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Introduction

One of the more uncommon congenital anomalies is cervical agenesis, or the absence of the cervix. These patients present with primary amenorrhea, cyclic chronic pelvic pain and a palpable pelvic mass resulting from a hematometra, due to the obstructed outflow of menstrual blood [13]. Reviews of the literature emphasize its rarity, with an estimated incidence of 1 in 80,000–100,000 births [16]. A 2004 review reported 116 cases since 1900 [8].

The cervix typically forms as a result of condensation of stromal cells around the fused müllerian ducts, which is in contact with the

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urogenital sinus. Embryologically, cervical agenesis is thought to result from a failure of canalization of the fusion of the ascending sinovaginal bulb with the descending Müllerian system. An adequate vagina and formation of the cervix also depends upon elongation of the müllerian ducts. The co-occurrence of cervical and vaginal agenesis could result from a failure of the elongation of the mullerian ducts [7].

Cervical agenesis or dysgenesis is often present with other genital or urogenital tract anomalies. Vaginal asplasia often occurs with cervical agenesis (60 out of 83 reported cases of cervical agenesis), but is much less commonly associated with cervical dysgenesis [8]. Cervical agenesis has also been found to be associated with renal anomalies, with an approximated incidence of 20 % (from a case review of 20 patients) [7]. Rock et al. found associated anomalies in 10 of their reported 30 cases: ovarian malposition (n=4), tubal abnormalities (n=4), endometrial hypoplasia (n=5), and a solitary kidney (n=2) [15].

Diagnosis

The diagnosis of cervical agenesis or dysgenesis can be facilitated with radiologic studies, including ultrasound and MRI. Valdes et al. first reported the diagnosis of cervical or vaginal atresia via ultrasound in 1984 [18]. Some have advocated the use of trans-rectal ultrasound, especially in

associated cases of vaginal canalization defects [6]. Other reports have found MRI to be extremely useful for pre-operative diagnosis [11, 12, 15]. MRI also has the advantage of imaging of the upper genito-urinary tract. All radiologic studies should be corroborated by pelvic examinations under anesthesia for a conclusive diagnosis. Pre-surgical diagnosis is helpful, however, for appropriate surgical preparation [13]. It is also crucial to differentiate these patients from those with an atretic segment of the vagina and those with a high transverse vaginal septum.

There are two broad categories of cervical anomalies: cervical agenesis and cervical dysgenesis. Patients with cervical agenesis have no uterine cervix and the lower uterine segment ends in a peritoneal sleeve [13]. Patients with cervical dysgenesis can be divided into four subtypes:

1. A cervical body consisting of a fibrous band extending towards the vagina that may have endocervical glands
2. Intact cervical body with obstruction of cervical os
3. Stricture of the midportion of the cervix, which is hypoplastic, with a bulbous tip
4. Fragmentation of the cervix with no portions connected to lower uterine segment [14] (Fig. 23.1).

Management

Current management recommendations are based on case reports and literature reviews; there have been no randomized trials to elucidate best surgical practice. This chapter summarizes surgical recommendations from several different reviews of the surgical literature with specific recommendations dependent on each patient's specific cervical anatomy.

Traditionally, hysterectomy was advocated as the treatment of choice for these patients. Early attempts to create uterovaginal anastomosis resulted in a variety of serious surgical complications, including endometritis, pelvic inflammatory disease, sepsis, and injury to other pelvic organs including bowel and bladder. Even if a passage is created through fibrous tissue between

the uterine cavity and the vagina, there are not typically functioning endocervical glands. The resulting absence of cervical mucus creates a difficult environment for sperm transport for patients desiring fertility.

Furthermore, patients are also subjected to long-term post-operative complications. Endometriosis can develop along the fistulous tract and these patients are also at higher risk for retrograde menstruation, increasing the likelihood of endometriosis in the pelvis [14]. The tract can re-stenose, requiring the need for repeat operations for further scar tissue [2, 14]. Recurrent pelvic infections after attempted fistulous tract formation can also eventually result in a hysterectomy and, if the infections are severe enough, bilateral oophorectomy [14].

Since the 1990s, however, there has been a shift towards attempting anastomosis of the utero-vaginal tract for reconstruction. This shift parallels advancement in surgical techniques and the availability of broad-spectrum antibiotics. There are, however, a few peri-operative considerations for patient selection to obtain higher rates of a successful surgical outcome, typically defined as long-term patency of the cervical canal, with subsequent cyclical menstruation and the possibility of pregnancy. Ideal surgical patients have a larger amount of cervical stroma and the presence of rudimentary endocervical glands. Rock et al. defines sufficient amount of cervical stroma as being at least 2 cm in diameter [15]. In addition, there should be a small discrepancy between the size of the uterine muscularis and vaginal stroma for ideal juxtaposition of the anastomotic site to decrease scarring [13]. Patients with vaginal agenesis tend to have more complicated surgeries due to the requirement of additional grafting of the neovagina.

As data for reconstructive surgery comes from smaller case reports, it is important to have an honest discussion pre-operatively with the patient disclosing the risks of surgery. In addition, it is helpful to obtain thorough imaging to attempt a pre-surgical diagnosis as noted above.

Once surgery is begun, the pelvic anatomy is carefully defined and the vesicouterine and recto-uterine space are fully developed. It is imperative

that the surgeon develop these spaces to determine whether there is sufficient cervical tissue for possible coring or anastomosis of cervical fragments. If reconstruction is not deemed feasible or if the uterine cavity is hypoplastic, a hysterectomy is performed [15].

If reconstruction is undertaken, surgical techniques vary depending on the amount of cervical
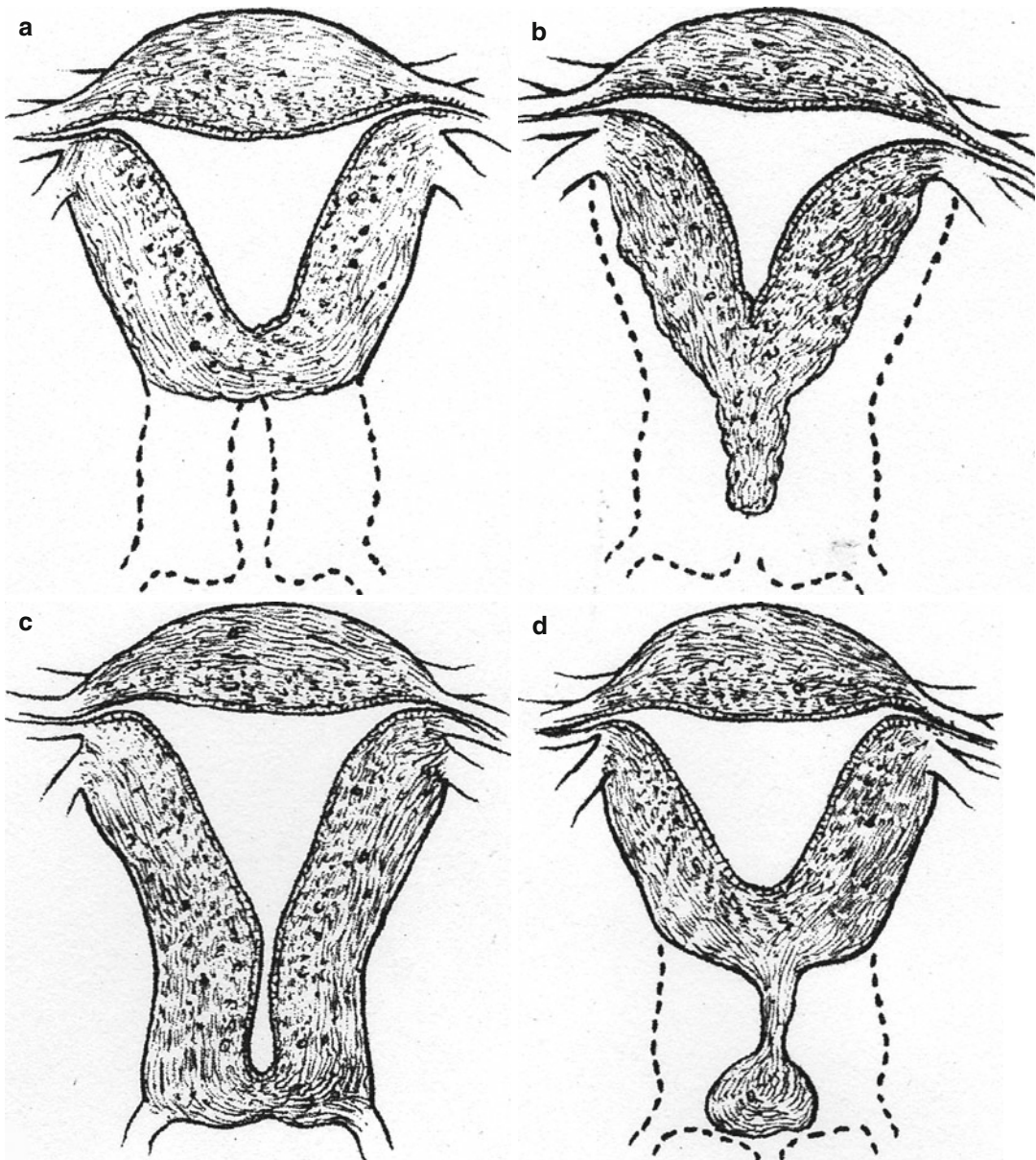

Fig. 23.1 Depictions of cervical agenesis and cervical dysgenesis (With permission from Rock and Jones [14]). (**a**) Cervical aplasia. (**b**) Cervical body consisting of a fibrous band of variable length and diameter that can contain endocervical glands. (**c**) The cervical body is intact with obstruction at the cervical os. Variable portions of the cervical lumen are obliterated. (**d**) Stricture of the midportion of the cervix, which is hypoplastic with a bulbous tip. No cervical lumen is identified. (**e**) Cervical fragmentation in which portions of the cervix are noted with no connection to the uterine body
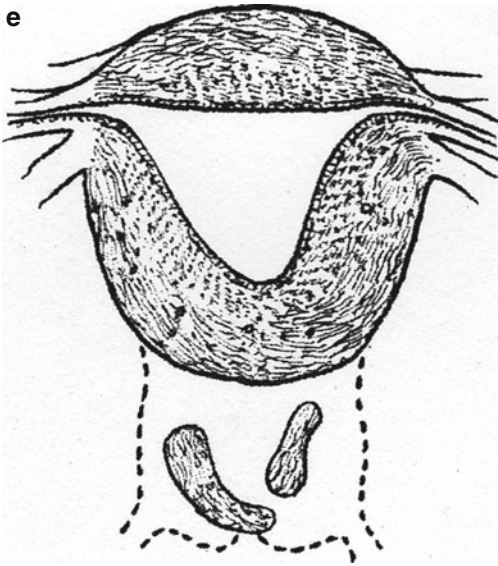
Fig. 23.1 (continued)

tissue present. If there is a small amount of cervical obstruction or a small atretic segment of the endocervical canal with a normal vagina, the surgeon can perform a coring or drilling procedure. During this procedure, the cervix is cored to remove the obstruction. A catheter is left in place, optimally with a full- thickness skin graft around the catheter to allow the tract to epithelialize more rapidly [15]. If there is accompanying vaginal aplasia, the surgeon can perform a vaginoplasty using the McIndoe technique [15].

In the presence of cervical agenesis or dysgenesis with cervical fragments or a fibrous cord, a more extensive surgery, such as an uterovaginal anastomosis, is advocated.

A large case series published by Deffarges et al. in 2000 described the surgical technique of uterovaginal anastomosis in 18 patients with cervical atresia. The patients underwent laparotomies with dissection of the vesicouterine and rectouterine space. An incision on the most superior portion of vaginal tissue was made and a channel formed between the bladder and the rectum until the abdominal anterior and posterior dissections were reached. A 10-mm dilator was inserted through an incision on the uterine fundus and placed at the most inferior portion of the uterus. The atretic vaginal tissue was resected in a similar technique

as with a cervical conization until the uterine cavity was entered. The uterus was then sutured in a circumferential manner with 3-0 polyglactine. A 16 French Foley catheter was placed in the canal to maintain patency for 15 days and patients were given Ampicilin for the duration [4].

A similar technique was described by Creighton et al. however the authors incorporated the use of laparoscopy. Laparoscopically, sutures were placed on the uterus for uterine suspension. An incision was made in the uterine fundus with a harmonic scalpel and a probe placed in the uterus to identify the lowermost portion of the uterus, which was incised horizontally. A second probe was placed in the vagina and a laparoscopic incision made over the most superior portion of the vagina. A Foley catheter was passed between the vagina and the uterus and the uterus closed with 2-0 polydioxanone suture circumferentially at 12, 3, 6, and 9 o'clock. In this case, the Foley was left in place for 4 weeks and the anastomotic site remained patent [3].

Other case reports have discussed the need for accompanying the cervical reconstruction with a graft of the neocervical canal to allow for improved healing and decreased stenosis. Possible graft tissue includes full thickness skin grafts [15], bladder mucosa graft, or amniotic membrane (from a case describing reconstruction after cesarean section) [10].

Another surgical technique involves the use of end-to–end anastomosis for the cases of cervical dysgenesis with cervical fragmentation. Grimbizis et al. describes a patient with cervical fragmentation in a symmetrical transverse fashion. They created an end-to-end anastomosis during a laparotomy, connecting the central and distal portions of the cervix and then using a Foley catheter as a stent in the endocervical canal [8] (Table 23.1).

Outcomes of Surgical Management

Reports of outcomes have differed between case series. In a retrospective 2010 review from Rock et al. describing surgical experience from 1940 to 2008, 30 patients with cervical agenesis and dysgenesis were described. Nineteen of the 30 patients underwent hysterectomy and

Table 23.1 Description of suggestive reconstructive surgical treatment

Anatomic findings	Suggested reconstructive surgical treatment
Cervical agenesis	
Present vagina	Anastomosis of lower uterine segment to vagina epithelium
Absent vagina	Suture uterine muscularis to stroma and graft material used to create neovagina
Cervical dysgenesis with cervical fragmentation	
Present vagina	Creation of a lumen by coring out center of cervical fragments or drilling a new lumen with insertion of a stent and optional grafting of neocervical canal. Sew fragments together or remove fragments and create a uterovaginal anastomosis
Absent vagina	Create neovagina using a graft
Cervical dysgenesis with fibrous cord	
Present vagina	Remove cord and perform uterovaginal anastomosis or create a neocervical canal with drilling or coring technique with insertion of a stent (if sufficient diameter cord)
Absent vagina	Removal of cord and uterus and create a vaginoplasty
Cervical dysgenesis with cervical obstruction	
Present vagina	Create a neocervical canal using a drilling/coring technique. Optional grafting of endocervical canal or remove cervix and perform uterovaginal anastomosis Create a neocervical canal using a drilling/coring technique. Optional grafting of endocervical canal.
Absent vagina	Vaginoplasty if required

Adapted from Rock et al. [15]

11 of the 30 had attempted uterovaginal anastomosis. Six of the 11 patients who underwent surgical reconstruction (55 %) eventually underwent a follow-up hysterectomy due to subsequent re-obstruction at the surgical site. The patients with the best outcomes were those with an intact cervical body and an obstructed cervical os; all of those patients (n = 4) had successful menstruation and one of the patients had two viable live births. Of those with cervical dysplasia consisting of a fibrous cord or cervical fragments, nine out of the ten patients ultimately required a hysterectomy [15].

In the case series from Deffarges, all 18 patients with cervical atresia (100 %) had restoration of menses though five (33 %) suffered from postoperative dysmenorrhea. Two of their patients had a low vaginal stenosis and one had secondary cervical stenosis, requiring multiple attempts at recanalization. Four of the patients (22 %) became pregnant spontaneously for a total of six spontaneous pregnancies. All required cesarean sections for delivery [4].

In 2008, Fedele et al. described uterovestibular anastomosis in 12 consecutive patients with cervical and vaginal aplasia. In their case series, all women (100 %) attained regular menstruation and had patency of the neovagina. Interestingly, all 12 of their patients were found to be producing mucus at the uterovaginal anastomosis despite complete cervical atresia on a pre-operative MRI in ten of the patients. The authors hypothesized the most caudal endometrial glands could undergo a "mucinous- secretive metaplasia." None of their patients had attempted pregnancy at the time of publication, so fertility and pregnancy outcomes were not reported [5].

Pregnancy outcomes have been incompletely reported. There have been some reports of pregnancy after reconstruction [4, 9, 15] and after IVF with laparoscopic zygote intra-Fallopian transfer (ZIFT) [17]. There have been two case reports of transmyometrial embryo transfer after in-vitro fertilization in patients with cervical agenesis that had not undergone surgical reconstruction [1, 10]. Most reports describe delivery by cesarean section [4, 9, 17].

Conclusion

Cervical agenesis and dysgenesis are rare Müllerian anomalies for which the management has been based upon small case series. Although hysterectomy has traditionally been the primary mode of surgical management, surgical reconstruction is a possibility for well- selected patients in the form of canalization, uterovaginal anastomosis or the creation of an end-to-end anastomosis depending on the anatomy observed. The majority of well- selected patients have good surgical outcomes with the attainment of cyclical menstruation and a few with subsequent live births.

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