Congenital stenosis of the anus. Pathological anatomy, embryology. Rationale for treatment.

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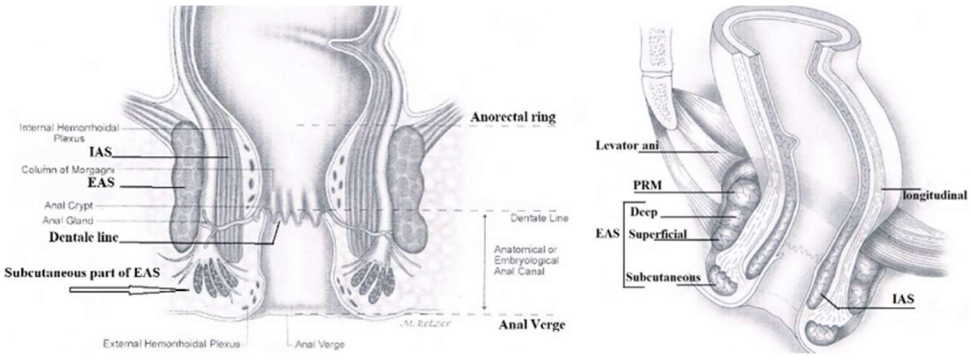
Abstract

All authors recognize the presence of an anal canal in congenital anal stenosis (CAS). However, there are different views on the pathological anatomy and physiology of CAS. Some believe the stenosis occupies a short distance between the anal canal wall and the anus. Others claim a long narrowing extending to the dentate line or the rectum. Material and Methods. We analyzed radiographic studies of CAS from our own experience and from literature sources to determine the pathological anatomy of CAS using radiometric methods. Results. It was shown that all forms, except for true cloaca, develop similarly during the embryological period, with the formation of the anal canal resulting from the craniocaudal advancement of the internal anal sphincter (IAS). The absence of anal opening, as well as its stenosis in its usual location, indicates that the exogenous IAS primordium does not move upward to meet the endogenous one. Penetration of the IAS beyond the anal canal leads to the formation of a narrow, rigid ring. If the IAS penetrates the skin within the subcutaneous portion of the external anal sphincter, a congenital anal stenosis (CAS) is formed. In other cases, the anus is displaced forward and upward, extending externally or into any cavity, creating an ectopic anus on the perineum, vaginal vestibule, urethra, or vagina. Conclusion: Congenital anal stenosis is a form of ARMs characterized by the presence of a normal anal canal, where the stenosis of the normally located anus ranges from 2 to 5 mm in length. Radiographic examination provides an accurate anatomical and physiological characterization of the defect. Treatment should be initiated as early as possible to prevent the development of megacolon. Dissection of the rigid ring with insertion of a tube into the anal canal can lead to complete recovery.

Keywords: anorectal malformations; congenital anal stenosis; pathological anatomy; congeninal stenosis of the anus; x-ray diagnosis; embryology.

Introduction Congenital anal stenosis (CAS) is defined as a rare condition in which the anus is narrow and located in its normal position, surrounded by the sphincter muscle complex. Sometimes a membrane near the dentate line may accompany [1]. The authors diagnosed stenosis based on the passage of a small-diameter bougie. None of the 27 patients had other

accompanying malformations, and no membrane near the dentate line was diagnosed. Members of the Arm-Net Consortium came to a consensus that with CAS, the anus lies in a normal position, completely surrounded by the sphincter muscle complex, but is too narrow. It may be partly covered by a median bar or membrane, usually located at the dentate line [2]. Instead of providing evidence, a reference is given to a review article, which also contains no studies of the pathological anatomy of this type of ARMs. This formulation contains several contradictions. First, the anus is the opening by which the anal canal ends. It cannot be surrounded by a muscular complex. Normally, the anus is surrounded by the subcutaneous portion of the external anal sphincter (EAS), which makes up approximately 1/10 of the anal canal [3] (**Figure 1**).



Second, the remaining 9/10 portion of the terminal intestine that is surrounded by a muscular complex is called the anal canal. Third, in the diagnostic algorithm presented in this article, the diagnosis is based solely on the location of the anus and the maximum caliber of the bougie that freely passes through the anus. The algorithm does not contain recommendations for identifying stenosis at the dentate line level. Fourth, in the discussion of the article it is stated 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 for fecal continence" [2]. Although this assertion is consistent with scientific evidence, it is not analyzed in this article. The references to the articles that supposedly proved this provide no evidence. Subsequent articles (post-2022) by the authors, who were members of the Consortium, contain no one in which they would consider fistulous forms of ARM as ectopic anal canal and preserved it. Saenz et al., on behalf of the Pediatric Colorectal and Pelvic Learning Consortium (PCPLC), discuss two types of anorectal stenosis. "Congenital anal stenosis is defined as an anus that lies within an intact sphincter muscle complex but is pathologically narrow. The narrowing of the anal canal is usually located at the dentate line, and patients often present with a skin-lined, 'funnel anus' and is frequently associated with a sacral anomaly." Congenital rectal stenosis can

be similarly defined as a well-developed, normally positioned anus within an intact sphincter complex but with a pathological narrowing located proximal to the dentate line [4]. There is no evidence for these definitions in the article by these authors, nor in the references they cite. Moreover, these definitions contain contradictory definitions. For example, if in CAS the anus is narrowed, it cannot simultaneously have a skin-lined, 'funnel anus'. In this case, it cannot be stenosis of the anus up to the dentate line. The definition of "congenital rectal stenosis" is not based on studies of the pathological anatomy of this type of ARMs. It has been around for 70-80 years, when pediatric surgeons, relying solely on their clinical experience and the Wangensteen-Rice-Invertogram, often identified anal stenosis with rectal stenosis. For example, Nixon believed that "In about half the cases, the stenosis is a string-like one at the junction of the proctoneum and endodermal lining of the upper part of the anatomical anal canal" [5]. At the same time, Browne believed that anal stenosis “is an obvious condition, although some of its anatomical features are puzzling, especially the frequent occurrence of the thick band of tissue running anterio-posteriorly across the anal site with minute opening on one side of it. Occasionally, this band is completely detached in its centre” [6]. This type of CAS is called "covered anus" in some classifications [7].

Confusion in definition can be seen in articles by pediatric surgeons of previous generations. For example, Wilkinson wrote: "Anorectal stenosis us usually limited to a narrow fibrous ring at any point in the anal canal, but most often near the anorectal junction: the anal canal and rectum are otherwise normal" [7]. Firstly, as we see from the articles cited above, stenosis can be either at the level of the anus or at the level of the dentate line, and not at any point in the anal canal. Secondly, it is most often observed in the anus. What did the author mean by stating that stenosis most often occurs near the anorectal junction if the anal canal is normally developed? Most likely, he meant the junction of the anus with the anal canal, because stenosis at the junction of the anal canal with the rectum has not been described. In Wilkinson's definition, the term "anal" refers to both the anus and the anal canal, and he refers to the anal canal as the rectum. This confusion is often found in modern literature, where, for example, perineal ectopia ani is called a rectal fistula. In an article by Kiely et al., 11 cases of delayed diagnosis of CAS were described. In two of them, the tight stenosis at the dental line, i.e., in the anal canal, was found [8]. As shown by radiographic analysis of 15 published cases of rectal atresia and stenosis, in all observations the membrane was found in the anal canal near the dentate line [9]. An analysis of the literature shows that authors who devote articles to anal stenosis combine two different diseases under this diagnosis: congenital stenosis of the anus, which is a result of a disturbance in the formation of the ectodermal rudiment (proctoneum) of

the IAS, and stenosis resulting from incomplete destruction of the anal membrane during the embryonic period. Normally, according to Nobles (1984), the anal membrane ruptures during embryos with a length of 13.5-135 cm [10]. At the site of membrane destruction, a dentate line is determined. If the anal membrane is not destroyed, not rectal, but anal atresia occurs. If the membrane is partially destroyed, not rectal, but anal stenosis occurs, which is an opening in the membrane of varying diameter with rigid fibrous walls [9]. **The purpose** of the present study is to analyze the literature on congenital stenosis of the anus (CSA) to determine the pathological anatomy and physiology of CSA.

Characteristics of the pathological anatomy and physiology of CSA according to literature data

1. All authors state that CSA is characterized by a normally functioning anal canal.

2. The anal opening is narrow. It is surrounded by fibrous rigid tissue. It is initially dilated with a #7 dilator and expanded to the width of a #12 dilator [6]. There are no studies determining the required size of anal dilation [4].

3. Kiely et al. showed that functional outcomes were good when treatment was initiated 4 months of age and poor at older ages. A study of poor outcomes revealed megarectum [8].

4. Saenz et al., for the treatment of CSA, dilation was used in 77% of patients, and surgery was performed in 23% of cases, including posterior sagittal anorectoplasty in 61% of them [4]. Some authors recommend dilation alone [1,2].

5. I did not find any studies about the long-term results of treatment.

Characterization of CSA according to embryology

"Normally, in the post-cloacal period, the endodermal internal anal sphincter migrates in the craniocaudal direction to meet the ectodermal portion" [11]. It passes within the pelvic floor muscles, through the puborectalis muscle (PRM) loop, and within the deep and superficial parts of the external anal sphincter (EAS). All patients with visible fistulas have stenosis of the ectopic anus, although to varying degrees. With congenital stenosis of the anus, stenosis is always present in its normal place. **Figure 2** shows the pathological anatomy and physiology of an ARMs with visible fistula.

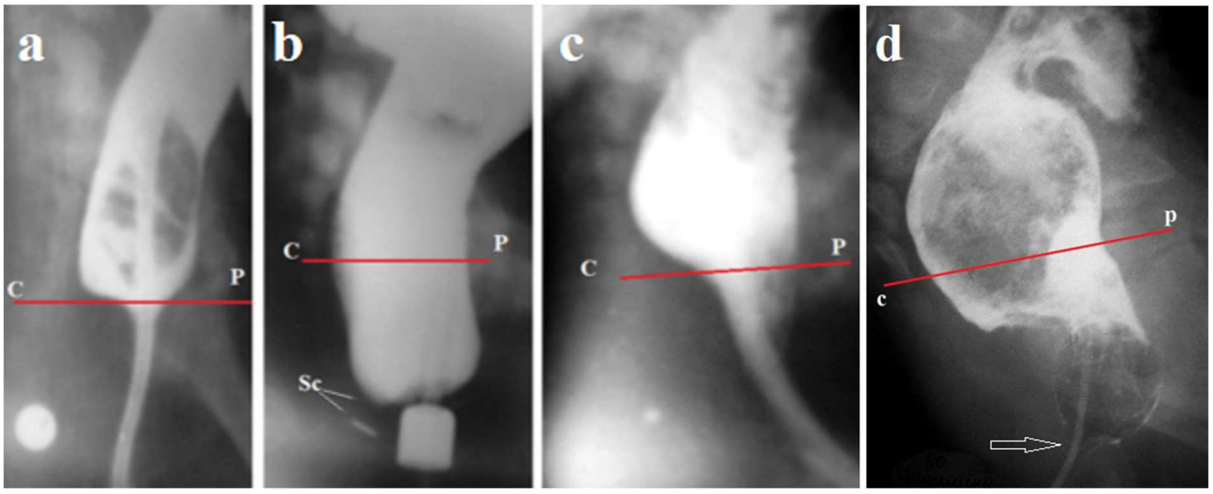


Figure 2. Anatomy and physiology of the ARMs with visible fistula. (a) Lateral radiograph of the anorectal area of a 9-month-old patient, performed after barium was introduced into the rectum through a catheter passed through the ectopic orifice. A round thumbtack was glued to the anal dimple. The sphincters of the anal canal located caudal to the pubococcygeal line (red) have contracted around the catheter, preventing barium leakage. (b) The same patient at 1.5 years of age. Radiograph taken during an attempted defecation. A wide opening of the anal canal occurred. The distance from the wall of the IAS to the thumbtack in the anal dimple (Sc) is 4 mm (the true diameter of the marker near the ectopic anus is 1.6 cm). The subcutaneous part of the EAS is in the subcutaneous fat tissue in the space between the skin and the IAS. As can be seen in Figure (a), its absence around the ectopic anus does not affect the function of barium (feces) retention. (c) During the introduction of barium through the endotracheal tube, relaxation of the IAS occurred, resulting in barium entering from the rectum into the upper part of the anal canal in front of the enema tip. At the same time, the posterior portion of the IAS was pressed against the tube due to contraction of the PRM. At the same time, the deep and subcutaneous parts of the EAS also contracted to prevent barium leakage. This is the radiographic equivalent of the rectoanal inhibitory reflex [12]. (d) A 7-year-old boy with congenital anal stenosis, who was treated with bougienage in early childhood. He complains about constipation and soiled underwear. A Foley catheter was inserted into the rectum, through which approximately 200 ml of barium was insufflated. Approximately 5 cm^3 of air was injected into the balloon. During an attempt to remove the catheter, the balloon became lodged above the anus (arrow).

Figure 2d shows that the stenosis is in the anus. Treatment was considered successful because the index finger could easily pass into the rectum. This congenital anal stenosis differs from other types of ARMs with visible fistulas only in that the narrow opening is not displaced

anteriorly but is in the anal dimple. A fecal stone with a diameter larger than the Foley catheter balloon is in the rectum. Therefore, despite a strong peristaltic wave, the fecal stone cannot pass through the narrow anal opening. Normally, the anal canal is located between the pubococcygeal line and the anus. In this case, because of repeated stretching of the PRM and the deep portion of the EAS by large fecal masses, the upper portion of the anal canal does not contribute to fecal continence. This is descending perineum syndrome, which causes fecal incontinence, since the upper part of the anal canal does not contract. Fecal retention inevitably leads to megacolon. It's not visible on an X-ray because the colon, cleaned before the barium enema, is in a collapsed state. The 200 milliliters of barium administered filled the rectum but did not expand it to its true size.

In CSA, the narrow opening is surrounded by a fibrous, rigid ring. This ring can be dilated to a certain diameter, the normal range of which is unknown [4]. However, after dilation, it remains fibrous, i.e., a rigid ring. This means that as the child grows, and the width of the rectum progressively increases, the width of the anus does not change. As a result, a secondary discrepancy occurs between the width of the stool, which forms in the wide rectum, and the throughput capacity of the rigid anus. For normal function, the anus must be elastic. An example of a normal, elastic anus is given in the article by Clayden and Lawson [13]. They performed anal dilatation under general anesthesia with alternate insertion into the anal canal of one to four fingers of the operator's hand for the treatment of children with functional megacolon. "Four of the 79 cases on anal dilatation were shown to have a minor degree of anal stenosis, with a string stricture at the mucocutaneous junction. This string stricture, approximately 1 cm from the anal verge, accepted only 2 fingers of the operator's hand during anal dilatation" [13]. Duhamel found in some patients with idiopathic megacolon fibrous changes in the IAS. He believed that "These histological aspects are the same as may be observed in congenital anal stenosis or in the biopsy of recto-vulvar fistulas" [14]. Histological examination was performed after dilatation of the anus and partial sphincterectomy of the IAS. These observations indicate that even a wide fibrous ring causes fecal retention with the development of megacolon. The observations described above raise doubts about the rationale for anal dilation for the treatment of CSA.

If the endodermal IAS does not encounter the ectodermal portion, it continues to move until it emerges (CSA, perineal and vestibular ectopia) or penetrates a cavity (the urethra in boys and the vagina in girls). However, by the time of penetration, in all cases, a functioning anal canal has already been formed (see Figure 2) (Figure 3).

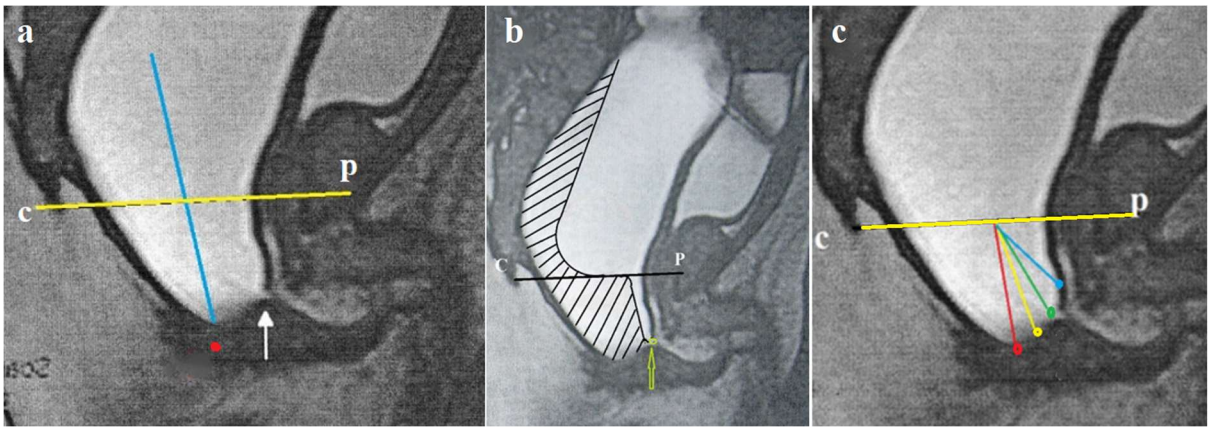


Figure 3. Augmented-pressure distal colostogram during MRI. **(a)** The anal canal located distally to the pubococcygeal line opened under high rectal pressure. The caudal wall of the IAS approached the anal dimple. The subcutaneous part of the EAS is in space between the wall of the IAS and marker near the anal dimple (red spot). The arrow shows the opening between the anal canal and urethra – bulbar ectopy of the anus. **(b)** Schematic diagram of anal canal contraction with low rectal pressure. The IAS is attached to the fistula opening. Therefore, IAS is displaced anteriorly and shorter than normal. **(c)** ARM variants: congenital stenosis of the anus – red, perineal ectopy – yellow, fistula into the bulbar urethra – green, fistula into the prostatic urethra – blue.

Comparison of treatment outcomes of boys with perineal fistula after cutback procedure and PSARP

The PCPLC registry concluded " that these patients (anal stenosis) can often be managed successfully with dilations alone. PSARP is the most common surgical repair chosen for those who undergo surgical repair" [4]. This conclusion was not substantiated, as long-term results were not presented. As previous analysis shows, dilation of the stenosis cannot lead to a cure. We can compare the treatment of stenosis in perineal ectopy using different methods: cutback procedure and posterior sagittal anorectoplasty (PSARP).

Cutback procedure. Denis Btowne рекомендовал “a simple backward incision from the displaced opening right across the normal situation of the anus, made by placing one blade of the pair of dissecting scissors in the bowel while the other lies on the skin. This opening is then kept dilated by passage of lubricated metal Hegars sound about number eight. No attempt should be made to suture the raw surfaces thus produced, and after a month or two they will be covered with supple and satisfactory new skin” [5]. Cutback procdure was widely used by other authors [6,7, 15,16,17,18, etc.].

To compare treatment outcomes after the cutback procedure with those after PSARP, I used the same objective assessment method. Before the adoption of the Krickenbeck classification

(2005), evaluation of functional treatment outcomes did not depend on subjective patient assessments. Ratings were deemed “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 [5,15,16].

Table 1. Сравнение результатов лечения мальчиков с промежностной эктопии после cutback procedure and PSARP.

Authors	Good (%)	Fair (%)	Poor (%)
1. Nixon [5]	98	0	2
2. Ackroyd et al. [15]	85	15	0
3. Kyrklund et al. [16]	90	8	2
4. de la Fuente [17]	90	?	?
A) Schmiedeke et al [18]			≈ 60
B) Lombardi et al. [19]			≈ 61.4
C) Stenström et al. [20]			≈100
D) Abo-Halawa et al. [21]			?

(1-4) – after the cutback operation; (A-D) – after PSARP.

A literature review confirms that complete transaction of the subcutaneous portion of the anus during a cutback procedure does not lead to fecal incontinence and eliminates the stenosis of the ectopic anus. Secondly, experience shows that suturing the dissected tissue leads to an inflammatory reaction, scar tissue formation, and secondary anal stenosis. If the diastasis between the wound edges is not sutured, the wound surface fills with elastic tissue within a few weeks, after which the anus can stretch to pass feces of normal diameter.

Dilation of stenosis in CSA.

Most pediatric surgeons believe that these patients can be successfully treated with dilation alone. However, as Saenz et al. demonstrated, surgical treatment was necessary in 23% of patients, including 62% with PSARP [4]. Secondly, as demonstrated above, it is not possible to cure CSA using dilation of the fibrous ring. Therefore, dilation of anal stenosis has no scientific basis. At the same time, as Table 1 shows, surgical treatment that destroys the anal canal leads to poor functional outcomes.

Rationale for surgical treatment of CSA

1. Congenital stenosis of the anus is like stenosis of the ectopic anus on the perineum and vestibule.

2. After a cutback procedure, in which together with stenosis, subcutaneous portion of the EAS is completely transected along its diameter, fecal continence and defecation are normal.

3. After PSARP, patients suffer from chronic constipation and fecal incontinence.

4. Dilation of the stenosis does not lead to recovery.

5. The diastasis between the dissected semicircles of the stenotic ring fills with elastic tissue within a few weeks.

6. Stenosis dissection is a simple procedure that is easily tolerated at any age and should be performed immediately after diagnosis to prevent the development of megacolon.

Method

Transversely dissect an anal stenosis with scissors, allowing the anus to be easily stretched to a diameter of 1-1.5 cm, depending on age. Insert a tracheostomy tube of the same diameter into the rectum and inflate the balloon with 5-7 cm of air to hold the tube in place for two weeks. The tube will ensure stool drainage from the rectum and prevent further stenosis (**Figure 4**)

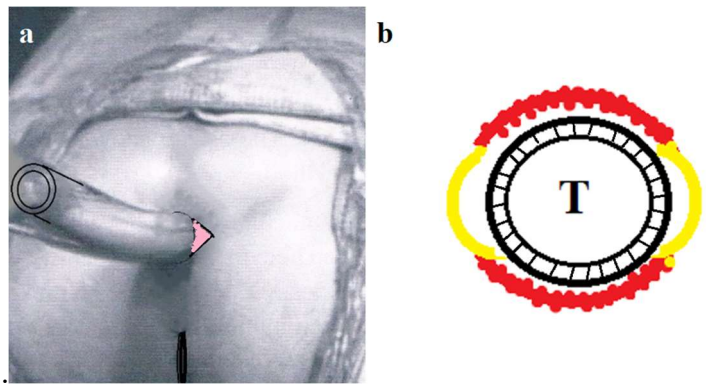


Figure 4. (a) Diagram of a transverse dissection of an anal stenosis, including the subcutaneous anus ring. **(b)** Transverse plane of the dissected anus. T – tracheostomy tube. Fibrous areas of the dissected anus – red. Diastasis between the fibrous walls, filled with elastic tissue over time – yellow.

Conclusion

Congenital stenosis of the anus is a form of ARMs characterized by the presence of a normal anal canal, where the stenosis, surrounded by the subcutaneous portion of the external anal sphincter, measures 2 to 5 mm in length, depending on age. Radiographic examination provides a precise anatomical and physiological characterization of the defect. Treatment should be initiated as early as possible to prevent the development of megacolon. Dilation of the stenosis does not resolve the rigidity of the fibrous ring. Dissection of the rigid ring with insertion of a tube into the anal canal can lead to complete recovery.

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