About the pathophysiology of anorectal malformations

Abstract

All congenital anorectal malformations (ARMs), except for very rare cases of true cloaca, have a normally formed anal canal. Different types of ARMs differ in the degree of anterior displacement of the anus, the opening of which is always narrow and rigid. Until 1982, most pediatric surgeons used a simple cutback procedure for perineal ectopia ani. Dissection of the fistula resulted in recovery. Currently, pediatric surgeons do not acknowledge the presence of an anal canal in ARMs. During posterior sagittal anorectoplasty (PSARP), they resect the internal anal sphincter (IAS), transect 90% of the external anal sphincter (EAS), and the puborectalis muscle (PRM). The bloodless and denervated rectum, torn from the levator plates, is brought down to the site of the removed IAS. As a result of extensive dissection, the neural connections of the pelvic organs are severed. Destruction of the anal canal leads to constipation, fecal incontinence, and megacolon. The intersection of neural connections leads to urological and sexual problems that worsen over time. Despite bowel management programs, antegrade enemas, and transitional-to-adult care, patients undergo repeated colorectal and urological surgeries and gradually disappear from follow-up. This article presents evidence that pediatric surgeons are ignorant of the anatomy and physiology of the anorectal area and analyzes the reasons for this. Pediatric surgeons are unfamiliar with the work of physiologists. Their knowledge is based on the "experience" of A. Peña. To bring pediatric surgeons back into the scientific mainstream, the American Pediatric Surgical Association should encourage physiologists and general gastroenterologists to publish articles in pediatric surgical journals on the anatomy and physiology of the anorectal area, which in children differs from that in adults only in size.

Keywords: anorectal malformations; physiology; cutback procedure; posterior sagittal anorectoplasty; internal anal sphincter; external anal sphincter; puborectalis muscle.

The internal anal sphincter (IAS) is a thickened continuation of the circular smooth muscle layer of the rectum, which plays an important role in the maintenance of fecal continence since it generates tone and is responsible for > 70% of resting anal pressure. It is in the anal canal, surrounded by striated muscles - the external anal sphincter (EAS) and puborectalis muscle

(PRM). Unlike the rectum, which expands to accommodate incoming feces, the IAS constantly contracts. Therefore, IAS and rectum have various electromechanical and biochemical characteristics [1]. Invisible nerve pathways connect all the muscles of the anal canal with each other and with the rectum. Long-term retention of feces in the rectum occurs because of tonic contraction of the IAS, EAS, and PRM. Periodic entry of a fecal bolus into the rectum leads to an increase in pressure within it. This causes a reflex relaxation (opening) of the IAS with simultaneous contraction of the IAS and PRM. This so-called rectoanal inhibitory reflex allows gas and liquid to penetrate the upper part of the anal canal. Since the communication between the rectum and anal canal is narrow at this moment, solid fecal matter remains in the rectum. The wall of the upper anal canal, into gas or liquid passes, contains sensory nerve elements that allow a person to differentiate their quality and act accordingly: pass gas or retain liquid. During relaxation of the IAS contraction of the EAS and PRM prevent involuntary defecation. The rectum relaxes, adapting to the new volume. Pressure in the rectum decreases, leading to contraction of the IAS and a decrease in the tone of the EAS and PRM. This situation can be repeated up to 18 times per hour [2] (Figure 1 b). When a volume of feces accumulates in the rectum that causes an increase in pressure capable of triggering the defecation reflex, a reflexive relaxation of the IAS, EAS, and PRM occurs with simultaneous contraction of the levator plates on both sides. They stretch the walls of the anal canal, creating a passageway for feces. At this point, the contracting rectum pushes the feces out (Figure 1c-e).

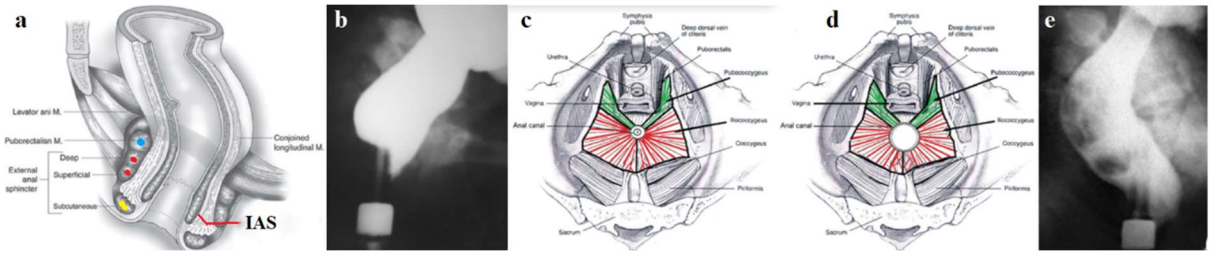

Figure 1. Normal anatomy and physiology of the anorectum. (a) Diagram of the anatomy of the anorectum from the book by Jorge and Habr-Gama [3]. The blue dot shows the PRM. The red dots show the deep and superficial parts of the EAS, and the yellow dot shows the subcutaneous part of the EAS. (b) Lateral radiograph of the infant's anorectum. A closed anal canal is visible between the rectum and the contrast marker placed near the anus. Since the marker's true diameter is 1.6 cm, the length of the anal canal behind the enema tip is 2.7 cm. Anterior to the enema tip, barium has entered the anal canal due to relaxation of the IAS. The posterior wall of the anal canal is pressed against the tip of the enema by the contracted PRM, and the lower part of the anal canal is closed by contraction of the EAS. This is the radiographic

equivalent of the rectoanal inhibitory reflex. (c-d). Scheme cross-section of the anorectal region at the level of the deep portion of the EAS. A scheme from the article of Bharucha was used [4]. (c) At rest anal canal is in a closed state. (d) During defecation, the anal canal opened (white circle) because of the contraction of the levator plates (pubococcygeus and iliococcygeus). (e) Lateral radiography of anorectum of the child during a barium enema. During defecation a wide channel arose, which corresponds to the width of the stool. A contrast marker strung on the tip of the enema is located near the anus.

At the request of the famous pediatric surgeon Duhamel Alamowitch, Gubler, and Roujeau (1966) examined the normal innervation of the internal anal sphincter. 100 rectums with anal canal were examined, 48 from deceased premature babies, 21 from stillborn full-term babies, 16 from deceased newborn babies and infants whose deaths were due to various causes, and 15 from adults. The IAS, which is a thickened extension of the internal circular layer of the rectal wall, was always found to be devoid of ganglionic cells. There is no Meissner plexus in the submucosa which covers it, and the rare ganglionic cells of the Auerbach intermuscular plexus (found in only 1 in 4) [5]. Based on this study, Duhamel concluded that "the internal sphincter itself has no autonomous innervation, unlike the rest of the digestive tube. Even if efferent nerves do exist at the level of this sphincter, there is a rupture of colo-rectal peristalsis at that point" [5].

Thus, the IAS differs from the rectal wall in the following characteristics: 1) It performs the function of retaining feces, but not its accumulation, ensuring, together with the striated muscles, a constant contraction of the anal canal2) it does not contain nerve ganglia and therefore does not participate in peristaltic; 3) it relaxes briefly with an increase in rectal pressure, which is designated as the rectoanal inhibitory reflex; 4) in a manometric study, this is manifested by a decrease in pressure in the upper anal canal in response to stretching of the rectum; 5) in an X-ray study, this is recorded as penetration of a contrast agent from the rectum into the upper anal canal in front of the enema tip. 6) IAS is part of the anal canal; 7) local neurological control is provided by the Cajal cells, which are considered the pacemakers of the IAS. They respond to pressure in the rectum [1,2].

The increase or decrease in sphincters tone, depending on the circumstances, is corrected by signals coming from postganglionic neurons originating from the inferior mesenteric ganglion or pelvic plexus and extending into the anal canal via the lumbar enteric nerves, hypogastric nerves or branches of the pelvic plexus [1,2]. An invisible nerve plexus surrounding the pelvic

organs provides communication between the organs, which is manifested by reflexes. Two anorectal reflexes are responsible: (1) for prolonged retention (the retention reaction), including the rectoanal inhibitory reflex, and (2) the defecation reflex [2].

About anal canal in ARMs in studies of pediatric surgeons.

I divided histological studies into 3 groups.

The first group of researchers compared the distal intestine with the anal canal of healthy individuals. Lambrecht and Lierse in 3 neonatal pigs with ARM found that the proximal region of the fistulae in ARM has most features of a normal anal opening. They consider that the fistula should be designated as an ectopic anal canal [6]. The most important result was the demonstration of a normal functioning IAS even in high and intermediate types of ARMs [6, 8]. Rintala et al have shown that in anorectal malformations the distal rectal pouch with the fistulous connection is anal canal ectopy [8]. The study by Uemura et al allowed them to conclude: "Epithelial and ganglionic distribution was similar in the distal rectal end of ARMs and in a normal anal canal. The anal transitional zone is the epithelial boundary between the rectum and skin in a normal anal canal. Anal transitional zone preservation could reproduce anal canal structure in ARM reconstruction" [9]. Docter et al presented micro-CT imaging to research resected material to provide new insights in microscale anatomy. The fistula, currently resected during surgical reconstruction for ARM, contains vital structures like the IAS, normal epithelial transition zone and normal ganglion cells. They believe that the fistula has a normal anal canal morphology and should be spared during ARM reconstruction if possible [10]. There are two types of articles in the literature that do not recommend using a fistula (rectal pouch) for ARM reconstruction.

In the second group, the authors based their recommendation on the absence of nerve ganglia or their insufficient number. For example, Holschneider et al. (1996) found that all fistulas were found to be aganglionic, including the adjacent part of the rectum involving the internal sphincter equivalent. In the authors' opinion, the recommendation to use the distal rectal pouch and parts of the fistula in the reconstruction of anorectal malformations should be reconsidered [11]. These authors for reconstruction of ARMs, used PSARP, which was proposed by Peña, who argued without any evidence that in ARMs the anal canal is absent [12]. Since then, the distal part of the intestine began to be called the "rectal pouch" or "fistula". Thus, Holschneider et al. did not know that the normal anal canal does not have ganglia. They believed that the

innervation of the anal canal should be the same as that of the rectum. Secondly, they did not conduct functional studies before surgery and did not compare the treatment outcomes of patients treated with PSARP with anal canal-preserving surgeries. Therefore, they had no scientific basis for not recommending the use of an ectopic anal canal for ARM reconstruction. In different studies, a total an aganglionic segment ranged from 70% [13] to 100% [14]. Fifty-six years have passed since the publication of Duhamel's article (1969) [4] and again (2025) in the article by Alexander et al, 86 pathological reports of anorectal fistulas were assessed for the presence of ganglion cells. Ganglion cells were absent in 5 (6%) of the 86 specimens. The conclusion states that: - "The majority of fistula specimens from patients with ARMs contain ganglion cells. Absence of ganglion cells in patients with ARM should prompt suspicion for HD (Hirschsprung disease)" [15].

To understand how the authors could have found ganglion cells in 94% of cases if, in all the articles in the terminal intestine agangliosis was observed in most cases, we turn to the research methodology. What scientific contribution did 12 authors from 10 different institutions make if all histological samples were collected at a single institution? Is it permissible for surgeons to retrospectively evaluate histological studies they did not perform? Paradoxically, these pediatric surgeons perform PSARP for all types of ARMs. This means that they do not acknowledge the presence of an anal canal in children with ARMs and are unaware of its characteristics. During this procedure, not only the anal canal but also a significant portion of the rectum is removed **(Figure 2).** Since there is no visual boundary between the rectum and the anal canal, and the rectal pouch is considered equivalent to the rectum, it becomes clear that ganglion cells were found in the rectum, not the IAS. This article, carried out with numerous methodological flaws, contradicts the conclusion of the study by Midrio et al, where ganglia were searched in 15/40 cases (37.5%) and resulted absent in 10/15 (66.5%). All patients have been followed and none developed signs or symptoms suggestive for Hirschsprung. They concluded that the practice of searching for ganglia in the terminal rectum/fistula in patients with ARM should be abandoned, as the incidence of associated diseases is rare [16].

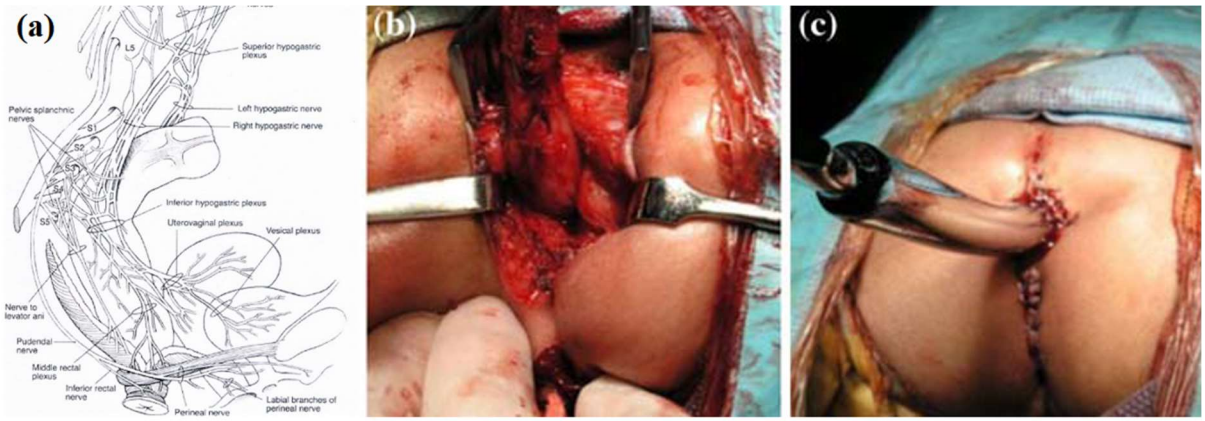

Figure 2. Results of PSARP. **(a)** Pelvic floor innervation from Bharucha's article [4]. **(b)** Extensive pelvic dissection with exposure of the IAS and rectum with dissection of invisible nerve endings and feeding vessels. **(c)** The photograph of the final stage of PSARP shows that a significant portion of the rectum has been exposed along with the IAS. Differentiation between them is impossible.

Radiological examination of the anorectum in ARMs

The third group of authors do not recommend using a fistula (rectal pouch) for ARMs reconstruction is that minor displacement of the anal orifice is associated with deeper anatomical aberrations in the form of anterior misplacement of the anorectum [17,18]. The results of radiographic examination of patients with ARMs provide insight into the pathophysiology of the anomaly (**Figure 3**).

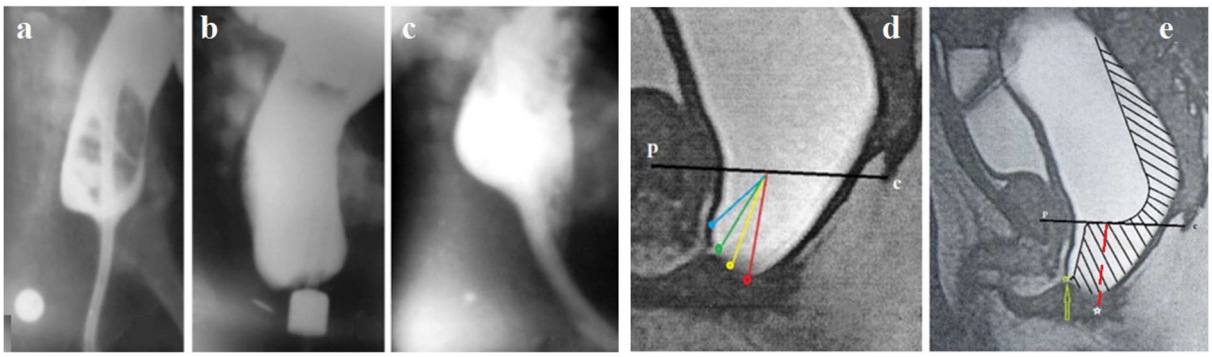

Figure 2. X-ray functional studies in patients with ARMs. **(a)** Lateral radiograph of a patient with vestibular ectopic of the anus. The anal canal has closed around the catheter through which barium was injected into the rectum. A pin is glued to the anal dimple. **(b)** During a repeat hospitalization, the same patient experienced anal canal disclosing during a contrast enema while attempting to defecate. Since the diameter of the contrast marker located near the fistula

opening is 1.6 cm, the distance between the button near the anal dimple and the anal canal wall is 4 mm. This gap, in addition to the skin and subcutaneous tissue, contains the subcutaneous portion of the EAS. **(c)** During the introduction of barium into the rectum through a endotracheal tube with an inflatable balloon connected to a manometer, the barium penetrated the upper part of the anal canal anterior to the tube, while the posterior wall of the anal canal was pressed against the tube by a contracted PRM. A decrease in basal pressure was recorded during this time. After a few seconds, the barium returned to the rectum, which was accompanied by a rise in pressure to the basal level. Thus, the radiological equivalent of the inhibitory anorectal reflex was identified [2, 19]. **(d)** MRI imaging during augmented-pressure distal colostogram with rectobulbar fistula (green). Different colors indicate possible ectopy types. **(e)** Because the IAS is fixed to the site of displacement, when closed, it is entirely displaced anteriorly from its usual location (red dotted line). The same anterior displacement of the anal canal with a catheter inside is seen in Figure 2a.

Analysis of typical X-ray functional studies demonstrates that, in patients with ARMs with visible fistulas, a normally functioning anal canal is present, which at rest contracts, preventing fecal incontinence, and during defecation it opens to allow the passage of feces. Periodic relaxation of the IAS and contraction of the EAS and PRM (rectoanal inhibitory reflux) allow to determine the quality of stool in the rectum. Despite the displacement of the anus relative to the anal dimple, as well as the displacement of the attachment of the IAS relative to other anal canal muscles at rest, during defecation, the IAS is surrounded by the pelvic floor muscles как в норме. In patients with anal ectopia, fecal continence and defecation are not impaired, since in the embryological period, anal ectopia occurs after the anal canal has already formed normally, reaching the subcutaneous tissue opposite the anal dimple [20]. What should the pediatric surgeon choose? Preserve the normally functioning anal canal created by nature or resect the IAS, calling it a fistula, pull in its place the denervated and bloodless rectum, cross the PRM, as well as the deep and superficial portions of the EAS, tearing the rectum away from the levator plates, and during dissection, severing the invisible nerve pathways that connect all reflexes of the urinary and genital tracts and the anorectum? Thus, the authors' (Group 3) assertion that the displacement of the IAS at rest relative to other anal canal muscles is a contraindication to preserving the so-called "rectal pouch" contradicts scientific data. **Manometric study.** I performed the study on 10 patients with visible fistulas who successfully inserted an endotracheal tube with a latex balloon through a narrow ectopic opening. Each

patient's basal pressure was within normal limits, and a positive anorectal reflex was observed [21,22]. The same results were published by Ruttenstock et al [23].

Comparison of PSARP outcomes with anal canal-preserving procedures.

I compared the long-term results of treatment of perineal fistula with the cutback procedure with results after PSARP, using the same assessment method that was used before 1982. Ratings were deemed as "good" when normal fecal retention and absence of constipation were achieved, "fair" when patients required laxatives or enemas, and "poor" when fecal incontinence and/or uncontrollable constipation occurred (Table 1) [12]. Authors 1-4 used the cutback procedure, and authors A-D used PSARP.

Authors	Good (%)	Fair (%)	Poor (%)
1. Nixon [24]	98	0	2
2. Ackroyd et al. [25]	85	15	0
3. Kyrklund et al. [26]	90	8	2
4. de la Fuente [27]	90	?	?
A) Schmiedeke et al [28]			≈ 60
B) Lombardi et al. [29]			≈ 61.4
C) Stenström et al. [30]			≈100
D) Abo-Halawa et al. [31]			?

Comparing the results shows a huge advantage of the cutback procedure compared to PSARP. In the study by den Hollander et al, patients have treated minor types of ARMs (i.e., anal stenosis, anterior anus, and recto-perineal fistula) consistently with non-surgical treatment. The authors identified that patients with minor forms of ARMs possess all the anorectal mechanisms present in healthy subjects. This study also demonstrates that patients with minor ARMs who undergo non-surgical treatment can achieve function outcomes significantly better than after surgery [32].

Discussion

Gone are the days when authors of articles sought to prove that PSARP was the ideal procedure for all types of ARMs. Recently, the ARM-Net Consortium Consensus concluded that: "According to present knowledge, the 'fistula' in ARM represents an ectopic anal canal and should be preserved as far as possible to improve the chance of fecal continence" [32]. The above evidence suggests that: 1) the absence of nerve ganglia in the terminal bowel confirms the presence of an IAS; 2) anterior displacement of the IAS during the fecal retention stage is evidence of IAS anterior ectopy, and not the absence of the anal canal; 3) X-ray examination reveals contraction of the anal canal around the catheter, indicating effective fecal continence.

The assertion that this portion must be resected so that the rectum after denervation and vascular ligation can be brought down into IAS place is unjustified. Moreover, the rectum performs the function of accumulation of feces, and not retention; 4) the presence of normal basal pressure and a positive anorectal inhibitory reflex confirms the presence of the IAS, i.e., a functioning anal canal; 5) Penetration of the contrast agent into the upper part of the anal canal in front of the enema tip indicates the normal function of the PRM, without which normal continence of feces is impossible [1]; 6) Wide opening of the anal canal during defecation is evidence of the normal function of the levator plates, which open the anal canal to reduce resistance to the passage of feces; 7) Functional results after preservation of the anal canal are significantly superior to the results after PSARP because PSARP destroys everything that functioned normally before it.

The ARM-Net Consortium Consensus, while stating that anal ectopia is present in ARMs, does not limit its presence to visible fistulas, as was previously believed following Stephens's studies [34]. As some studies have shown, IAS was detected in all experimental animals [5] and in patients with intermediate and high types of ARMs [7, 8]. Compelling evidence for the presence of a functioning anal canal in the so-called high types is presented in the article by Kraus, co-authored by Levitt and Peña [35]. In the article, on augmented-pressure distal colostogram in boys, state: - "… it is extremely important in this regard to understand that the lowest part of the rectum is usually collapsed from the muscle tone of the funnel-like striated muscle mechanism that surrounds the rectum in 90% of cases…" [35]. Meanwhile, it is known from anatomy that there are no muscles around the rectum that are compressing it. The terminal section of the intestine, which is surrounded by muscles, the tone of which overlaps the intestinal lumen and does not allow contrast agent to pass through, cannot be the rectum. The characteristics of this section correspond to the idea of the anal canal, the contraction of which performs the function of the fecal retention. The article provides radiographs showing that with low pressure in the rectum, the distal intestine is in a closed state. On radiographs with high-pressure in the rectum, this region opens widely, with the distal contour approximately 1 cm from the anal dimple [35]. This is the normal functioning of the anal canal, which is destroyed in PSARP only because it is mistakenly referred to as a rectum or fistula.

Recently, a trend has emerged where authors of articles recently promoting PSARP as the ideal treatment for ARMs have begun to look for other options. In the article by Halleran et al., for the first time in many years, Levitt proposed an operation that has an advantage over PSARP. "The main technical advantage of the PRAA is that it obviates the need for any anterior rectal

dissection, thereby mitigating the risk of urethral injury in males or vaginal injury in females" [36, 37, 38]. The authors believe that after PRAA “there is virtually no distal rectum that is discarded, thus the inherent value of the distal rectum (the internal sphincter present within the anorectal wall) is preserved”. It is surprising that pediatric surgeons operate on children with ARM without knowing that the rectum and the IAC are different anatomical regions. It follows that the new operation (PRAA) has no scientific basis. The new trend is also noticeable on the European continent [39]. In the article Mašić et al., note that "Despite good outcomes, PSARP risks sphincter transection, perineal body dehiscence, and stenosis." Therefore, they proposed sphincter-preserving anorectoplasty from the anterior approach [38]. A study by den Hollander et al. showed that patients with minor types of ARMs can achieve optimal anorectal function after non-surgical treatment. In addition, they demonstrated that patients who did not undergo surgery have the same fecal continence mechanisms as healthy individuals [32]. A detailed analysis of trends in the treatment of APM is given in my article [40].

This review shows that pediatric surgeons operating on children with ARMs are ignorant of the anatomy and physiology of the pelvic floor and do not cite scientific physiologists and practicing gastroenterologists, as if the anatomy and physiology of the pelvic floor in childhood are different from those of adulthood. Pediatric surgeons do not cite scientists from previous generations, as if pediatric colorectal surgery was created by Alberto Peña. I analyzed Peña's articles on ARMs [12], functional constipation [41], and persistent cloaca [42] and did not find a single scientific study or reference to scientific evidence. In numerous articles, he declared his experience, which had no scientific basis. In fact, he was talking about experiments on children with ARMs. He surrounded himself with pediatric surgeons who do not value scientific evidence. During this period, beginning in 1982 (43 years), several generations of pediatric surgeons grew up with misconceptions about colorectal surgery. Peña's entire practice was a successful business, which, albeit in a truncated form, continues to this day.

The following advertisement no hints about the need to preserve the anal canal in children with ARMs [43]. "Dear Colleague,

This is your last chance to secure the best price for the 70th Workshop on the Surgical Treatment of Anorectal Malformations and Hirschsprung Disease in Children. Early bird registration ends December 31, 2025, and prices will increase on January 1, 2026. If you plan to attend, we encourage you to register soon to take advantage of the lowest available rate.

The workshop will take place April 27–30, 2026, at Children's Hospital Colorado" (Alberto Peña, Andrea Bischoff and Luis De La Torre).

Conclusion

Fifty-six years have passed since the renowned surgeon Duhamel published a study proving that in the anal canal healthy people has no ganglia. This fact has been confirmed by other researchers, making it a scientific indisputable fact. Currently, a group of pediatric surgeons who are members of the Pediatric Colorectal and Pelvic Learning Consortium periodically publish their recommendations. The last article described a retrospective analysis of histological specimens resected after PSARP. They did not subdivide the histological specimens into anal canal and rectum, as a priori, based on Peña's opinion, do not recognize the presence of the anal canal in ARMs. They falsely concluded that most fistula specimens from patients with ARMs contain ganglion cells, because 96% of cases were examined in the rectum specimens. They considered the absence of ganglion cells in patients with ARMs to be a suspicion of Hirschsprung disease, ignoring scientific research. They did not conduct manometric or radiographic studies, making their study devoid of scientific status. All articles by The Pediatric Colorectal and Pelvic Learning Consortium promote PSARP and the treatment of severe complications from this destructive procedure. The reaction of reviewers, journal editors, and the American Pediatric Surgical Association is astonishing, as they allow these surgeons to publish non-scientific texts and operate on children with a method that leads to lifelong disability. There is an urgent need to organize a multidisciplinary review of the state of pediatric colorectal surgery to protect patients.

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