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Anomalies Associated with Anorectal Malformations

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Rezumat

Anomalii congenitale asociate malformațiilor ano-rectale

Scopul lucrării este acela de a evalua incidența anomaliilor congenitale întâlnite la pacienții diagnosticați cu malformații ano-rectale şi de a compara aceste rezultate cu cele publicate în literatura de specialitate. Un număr de 50 de pacienți diagnosticați cu malformații ano-rectale internați în clinica de chirurgie a Spitalului de Urgență pentru Copii "Grigore Alexandrescu" în perioada 2005-2011 au fost evaluați, fiind adunate informații cu privire la tipul de malformație ano-rectală şi anomaliile congenitale asociate, acestea din urmă fiind apoi împărțite pe sisteme. Din cei 50 de nou-născuți, 28 au fost băieți şi 22 fete. Mai mult de jumătate din pacienții incluşi în studiul de față au prezentat anomalii asociate, cele mai frecvente dintre acestea fiind întâlnite la nivel gastro-intestinal (36%), uro-genital (24%) şi cardio-vascular (16%). S-a observat o incidență crescută a patologiei gastro-intestinale asociate la pacienții diagnosticați cu persistență de cloacă, în timp ce grupul celor cu fistulă recto-vestibulară au prezentat cel mai frecvent anomalii cardio-vasculare. Incidența patologiei uro-genitale la copiii diagnosticați cu fistulă perineală a fost mult mai mare decât rezultatele întâlnite în literatura de specialitate.

Cuvinte cheie: malformație ano-rectală, patologie asociată, malformații gastro-intestinale, clasificare Krickenbeck

Abstract

Background: The purpose of the paper is to review the incidence of associated congenital anomalies that are encountered in patients presenting anorectal malformations and compare these results with those previously published.

Material and Methods: A number of 50 cases with ARM from our institution were reviewed (from 2005 to 2012) and information was collected on patient demographics, type of ARM and associated congenital anomalies, the latter being then categorized according to organ systems.

Results: Out of 50 newborns, 28 were males and 22 females (1.27:1). 34 (68%) had at least one associated abnormality. The majority of patients (40%) had imperforated anus without fistula. The most frequent seen anomalies were gastrointestinal (36%), urogenital (24%) and cardiovascular (16%).

Conclusions: More than half of the children included in our series have other associated abnormalities. We found gastro-intestinal anomalies to be the most common associated congenital defects in our patients. A higher incidence of this type of anomalies was encountered in newborns with persistent cloacal anomaly. The rectovestibular fistula group was most likely to present cardiac abnormalities. The incidence of genitourinary anomalies in the perineal fistula group is higher than the one described in other studies.

Key words: anorectal anomaly, associated malformations, gastrointestinal abnormalities, Krickenbeck criteria

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Introduction

Anorectal malformations (ARMs) are among the frequently encountered congenital abnormalities that represent an

important part of paediatric surgical practice (1). They include a series of defects ranging from a slight malposition of the anus to more complex anomalies of the hindgut and urogenital organs (2), with an estimated incidence of 1 in 2500 to 5000 live births; they are slightly more common in males (3,4).

Management of ARM in the neonatal period is essential and it focuses on the accurate classification of anorectal malformations and the best way to restore the normal intestinal anatomy and function. However, surgeons must be aware of the high incidence of associated abnormalities in cases of ARMs (5). About 20-80% of patients born with anorectal malformations also present one or more associated defects, including gastrointestinal, genitourinary, cardiovascular and of the central nervous system (6,7). These anomalies have an important impact on the survival of such patients.

Because the classification of ARM has been repeatedly revised over the years, problems in comparing the outcome results of different studies have arisen, due to confusions related to the classification and assessment systems. In previous studies, the prevalence of other congenital anomalies has been associated with the type of ARM according to other classifications, such as the Peña classification based on the presence and position of the fistula or the Wingspread classification (low, intermediate and high anorectal malformations) (8,9). In May 2005, during an international congress, a new system of classification (the Krickenbeck system) was devised; this system incorporates the anatomic description of the malformation, the type of surgical procedure performed and postoperative assessment of bowel movement, constipation and soiling (1,9).

The aim of our study is to evaluate the incidence of associated congenital anomalies in relation to the anatomic type of ARM as defined by the Krickenbeck classification.

Materials and Methods

This was a retrospective observational study, conducted by the Department of Paediatric Surgery at "Grigore Alexandrescu" Children's Hospital. The study period lasted from January 2005 to December 2011. Data regarding the patient demographics, type of ARM and associated anomalies were collected and then classified according to the Krickenbeck criteria. Abnormalities were categorized as cardiovascular, gastrointestinal, genitourinary, of the central nervous system, musculoskeletal and craniofacial. Chromosomal abnormalities were also recorded.

After having been stabilized, the patients were clinically examined in order to identify the type of anorectal malformation, visible associated anomalies and other abnormalities. For patients with imperforate anus without fistula, an invertogram was performed so that the level of distal gas shadow could be identified.

An ultrasound was performed in most cases in order to rule out urogenital anomalies. For those patients identified with genitourinary abnormalities during the ultrasound investigation, a micturating cystourethrogram was used in order to evaluate the defect.

If there were positive findings during auscultation of the chest, an echocardiogram was requested. Plain abdominal and thoracolumbosacral radiographs were also performed.

Based on the presence or absence of associated congenital anomalies, the patients were separated into two groups.

Table 1. *Patients with ARM according to the Krickenbeck anatomic classification*

Type of ARM	Number	Male	Female
Imperforated anus without fistula	20	15	5
Perineal fistula	9	9	NA*
Cloaca	6	NA	6
Rectovestibular fistula	11	NA	11
Anal stenosis	2	2	NA
Rectourethral fistula	1	1	NA
Rectovesical fistula	1	1	NA
TOTAL	50	28 (56%)	22 (44%)

*NA= Not applicable

Results

A total of 50 patients fulfilled the criteria for inclusion in this series (*Table 1*). There were 28 males (56%) and 22 females (44%) (ratio 1.27:1). Weight at birth was 2.76 kg (range 1.36-4.1 kg). Age at presentation varied from 4 hrs to 14 days (mean 1.12 days). There were 20 premature infants, born within 32 to 36 weeks of gestation.

The majority of the patients were diagnosed with imperforated anus without fistula (40%), followed by rectovestibular fistula (22%), perineal fistula (18%) and persistent cloacal anomaly (12%). There were 2 cases with anal stenosis and one case with rectourethral and rectovesical fistula.

Associated anomalies according to organ system

34 (68%) of the 50 patients diagnosed with ARM had associated congenital anomalies and syndromes. The major groups of systems involved were gastrointestinal (36%), genitourinary (24%) and cardiovascular (16%). *Table 2* shows the frequency of malformations seen in association with the anatomic type of ARM according to the Krickenbeck classification.

Out of the 34 patients presenting associated congenital abnormalities, 14 (41.17%) had multiple congenital anomalies, affecting different or even the same system (*Table 3*).

The most common gastrointestinal anomaly we encountered in our series was oesophageal atresia with tracheo-oesophageal fistula, which was found in almost half of the patients presenting gastrointestinal defects.

Among genitourinary malformations, hypospadias, hydronephrosis, renal aplasia and dysplasia and bladder exstrophy were the most common pathologies encountered in the series.

As for the cardiovascular anomalies, the most frequent abnormalities found in our patients were patent foramen ovale and atrial and ventricular septal defects.

Table 2. *Frequency of malformations seen in association with the type of ARM*

Type of ARM	Number	Gastrointestinal	Genitourinary	Cardiac	Others
Imperforated anus without fistula	20	9 (45%)	6 (30%)	0	3a (15%)
Perineal fistula	9	0	3 (33.33%)	0	2b (22.2%)
Rectovestibular fistula	11	3 (27.27%)	0	5(45.5%)	2c(18.18%)
Cloaca	6	5 (83.33%)	2 (33.33%)	1(16.7%)	1d(16.7%)
Anal stenosis	2	1 (50%)	0	0	0
Rectourethral fistula	1	0	0	1(100%)	0
Rectovesical fistula	1	0	1(100%)	1(100%)	0
TOTAL	50	18 (36%)	12 (24%)	8 (16%)	8 (16%)

[a]Includes 1 patient with Down syndrome and 1 patient with musculoskeletal anomalies and 1 with cerebral malformations.
[b]Includes 1 patient with hemangiomas and 1 with cerebral malformations.
[c]Includes 1 patient with cleft palate, limb anomalies and spina bifida and 1 with hemangiomas.
[d]Includes 1 patient with cleft lip and limb anomalies.

Table 3. *Anomalies associated with anorectal malformations according to organ system*

System	Anomaly	Number of cases
Genitourinary	Hypospadias	3
	Hydronephrosis	2
	Renal aplasia and dysplasia	2
	Renal hypoplasia	1
	Pseudohypospadias	1
	Renal agenesis	1
	Ectopic testicle	1
	Sexual ambiguity	1
	Atresia of urethra	1
	Penile agenesis	1
	Bladder exstrophy	2
Gastrointestinal	Esophageal atresia with tracheoesophageal fistula	8
	Colon agenesis	2
	Ileal atresia	1
	Malrotation	2
	Duodenal stenosis	1
	Meckel's diverticulum	1
	Omphalocele	4
Cardiovascular	Patent foramen ovale	4
	Atrial septal defect	3
	Ventricular septal defect	2
	Dextrocardia	1
	Bilateral superior vena cava	1
Musculoskeletal	13th rib	2
	Limb anomalies	3
	Vertebral malformations	2
Craniofacial	Cleft lip	1
	Cleft palate	1
Central nervous system	Spina bifida	1
	Cerebral malformation	2
Chromosomal abnormalities	Down Syndrome	1

The imperforated anus without fistula group had the highest proportion of other associated anomalies, 15% of this group being affected by musculoskeletal anomalies, cerebral malformations and chromosomal abnormalities, represented by a patient diagnosed with Down syndrome.

Comparison of associated congenital abnormalities between types of anorectal malformation

Due to the fact that there were no normal controls for comparison, patients with rectourethral fistula were used as the base group for comparison between each group, as this group has the lowest number of associated malformations out of all categories. (*Table 4*)

In our study, the patients diagnosed with persistent cloacal anomaly were most likely to have associated gastrointestinal malformations (19 times more likely than the base group), followed by the patients with imperforated anus without fistula (3 times more likely than the base group).

In the present series, patients with perineal fistula were most likely to have a genitourinary malformation. We also found that the patients presenting rectovestibular fistula were most likely to have cardiac abnormalities (70 times more likely than the base group) followed by those persistent cloacal anomalies (7 times more likely than the base group).

Discussion

Our study aims to demonstrate the high incidence of associated congenital anomalies seen in patients presenting ARM, by associating these abnormalities to the anatomic type of malformation as described in the Krickenbeck classification.

ARMs represent a frequently encountered pathology in the general paediatric surgical practice. The morbidity and mortality associated with the evolution of these patients are mostly related to the wide spectrum of other associated malformations.

The reported range of other anomalies varies between 28% and 70% (6,7). In our series, 68% of the patients had associated anomalies; that lies within the indicated range.

Previous series that have studied the association between ARM and other congenital anomalies have used older classifications of anorectal malformations, such as Wingspread or Peña, but by following the guidelines of the Krickenbeck criteria, a better comparison between groups of patients with ARM from different centers and a more precise evaluation of factors that may modify the outcome of the surgical treatment can be done (10,13).

Table 4. *Comparison of associated congenital anomalies between the types of ARM by using rectourethral fistula as base group*

Type of ARM	Gastrointestinal		Genitourinary		Cardiac		Others	
	OR	95% CI	OR	95% CI	OR	95% CI	Others	95% CI
Imperforate anus without fistula	3.016	0.588-6.196	2.270	0.493-6.351	5.126	0.591-0.910	1.054	0.186-4.192
Perineal fistula	1.136	0.579-0.893	2.355	0.370-8.553	5.629	0.692-0.936	1.992	0.277-10.025
Rectovestibular fistula	0.948	0.137-2.624	0.917	0.562-0.853	69.930	1.879-53.230	1.460	0.210-7.117
Cloaca	18.836	1.266-112.287	2.252	0.271-10.683	7.392	0.107-10.481	1.263	0.107-10.481
Anal stenosis	2.882	0.107-31.031	0.993	0.637-0.883	5.825	0.734-0.946	0.995	0.734-0.946
Rectourethral fistula	1.000	NA	1.000	NA	1.000	NA	1.000	NA
Rectovesical fistula	1.000	0.511-0.784	0.297	0.133-0.378	1.000	0.072-0.285	1.000	0.739-0.948

OR = Odds Ratio; 95% CI = 95% confidence interval; NA = not applicable

Genitourinary abnormalities

The most common associated anomalies reported across the world were related to the urogenital system (1), approximately 50% of patients with ARM being also found to have urinary defects (10).

In this series, only 24% of patients were diagnosed with this type of malformations, the most common pathology among the genitourinary system being hypospadias and hydronephrosis.

Goossens et al have shown in their study that the incidence of genitourinary anomalies decreases with the diminishing complexity of the anorectal malformation. As such, the patients with perineal fistula had the lowest incidence of genitourinary abnormalities, while patients with more complex malformations, such as rectovesical fistula had the highest incidence of associated genitourinary defects.

Our findings also showed a high incidence of genitourinary defects in the rectovesical fistula group (although it must be specifies that in our series, only 1 patient was diagnosed with this type of anorectal malformation), but the incidence of genitourinary abnormalities in patients with perineal fistula was second highest with 33.33%, the same as for patients with persistent cloacal anomaly. These results are noticeably different from the ones presented in other studies.

Gastrointestinal anomalies

In our series, the system which was most affected was the gastrointestinal tract; these anomalies were seen in 36% of our patients, which is higher than the reported 9% to 24% encountered in other studies (1). The most frequent gastrointestinal malformation seen in our patients was oesophageal atresia with tracheo-oesophageal fistula with an incidence of 57%. This number was higher than the one found in other reports, where the incidence of this association was between 11-15% (11,13).

The highest incidence of gastrointestinal malformations was found in out persistent cloacal anomaly group (83.33%), while the lowest incidence was in the rectovestibular fistula group. The anal stenosis and imperforated anus without fistula groups had similar numbers of gastrointestinal defects.

Cardiac anomalies

The incidence of cardiac anomalies found in patients with ARM varies between 7-10% (1), with the most frequent pathology being ventricular septal defect. Other studies have also shown an increased association between ARM and tetralogy of Fallot (14).

In our series, the most common cardiac defects were patent foramen ovale and atrial septal defects. Ventricular septal defects were found in only 2 cases, while no cases of tetralogy of Fallot were encountered.

Both patients with rectourethral and rectovesical fistula included in our study were diagnosed with atrial septal defects. We also found that 45.5% of the newborns included in the rectovestibular fistula group had associated cardiac anomalies, while no cardiac abnormalities were encountered in the imperforated anus without fistula, perineal fistula and anal stenosis groups.

Chromosomal anomalies

Other studies have shown that nearly 50% of patients with imperforated anus without fistula also presented chromosomal abnormalities (especially Down syndrome) (4,5). In our study, out of the 20 patients who were diagnosed with this type of ARM, only 1 (5%) was also diagnosed with Down syndrome.

Conclusion

In our study we have concluded that 68% of the newborns with anorectal malformations had associated anomalies, which lies within the reported range. We found gastrointestinal anomalies to be the most common associated congenital defects in our patients; we encountered a higher incidence of this type of abnormalities in newborns with persistent cloacal anomaly, while the groups presenting rectovestibular fistula were most likely to present cardiac abnormalities. The incidence of genitourinary anomalies found in the group with perineal fistula is higher than the one described in other studies.

Regardless of the type of anorectal malformation, it is imperative that a thorough clinical evaluation and systemic investigations of all patients be done in order to exclude or

confirm the presence of gastrointestinal, genitourinary and cardiac abnormalities.

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