Review Article
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The role of Alberto Peña in the modern concept of the pathological physiology of the persistent cloaca

Abstract

Introduction: While some authors suggest that anorectal malformations (ARMs) arise from abnormal development of the cloaca, only the true cloaca arises because of the cessation of the division of the cloaca stopping at the stage of development characteristic of female birds and reptiles. It is characterized by the confluence of the rectum, uterus, and bladder into a wide space, resulting in the complete absence of the urethra, vagina, and anal canal. Currently, following the articles by Hendren and Peña, persistent cloaca is the name given to the pathology that was previously called ARM with vaginal fistula. These patients have a urethra, vagina, and anal canal that supposedly merge into a common narrow channel. These patients are operated on as if they were patients with a true cloaca.

Methods: The experience of Peña and colleagues on the theoretical basis, diagnosis, and treatment of the so-called persistent cloaca is analyzed in comparison with scientific data and treatment results.

Results: As a result of an analysis of literature and our own research, it has been shown that with all types of ARMs, except for the true cloaca, the pathology develops at a later embryonic period because of impaired development of the exodermal rudiment of the anal canal. By this time, normal development of the anal canal occurs up to the subcutaneous tissue. When the IAS does not encounter the ectodermal rudiment, as evidenced by the absence of the anus, IAS deviates forward and upward, forming a narrow, rigid fistula at the point of penetration outward or in the wall of the organ. Outside the anal canal, it forms a long and narrow canal. This process leads to the ectopy positioning of the anus on the perineum, vestibule, urethra, or vagina. What surgeons typically remove during the pull-through procedure, under the guise of a fistula, is a functioning anal canal.

Conclusion: Unlike a true cloaca, so-called persistent cloaca develops due to the penetration of the IAS the vagina before a cavity has formed within it. As a result, the IAS creates a narrow long fistula that, in some cases, obstructs the emptying of the upper part of the vagina, leading to hydrocolpos. These patients typically have a normally functioning bladder and urethra. It is likely that in most cases there is a functioning anal canal too. Poor outcomes are the result of mutilating surgeries.

Keywords: Embryology Anorectal Malformations, Embryology Persistent Cloaca, Pathophysiology ARM, Diagnosis, Treatment, Hypothesis

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Introduction

It is no exaggeration to say that Peña revolutionized the understanding of the pathological anatomy and physiology of Persistent Cloaca (PC), which changed the diagnosis and treatment of these children. Therefore, before moving on to his innovations, it is necessary to dwell on the scientific knowledge that preceded Peña's appearance, as well as on reliable scientific facts, without which it is impossible to appreciate Peña's contribution to the diagnosis and treatment of PC. Knowledge of embryology is crucial for understanding anorectal malformations (ARMs).[1] However, there are more hypotheses in the literature than established scientific facts. The embryologist Kluth described this problem as follows: "Today, the normal and abnormal development of the hindgut is still a matter of speculation. (1) the embryonic cloaca never passes through a stage resembling any form of anorectal malformation in neonates, including the so-called 'cloacas' in female embryos".[2]

The true cloaca

From an embryological point of view, "Persistent Cloaca is a rare condition that occurs only in female infants. It results from the total failure of the urorectal septum to descend and therefore occurs at an exceedingly early stage of development (10-mm stage)".[3] This condition is an example of atavism, where development halts at a stage typical of birds and reptiles. It is characterized by the confluence of the rectum, uterus, and bladder into a wide space called a urogenital sinus, resulting in the complete absence of the urethra, vagina, and anal canal. Because of this, children with this defect cannot control urination and bowel movements. Until 1982, isolated cases of cloaca were described. Okonkwo and Crocker wrote in 1977 that "Cloacal dysgenesis is a rare anomaly. Fifty cases have been reported in the literature".[4]

Indisputable scientific facts embryological development of the anorectum

a) The genitourinary septum descends and separates the cloaca into two channels: the rectum posteriorly, and the bladder and urethra (urethra and vagina in females) anteriorly.[3,5]

b) "Normally, in the post-cloacal period, the endodermal internal anal sphincter (IAS) migrates in the craniocaudal direction to meet with the ectodermal portion".[6] This refers to the unique ability of the IAS to create a elastic channel within the striated muscles.

c) The anal canal is divided into superior and inferior parts. The superior two-thirds are derived from the endoderm and are lined by simple columnar epithelial cells. The inferior one-third is derived from ectoderm and is lined by stratified squamous epithelial cells. The junction between these two types of epithelia is called the pectinate line or the dentate line.[1,2,4]

d) During the 10th week, the anal tubercles, a pair of ectodermal swellings around the proctodeal pit, fuse dorsally to form a horseshoe-shaped structure and anteriorly to create the perineal body.[3] This suggests that the formation of the distal ectodermal part of the IAS begins from the perineum and occurs through upward migration to meet the endodermal part of the IAS.

e) According to the study of Nobles (1984), at the junction of the endodermal and exodermal rudiments, the anal membrane at first appears, which ruptures during embryos with a length of 13.5–135 mm.[7] Nievelstein et al. obtained the same data.[8]

f) Nievelstein et al based on embryological observations, concluded that the abnormal communications, usually called fistulae, should be regarded as ectopic anal orifices. Anorectal malformations with the anus in normal position are best explained as late embryonic defects.[8]

These undeniably reliable data lead to the conclusion that ARMs may occur during two periods of embryological development: (1). Only the true cloaca develops because of the cessation of the development of the anorectum in females, since the cloaca normally develops in female birds and reptiles for fertilization and egg release. Therefore, the cloaca has wide lumen and elastic walls. (2). All other types of ARMs occur at a later period, when the cloaca has already been divided into the urethra, vagina, and anal canal. In these cases, the pathology occurs because of a violation of the connection between the endodermal and ectodermal parts of the anal canal. Evidence of this is the absence of the anal opening in the ring of the subcutaneous portion of the external anal sphincter (EAS). Atresia/stenosis of the rectum, and ARM type H also develop in the post-cloacal embryonic period with a violation of the ectodermal part of the anal canal of another nature.[9-11]

Regularities of pathological anatomy and physiologists of ARMs

A. All the fistulas are in the projection of the central sagittal plane. They are always displaced forward and upward from where the anus is normally located.

B. In cases of visible (perineal and vestibular) fistulas, they are slightly displaced upward from the anal dimple. The degree of upward displacement increases in cases of invisible fistulas.

C. In all cases of visible (perineal and vestibular) fistulas, there is a normally functioning anal canal, as evidenced by X-ray[12] and manometric studies,[12,13] as well as the successful outcomes of the cutback procedure, which does not damage the anal canal.[14] These postulates are discussed in detail below.

Radiological studies at ARMs

A. ARMs with visible fistula

On X-ray examination of patients with perineal and vestibular fistulas at rest, the anal canal is defined as the zone of constant contraction between the rectum and anus, near which a contrast marker is located. It contracts around the catheter inserted into the rectum, and as it does normally, preventing fecal leakage (Figure 1a,c). Its length corresponds to the normal length of the anal canal for the child's age. During attempts at bowel movements, the anal canal opens widely, as it should, to reduce resistance to the passage of feces (Figure 1b**).**

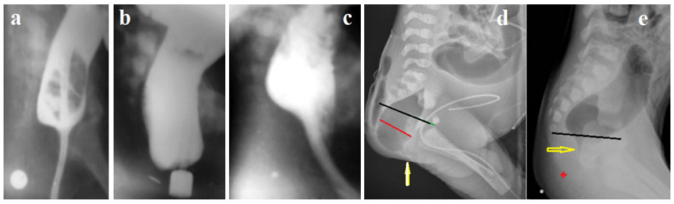

Figure 1 Lateral radiographs of the anorectum in patients with ARMs with visible fistulas.

(ab). Images of the same child taken at various times. **(a).** A catheter was inserted through the vestibular fistula into the rectum to enter contrast substance into the intestine. At rest, the anal canal is contracted around the catheter. **(b).** During a follow-up examination, an attempt to defecate resulted in a wide opening of the anal canal. The distance between the marker in the anal dimple and the wall of the open anal canal is 4 mm. It is equal to the thickness of the skin and subcutaneous tissue. **(c).** During the administration of barium through the endotracheal tube the relaxation of the IAS occurred, allowing barium to enter the upper part of the anal canal, in front of the rectal tube. This was accompanied by a drop in rectal pressure, which was measured by a balloon connected to a pressure gauge [12]. At this moment, the posterior wall of the anal canal is pressed against the rectal tube by the contraction of the PRM, which together with external anal sphincter closes the anal canal. **(d).** The arrow shows a perineal fistula when the anal canal is opened. **(e).** The arrow shows a closed anal canal. The red asterisk is in the anal fossa area.

Thus, in X-ray examinations of patients with visible fistulas, a functioning anal canal was always observed. It differed from a normal anal canal only by the absence of a distal section 2-4 mm in length, which is typically surrounded by the subcutaneous portion of the EAS. The anal canal is opened through a narrow, rigid fistula located in front of the anal dimple on the perineum or in the vestibule. The presence of a normally functioning anal canal is further confirmed by manometric studies. Both the basal anal pressure, and the rectoanal inhibitory reflex were consistent with a normal anal canal function.[12,13]

In the modern literature, starting with the publication of Peña,[15] the distal part of the intestine is called the rectal pouch or fistula, which is removed during all pull-through operations, including posterior sagittal anorectoplasty (PSARP). The theoretical basis for this misconception is the absence or marked reduction of nerve ganglia in the distal part of the intestine, compared to the rectum in healthy individuals. These authors called the anal canal a rectal sac and compared the anal canal with the rectum.[16,17] However, the anal canal, unlike other parts of the intestinal tube, does not have an intermuscular plexus.[18] Uemura et al. again after Duhamel, Alamowich, Gubler and Roujeau (1966) proved that: - "Epithelial and ganglionic distribution was similar in the distal rectal end of ARMs and in a normal anal canal".[19]

All the above scientific studies of patients with visible fistulas indicate that the distal part of the intestine, erroneously called the rectal pouch or fistula, is a normally functioning anal canal. The rigid and narrow channel between the anal canal and the anteriorly displaced opening is in the subcutaneous tissue and skin and has a length from 2 mm in newborns to 4 mm in infants. These data confirm the embryological studies of Nievelstein et al., that the abnormal communications, usually called fistulae, should be regarded as ectopic anal orifices.[8]

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B. ARM with long anopenile fistula

Craniocaudal penetration of IAS along the natural path through the center of the puborectalis muscle (PRM) and external anal sphincter (EAS) causes the formation of a normal anal canal. However, the displacement of IAS beyond the limits of this path leads to the formation of a narrow rigid fistula. This channel can be very short if IAS penetrates through the subcutaneous tissue and skin inside the subcutaneous portion of the EAS, which leads to a congenital stenosis of the anus.[20] But this fistula can be long when IAS is created by a long narrow rigid canal in the subcutaneous tissue until it penetrates outward. This is how fistula exits on the penile skin (Figure 2 a,b)[21] or rectopenile fistula (Figure 2c).[22]

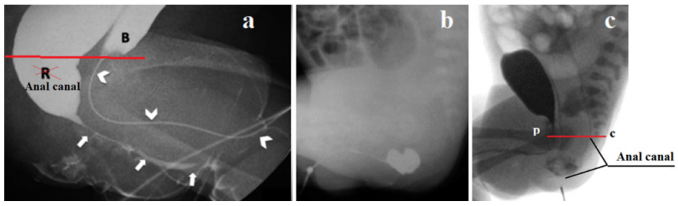

Figure 2(a) Sfoungaris et al presented the case of a male neonate with imperforate anus and a fistula exiting on the penile skin. "Colostogram and urethrography. Long and narrow recto-penoscrotal fistula (arrows) and urethra (arrowheads). R: rectum, B: urinary bladder.[21] (b, c) Yang et al. described rectopenile fistula.[22] (b). Contrast medium introduced through the fistula opening filled the narrow canal and intestine located near the anal dimple. (c). During disclosure of the anal canal below the pubococcygeal line (p-c) gas from the rectum approached the anal dimple.

I drew the pubococcygeal line (red). In both cases, an open anal canal is visible below the pubococcygeal line. In Figure 2a, the anal canal is mistakenly labeled as the rectum. A long, narrow fistula extends under the skin from the already formed anal canal to the root of the scrotum. These typical cases demonstrate that when the IAS penetrates tissues beyond its natural path, it creates a narrow, rigid fistula, the length of which depends on the distance between the anal canal and the point of entry into any cavity or outward. This suggests that the IAS first reaches the subcutaneous tissue opposite the anal fossa. If a normally formed anal canal does not encounter the ectodermal rudiment coming towards it, it turns forward and continues until it penetrates a cavity. The findings described above are consistent with a recent statement by pediatric surgeons from the ARM-Net Consortium: "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".[23] As this definition indicates, the authors do not limit the ectopia of the anal canal to visible fistula alone, which is confirmed by both manometric and radiological studies.[24]

C. ARMs Without Visible Fistula

ARMs without visible fistulas (whether without a fistula, with urethral or vaginal fistulas) differ from ARMs with visible fistulas only by higher ectopia of the anus. Stephens (1953) proposed the concept of a pubococcygeal line from the lower part of the pubic bone to the distal coccygeal vertebra. He showed that this line corresponds to the location of the puborectalis muscle (PRM) separating the rectum and anal canal.[25] Since then, it was believed that if the intestine was visible below the PRM line, then in this case there was an anal canal, and the defect was classified as a low type, and if it was at or above this line, this indicated the absence of an anal canal. Over time, all cases with visible fistulas were classified as low type, and efforts were made to preserve the anal canal. Cases with invisible fistulas were classified as high types and pull-through surgery was used for treatment. However, such a division into high and low types turned out to be insufficiently precise. For example, Ohama et al. demonstrated that the functional IAS, which exists at the rectal end even in high and intermediate anorectal malformations, should be preserved during reconstruction due to its critical role in maintaining anal continence.[26] When it became clear how the anal canal functions and we began to use provocation of high rectal pressure to open it during radiographic examination, it turned out that high types also have an anal canal (Figure 3).[27,28] What now is meant by a high type of ARM? Since all ARMs have an anal canal, they all belong to low types. We call ARMs high or low based on the degree of ectopy of the anus.

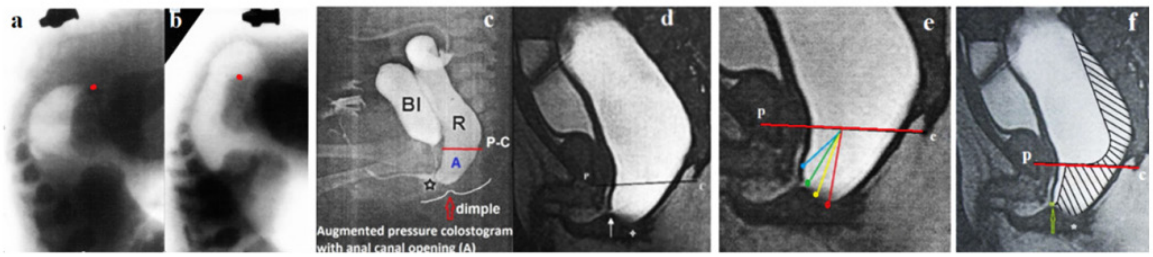

Figure 3 Radiologic studies in males with ARM without visible fistulas. (a). On the radiograph of a newborn 30 hours after birth, the gas reaches the caudal part of the ischium (red dot), (b). In the same patient, during abdominal compression, the gas penetrated the anal canal and approached the contrast marker glued to the anal dimple. (c, d) Augmented-pressure distal colostograms: (c-X-ray) and (d-MRI). In both cases, under the high pressure, the anal canal (caudal to P-C line – red) opened, and its caudal wall approached the anal dimple. (e). Scheme of anal ectopy variants at high pressure in the rectum. The anal canal is wide open: red indicates the normal position of the anus (congenital anal stenosis); yellow - perineal fistula; green - bulbar fistula; blue - prostatic fistula. (f). Scheme at low rectal pressure. The rectum is narrow. The anal canal is in a contracted state.

Although the site of IAS penetration in urethral fistulas is significantly higher than in cases with visible fistulas, by the time of displacement, the anal canal is already formed, and its blind ending is located 2-4 mm from the anal dimple. A fistula is the short channel in the wall of the organ through which the IAS penetrates the organ. Its length is equal to the thickness of that wall of the organ through which it penetrates. Since ARMs with urethral fistulas, as well as those without fistula, have a normally formed anal canal, they should be considered low types. Peña and colleagues confirm these observations. They claim: - «It is extremely important in this regard to understand that the lowest part of the rectum is usually collapsed due to the muscle tone of the funnel-like striated muscle mechanism that surrounds the rectum in 90% of cases…".[29] However, it is well known from anatomy that there are no muscles around the rectum. The muscles that compress the intestine and prevent the leakage of intestinal contents are located around the anal canal. Since the authors

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do not cite any specific studies, the 90% figure should be interpreted with caution. From the above data, it follows that the IAS is involved in the formation of the anal canal until it reaches the subcutaneous tissue. It can penetrate outward through the ring of the subcutaneous portion of the EAS. Any such penetration is almost always accompanied by the formation of stenosis at the penetration site, leading to congenital anal stenosis. More often, the IAS moves anteriorly and upward, each time leaving behind a functioning anal canal.

In females without visible fistulas, the only thing that is in the way of the ectopy of the anus is the vagina, so from an embryological point of view, only ectopia of the anus in the vagina is possible, which, as shown in the diagram proposed by Stephens, can be lower or higher (Figure 4). [25,30]

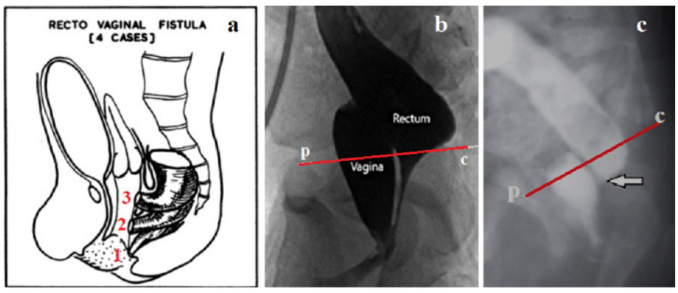

Figure 4 ARMs without visible fistulas in females. **(a).** Scheme of ectopy of the anus in girls: 1 - vestibular, 2 - low vaginal, 3 - high vaginal. **(b).** In a female with a low vaginal fistula, the rectum is not dilated. The contrast agent introduced through the colostomy passes through the anal canal (caudal of pubococcygeal line) and comes out of the vagina in a wide stream. The moment of contraction of the anal canal is recorded. **(c).** In a female with high vaginal fistula, the contrast agent introduced through the colostomy passes through the anal canal to the high part of the vagina. The upper part of the anal canal is widely open to the level of the fistula (arrow), and the lower part is in a closed state since the pressure in the rectum is not high enough.

Literature review and our own research indicate that the above types of ARMs develop in the post-cloacal period because of impaired development of the ectodermal rudiment of the anal canal. The upper part of the anal canal develops normally because of the cranio-caudal movement of the IAS. Having reached the subcutaneous tissue and not encountering the ectodermal rudiment on its way, the IAS continues to create a canal, moving forward and upward until it penetrates the outside or into some cavity. Beyond its natural path, it creates a narrow and rigid fistula. If it penetrates through the subcutaneous tissue and skin inside the subcutaneous portion of the EAS, stenosis of the anus (not the anal canal) occurs, ranging in length from 2 mm in a newborn to 4 mm in an infant. In other cases, ectopia of the anus occurs to varying degrees, but the distance from the caudal wall of the anal canal to the imperforate anal pit remains within the same limits. Thus, in all cases, regardless of the degree of ectopia, there is a normally functioning anal canal. If the anal canal is in a contracted state, it is impossible to see it. It opens only when the pressure in the rectum is high. During surgery under anesthesia, the pressure in the rectum decreases, which leads to a contraction of the anal canal. Those surgeons who do not know about the existence of the anal canal and find a narrow channel, think that it is a fistula, and resect it, and in its place, they lower the rectum, which has a completely different function.[12]

Modern ideas about the so-called "persistent cloaca"

An ARM, which is now called persistent cloaca, before Hendren's classification was known as ARM with recto-vaginal fistula because feces were expelled from an opening in the perineum in the absence of an anus. The reason for highlighting this anomaly was the existence of two similar types of anomalies. In rare cases, the vagina had a wide lumen (see Figure 4). Sometimes a narrow, rigid channel was observed, and in some cases, a closed cavity formed in the upper part of the vagina, leading to the accumulation of a large amount of fluid (hydrocolpos). Hendren termed this second type of anomaly persistent cloaca.[31] Although the anatomical and physiological descriptions of this defect lack a scientific basis, these children are treated as though they suffer from a true cloaca.[32]

Hendren did not conduct any studies to assess the function of the urinary system and lacked a clear understanding of the pathological anatomy of the defect. For example, he believed that hydrocolpos resulted from vaginal distension by urine. He wrote: "Intermittent catheterization of the bladder and/or urine-filled vagina can often provide adequate decompression".[19] Peña adopted the idea of persistent cloaca. However, neither he nor his colleagues conducted studies on the state of the urinary system. In 1997, Peña proposed the total urogenital mobilization (TUM) operation to repair cloaca. During the presentation of Peña's article, Hendren asked, "Do you think the external urethral sphincter is of any importance in these patients? And if it is, do you think this mobilization risks any injury to the external sphincter?" Peña responded, "My experience in managing cloaca is that girls who suffer from urinary incontinence do so not because of a lack of urinary sphincter but rather due to the lack of bladder contractility".[23] Of the numerous articles by Alberto Peña, I have not found a single scientific study. All his innovations are the result of his "experience". Even though experience is not scientific evidence, he and his followers do not recognize scientific data that contradicts Peña's experience. Below is an analysis of the situation with "persistent cloaca".

A. The modern description of the anatomy and physiology of the "persistent cloaca" contradicts known patterns of embryology

Currently, following Hendren and Peña, it is believed that the already formed the urethra, vagina, and rectum are connected during the embryonic period into a single canal called the cloaca.[33] However, the presence of the urethra and vagina indicates that patients have successfully passed the cloacal stage and therefore have a urethra, vagina, and must also have an anal canal. In the post-cloacal period three separate canals form within the perineal tissues, with their walls adjacent to each other. There is no evidence to suggest that these three channels can fuse into a single channel, especially not in one specific location. This assumption contradicts common sense. Furthermore, in a study by Runck et al., the molecular pathologies underlying cloacal malformations were examined. Their findings using Shh knockout mice showed that "...the epithelium from the common channel has matured and does not exhibit the same characteristics as cloacal epithelium before septation...".[34] This means that the epithelial samples taken by pediatric surgeons from the so-called "common canal" were more mature than what would be expected in an undivided cloaca. From an embryological perspective, therefore, the "persistent cloaca" has no relation to the cloaca. By calling the anal canal the rectum (rectal pouch or fistula), these authors, as shown above, justify its extirpation during PSARP.

B. The generally accepted description of radiological studies of the persistent cloaca does not correspond to the true anatomy

Radiological diagnostics are used to determine the supposed confluence site of the urethra, vagina, and rectum. The surgical treatment method depends on the fusion level.[35,36] Figure 5 shows sample MRI studies.

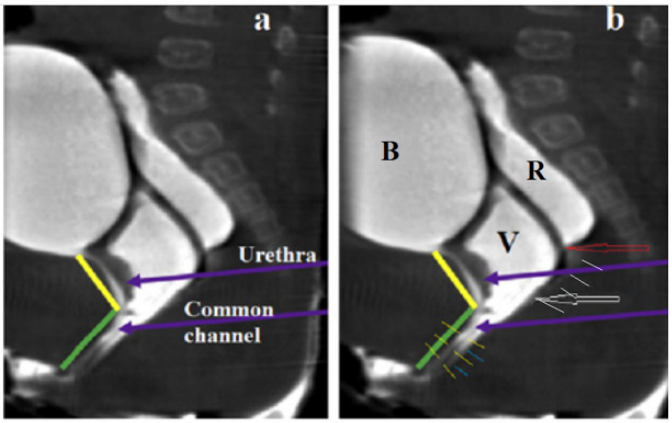

Figure 5(a) MRI study from the article by Wood et al.,[35] with the caption "Short common channel cloaca with adequate urethra." The authors suggest that, in the angle between the yellow and green lines, the urethra merges with the vagina to form a common channel. In the copy of the same image **(b)** between the yellow lines, you can see the continuation of the urethra up to the perineum, and next to it, the blue lines indicate the narrow vaginal canal. Between them, a dark dividing line can be observed, representing the fusion of the walls of the urethra and vagina. The upper part of the vagina (V) is dilated (hydrocolpos). The red arrow points to a rectum above the contracted anal canal, which is shown with white lines. It is visible between the red and white arrows. The site of IAS penetration into the vagina divides the vagina into an upper, widened part and a lower, narrow part.

The MRI description from the article by Wood et al.,[35] which became a tracing paper for other surgeons,[36] does not correspond to the true anatomy of the anorectum. First, there is a urethra, vagina and anal canal formed in the postcloacal period. Second, the urethra is of normal length. It does not connect with the vagina but reaches the perineum in the vestibule. Third, there is a high ectopia of the anus in the vagina, which divides the vagina into two parts: an upper widened part (hydrocolpas) and a lower narrow part that opens near the urethra. It follows that this defect is an ectopia of the anus in the vagina with stenosis at the level of the ectopia, above which there is a hydrocolpas, and below which there is a narrow rigid canal. This pathology is not related to the cloaca. Her urinary system, contrary to Peña's assertion, is not impaired. The presence of an anal canal makes it possible to perform surgery to preserve it to achieve normal fecal continence and defecation.

C. Comparison of the results of different treatment methods for "Persistent Cloaca".

In a retrospective review by Rosen et al. of all girls with ARM treated by Peña from 1980 to 2000, only 6 out of 617 patients were found to have a true rectovaginal fistula, an incidence of 1%. In numerous cases where a vaginal fistula was diagnosed at other institutions, the authors considered it a misdiagnosis, asserting that it was a persistent cloaca.[37] To evaluate the value of a novel approach to treating children with so-called "persistent cloaca" it is important to compare the treatment results before and after the diagnosis was changed.

As shown in the systematic review by Versteegh et al., after treating a "persistent cloaca" as if it were a true cloaca, voluntary bowel movements were reported in 57% of patients, soiling in 71%, and constipation in 51%. Spontaneous voiding was reported in 46%. Additionally, 42% of patients used intermittent catheterization, and 22% had a urinary diversion.[38] First, in the treatment of vaginal fistulas before categorizing them as "persistent cloaca", there were no issues with urinary diversion. Comparisons of outcomes before and after changes in diagnosis from vaginal fistula to persistent cloaca are not described in the literature. Secondly, Kittur and Vora conducted a follow-up study involving one adolescent and eight adult female patients born with cloaca, who had undergone rectal pull-through surgery in infancy, leaving the urogenital region untouched. After full growth, the common channel was used as a vagina with introitoplasty and dilatation, or with additional surgical intervention. All patients retained an intact urogenital complex, and none experienced recurrent urinary tract infections until adolescence. All patients are now beyond adolescence, and five have married. Three reported regular sexual intercourse, and one has conceived. Of the three patients who are not married, introitoplasty was performed to allow for free menstrual flow. Only one of the nine patients reported urinary and fecal incontinence. The authors argue that there is a strong case against subjecting cloaca patients to TUM, as it has significant potential to cause urinary incontinence. Furthermore, the common channel can be used as a vagina in conjunction with introitoplasty and dilatation.[39] AbouZeid, based on the analysis of long-term results, concluded that in some cases of cloaca, it might be better from both functional and cosmetic perspectives to perform only introitoplasty while leaving the common urogenital sinus uncorrected.[36]

Based on the literature review and our own research, the following was established:

1) The expanded concept of the so-called "persistent cloaca" has nothing to do with the true cloaca. This pathology develops in the post-cloacal period, when the urethra, vagina and anal canal have already emerged. The only hole in the crotch is displaced forward from the usual location of the anal dimple.

2) There is no evidence that these patients have congenital pathology of the urinary and anorectal functions. Preservation of the urinary system without intervention leads to significantly better functional results than scientifically unsubstantiated correction.

3) It is known that the vagina is narrow and has rigid walls. Sometimes there is obstruction of the vaginal lumen, because of which fluid accumulates in the upper part of the vagina.

I propose a hypothesis of the embryonic development of the so-called "persistent cloaca", which explains how this type of ARM arises and eliminates contradictions in the description of pathological anatomy and physiology.

The hypothesis of embryonic development of the so-called "persistent cloaca"

The pathology develops in the post-cloacal period, when the cranio-caudal advancement of the IAS reaches the subcutaneous tissue, but without encountering the ectodermal rudiment of the anal canal on its way, it begins to shift forward and upward. The IAS penetrates the vagina and can create two types of vaginal fistulas. (a). If a cavity has already formed in the vagina by this time, a typical ARM with a short narrow fistula is formed, the length of which is equal to the thickness of the vaginal wall. In such cases, the vagina has a wide cavity and elastic walls (see Figure 4). Depending on the place of penetration, the fistula can be located lower or higher. (b). If at the time of penetration of the IAS into the vagina the cavity has not yet formed, then the advancement of the IAS creates a narrow and rigid canal, the same as in boys with rectopenile fistula (see Figure 2). In both cases, by the time the IAS penetrates the vagina, the urethra and anal canal are already normally formed. Figure 6 shows the anatomical diagrams of the so-called "persistent cloaca" anatomy.[25,33]

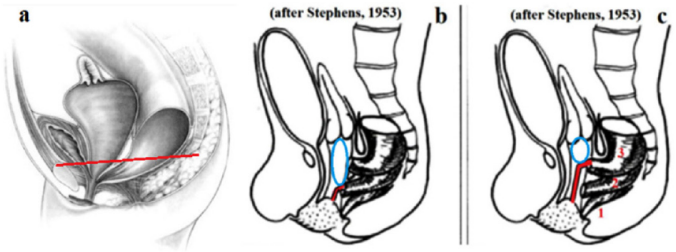

Figure 6(a) The diagram of the "persistent cloaca" from the article by Levitt and Peña.[33] Caudal to the pubococcygeal line, which I have drawn (in red), are the urethra, vagina, and anal canal. In the diagram, all channels merge in one place. However, as shown above, the urethra does not merge with the vagina but is located next to it. The upper part of the vagina above the ectopia of the anus is dilated (hydrocolpos). **(b-c)**. On the diagram of fistula locations in girls, proposed by Stephens (1-vestibular; 2-vaginal low; 3-vaginal high), I have drawn a channel in red, which is created by the IAS in the vagina in cases where an internal cavity has not yet developed. The blue oval shows the location of the hydrocolpos above the site of the IAS ectopy, where the release of fluid from the upper part of the vagina is blocked. **(b).** Low vaginal ectopy. **(c).** High vaginal ectopy with narrow and rigid vagina.

Discussion

Two of Peña's statements, made without any evidence and contrary to the available scientific research, fundamentally changed the entire pediatric colorectal surgery. First, to justify posterior sagittal anorectoplasty (PSARP), he announced that in ARM the anal canal is absent. Second, he announced that the type of ARM previously thought to be a vaginal fistula was persistent cloaca. After that, such patients began to operate as patients with a true cloaca and the results of these operations were as if they had no urethra, vagina, and anal canal.[33] As shown above, from an embryological point of view, it is impossible to reunite the already separately created urethra, vagina, and anal canal. It was shown that the histology of tissues from the so-called common canal showed a later development compared to the true cloaca.[34] The change in the name and the idea of the pathological anatomy of the "persistent cloaca" were not justified by scientific research, but only by Peña's "experience". Even if we consider the operations proposed by Peña to be experiments, they were not analyzed and did not lead to an improved understanding of the pathophysiology of the disease or to improved correction of the defect. The absence of an anus in the subcutaneous part of the EAS, through which the rectum was pulled during PSARP, indicates a late development of the defect. There were no studies and no evidence that the function of the bladder and urethra was impaired.

As the analysis of CT and MRI, based on which the authors determined the length of the common canal, shows, the method of determination was incorrect. The authors grossly ignored the obvious facts about the presence of a normal length of the urethra. Peña's colleagues describe various properties of the allegedly persistent cloaca that have no scientific basis but are used to select Peña's proposed surgical treatment. For example, in the article by Halleran et al., the goals of urinary reconstruction are positioning of the bladder neck above the urogenital diaphragm to maximize future urinary continence and create a visible urethra that can be catheterized if needed.[40] However, there is no study that has proven that with persistent cloaca the bladder neck is not positioned at the proper level. In the figure from the article by the same authors (See Figure 5), the bladder neck is positioned normally - at the level of the pubococcygeal line. The same article presents alleged variations in urethral length measured on voiding cystourethrograms of healthy female patients between ages 6 and 36 months. The mean urethral length for patients age 6-12 months was 2.50 cm without significant changes up to 36 months. Of 91 patients, 87 (96%) had a urethral length >1.5 cm.[40] However, these measurements, which are the basis for choosing surgery, are incorrect, since it is known that in adult healthy nulliparous women the length of the muscular region of the urethra, measured on MRI, ranged from 20 to 35 mm (mean 24 mm), and striated urethral sphincter was observed 10 and 15 mm below the bladder base.[41] If in 29% of healthy adult women the length of the urethra is 2 cm, then in girls in the first year of life the normal urethral length should be significantly less. This error can only be explained by the fact that on CT and MRI the computer shows the true dimensions, while cystourethrograms are performed with projection magnification, which Halleran et al. did not consider.

Peña and Levitt without any evidence believe that a urethra less than 1.5 cm long is the cause of urological problems. However, if 29% of healthy adult women have a 2 cm urethra, why is a urethra less than 1.5 cm a problem in girls under 3 years of age? The suggestion that isolating the urethra and lengthening it by creating an additional canal can improve its function is unscientific. The urethra, which is a sphincter, after isolation from the surrounding tissue it loses neural regulation and stops responding to high bladder pressure, necessitating intermittent bladder catheterization. Violation of the blood supply to the urethra turns it into a fibrous tissue with possible narrowing, up to stenosis. These operations turn the functioning urethra into a fibrous channel for catheterization, which results in a lifelong disability for children who previously had a normally functioning urethra.

The goal of changes in the field of medicine is to improve the results of diagnosis and treatment. To justify the change in diagnosis and treatment of the so-called persistent cloaca, it was necessary to compare the results of treatment of these patients during the period when they were diagnosed as ARM with vaginal fistula with the results of the operations proposed by Peña. First, the analysis of the literature does not confirm that there were special problems in the treatment of patients with vaginal fistulas that go beyond the problems of other types of ARM. Second, Peña and his followers did not compare their results with the results of previous generations. Third, at least two studies indicate that leaving the urogenital tract intact in the so-called persistent cloaca shows normal function.[36,39] In other words, the poor results after the operations proposed by Peña are not due to congenital pathology but are a consequence of destructive operations proposed without scientific justification.

Results of treatment of persistent cloaca.

In a review article by Versteegh et al., long-term results after surgical treatment of persistent cloaca are presented, using the methods proposed by Peña and Levitt. Nine percent to 41% of cloaca patients have been reported to suffer some degree of urinary incontinence, with 18%-27% having undergone urinary tract surgery. Studies show that 11%-75% of cloaca patients have abnormal or deteriorating renal function. Some of these patients developed established renal failure and underwent kidney transplantation. Deaths because of chronic kidney disease are reported but the true incidence remains unknown.[42] In an article by Wood and Levitt, the reasons for poor results after treatment of persistent cloaca are cited as allegedly unrelated to surgery: (1) The type of malformation, (2) sacral development or lack thereof as suggested by the sacral ratio, and (3) spinal abnormalities all have an influence of the potential for bowel control. The interaction between these three (the ARM continence index) and their individual contribution is the subject of ongoing prospective research.[43] Thus, these statements have no scientific basis, since the authors state the reason for the poor results even before the results of the "prospective research" were obtained.

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To evaluate the results of treatment, it should be borne in mind that in the case of the so-called persistent cloaca, two surgeries are performed: PSARP and on the urinary tract. Following Peña, pediatric surgeons ignore the presence of the anal canal, resect the IAS and in its place lower the devascularized and denervated rectum, transect the PRM and all parts of the EAS except the subcutaneous, and disconnect the rectum from the levator plates, which normally open the anal canal to reduce friction when passing feces. As shown by Chong et al., only PSARP leads not only to fecal incontinence and severe chronic constipation, but also to serious damage to the urinary system. From 50 patients in median age at last follow up was 18 years (range 12–34 years) after ARMs correction (4 with cloaca), chronic kidney disease stage II or above was found in 14 (28%) patients, of whom four required a renal transplant. Abnormal bladder outcomes were found in 39 (78%) patients. Augmentation cystoplasty with Mitrofanoff had been performed in 12. Of those who had not undergone cystoplasty, 17 had urinary symptoms, including urinary incontinence in 12. Of the 39 patients with abnormal bladder outcome, 19 (49%) did not have a spinal cord abnormality. There was also no significant statistical association between level of ARM and abnormal renal outcome or presence of bladder abnormality.[44] According to Lane et al., two from 21 patients have required renal transplants for congenital renal dysplasia, and two have developed chronic renal failure associated with the sequalae of vesicoureteral reflux. Eleven (52.3%) of the patients manage their bowels with an antegrade continent enema (ACE), and two of the LCC cloaca are defunctioned with a colostomy. Clean intermittent catheterization is performed by 12 (57%) of the patients, either per urethra or via a Mitrofanoff channel.[45] In the article by Zamir of 60 patients with cloaca, bowel diversion was done as transverse colostomy in 39 (65%) patients, sigmoid colostomy in 17 (28%) patients, and ileostomies in three patients (5%) patients, to facilitate subsequent vaginal reconstruction. In patients with low confluence, anoplasty served the purpose. Regarding hydrocolpos, tube vaginostomy was done in 14 patients. Three (5%) patients had dilated floppy bladders. It was drained per urethra, and no urinary diversion was needed. Hydronephrosis and hydroureter gradually subsided postoperatively in nine patients.[46] This study proves that at least some of the girls had a functioning anal canal. Meanwhile, it is known that at low pressure in the rectum, the anal canal is in a closed state. If the author had applied abdominal compression, he would have been convinced that the anal canal was present in all patients. Secondly, the author has proven that bladder dilation and hydronephrosis are caused by compression of the hydrocolpos. They disappeared after drainage. Thirdly, the author had no problems with bladder catheterization, and no urinary diversion was needed in his patients. These observations confirm the assertions of other authors that severe urinary system disorders in patients with cloaca are caused by unnecessary operations.[36,39]

The proposed hypothesis of the embryonic development of the so-called persistent cloaca proves that these patients represent an ectopia of the anus in the vagina before the appearance of a cavity in the vagina. Therefore, as with all other ectopia of the anus, they have an anal canal and a normally functioning urinary system. The distal part of the vagina is represented by a narrow rigid fistula, which can be blocked by the fistula wall and cause obstruction of the canal with accumulation of fluid in the upper part of the vagina (hydrocolpos). The latter compresses the urinary tract, causing hydronephrosis. Untimely or incomplete removal of fluid leads to kidney damage. Drainage of hydrocolpas is possible by passing a tube through the narrow part of the vagina, which will eliminate the septum and will facilitate stretching of the vagina. These children can be healthy if the anal canal is preserved and the urinary system is left untouched.

Conclusion

Unlike a true cloaca, so-called persistent cloaca develops due to the penetration of the IAS the vagina before a cavity has formed within it. As a result, the IAS creates a narrow long fistula that, in some cases, obstructs the emptying of the upper part of the vagina, leading to hydrocolpos. These patients typically have a normally functioning bladder and urethra. It is likely that in most cases there is a functioning anal canal too. Poor outcomes are the result of mutilating surgeries.

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