DOI: 10.34057/PPj.2022.41.02.2021-9-1
Pelviperineology 2023;42(2):74-79

Pathological physiology of the anorectal malformations without visible fistula. A short review

Michael LEVIN

Department of Pediatric Radiology of the 1st State Hospital, Minsk, Belarus

Citation: Levin M. Pathological physiology of the anorectal malformations without visible fistula. A short review. Pelviperineology 2023;42(2):74-79

ABSTRACT

Until 1982, pediatric surgeons came to a consensus that in patients with anorectal malformations (ARM), the intestine that is located caudal to the pubococcygeal line is the anal canal and, to achieve the best functional result, it must be preserved during surgery. Simultaneously with the publication of posterior sagittal anorectoplasty, it was stated that except for patients with rectal atresia and anal stenosis, patients with ARM are born without an anal canal. It is believed that the rectal pouch or fistula should be removed. We analyzed 41 articles, including 2 of our own studies, which reflect the entire palette of ideas about the pathological anatomy and physiology of ARM without a visible fistula (females and males without fistula and males with urethral fistula). On histological, manometric and radiological examinations, in most patients, the intestine located caudal to the PRM has the characteristics of a functioning anal canal. This literature review proves that most ARM patients without a visible fistula have a functioning anal canal, the preservation of which can ensure normal anorectal function.

Keywords: Anorectal malformations; imperforate anus; ARM without fistula; ARM with urethral fistula; physiology, anorectal malformations; pathophysiology; rectourethral fistula; without fistula; imperforate anus; anal ectopy, pelvic floor

INTRODUCTION

In 1953, Stephens[1] proposed the concept of a pubococcygeal (P-C) line, which runs from the lower limit of the pubic bone to the distal coccygeal vertebra. He showed that this line corresponds to the location of the puborectalis muscle (PRM), which plays a large role in stool retention. If the blind end of the intestine is located above this line, these cases are considered a high type of anorectal malformations (ARM), and if more caudally of this line it is a low type.[1] This understanding of the pathological physiology of ARM was reflected in the Wingspread classification (1984). Since then, it was believed that if the gut is located below the P-C line, it means the patient has an anal canal that needs to be preserved during surgery.[2]

For many years, for diagnosis of the level of ARM used an invertogram. Recently, this method has not been used since the overall sensitivity of invertograms in detecting low anomalies was 33.3%, whereas specificity was 66.7%.[3-5] The low reliability of this method is due to erroneous ideas about the physiology of the anorectal zone. It was assumed that gas in the rectum rises and is retained in the blindly ending gut. However, firstly,

Address for Correspondence: Michael Levin, Department of Pediatric Radiology of the 1st State Hospital, Minsk, Belarus
E-mail: nivel70@hotmail.com **ORCID ID:** orcid.org/0000-0001-7830-1944
Received: 03 July 2022 **Accepted:** 27 October 2022

the contents of the intestine move only by a peristaltic wave. Secondly, the gas cannot pass into the closed anal canal if the rectal pressure is less than the threshold level. On radiographs in newborns, it is often difficult to determine bone landmarks for drawing the P-C line. Cremin et al.[6] showed that this line runs between the caudal and middle third of the pear-shaped ischium.

In 2005, the Krickenbeck classification was adopted, which is a listing of the main types of ARM without a division into high and low types.[2] The main idea, proposed by Peña and accepted by the community of pediatric surgeons, is that in ARMs the anal canal is absent, and the rectal pouch or fistula is so different from the rectum that it cannot be used for defect correction.[5] Neither Peña's articles nor other scientific sources provide conclusive evidence for this claim. Moreover, it contradicts all research from previous generations. Since it had been considered that all patients without a visible fistula a priori do not have an anal canal, the studies to determine the level of anomaly have lost all meaning.

The purpose of this review is to study materials on the pathological anatomy and physiology of ARM without visible fistula.

MATERIALS AND METHODS

We analyzed 41 articles, including 2 of our own studies, which reflect the entire palette of ideas about the pathological anatomy and physiology of ARM without a visible fistula (females and males without fistula and males with urethral fistula).

Histological Studies in ARM

In a study by Holschneider et al.[7] it was shown that in patients with ARM "Classical aganglionosis was found in 31% of the rectal pouch specimens, hypoganglionosis in 38%, neuronal intestinal dysplasia (NID) type B in 14%, and dysganglionosis in 10%". In the authors' opinion, "... the recommendation to use the distal rectal pouch and parts of the fistula in the reconstruction of ARMs malformations should be reconsidered".[7] These histological results were confirmed by other researchers, who believed that the histological structure of the anal canal should be consistent with the structure of the rectum.[8,9]

Alamovich et al. (citation from Duhamel[10]) investigated the innervation of the normal IAS. This study shows that the IAS itself has no autonomous innervation unlike the rest of the digestive tube. Lambrecht and Lierse[11] in 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. The most important result was the demonstration of a normal internal sphincter even in high and intermediate types of ARM.[11] A study by Uemura et al.[12] showed that "Epithelial and ganglionic distribution was similar in the distal rectal end of ARMs and in a normal anal canal". The mistake of pediatric surgeons is that when describing the normal innervation of the anal canal, they compared it with the innervation of the rectum and concluded that this is not a normal rectum. They did not know that the anal canal had a different histological structure from the rectum. Histological studies show that in most patients with ARM, the distal intestine, which is still called a fistula or rectal sac, has the histological structure of a normal anal canal.

Manometric Study

In 5 infants with ARMs (high type 2, intermediate type 3), a preoperative manometric study at the rectal end was performed with a probe introduced from the distal colostomy. This study showed the presence of rhythmic activity in all, and positive reflexive pressure fall by rectal distension in 4.[13] The presence of a rectoanal inhibitory reflex is a characteristic of the anal canal. Preoperative rectal manometry of rectoperineal or rectovestibular fistula showed the presence of functional anal structures within the fistula in all patients.[14]

X-ray Examinations

In a newborn's first hours of life, the rectal pressure is below a threshold level. Therefore, the anal canal is in a closed state, and meconium with gas is in the rectum. Only after 30 hours of birth does the rectum collect enough gas and meconium to create pressure that opens the anal canal. An article by Levitt and Peña[5] suggests doing a crosstable lateral radiograph after 16-24 hours after birth. However, they claim that this study can help show the air column in the distal rectum in the small percentage of patients.[5] Hosokawa et al.[15] on the sonograms found, that the pouch-perineum distance on the next day was significantly shorter than on the birthday (p=0.001). Such a significant shortening can only be explained by the fact that in some newborns, additional content entered the rectum that led to an increase in rectal pressure and the opening of the anal canal. This is the reason of increase in the average the pouch-perineum distance. In Figure 1 can be seen the importance of research time.

The reflex opening of the anal canal takes several seconds. Then, the rectum, adapting to the increased volume of contents, relaxes, which leads to a drop in rectal pressure. This causes a reflex contraction of the anal canal and the displacement of gas from the anal canal into the rectum. In the process of increasing the volume of rectal contents, this situation is repeated several times.[14]

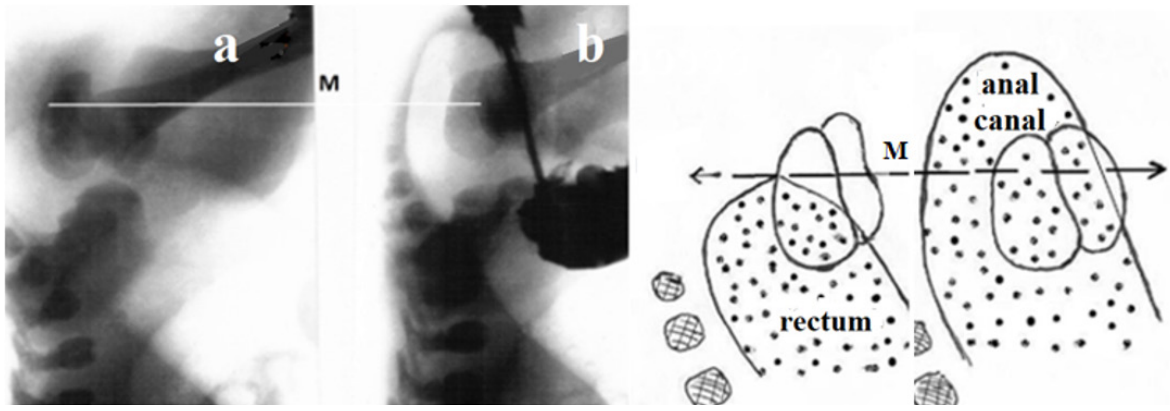

Figure 1. Radiographs of a newborn with ARM without a visible fistula. (a) Invertogram took 12 hours after birth. The distal contour of the rectum is located on a horizontal line (M) between the middle and distal third ischium, which has a typical pear shape. According to Cremin et al.[6] data, this line corresponds to the pubococcygeal line (see scheme). (b) Thirty hours after birth the erroneous introduction of contrast medium into the perineal tissue (instead of to the rectum) was produced. The anal canal opened, and gas is visible close to the perineal skin. The rectal width is noticeably larger than in Figure 1a. Line "M" was inscribed because there are no other bony landmarks on radiographs. Thus, 12 hours after birth, the X-ray picture corresponded to the intermediate type of ARM (this is the normal position of the rectum over the contracted anal canal), and after 30 hours during the tension of the abdomen, as a reaction to pain, the anal canal opened, which indicates a low type

Therefore, X-ray even 30 hours after birth does not guarantee that at the time of the radiograph the opening of the anal canal will be recorded. The threshold pressure at which the anal canal opens depends on the volume of meconium and gas, as well as on intra-abdominal pressure.[16] After 30 hours there is a theoretical risk of intestinal perforation and/or vomiting with aspiration hazard. Limiting the time of the study, limits the increase in rectal volume. The abdominal compression increases rectal pressure and causes the anal canal to open at the time of fluoroscopy (Figure 2).[17]

The approach of gas to the perineum in the restless newborn is a known phenomenon. In the literature, it is mistakenly explained with the descending perineum during an increase in intra-abdominal pressure. It is considered a mistake to assess the level of ARM by the descending perineum since in a calm state the perineum returns to its place.[5,18] As is known from physiology, during an increase in intra-abdominal pressure, the muscles of the pelvic floor do not descend but rise.[19] Some authors call this phenomenon a "well-descended rectum",[18] ignoring the fact that the rectum is fixed in the tissues of the pelvis and cannot move. In the article by Nagdeve et al.[18], of 12 male neonates with high ARM who on invertogram showed well descended rectum, with lower limit of rectal gas bubble at or below the ossified fifth sacral vertebra the fistula with urinary tract was found in 11 patients (seven had fistula to bulbar urethra and four to prostatic urethra). It is believed that the exact level of ARM is determined at the time of surgery. However, during surgery, the rectal pressure decreases, so the anal canal closes. A closed IAS both in norm and low ARM is a canal that looks like a fistula. Koga

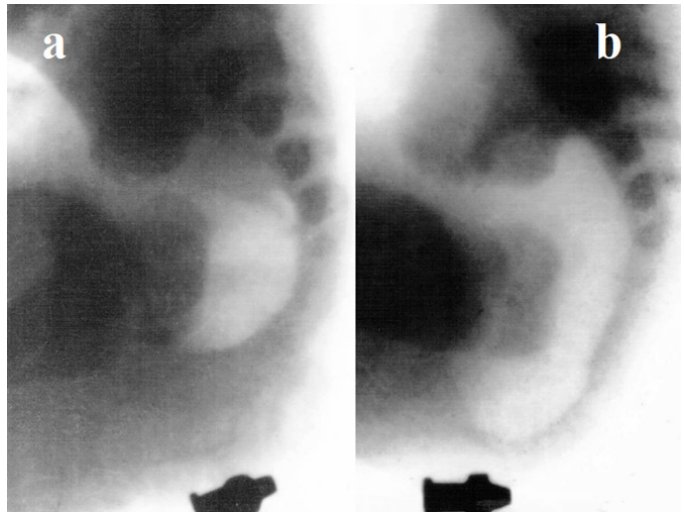

Figure 2. Radiographs of a newborn with ARM without fistula were performed horizontally. A radiopaque marker was glued to the anal dimple. (a) At rest, (b) During abdominal compression, the gas approached the marker. The distance between the marker and the intestine is the thickness of the skin and subcutaneous tissue

et al.[20] proposed a method for measuring the urethral fistula during surgery to remove it without residue. In fact, under the guise of a fistula, they removed IAS. However, "the internal anal sphincter is currently regarded as a significant contributor to continence function".[21] Thus, what is commonly called the well-descended rectum in the literature is the anal canal. This picture is no different from an open anal canal in healthy infants.[16] It follows that patient with bulbar and prostatic urethral fistulas, as well as in patients without fistula have an anal canal.

Hence these cases are the low type of ARM. The presence of the anal canal is especially evident in visible fistulas, which differ from the invisible by less displacement of the anus (Figure 3).

The inability to open the anal canal 30 hours after birth during abdominal compression indicates the presence of a high type of ARM.

Augmented-pressure Distal Colostogram

Kraus et al.[22] 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 (ARM without visible fistulas) is usually collapsed from the muscle tone of the funnel-like striated muscle mechanism that surrounds the rectum in 90% of cases …". Meanwhile, it is known from anatomy that there are no muscles around the rectum. Muscles surround the anal canal, participating in fecal retention and defecation. The authors, describing the normal function of the anal canal, call it the rectum. In fact, this statement suggests that at least 90%

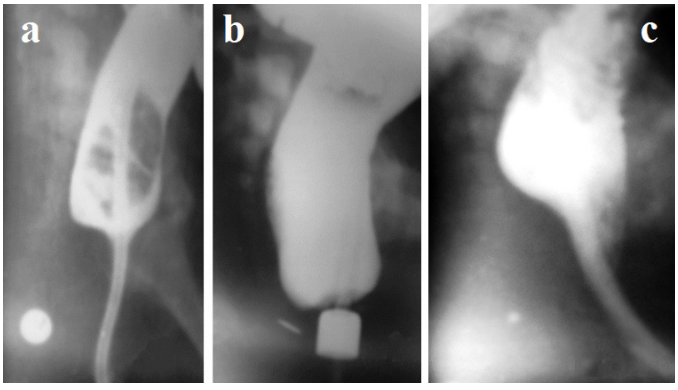

Figure 3. The radiographs of the same girl with vestibular fistula performed at different ages. (a) At the age of 3 months, the rectum was filled with barium through the catheter, conducted through the fistula. A pushpin is located near the anal dimple. The distal intestine, with a length equal to the length of the normal anal canal, constantly contracted around the catheter, preventing leakage of barium. (b) At the age of 9 months, during a barium enema, the wide opening of the anal canal occurred. The distance from the pushpin to the distal wall of the open anal canal equals 4 mm. Barium does not penetrate outward, since the tip of the enema occluded the narrow and rigid ectopic anus. The true diameter of the marker on the enema tip is 1.6 cm. The width of the rectum is 4.3 cm (the maximum rate for children 1-3 years is 3.7 cm). Conclusion: Ano-vestibular ectopy, megarectum. The diastasis between the anal canal and anal dimple is (4 mm), which corresponds to the thickness of the skin and subcutaneous tissue. (c) Barium was injected into the rectum through an intubation tube (no: 8), passed through the vestibular fistula. The penetration of barium into the upper part of the anal canal in front of the intubation tube is determined. The posterior wall of the anal canal is pressed against the tube of the contracted PRM. The contraction of the PRM and EAS provide normal liquid barium retention. This is a typical X-ray picture of a rectoanal inhibitory reflex

of boys without a visible fistula have a functioning anal canal. The augmented-pressure distal colostogram is characterized by high uncontrolled hydrodynamic pressure, which significantly exceeds the threshold pressure of the anal canal opening during defecation. Unlike reflex opening, which typically lasts less than a minute, this pressure results in the mechanical, permanent opening of the anal canal. A serious disadvantage of this method is the danger of perforation of the sigmoid colon. Therefore, most authors use a distal colostogram without high pressure.[23-26] However, only the augmented-pressure distal colostogram, with X-ray examination or with the use of CT or MRI, shows the presence of an anal canal (Figure 4).

Comparison of Treatment Outcomes for Low-type ARM

Preserving the anal canal for "ectopic anus by the simple cut-back is all that is needed to make the imperfect anus large enough to work where it lies".[27-33] For example, "All males treated for low ARMs outcomes by bowel function scores were good at 85% and satisfactory in 15%".[27] After PSARP, during which the IAS is excised, a large proportion of the patients have persistent fecal incontinence, constipation, and sexual problem.[34-41]

DISCUSSION

Following a study by Stephens[1], who proved that if in newborns with ARM gas penetrates below the P-C line, this indicates the presence of the anal canal, which must be preserved to obtain good functional results, it was recorded in the Wingspread classification (1984).[2] After 1982, Peña's articles claimed the absence of the anal canal in patients with ARM, but this statement

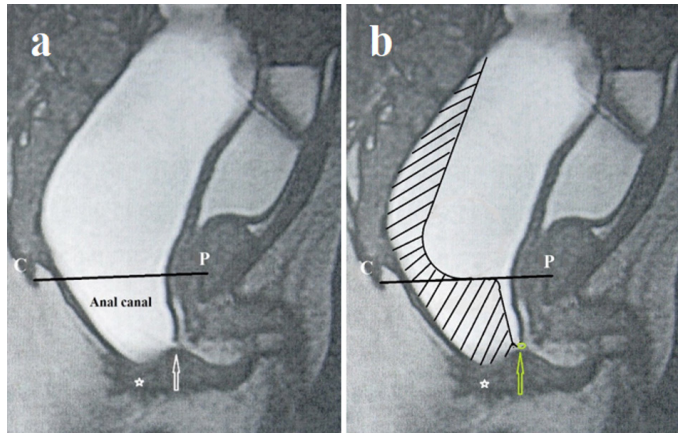

Figure 4. (a) MRI imaging during augmented-pressure distal colostogram in a male with recto-bulbar fistula (arrow). Distal to the pubococcygeal line, a wide-open anal canal is visible. Its blind end is located ≈2-4 mm from the proposed site of the anal fossa (asterisk). (b) MRI reconstruction scheme with low rectal pressure. The anal canal is closed. Conclusion: Ectopia of the anal canal into the bulbar part of the urethra

was unfounded, since it was not the result of any research. Since then, the intestine located caudal to the P-C line has been called a fistula or rectal pouch. During pull-through operations (posterior sagittal approach, anterior sagittal approach or by laparoscopy) this "fistula" is removed, as it is believed that its function is impaired. However, the authors compared histological studies indicating the presence of a normal anal canal with the innervation of the rectum. The conclusions of these authors were erroneous, since the innervation of the rectum and anal canal is different. Histological studies of the so-called fistula completely coincide with the structure of the normal anal canal.[10-12]

The idea of a functional inferiority of the "rectal pouch" was based solely on the erroneous conclusions of histological studies. Meanwhile, manometric studies indicate the presence of a normally functioning anal canal with normal basal anal pressure and a positive rectoanal inhibitory reflex.[13,14,17]

X-ray studies confirm that the so-called fistula functions like a normal anal canal. At a rectal pressure below the threshold, it is in a closed state, i.e., it performs the function of the fecal retention. With an increase in rectal pressure above the threshold level, it opens, i.e., an attempt to defecate. The presence of the anal canal is especially evident in visible fistulas, which differ from the invisible by less the anus displacement. Levitt and Peña[5] refer to the anal canal as a fistula, which is in a contracted state of the surrounding muscles. This is a description of a normal anal canal. The point of this unreasonable name change is to justify the removal of the "fistula" in posterior sagittal anorectoplasty (PSARP). Second, to justify poor long-term results. They are supposedly the maximum possible since children with ARM do not have an anal canal from birth.

However, comparing the results of a PSARP treatment in which an anal canal is destroyed, with a cutback operation in which the anal canal is completely preserved, one can see the enormous advantage of cutback operation.

CONCLUSION

Histological, manometric, and radiological studies show that most patients with ARM without visible fistulas have an anal canal. This means they have an IAS, sensitive to rectal pressure and which is located caudally to a PRM. In response to pressure in the rectum, there is a temporary relaxation of the IAS (rectoanal inhibitory reflex) and contraction of the PRM, as well as the deep and superficial parts of the external anal sphincter. Higher rectal pressure, caused by abdominal compression, stimulates the defecation reflex with a wide opening of the anal canal, which makes it possible to diagnose a low type of ARM.

ETHICS

Peer-review: Internally and externally peer-reviewed.

DISCLOSURES

Financial Disclosure: The author declared that this study received no financial support.

REFERENCES

1. Stephens FD. Imperforate rectum. A new surgical technique. Med J Aust 1953; 1: 202-3.
2. Holschneider A, Hutson J, Peña A, et al. Preliminary report on the International Conference for the Development of Standards for the Treatment of Anorectal Malformations. J Pediatr Surg 2005; 40: 1521-6.
3. Carroll AG, Kavanagh RG, Ni Leidhin C, Cullinan NM, Lavelle LP, Malone DE. Comparative Effectiveness of Imaging Modalities for the Diagnosis of Intestinal Obstruction in Neonates and Infants: A Critically Appraised Topic. Acad Radiol 2016; 23: 559-68.
4. Horsirimanont S, Sangkhathat S, Utamakul P, Chetphaopan J, Patrapinyokul S. An appraisal of invertograms and distal colostograms in the management of anorectal malformations. J Med Assoc Thai 2004; 87: 497-502.
5. Levitt MA, Peña A. Anorectal malformations. Orphanet J Rare Dis 2007; 2: 33.
6. Cremin RJ, Cywes S, Louw JH. A rational radiological approach to the surgical correction of anorectal anomalies. Surgery 1972; 71: 801-6.
7. Holschneider AM, Ure BM, Pfrommer W, Meier-Ruge W. Innervation patterns of the rectal pouch and fistula in anorectal malformations: a preliminary report. J Pediatr Surg 1996; 31: 357-62.
8. Gangopadhyay AN, Upadhyaya VD, Gupta DK, Agarwal DK, Sharma SP, Arya NC. Histology of the terminal end of the distal rectal pouch and fistula region in anorectal malformations. Asian J Surg 2008; 31: 211-5.
9. Xiao H, Huang R, Cui DX, Xiao P, Diao M, Li L. Histopathologic and immunohistochemical findings in congenital anorectal malformations. Medicine (Baltimore) 2018; 97: e11675.
10. Duhamel B. Physio-pathology of the internal anal sphincter. Arch Dis Child 1969; 44: 377-81.
11. Lambrecht W, Lierse W. The internal sphincter in anorectal malformations: morphologic investigations in neonatal pigs. J Pediatr Surg 1987; 22: 1160-8.
12. Uemura K, Fukuzawa H, Morita K, Okata Y, Yoshida M, Maeda K. Epithelial and ganglionic distribution at the distal rectal end in anorectal malformations: could it play a role in anastomotic adaptation? Pediatr Surg Int 2021; 37: 281-6.
13. Ohama K, Asano S, Nanbu K, Kajimoto T. The internal anal sphincter in anorectal malformation. Z Kinderchir 1990; 45: 167-77.

14. Ruttenstock EM, Zani A, Huber-Zeyringer A, Höllwarth ME. Pre- and postoperative rectal manometric assessment of patients with anorectal malformations: should we preserve the fistula? Dis Colon Rectum 2013; 56: 499-504.

15. Hosokawa T, Yamada Y, Sato Y, et al. Changes in the Distance Between the Distal Rectal Pouch and Perineum From the Birth Day to the Next Day in Neonates With an Imperforate Anus. J Ultrasound Med 2017; 36: 601-6.

16. Levin MD. Anatomy and physiology of anorectum: the hypothesis of fecal retention, and defecation. Pelviperineology 2021; 40: 50-7.

17. Levin MD. [The pathological physiology of the anorectal defects, from the new concept to the new treatment]. Eksp Klin Gastroenterol 2013: 38-48.

18. Nagdeve NG, Bhingare PD, Naik HR. Neonatal posterior sagittal anorectoplasty for a subset of males with high anorectal malformations. J Indian Assoc Pediatr Surg 2011; 16: 126-8.

19. Bharucha AE. Pelvic floor: anatomy and function. Neurogastroenterol Motil 2006; 18: 507-19.

20. Koga H, Kato Y, Shimotakahara A, et al. Intraoperative measurement of rectourethral fistula: prevention of incomplete excision in male patients with high-/intermediate-type imperforate anus. J Pediatr Surg 2010; 45: 397-400.

21. Zbar AP, Khaikin M. Should we care about the internal anal sphincter? Dis Colon Rectum 2012; 55: 105-8.

22. Kraus SJ, Levitt MA, Peña A. Augmented-pressure distal colostogram: the most important diagnostic tool for planning definitive surgical repair of anorectal malformations in boys. Pediatr Radiol 2018; 48: 258-69.

23. Madhusmita, Ghasi RG, Mittal MK, Bagga D. Anorectal malformations: Role of MRI in preoperative evaluation. Indian J Radiol Imaging 2018; 28: 187-94.

24. Zhan Y, Wang J, Guo WL. Comparative effectiveness of imaging modalities for preoperative assessment of anorectal malformation in the pediatric population. J Pediatr Surg 2019; 54: 2550-3.

25. Midrio P, van Rooij IALM, Brisighelli G, et al. Inter- and Intraobserver Variation in the Assessment of Paola Preoperative Colostograms in Male Anorectal Malformations: An ARM-Net Consortium Survey. Front Pediatr 2020; 8: 571.

26. Tang ST, Cao GQ, Mao YZ, et al. Clinical value of pelvic 3-dimensional magnetic resonance image reconstruction in anorectal malformations. J Pediatr Surg 2009; 44: 2369-74.

27. Kyrklund K, Pakarinen MP, Taskinen S, Rintala RJ. Bowel function and lower urinary tract symptoms in males with low anorectal malformations: an update of controlled, long-term outcomes. Int J Colorectal Dis 2015; 30: 221-8.

28. Nixon HH. Anorectal anomalies: with an international proposed classification. Postgrad Med J 1972; 48: 465-70.

29. Wilkinson AW. Congenital anomalies of the anus and rectum. Arch Dis Child 1972; 47: 960-9.

30. Scott JE. The microscopic anatomy of the terminal intestinal canal in ectopic vulval anus. J Pediatr Surg 1966; 1: 441-5.

31. Swain VA, Tucker SM. The results of operation in 46 cases of malformation of the anus and rectum. Gut 1962; 3: 245-51.

32. Nixon HH, Puri P. The results of treatment of anorectal anomalies: a thirteen to twenty year follow-up. J Pediatr Surg 1977; 12: 27-37.

33. de la Fuente AQ, Arance MG, Cortés L. [Low ano-rectal malformations (author's transl)]. An Esp Pediatr 1979; 12: 603-6.

34. Danielson J, Karlbom U, Graf W, Wester T. Outcome in adults with anorectal malformations in relation to modern classification - Which patients do we need to follow beyond childhood? J Pediatr Surg 2017; 52: 463-8.

35. Bukarica S, Marinković S, Peković-Zrnić V, Dobanovacki D, Borisev V, Likić J. [Clinical evaluation of fecal continence after posterior sagittal anorectoplasty in anorectal abnormalities]. Med Pregl 2004; 57: 284-8.

36. Stenström P, Kockum CC, Emblem R, Arnbjörnsson E, Bjørnland K. Bowel symptoms in children with anorectal malformation - a follow-up with a gender and age perspective. J Pediatr Surg 2014; 49: 1122-30.

37. Schmiedeke E, Zwink N, Schwarze N, et al. Unexpected results of a nationwide, treatment-independent assessment of fecal incontinence in patients with anorectal anomalies. Pediatr Surg Int 2012; 28: 825-30.

38. Schmidt D, Jenetzky E, Zwink N, Schmiedeke E, Maerzheuser S. Postoperative complications in adults with anorectal malformation: a need for transition. German Network for Congenital Uro-REctal Malformations (CURE-Net). Pediatr Surg Int 2012; 28: 793-5.

39. Grano C, Aminoff D, Lucidi F, Violani C. Long-term disease-specific quality of life in adult anorectal malformation patients. J Pediatr Surg 2011; 46: 691-8.

40. Hashish MS, Dawoud HH, Hirschl RB, et al. Long-term functional outcome and quality of life in patients with high imperforate anus. J Pediatr Surg 2010; 45: 224-30.

41. Lombardi L, Bruder E, Caravaggi F, Del Rossi C, Martucciello G. Abnormalities in "low" anorectal malformations (ARMs) and functional results resecting the distal 3 cm. J Pediatr Surg 2013; 48: 1294-300.