

Gynecologic Concerns in Patients Born with Anorectal Malformations

Author: [Lesley Breech, MD](#)

Associate Professor Pediatrics, Obstetrics and Gynecology and Surgery

There are several important gynecologic and obstetric concerns that girls and young women with [anorectal malformations](#) may have. Unfortunately, at this point in time many unanswered questions remain. We are learning more everyday about these issues and with collaboration between patients, families and providers, we can help subsequent patients by continuously sharing what we've learned. The following is a summary of what we know now.

Delivery

At delivery, determination of gynecologic abnormalities is not urgent because bowel and urologic problems can be serious or even life-threatening. Gynecologic issues may not seem quite as important immediately, yet delineating the reproductive anatomy can be very important to prevent problems in the newborn period or with pubertal development.

Sixty percent of female cloaca patients have some degree of septation, (division or separation) of the uterus and/or the vagina. It can be fairly minimal such as a partial septum or a partial division within the vagina, or it can be much more significant with a duplicated vagina and double uterus and cervix (two hemiuteri).

View Videos

[Introduction](#)

[Infancy](#)

[Puberty and Menarche](#)

[Sexual Function](#)

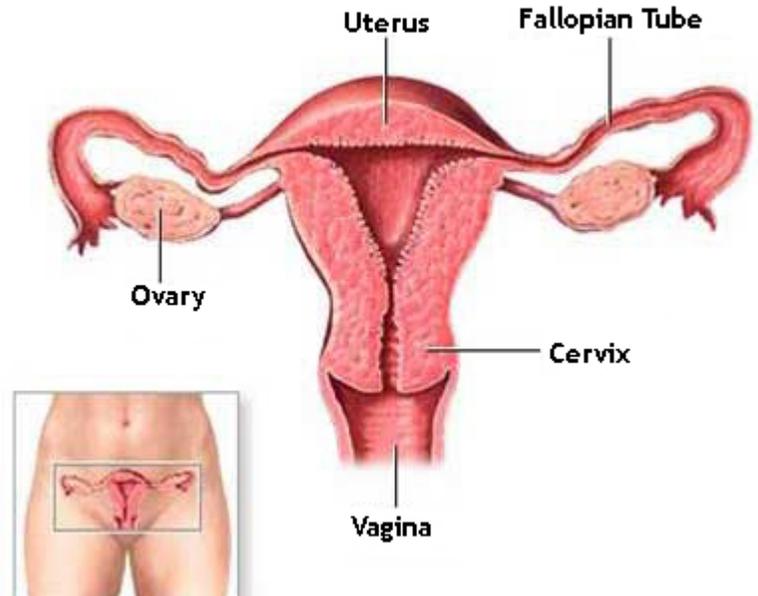
[Obstetrics](#)

[Frequently Asked Questions](#)

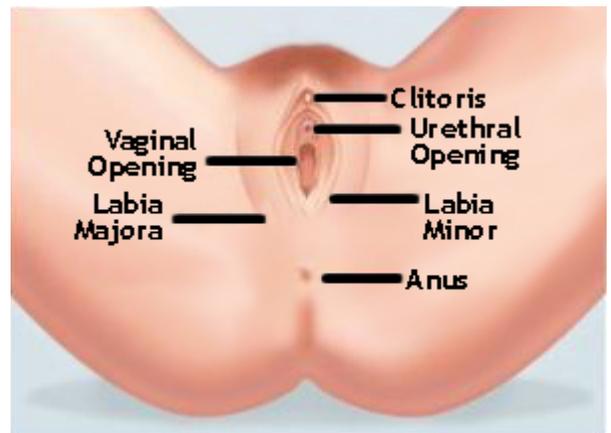


Genital tract anomalies are less common in conditions such as an **imperforate anus with rectoperineal or vestibular fistula (add link)**. Historically, an assessment of the reproductive structures was not included in the repair of less complex anorectal malformations, so the incidence of reproductive abnormalities may be falsely low. Vaginal septa occur in patients with perineal or vestibular fistula in about three percent of cases. If identified, the vaginal septum (divider) may be removed during the initial surgery.

The internal female structures: uterus, fallopian tubes and the upper part of the vagina grow and develop at the same time as the urologic system and gastrointestinal systems. Consequently, any problem in embryologic development in either the urologic or gastrointestinal system could also affect the reproductive system.



The Mullerian ducts exist on either side of the embryo and migrate down into the pelvis. The ducts meet in the middle and fuse to form one midline uterus. In patients with an abnormality in development, one hemiuterus with an attached fallopian tube may develop on each side. Canalization occurs to create the central portion of the uterus, the cervical canal, and the vagina. Finally, the endometrium (uterine lining) grows, which completes uterine development. The endometrial lining, which develops each month and sheds as the menses (period), requires an open tract to avoid blockage of flow. At the time of puberty and menarche (the onset of periods), the patency of the reproductive structures should be considered.



Importantly, the ovaries develop from a different embryologic origin than the fallopian tubes, the uterus, and the upper vagina, so they remain unaffected. Hormone production, (estrogen or

estradiol) at the time of puberty, as well as the capacity to release the genetic material (the egg) should be normal.

The external genitalia are affected in patients with a cloacal anomaly. Externally, young women with a cloaca will have only one visible opening on the perineum, which is different than the normal female perineum. Normally, each system (urologic, the gynecologic, and the colorectal) exits separately on the perineal skin. The urethra and vagina are located right next to each other, and the rectum is separated from these by a bridge of skin called the perineal body.

Eventually after surgical separation of all three systems, each opening will be present, a urethra, a vagina and the rectum / anus. Surgical attempts are made to make the openings and perineum aesthetically pleasing to patients, but since surgery is performed early in life, occasionally some surgical intervention may be necessary later. Discussions about the appearance of the external genitalia are sensitive and require a special relationship between a patient and her pediatric surgeon and gynecologist.

In patients with cloacal extrophy, the pubic bone, mons pubis (a fatty layer overlying the pubic bone), or clitoral area may be more affected. The pubic bones could be separated or there can be atypical development of the mons pubis which increases the likelihood of abnormal development of the clitoris. The clitoris is responsible for the majority of sexual sensation. If clitoral development is impaired, concerns about sensation or the potential for orgasm in the future may exist.

It is imperative to accurately determine the reproductive anatomy. The initial time to accomplish this is during the definitive surgical repair. The reproductive structures can be visualized and the patency confirmed if the surgical repair requires an incision on the abdomen. If, as is the case for the majority of cases, the repair is accomplished exclusively through a posterior sagittal approach (which is through an incision along the buttocks and does not enter the abdominal cavity), a complete assessment is impossible. If the child has a colostomy, the colostomy closure is the next opportunity to be able to assess the internal gynecologic structures. Later in life an examination under anesthesia of the external perineum and the vagina and cervix (cervices) can be performed with vaginoscopy (a telescope which allows visualization with distension of water).

Gynecological issues to consider may be divided into several categories:

1. Infancy
2. Puberty

3. Sexual intimacy
4. Obstetrics

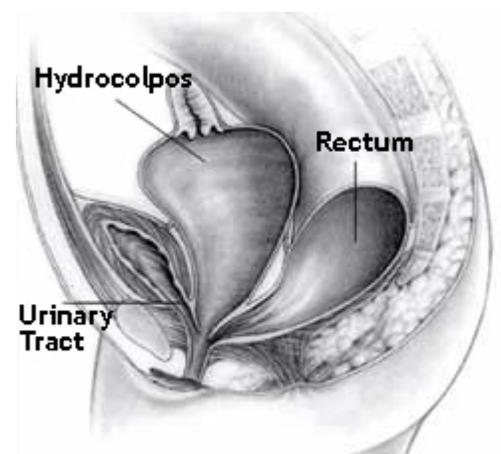
It is easiest to consider the reproductive concerns chronologically in a girl or young woman's life. The major newborn concern is the development of hydrocolpos (which is a very large fluid filled vagina). Menarche or the onset of menstrual periods is the next event with the potential for problems. Sexual intimacy should be discussed after puberty and before the initiation of sexual activity. Lastly, obstetrical issues should also be considered as young women become sexually active and consider childbearing.

Infancy

Hydrocolpos is the collection of fluid within the vaginal space. Hydrocolpos is more likely to occur in patients with a complex cloacal anomaly. As the vagina fills it can compress the ureter causing hydronephrosis of the kidney which can be dangerous. Drainage of the fluid from the vagina with an indwelling tube until the definitive surgical repair (vaginostomy) is the usual solution to the problem; however sometimes, it may be necessary to drain the urine directly from the urinary system (vesicostomy), bypassing the blockage caused by the distended vagina. Following repair of the cloaca, the rest of early childhood is just like every other little girl's childhood from a gynecologic standpoint.

Puberty and Menarche

At puberty, because the ovaries should be functioning normally, pubertal and breast development should occur as expected. The earliest secondary sexual characteristic in most girls is breast development (thelarche). A 1997 study written by Dr. Herman-Giddens and colleagues from the Pediatric Research in Office Settings Network defined the timing of pubertal development in more than 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 white girls. Adrenarche (hair growth) is triggered by the function of the adrenal gland so this also progresses normally. The age of menarche (the first period) has remained relatively stable over the past 20 years at 12.0 to 12.5 years in the US. Therefore a window of opportunity exists to prevent the accumulation of obstructed menstrual fluid if a girl's gynecologic anatomy is not patent (open). Menarche will depend upon several factors



and generally occurs 1 – 2 years after the onset of breast development. The time between breast development and the onset of periods is the ideal time for evaluation.

Menstrual Periods

There are several important issues to consider. First, is the consideration that menarche may be influenced by genetic factors. If a young woman with a cloacal anomaly is 15 years old, and has not started periods; her provider may become concerned. However, consideration of the age of menarche of other first degree relatives (mother, sister) may be helpful and reassuring.

Second, the onset of periods relies on the production of hormones by the ovary. Most patients with anorectal malformations, have normally developed ovaries. It is unlikely that the ovaries will have any difficulty producing estrogen. If one ovary was surgically removed, the remaining ovary will take over and make enough hormone for the entire body and release eggs (genetic material). One ovary has enough genetic material to allow children for the future. Sometimes a surgeon or physician may think that ovarian tissue was removed yet there still may be enough of a remnant to produce breast development. A portion of an ovary is all that is necessary.

Next, the inner lining of the uterus (endometrium) is stimulated to produce menstrual bleeding. The absence of menses can occur in a significant number of patients with cloacal anomalies due to the absence of a uterus or an under-developed uterus. Many young women may have two under-developed hemi-uteri instead of one fully developed uterus. An under-developed uterus could have very little endometrium or even none at all. If there is no endometrium and only uterine muscle is present, menstrual flow will not occur because the endometrium is what constitutes the menstrual flow. It is very difficult to make this determination until puberty, so it is recommended to undergo an ultrasound of the pelvis within about 6 months after the onset of breast development. This allows determination of whether the lining is beginning to develop or if the lining is present at all seen on ultrasound as a "uterine stripe". Ultrasound is not invasive or painful, so it is an easy way to evaluate the reproductive structures.

Lastly, the patency of the reproductive tract is essential for menstrual blood to drain. The cervix (opening to the uterus) and the vagina must openly communicate to allow flow of the menses. Occasionally there could be a well developed uterus and cervix that appears to be patent; however, if not attached to an open, adequate vagina, obstructive complications may occur. This may manifest as monthly pain on either the right or left side. Optimally communication is established prior to the development of a significant obstruction of menstrual fluid and severe

pelvic pain. If adequate vaginal tissue is not available, the cervix and uterus may be attached to a vaginal replacement to allow a patent outflow tract to the skin.

Vaginal replacements may be created using a variety of tissue including segments of small bowel, large bowel, rectum, or even skin or bladder. This allows a connection for the menstrual flow and the potential for sexual intercourse. Continued gynecologic follow-up after menarche is important because progressive discomfort or pain with periods (dysmenorrhea) can be suggestive of blockage of only part of the reproductive tract. This could also be demonstrated on [pelvic ultrasound](#) or an [MRI](#). Accumulation of menstrual fluid in the obstructed portion of the uterus, tube, or vagina could cause pain and necessitate removal.

If a patient has two hemi-uteri, will she have two menstrual periods each month?

The ovaries respond to signals from the brain in all females, therefore only one ovary will ovulate (release an egg) and trigger one menstrual period. Both hemiuteri respond to the same signal and the shedding of both endometrium occurs simultaneously. Menstrual periods may be a day or so longer, but otherwise normal. Irregular bleeding after menarche is caused by the same diagnoses as other adolescents without anorectal or reproductive issues.

Is the patient at risk for any other gynecological problems?

After puberty, young women may develop cysts (fluid collections) in the pelvis. Cysts often occur on one side of the pelvis and may be quite painful. The fluid collections may originate from the an ovary or the lining of the pelvis itself. Hormonal stimulation is felt to play a role in the development of cystic structures. Pelvic cysts are more common in young women after repair of cloacal extrophy. Hormonal suppression or x-ray guided drainage are the standard treatment for these cysts.

Endometriosis is a gynecologic condition in which the lining of the uterus (endometrium) is found outside the uterus. It is more likely to occur in patients with a history of retrograde (reverse) menstrual flow, as in cases of obstruction to menstrual drainage. Chronic pain and discomfort, in addition to possible effects on fertility, are the major concerns with this condition. Lastly, chronic pelvic pain has also been described and may be attributed to the development of scarring after numerous surgical procedures.

Sexual Function

Comfortable, satisfying sexual intimacy is a goal for all women. Sensation of the external genitalia is primarily controlled by nerves located in the clitoral region. Clitoral development is usually unaffected in patients with anorectal malformations; sexual satisfaction and orgasm are expected to be normal. Sometimes at the time of the definitive repair the area of the clitoris can be cosmetically optimized. In patients with a complex cloacal anomaly, anatomic positioning of the clitoris and vagina may require physical adjustments for full sexual satisfaction or orgasm with vaginal intercourse.

If vaginal development is incomplete or preservation of the natural vagina cannot be accomplished surgically, a vaginal replacement (neovagina) may be necessary. Goals of creation of a neovagina include the creation of a structure of adequate size and position to accommodate sexual intercourse, the production of a "normal" amount of secretion and lubrication, and the necessity for minimal care for maintenance. Sometimes patients require additional minor outpatient vaginal surgery after puberty to facilitate comfortable intercourse. Even if menses occur without difficulty, the vagina should still be evaluated by a gynecologist familiar with the patient's history and experienced in reconstructive surgical procedures.

The initial surgery is performed to separate the colorectal, urologic, and gynecologic systems, so revision of the vaginal opening at the time of the onset of sexual activity may be indicated. Some patients may desire minor revisions of the anatomic appearance of the external genitalia. Growth and development of the genitalia continues well into puberty, so consideration of possible surgery should be reserved until the completion of puberty. This is possible if it will not influence the function of the area. The goal is a good functional outcome for young women including fecal and urinary continence and a satisfying sexual life. Interaction with a trained, experienced psychologist may also be beneficial when discussing such personal components of sexual life.

Obstetrics

Unless unusual scarring occurs after definitive surgical repair, fertility issues are usually not the primary concern. As previously described, the ovaries, containing the genetic material (eggs), develop normally. Proper communication with the fallopian tube is necessary for fertilization. Adequate development of the uterus, with an endometrial lining, is necessary for implantation of an embryo. Sufficient uterine muscular development is also important to carry a pregnancy to a viable gestational age. The implications of a poorly developed cervix are at present unknown.

Limited data is currently available; however, a number of women born with cloacal malformation have delivered healthy babies, even with hemiuteri.

What problems may she be at risk for during pregnancy?

Patients may develop preterm labor or early delivery. Uterine didelphis (two hemiuteri positioned close together) has the best obstetrical outcome of any major congenital uterine anomaly in patients without a cloacal anomaly. It is desirable to apply this data to women who have two hemiuteri with a cloaca; however, it is important to determine if other factors may influence obstetrical outcomes.

If a patient has two hemi-uteri, is she more likely to have twins?

As in all women, the ovaries respond to the same signal from the brain triggering the release of one egg each month (ovulation). If the egg is fertilized, the embryo will implant in the uterus or one of the hemiuteri. It is the egg release and the fertilization process that determine if a twin pregnancy will develop, so the presence of two hemiuteri will not increase the chance of having twins.

If a patient with an imperforate anus with a rectoperineal or vestibular fistula has been properly repaired, a vaginal delivery may be possible. Examination by an experienced obstetrician-gynecologist (ob-gyn) is important to confirm appropriate anatomic repair and healing of the perineum. With advanced reconstructive surgery, such as in cases of cloacal repair, bladder augmentation, bladder neck reconstruction, or vaginal replacement, a cesarean section may be recommended. Because of the complex nature of the reconstructive procedures performed during infancy, the urologic or gastrointestinal system may be in jeopardy during a cesarean section; therefore, a planned delivery by a multidisciplinary team, including an ob-gyn experienced in pelvic reconstructive surgery, is strongly recommended. The Colorectal Center for Children at Cincinnati Children's Hospital Medical Center works in conjunction with the [Fetal Care Center of Cincinnati](#) for the care of such patients and is the only center in the world to offer such advanced specialty care.

Management of Reproductive Structures

Observation

Most girls should have undergone an assessment of the development and patency of the reproductive structures in infancy, either during the definitive surgery or during colostomy closure. In circumstances where the anatomy has not been previously discussed with family, detailed information can be obtained by reviewing previous operative records with the pediatric surgeon or gynecologist. Physical examination in the office or under anesthesia (with vaginoscopy) can provide information regarding the vagina and cervix or the external genitalia. Imaging studies (ultrasound) performed after puberty can also provide helpful information. Occasionally, laparoscopy to look at the reproductive structures may be needed.

Although the American College of Obstetrician and Gynecologists recommends all adolescent females age 13 – 15 years have an initial reproductive health visit, young women with a history of an anorectal malformation should have an earlier visit. This allows a review of normal pubertal development and a discussion regarding possible gynecological issues to expect. An ultrasound of the pelvis within 1 – 2 years of the onset of breast development is also recommended. Serial ultrasounds provide information about the degree of development of the uterus and endometrial lining and can exclude the presence of an obstructed reproductive tract. Even if mildly obstructed structures are diagnosed, medications can be used to suppress hormonal stimulation and prevent continued accumulation of menstrual blood. Such suppression may increase the chance that reproductive structures may be preserved instead of removed.

Removal

If poorly developed reproductive structures are identified as an infant, especially if patency cannot be confirmed or established surgically, the uterus or fallopian tubes may be resected. If significant obstruction and distension with menstrual blood occurs, most reproductive structures will need to be removed. It seems that if one side of the reproductive system is considerably less developed, it is of great risk for obstruction and complications; and thus removal of the abnormal side is recommended.

Reconstruction

From a gynecological standpoint, establishment of adequate communication between the uterus (hemiuteri), vagina, and perineum is the main goal. When the natural vagina is not adequate, a



vaginal replacement (neovagina) can allow the proper outflow of menstrual blood and provide an adequate space for vaginal intercourse. Minimizing the risk of an obstruction of the gynecological system remains a priority.

Conclusion

Gynecologic concerns are particularly important at several intervals: infancy, puberty and the onset of menses, the debut of sexual intimacy, and pregnancy. Definition of reproductive anatomy provides the key to appropriate counseling and preparation. We continue to learn more daily as we care for a growing population of young adult women born with anorectal malformations.

Contact the Colorectal Center at Cincinnati Children's

For more information or to request an appointment for the Colorectal Center at Cincinnati Children's Hospital Medical Center, please [contact us](#).