Uterovaginal Anomalies: Diagnosis and Current Management in Teens

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Introduction
Genital anomalies in the adolescent population are a challenging group of conditions. Not only does the understanding of normal embryologic development essential but also the appreciation that her immaturity and budding sexuality will influence the nature and success of her treatment is important. The ideal care for these young females requires a team approach of both medical and supportive professionals. Normalizing her reproductive tract surgically and supporting her psychologically can usually be accomplished and, in most situations, reproductive capacity is preserved.

Embryology
To understand the nature and complexity of these congenital malformations, a basic understanding of embryology of the organs involved is needed [1]. In humans, gender is established at fertilization, but the gonad is initially undifferentiated. The Y chromosome harbors the gene that induces testicular development, called testicular determining factor (TDF), and is located in the gender-determining region on the small arm of the Y chromosome (SYR region). TDF acts on the undifferentiated gonad as an organizing agent to promote testicular development, which leads to the subsequent differentiation into both Leydig and Sertoli cells. By the eighth week, the Leydig cells elaborate testosterone and the Sertoli cells produce müllerian-inhibiting factor (MIF). Gender differentiation is dependent on the presence or absence of testosterone and MIF.

The urogenital system is composed of two separate but interrelated organ systems: the urinary excretory system and the genital system of reproductive organs and ducts. Although these two systems serve diverse functions, they are intimately related embryologically, and any defect in one component often affects the other. In addition, the cloaca, an endodermally lined chamber into which the primitive gastrointestinal tract empties, and the urogenital sinus contribute to the formation of the rectum, vulva, and lower vagina.

By the eighth week, the testes produce testosterone from the Leydig cells, which stimulates the mesonephric (wolffian) duct and masculinizes the external genitalia. The testes elaborate müllerian-inhibiting factor from Sertoli cells, which suppresses paramesonephric (müllerian) duct development. Embryos without a Y chromosome, as found in the normal 46 XX female embryo, do not produce TDF, testosterone, or müllerian-inhibiting factor. In this situation, the gonad develops into the ovary, and it is recognized as such by the 10th week.

In the undifferentiated stages of early development, both male and female embryos have two pairs of rudimentary sexual ducts: the mesonephric duct (wolffian duct), which drains the primitive mesonephric kidney, and its lateral counterpart, the paramesonephric (müllerian) duct (Fig. 1). The wolffian ducts play an essential role in the male reproductive system, becoming the epididymis, vas deferens, and ejaculatory duct, but are usually only recognizable in the female as vestigial remnants. The müllerian tract of mesodermal origin is the critical embryologic structure that develops into the internal genitalia of the female. These paired bilateral müllerian ducts pass caudally, parallel to the mesonephric duct. In the pelvis, they cross ventral
to the mesonephric duct and grow medially and caudally to contact and fuse with their counterparts to form a Y-shaped uterovaginal bulb or tubercle that abuts with the urogenital sinus, which is the anterior portion of the primitive, but now divided, cloaca. The divided cranial portion of the müllerian duct forms the fallopian tubes and ostium, and the caudal fused portion, the uterus, cervix, and the upper vagina. The primitive lower vagina is derived from the endodermal urogenital sinus, which is in direct contact with the uterovaginal tubercle. The embryologic vagina develops from paired endodermal outgrowths, called sinovaginal bulbs, which fuse to form a solid vaginal plate that later breaks down to form a single vagina. The lumen of the vagina is separated from the urogenital sinus by a membrane called the hymen, which normally ruptures in the late perinatal period. It remains a distinct entity as a fold of mucous membrane surrounding the entrance to the vagina [2].

### Psychological Issues in Genital Tract Malformations

Adolescents diagnosed with genital-tract malformations are at psychological risk and ideally should be referred to an appropriate counselor, be that a psychiatrist, psychologist, or social worker as part of their care [4••]. Psychological supports vary based on the severity and consequences of the malformation. Immediate reasons for referral usually relate to the immaturity of the patient and her ability to fully cooperate with medical treatment. However, it is the long-term physical and psychological impact of the diagnosis and treatment that could affect both sexual and reproductive function that is of prime concern to the medical team and her family. The adolescent’s perception of the genital anomaly might differ from that of her parents. The definitive diagnosis usually occurs in the adolescent period when the patient is between 10 and 17 years of age, a developmental time associated with wide swings in maturity and understanding of psychosexual function [5]. Events that are perceived as far in the future and are not clearly understood have far less meaning to the patient than immediate worries, such as being different from her peers or embarrassment from the medical attention focused on her reproductive organs. Factors such as culture, the degree

### Classification of Reproductive Tract Malformations

Alterations in the normal sequence of development due to genetic, hormonal, vascular, or other causes can lead to a wide spectrum of reproductive tract abnormalities. The American Fertility Society has a detailed classification system [3•]. Clinically, however, the reproductive malformations can best be appreciated based on simple embryologic principles (Table 1). Lack of development or lateral or vertical fusion defects, which might or might not communicate, can help the clinician devise an appropriate management strategy. Serious reproductive tract abnormalities in the female are rare (ie, <1%); however, when they present themselves at puberty masked as pelvic pain, a pelvic mass, or primary amenorrhea, they can be very disturbing and have potentially serious consequences if not dealt with expeditiously and competently.

### Table 1. Classification of female genital-tract abnormalities

<table>
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<th>Aplastic or hypoplastic abnormalities</th>
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<td>Lateral fusion abnormalities</td>
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<td>Other abnormalities</td>
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<td>Imperforate hymen</td>
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<td>W olffian duct remnants</td>
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Figure 1. Embryology of the female urogenital tract.
of openness in the family, and the way in which the family deals with problems can influence her understanding and acceptance of diagnosis and proposed treatment. Her grasp of the issues will change with time, and as she matures she will become more concerned over relationships and eventually her own reproduction.

The psychological concerns of parents cannot be overestimated. Parents often express a great deal of guilt, despite reassurance. They grieve the loss of the normal transition into sexuality and reproduction for their daughter. They have an immediate understanding of potential life-long consequences that the early adolescent might not share. They also have to cope with diagnosis and treatment while supporting their child. Psychological referral for parents, especially at the time of diagnosis of a severe anomaly, can be very helpful, not only for them but for their affected teenager and the medical team. Counseling initiated after diagnosis can clarify the expectations, thus ensuring smooth, informed family functioning and cooperation with the treatment protocols. It might be helpful to introduce the patient and her parents to another family who have gone through a similar circumstance, knowing that they are not alone can be great comfort and provide valuable support and guidance. Denial and rebellion are common in this age group. Meeting others decreases the sense of isolation that many patients and their families experience.

Ideally, the counselor, preferably a woman, should be a member of the team who cares for these patients. She should have a working knowledge of the specific anomaly as well as experience with adolescent developmental issues and family dynamics. The need for open communication between counselor and the medical team is paramount.

Severe genital-tract anomalies can have a lasting impact on the patient and her family. The goal of psychological treatment is to aid adjustment and coping with time. It is tied to the development of the patient, her wishes and desires as she grows, her functioning within the family, and eventually, her own family. The concerns of the 12-year-old are different from those of the 17-year-old and different again from those of the 24-year-old. It is the role of the counselor to help smooth the transition from the adolescent diagnosed with a congenital anomaly into adulthood, a dialogue that often can take years, if done properly.

Genital-tract Abnormalities in the Adolescent

Aplastic and hypoplastic abnormalities

Bilateral agenesis of the müllerian duct

Patients with congenital absence of the vagina, cervix, and uterus are classically described as having the Rokitansky-Kuster-Hauser syndrome [9]. The patient presents with normal secondary sexual development and primary amenorrhea. The müllerian ducts failed to develop, but the ovaries are anatomically normal and physiologically functional, accounting for the normal appearance of these young women. Interestingly, there is usually a dimple of a vagina formed from the urogenital sinus, but the rest of the genital-pelvic organs derived from the müllerian duct or genital tubercle are absent or rudimentary. Surprisingly, fallopian tubes can usually be found coming off bilateral atretic uterine horns, which are attached to the pelvic peritoneum. Occasionally, one or both of these hypoplastic primitive uteri has a functional endometrium. Not infrequently associated with this condition are renal tract anomalies (eg, pelvic or absent kidney), skeletal anomalies of the spine, and middle-ear problems. This condition might have a genetic etiology [10].

Hyoplastic uterus

Occasionally a hypoplastic uterus is formed that is connected to the contralateral, more normal unicornean uterus. These patients present with pelvic pain and dysmenorrhea, especially if the defect is noncommunicating. The blood is trapped, and during menses, the lack of outflow causes distention of the hypoplastic uterus (ie, hemihematometria) or reflux, resulting in endometriosis. Most worrying is if a pregnancy occurs in an isolated rudimentary uterus from transperitoneal migration of sperm.

Lateral fusion anomalies

Noncommunicating defects

Embryologically, the müllerian ducts unite in the midline to form the uterus, cervix, and upper two thirds of the vagina. After initial fusion in the normal female, the medial portion is reabsorbed. In some individuals, the
medial portion does not disappear, resulting in uterine, cervical, or vaginal duplication, which can be partial or complete. If the lateral duct communicates, no or few symptoms are present in the adolescent. Occasionally, patients with lateral fusion defect either do not fuse, or the medial walls do not reabsorb, and one side remains isolated from the normal patent paired duct. In this situation, functional endometrium causes symptoms from the accumulation of blood. This can be an isolated uterine horn or in a separate, noncommunicating cervix and vagina. These patients present after menarche with severe and often incapacitating dysmenorrhea, pelvic pain, and a fluctuant pelvic mass. The diagnosis can be difficult and often delayed, because the patient appears normal and has functional menstrual bleeding. Urgent care is essential to alleviate the obstruction and to prevent long-term consequences.

Vertical fusion abnormalities

In the normal female embryo, the fused müllerian ducts unite with the urogenital sinus at the genital tubercle. Occasionally this fusion of the upper mesodermal müllerian duct with the lower endodermal urogenital sinus falls short or the sinovaginal bulbs failed to develop. This failure of fusion results in a transverse vaginal separation of the upper müllerian tract from the lower primitive vagina (Fig. 2). The site of this obstruction can occur anywhere along the vaginal canal but is usually encountered between the upper and middle third. The septum can be a few millimeters thick to almost the length of the entire vagina. These patients have a functional uterus and, with a complete transverse vaginal septum, total obstruction occurs, resulting in primary amenorrhea and pelvic pain. Correction of the long vaginal septum can be very challenging. In some patients, a small orifice is present in the transverse septum, and the diagnosis can be made during a pelvic examination when the cervix cannot be visualized. Young patients might present with difficulty using tampons or lack of adequate vaginal penetration.

Other anomalies

Imperforate hymen

The hymenal membrane is a derivative of the urogenital sinus. It is unfenestrated in early intrauterine life but ruptures in the prenatal period and persists as a vascular membrane encircling the vaginal orifice. Failure of the breakdown of the hymen results in an isolated outflow genital tract. Mucus in the postnatal period can accumulate behind the intact hymen as a mucocele or, postpubertally, blood causes a hematocolpos.

Wolffian duct remnants

The vestigial portion of the embryonic wolffian duct can persist as cystic structures that run parallel with and are intimately related to the müllerian duct. Usually, these cysts are small and are of no consequence, but occasionally are seen on ultrasound or can be very large and produce a mass affect. Most commonly noted in the adolescents are paraovarian broad ligament cysts or, on vaginal examination, Gartner’s duct cysts.

Presentation

Most malformations of the reproductive tract are not diagnosed in young adolescents. Only when there is obstruction to the menstrual outflow, pelvic pain, dysmenorrhea, or abdominal or pelvic mass in the reproductive tract is an abnormality diagnosed. Silent bleeding can occur for months to years before causing symptoms. When the diagnosis is entertained or established, the young patient and her family are best referred to a multidisciplinary center.
that has had significant experience with these conditions. Failure to do so leads to inappropriate and often inadequate treatment. The best opportunity for success is the first surgical attempt.

Clinical Management
Supportive care
All patients with genital-tract abnormalities have both a physical and a psychological component to their condition. It is best to treat the patient and her family with a team, which should include a reproductive health psychologist, experienced nurses, and a competent group of well-trained surgeons. The patient and her family are naturally concerned about her body image, sexuality, and future fertility. Before any treatment begins, the surgeon should thoroughly discuss the abnormality and its implications with the parents and, if appropriate, with the young patient. The discussion must be accurate and presented in terms all to understand. An experienced psychologist, comfortable with dealing with young women with reproductive abnormalities, is invaluable to help allay any anxieties. A knowledgeable nurse also lessens the medical concerns and, as a team member, will guide, comfort, and support her before and after surgery. If dilatation of the introitus and the vagina are required, it is critical that the patient be made familiar with these and “practice” before their use becomes essential to the success of the procedure. Also, the patient should meet and have ample opportunity to discuss treatment plans with the surgeon involved. This preoperative supportive care surrounding the patient with reproductive abnormalities cannot be overstated; without dealing with both the physical and emotional side of the condition, the result will be less than ideal. A poor result can have life-long consequences.

Surgical Management
A full discussion of each surgical procedure for all reproductive-tract obstructive malformations is beyond the scope of this paper. Detailed surgical procedures are best learned from the original papers or standard textbooks of reproductive surgery [11, 12]. However, certain principles of treatment are indispensable if the surgery is to have an acceptable and hopefully ideal result. The most important decision to make is whether surgery is truly required and, if so, when would be the most opportune time to perform it from the patient’s perspective. Many anomalies of the reproductive tract are found when the patient was assessed for another condition. If she has no symptoms and no functional disturbance, the surgery should be delayed until the patient is more mature. Surgery too early is often more difficult and can have inferior results because of lack of cooperation of the patient. If, however, the abnormality needs repair in the young patient, the next decision is whether or not the patient or the medical team would benefit from a short delay and the implementation of ancillary therapy. If menstrual outflow obstruction is causing the pain and mass effect, gonadotropin-releasing hormone agonists or oral contraceptives provide the necessary relief of symptoms and time for education of the patient and her family. This hiatus provides the necessary interval to assess the situation fully as well as prepare the teenager for what will be required. The young female can familiarize herself with her own anatomy and if a dilator or mold is to be used, to become comfortable with it. This preoperative time is not wasted because it removes much of the anxiety that naturally would occur postoperatively had not this preparation been taken. If the lower portion of the vagina needs to be anastomosed to the upper vagina, preoperative use of dilators not only permits the patient to become comfortable with the dilator but also is helpful clinically, narrowing the distances involved by compression of the transverse vaginal septum and stretching the lower vaginal mucosa so that the anastomosis is much easier and over a shorter distance [13]. The surgeon and the entire operating room staff should be well prepared. The only consistent finding about congenital malformations is that they are not totally predictable, and each and every patient has to be extensively evaluated before making the surgical plan. The entire team must be prepared for any eventuality. Never should the surgery be rushed, as the best chance of repair is the present one. Some procedures might require staging. In obstructive disorders that are being done vaginally, an ultrasound probe placed on the lower abdomen can be very helpful in visualizing the bladder, and, if accompanied by a metal or firm object in the rectum, both bowel and bladder injuries can be avoided.

Postoperative Care
Most reconstructive reproductive-tract abnormalities encountered in the young adolescent are amenable to surgery. The patient and her family can expect a good result. However, obstructive lesions of the vagina or creation of a neovagina require daily and constant supervision for days, weeks, and months to obtain a functional vagina. Vaginal molds and, subsequently, dilators should be used, and the newly created vagina should be meticulously cared for, or the operative area will contract, hourglass, stenose, or become obliterated. Molds should be left in place for weeks before reverting to frequent dilations. The problem of compliance is a significant issue in this young age group. The pubescent female is often immature, not motivated, and not interested in cooperating to maintain her vagina. In this situation, the female psychologist and nurse can be very helpful. Constant supervision and encouragement are needed for good results. Any lack of diligence, even for short periods, can be disastrous for the patient and her future sexual satisfaction and function.
Conclusions
Fortunately, obstructive reproductive-tract abnormalities are rare. Properly managed, most patients do well, and they and their families can expect a normal sexual and reproductive life. With a few of these abnormalities, such as absent uterus, full reproductive potential cannot be realized, but with advances in reproductive technologies, most of these patients can fulfill their reproductive desires [14,15]. Psychological support for months and years helps the patient accept her abnormality and become a normally functioning adult woman.

References and Recommended Reading
Papers of particular interest, published recently, have been highlighted as:
• Of importance
•• Of major importance
Internationally accepted classification of all müllerian duct anomalies.
A comprehensive review of all congenital malformations by one of the world's most experienced surgeons.
Classic textbook for surgical approaches to müllerian duct abnormalities.