The diagnosis and management of girls and young women with Müllerian anomalies requires not only knowledge of embryologic development, but an awareness of the known associations of structural anomalies of the female reproductive tract with other congenital anomalies, including renal anomalies and anorectal malformations. Recognition of such associations appropriately guides the diagnostic evaluation and allows potential simultaneous operative treatment. Because reproductive anomalies are not rare, most clinicians will encounter these abnormalities within routine gynecologic practice and an organized discussion is advantageous for appropriate diagnosis, management, and possible referral. Familiarity with the anomalies, associations and optimal treatment allows providers to offer the recommended clinical care in a timely way, avoiding unnecessary delays and potential compromise to reproductive success.

EMBRYOLOGY

The reproductive organs in the female consist of external genitalia, gonads, and an internal duct system, the Müllerian ducts. These three components originate embryologically from different primordia and in close association with the urinary system and hindgut. Thus, the developmental history is quite complex. The Müllerian (paramesonephric) duct system is stimulated to develop preferentially over the Wolffian (mesonephric) duct system, which regresses in early female fetal life. The cranial parts of the Wolffian ducts can persist as the epoöphoron of the ovarian hilum; the caudal parts can persist as Gartner’s ducts. The Müllerian ducts persist and, with complete development, form the fallopian tubes, the uterine corpus and cervix, and a portion of the vagina.

About 37 days after fertilization, the Müllerian ducts first appear lateral to each Wolffian duct as invaginations of the dorsal coelomic epithelium. The site of origin of the invaginations remains open and ultimately forms the fimbriated ends of the fallopian

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**KEYWORDS**

- Müllerian anomalies
- Utero-vaginal anomalies
- Vaginoplasty
- Vaginal reconstruction
- Vaginal anomalies
tubes. At their point of origin, each of the Müllerian ducts forms a solid bud. As the solid buds elongate, a lumen appears in the cranial part, beginning at each coelomic opening. The lumina extend gradually to the caudal growing tips of the ducts.

The paired Müllerian ducts continue to grow in a medial and caudal direction until they eventually meet in the midline and become fused together in the urogenital septum. A septum between the two Müllerian ducts gradually disappears, leaving a single uterovaginal canal lined with cuboidal epithelium. Failure of reabsorption of this septum can result in a septate uterus. The most cranial parts of the Müllerian ducts remain separate and form the fallopian tubes. The caudal segments of the Müllerian ducts fuse to form the uterus and part of the vagina. The cranial point of fusion is the site of the future fundus of the uterus. Variations in this site of fusion can result in an arcuate or bicornuate uterus. Complete failure of fusion can result in a didelphic uterus. Although isolated case reports continue to challenge established embryologic mechanisms of Müllerian development, additional reports are necessary to fully evaluate potential variations in embryologic development.

The vagina is formed from the lower end of the uterovaginal canal, which developed from the Müllerian ducts and the urogenital sinus. The point of contact between the two is the Müllerian tubercle. A solid vaginal cord results from proliferation of the cells at the caudal tip of the fused Müllerian ducts. The cord gradually elongates to meet the bilateral endodermal evaginations (sinovaginal bulbs) from the posterior aspect of the urogenital sinus below. These sinovaginal bulbs extend cranially to fuse with the caudal end of the vaginal cord, forming the vaginal plate. Subsequent canalization of the vaginal cord occurs, followed by epithelialization with cells derived mostly from endoderm of the urogenital sinus. Most investigators suggest that the vagina develops under the influence of the Müllerian ducts and estrogenic stimulation. There is general agreement that the vagina is a composite formed partly from the Müllerian ducts and partly from the urogenital sinus.

At about the 20th week, the cervix takes form as a result of condensation of stromal cells at a specific site around the fused Müllerian ducts. The mesenchyme surrounding the Müllerian ducts becomes condensed early in embryonic development and eventually forms the musculature of the female genital tract. The hymen is the embryologic septum between the sinovaginal bulbs above and the urogenital sinus proper below. It is lined by an internal layer of vaginal epithelium and an external layer of epithelium derived from the urogenital sinus (both of endodermal origin), with mesoderm between the two. It is not derived from the Müllerian ducts.

CONGENITAL HYMENAL ABNORMALITIES

The normal hymen can have multiple configurations including annular, crescentic, and fimbriated/redundant. However, failure of the hymen to perforate completely in the perinatal period can result in varying anomalies including imperforate, microperforate, cribiform (sievelike), navicular (boatlike), or septated (Fig. 1). Such anomalies are ideally recognized at birth as part of the newborn examination or seen in childhood as part of well-child genital examinations. An abnormal hymen that results in a small orifice may preclude the use of tampons, insertion of vaginal cream or suppositories, or the ability to have vaginal intercourse. Providers should be aware that there have been reports of familial occurrences of hymenal abnormalities, and thus alert young women that their daughters may have a similar abnormality.1

Imperforate hymen (Fig. 2) has an incidence of 1 in 1000 and may present in the neonatal period as hydrocolpos or mucocolpos, resulting from stimulation of the vaginal epithelium. Significant amounts of mucous can be secreted during the
perinatal period secondary to maternal estradiol stimulation. The newborn may have a bulging, translucent yellow-gray mass at the vaginal introitus. Most hydro/mucocolpos is asymptomatic and resolves as the mucous is reabsorbed and estrogen levels decrease. However, large hydro/mucocolpos may obstruct the ureters, resulting in hydronephrosis or even respiratory distress. Neonatal ultrasound

Fig. 1. Septate hymen in menarchal female.

Fig. 2. Imperforate hymen. Notice the thin, transparent hymen stretched over the dark-colored accumulated menstrual blood.
can demonstrate the fluid collection. Resection of the hymen is recommended in the symptomatic infant. Aspiration, without definitive enlargement or a vaginostomy for continued drainage, should be avoided owing to the risks of reaccumulation with recurrence of a mass or ascending infection. Asymptomatic girls with an imperforate hymen can be monitored throughout childhood and avoid the risks of surgery in infancy. The optimal time for surgery is after the onset of puberty (as evidenced by thelarche) but before menarche. The hymen should then be resected to prevent the development of hematocolpos, pain, and possible retrograde menstruation. Pubertal timing allows adequate resection with less risk of scarring and a decrease in the potential need for repeat procedures, given the presence of adequate on-going endogenous estrogen. In the unestrogenized state of childhood it may be difficult to differentiate vaginal agenesis and an imperforate hymen.

Unfortunately, the most common presentation of imperforate hymen is in a pubertal girl with cyclic or persistent pelvic pain and an abdominal mass or perineal bulging with a translucent bluish-tinged hymen secondary to significant hematocolpos, and in severe cases, additional hematometra. Less commonly, mass-effect symptoms including urinary retention or constipation are the patient complaints leading to evaluation. Hematocolpos may become quite large because the vagina is distensible and the obstruction is so distal.

Definitive surgery should take place only after the appropriate evaluation. This should always include an examination of the external genitalia, and a digital rectal examination, if indicated, with radiographic imaging only as necessary to confirm the diagnosis. Pelvic and abdominal ultrasonography will confirm a distal obstruction. The procedure is usually performed under general anesthesia with the patient in high lithotomy. After the usual sterile preparation and draping, a straight or Foley catheter is used to drain the bladder and properly delineate the urethra. Stay sutures can then be placed to provide ample traction for the hymenotomy and allow stabilization for quick insertion of a suction device to collect the copious amount of thickened, chocolate-colored old blood and menstrual fluid. After evacuation of the hematocolpos, the hymenal orifice can be enlarged with removal of excess tissue to create an orifice of “normal” size. In treatment of the imperforate hymen, puncture without definitive surgical repair should be avoided, because the viscous fluid may not drain adequately and the small perforations will allow ascension of bacteria and the possibility of infection, such as pelvic inflammatory disease or tuboovarian abscess.

Other hymenal anomalies with some degree of perforation are rarely clinically significant in childhood. A navicular configuration may be associated with postvoid dribbling and complaints of incontinence or, in rare circumstances, recurrent urinary tract infections. Most young women present after menarche with complaints of difficulty inserting or removing a tampon or, less commonly, with significant dyspareunia with penetration (Fig. 3). These are also usually surgically corrected with a simple outpatient excision.

**ANOMALIES OF THE UTERUS, CERVIX, AND VAGINA**

Anomalies of the female reproductive tract may result from one of several developmental abnormalities, including agenesis/hypoplasia, vertical fusion (canalization abnormalities resulting from abnormal contact with the urogenital sinus), lateral fusion (duplication), or resorption (septum). Clinicians should be cognizant that patients with each abnormality will present with different symptoms, and will require specific
individualized therapy. Common symptoms seen in uterine and vaginal malformations include primary amenorrhea, acute and/or chronic pelvic pain, abnormal vaginal bleeding, or a foul-smelling vaginal discharge (often worse at the time of menses). Non-obstructive anomalies may even be found incidentally on routine examination. Young women with obstructive anomalies are more likely to present with acute symptoms of pain and discomfort.

**Genetics**

The etiology of anatomic defects of the female genital tract is not fully understood. Most forms of isolated Müllerian duct and urogenital sinus malformations are inherited in a polygenic/multifactorial fashion. Mendelian forms of inheritance with a single gene mutation have been described to explain specific syndromes including the McKusick-Lauffman syndrome (MKS), which uncommonly includes vaginal atresia and more commonly includes transverse vaginal septa and the hand-foot-genital syndrome.¹ Hand-foot-genital syndrome includes bilateral great toe and thumb hypoplasia and small hymenal opening with various degrees of incomplete Müllerian fusion with or without two cervices or a longitudinal vaginal septum.

**Incidence**

The actual incidence of Müllerian anomalies is not definitively known. Reporting issues skew our knowledge of the true incidence. These abnormalities are relatively underreported in infants and are likely overreported in patients being evaluated for reproductive concerns, especially in adult women struggling with infertility.³⁻⁶ In a study of fertile women who were evaluated for Müllerian duct anomalies at the time of tubal ligation, an incidence of 3.2% was identified.⁴ Many young women may have an underlying asymptomatic Müllerian duct anomaly; yet, since they have no pain, pelvic mass, or reproductive compromise, they may not come to diagnosis.

Many uterine abnormalities are asymptomatic until attempts at childbearing and thus, are often not diagnosed during adolescence. It is not until adulthood, when patients experience multiple spontaneous pregnancy losses, persistent menstrual abnormalities, or infertility that a definitive diagnosis is made. Patients with segmental agenesis/hypoplasia usually present with primary amenorrhea: however, if a normally functioning endometrium is present, significant cyclic or chronic pelvic pain may develop. It is only if a mucocolpos significant enough to cause symptomatic

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*Fig. 3. Redundant posterior hymen causing difficulty with tampon use.*
obstruction occurs as an infant, or hematocolpos at the time of menarche, that children or adolescents with obstructive phenomenon come to diagnosis.

**Diagnosis**

Identification of recognizable reproductive symptoms is an important key to the diagnosis of a structural defect of the female genital tract. The symptoms include cyclic or constant pain, pain with insertion and removal of a tampon, or the absence of menses. A gynecologic history of amenorrhea, the inability to remove a tampon, or persistent bleeding despite using a tampon is suspicious for a uterovaginal anomaly. Last, the physical examination may reveal an imperforate hymen or only a vaginal dimple with vaginal agenesis. The discovery of a blind vaginal pouch can suggest that the Müllerian contribution to the vagina has been impaired.

**Imaging Studies for Reproductive Tract Anomalies**

Radiographic imaging provides much needed information to aid in making the correct diagnosis. Delineation of the patient’s individual anatomy is essential to develop an appropriate plan for surgical correction. Ultrasound is helpful in identifying the anatomy in all cases of reproductive tract anomalies and can be used as a transabdominal, transvaginal, or transperineal approach. MRI can be helpful in determining the anatomy in cases of complicated obstructive anomalies, and is often considered the “gold standard” for imaging of anomalies of the reproductive tract. MRI is especially useful in determining the presence or absence of the cervix in complex anomalies or the presence of functioning endometrium in cases of a noncommunicating obstructed rudimentary uterine horn, however, MRI may miss a rudimentary uterine horn if it is located laterally along the psoas muscle and pelvic sidewall. Such a condition may be more likely in prepubertal patients who have very widely spaced uterine horns, as seen in cloacal anomaly or cloacal exstrophy. A hysterosalpingogram (HSG) can be helpful in determining the patency of the Müllerian structures and delineating complex communication with the urologic or colorectal system. An HSG can be uncomfortable unless performed under anesthesia and should be primarily reserved for use in older adolescents and young adults. In cases of complicated Müllerian anomalies, especially in combination with anomalies of the urologic and colorectal systems, additional information regarding anatomy and diagnosis may be obtained during an examination with the patient under anesthesia using cystoscopy, vaginoscopy, laparoscopy, and/or hysteroscopy.

**Renal and Other Anomalies**

Since the development of a normal Müllerian duct occurs in association with the normal development of the mesonephric duct, urinary tract anomalies are the most common abnormality associated with congenital anomalies of the female reproductive tract. Urinary tract abnormalities in patients with Müllerian duct anomalies include ipsilateral renal agenesis, duplex collecting systems, renal duplication, and horseshoe-shaped kidneys. In the general population, the incidence of unilateral renal agenesis has been estimated to be between 1 in 600 and 1 in 1200, on the basis of autopsy studies. The incidence of associated genital abnormalities in female patients with renal anomalies is estimated to be between 25% and 89%.

**TRANSVERSE VAGINAL SEPTUM**

Transverse vaginal septa are believed to arise from a failure in fusion and/or canalization of the urogenital sinus and Müllerian ducts. Approximately 1 in 80,000 females are
born with a transverse vaginal septum. The complete transverse vaginal septum may be located at various levels (low, middle, or high) in the vagina. Approximately 46% of vaginal septa occur in the upper vagina, 40% in the middle vagina, and 14% in the lower vagina.\textsuperscript{23} On examination, the vagina may seem foreshortened ending in a blind vaginal pouch. A recto-abdominal bimanual examination may elicit an abdominal/pelvic mass. Transverse vaginal septa are rarely associated with uterine anomalies.\textsuperscript{2}

The septa are usually less than 1 cm thick and may completely or incompletely extend from one vaginal sidewall to the other. Transverse vaginal septa commonly have a small central or eccentric perforation.\textsuperscript{24} Rarely, pyohematocolpos may be caused by ascending infection through the small perforation. If there is no perforation in the transverse septum, there is a resultant obstruction with hematocolpos from concealed menses; a mass is palpable above the examining finger on rectoabdominal palpation. An obstruction with a transverse septum resulting in hematometra may lead to endometriosis.\textsuperscript{25}

Ultrasound or MRI may help define the septum and its thickness preoperatively. A transperineal ultrasound may also confirm the thickness of the distal obstruction or distance from the perineal skin to allow for appropriate preoperative planning. It is also extremely important to identify a cervix, most reliably seen on MRI, to differentiate between a high septum and congenital absence of the cervix. Management varies between a high transverse vaginal septum and cervical agenesis, and attempts of surgical correction of cervical agenesis have been associated with significant complications, including death.\textsuperscript{26}

After menarche, patients with an obstructive anomaly present similarly; however, patients with a high septum present more quickly with significant pain because the uterus distends more quickly. Those adolescents with a microperforation in the transverse septum may have egress of some menstrual blood with continued discharge but may present with pyohematocolpos secondary to ascending infection or painful tampon insertion and dyspareunia.

Aspiration of the hematocolpos should always be avoided because of the risks of ascending infection and pyocolpos. Definitive surgical correction is the therapy of choice. Before surgery, some distention of the upper vagina with menstrual blood (hematocolpos) before the development of significant hematometra may be advantageous. This allows the potential to increase the amount of upper vaginal tissue available for reanastomosis. Furthermore, preoperative dilation therapy may decrease the thickness of the septum and increase the amount of lower vaginal tissue available. Thin septa can then be resected followed by primary end-to-end anastomosis of the lower and upper vagina. Thicker septa may require undermining and mobilization of the upper and lower vaginal mucosa before anastomosis, in addition to resection of the fibrotic septal tissue. A common complication of resection of thick septa is scar contracture and vaginal stenosis. A circumferential “Z”-plasty technique allows for scarring along the suture line to contract the incision in a longitudinal fashion rather than a transverse one.\textsuperscript{26} Postoperatively, wearing a vaginal mold or initiating early vaginal dilation may further decrease the risk of vaginal stenosis.\textsuperscript{27} If there is not enough vaginal mucosa to accomplish a pull-through procedure and reanastomosis of the vaginal mucosa, a graft may be necessary to create a patent vaginal tract.

**LONGITUDINAL VAGINAL SEPTUM**

Longitudinal vaginal septa may be associated with one of several uterine anomalies including a complete septate uterus, uterine didelphi, and rarely bicornuate uterus. Longitudinal vaginal septa have also been reported to occur in association with
anorectal malformations, including imperforate anus with rectovestibular fistula and cloaca. Septa may be partial or extend the complete length of the vagina. As many as 20% of patients will have renal abnormalities. Most reproductive-age patients present with complaints of dyspareunia and/or persistent bleeding despite tampon use, yet many women may be asymptomatic. Prepubertal girls undergoing evaluation and surgical management of anorectal or genitourinary anomalies are most often asymptomatic but, because surgical exposure is best at the definitive repair, most are treated then. The presence of a vaginal septum can usually be visualized on examination of the vaginal introitus. However, if the septum is incomplete and does not occupy the entire vagina, vaginoscopy may be necessary to evaluate the full extent in prepubertal patients (Fig. 4). Pelvic examination, with a speculum examination, in reproductive-age young women is usually adequate for visualization. Treatment is by surgical resection. Resection should be performed in childhood if undergoing other genitourinary procedures or repair of an anorectal malformation; however, when the vaginal septum is an isolated anomaly, it is most often corrected after menarche owing to the later pubertal diagnosis. The septum should be completely removed while taking care to avoid damaging the cervix or cervixes, which commonly insert along the septum at a similar level bilaterally (Fig. 5). Trauma to the bladder, rectum, and cervical blood supply should be avoided. After resection of the midline septum, closure of the vaginal mucosal defect is performed by suturing the mucosal surfaces together. In reproductive-age women the use of the hand-held harmonic scalpel for excision followed by reapproximation of the vaginal mucosa with interrupted sutures is effective.

**OBSTRUCTED HEMI-VAGINA WITH IPSILATERAL RENAL AGENESIS**

Longitudinal vaginal septa are often present with uterine didelphys; yet in some cases an obstructing septum is present. The association of the obstructed hemivagina with ipsilateral renal agenesis has been well described in the literature. In fact, an acronym has been proposed, OHVIRA (Obstructed Hemivagina and Ipsilateral Renal Anomaly), to describe the syndrome. Patients with an obstructed hemivagina present later than other obstructing vaginal anomalies, presumably because menstrual flow occurs normally from the nonobstructed hemiuterus. However, these patients also complain

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**Fig. 4.** Vaginal septum viewed on vaginoscopy of an infant undergoing surgical repair of a cloacal anomaly. Notice the rectal communication at the base of the vaginal septum.
of unilateral cyclic, followed by constant, pain. There may be a microperforation allowing for communication from the obstructed to the nonobstructed side resulting in prolonged or intermenstrual discharge, or very rarely, even pyocolpos.

On physical examination, a mass is felt to bulge from the lateral wall of the vagina toward the midline. After the pelvic examination, radiographic evaluation may also be helpful. Ultrasound is an excellent modality to demonstrate both the uterovaginal anomaly and the urinary anatomy. MRI can provide beautiful images; however, may add significant expense (Fig. 6). Diagnostic laparoscopy is rarely necessary. Most

Fig. 5. Vaginoscopy of a prepubertal girl after repair of an anorectal malformation. She had an associated longitudinal vaginal septum that was removed during her definitive repair. Notice the close proximity of the two cervices and the small amount of residual septum remaining posteriorly.

Fig. 6. MRI of the pelvis demonstrating an obstructed right hemivagina in a patient with OHVIRA.
patients with this anomaly can undergo resection of the obstructing septum (the common wall of the hemivaginas) in a fashion much like that described previously for longitudinal vaginal septa. The septal wall is resected to create a single vaginal vault. After resection of the septal wall of the obstructed hemivagina, the patient has normal function with a single vagina, two cervices, and two hemiuteri (uterine didelphis).

VAGINAL ATRESIA/DISTAL VAGINAL AGENESIS/SEGMENTAL VAGINAL AGENESIS

Vaginal atresia, distal vaginal agenesis, or segmental vaginal agenesis occurs when the urogenital sinus fails to contribute to the lower portion of the vagina. The uterus, cervix, and upper vagina develop normally, and the absent mid to lower section of the vagina is replaced by fibrous tissue. The most common presenting symptom is primary amenorrhea. Patients may develop cyclic or chronic pain and a pelvic or abdominal mass as the upper vagina fills with cervical mucus secreted after puberty or obstructed blood after concealed menarche. A physical examination confirms normal secondary development; however, the examination of the external genitalia reveals only a small dimple at the vaginal introitus. A rectoabdominal examination can be helpful to determine the presence of any midline structures (a distended upper vagina, a cervix, and/or uterus). In patients who cannot tolerate a rectoabdominal exam, an ultrasound can provide similar information to assist in differentiating this condition from vaginal agenesis. Ultrasonography, which should be done to assess renal status, also can confirm the presence of an obstructed upper vagina, or the presence of a normal cervix and uterus. Ultrasound with a transperineal approach also will demonstrate the thickness of the segment of agenesis which can be crucial information to guide the surgical approach; MRI is also an excellent modality to determine the presence of a cervix and exclude the diagnosis of cervical agenesis.

In individuals with agenesis or atresia of the distal/lower vagina, an obstruction with a hematocolpos of the upper vagina will result at the time of menarche and requires a surgical procedure to relieve the obstruction at the time of diagnosis. This should ideally be performed when a large hematocolpos has formed but before development of hematometra. The hematocolpos will act as a natural tissue expander hopefully decreasing the need for grafts. After appropriately positioning the patient, a crescentric incision is made where the hymenal ring is or should be located. Careful dissection is performed superiorly through the fibrous area of the absent segment taking precautions to avoid the urethra/bladder and rectum, until the bulging upper vagina is reached. Intraoperative ultrasound guidance can be useful to identify the distended upper vagina in difficult cases. The obstruction is then drained; the vaginal mucosa is identified and a pull-through procedure brings the distended upper vaginal tissue down to the introitus. Foley catheters or urethral sounds, Hegar dilators, or simultaneous rectal examinations are all helpful to determine the limits of the vaginal dissection.

If the distance to the vagina is too great and the vagina will not reach the introitus, an interposition graft has been used to join the introitus and upper vagina. When a bowel graft is necessary, the technique mandates a combined abdominal and perineal approach (Figs. 7–9). This surgical technique is more readily performed in childhood, especially if it is performed in the context of more complex reconstruction, such as genitourinary or anorectal surgery.

VAGINAL AGENESIS (MÜLLERIAN APLASIA)

Vaginal agenesis, Müllerian aplasia, or MRKH (Mayer-von Rokitansky-Küster-Hauser) syndrome all include the congenital absence of the vagina and are associated with variable Müllerian duct development. Aplasia of the Müllerian ducts results in not
only absence of the uterine corpus and cervix but also the upper portion of the vagina, which may result in near total or complete vaginal agenesis. Müllerian aplasia is estimated to occur in about 1 in 1000 to 1 in 83,000 female births, but the most widely cited incidence is approximately 1 in 5000 female births.\textsuperscript{38} In fact, the most common cause of vaginal agenesis is Müllerian aplasia or MRKH syndrome. Given the close association between the genital and urinary systems throughout fetal development, it is not surprising that approximately one third to one half of patients have associated renal anomalies, including renal agenesis, malrotation, or ectopic kidney.\textsuperscript{39–41} Other anomalies associated with vaginal agenesis include anomalies of the skeletal and auditory systems.

Vaginal agenesis is usually accompanied by cervical and uterine agenesis, although approximately 7% to 10% of affected individuals may have a normal but obstructed uterus or a rudimentary uterus with functional endometrium.\textsuperscript{42–45} Vaginal agenesis should be differentiated from vaginal atresia/distal vaginal agenesis/segmental vaginal agenesis. Another variant, cervical agenesis or atresia, is the presence of a uterus but agenesis of both the vagina and cervix. Approximately 90% of patients have some degree of Müllerian development, most commonly bilateral fibromuscular uterine

![Fig. 7. Intraoperative images of a patient undergoing surgical treatment of distal vaginal agenesis. The native upper vagina would not reach to the introitus.](image)

![Fig. 8. A segment of left colon with adequate length and blood supply was identified for use as an interposition graft.](image)
remnants located along the pelvic sidewall. However, only 2% to 7% of the uterine remnants will have a functional endometrial lining requiring intervention.46

Most patients present with primary amenorrhea around age 15 years, given that they have normal secondary sexual characteristics. Müllerian aplasia is the cause of 10% of cases of primary amenorrhea, second only to gonadal dysgenesis in a tertiary care center.11 Girls with Müllerian remnants with a functional endometrium may present with cyclic or chronic pain secondary to the accumulation of obstructed menstrual blood.

Evaluation of the patient with near total or complete vaginal agenesis, like other Müllerian anomalies, begins with a genital examination. Most patients will have a vaginal dimple or very foreshortened, blind-ending vagina. The hymenal fringe is usually present along with the small vaginal pouch, as they are both derived embryologically from the urogenital sinus (Fig. 10). A pelvic mass will usually be absent and occasionally a peritoneal fold can be palpated on rectoabdominal bimanual examination. Transabdominal pelvic and renal ultrasound will further aid in assessing the reproductive and urologic anatomy and frequently help to determine if a functional

Fig. 9. The colonic interposition graft was then anastomosed to the upper native vagina in an isoperistaltic fashion to reach the introitus comfortably.

Fig. 10. A view of the introitus of a patient with vaginal agenesis demonstrating the small amount of hymenal fringe that can often be present.
endometrium is present. An MRI may be more accurate in the evaluation of Müllerian structures, but given the expense, it can be reserved for when ultrasound is indeterminate.\textsuperscript{36} Laparoscopy may be necessary if noninvasive imaging fails to make an accurate diagnosis and it allows for the removal of obstructed uterine structures. Furthermore, a karyotype will definitively differentiate androgen insensitivity from Müllerian aplasia, if necessary.

**Counseling**

Of all the Müllerian anomalies, this diagnosis is by far the most upsetting to a girl, young adolescent, or adult woman, and her family. Counseling by an experienced team including nurses, social workers, psychologists and/or psychiatrists is recommended and should be strongly reinforced by the treating health care provider when interacting with patients and families.\textsuperscript{47} Although fertility concerns, primarily the inability to carry a pregnancy, are the overriding issue, it is also important to stress to the young woman and her family that she has normal ovarian function with normal production of sex steroids and normal amounts of genetic material. Emphasis on the possibility of fertility options with assisted reproductive technologies and a gestational carrier is crucial\textsuperscript{46,48} in addition to confirming the ability to create a functional vagina.

**Treatment**

Counseling for the young woman and her family is not only the first step in treatment of Müllerian aplasia, but likely the most important for a long-term successful outcome. Although patients and families may be hesitant to engage in psychotherapy, attention should be given to the psychosocial issues as well as to the correction of the anatomic abnormality. The patient’s cooperation and positive attitude are vital to the ultimate success of the creation of a functional vagina.\textsuperscript{27} The timing for the nonsurgical or surgical creation of a vagina is purely elective, although many patients feel it is emergent at the time of diagnosis. Because this procedure is not a surgical emergency and it is a decision with lifelong impact, surgical intervention should be delayed until the young woman chooses to proceed with a self-selected specific treatment plan. Because primary nonsurgical creation of a vagina requires continued vaginal dilation, it clearly is inappropriate to consider this approach in children. Surgical procedures to create the vagina which require postoperative dilation should not be performed during childhood. Parents who have been instructed to dilate their daughter’s vagina often present distraught and uncomfortable with the procedure. Thus, treatment should wait until the adolescent or young woman is able to determine the treatment and timing of the treatment. Parents may feel anxious and isolated at the time of diagnosis, precipitating interest and inquiry regarding surgical correction. Any technique for creation of a functional vagina should be delayed until the mid to late teens when the young woman can make her own decision, with the support of her family, and is comfortable with and willing to participate in the process. Some women may elect never to create a vagina. It is important to provide patients and their families with resource information presenting all options and support regardless of the treatment selected.

**Creation of a Functional Vagina**

A vagina may be created by both nonsurgical and surgical approaches.\textsuperscript{49,50} A successful outcome, using any method, is a vagina of adequate caliber (diameter and length), with an appropriate amount of secretion or lubrication, positioned as close as possible to an anatomic axis, to accommodate comfortable sexual intercourse. In addition, the vagina should require minimal care for maintenance. None
Nonsurgical Repair

The nonoperative approaches attempt to use progressive invagination of the vaginal dimple to create a vagina of adequate diameter and length. The nonoperative method of creating a functional vagina involves the use of pressure against the vaginal dimple to create a progressive invagination of the mucosa. The Frank approach for creation of a vagina involves the use of graduated hard dilators. The use of vaginal dilators should be offered as first line of therapy to patients with this condition. The rationale for this approach is related to the elimination of surgery, surgical scars, and exposure to anesthesia. Recent studies have shown that the use of dilators is greater than 85% to 90% successful for the creation of a functional vagina. It may take months to years to create a functional vagina depending on patient motivation and the frequency of dilation. The Ingram modification of this technique, which involved sitting on a bicycle seat stool, took an average of 11.8 months with a range of 3 to 33 months, but more than 90% were successful in dilation. Although this modification is an interesting adaptation of the Frank method, many young women initially find this technique very awkward. However, as the vagina gains length it can be helpful. The greatest success can be achieved through a combination of education, nursing support, mental health counseling, and a mentor program.

Surgical Repair

Surgical creation of a neovagina is another option for young women who fail nonoperative dilation therapy or who choose surgery after a thorough discussion with the patient (and parents/guardians as indicated) regarding the advantages and disadvantages. Surgical creation of a vagina can be accomplished by one of several techniques. Discussion should fully describe that many surgical techniques will require postoperative dilation to maintain vaginal adequacy. Generally, surgery should not be used as a first-line option. Many young women are interested in a “quick fix” and think that surgery will achieve this goal. The goals of surgery are to create a neovagina that is of adequate length and width and placed in an anatomically correct axis to provide comfortable, mutually satisfying sexual intercourse. Ideally the neovagina should not require exogenous lubrication, continued postoperative dilation, or have significant surgical complications. Currently, there are multiple operations appropriate for the creation of a neovagina in patients with vaginal agenesis but no consensus on the best approach. The procedure of choice should also be determined by the surgeon’s experience and success with the procedure because reoperation increases the risks of injury to surrounding organs.

The modified Abbé-McIndoe is the most commonly performed surgical technique by gynecologists in the United States. It involves harvesting a split-thickness skin graft from the patient’s buttocks and placing it over a vaginal mold. An incision is made at the vaginal dimple and adequate space created to the level of the peritoneum for the mold and graft. The labia minora are then temporarily sewn together to prevent the mold from expulsion. The patient is then placed on strict hospital bed rest for 7 days. The mold is then removed and the patient wears a stent continuously for 3 to 6 months, then nightly for an additional 6 months. The patient then must perform regular dilations, continuously wear the surgical mold, or engage in vaginal intercourse to prevent skin-graft contracture and loss of the neovagina. This technique is felt to have a greater than 80% functional success rate. However, it leaves the patient...
with a potentially disfiguring scar from the donor site. Other complications include graft failure, wound infection, hematoma, and fistula formation.

More recently, investigators have used the modified Abbé-McIndoe technique but substituted other tissues as graft material to avoid unsightly graft site scarring. They include artificial dermis with human recombinant basic fibroblast growth factor spray, autologous buccal mucosa, Interceed absorbable adhesion barrier (Ethicon, Inc, Somerville, NJ), and human amnion.

The modified laparoscopic Vecchietti procedure creates a dilationlike neovagina in 7 to 9 days. It involves placement of an acrylic 2-cm olive-shaped bead onto the vaginal dimple that is gradually pulled superiorly by threads laparoscopically placed that are then connected to the traction device placed on the patient’s abdomen. The threads are then gradually tightened approximately 1.0 to 1.5 cm per day for a week. Postoperatively, the patients must comply with daily vaginal dilation until regularly sexually active.

The laparoscopic Davydov technique uses the patient’s own pelvic peritoneum to line the neovagina. It involves dissection of the perineum to create a neovaginal space while laparoscopically mobilizing the peritoneum. The peritoneum is then sutured to the introitus and a purse-string suture closes the cranial end of the neovagina. A vaginal mold is left in situ for 6 weeks and the patient then begins daily dilation until regularly sexually active. Complications related to laparoscopic injury and fistula formation have occurred; however, patients report similar sexual function to women with a native vagina.

Patients who fail nonoperative dilation therapy or those seeking little to no postoperative upkeep may be candidates for a bowel vaginoplasty. Bowel vaginoplasty is the preferred method of most pediatric general surgeons, given the immediate and long-term correction of the anomaly. Each bowel segment has advantages and disadvantages. Other than the consideration given to a segment of adequate length and its ability to reach the perineum, the segment chosen should not interfere with fecal continence or the ability to perform necessary simultaneous reconstructive procedures.

Bowel vaginoplasty is performed by selecting an approximately 10-cm segment of bowel that can be mobilized but retain an adequate vascular pedicle to reach the perineum without traction on the pedicle or graft. It may be placed in an isoperistaltic or antiperistaltic fashion in the space dissected from the perineum to the pouch of Douglas between the bladder and rectum. Most authors advocate anchoring the proximal bowel neovagina to avoid graft prolapse.

Sigmoid vaginoplasty has been the most commonly used bowel segment, given its proximity to the perineum and therefore little difficulty performing a tension-free anastomosis to the introitus. The sigmoid also produces mucous that acts as a natural lubricant. However, some patients have found it excessive and at times malodorous requiring postoperative neovaginal irrigation. Patients do not require routine vaginal dilation given the large lumen, but the anastomosis at the perineum may stenose. As long as the introitus at least retains some patency, this can be addressed after puberty with an outpatient introitoplasty (Fig. 11). The disadvantages include the need for a laparotomy (and the resultant scar) unless a skilled laparoscopist is available who can isolate an adequate vascular pedicle. Also included in this procedure are the inherent risks of wound problems at the bowel anastomosis and graft prolapse or even failure.

When an adequate segment of colon is not available for vaginoplasty, small bowel has been used. It is more difficult to mobilize to the perineum, has a smaller lumen, produces excessive mucous, and may be less durable when exposed to the potential trauma of sexual intercourse. Complete stenosis has also been reported.
Postoperative sexual satisfaction has been assessed in a validated fashion in only a relatively small number of patients who underwent bowel vaginoplasty. Communal and colleagues administered the standardized Female Sexual Function Index to 16 patients, and Hensle and colleagues administered a validated Female Sexual Dysfunction Questionnaire to 44 patients. Eight of the 11 who responded and who were sexually active reported a 75% “very satisfied” rate, whereas 78% of the 36 who responded endorsed sexual satisfaction, respectively.

The Williams vulvovaginoplasty uses full-thickness skin flaps from the labia majora, creating a neovaginal pouch. Vaginal dilators are then inserted daily postoperatively for a month. Unfortunately, the kangaroo-like pouch is positioned at an anatomically odd angle and may have a relatively short length. Furthermore, the labia majora is hair-bearing skin, which will usually result in undesirable cosmesis. Attempts to avoid some of these concerns have included preoperative labia majora tissue expanders, the Creatsas modification, and substituting labia minora.

Full-thickness skin grafts also have been performed using the myocutaneous rectus abdominis or gracilis and pudendal thigh fasciocutaneous flaps. Although potentially necessary in patients with more diffuse anomalies, like cloacal extrophy, these neovaginal grafts are usually one of last resort for a diagnosis of isolated vaginal agenesis given the disfiguring scarring and risks of harvest site wound concerns and graft failure.

**RUDIMENTARY UTERINE HORMS**

Since approximately 7% to 10% of patients with vaginal agenesis (Mu¨ llerian aplasia) may have a rudimentary uterus with some functional endometrium and no outflow tract, it is important to maintain a high index of suspicion for an obstructed rudimentary uterine horn as a cause for recurrent pain in patients with this diagnosis. Ultrasound and/or MRI may be useful in identifying the noncommunicating uterine horn and determining whether functional endometrium is present, as evidenced by a visible endometrial stripe. Not uncommonly, laparoscopy is necessary to diagnose and remove the obstructed rudimentary noncommunicating uterine horn. Patients who have any obstruction to menstrual flow, including an obstructed uterine horn, are at
increased risk of endometriosis, but experts believe the endometriosis resolves after relief of the obstruction. Early identification and excision of a blind rudimentary horn will prevent endometriosis by eliminating reflux of menstruation. If a rudimentary is horn is known to be present, patients should be counseled about symptoms of an ectopic pregnancy and the need for immediate evaluation, because spontaneous pregnancies have been reported.79

CERVICAL ATRESIA/ HYPOGENESIS

Cervical agenesis and dysgenesis are rare54,80–82 but are extremely important to differentiate from diagnoses like transverse vaginal septum or vaginal atresia/segmental vaginal agenesis. A correct diagnosis allows appropriate counseling before surgical intervention as the classic management recommendation for this condition is hysterectomy. Patients may present with primary amenorrhea, cyclic or chronic abdominal or pelvic pain, and/or a distended uterus. Ultrasonography can aid in defining the anatomy, yet an MRI allows superior identification of the presence and integrity of the cervix.18 The adequacy of the cervix is the crucial part of the decision to leave the uterine structure in place and anastomose to the native lower vagina or a newly created vagina. The diagnosis and management of this Müllerian anomaly are both challenging and controversial.

DISORDERS OF THE UTERUS

Complete Uterine Septum

The septate uterus has a smooth, normal external surface at the fundus, but the endometrial cavity is split into two by a midline septum. Most uterine fusion/duplication abnormalities do not require surgical intervention; however, the septate uterus is well known for the associated obstetric concerns. If patients experience pain, recurrent miscarriage, infertility, or premature labor, the abnormality should be repaired by hysteroscopic resection.83–89 The situation is more controversial in young women in whom the diagnosis is made in late adolescence or early adulthood. The decision to surgically intervene before attempts at pregnancy still remains unclear. The presence of a concomitant vaginal septum may influence the timing of intervention. A combined approach may be considered.

Bicornuate Uterus

The uterine fundus is deeply indented, often heart-shaped, in patients with a bicornuate uterus. In most cases, a single cervix is present. The level of the indentation of the fundus can be complete, partial or arcuate. Historically, treatment in the form of a metrorrhaphy, had been recommended. Presently, no surgical intervention is recommended and patients are followed closely for obstetric concerns. In most cases the vagina is normal.

Unicornuate Uterus

A unicornuate uterus is a single uterine horn that has only a single round ligament and fallopian tube, sometimes referred to as a hemiuterus. The opposite uterine horn (hemiuterus), round ligament, and fallopian tube, derived from the opposite Müllerian duct, may be absent or underdeveloped. Variations in the degree of hemiuterine development can occur producing a noncommunicating uterine horn (hemiuterus) on the contralateral side with or without active endometrium. A single unicornuate uterus communicates with a single cervix and a normal vagina. As suspected with unilateral impairment in Müllerian development, associated renal anomalies are common.
Patients with a unicornuate uterus are at increased risk of premature labor and breech presentation. As in other obstructive anomalies, endometriosis and subsequent fertility issues may be significant in patients with an associated obstructed uterine horn or hemiuterus.

PREGNANCY OUTCOME IN WOMEN WITH MÜLLERIAN DUCT ANOMALIES

Women with Müllerian duct anomalies seem to have an increased rate of unexplained infertility, endometriosis, spontaneous abortion, breech presentation, and premature delivery. More long term outcome data are necessary to fully determine the obstetric prognosis of women with Müllerian duct anomalies associated with other urogenital or anorectal malformations. Women who have had vaginal atresia/lower vaginal agenesis/segmental vaginal atresia corrected by the creation of a neovagina are able to become pregnant and to maintain a pregnancy, if a well developed cervix and uterus are present. Patients with vaginal agenesis always should be counseled regarding opportunities for adoption and surrogacy. The wider use of assisted reproductive technologies also will enhance the reproductive capacity of women with congenital abnormalities of the reproductive tract.

ADDITIONAL READING


REFERENCES