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Gynecologic concerns in patients with anorectal malformations

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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