Persistent cloaca. Pathophysiology, diagnosis, and treatment.

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From the point of view of embryogenesis cloaca results from the total failure of the urogenital septum to descend. As a result, there is no division into the urogenital and rectal parts and there are no urethral, genital, or anal openings. The bladder, uterus, and rectum open into a common canal called the cloaca [1]. This is the only correct definition of a cloaca malformation, which was published on Wikipedia. But after I drew the attention of the forum members, citing Wikipedia, Levitt changed the Wikipedia article, and now it is very difficult to find the correct interpretation of this name.

Until 1982, isolated cases of cloaca were described [2,3,4]. Okonkwo and Crocker wrote in 1977 that “Cloacal dysgenesis is a rare anomaly. Fifty cases have been reported in the literature” [4]. In 1982, Hendren "described13 more cases of urogenital sinus malformation with an anorectal anomaly, usually rectal atresia with rectovaginal fistula", which he called the cloaca [5]. Since then, the persistent cloaca is a defect in which there is the **confluence** of the rectum, vagina, and urethra [6,7].

Firstly, if the cloacal membrane already has divided the cloaca into three functional parts, then this defect has nothing to do with the cloaca. Just as it is impossible to call the anal canal a rectal sac and remove it during an operation, it is also impossible to call the described defect by cloaca, immediately endowing it with functional problems of the cloaca and justifying severe complications. **Secondly**, if there was a division of the cloaca into three functional parts, then their confluence in the future is theoretically impossible.

Third, before the change name and treatment, neither journal articles nor textbooks found any difference in the functional results of pull-through operations in patients with vaginal fistulas (read persistent cloaca) compared with other types of high defects.

Fourth, I have not found a single study that determined the function of the bladder, internal and external urethral sphincters, or the level of rectal anomaly in these patients before surgery.

In 1989, Peña described the author's personal experience in the surgical treatment of 54 patients with posterior sagittal ano recto vagino urethroplasty (PSARVUP). The length of the common channel varied from 0.5 to 7 cm. Common channels longer than 3 cm usually required some technical alternative to replace the vagina. In at least 34 cases, the vagina was reconstructed primarily without any additional technical manoeuvres. Different degrees of vaginal and uterine septation were found in 25 of 50 cases. Hydrocolpos was an associated defect in 14 of 49 patients. Sixty-eight percent of the patients had an important associated urological defect. Five (9%) patients had urinary incontinence that was successfully managed by intermittent catheterization. Only seven patients required a laparotomy in addition [6].

In 1997, Peña offers the operation total urogenital mobilization to repair cloaca that avoids the separation of the vagina from the urinary tract with associated morbidity {urethrovaginal fistula (7% to 11%), vaginal strictures (10%), and acquired vaginal atresia (2%)}, which he did not write about in 1989. During the presentation of the article, Hendren asked the question: Do you think the external sphincter is of any importance in these patients? And if there is any importance to it, do you think that this mobilization risks any injury to the external sphincter? Peña's answer: "My experience in the management of cloaca is that the girls who suffer from urinary incontinence don't suffer from the lack of urinary sphincter but rather because of lack of contractility of the bladder" [8].

It is known that “the striated urogenital sphincter muscle can contract to assist in maintaining continence in incontinent women whose vesical neck is not competent” [9,10]. However, patients operated on with PSARP have two other equally serious problems. (1) Dissection of the puborectalis muscle, and especially its pubourethralis portion, reduces the ability to retain urine [10]. (2) Impaired bladder emptying is due to the fact that the separation of the vagina from the urinary tract leads to denervation of the urethra. The urethra is a sphincter, not just a tube. Therefore, the denervated urethra does not receive information from an overflowing bladder and does not open. The total urogenital mobilization proposed by Peña, which was called upon to correct the mistakes of the previous tactics, leads to an even greater denervation of the pelvic floor.

Let us take turns looking at the anatomy and physiology of each of the three systems.

Anorectum

In the diagram of ARM with vaginal fistula Stephens shows three levels (**Figure 1**) [11].

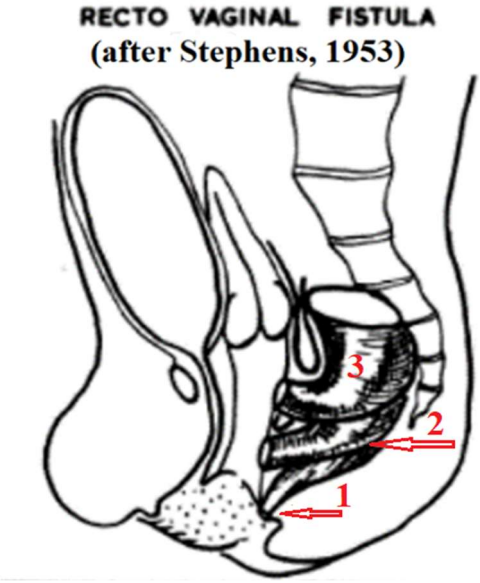

Figure 1. (1) vestibular ectopy, (2) low vaginal ectopy, (3) high vaginal anomaly.

Wood et al believe that in persistent cloaca “The P-C line is a good guide as to whether the true rectum can be mobilized from a posterior sagittal approach or will require a trans-abdominal approach. If the normal lumen of the rectum lies below the P-C line it can reliably be mobilized through a posterior sagittal incision” [12]. “Persistent cloaca malformations can be considered as the female equivalent of high anorectal malformations which correspond to rectal atresia accompanied by congenital recto urethral fistula in male” [7]. Meanwhile, Kraus et al showed that in at least 90% of boys with urethral fistulas, the distal gut is located below the last coccygeal vertebra [13].

Incorrect definitions lead to false conclusions. According to scientific research, the intestine located distal to the P-C line is the anal canal, not the rectum [11]. Based on the above, urethral fistulas, as well as vaginal fistulas, are mostly low ectopy. Someone calls urethral fistulas a defect with a good prognosis, someone a high anomaly, but from a scientific point of view, in most cases, this is an ectopy of the anal canal. Scientific facts are verifiable.

It is known that the anal canal is formed as a result of the fusion of the proximal endogenous and distal exogenous primordia. All low ARMs are characterized by the absence of canalization of the distal anal canal, i.e., exogenous primordium. The endogenous anlage moves caudally in the perineal tissues, forming the internal anal sphincter. Not meeting an exogenous channel on its way, it deviates anteriorly until it penetrates some cavity. Outside the perineal tissue, the canal acquires the property of a fibrous canal of various lengths - from 2 mm when penetrating through the skin and subcutaneous tissue in perineal fistulas, to several centimeters in scrotal or penile fistulas. It is important to understand that the formation of an ectopic anus is the final stage in the formation of a defect. By this time, in all the low anomalies, a functioning anal canal had already formed.

There are clear patterns of ectopy of the anal canal. Deviating from the obstacle (subcutaneous tissue), it always goes only forward and strictly in the middle of the sagittal plane. It cannot be assumed that urethral fistulas form in the cloacal

stage, as a result of improper separation. Subcutaneous passages several centimeters long also pass strictly along the perineal suture, but it is obvious that they are not related to the cloacal stage. I hypothesize that vaginal fistulas result from ectopy of the anus with penetration into the vaginal wall. In cases where an internal canal has formed in the vagina, a typical vaginal ectopy occurs with a fibrous canal in the vaginal wall. In those, more frequent observations, when the internal vaginal canal has not yet been realized, a narrow fibrous canal is formed, which descends and penetrates the skin of the perineum **(Figure 2).**

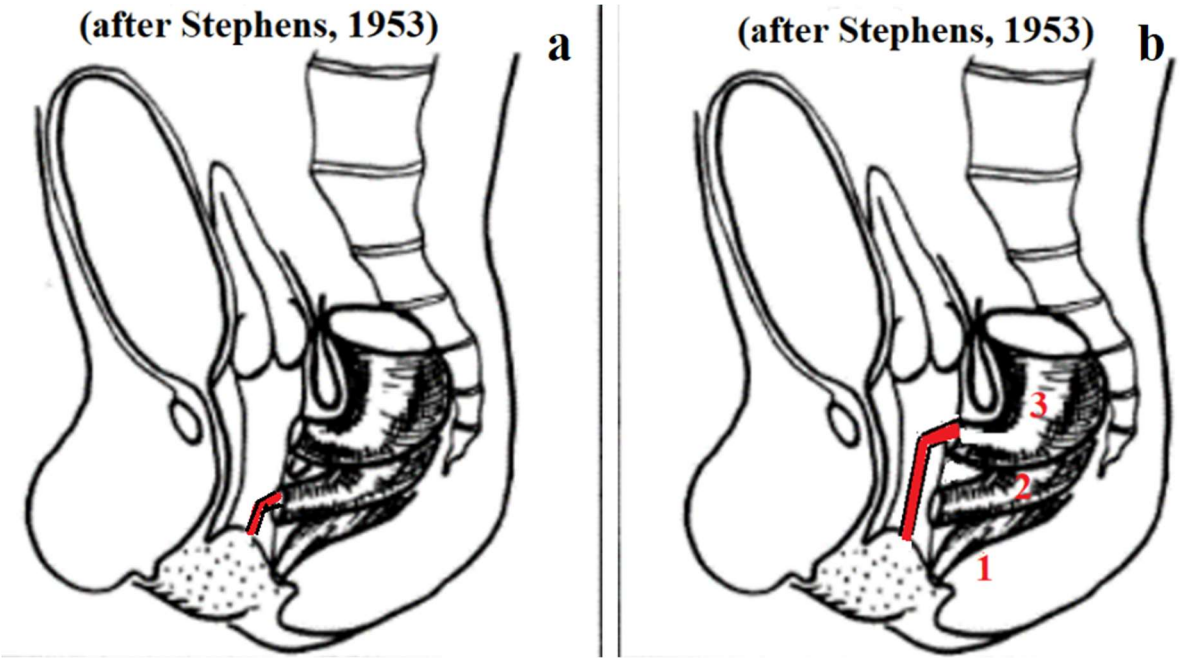

Figure 2. Stephens's scheme of the ARM with vaginal fistulas was used [11]. (a). Low vaginal fistula {2}; (b) high vaginal fistula {3}. {1} vestibular fistula. On this diagram, two options for the formation of long narrow fibrous canals with red lines are shown inside the vagina.

Vaginal malformations. This hypothesis explains the development of the so-called persistent cloaca and as well as its typical symptoms.

1. Embryonic development of ARM with vaginal fistula occurs after the cloaca is divided into three functional sections. By analogy with other types of ARM, one should expect that the bladder and urethra are normally developed in them.

2. A significant percentage of patients have a functioning anal canal since the intestine is located caudal to the P-C line.

3. The entrance to the vagina is occupied by a narrow rigid fistula, which has different lengths in different patients.

4. This canal in a significant part of patients causes obstruction of the exit from the upper part of the vagina, which leads to hydrocolpos.

Urinary system. There are no studies in the literature on the state of the urinary system in these patients before surgery. In patients with vestibular fistulas after operations that preserve the anal canal, no disorders of the urinary system have been described [14,15,16].

However, after PSARP the range of reported prevalence of long-term active problems was as follows: fecal incontinence, 16.7% to 76.7%; chronic constipation, 22.2% to 86.7%; urinary incontinence, 1.7% to 30.5%; ejaculatory dysfunction, 15.6% to 41.2%; and erectile dysfunction, 5.6% to 11.8%" [17] . Similar results are described in other studies [18,19,20].

Changes in the name of the pathology and treatment tactics were made without any substantiating functional studies. How did this treatment affect the results? There is not a single study comparing the results of the treatment of those numerous patients who were treated for vaginal fistula with those of the same patients, but already diagnosed with persistent cloaca.

Analysis of the literature indicates that there is no evidence of congenital pathology of the urinary system. There is every reason to believe that poor results of operations are due to (1) damage to the puborectalis and pubourethralis muscles, (2) extirpation of the internal anal sphincter, (3) cutting off the levator plates from the rectum, (4) denervation of the pelvic organs, including the internal urethral sphincter.

After total urogenital mobilization (TUM) in infancy even in the best of hands can be followed by serious complications such as urethral stenosis, complete vaginal and anal closure, tight introitus, neurogenic bladder, and urinary

incontinence. Up to 50% of patients may have urinary incontinence or may be dependent on clean intermittent catheterization after cloaca repair [21,22].

Kittur and Vora have done a follow-up study of 1 adolescent and 8 adult female patients born with cloaca, who had rectal pull-through alone in infancy, leaving the urogenital region untouched. Their urogenital problems were treated as they came up in adolescence and adulthood. The common channel after full growth was used as vagina with introitoplasty and dilatation or with surgical intervention for treatable genital anomalies if any.

All patients had an intact urogenital complex and none had documented recurrent urinary tract infections until adolescence. All patients are now beyond adolescence and five got married. Three have reported regular sexual intercourse and one has conceived. Three patients, who are not married, had introitoplasty done for free menstrual flow. They believe that nearing puberty, the introitus should be assessed and thereafter before marriage, a dilatation program can be instituted. Four patients needed introitoplasty beyond 20 years of age. With these measures, the common channel was used as a vagina for regular intercourse.

Only one patient out of nine reported urinary and fecal incontinence. All the others were continent for urine and stool. Although their series is small, they believe that there is a strong case for not subjecting cloaca patients to TUM which has a sizeable potential of developing urinary incontinence. Furthermore, the common channel can be used as vagina along with introitoplasty and dilatation [23]. AbouZeid, as a result of the analysis of long-term results, came to the conclusion that "in some cases of cloaca, it might be better from the functional and cosmetic point of view to perform just introitoplasty while keeping these girls with the common urogenital sinus uncorrected" [24].

Recently, Fuchs al, in a retrospective analysis of ARM patients who were operated on in their institution, found that urologic anomalies occur at a high rate in children with ARM and appear to increase in frequency with increasing complexity of the ARM subtype [25]. This relationship can be due to two reasons.

(1) either complex, i.e., high types of abnormalities, more often combined with congenital urinary tract pathology, (2) or more extensive dissection of the perineum leads to impaired urinary function. The majority of the author, following Peña, claim without any evidence that PSARP is an ideal operation and that all postoperative problems, including urological ones, are due to birth defects. This statement is not true, as it has no scientific evidence. On the contrary, as shown above, when comparing the results of ARM treatment with perineal and vestibular fistulas operated on by different surgical methods, the following situation was found. Authors who used cutback operation or perineal procedure describe good results in anorectal function in 85% and satisfactory in 15% of patients. They did not have bad results, including urological [14,15,16,26].

Conclusion

I believe that the pathophysiology of the so-called persistent cloaca does not live up to the name. According to the pathophysiology, all ARM with vaginal fistula is divided into two types: (1) ARM with a short vaginal fistula (low and high), and (2) ARM with a long vaginal fistula (low and high). It is fundamentally important to establish whether there is an anal canal. For this, you need to create high pressure in the rectum in order to induce a defecation reflex. Sometimes it is enough to apply compression of the abdomen, 1-2 days after birth, or inject a contrast agent through a colostomy. An operation that preserves the anal canal will avoid both anorectal and urological complications. I consider the perforation of the perineum more physiological [27]. The fistula can be closed through the created anus. Drainage of hydrocolpos and vaginal bougienage, as well as further management of patients, is a problem for gynecologists.

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