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ORIGINAL ARTICLE

Gynecological anomalies in patients with anorectal malformations

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Abstract

Purpose The association of gynecological anomalies in all anorectal malformations (ARM) is firmly established. Our goal is to study this pathology in our patients to focus attention to this important issue.

Methods Retrospective study of female patients operated for ARM and who underwent magnetic resonance imaging in our center. The type of malformation, the presence and type of vaginal, uterine, tubaric and urological anomalies were studied.

Results 63 patients were included: 34.9% cloaca, 28.6% vestibular and 12.7% perineal. Half of patients had some type of müllerian anomaly; 19 vaginal, most frequent being the longitudinal vaginal septum (66.7%); 30 had uterine alterations, most frequent being the uterus didelphys (60%). Eighty percent of patients with complex ARM (cloaca, exstrophy) presented some type of gynecological malformation compared to 21.8% found in simple ARM (stenosis, perineal, vestibular) ($p < 0.001$). Vaginal anomalies are associated with a uterine anomaly in 100% of cases. Conversely, patients with uterine anomalies have concurrent vaginal anomaly in 63.3% of cases.

Conclusion Screening for gynecological anomalies is indicated in all patients with ARM. We recommend a vaginal examination in any girl with ARM during definitive repair and a subsequent MRI during follow-up. Collaboration with a gynecologist is essential.

Keywords Anorectal malformation · Müllerian · Gynecological anomalies

Introduction

Anorectal malformations (ARMs) affect between 1:2000 and 1:5000 live births, and represent a broad spectrum of malformations that considerably differ in their anatomical characteristics, complexity, and functional prognosis [1]. The surgical reconstruction of any ARM aims to achieve adequate fecal, urological, and gynecological or sexual function.

The association of gynecological anomalies in all anorectal malformations (ARM) is firmly established and ranges from 26 to 39% [1] and, therefore, one must actively check for this possibility in every female child presenting with ARM. Early detection is essential to allow adequate treatment and follow-up into young adulthood. A delayed or missed diagnosis may result in adverse late consequences for the patient. The definitive repair of the anorectal malformation offers an ideal opportunity for adequate diagnosis and management of most of these müllerian anomalies.

The purpose of this report is to study the associated gynecological pathology in our female patients with anorectal malformations to focus attention to this important issue.

Methods

A retrospective review was performed of the medical records of 63 patients with ARM and who underwent magnetic resonance imaging (MRI) in our center. The type of malformation, the presence and type of vaginal, uterine, tubaric and urological anomalies were studied.

Results

A total of 63 patients were included. The distribution of the type or ARM is shown in Fig. 1 and was as follows: 34.9% cloaca, 28.6% vestibular and 12.7% perineal. The

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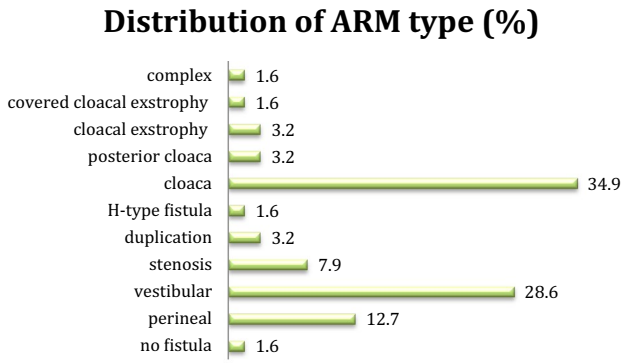

Fig. 1 Types of ARM in our series (%)

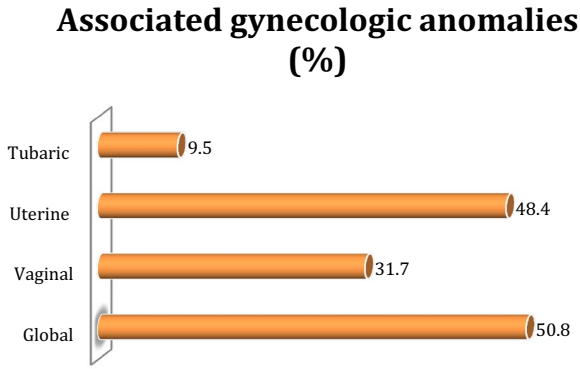

Fig. 2 Associated gynecologic anomalies in female ARM patients (%)

mean age at study was 12 years and the mean age at MRI was 6.6 years old.

Of these 63 patients, 32 (50.8%) had some type of müllerian anomaly; 19 (30.2%) were found to have a congenital vaginal anomaly, 30 (48.4%) presented some form of associated uterine malformation and 6 (9.5%) tubaric (Fig. 2).

Within the vaginal anomalies, the most frequent defect found was the complete longitudinal vaginal septum (66.7%) (Fig. 3) followed by vaginal agenesis (27.8%); and within the uterines the most frequent was the uterus didelphys (60%) (Fig. 4) and septate uterus (13.3%) (Figs. 5, 6).

Eighty percent of patients with complex ARM (cloaca, cloacal exstrophy) presented some type of gynecological malformation compared to 21.8% found in more simple forms of ARM (stenosis, perineal fistula, recto-vestibular fistula), this difference was statistically significant ($p < 0.001$). More specifically, of 18 patients with recto-vestibular fistula 5 (27.8%) had associated müllerian anomalies, 2 (11.1%) vaginal (5.6% septum and 5.6% agenesis) and 5 (27.8%) uterine, while out of 22 patients with cloaca 18 (81.8%) had

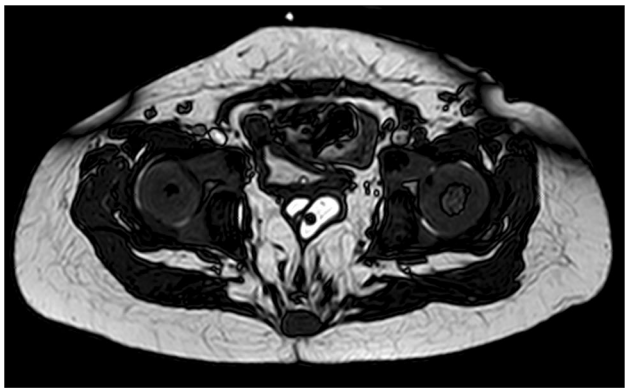

Fig. 3 Axial 3D BFEE section through pubic symphysis showing longitudinal vaginal septum in a girl with cloacal malformation

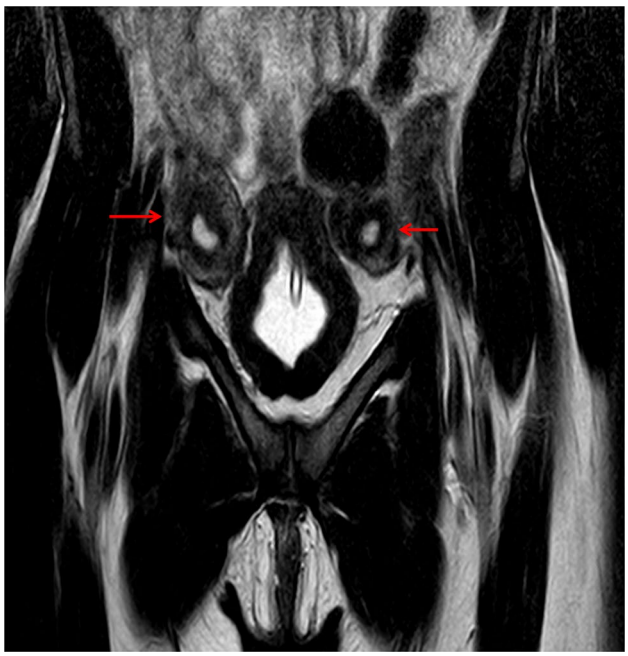

Fig. 4 Coronal T2 HR image showing double uterus (red arrows) in a girl with cloacal malformation

congenital gynecological malformations (59.1% vaginal, 77.3% uterine and 22.7% tubaric) (Fig. 7).

We found that vaginal anomalies were associated with a uterine anomaly in 100% of cases. Conversely, patients with uterine anomalies had a concurrent vaginal anomaly in 63.3% of cases.

Cystic problems in the internal genital organs were found in 2 (20%) of the senior author's 10 adolescent cloaca patients studied.

Thirty-four percent of cases had associated renal anomalies (half of them were hydronefrosis). Patients with gynecological malformation presented more frequently associated urological anomalies than those who did not ($p < 0.01$).

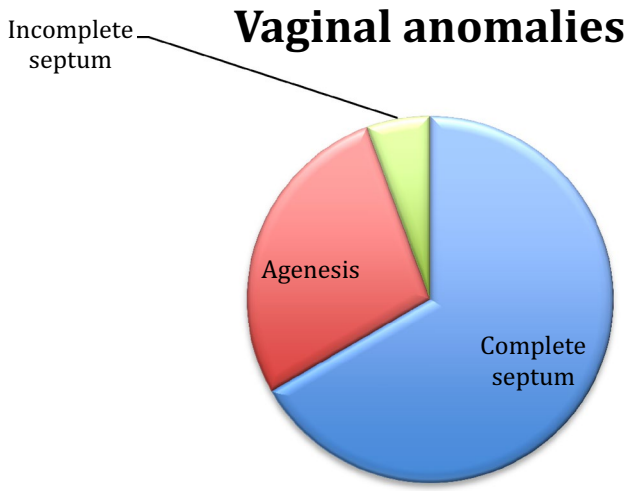

Fig. 5 Type of vaginal anomalies (%)

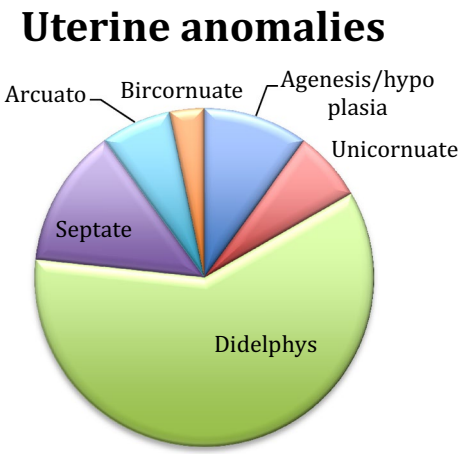

Fig. 6 Type of uterine anomalies (%)

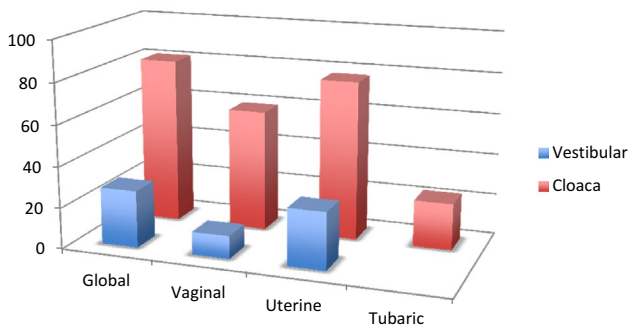

Fig. 7 Associated gynecologic anomalies in vestibular fistula and cloaca (%)

Discussion

Female children with ARM often associate gynecologic anomalies, which should be identified and addressed. The time of definite repair of the anorectal malformation represents a unique opportunity for the evaluation and treatment of vaginal defects and prompt recognition of uterine anomalies allows important counseling in reproductive and obstetric prognosis. This chance may be missed unless the pediatric surgeon is aware of this association with potential for negative consequences for future sexual and reproductive function.

Associated utero-vaginal anomalies in patients with recto-vestibular fistula are more frequent than previously thought and high index of suspicion is appropriate in girls with this type of anorectal malformation. We found that 27.8% of our patients had some form of müllerian anomaly. Others authors have reported similar findings. Levitt et al. [2] described that 17% of 272 patients with recto-vestibular fistula had a gynecologic anomaly, and, as in our cases, vaginal septum occurred in 5% and 8% had an absent vagina, thereby justifying the search for this defect, at the time of the main repair of the anorectal malformation. In consequence, as other authors have already pointed out, vaginoscopy should be performed on all girls during the definitive repair of any type of ARM with preparation to undertake vaginal septum resection if diagnosed [3, 4].

There is a strong association of gynecologic anomalies with a cloacal anomaly. Breech et al. [5] found that 53–67% of female cloaca patients were born with utero-vaginal anomalies and Rintala et al. [6] found that all patients with cloacal anomalies had abnormal genital tract and 40% of patients had duplicated müllerian system with two vaginas and uteri. In line with this data 81.8% of our cloaca patients had some sort of congenital gynecological malformations and we found that utero-vaginal duplication was the most frequent genital anomaly. The time of colostomy closure in these patients may be an opportunity for exploring the appearance of the uterus. This may be also the right time to check the patency of the fallopian tubes since up to 19% our patients had tubaric anomalies with further complications. Peña et al. [7] routinely check the patency of the müllerian structures whenever they have the opportunity to be in the abdomen of their patients.

A significant characteristic of the complete vaginal septum is the fact that, in our series, it was always associated with the presence of 2 hemiuteri. These findings have been also reported by others. Breech et al. [8] found that longitudinal vaginal septums were associated with a uterine anomaly (septate or didelphys) in 95% of cases and, similar to our series, patients with uterine duplication had a concurrent longitudinal vaginal septum in 75% of cases

[8]. A routine examination as simple as a vaginal exploration at the time of the repair of the rectum can, therefore, detect congenital uterine anomalies with future obstetric implications.

Most young women who were operated on for ARM during childhood enjoy healthy sexual relationships and pregnancy is possible even in the most complex ARM [6, 9]. In women with an associated reproductive anomaly, the specific type of anomaly will influence the potential for conception and pregnancy risks: spontaneous pregnancy loss ranges from 32% in uterine didelphys to 36% in unicornuate uterus and preterm delivery from 23% in didelphus to 45% in unicornuate [5]. Knowledge of reproductive anatomy provides the pediatric surgeon and the gynecologist with optimal medical and surgical management during the follow-up of these patients.

In the light of these data and their possible future consequences in terms of sexual activity, pregnancy and mode of delivery, in the management of females with an ARM it is mandatory to obtain a description of the reproductive anatomy as completely as possible during childhood. The diagnosis can be made with MRI [8]. Imaging should include an assessment of the kidneys and urinary tract since we found up to 34% of cases had associated renal anomalies.

We have learned throughout our practice that MRI in very young patients is sometimes not able to assess the internal genital anatomy accurately and based on our experience we have postponed the indication of MRI to 7–8 years of age, which has the added benefit of being performed without anesthesia. According to our imaging protocol, the MRI is also performed to evaluate the urinary tract and spinal cord as well as possible postoperative complications.

In addition, at the time of puberty (which begins with breast development) a standardized follow-up is necessary. Ultrasound surveillance of the reproductive structures should begin after thelarche and continue every 6–9 months through menarche [5, 9].

Other gynecologic concerns for pubertal females include the development of pelvic cysts [6]. In line with our results, Breech et al. found that 19% of their adult or adolescent cloacal patients required surgical interventions for large cystic collections that involved uteri and adnexal tubes. These cysts were not related to menstruations and were histologically benign paraovarial cysts [5]. In our experience we found that some of these cysts are postoperative pseudocyst formed by peritoneum rather that real cyst. Whether these cysts need to be operated on has yet to be established.

In brief, the recognition of gynecological anomalies in females with ARM allows for more optimal medical and surgical care. Our management of these patients includes a vaginoscopy at the time of definitive repair surgery, MRI after 7–8 years of age, a pelvic ultrasound every 6–9 months after thelarche and follow-up by a specialized gynecologist.

Conclusions

In conclusion, it is important to be aware of the association between anorectal malformations and müllerian anomalies. Screening for gynecological-related concerns is indicated in all patients with ARM. We recommend a vaginal examination in any girl with ARM during definitive repair and a subsequent MRI during follow-up. Knowledge of reproductive anatomy in females with ARMs provides optimal planning and surgical management, avoiding negative consequences to future sexuality and fertility. Collaboration with an experienced gynecologist is essential in these patients.

Author contributions MF, AL and EM conceived the idea. MF and AL drafted the database, which was further completed with the contribution of all other authors. MF wrote a first draft of the introduction and of the results. MF, AL, EM and JC wrote the discussion and contributed to shape the final version of the manuscript.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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