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Abnormalities in "low" anorectal malformations (ARMs) and functional results resecting the distal 3 cm

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Abstract

Purpose: "Low" anorectal malformations (ARMs) are considered minor anomalies of the distal rectum and anal–canal development. Nonetheless, the prognosis of affected patients is far from excellent, as some degree of constipation is a frequent complaint in the long-term follow-up. Constipation in "low" ARM has been reported in 42%–70% of cases. Vestibular fistulas seem to have the highest rate of constipation (not less than 61.4%). The aim of this study was to evaluate all the histological wall abnormalities of ARM with recto-perineal and recto-vestibular fistulas in order to identify features that could explain the bowel dysfunctions. Moreover, the resection of distal perineal and vestibular fistulas (last 3 cm) allowed evaluating functional results in "low" ARM series with extensive fistula resection.

Methods: One hundred four specimens were collected from 52 patients (32 recto-perineal and 20 recto-vestibular fistulas) during the posterior sagittal anorectoplasty (PSARP). The distal 3 cm of aberrant anorectal canals (fistulas) was systematically resected and divided longitudinally. One portion was fixed for immuno-histochemical stainings (PGP 9.5, S-100, NSE), H&E, and tricromic stainings. The frozen sections of the second portion were incubated for enzyme histochemical stainings (AChE, etc.). The follow-up of 42 of 52 ARM was postoperatively evaluated at 3–8 years of age, and the assessment of the outcome after PSARP repair was in line with Krickenbeck's 2005 meeting parameters.

Results: Muscle coat was abnormal in all cases (100%), showing aspect and absence of organization into the circular and longitudinal layers. The connective tissue was found to be irregular and abnormally represented in 100% of cases. Abnormal vascularization was detected in 5 cases (9.6%). All vestibular (100%) and 71.8% of perineal fistulas showed different degrees of enteric nervous system (ENS) anomalies. In the series of 42 patients followed up at least after 3 years of age, 40 cases (95.2%) showed postoperative good continence without use of laxatives (according to Krickenbeck's 2005 criteria).

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Conclusion: Every wall component of the distal rectum can be affected by different structural abnormalities in "low" ARMs. Pediatric surgeons should take into consideration the implications of these structural abnormalities during radical treatment. The resection of a significant portion of the distal fistula seems to permit better functional results.

Anorectal malformations (ARMs) are relatively frequent congenital abnormalities of the distal rectum and anal canal that range from minor anomalies such as recto-perineal fistula to major ones such as cloaca or recto-vesical fistula. The most recent classification was published after Krickenbeck's Consensus Conference in 2005 [1]. The outcome of patients with ARMs has greatly improved during the last three decades since Posterior Sagittal Ano-Recto Plasty (PSARP) was introduced by Alberto Peña in the early 80s [1–3]. Nonetheless, surgical treatment of ARMs remains far from being perfect [4]. In particular, postoperative constipation is a common problem in the long-term follow-up of treated patients. The incidence of constipation varies depending on the severity of the malformation. ARM with vestibular fistula and recto-perineal fistula, defined "low," presents constipation in 42%–70% of cases [5,6]. Vestibular fistulas seem to have the highest rate [7–9]. The reasons for these different functional results are still controversial and the conviction that this group of ARMs represents a benign form which leads most of the surgeons to a less invasive approach. In fact, a minimal dissection has been recommended in these patients during the PSARP, allowed by a very small incision "mini-PSARP" [10]. Moreover some authors proposed "fistula-transposition" as a more physiological technique [11]. The present study was aimed to investigate accurately the microscopic structure of recto-perineal and recto-vestibular fistulas in order to identify possible intrinsic abnormalities of the distal rectum that could explain the high rate of postoperative chronic constipation in patients with "low" ARM.

1. Materials and methods

1.1. ARM patients and specimens

Specimens were harvested from 52 consecutive "low" ARM patients during the PSARP procedure (age range at surgery: 4 days to 2 years). A total of 32 ARM with recto-perineal and 20 ARM with recto-vestibular fistulas (10 males and 42 females, M:F ratio 0.2) were included in the study. Our series presented normal sacral ratio at lateral view (LSR) ranging from 0.58 to 0.81, according to the studies of Torre et al. [12]. In any case we performed MRI evaluation to exclude intraspinal pathologies in those cases which presented a LSR < 0.77, according to the normal value assessed by Peña [13]. No cases of the series of ARM patients were associated with major anomalies (cardiac malformations, meningocele, sacral agenesia, Currarino syndrome, etc…). However 9 cases (17.3%) were associated with urogenital anomalies (6 vescicoureteric reflux, 2 hydronefrosis, 2 unilateral renal hypoplasia, and 1 cryptorchidism). One ARM case (vestibular fistula) was associated with both unilateral renal hypoplasia and unilateral radial hypoplasia.

Most of the patients primarily underwent to PSARP procedure without a protective colostomy.

Nevertheless, 6 patients (11.5%) came to our observation with a previously performed diverting colostomy. In other 4 ARM cases (7.7%) with vestibular fistula the patients came to our observation when they were more than 8 months old and we decided to perform a divided descended colostomy before the radical management of the anomaly.

The standard PSARP approach, in ARM with perineal and vestibular fistulas, plans a short incision of about 2 cm that is created dividing the distal portion of the sphincter structures located posterior to the fistula. In our series we always performed a more complete PSARP approach. In every case a Foley catheter was inserted through the urethra, in order to avoid possible urethra injuries. The posterior sagittal incision was more extended and deeper than in the classical mini-PSARP procedure. We obtained to divide the entire sphincter mechanism (parasagittal fibers, muscle complex). In these "low" anomalies the *levator ani* is not located very deep into the perineum ("the higher the malformation, the deeper the levator muscle is located" [10]); in every case it was possible to practice a partial sagittal section of levator muscle in order to mobilize a longer portion of the fistula. Final anoplasty was performed using a new distal anorectal canal derived from the transitional portion that is present at the end of the fistula and the upper rectum. Tapering of the rectum was never performed.

Each patient underwent the resection of the last 3 cm of the distal aberrant anorectal canal (including the perineal or vestibular opening) (Fig. 1A and B). Each specimen was subsequently divided longitudinally into two distinct portions (Fig. 1B). One portion was fixed in formalin solution (10% in acetate buffer, pH 7), and paraffin-embedded tissue sections were stained for immuno-histochemical and histological study. The second portion was frozen in liquid nitrogen-cooled isopentane and then 15-μm cryostat sections were incubated for enzyme histochemistry. The use of different complementary immuno-histochemical, enzyme histochemical and conventional histology techniques allowed the comprehensive assessment of different components of the wall of the resected rectal fistulas: muscularis propria, connective tissue, blood and lymphatic vessels and enteric nervous system (ENS).