Rectovestibular Fistula With Absent Vagina: A Unique Anorectal Malformation

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Background/Purpose: A very unusual anorectal defect consisting of a vestibular fistula and absent vagina is presented. Only two previous isolated reports were found in the literature describing a repair that included both abdominal and perineal approach. The authors present another technical alternative that allows the repair via posterior sagittal.

Methods: Of 1,007 patients with anorectal malformations analyzed, eight female cases shared the common anatomic features of a vestibular fistula and absent vagina. What appeared to be the vagina was actually the rectal orifice ending in the vestibule, and the vagina was absent. With the posterior sagittal approach, the distal rectum was used to create the neovagina. The proximal rectum was mobilized and placed within the limits of the sphincter mechanism. Two of the eight patients were born with a patent upper third remnant of the vagina. In one case, the remnant was anastomosed to the neovagina at the time of colostomy closure; in the second one, the anastomosis was performed during the main repair.

Results: Four patients are continent of urine and have voluntary bowel movements. One patient is younger than 3 years and therefore has not been evaluated. One patient has a very poor sacrum and therefore is incontinent. One patient had a primary repair elsewhere and therefore is excluded, and another patient is lost to follow-up. One patient is already sexually active.

Conclusions: It is mandatory to perform a meticulous inspection of the perineum in female patients with anorectal malformations to detect unusual defects. Also, a high index of suspicion is necessary to establish the diagnosis of this defect and avoid an inadequate treatment. The posterior sagittal approach represents another alternative to treat these defects without the need for a laparotomy.
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INDEX WORDS: Anorectal malformation, imperforate anus, absent vagina, vaginal agenesis, vaginal replacement.

WE DESCRIBE a type of anorectal malformation in girls in which two perineal orifices are seen within the vulva. The defect can be misdiagnosed as imperforate anus with rectovestibular fistula or imperforate anus without fistula and with normal urethral and vaginal openings. However, what appears to be the vagina is actually the rectal orifice ending in the vestibule where the vagina would normally be, and the vagina is absent.

It is important for clinicians to be aware of this defect because of the ease of misdiagnosis and potential incorrect management. We have learned that these patients, in general, have good sphincters and sacra and therefore good prognosis. The treatment of this malformation represents a unique challenge, and we use special surgical maneuvers to repair it. The association of anorectal malformations and absent vagina was mentioned previously[1] without description of the treatment used. Two subsequent isolated reports were found in the literature.[2,3] Those investigators proposed a repair that included a laparotomy. Here we describe a similar technique; however, the repair is achieved via posterior sagittal without opening the abdomen.

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MATERIALS AND METHODS

Records of 1,007 patients with anorectal malformations in the senior author's personal series were reviewed. Eight patients shared the anatomic features of imperforate anus, absent vagina, and a rectum ending in the vestibule (Fig 1A). We reviewed the clinical and radiological records of the patients and studied their sacra, perineal muscles, gynecologic anatomy, associated anomalies, and functional results regarding fecal and urinary continence.

Anatomic Characteristics

These patients had imperforate anus with a prominent midline groove and a noticeable anal dimple, consistent with good clinical prognosis.[4,5] The external appearance of the genitalia seemed normal. A more meticulous inspection showed that immediately behind a normal urethra is a rectal opening occupying the place in which the vagina is normally found (Fig 2). The rectum and urethra are intimately attached because they share a common wall. All patients had normal ovaries. In six

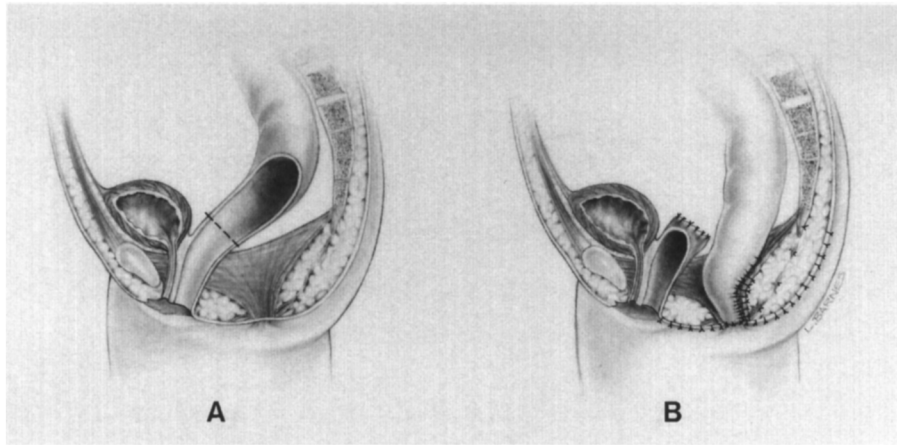

Fig 1. (A) Diagram shows a sagittal view of the defect. (B) The sagittal view of the surgery used to repair this defect.

patients the fallopian tubes gradually became cordlike structures as they approached the midline (Fig 3), and the uterus and vagina were completely absent. In two cases the fallopian tubes connected to a uterus that connected to a vaginal remnant representing approximately its upper third (Fig 4).

A colostomy had been opened at birth in all eight patients at other institutions. The patients were referred to our institution for definitive repair of the anorectal anomaly. In six patients we agreed with the original diagnosis of rectovestibular fistula, but at the time of the main repair we could not find a vagina. In the last two patients the diagnosis of imperforate anus with absent vagina was made preoperatively.

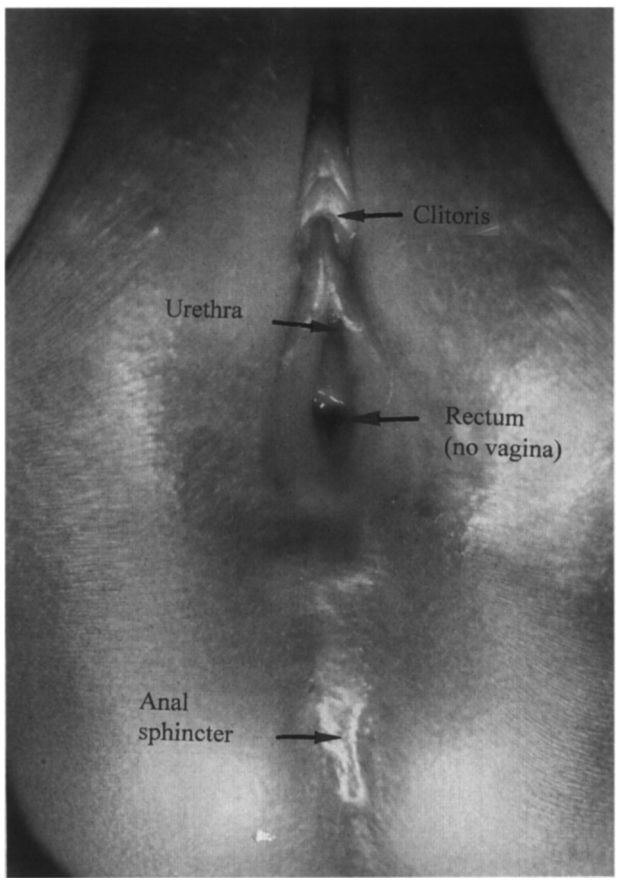

Fig 2. Photograph of the perineum of one of our patients. The anterior orifice is the urethra, and the posterior orifice is the rectum.

Surgical Technique (Main Repair)

All patients were approached posterior sagittally. We divided the entire sphincteric mechanism in the midline until the posterior wall of the rectum was identified. The rectum was dissected in a circumferential manner about 8 cm proximal to the opening at the genitalia, and it was then divided at that same level. The distal end of the rectum was left in place as a neovagina and its dome was closed in two layers of interrupted long-term absorbable sutures (Fig 1B). Its blood supply remained excellent and we assumed that was provided through the neighboring structures.

Multiple temporary silk sutures were placed circumferentially in the distal edge of the proximal rectum. This maneuver allows the surgeon to handle and exert uniform traction on the rectum, which facilitates its dissection. While applying traction to the rectum, a circumferential dissection was performed, dividing and ligating the bands and vessels that hold the rectum in the pelvis. The dissection was continued until the rectum reached the perineum without tension. An anoplasty was then performed within the limits of the sphincter mechanism.

The colostomy is closed approximately 2 months after the main repair. During the colostomy closure the surgeon must inspect the internal genitalia. If the patient has a remnant of vagina and uterus (as was the case in two patients, Fig 4A), the neovagina (distal rectum) must be anastomosed to the upper third (remnant) of vagina (Fig 4B). For this, a Hegar dilator is introduced through the neovagina to show its dome behind the bladder inside the abdomen. Traction sutures are placed in that dome and an end-to-end anastomosis is performed between the upper third remnant of the original vagina and the neovagina. This anastomosis was performed with interrupted 5-0 long-term absorbable sutures.

All patients underwent repair with the posterior sagittal approach, and all underwent vaginal reconstruction using the distal rectum as neovagina. Two patients underwent anastomosis of the upper third of their native vagina to the neovagina. In both cases, this anastomosis was performed through a laparotomy; in one case it was performed at the time of colostomy closure, and in the remaining case it was performed at the time of the main repair. In that specific case the abdomen had to be open to repair other associated urologic problems.

Two patients had very dysplastic sacra, and four had relatively normal sacra. Sacral radiographs were not available for two patients. The sacrum was evaluated using a sacral ratio developed at our institution.[4] The sacral ratio in anterior-posterior view (AP film) is determined by tracing a line from the uppermost portion of one iliac

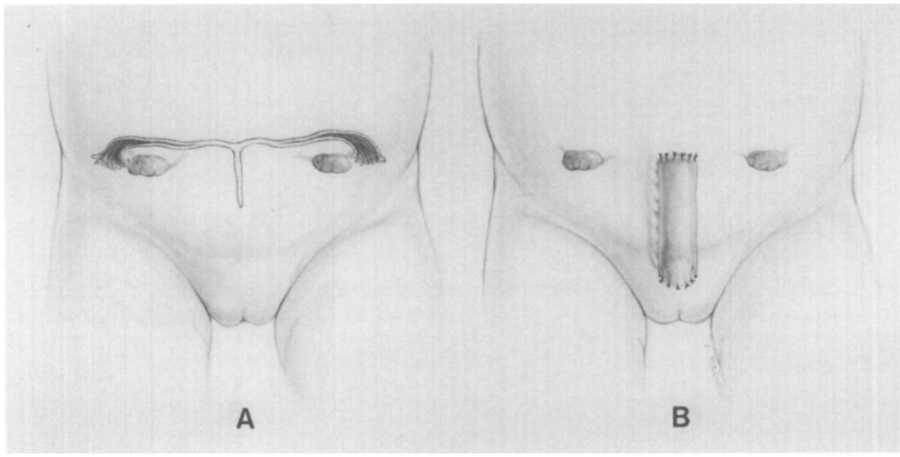

Fig 3. (A) Diagram shows the internal anatomy of six of the patients. The fallopian tubes became cordlike as they approached the midline, and the uterus and vagina were completely absent. (B) After the repair, the distal rectum becomes the neovagina.

crest to the other. A second line is drawn across between the lowest point of each sacroiliac joint (posterior and inferior iliac spine). A third line runs parallel to the other two and touches the lower most radiologically visible point of the sacrum. A ratio is obtained by dividing the distance between the two lower lines by the distance between the two upper lines. In 100 normal children at the author's institution, the average ratio was 0.74; children with anorectal malformations show a spectrum of lower values. Lower ratios represent different degrees of sacral hypodevelopment and are associated with defects that have a poor functional outcome. The average sacral ratio for the group of patients of this presentation was 0.60, which is similar to the ratios obtained for patients born with persistent cloaca.[4] However, if the two patients with abnormal sacra are excluded, the average ratio is 0.76, a ratio similar to that in patients with rectoperineal fistula or normal individuals. The perineal muscles were well developed but were slightly less developed than those found in patients with rectovestibular fistula. Five of the eight patients had an absent kidney.

RESULTS

Four patients are continent of urine and have voluntary bowel movements. One patient is under 3 years of age and is therefore not old enough to determine a clinical result. One patient, who also has a very poor sacrum (lateral sacral ratio, 0.19), receives enemas and intermittent catheterization. This patient has also required bladder augmentation and ureteral reimplantation. One patient was excluded from this analysis because she underwent her original repair elsewhere and required reoperation at our institution, and one patient has been lost to follow-up.

Two of our patients have reached puberty. One belongs to the group with no uterus. She reports having sexual intercourse but is unable to experience orgasm. The other patient has a uterus, is not sexually active, but is menstruating normally through the neovagina. The rest of our patients have not reached puberty.

DISCUSSION

The eight patients we have described have a relatively rare anorectal defect. During the period in which we saw these eight cases, we also saw more than 100 girls with rectovestibular fistula and more than 200 patients with persistent cloaca.

Vaginal atresia was only mentioned in two cases among 507 with anorectal malformations by Gross in 1953,[1] without any description of the surgical repair.

We found two previous reports related to the surgical repair of this defect. Cohn and Murphy[2] in 1954 operated via abdominoperineal on a 7-year-old patient who had the same defect. Subsequently, Ein and Stephens[3] published

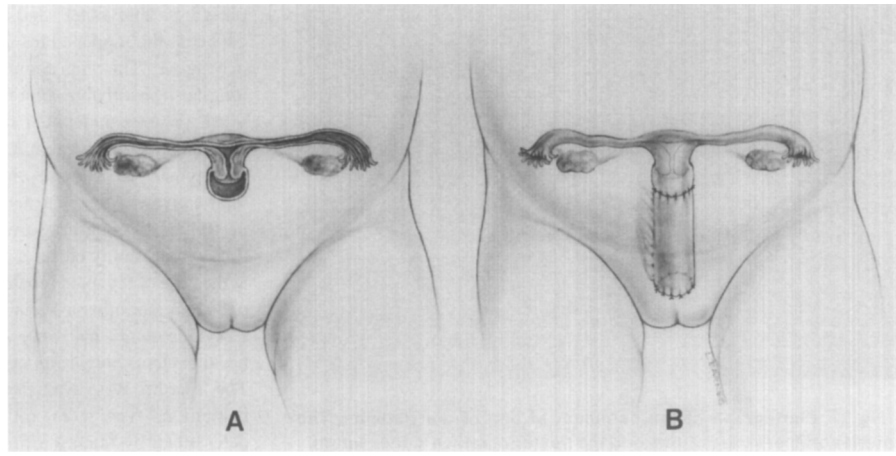

Fig 4. (A) Internal anatomy of two of the patients. The fallopian tubes connected to a uterus, which connected to a vaginal remnant (approximately its upper third). (B) After the repair, the distal rectum (neovagina) is anastomosed to the upper third of the patients' original vagina.

in 1971 their experience with two patients also operated on with an abdominoperineal approach, preserving the lower rectum as a neo-vagina as shown in this paper. The neovagina was closed at the level of the peritoneal reflection because the investigators did not find any viable upper vagina with which to connect. The basic therapeutic principles used by those investigators were followed by us but without opening the abdomen; the repair was performed only via posterior sagittal.

It is imperative that clinicians be aware of the possibility of imperforate anus with absent vagina. Externally, the patients look like typical girls with imperforate anus, either with a rectovestibular fistula or with no fistula. At birth if the patient passes meconium, the surgeon may assume that the patient has a rectovestibular fistula and will open a colostomy. This assumption is understandable because rectovestibular fistula is by far the most frequent defect seen in girls.[4-6] The labia are often swollen in the newborn period and it is difficult to delineate precisely the anatomy of the female genitalia. At the time of the main repair, the surgeon will be puzzled when finding that there is no vagina.

Another possible scenario is a patient who does not pass meconium in the first 24 hours of life. The diagnosis of imperforate anus without fistula is presumed, and a colostomy is opened. At the definitive repair the surgeon may be unable to find the rectum because of the incorrect assumption that the structure located behind the urethra is the vagina.

A distal colostogram probably could help to suspect this malformation. However, this study will show a compressed lower rectum by the sphincter mechanism as seen in cases of rectovestibular fistulas. The rectum cannot be distended by contrast material because this leaks through the lower opening, which prevents an increased hydrostatic pressure.

At the time of the main repair, the surgeon who sees a patient with this type of malformation has two surgical alternatives. The rectum can be separated from the urethra and moved back within the limits of the sphincter mechanism. In that case, some technique of vaginal replacement must be implemented, either during the main repair or later in life.

A second option, which we have used in these cases, is to leave part of the lower rectum in situ, attached to the urethra. This piece of rectum is used as a neovagina, and the upper rectum is pulled down to be placed within the limits of the sphincter mechanism.

The major question is which of the two techniques will give better functional results. In favor of the first approach, one can argue that there is inherent value in using the distal most rectum as rectum. The fact that it has a pectinate line may mean that it has functional characteristics necessary for bowel control, such as sensation and reservoir function. The major disadvantage of this method is the need for vaginal reconstruction, which if performed, should be done by using one of the described techniques.[7,8] In addition, separation of the rectum from the urethra may provoke denervation of the lower urinary tract as well as denervation of the rectum.

The approach we used in this group of patients moves a more proximal portion of rectum posteriorly, leaving the distal most rectum to become the neovagina. This maneuver is relatively simple. It can be performed with a posterior sagittal approach without a laparotomy and avoids the need for vaginal reconstruction.

It is important at the time of colostomy closure to look for an upper third of vagina in the pelvis as we found in two of our patients. The neovagina should be anastomosed to this vaginal remnant to restore continuity between the internal genitalia (uterus and fallopian tubes) and neovagina.

Preliminary results regarding fecal and urinary continence indicate that our patients with good sacra have good functional results. Currently, we know very little about the sexual function and fertility potential of these patients. We do have evidence that the neovagina functions as a vagina in the one patient old enough to be sexually active and in another to allow normal menstruation.

We found a high incidence of urologic anomalies. We know that absent kidney is very common in patients with anorectal malformations.[9] Also, patients without anorectal malformations but with absent vagina have been found to have a high incidence of absent kidney.[10]

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Discussion

M.A. Levitt (response): The interesting thing about this group of patients is the first six were diagnosed as the typical rectovestibular fistula. Colostomies were opened and then they were referred for definitive repair. And the novel approach, which you just mentioned, was to leave the distal rectum in situ, which of course prevents the problems of denervating the urethra and the distal rectum.

S.H. Ein (Toronto, Ontario): I rise with some hesitation, but I must tell you about my experience with this problem. When I was a resident, the late Dr Stevens and I had two similar cases in Toronto that I wrote up and presented as a first. I was subsequently embarrassed to find out that the late Burt Cohn, who was a pediatric surgeon in Brooklyn, had reported a similar case from the Montreal Children's Hospital in the 1940s. So it is *deja vu.* But it is still a novel way to treat a very difficult problem.