additional anomaly-(ies) among patients.

Additional anomaly-(ies)	Intermediate	Low-type ARM	High-type ARM	Number of patients: n (%)
Genitourinary system anomaly-(ies)	1	6	11	18
Cardiovascular system anomaly-(ies)	3	2	10	15
Gastrointestinal system anomaly-(ies)	2	4	12	18
Musculoskeletal system anomaly-(ies)	3	2	7	12
Central nervous system anomaly-(ies)	-	-	2	2
Endocrine system anomaly-(ies)	-	1	2	3
Chromosome anomaly-(ies)	-	-	2	2
Other anomaly-(ies)	2	1	1	4
Total	**11**	**16**	**47**	**74**

ARM: Anorectal malformation

Table 3. Association between anorectal malformations and voluntary bowel movements

		Voluntary bowel movements			Total
		Death	Yes	No	
Type of anorectal malformation	Intermediate	2	5	3	10
	Low	3	35	7	45
	High*	13	18	10	41
Total		18	58	20	96

*: There was no significant difference between the groups (p=0.146).

Table 4. Association between anorectal malformations and faecal soiling

		Faecal soiling					Total
		Death	None	Once or twice a week	No social problems	Serious social problems	
Type of Anorectal Malformation	Intermediate	2	4	1	2	1	10
	Low	3	35	0	7	0	45
	High*	13	16	2	4	6	41
Total		18	55	3	13	7	96

*: There is a significant difference between high-type and low-type anorectal malformation (p=0.001).

Table 5. Association between anorectal malformations and constipation

		Constipation					
		Death	Can be Resolved With Diet	Laxatives are Needed	Resistant to Diet and Laxatives	None	Total
Type of Anorectal Malformation	Intermediate	2	1	1	3	3	10
	Low	3	7	5	0	30	45
	High*	13	14	4	2	8	41
Total		18	22	10	5	41	96

*: There is a significant difference between high-type and low-type anorectal malformation (p=0.001).

Results

During the past 15 years, of 7663 neonates admitted to the NICU. 96 (1.2%) of them were diagnosed with ARM. Of these 96 patients diagnosed with ARM, 67 were male and 29 were female (M/F: 3/2). According to the Wingspread classification, 41 patients had high-type (43%), 10 patients had intermediate-type (10.5%) and 45 patients had low-type (46.5%) ARMs. Low-type ARM was more common in males (52.2%), whereas high-type ARM was more common in females (44.8%). Further, 3 patients had cloacal anomalies, and they were included in the high-type ARM group. Distribution by birth weight was as follows: of the total, 19 (19.8%) patients were <1800 grams, 47 (49%) patients were between 1800 and 2500 grams and 30 (31.2%) patients were >2500 grams. According to the gestational age, 52.1% (n=50) of the patients were premature and 47.9% (n=46) were mature. Fistula was present in 77% (n=74) of the patients. Of patients with low-type ARM, 71.1% (n=32) had perineal fistula, 6.6% (n=3) had anovestibular fistula and 22.2% (n=10) had no fistula. Of patients with intermediate-type ARM, 50% (n=5) had rectovestibular fistula, 10% (n=1) had rectobulbar uretral fistula, 10% (n=1) had rectovaginal fistula and 30% (n=3) had no fistula. Of the patients with high-type ARM, 34% (n=14) had rectoprostatic uretral fistula, 22% (n=9) had rectovesical fistula, 12.1% (n=5) rectovestibular fistula had, 9.7% (n=4) had rectovaginal fistula and 22% (n=9) had no fistula. Ostomy was performed in 53 patients. Of the patients who underwent ostomy, 73.5% (n=39) had high-type, 17% (n=9) had intermediate-type and 9.4% (n=5) had low-type ARMs. Of the patients who underwent ostomy, sigmoid separe colostomy was performed in 79.2% (n=42), transverse loop colostomy was performed in 7.5% (n=4), ileostomy was performed in 7.5% (n=4), Hartman colostomy was performed in 3.7% (n=2) and gastrostomy was performed in 1.9% (n=1). Isolated oesophageal atresia (OA) was present in the patient who underwent gastrostomy. According to the type of definitive surgery, 50% (n=44) of the patients were treated with V-Y anoplasty, 47% were treated with PSARP via abdominal and sacroperineal approach and

3.4% (n=3) were treated with cloacal repair. Of the 53 patients who underwent colostomy, 13.2% (n=7) had dermatitis, and 5.6% (n=3) had stoma prolapse. Of patients with stoma prolapse, 2 had undergone transverse loop colostomy and one had undergone diverting sigmoid colostomy. Of the 88 patients who underwent definitive surgery, 3.4% (n=3) had rectal prolapse and 3.4% (n=3) had anal stenosis. The complication rate was 16.6% (n=16). The rate of comorbid anomalies of GIT was 14% (n=11) for the 78 patients who survived, whereas the rate of comorbid anomalies of GIT was 38.8% (n=7) for the 18 patients who died. The incidence of comorbid anomalies of GIT was significantly higher in non-survivors (p=0.026). The distribution of other congenital anomalies according to the type of ARM is presented in Table 2. Of the 18 patients who died, 12 (66.6%) had VACTERL association. No chromosomal anomaly was observed among the survivors; however, 2 patients had adenoids and borderline mental retardation (n=1) and right ear pinch abnormality (n=1). Trisomy 21 was detected in 2 (11%) of the 18 patients who died. Further, 2 patients had iris coloboma and nystagmus. There was a significant correlation between the presence of a chromosomal anomaly and mortality (p=0.0394). Mortality was significantly increased in the presence of an accompanying congenital anomaly (p=0.049). Of the 78 surviving patients, 58 (74%) had VBM, whereas 20 (26%) had no VBM. VBM status according to the type of ARM is presented in Table 3. Of patients without VBM, 50% (n=10) had high-type, 35% (n=7) had low-type and 15% (n=3) had intermediate-type ARMs. There was no significant difference between the ARM types with respect to VBM (p=0.146). Table 4 shows the distribution of faecal soiling (FS) according to the type of ARM. Of patients with FS, 50% (n=6) had continuous FS causing serious social problems. There was a statistically significant difference between patients with high-type and patients with low-type ARMs with respect to FS (p=0.001). The distribution of patients with chronic constipation according to the type of ARM is presented in Table 5. In total, 47.4% (n=37) of the patients had constipation. There was a statistically significant difference between the type of ARM with respect to constipation (p=0.001). The mortality rate was 18.8% (n=18). Of 18 non-survivors, 16.6% (n=3) died of respiratory failure, 38.8% (n=7) died of heart failure, 11.1% (n=2) died of sepsis and 33.3% (n=6) died of reasons related to the urinary tract. The mortality rate was 31.7% (n=13) for high-type ARM, 20% (n=2) for intermediate-type ARM and 6.6% (n=3) for low-type ARM. Mortality was significantly higher for high-type ARM compared to that for low-type ARM (p=0.012).

Discussion

Congenital anorectal malformations occur in 1 in 4,000–5,000 live births (1). Approximately two-thirds of the cases of ARMs occur in boys and one-third occur in girls. It is reported that two-thirds of male patients with ARM have the high type, whereas two-thirds of female patients with ARM have the low type (6). In our study, 43% (n=41) of the patients had high-type, 10.5% (n=10) had intermediate-type and 46.5% (n=45) had low-type ARMs. Low type was more common in males (52.2%), whereas high type was more common in females (44.8%). The incidence of ARM in the NICU was 1.56%. The results obtained in this study are consistent with those of literature.

The incidence of comorbid anomalies in patients with ARM ranges from 25% to 75% (7). The incidence of comorbid anomalies in patients with high- and intermediate-type ARMs is two times higher than that in patients with low-type ARM (5). The incidence of urogenital anomalies accompanying ARMs is reported to be 28%–89% (3). In our study, urogenital system anomalies were found in 18.7% (n=18) of the patients. In their study, Stoll et al. encountered cardiovascular anomalies in 15 (8.6%) out of 174 patients (8). In the present study, 15.6% (n=15) of the patients were found to have cardiovascular system anomalies, and this rate was higher than that reported in literature. Skeletal system anomalies have been observed in 12%-44% of patients with ARM (9). In our study, skeletal system anomalies were detected in 12.5% (n=12) of the patients, a finding consistent with that reported in literature. These results further support the fact that patients with ARM must be screened for VACTERL association.

The rate of comorbid anomalies of GIT was 14% (n=11) in the 78 patients who survived and 38.8% (n=7) in the 18 non-survivors. In previous studies, the most common comorbid anomaly of GIT in patients with ARM was OA (9). In our study, 18.8% (n=18) of the patients had comorbid anomalies of GIT. OA was observed in 7.29% of these patients. Of these patients, 6 had OA with tracheoesophageal fistula, and one had isolated OA. We observed that comorbid anomalies of GIT increased the mortality. It is necessary to screen patients with ARM for the presence of anomalies of GIT.

In patients with ARM, the incidence of Down syndrome was found to be between 0.36% and 2.7% (10). In our study, this rate was 2%, and it was consistent with that reported in literature.

In patients with high and intermediate-type ARM, the first session of surgical treatment generally involves opening a stoma. If a diverting sigmoid colostomy has been performed at the junction of the descending colon and the sigmoid colon, the likelihood of encountering complications, such as prolapse, passage of faeces into the distal colon and metabolic acidosis, is minimal (11). Of the 53 patients who underwent colostomy in our series, 42 underwent diverting sigmoid colostomy, 4 underwent transverse loop colostomy, 2 underwent Hartman colostomy, 3 underwent ileostomy, one underwent ileostomy–gastrostomy and one underwent gastrostomy. The patient who underwent gastrostomy had isolated OA. However, the number of surgeons who perform definitive surgery without colostomy is increasing day by day (12-14). High-type ARMs, such as rectovesical fistula, can be treated with laparoscopy-guided anorectal pull-through method without any need

for posterior sagittal incision, as mentioned by Georgeson et al. in 2000 (15).

Pena proposed the assessment of three parameters to evaluate the outcomes of surgery in patients with ARM. These are VBM, FS and constipation. These criteria have gradually gained recognition over the years and were accepted as the Krickenbeck criteria in May 2005 (4). According to the results of the study by Pena, the rate of achieving full continence is 41% (1). The rate of achieving full continence in the present study was 74%. This was due to the high percentage of patients with low-type ARM in our study. In a study by Çavuşoğlu et al. (16), the rate of VBM was 89.5%, and the mean age of their patients was 86 months. In our study, the rate of VBM was 74% (n=58), and the mean age of the patients was 89 months. In our study, VBM was present in 35 out of 42 surviving patients with low-type ARM, in 5 out of 8 surviving patients with intermediate-type ARM and in 18 out of 28 surviving patients with high-type ARM. Although the rate of VBM was higher among patients with low-type ARM, no statistically significant difference was found between the types of ARM with respect to VBM. In a study conducted by Çavuşoğlu et al. (16), the percentage of patients without FS was reported as 26.3%, whereas in our study, this percentage was 70.5%. The low rate of FS in our study was due to the high number of patients with low-type ARM in comparison with the high number of patients with high-type ARM in literature. In our study, a statistically significant difference was found between the types of ARM with respect to FS (p=0.001). In a study by Çavuşoğlu et al. (16), the rate of constipation was found to be 79%. In our study, the rate of constipation was 47.4% (n=37). Of these patients, 20 high-type, 4 had intermediate-type and 12 had low-type ARMs. The constipation rate in our study is below the rates reported in literature. We believe that this is due to the fact that the patients were subjected to the dilatation programme with 15-day intervals in the post-operative period.

In a previous study, dermatitis was the most common (53%) complication in patients with colostomy (17). This was the case in our study also (13.2%). In another study, the rate of colostomy prolapse was 16.3% (18). This is due to the fact that the transverse colon is more mobile. In our study, colostomy prolapse was observed in 5.6% of the patients who underwent colostomy. Further, 2 patients with a prolapse had undergone transverse loop colostomy, and 1 patient had undergone diverting sigmoid colostomy. We believe that diverting sigmoid colostomy is effective in decreasing the incidence of prolapse in patients with ARM.

One of the complications in the late period is the prolapse of the rectal mucosa. In a study of 833 patients, rectal mucosa prolapse was observed at a rate of 3.8% (19). In our study, rectal mucosa prolapse was observed in 3.4% of patients who underwent definitive surgery. These patients had high-type ARM. Another common complication observed in patients after definitive surgery is anal stenosis. In patients undergoing long-term follow-up, the rate of anal stenosis can be as high as 30% (20). In our study, the rate of anal stenosis was 3.4%. We believe that the low rate of anal stenosis in our study in patients who underwent definitive surgery was due to the fact that the patients were subjected to regular anal dilatation programme, and the location of the neo-anus opening was determined by an electromyostimulation device. The mortality rate is reported to be 5% in highly developed countries, and 35% in underdeveloped and the developing countries (21, 22). Of the 96 patients who were operated in our clinic due to ARM, 18 (18.8%) died. Of these, 3 patients died of respiratory failure, 7 died of heart failure, 2 died of sepsis and 6 died of causes related to the urinary tract. Further, 12 (66%) out of 18 patients who died in our study had VACTERL association. The incidence of comorbid anomalies in patients with ARM causes a significant increase in mortality.

In conclusion, comorbid anomalies that are associated with increased mortality in patients with high and intermediate-type ARMs should be carefully investigated, and these anomalies should be considered during treatment and follow-up. In patients with high and intermediate-type ARMs, stool incontinence, FS and constipation remain the most serious problems.

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