ACOG committee opinion

Nonsurgical Diagnosis and Management of Vaginal Agenesis

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ABSTRACT: Vaginal agenesis occurs once in every 4,000–10,000 females. The most common cause of vaginal agenesis is congenital absence of the uterus and vagina, which is also referred to as müllerian aplasia, müllerian agenesis, or Mayer–Rokitansky–Küster–Hauser syndrome. The condition usually can be managed nonsurgically with the use of successive dilators if it is correctly diagnosed and the patient is sufficiently motivated. Besides correct diagnosis, effective management also includes evaluation for associated congenital, renal, or other anomalies and careful psychologic preparation of the patient before any treatment or intervention. If surgery is preferred, a number of approaches are available; the most common is the Abbe-McIndoe operation.

Vaginal agenesis is an uncommon, but not rare, condition. Given an incidence ranging from 1 per 4,000 to 1 per 10,000 females (1), vaginal agenesis is a condition that general gynecologists will encounter once or twice during their professional careers. The most common etiology of vaginal agenesis is congenital absence of the uterus and vagina, which also is referred to as müllerian aplasia, müllerian agenesis, or Mayer–Rokitansky–Küster–Hauser syndrome. The term müllerian aplasia will be used to describe this congenital reproductive anomaly throughout this document. Müllerian aplasia is caused by embryologic failure of the müllerian duct with resultant anomalies in the müllerian structures. The ovaries, given their separate embryologic source, are normal in structure and function.

To manage vaginal agenesis effectively in young women, correct diagnosis of the underlying condition is important. Evaluation for associated congenital, renal, or other anomalies also is essential. Both diagnosis and evaluation usually can be completed without surgery. Careful patient counseling should be provided before any treatment or intervention. Nonsurgical creation of the neovagina should be the first-line approach. These recommendations, as well as surgical approaches for use in the rare situations when nonsurgical approaches are unsuccessful, are discussed.
Differential Diagnosis

Patients with müllerian aplasia have a normal 46,XX karyotype, normal female phenotype, and normal ovarian hormonal and oocyte function. Puberty and development of secondary sexual characteristics progress normally except that menarche does not occur. Therefore, patients with müllerian aplasia typically present in adolescence with primary amenorrhea. Müllerian aplasia is the second most common etiology of primary amenorrhea, next to gonadal dysgenesis (2).

On physical examination, patients with müllerian aplasia have normal breast development, normal secondary sexual body proportions, body hair, and hymenal tissue. A vagina is absent unless it has been created by sexual encounters. Differential diagnosis of vaginal agenesis includes congenital absence of the uterus and vagina, androgen insensitivity (absence or alteration of androgen-receptor function), a low-lying transverse vaginal septum, and imperforate hymen.

In androgen insensitivity, the gonads are testes producing normal androgens in karyotypic 46,XY individuals. The lack of androgen tissue receptor results in sparse or no pubic and axillary hair. Patients with androgen insensitivity typically have normal breast development at puberty due to peripheral conversion of circulating androgens to estrogen; however, no internal female organs exist. In pubertal females, the differential diagnosis between androgen insensitivity and müllerian aplasia is easily made by assessing serum testosterone levels. A testosterone level in the pubertal male range confirms the diagnosis of androgen insensitivity.

In postpubertal patients, the presence of functioning ovarian tissue seen on pelvic ultrasound examination may serve as a secondary confirmation of the diagnosis of müllerian aplasia, excluding the diagnosis of androgen insensitivity. Chromosomal studies, which are significantly more costly than ultrasonography, are required only in prepubertal children who do not yet produce gonadal steroids.

The differential diagnosis of vaginal agenesis also includes the diagnosis of imperforate hymen and transverse vaginal septum. Patients with these latter conditions will have a normal cervix and fundus, both of which are palpable on rectal examination. In contrast to most patients with müllerian aplasia, the patient with an imperforate hymen will not have the typical fringe of hymenal tissue. The patient with a low transverse vaginal septum will have a normal hymen, like the patient with müllerian aplasia. Ultrasonography can be used to define the müllerian structures in cases where palpation is unrevealing.

Correct diagnosis of the underlying condition affecting the genital anatomy is crucial before any surgical intervention. If the patient is operated on for an incorrect diagnosis, it can be extremely difficult to create a vagina without surgery because of scar tissue.

Evaluation of the Patient with Müllerian Aplasia

Most patients with müllerian aplasia have small rudimentary müllerian bulbs without any endometrial activity. In 2–7% of patients with müllerian aplasia, active endometrium is found in these uterine structures (1). These patients will present with cyclic abdominal pain. Magnetic resonance imaging has been suggested to assess the internal genitalia, although it is rarely needed in the initial evaluation unless ultrasound evaluation for the presence of functional endometrium in a müllerian structure is equivocal (3). Although laparoscopy is not necessary to diagnose müllerian aplasia, it may be useful in the evaluation of patients with cyclic abdominal pain to exclude the possibility of endometrial activity in müllerian structures (4).

Patients with müllerian aplasia often have concomitant congenital malformations, especially of the urinary tract and skeleton, and should be evaluated for these anomalies. Ultrasonography can be used to screen for the more common findings of renal agenesis or a pelvic kidney. This evaluation can be performed during the study of ovarian and müllerian structures. The implications of ureteral duplication in the case of later abdominal or pelvic surgery can be discussed or intravenous pyelography can be used to exclude this possibility.

After the diagnosis of müllerian aplasia, the adolescent should be offered psychologic support to help her adjust. Counseling should emphasize that a normal sex life will be possible after a neovagina has been created. Ultimately, however, infertility will be the most difficult aspect of this disorder for the patient to accept. Discussion of assisted reproductive techniques and use of surrogacy is appropriate. Specifically, it is important to explain that eggs can be harvested from patients with müllerian aplasia. This allows young teens to understand their repro-
ductive potential for becoming a biologic parent and may help them accept the diagnosis and its implications. Referral to a mental health professional may be necessary for some patients.

The best predictor of good emotional outcome after diagnosis and vaginoplasty is a good relationship between the patient and her parents and the ability to show feelings with family and friends (5). Contact with a support group or young women with the same diagnosis may be helpful (4). (More information is available at http://www.mrkh.org.)

Patients should be given a brief, written medical summary of their condition, including a summary of concomitant malformations. This information may be useful if the patient requires urgent medical care or emergency surgery from a provider unfamiliar with müllerian aplasia.

**Nonsurgical Creation of a Neovagina**

Timing for nonsurgical or surgical creation of a neovagina is elective; however, it is best planned when the patient is emotionally mature. Nonsurgical creation of the vagina is the appropriate first-line approach in the majority of patients because it is the least morbid procedure. In a recently reported series of patients with müllerian aplasia, more than 90% were able to achieve anatomic and functional success by vaginal dilation (6).

Patients are asked to place successive dilators on the perineal dimple for 30 minutes–2 hours a day. Sitting on a race bicycle seat stool provides the perineal pressure and allows the patient to participate in simultaneous productive activities, such as doing homework or practicing a musical instrument (7). If sitting on the bicycle seat stool is too uncomfortable, the patient may have better success using dilators while reclining on a bed after a bath while still wet from bathing. Use of dilators in the management of vaginal agenesis is appropriate and successful in the majority of patients. Mature, highly motivated patients who wish to avoid surgery and are aware that it will take several months to achieve their goal are likely to be successful (7, 8). Because the nonoperative approach is noninvasive and often successful, it is strongly recommended as first-line therapy.

Clinicians often use “buddies,” other patients with vaginal agenesis who have successfully dilated, as support to the young woman attempting dilation. Young married patients make excellent buddies. If fertility issues are a major concern to the patient or her family, it may be helpful to find a buddy who has used assisted reproductive techniques to become a mother.

**Surgical Creation of a Neovagina**

Surgery becomes an option in patients unsuccessful with dilators or if the patient prefers surgery after a thorough discussion of the risks and benefits of the procedure and the available nonsurgical alternatives. The aim of surgery is the creation of a vaginal canal in the correct axis of adequate size and secretory capacity to allow intercourse to occur without the need for continued postoperative dilation. The timing of the surgery depends on the patient and the type or procedure planned. Surgeries often are performed in late adolescence when the patient is more mature and better able to comply with postoperative dilation or instructions. Surgery usually is scheduled during summer vacation to allow for an adequate recovery time without missing school while limiting questions from peers (4, 9).

A number of operations are appropriate for the correction of vaginal agenesis. The approach usually is based on the experience of the operating surgeon. Pediatric surgeons are more likely to use bowel segments for the creation of a neovagina; gynecologists are more likely to use a perineal approach. Whatever technique is chosen, the surgeon must be experienced with the procedure, because the initial surgery is more likely to succeed than are follow-up procedures. Reoperation in these cases increases the chance of operative injury to surrounding tissues and the possibility of a poor functional outcome. At present, there is no consensus in the literature regarding the best option for surgical correction (10).

The most commonly used surgical procedure employed by U.S. gynecologists to create a neovagina is the Abbe-McIndoe operation. This involves the dissection of a space between the rectum and bladder, placement of a mold covered with a split thickness skin graft into the space, and the diligent use of vaginal dilation postoperatively. Postoperative dilation must be continued to prevent significant skin graft contracture. This surgery is inappropriate if the patient rejects the nonsurgical technique because she has concerns about or objections to dilation. If postoperative dilation is not done, the patient will have a nonfunctional vagina.
Newer procedures for the creation of the neovagina are the Vecchietti procedure and laparoscopic modifications of operations previously performed by laparotomy. The Vecchietti procedure involves the creation of a neovagina via dilation with a traction device attached to the abdomen, sutures placed subperitoneally via laparotomy, and a plastic “olive” placed on the vaginal dimple. In the laparoscopic modification, traction sutures are placed laparoscopically. The two techniques are comparable in terms of producing a functional neovagina (11).

Davydov developed a three-stage operation involving dissection of the rectovesical space with abdominal mobilization of the peritoneum with creation of the vaginal fornices and attachment of the peritoneum to the introitus. The newer adaptation involves dissection of the rectovaginal space, with mobilization of the peritoneum from below and laparoscopic assistance from above. This is followed by closure of the abdominal end of the neovagina with a laparoscopically placed pursestring suture (12–14).

Summary
The most important steps in the effective management of vaginal agenesis are correct diagnosis of the underlying condition; evaluation for associated congenital, renal, or other anomalies; and careful psychologic preparation of the patient before any treatment or intervention. If any of these are neglected, the success of the intervention will be compromised.

Laparoscopy is seldom required to make the diagnosis, but may be appropriate in the rare patient presenting with cyclic pelvic pain. Nonsurgical creation of the neovagina should be the first-line approach. In cases where surgical intervention is required, referrals to centers with expertise in this area should be considered. Few surgeons have extensive experience in construction of the neovagina, and the initial surgery has the greatest chance for success. In addition, experts at these centers may be more successful in promoting the nonsurgical approach, given their experience.

References