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ORIGINAL ARTICLE

Anorectal malformation without fistula: a defect with unique characteristics

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Abstract
Introduction Anorectal malformations (ARMs) without fistula occur in approximately 5 % of all cases of ARM. The high frequency of Trisomy 21 associated to this type of malformation has been previously described. A review of the literature revealed only one previous publication discussing ARM without fistula with or without Trisomy 21; all other publications focused their discussion in patients with Trisomy 21. We felt that ARM without fistula has very specific characteristics and therapeutic implications that deserved a special discussion, which prompted us to review our experience.
Methods A retrospective review of the medical records of patients with ARM without fistula was performed between September 1980 and March 2014. From a series of 2,189 cases of ARMs, 92 had no fistula identified. Information related to demographic, anatomic, and prognostic factors, as well as outcome variables was obtained and compared to those results obtained from other types of ARMs.
Results Seventy-six patients were males and 16 females. Thirty-seven patients (40 %) had Trisomy 21. Eighty-six patients were primarily operated on and six had a reoperation after a failed attempted repair at another institution. Of the six patients that were reoperated, five had an attempted repair in the newborn period without a colostomy and the operation was aborted after the rectum could not be found. The location of the blind rectum was at the level of the bulbar urethra in males or 1–2 cm from the perineal skin in females in 80 patients; and in 9 patients it was found at the level of the prostatic urethra. In five patients, during the repair, there was an incidental opening of the urethra, which was repaired with uneventful recovery. The sacrum was normal in 61 patients; 4 patients had a sacral ratio <0.4, which indicated poor prognosis for fecal continence. Sixty-four patients had normal urinary tract, four patients had an absent kidney, ten had bilateral hydronephrosis, and three unilateral. Long-term outcomes related to bowel control were available in 52 cases: 11 of 18 patients with Trisomy 21(61 %) had voluntary bowel movements and 29 of 34 (85 %) without Trisomy 21 had voluntary bowel movements. All patients without Trisomy 21 had urinary control.
Conclusion Anorectal malformation without fistula is a unique defect. In our series, it occurs in 4 % of all ARMs. Even when patients do not have a fistula, the operation is not technically easier due to the presence of an extensive common wall between the rectum and urethra in males or vagina in females. The ARM with no fistula has a good reasonable functional prognosis, even in patients with associated Trisomy 21 and conveys a low frequency of associated urologic defects.

Keywords Anorectal malformation · Imperforate anus · No fistula · Trisomy 21 · Down syndrome

Introduction

Anorectal malformations (ARMs) without fistula occur in approximately 5 % of all cases of ARM. The high frequency of Trisomy 21 associated to this type of malformation has been previously described [1, 2]. A review of the literature revealed only one previous publication

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discussing ARM without fistula with or without Trisomy 21 [3]; all other publications focused their discussion in patients with Trisomy 21. We felt that ARM without fistula has very specific characteristics and therapeutic implications that deserved a special discussion, which prompted us to review our experience.

Methods

A retrospective review of the medical records of patients with ARM without fistula was performed for the period between September 1980 and March 2014. From a series of 2,189 cases of ARMs, we identified 92 without a fistula. Information related to demographic, anatomic, and prognostic factors, as well as outcome variables was obtained and compared to those results obtained from other types of ARMs.

IRB approval was obtained for this study (#2013-0737).

Results

Seventy-six patients were males and 16 were females. Thirty-seven patients (40 %) had Trisomy 21.

Forty-four patients had a descending colostomy with separated stomas, 25 had a loop colostomy, 11 patients had transverse colostomies, and in 12 patients this information was not available.

Eighty-six patients were primarily operated on and six underwent a reoperation after a failed attempted repair at another institution. Of the six patients that were reoperated, five had an attempted repair in the newborn period without a colostomy and the operation was aborted after the rectum could not be found.

The location of the blind rectum was at the level of the bulbar urethra in males or 1–2 cm from the perineal skin in females in 80 patients (90 %). In nine patients (10 %) the blind rectum was found at the level of the prostatic urethra, and in three patients this information was not available.

In five patients, there was an incidental opening of the urethra during the main repair, which was immediately repaired with uneventful recovery.

The sacrum was normal (sacral ratio >0.7) in 61 patients. Thirteen patients had a sacral ratio between 0.5 and 0.69. Four patients had a ratio lower than 0.4, which indicated poor prognosis for fecal continence. There was no information about the sacrum in 14 patients. The average AP sacral ratio of patients without fistula was 0.55 and 0.65 in lateral view. These ratios were not different from those found in cases of recto-urethral bulbar (AP 0.6, Lateral 0.76) or prostatic fistula (AP 0.56, Lat 0.65).

Urologic associated defects

Four patients had a single kidney (4.3 %). This defect occurred more frequently in other malformations (bulbar 11 %, prostatic 16 %, and bladderneck 31 %).

Hydronephrosis (10 bilateral, 3 unilateral) occurred in 14 % of this series which was not significantly different from other malformations including bulbar fistula (9 %), prostatic (6 %), and bladderneck (24 %).

Vesico-ureteral reflux occurred in four cases (4.3 %) of patients without fistula; 13 % of bulbar fistula, 26 % of prostatic and 29 % of bladderneck.

Spinal and vertebral associated defects

Tethered cord occurred in 25 % of the patients without fistula and evaluated for this condition. This figure is similar to the average of frequency of tethered cord in all other defects.

Hemivertebrae were present only in two cases (2.1 %) of patients without fistula; 8 % of cases with bulbar fistula, 9 % of prostatic and 15 % of bladderneck.

Cardiovascular associated defects

Twenty-two patients without a fistula (23.9 %) suffered from 27 cardiovascular malformations. This association was present in 19 % of the bulbar urethral fistula cases; 21 % of prostatic and 23 % of bladderneck. However, the incidence of cardiovascular associated defects in Trisomy 21 was 43 % (16/37) whereas in those patients without Trisomy 21 it was 10.9 % (6/55).

The most common cardiovascular malformations were: patent ductus arteriosus (6), ventricular septal defect (6), atrial septal defect (4), patent foramen ovale (4), A/V canal (3), tetralogy of Fallot (1), bicuspide aortic valve (1), and a non-specified "cardiac malformation" (1).

Gastrointestinal associated defects

Only one patient without fistula had esophageal atresia, whereas this associated malformation occurred in 8 % of the general series. Interestingly, there were no duodenal atresia cases in the present series of patients without fistula.

Associated syndromes or conditions that may produce developmental delay occurred in seven patients without Down syndrome. These included: three demonstrated developmentally delayed patients, bilateral coloboma (1), absent corpus callosum (1), Opitz syndrome (1), and Apert syndrome (1).

Long-term outcomes related to bowel control were available in 52 cases. Forty patients (76.9 %) had voluntary bowel movements: 11 of 18 patients with Trisomy 21(61 %)

had voluntary bowel movements and 29 of 34 (85 %) without Trisomy 21 had voluntary bowel movements.

Long-term outcomes related to urinary control were available in 48 cases. Forty-five patients were urinary continent. The three patients that were urinary incontinent had Down syndrome. In other words, all patients without Trisomy 21 had urinary control and 10 out of 13 (76.9 %) with Trisomy 21 had urinary control.

Three patients underwent unnecessary investigation for Hirschsprung disease due to constipation problems and all resulted negative.

Discussion

The incidence of ARMs without fistula in our series was 4.2 %, which is not different from previous reports. The preponderance of this defect in males with this defect (76 males/16 females) was surprisingly higher than in our own general series that include all other malformations.

Traditionally, we liked to say that half of these patients suffer from Down's syndrome; our findings showed a slightly lower frequency, since 40 % of these cases were affected by that syndrome.

We were able to confirm that the overwhelming majority of cases without fistula have the rectum located in a position that can be reached comfortably via a posterior sagittal approach on males and females. The old literature [4] frequently mentioned "high" malformations without a fistula. We believe that most likely; those cases represented misdiagnosis, consecutive to a technically deficient high pressure distal colostogram.

Even when we recognize that ARMs without fistula belong to the "benign" side of the spectrum of anorectal defects, we were surprised by the fact that we experienced five incidental openings of the urethra. Those cases happened in the beginning of our experience. We feel that it was a consequence of our over confidence and wrong assumption that the dissection would be easier in a case without a fistula to the urinary tract. We now recognize that the fact that these patients have no fistula, does not mean that their repair is necessarily simpler. Actually, the common wall without a natural plane of separation, between the posterior urethra and the anterior rectal wall, is longer than in higher defects and that makes the repair a technically demanding procedure. In females, the common wall is with the vagina.

The average sacral ratio was not different from those found in cases of recto-urethral bulbar and prostatic fistulas. These findings confirm our impression that we were dealing with a malformation with reasonably good functional prognosis.

In general, the most common anatomic urologic defect associated with anorectal malformations is absent kidney. Our patients without a fistula had a significantly lower frequency of association with this condition when compared with other malformations. Hydronephrosis which is the second most common defect seen in association with ARM, in our series of cases without fistula had an incidence not much different from other defects. Vesico-ureteral reflux, on the other hand, had a much lower incidence than in other malformations which seems consistent with our initial impression that ARM without fistula appears to be a more benign malformation when compared to others; except cases of recto-perineal fistulas.

Tethered cord occurred with the same frequency in cases with or without a fistula; however, we feel that our figures are not very significant because very few patients were evaluated to rule out this condition.

The frequency of association of cardiovascular defects was not significantly different between patients with fistula and no fistula; however if one considers only the patients with Down's syndrome, the frequency appears to be higher.

A significant difference was also found in cases with esophageal atresia. This important malformation occurs in about 8 % of all ARM cases. Yet, only one case was found in the series without a fistula. We were also surprised by the fact that in these series there were no patients with duodenal atresia.

The group of patients with ARM without fistula and without Down syndrome, has a good functional prognosis, yet it seems to have a peculiar pattern of association with unusual defects and syndromes. A significantly high percentage of cases (12.7 %) were associated with syndromes and (or) conditions that have a well-known impact on the intellectual development (microcephaly, absent corpus callosum, bilateral coloboma, Opitz, and Apert syndromes).

In general, the functional prognosis for ARM without fistula is reasonably good (75 % bowel control), particularly for patients without Trisomy 21 (85 % bowel control). Understandably, the patients with Down syndrome have a less favorable prognosis. We know that the degree of developmental delay in those patients is variable, which is a very important factor to have control of bowel movements. However, this finding confirms our belief that patients with Down syndrome are not necessarily candidates for a permanent colostomy.

As expected, urinary control was normal in all patients without Trisomy and in 76 % of patients with Down's syndrome.

Conclusion

Anorectal malformations without fistula represent a unique group of defects with special characteristics that include:

A high frequency of Trisomy 21 and other syndromes, that may affect intellectual development.

All patients have a blind rectum located low enough to be reached comfortably with a posterior sagittal approach.

The complexity of the surgical repair of this defect must not be underestimated; the surgeon must keep in mind that the rectum and urethra (or vagina) shares a long common wall without a plane of separation.

The frequency of association with urologic defects and esophageal atresia is lower than in other ARMs.

Patients born with ARM without fistula have a good functional prognosis, particularly if they do not suffer from Down syndrome. However, they have a significantly high risk of suffering from other conditions that may produce developmental delay.

References

1. Black CT, Sherman JO (1989) The association of low imperforate anus and Down's syndrome. J Pediatr Surg 24:92–94
2. Torres R, Levitt MA, Tovilla JM et al (1998) Anorectal malformations and Down's syndrome. J Pediatr Surg 33:194–197
3. Fanjul M, Molina E, Cerda J et al (2009) Characteristics of the anorectal atresia without fistula. Based on 12 cases. Cir Pediatr 22:45–48
4. Stephens FD, Smith ED (1971) Individual deformities in the male. In: Ano-rectal malformation in children. Year Book Medical Publishers, Chicago, pp 33–35