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Vaginal agenesis and rectovestibular fistula. Experience utilizing distal ileum for the vaginal replacement in these patients, preserving the natural fecal reservoir

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ABSTRACT

Background: The association of rectovestibular fistula (RVF) and vaginal agenesis (VA) presents a diagnostic and management challenge. The vaginal replacement is usually performed with rectum or sigmoid, which are the natural fecal reservoirs; thus, the fecal control could be affected. We present our experience utilizing ileum to preserve the rectum and sigmoid.
Methods: We performed a retrospective study of eight patients with RVF and VA treated from May 2011 to June 2015 at two colorectal centers, at Pittsburgh and Mexico. We recorded the age at diagnosis of VA, treatment, presence of other associated malformations and outcome.
Results: Eight of forty-nine girls with RVF had an associated VA (16.3%). Three patients had a timely diagnosis and five a delayed diagnosis. Six patients were submitted to a vaginal replacement with ileum and achieved fecal control. Two are waiting for surgery.
Conclusions: A high index of suspicion of vaginal agenesis helps in a timely diagnosis in girls with RVF. The use of ileum allows for preservation of the fecal reservoirs, thus optimizing the chance for fecal control in patients with anorectal malformations.

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1. Background

Rectovestibular fistula is one of the most common types of anorectal malformations seen in females. Vaginal agenesis coexists with this anorectal malformation in some patients [1]. The timely diagnosis of gynecological anomalies in girls with rectovestibular fistula is important for creating the proper surgical plan. The reported frequency of vaginal agenesis in the setting of imperforate anus with rectovestibular fistula is 9.5% [2]. Rectovestibular patients in whom the accompanying diagnosis of vaginal agenesis is missed may suffer several problems during and after puberty [3–5].

The posterior and anterior sagittal approaches are the most commonly used techniques to repair imperforate anus with rectovestibular fistula. However, for the patient with vaginal agenesis there are multiple options for vaginal replacement. The standard procedure entails the use of a segment of bowel. The rectum and the sigmoid are the most frequent intestinal segments used. The use of a segment of ileum for the vaginal replacement is also a viable option; the latter allows for preservation of the natural fecal reservoir, but only four such cases have been reported [2,4,6].

The purpose of this report is to share our surgical experience using distal ileum for vaginal replacement in patients with vaginal agenesis and rectovestibular fistula in order to preserve the rectum and sigmoid since they are the natural fecal reservoirs. Also, to convey the importance of having a high index of suspicion of vaginal agenesis in patients with rectovestibular fistula.

2. Methods

We performed a retrospective cross-sectional study approved by our institutional review board (PRO14020665). Data were collected from the database of the senior author and included patients with rectovestibular fistula and vaginal agenesis referred from May 2011 to June 2015 to the Colorectal Center at Children's Hospital of Pittsburgh and the Colorectal Center of Mexico. In these centers we prefer to use ileum for vaginal replacement in these patients in order to offer the opportunity to achieve fecal control since we preserve the natural fecal reservoir.

The recorded variables included age at diagnosis of the rectovestibular fistula, age at diagnosis of vaginal agenesis, age at anorectoplasty, age at vaginal replacement, the approach for the vaginal replacement, the

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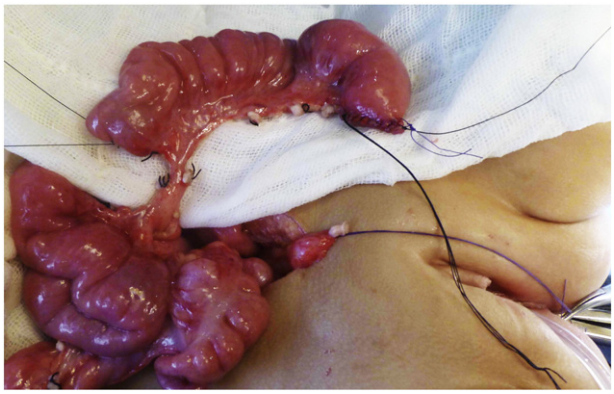

Fig. 1. A segment of distal ileum used for vaginal replacement during a posterior sagittal approach to repair the rectovestibular fistula.

timing of the vaginal replacement related to the PSARP, associated anomalies, sacral ratio, and bowel function.

We have defined for this article "Timely diagnosis of vaginal agenesis" as the diagnosis being made at or about the same time as the diagnosis of the rectovestibular fistula. "Delayed diagnosis" was defined by the vaginal agenesis being diagnosed significantly after the diagnosis of rectovestibular fistula, or diagnosed during or after the anorectoplasty.

The surgical technique employed for the vaginal replacement with ileum in these patients is briefly described. First, we determine the location of the ileum with the longest mesentery. To determine this location, we make traction of the cecum, and at the same time, we are doing proximal traction in the ileum until we observe the longest mesentery [7]. The ileum with the longest mesentery is usually between 15 and 20 cm from the ileocecal valve. Once we found this location, the ileum is transected. To obtain the graft of ileum, we continue dissecting the mesentery to gain length of the ileum usually until we can get 25 cm with adequate irrigation. Then, we cut the bowel again and create a Hartmann pouch in the proximal end of the graft. Then, we perform an isoperistaltic pull-through of the ileum between the bladder and the rectum placing the distal end of the graft in the vulva. During the pull-through, it is important to observe a proper mobilization of the vascular pedicle to prevent the minimal twist or torsion that compromises the viability of the graft (Fig. 1). Extreme care in the division and the management of the vessels allows gaining length preserving the blood supply; during these steps we use vascular bulldog clamps to test the viability of the graft. Then, an end-to-end anastomosis of the ileum is performed, the mesenteric defect is repaired avoiding damage to the vascular pedicle, and the laparotomy is closed. The anastomosis of the ileum to the vulva is completed using 6-0 Vicryl (Ethicon, USA) and the perineal body is repaired.

3. Results

From 2011 to 2015, 369 patients with anorectal malformation were registered in the database. Forty-nine girls had a rectovestibular fistula, eight of these (16.3%) had associated vaginal agenesis; 6 of these 8 patients have undergone a vaginal replacement using a segment of distal ileum.

Three patients had a "timely diagnosis" (Table 1). Two of these patients had a vaginal replacement during the repair of the rectovestibular fistula, through a posterior sagittal approach and a laparotomy using a transverse suprapubic incision (patients 1 and 4); one patient has not been repaired (patient 5).

Five patients had a "delayed diagnosis". Two patients had a vaginal replacement during the repair of the rectovestibular fistula, through the approach described above (patient 2 and 3), two had a vaginal replacement after the PSARP through an anterior perineal approach and laparotomy (patient 6 and 7), and one patient is awaiting the vaginal replacement (patient 8).

Three patients were referred to the colorectal center for bowel management because of pseudo-fecal incontinence. Two of these girls came without the diagnosis of vaginal agenesis and were diagnosed during the work up.

3.1. Associated anomalies

Out of 8 patients, 7 had uterine abnormalities, agenesis or hemiuterus, one patient's uterine anatomy is unknown (patient 8); she was referred for bowel management after the PSARP and the diagnosis of the vaginal agenesis was made at that time. Left renal agenesis was the second most frequent associated defect in 3 patients. The range of the sacral ratios in the anterior–posterior view was between 0.3 and 0.8, and in the lateral view was between 0.6 and 1. None of the patients had tethered cord.

3.2. Outcome

Follow-up is between 4 and 1 years. Five patients with vaginal replacement are older than three years and have voluntary bowel movements using sennosides. One patient is younger than three years and has two bowel movements per day using oral sennosides. All vaginal replacements are viable and patent.

4. Discussion

4.1. Frequency of rectovestibular fistula and vaginal agenesis

Robert Gross described two cases of vaginal atresia among 507 patients with anorectal malformation, but without a description of a surgical repair [8]. Levitt et al. [1] in 1998 reported eight girls with rectovestibular fistula and absent vagina among 1007 patients with anorectal malformations. The only two reported frequencies are 9.5% [2] and 16.3% from our series. These frequencies from pediatric colorectal centers do not reflect the actual incidence. We found in the literature 79 patients with rectovestibular fistula and vaginal agenesis in 23 publications [2–6,9–26], most of these series are case reports (Table 2). Thus, the frequency of rectovestibular fistula with vaginal agenesis is unknown.

Table 1
Age at time of diagnoses, management, and sacral ratio in eight girls with rectovestibular fistula with vaginal agenesis.

Patient	Diagnosis of rectovestibular fistula	Diagnosis of vaginal agenesis	Colostomy	Anorectoplasty	Vaginal replacement with ileum	Sacral Ratio AP/L
1	3 d	3 d	3 d	7 mo	7 mo	0.3/0.6
2	2 d	4 mo	2 d	16 mo	16 mo	0.6/1.0
3	3 d	6 mo	3 d	11 mo	11 mo	0.4/0.7
4	1 d	1 d	11 mo	12 mo	12 mo	0.8/0.8
5	3 mo	3 mo	3 mo	Awaiting	Awaiting	0.3/0.7
6	1 d	3 y	No. primary anorectoplasty	10 d	4 y	0.4/0.7
7	5 mo	7 mo	5 mo	7 mo	11 y	0.5/0.8
8	1 d	6 y	1 d	6 mo	Awaiting	0.7/0.8

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Table 2
Type of vaginal replacement in 87 females with rectovestibular fistula and vaginal agenesis.

Author	Year	Cases	Rectum	Sigmoid	Vagina	Ileum	Skin flaps	Dilatations	Colon pouch	Flaps of peritoneum	No vaginal replacement	Not mentioned
Cohn and Murphy [9]	1956	1	1									
Ein and Stephens [10]	1971	2	2									
Tolete et al. [11]	1989	4	1	1			1					1
Digray et al. [12]	1999	1	1									
Adejuyigbe et al. [3]	2002	1			1							
Sarin and Sinha [4]	2002	4		1		1	1				1	
Tei et al. [13]	2003	2	2									
Günsar et al. [14]	2003	1		1								
Deshpande et al. [15]	2003	1	1									
Patankar et al. [6]	2004	1				1						
Banu et al. [5]	2006	5	1		2			1			1	
Wester et al. [16]	2006	1							1			
Komura et al. [17]	2007	1									1	
Matignas and Delos reyes [18]	2008	1	1									
Chatterjee [19]	2008	2										2
Levitt et al. [1]	2009	26	13	8	3	2						
Wang et al. [20]	2010	3						1		1	1	
Wester et al. [21]	2012	6		3	3							
de Blaauw et al. [22]	2013	2		2								
Kapczuk et al. [23]	2014	1					1					
Kisku et al. [24]	2014	7	2	5								
Teo et al. [25]	2015	1									1	
Pandya et al. [26]	2015	5	1	1	3							
De la Torre et al.	2016	8				6					2	

4.2. Age at diagnosis of the vaginal agenesis

Making the diagnosis of vaginal agenesis with rectovestibular fistula during the neonatal period is of course ideal to allow proper surgical planning [21]. However, it is difficult to achieve such an early diagnosis as we have only two in our series, and just 7 in the 79 reported cases. In the literature six patients were diagnosed between 2 months and 12 months and eighteen between 14 months and 26 years. Five series did not report the age of diagnosis [2,10,19,24,26].

4.3. "Timely or delayed" diagnosis of the vaginal agenesis

Failure to make the diagnosis of vaginal agenesis in the setting of rectovestibular fistula can lead to improper surgical decision-making. Such a delayed diagnosis occurred in 20 patients in the literature; ten during the pull-through and ten after the pull-through. No mention is made of the time of diagnosis in 37 patients [2,10,19,26]. In our series a delay occurred in 5 of 8 patients. Three reasons may explain this delay: 1) the lack of awareness regarding the association between these two entities; 2) imprecise evaluation of the vulva; and 3) the complex anatomy of the area in small patients. The vulva of the newborn may have soft tissue edema, secretions, and meconium from the rectovestibular fistula. These circumstances make the initial inspection problematic, and the surgeon may mistakenly assume the presence of a urethra and vagina. Any uncertainty about the presence of a vagina mandates careful reexamination of the vulva. We have learned that vaginal agenesis may have several different clinical appearances. When there are only two orifices in the corners of the vulva, and in between there is a flat surface the diagnosis is relatively clear (Fig. 2), this clinical picture in our experience was unusual, where there was intricate anatomy of the vulva that resembled a vagina (Fig. 3). We advocate a thorough examination of the vulva to rule out a vaginal agenesis in all patients with rectovestibular fistula. "Timely diagnosis" occurred in only 22 patients of the 79 patients [2–6,9–26] and 3 of 8 in our series.

4.4. Time of vaginal replacement

The confirmation of two orifices in the corners of the vulva, or the absence of a patent vagina strongly suggest the diagnosis of vaginal agenesis. When the anatomy is challenging our approach is to perform a cystoscopy and a vaginoscopy in the operating room. Once we confirm that there is not a patent vagina, we create a diverting colostomy. During the laparotomy, we document the anatomy of the ovaries and uterus. In one of our patients with severe cardiac disease, to avoid surgery we dilated the rectovestibular fistula, and an MRI confirmed the vaginal agenesis. Usually, after six months of age, we proceed with the anorectoplasty and the vaginal replacement with ileum by a posterior sagittal approach and laparotomy. This operation is significantly easier than vaginal replacement in a patient with a previous anorectoplasty. In these latter patients, we perform the vaginal replacement through an anterior perineal approach and a laparotomy, but this procedure is much more laborious and complicated. Other authors suggest similar approaches [2,5,9,16,24,26].

4.5. Type of vaginal replacement and outcome of bowel function

The bowel control in patients with anorectal malformation may be affected depending on the segment of bowel utilized for the vaginal replacement. In 1956 Cohn and Murphy [9] performed the repair of a patient with this association. They did not mobilize the rectum leaving the fistula as the vaginal introitus and the rectum was transected and closed at the peritoneal reflection and left in situ to function as a vagina. The proximal colon was mobilized and used to perform the anorectoplasty. This technique has been the most commonly used; probably because it is easy and reproducible. Twenty-five more patients have been treated with this technique [2,5,10–13,15,18,24,26]. The reported bowel function when the rectum is left in situ as a vagina varies from fecal incontinence to poor fecal control [2,9,10,13,24,26] to voluntary bowel movements [2,12,15]. Three authors did not report on these data [5,11,18]. The rectum is the natural reservoir of stool because it possesses a different anatomy and motility than the proximal segments of the colon. Toilet training and bowel movements should be more normal when the rectum is preserved [2]. We prefer to use the rectum for the anorectoplasty and mobilize a segment of ileum for the vaginal replacement.

In the literature the sigmoid was used to create the vaginal replacement in 22 patients, only three authors report voluntary bowel movements [21,22,24]. When there is a partial vaginal agenesis, a vaginal pull-through is feasible; twelve patients had this procedure [2,3,5,21,26]. One has voluntary bowel movements [3], and the other

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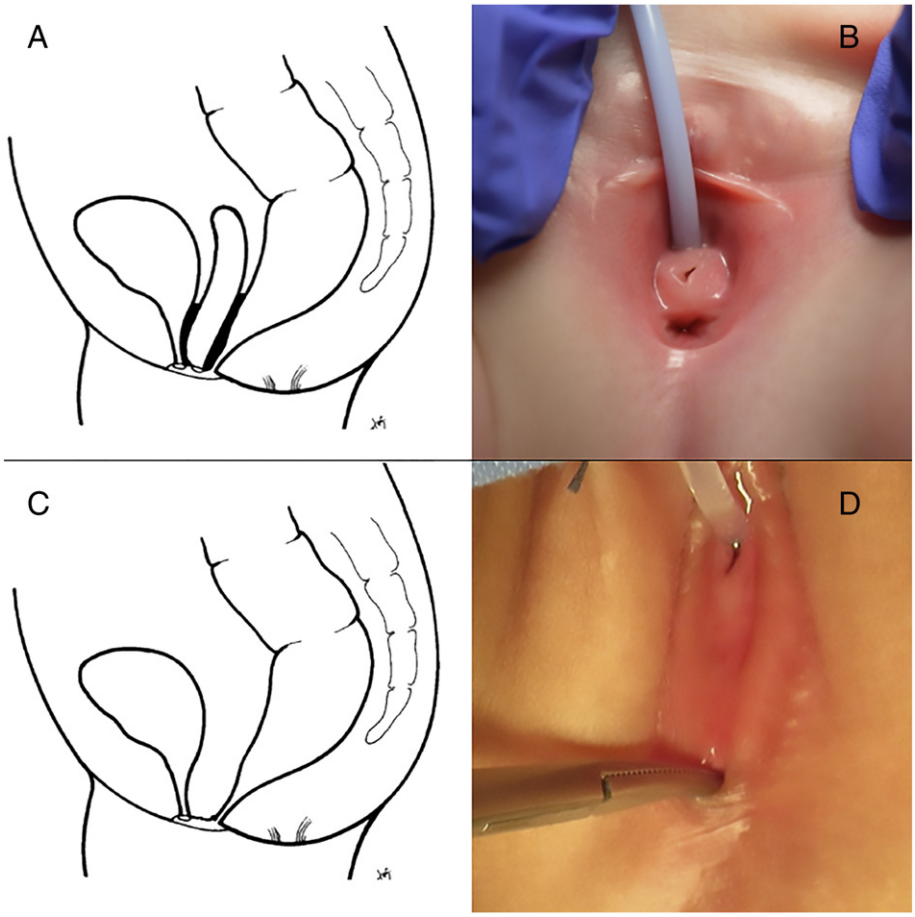

Fig. 2. Comparison of the typical anatomy observed in girls with rectovestibular fistula and the anatomy of this malformation coexisting associated with vaginal agenesis. (A) Illustration of a rectovestibular fistula showing the common wall shared by the posterior wall of the vagina and anterior wall of the rectum. (B) The clinical picture of a rectovestibular fistula. Urethra with a blue Foley, directly below the urethra is the patent vagina, and in the vestibule is the rectovestibular fistula. (C) Illustration of a rectovestibular fistula and vaginal agenesis. (D) The clinical picture of vaginal agenesis coexisting with rectovestibular fistula (forceps). The urethra has a clear Foley, and the vulva shows a smooth flat surface.

authors did not address this issue. Four cases [2,4,6] used the ileum to create the vagina; in three cases the ileum was used because the colostomy location did not allow the sigmoid to be used, and in the other the surgeon decided to use ileum during the anorectoplasty for unclear reasons. In these patients one author reported voluntary bowel function [4], and the others did not report the bowel function. In our series five patients are older than three years and have regular voluntary bowel movements with the use of sennosides, and the sixth patient is two years old and use also sennosides, and has two bowel movements per day. None have signs of fecal incontinence.

Other procedures for vaginal replacement used are skin flaps [4,11,23], dilation of a vaginal pouch [5,20], colon pouch [16] and peritoneum [20]. In three cases the technique for vaginal replacement was not mentioned [11,19] Five cases did not have a vaginal replacement [4,5,17,20,25]. In these cases, the bowel function was not mentioned.

There is not a perfect vaginal replacement technique other than the neo-vagina with tissue engineering. It is true that rectum and sigmoid are the most common segment of bowel used in patients with anorectal malformation. Even in patients with other conditions such as Mayer–Rokitansky–Küster–Hauser syndrome, where a vaginal replacement is performed more with sigmoid than ileum. However, these last patients do not have anorectal malformation and the graft obtained from sigmoid does not affect the fecal continence since they have normal rectum and normal anal canal. That is probably why many surgeons prefer sigmoid since their experience using sigmoid is greater than using ileum [27–30] To perform a vaginal replacement with ileum, surgeons need to realize a delicate, challenging and laborious operation. Our experience using ileum is very satisfactory, and our patients have preserved their natural fecal reservoirs. In the end, always the decision to select rectum, sigmoid, colon or ileum is based on the experience of the surgeon.

4.6. Associated anomalies

Uterine and renal agenesis are the most frequent associated anomalies. It is interesting to note that it is predominantly the left kidney that is absent [5,14,17,18,24].

4.7. Sexual function and complications

Patients with rectovestibular fistula and unrecognized vaginal agenesis may suffer from amenorrhea, cyclic abdominal pain, acute abdomen with hydrometrocolpos or an inability to have sexual intercourse after puberty [3–5]. It is vital to follow patients with a uterus or hemiuterus since they are at risk for these complications, even after vaginal replacement. There are reports of adult patients with unrecognized anorectal malformation and vaginal agenesis that undergo vaginal replacement at a later stage to become sexually active. [22] In our series, the grafts possess perfect viability and patency; we do not have long-term results concerning functionality, but since the neo-vaginas are in good condition, we can expect sexual functionality in the future. The aim is to provide fecal control; 5 of our six patients are at this age and have perfect fecal control. This supports our principles to preserve sigmoid and rectum to offer a better opportunity to our girls.

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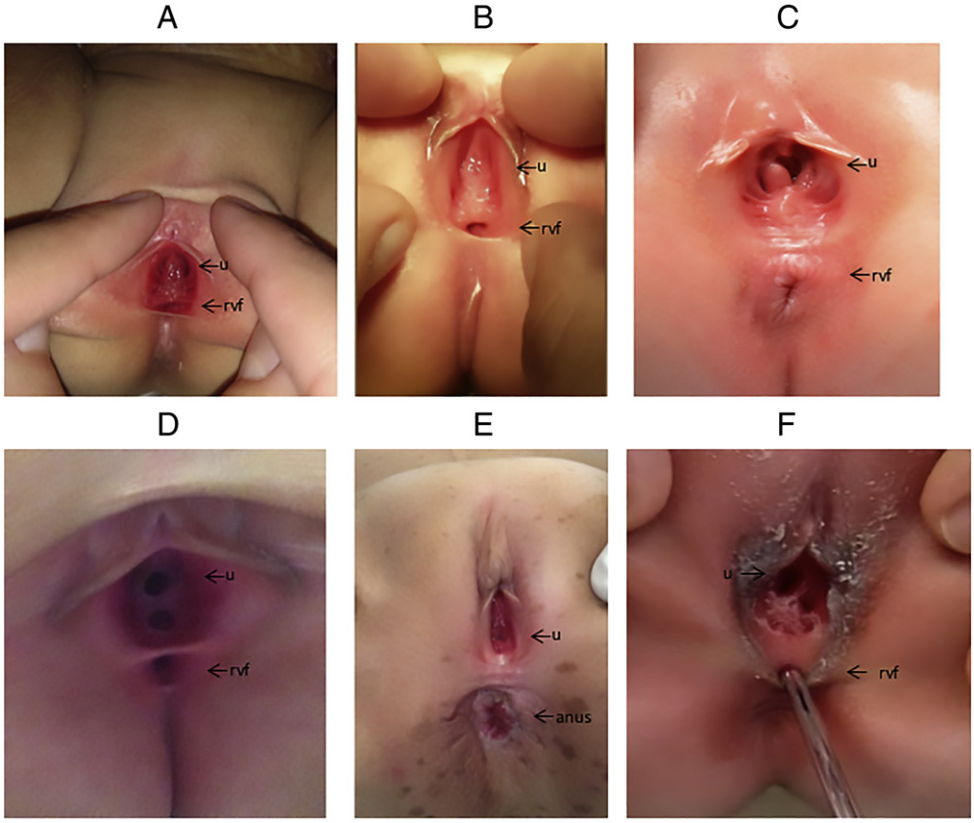

Fig. 3. Clinical pictures of six patients with rectovestibular fistula and vaginal agenesis. These images show the challenging anatomy. (A) Three days old. The urethra is in the left fold. Rectovestibular fistula. (B) Three months old. Urethra, no vagina, and rectovestibular fistula. (C) Eleven months old. Abnormal urethral opening, no patent vagina and rectovestibular fistula. (D) Six months old, the orifice between the urethra and fistula is a blind pouch. (E) Four-year-old patient with neurofibromatosis. Urethra in the vulva and no vagina. This patient had a neonatal anorectoplasty. (F) Newborn, the perineum has two openings. The urethra is the orifice in the upper right part of the vulva. Rectovestibular fistula (Hegar 4). No vagina.

5. Conclusions

1.- Timely diagnosis should be the goal in these babies to plan the surgical procedure. All surgeons must be aware of the existence of this association, and perform a thorough physical exam of the vulva in all patients with rectovestibular fistula.

2.- Vaginal replacements are feasible at any age, but we have learned that it is easier at the time of the anorectoplasty, and that is clearly our preference. We have learned from patients with Müllerian agenesis, that these girls need to develop with normal external genitalia, which is critical for optimal psychological development.

3.- Vaginal replacements when the rectovestibular fistula was already performed are feasible. However, the perineal approach in these girls increases the risk of injury to the urethra and the rectum, since we need to create a vaginal space amidst the scarring between these two structures. This risk of injury is why we recommend correcting the vaginal agenesis in the same procedure as the anorectoplasty.

4.- The use of ileum for a vaginal replacement is our preference since this approach preserves the natural reservoirs. The use of the rectum or sigmoid as a vagina may predispose to fecal incontinence in patients with this association.

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