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Gynecologic concerns in patients with anorectal malformations

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Reproductive congenital anomalies;
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Imperforate anus;
Vestibular fistula;
Anorectal malformation

Children with anorectal malformations (ARMs) constitute a significant group within a pediatric surgery practice. In females, the most common ARM is an imperforate anus with a rectovestibular fistula, followed by an imperforate anus with a rectoperineal fistula and then cloacal anomalies. Some malformations, such as an imperforate anus with a rectovestibular fistula, may seem straightforward, treated with a posterior sagittal anorectoplasty; however, it is vital to recognize the association with gynecologic anomalies. Girls with the most complex anorectal defect, cloacal malformation, have significant associated urological and gynecologic anomalies, which should be recognized and treated. An opportunity to diagnose and treat such anomalies may be missed in the newborn period or at the definitive surgery, unless the pediatric surgeon is aware of this association. With the knowledge of the associated anomalies and long-term sequelae, surgeons can provide better care for girls and important counseling for parents. Important gynecologic issues to consider may be divided into several categories, such as infancy, puberty, sexual intimacy, and obstetrical concerns. Knowledge of reproductive-related issues in females with ARMs allows the pediatric surgeon and their gynecology colleagues to provide optimal surgical management throughout infancy, childhood, and into young adulthood. Appropriate counseling for patients and families about potential reproductive concerns that may develop many years after the definitive surgical repair allows preparation and planning to preserve future fertility.

It is highly likely that most pediatric surgeons will treat both boys and girls with anorectal malformations (ARMs), as they constitute a significant group within a pediatric surgery practice. In females, the most common ARM is an imperforate anus with a rectovestibular fistula,[1] followed by an imperforate anus with a rectoperineal fistula and the cloacal anomaly. Some malformations, such as an imperforate anus with a rectovestibular fistula, may seem straightforward, treated with a posterior sagittal anorectoplasty; however, the association of gynecologic anomalies is vital to recognize. Girls with the most complex anorectal defect, the cloacal malformation, often have associated urological and gynecologic anomalies, which should be recognized and treated. An opportunity to diagnose and treat such anomalies may be missed in the newborn period or at the definitive surgery, unless the pediatric surgeon is aware of this association. Important gynecologic issues to consider may be divided into several categories, such as infancy, puberty, sexual intimacy, and obstetrical concerns. The presence of hydrocolpos is the major concern in the newborn period, followed many years later by the onset of menstruation as the next time for possible gynecologic problems. Sexual intimacy should be addressed after puberty and before the onset of sexual activity. Obstetrical issues should also be approached as young women contemplate reproductive potential. Knowledge of reproductive-related issues in females with ARMs and collaboration with experienced gynecologic colleagues allows the pediatric sur-

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geon to provide optimal medical and surgical management throughout infancy, childhood, and into young adulthood.

Evaluation for associated anomalies

Many pediatric surgeons report that they customarily repair most ARMs (including rectoperineal and rectovestibular fistula), other than a cloaca, without evaluating for the presence of a gynecologic anomaly. A recent review at our institution reinforced the importance of such evaluation and the deficiency in this approach.[2] In a series of 272 patients treated for imperforate anus with a rectovestibular fistula, 5% had an associated vaginal septum and 9% had an absent vagina. In that series, the authors treated a young woman who presented with primary amenorrhea 18 years after the initial anoplasty for an imperforate anus with a rectovestibular fistula. She and her parents were unaware of her diagnosis of absent vagina as the diagnosis was not made until young adulthood. Most pediatric surgeons are aware of the strong association of gynecologic anomalies with a cloacal anomaly, which is cited as 53%-67% of female patients with uterovaginal anomalies.[3-5] Gynecologic abnormalities seem to be uncommon in patients with imperforate anus and a rectoperineal fistula.

Why is it important to learn of such anomalies so early? Of course, in cloacal anomalies, it is necessary to separate both the urinary and colorectal systems from the reproductive tract; however, it is also essential in such cases to appreciate the uterovaginal anatomy to prevent adverse outcomes at puberty and menarche. A high rate, 36%-41%, of menstrual obstruction at puberty is well described.[6,7] If not diagnosed early, not only can this complication produce significant pain, often requiring urgent surgical intervention, but can also lead to infertility by acute removal of the reproductive structures to relieve pain, or through the development of endometriosis. In less complex ARMs such as imperforate anus with rectovestibular or rectoperineal fistula, a vaginal septum is the most common associated finding and can be most effectively treated during the initial repair of the rectum, without trauma to the hymen or introitus. This can be accomplished with one anesthetic, with optimal surgical exposure, and without the possible psychological concerns with a "vaginal surgery" as a young teenager.

Evaluation of reproductive anatomy

Vaginoscopy should be performed on all girls during the definitive repair of any ARM. This can be performed under coincident anesthesia at the time of the posterior sagittal anorectoplasty, with preparation to undertake vaginal septum resection if diagnosed. In more complex malformations, an examination under anesthesia with both cystoscopy and vaginoscopy can often be beneficial for surgical planning. If girls or young women were treated in infancy, without a complete gynecologic evaluation, endoscopy can be combined with any other indicated surgical procedures during childhood. The goal is to have as much information as possible regarding reproductive anatomy before the onset of menarche. Accurate knowledge of the reproductive anatomy allows both parents and providers to adequately prepare for pubertal changes and necessary surveillance.

Vaginoscopy allows evaluation of the vaginal anatomy, documentation of vaginal duplication with 2 hemivaginas and a septum, documentation of the septal length, and total vaginal length. The cervix or cervices can also be visualized. This allows documentation of the cervical appearance, including cervical development, the position of the cervix/cervices in the vagina(s), and the presence or absence of mucus at the ectocervix (from which one can infer patency).

Assessment of the internal reproductive anatomy should be performed whenever an intra-abdominal procedure is performed. In patients who undergo diversion with creation of a colostomy before the definitive surgery, evaluation of the reproductive anatomy can be performed at the time of colostomy closure if entry into abdomen was not needed at the definitive repair. If a colostomy was not necessary, this assessment can be deferred until later, perhaps at the time the patient undergoes the creation of an appendicostomy if indicated for bowel management. It is advantageous to perform an assessment of the internal anatomy before the onset of menses, to assess for the possible risk of obstruction to menstrual flow. The uterus/uteri should be identified. The insertion of the fallopian tubes into the uterine body should be documented, in addition to the communication of the fallopian tubes with the ovaries.

Patency of the Müllerian system should also be confirmed. Pediatric feeding tubes can be used to cannulate the distal aspect of each fallopian tube. Gentle compression of the fimbriae around the tube allows the antegrade instillation of saline through the fallopian tube, uterus, cervix, and vagina of the Müllerian system bilaterally (Figure 1). This assessment of patency can provide reassurance regarding the future outflow of menstrual products. Although almost all females with ARM have normal ovaries, this should also be confirmed. Assessment of the Müllerian system can also be performed during laparoscopic procedures. Each cervix can be cannulated with a ureteral catheter during vaginoscopy; saline or dye is then instilled in a retrograde fashion while the fallopian tubes are observed with the laparoscope for spillage from each fallopian tube, thus confirming retrograde patency.

Important gynecologic issues to consider may be divided into several categories such as, infancy, puberty, sexual intimacy, and obstetrical concerns. The presence of hydrocolpos is the major concern in the newborn period, followed many years later by menarche (the onset of menstruation) as the next time for possible gynecologic problems. Sexual intimacy should be addressed after puberty and before the onset of sexual activity, and obstetrical issues should also be

considered, as young women contemplate reproductive potential. It is logical to address these issues in a chronologic manner, beginning with infancy.

Infancy

Hydrocolpos

The most significant gynecologic concern during infancy is the presence of hydrocolpos. Hydrocolpos involves the accumulation of fluid, urine, and mucus within the vagina, causing distension of the vagina or hemivaginas. Secondary obstruction of the urinary tract can occur with the development of hydroureter and/or hydronephrosis due to compression of the ureters at the trigone by the hydrocolpos. The definitive etiology of hydrocolpos is unclear; however, it seems to occur more commonly in females with vaginal duplication, a cloaca with a long common channel, or a posterior type cloaca.[8] In our experience of 411 females with a persistent cloaca, the incidence was 28% or 116 cases.[8] Hydrocolpos may be identified antenatally. With an appropriate index of suspicion, abnormal prenatal ultrasound findings can be suggestive of hydrocolpos in a baby with a cloacal anomaly. Prenatal magnetic resonance imaging may provide a more definitive view of the anatomy.[9]

In newborns, the treatment of choice is drainage of the vagina with vaginostomy, which can be performed at the same time as creation of the colostomy.[10] A pigtail catheter can be placed transabdominally that should be left in place until the definitive surgery (Figure 2). Single puncture drainage or intermittent attempts to drain a hydrocolpos by transiently passing a catheter from a perineal approach are discouraged. Such techniques are ineffective at complete drainage and can put the patient at significant risk for infection, including sepsis, and persistent inflammation, which may adversely affect mobilization of the vagina(s) at the definitive surgical repair. If a vaginal septum is present with 2 hemivaginas, a window or defect should be made within the septum to allow adequate initial and continued drainage of both hemivaginas (Figure 3). When creating the window, attention should be paid to the location of the cervix on each side of the septum. On each side of the septum, most cervices are located close and adherent to the septum at the superior aspect and often located at the same level of the vagina. Trauma or damage to the cervix even in infancy has the potential to increase the risk of cervical insufficiency as an adult woman. Cervical insufficiency is a recognized cause of mid-trimester pregnancy loss as an adult woman.[11] In rare cases, vesicostomy may be necessary in addition to vaginostomy, when a patient has atresia or stenosis of the common channel, poor spontaneous bladder drainage, or other associated urological indications.

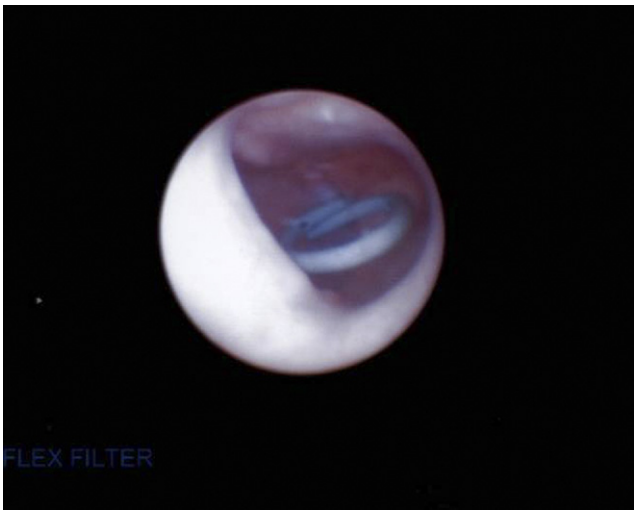

Figure 2 A view of the lower vagina during vaginoscopy for a patient with a history of hydrocolpos. The pigtail catheter is seen bridging both hemivaginas through a defect in the upper aspect of the vaginal septum. (Color version of figure is available online.)

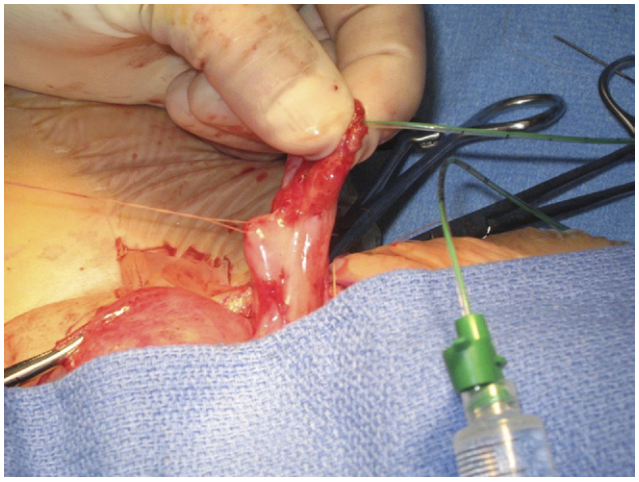

Figure 1 A #3 pediatric feeding tube is gently placed into the distal aspect of the fallopian tube to instill saline when testing patency of the Müllerian system. (Color version of figure is available online.)

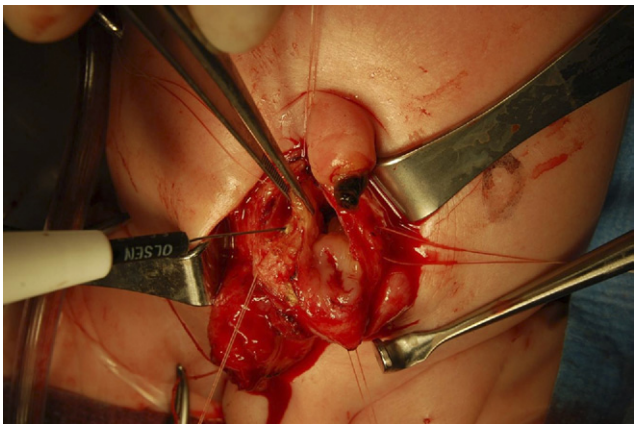

Figure 3 An intraoperative view of the creation of a window in the vaginal septum to allow the pigtail catheter to drain both hemivaginas until the definitive cloacal repair. (Color version of figure is available online.)

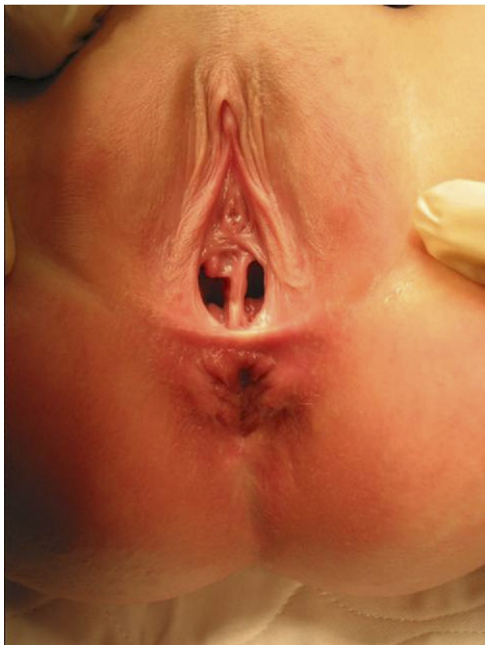

Figure 4 A perineal view of a vaginal septum. (Color version of figure is available online.)

Vaginal septum

As previously noted, about half of the females with a cloaca and 5% of girls with an imperforate anus and rectovestibular fistula will have vaginal duplication and a vaginal septum (Figure 4). During infancy and childhood, the presence of the septum has no clinical ramifications. As young women enter puberty and become menarchal, they may desire to use tampons for menstrual hygiene. The presence of a vaginal septum prevents effective use of a tampon for menstrual flow. In adult women, it may cause painful vaginal intercourse. The posterior sagittal approach allows excellent surgical exposure to resect the distal aspect of the septum and unify the distal vagina. The advantages of resecting the septum at the definitive repair include a single anesthetic exposure, optimal surgical exposure, and elimination of the possible psychological concerns of a "vaginal surgery" as a young teenager. If the septum is not resected at the definitive repair of the ARM, surgical management should be deferred until after puberty. Estrogen produced by the ovaries during puberty increases the pliability and flexibility of the hymen thus minimizing any trauma to the introitus while accessing the internal vaginal area.

Puberty and menarche

At puberty, ovarian function is normal so pubertal and breast development should occur as expected. Breast development signifies the "turning on" of the hypothalamus-pituitary-ovarian axis. Estrogen is produced at the ovaries, stimulating breast development and growth, as well as endometrial stimulation in the uterus. The earliest secondary sexual characteristic in most girls is breast development (thelarche). A 1997 paper written by Herman-Giddens et al described the timing of pubertal development in >17,000 healthy young girls in the United States. The mean ages for the onset of breast development were 8.9 years in African-American girls and 10.0 years in Caucasian girls.[12]

Adrenarche (hair growth) is triggered by the function of the adrenal gland so is unaffected by Müllerian or ovarian development. Despite the earlier onset of breast and pubic hair development, the onset of menses or menarche has remained stable over the past 25 years at 12.0-12.5 years in the United States.[13] Therefore, a window of opportunity exists to prevent the accumulation of obstructed menstrual fluid if the reproductive anatomy is not patent. Generally, the period from the onset of breast development until menarche is from 1 to 3 years, which is the ideal time for evaluation. However, the timing of menarche can be affected by several factors, including family history, ovarian function, development of the uterine structure, and the persistence and patency of the reproductive tract. It is important to inquire about the onset of menses in pubertal females, especially if a reproductive anomaly is known to be present. Young women with a previous cloacal anomaly have been reported to experience an obstruction to menstrual flow in 36%-41% of patients.[6,7]

Effective menstruation requires adequate uterine development, with the presence of endometrium within the uterine body and a patent outflow tract. The cervix and vagina must be patent to allow the menstrual flow without obstruction. In some patients with cloaca, as many as 20% of patients experienced amenorrhea because of the absence of or the underdevelopment of the Müllerian structures.[7] Underdeveloped structures may have variable amounts of endometrium, but if present, require an adequate outflow tract. Confirmation of the patency of the reproductive tract before menarche is important to avoid obstruction, pain, and risk to reproductive organs and fertility.

Methods to assess patency of the reproductive structures include evaluation of the presence of mucus at the ectocervical region on vaginoscopy and antegrade/retrograde perturbation of the Müllerian structures during any open or laparoscopic abdominal procedures. Such assessments can and should be combined with other surgical intervention and coincident anesthesia during childhood or early puberty. During vaginoscopy, perhaps at colostomy closure or creation of an appendicostomy, documentation of the presence of mucus at the outer part of the cervix is reassuring. Mucus is produced within the endocervical canal and may pass through the cervix and into the vagina if the cervix is patent. As previously described, saline perturbation can be performed in an antegrade manner by cannulating the distal fallopian tube and instilling saline. Visualization of the egress of the saline from the vagina can confirm patency of the Müllerian system. During laparoscopic procedures, retrograde perturbation is the procedure of choice because the distal fallopian tube cannot be adequately compressed for antegrade perturbation, making definitive confirmation of patency impos-

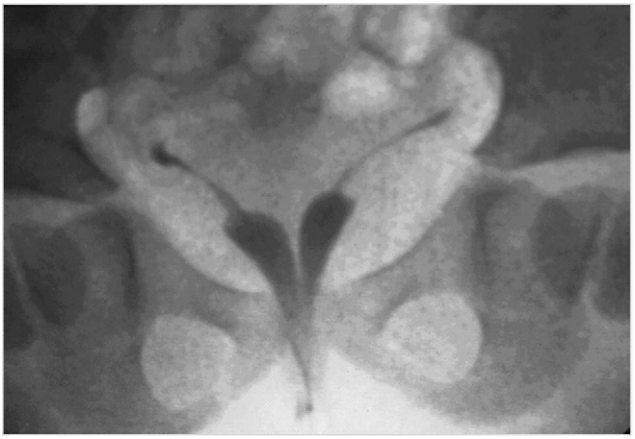

Figure 5 If the uterine anatomy remains unknown into adulthood, an hysterosalpingogram (HSG) can be performed.

sible. Cannulation of the cervix/cervices is necessary, followed by perturbation in a retrograde manner. If young women reach puberty without such an assessment, retrograde instillation of contrast material using fluoroscopy can provide beneficial information (hysterosalpingogram) (Figure 5).

Recommended management of females with an ARM begins with delineation of the reproductive anatomy as completely as possible during infancy. Parents and other members of the medical team should be counseled regarding expectations and precautions. Ultrasound surveillance of the reproductive structures should begin 6-9 months after the onset of breast development and continue every 6-9 months through menarche. The actual images should be reviewed with special attention to endometrial thickness in each uterine body. If an obstruction to menstrual flow is detected by visualization of a thickened endometrium with hematometra and/or hematocolpos, medical intervention should be initiated immediately to minimize adverse sequelae.

Hormonal suppression of menses and endometrial stimulation prevent continued accumulation of obstructed menstrual products. It allows symptom relief, resolution of inflammatory changes, and increases the potential for salvage of reproductive structures. After inflammatory changes have resolved and retained menstrual fluid has resorbed, surgery will be necessary to establish an adequate outflow tract.

Other gynecologic concerns for pubertal females with an ARM include the development of adnexal cysts, hydrosalpinges, endometriosis, and chronic pelvic pain. The risk of endometriosis significantly increases with an obstruction of the Müllerian system, but may also be seen in patients with congenitally displaced endometrium. Chronic pelvic pain may be related to the development of endometriosis or pubertal stimulation of chronically adhesed reproductive anatomy.

Reproduction

Even when their daughter is only an infant, reproductive potential is already on the minds of parents. Parents are anxious to learn if their daughter will be able to have normal sexual intimacy and carry a pregnancy as an adult woman. Most young women who were born with ARM enjoy healthy sexual relationships and may be able to become pregnant.[14,15] Warne et al[7] reported that 12 of 21 (57%) adult cloacal have had vaginal intercourse. In our institution, we are following up nearly 500 females with a cloacal anomaly as they reach adulthood. Continued follow-up is necessary to fully appreciate the sexual outcomes in this population.

A complete physical examination after puberty is necessary to evaluate secondary sexual development and determine the adequacy of the reproductive anatomy for sexual intimacy. Examination of the introital area should be done to assess for possible stenosis. The perineal body should also be examined with attention to the quality of the repair, the presence of scarring, and the size (Figure 6). An inadequate perineal body may be problematic for sexual intimacy or vaginal delivery of an infant. A review by Warne et al[7] reported that 4 of 21 (20%) adult patients who underwent cloacal repair required vaginal surgery to facilitate comfortable sexual activity.

Even if the introitus is adequate for menstrual flow, it may be inadequate for intercourse. The vaginal introitus should be able to accommodate at least a 24-26 size Hegar dilator for comfortable sexual activity. Many young women can perform self-dilation using vaginal or Hegar dilators, to stretch a small band of scar that may preclude comfortable sexual relations. In cases of more significant scarring, an introitoplasty may be necessary. This is an outpatient procedure in which the vaginal introitus is circumferentially mobilized to excise the scarred segment and exteriorize the soft pliable upper vagina (Figures 7A and 7B). Preopera-

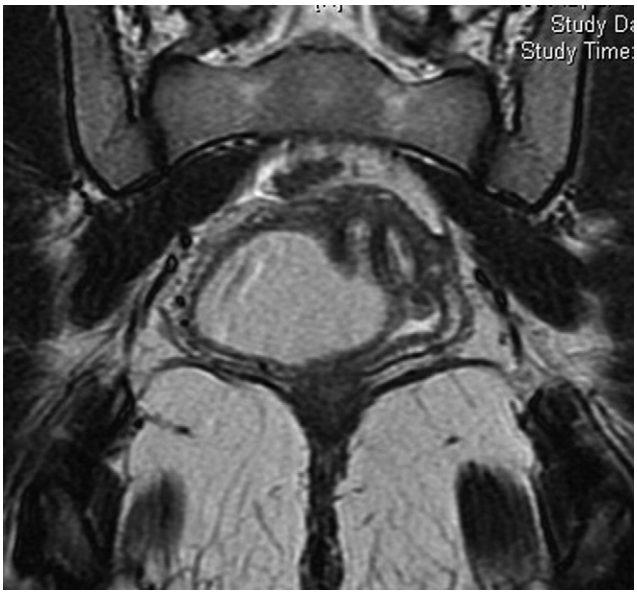

Figure 6 The magnetic resonance imaging view of an obstructed right hemivagina in a patient with a history of a perineal fistula and a duplicated reproductive system. The 2 hemiuteri are seen deviated to the left side of the patient with the enlarged, obstructed vagina to the right.

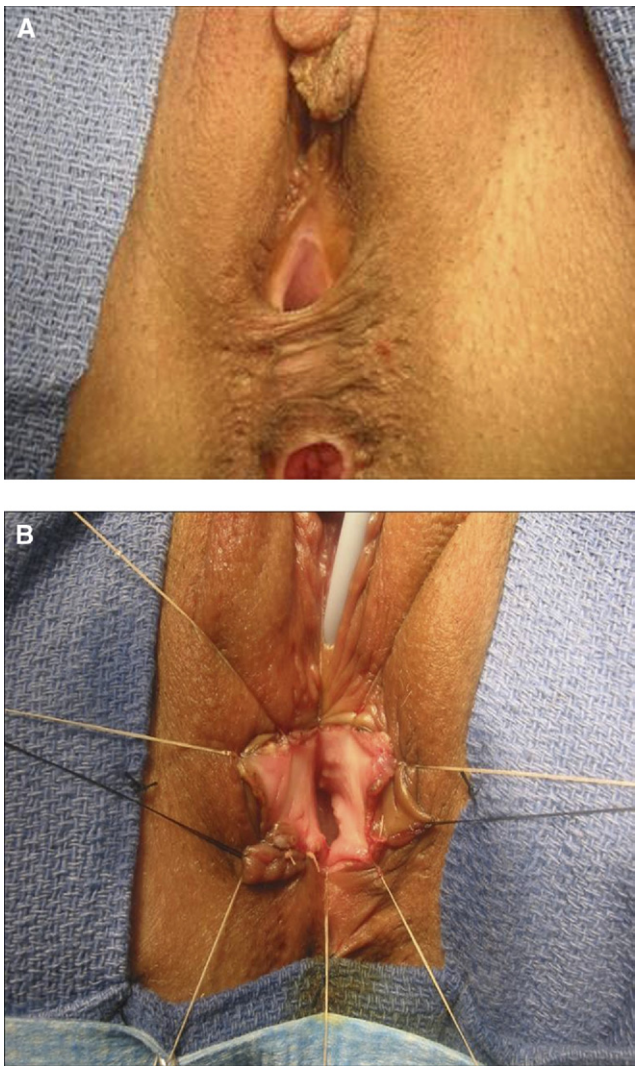

Figure 7 (A) Perineal photo of a 14-year-old female who had previously had a cloacal repair. The introitus is small and obstructed by scarring at the perineal body. (B) The final sutures are placed at the completion of the introitoplasty in this patient. (Color version of figure is available online.)

tively, an assessment of the degree of stenosis, including the amount and depth of scarring and the adequacy of the perineal body is essential. If the segment of stenosis is too thick and/or an inadequate segment of vagina exists above it, consideration of distal vaginal replacement may be indicated. Such a procedure requires a full bowel preparation and may include an abdominal exploration for a bowel graft or the use of a rectal patch. If the perineal body is too short to accommodate a larger vaginal introitus, a repeat anoplasty may be necessary to reposition the anus more posteriorly. Anal mislocation is a common indication for reoperation, even in younger patients, which also lengthens the perineal body.[16] It is imperative to know how much the patient relies on the sphincteric placement of the rectum for bowel control. Posterior relocation may not be an option for young women who have bowel control and rely on proper position within the sphincter to be clean and dry. Often short-term vaginal dilation may be necessary postoperatively.

Vaginal reconstruction is necessary in cases in which the native vagina is congenitally absent or unable to be mobilized to reach the perineum at the time of surgical reconstruction. It is always preferable to retain native vagina whenever possible; however, other tissue sources have been used to construct a functional vagina. Our group reported on 131 cases of vaginal replacement using a segment of bowel to replace some or all the native vagina.[17] Several complications were noted in the retrospective review, including mucus production, stenosis, neovaginal prolapse, and fistula formation. The most common complication was introital stenosis, affecting 14% of all cases. Mucus production was reported only in patients with a rectal neovaginal segment.

Pregnancy is possible in women born with an ARM, even in the most complex ARM, cloaca.[14,15] In adult women born without any associated reproductive anomaly, pregnancy rates and outcomes should be similar the rest of the population. In women with an associated reproductive anomaly, the specific type of anomaly will influence the potential for conception and pregnancy risks (Table 1). A unicornuate uterus refers to a single uterine horn. A uterine didelphis describes 2 uterine horns, each with a cervix, usually contiguous in the midline. In females with 2 hemiuteri and 2 cervices, with 1 on each side of the pelvis, it is not clear whether comparison to the uterine didelphys or the unicornuate uterus is most appropriate. Finally, a bicornuate uterus refers to a bilobed (or heart-shaped) uterine body with a single cervix.

Recommended obstetrical management begins before pregnancy, with a preconceptual counseling visit with a maternal fetal medicine physician. Such consultation allows a review of previous operative procedures, determination of the known reproductive anatomy, and evaluation for concurrent medical issues, such as the development of renal insufficiency, which may influence plans for pregnancy. Awareness of the necessity for intermittent catheterization is also important because the anatomic distortion of an enlarging gravid uterus may preclude reliable access through an abdominally placed continent conduit, with the potential need for long-term indwelling catheterization until delivery. During pregnancy, patients should also be followed up closely for signs or symptoms of preterm labor. Uterine anomalies increase the risk for preterm labor, preterm delivery, and abnormal presentation. Decisions about mode of delivery should be addressed individually, with consideration of previous surgeries, the presence of any

Table 1 Conception and pregnancy risk potential in women born with an associated reproductive anomaly[14]

	Spontaneous pregnancy loss	Preterm delivery	Term delivery	Live birth
Unicornuate uterus	36%	45%	45%	54%
Bicornuate uterus	36%	23%	40%	55%
Uterine didelphus	32%	28%	36%	56%

stomas, and repair of the ARM. As a general recommendation, women with a history of an imperforate anus and a rectovestibular or rectoperineal fistula are candidates for a vaginal delivery. The adequacy (size and scarring) of the perineal body should be assessed by the obstetrical provider before making that decision. Women who had a history of a cloacal anomaly should be delivered by Cesarean section. This is especially important in women who may have had a long common channel and required an extensive surgical repair for good functional outcome. Also, patients who have a neovaginal replacement or interposition graft require Cesarean section to avoid irreparable damage to the graft. The obstetrician should consult with a pediatric surgeon knowledgeable about cloacal reconstruction to discuss the approach. Intraoperative consultation may be beneficial to protect continent conduits, like a Mitrofanoff or an appendicostomy, and the essential vascular supply of these structures or even the gastric, ileal, or colonic segment of a bladder augmentation.

Knowledge of reproductive-related issues in females with ARMs allows the pediatric surgeon to provide optimal surgical management in infancy, childhood, and into young adulthood. Collaboration with a gynecologist is essential. Appropriate counseling for patients and families about potential reproductive concerns that may develop many years after the definitive surgical repair allows preparation and planning to preserve future fertility.

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