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INVITED MINI REVIEW

Management of Reproductive Tract Anomalies

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About the Author

Dr. Garima Kachhawa is a consultant Obstetrician and Gynaecologist in Delhi since over 15 years; at present, she is working as faculty at the premiere institute of India, prestigious All India Institute of Medical Sciences, New Delhi. She has several publications in various national and international journals to her credit. She has been awarded various national awards, including Dr. Siuli Rudra Sinha Prize by FOGSI and AV Gandhi award for best research in endocrinology. Her field of interest is endoscopy and reproductive and adolescent endocrinology. She has served as the Joint Secretary of FOGSI in 2016–2017.

Abstract Reproductive tract malformations are rare in general population but are commonly encountered in women with infertility and recurrent pregnancy loss. Obstructive anomalies present around menarche causing extreme pain and adversely affecting the life of the young women. The clinical signs, symptoms and reproductive problems depend on the anatomic distortions, which may range from congenital absence of the vagina to complex defects in the lateral and vertical fusion of the Müllerian duct system. Identification of symptoms and timely diagnosis are an important key to the management of these defects. Although MRI being gold standard in delineating uterine anatomy, recent advances in imaging technology, specifically 3-dimensional ultrasound, achieve accurate diagnosis. Surgical management depend on the type of anomaly, its complexity and the proper embryological interpretation of the anomaly and involves multiple specialties; thus, patients should be referred to centres with experience in the treatment of complex genital malformations.

Keywords Genital tract abnormality · Uterine malformation · Mullerian anomaly

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Malformations of reproductive tract result from mal-development or developmental arrest at a critical stage of embryonic development but may also result from genetic mutations or environmental insults during the period of organogenesis [1]. The clinical signs and symptoms and reproductive problems depend on the anatomical distortions, which may range from congenital absence of the vagina to defects in the lateral and vertical fusion of the Müllerian duct system [2].

Although the incidence of female genital tract anomalies is not known in general population, prevalence is around 4–6.9% [3] while in women with infertility, recurrent pregnancy loss, the proportion affected is much higher (15–27%) [4]. Congenital uterine anomalies resulting from abnormal formation, fusion or resorption of the Müllerian ducts comprise 5.5% of an unselected population, 8.0% of infertile women, 13.3% with miscarriage and 24.5% of women with infertility and miscarriage [5].

Management

The clinical presentation varies over a wide range and is related to both the complexity and spectrum of underlying and associated conditions. Knowledge of genitourinary embryology is essential for the understanding, diagnosis and subsequent treatment of genital malformations, and those that generate gynaecological problems in young patients. The clinical presentation and treatment of FGTM are directly related to the anatomical status of the defect, the new European Society of Human Reproduction and Embryology (ESHRE)/European Society for Gynaecological Endoscopy (ESGE) classification of female genital anomalies a clear definition of all types of anomaly is provided, and the anomalies were categorized in well-described classes and sub-classes [6, 7].

Diagnosis

The typical age of presentation is around puberty and is primarily related to the fact that these conditions typically affect the internal reproductive tract and do not affect the appearance of the external genitalia in most of these cases. These girls seek advice for delayed puberty, primary amenorrhoea, cyclical pain abdomen (cryptomenorrhea) or may present with painful menstruation from the onset of menarche that progressively worsens over time [8].

Identification of symptoms is an important key to the diagnosis of an anatomical defect of the female genital tract. USG is helpful in delineating detailed anatomy of abnormality as an essential prerequisite for appropriate planning of surgery. Current advances in ultrasound technology, specifically three-dimensional ultrasound, achieve the same benefits of MRI in being accurate and non-invasive but also offer the following advantages: they are available in the office, they are cost-effective, and they provide immediate results [9].

Magnetic resonance imaging (MRI) is considered gold standard in complicated higher obstructive uterocervical anomalies offering accuracy, but entailing cost, patient discomfort, and inconvenience, as three-dimensional technology is more accessible and more providers become proficient in using it, ultrasound may replace MRI as the new gold imaging standard in diagnosing mullerian anomalies [10]. Laparoscopy is done to confirm the anomaly and to assess the tuboovarian relation and patency if expertise available and preferred over HSG.

Owing to the close association of genital and urinary systems throughout foetal development, approximately 30–50% of patients have associated renal anomalies; the entire genitourinary tract must be evaluated in detail.

Surgical management depends on the type of anomaly, its complexity, the patient's symptoms and the proper embryological interpretation of the anomaly. Some anomalies may require complex surgery involving multiple specialties; thus, patients should be referred to centres with experience in the treatment of complex genital malformations. Most malformations can be resolved vaginally or by hysteroscopy, but laparoscopy or laparotomy is often needed; however, the approach and procedure must be carefully chosen and planned. Finally, if there are fertility problems (recurrent miscarriages or immature or premature deliveries) or breech or transverse foetal presentation, a uterine anomaly should always be excluded.

Hymeneal Defects

Imperforate Hymen is the complete failure of the inferior plate of the vagina to canalize and is noted in approximately 1 in 2000 females. Typically, adolescents present after menarche when menstrual blood trapped in the vagina causes cyclical pain as cryptomenorrhea (hidden menses). After few months, the vagina distends greatly followed by cervix, uterus and tubes and allowing the formation of hematocolpos. In neglected cases, hematometra and hematosalpinx may be encountered (Fig. 1) and may present as acute abdomen with difficulty in urination and defaecation. Retrograde menstruation may lead to the development of endometriosis or rarely tuboovarian masses.

Definitive surgery should take place after appropriate evaluation of the external genitalia that reveals bluish distended bulge at the introitus and a digital rectal examination. USG is required only to confirm obstruction at a higher level.

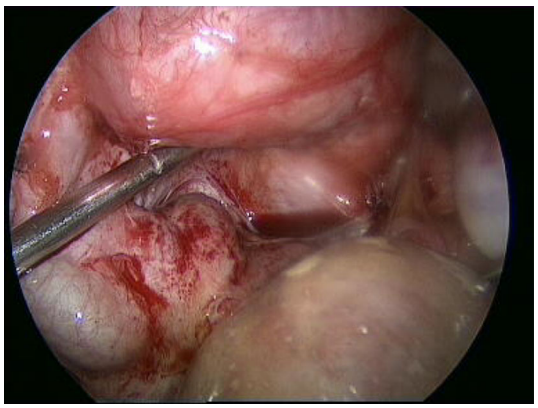

Fig. 1 Laparoscopic view showing hematometra, hematosalpinx and endometriosis

The goal of hymenotomy/hymenectomy is to allow the egress of menstrual flow, tampon use and eventually comfortable sexual intercourse. Simple vertical, T-shaped, cruciform, X-shaped and cyclical incisions may be used, but X-shaped incision has the advantage of reduced risk of injury to the urethra—which should be stented during the procedure [11].

Pressure on the uterus in order to expel more blood is discouraged as it can lead to retrograde flow through the tubes causing endometriosis and tubal adhesions [12]. Hymenal abnormalities can be recognized at birth or neonatal period as hydrocolpos or mucocolpos but asymptomatic girls can be monitored throughout the childhood and ideally surgery in these girls should be done after the onset of puberty.

Vaginal Septum

Transverse vaginal septum causes obstructive symptoms of cryptomenorrhea and is reported with an incidence of 1 in 80,000 females. It can occur at various levels in the vagina but appears to be more common in the upper part (46%), followed by middle (35%) and lower part (19%) of the vagina. The thickness of the septum is usually 1 cm but varies up to 5 cm if present at higher level near the cervix and then it is associated with cervical anomalies.

The diagnosis of transverse septum is suspected when a foreshortened vagina with inability to visualize cervix and presence of hematometra is encountered. MRI is especially useful in identifying whether cervix is present, thereby differentiating a high transverse vaginal septum from cervical agenesis.

The thin septum is incised in the middle of a bulge that may be created with abdominal pressure and the initial incision is stretched to evacuate the old blood. After digital dilatation, the vaginal walls above the septum are palpated and the septum is resected laterally paying great attention to the bladder and the rectum [13].

Resection of a thick septum is technically much harder and requires an experienced, possible multidisciplinary surgical team. For a thick transverse vaginal septum (or for partial vaginal agenesis), the initial opening into the obstructed space may be difficult to create, as the bulge is not palpable. The septum can be negotiated under transabdominal ultrasound guidance. With a very high septum, a combined abdominal/vaginal or laparoscopic approach may be used. Some distension of the upper vagina with menstrual blood before the development of significant hematometra may be advantageous as it allows the potential to increase the available amount of upper vaginal tissue for reanastomosis and may also decrease the thickness of the septum. If there is inadequate mucosa to cover the entire area, then a split-thickness or full-thickness skin graft may be used [14]. Occasionally, a z-plasty can be used to decrease the risk of stenosis. Injury to bladder and bowel are the most common surgical complications.

Longitudinal Vaginal Septum is often associated with complete uterine septum, uterine didelphys and rarely bicornuate uteri. In addition to ipsilateral renal agenesis, associated anorectal anomalies like imperforate anus with rectovestibular fistula may be present. An adolescent with longitudinal septum presents with normal menarche with the need for two tampons and dyspareunia. Pervaginal examination with a speculum is usually adequate for visualization of longitudinal septum with patent vagina and cervix, but a unilateral vaginal and pelvic mass due to obstructed hemi-vagina may be felt. The most common obstructed lateral fusion defects is unilateral obstruction and is most commonly seen in women with complete duplication of the reproductive tract (double uterus, cervix and vagina) [15]. In longitudinal septum, a wide excision of the vaginal septum is done after evaluation of renal system as outpatient surgery, under general or regional anaesthesia. After identifying both cervices, the septum is excised completely just short of cervices, and the resected ends are sutured with small interrupted delayed absorbable material. If the patient also has a uterine septum, it is resected hysteroscopically in the same setting [16].

Vaginal Agenesis (Müllerian Aplasia)

Varying degree of Müllerian hypoplasia or agenesis affects one in every 5000 women and results not only in the absence of uterus and cervix but also upper vagina, leading to complete vaginal agenesis also referred to as Mayer–Rokitansky–Kuster–Hauser (MRKH) syndrome (Fig. 2). Vaginal atresia, distal or segmental vaginal agenesis with normally developed uterus, cervix and

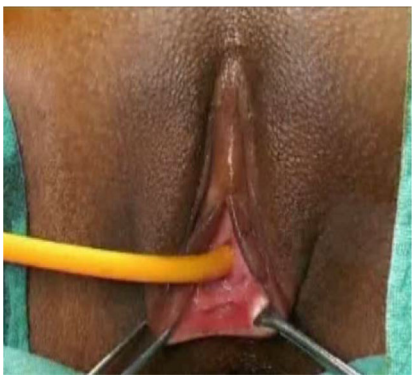

Fig. 2 Vaginal agenesis (Mayer–Rokitansky–Kuster–Hauser syndrome)

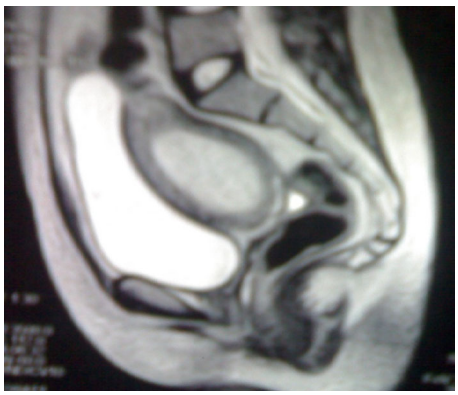

Fig. 3 T2-weighted MR imaging of sagittal view showing uterus with hematometra in a case of cervical agenesis

upper vagina are managed as transverse vaginal septum and vaginoplasty.

MRKH accounts for 10% cases of primary amenorrhoea, and usual presentation is failure to attain menarche with normally developed secondary sexual characteristics with a hymenal ring and a vaginal dimple or shallow pouch up to 1.5 inches deep with complete absence of uterus, cervix and upper vagina. MRKH with functional endometrium is present in 7–10% women, and this subgroup of patients may require excision of rudimentary horn due to cyclic pain after evaluation of upper reproductive organs by imaging.

The goal of treatment is development of a functional vagina for sexual intercourse that may be accomplished either by progressive invagination of the vaginal dimple and gradual dilatation. Although this technique is 85–90% successful, it may take months to create adequate vagina; thus, proper counselling is important [17]. Most commonly modified McIndoe vaginoplasty is done. A canal is created within the connective tissue between the bladder and rectum, which is lined by split-thickness skin graft, human amnion, peritoneum, buccal mucosa or intercede absorbable adhesion barrier. Cutaneous and musculocutaneous flaps are also used to line the neovagina, e.g. Williams vaginoplasty. Laparoscopic Davydov with peritoneal pull through also yields good results [18]. The patient subsequently wears a stent continuously for 3 months to prevent stenosis [19].

Cervical Atresia/Dysgenesis

Congenital agenesis of the uterine cervix is a rare Müllerian anomaly (Fig. 3), which is associated with both partial or complete vaginal aplasia in 52% and renal anomalies in 17.8%. Amenorrhoea and severe rapidly progressive lower abdominal pain are the first symptoms with normally developed external sex organs, palpable vaginal recess, varying degrees of uterine enlargement with endometriosis and in some cases tuboovarian masses [20].

It is important to differentiate from high transverse vaginal septum or segmental atresia. Traditionally, hysterectomy has been recommended due to high failure rate of canalization procedures, risk of serious and sometimes fatal ascending infection, and persistent low fertility. With the advancement in modern assisted reproductive facilities, the conservative management seems feasible but depends upon adequacy of cervix. These methods have included creation of neovagina and reconstruction of cervix around various stents, which is both challenging and controversial [21, 22].

With a very high septum, some surgeons have used a combined abdominal/vaginal or laparoscopic approach to put a probe through the fundus and into the obstructed vagina to press on the septum [22].

Congenital Uterine Anomalies (CUA)

Arcuate uterus is the most common uterine anomalies in the unselected population (3.9%) while septum is commonly seen in high-risk women comprising one-third cases, 10% didelphis and unicornuate, and <5% uterine and vaginal aplasia. 3D/2D USGor MRI confirms diagnosis and is considered even better than laparoscopy in differentiating a rudimentary horn with functional endometrium.

A meta-analysis indicated that the spontaneous abortion (RR 1.68), preterm delivery (RR 2.21), malpresentation at delivery (RR 4.75), as also low birth weight (RR 1.93) and perinatal mortality rates (RR 2.43) were significantly higher in women with CUA. This means that further to the increased probability of subfertility and spontaneous abortion, these women have also a higher risk of not having a take-home baby, despite giving birth [23, 24].

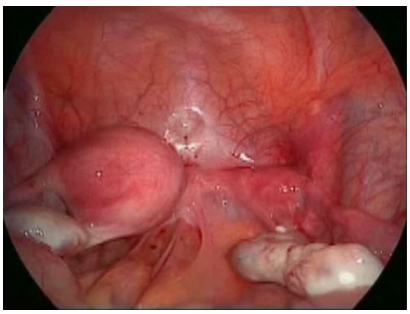

Fig. 4 Laparoscopic view showing unicornuate uterus (left horn) with rudimentary horn (right horn)

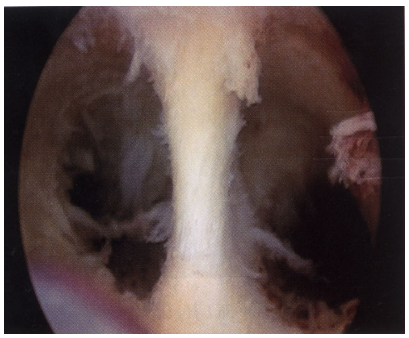

Fig. 5 Hysteroscopic view of a complete uterine septum

Unicornuate Uterus

Arrested development of one of the Müllerian ducts results in unicornuate or hemi-uterus which is present in 14% of a series of HSG cases during evaluation for infertility or bad obstetric outcome as a deviated banana shaped cavity with a single fallopian tube. Reproductive performance is severely impaired with an increased incidence of dysmenorrhoea, infertility and endometriosis. Preterm labour occurs in 20% of all pregnancies, and live birth rate is 29%.

Unicornuate uterus is associated with rudimentary horn in 65% of cases (Fig. 4) and 31% contained endometrial tissue and only half of these communicated with the main uterine cavity. Pregnancy in noncommunicating horn may be conceived by intra-abdominal transit of sperm from the normal horn associated with a high incidence of uterine rupture. Rupture in rudimentary horn occurs usually before 20 weeks and in 80% prior to third trimester. Owing to the high risk for maternal morbidity secondary to rupture and intraperitoneal haemorrhage excision of a cavitary rudimentary horn is indicated when identified.

Bicornuate Uterus

Incomplete lateral fusion of Müllerian ducts results in bicornuate uterus and the level of indentation of the fundus can be complete, partial or arcuate. Two separate but communicating endometrial cavities and a single cervix characterize it. Women with bicornuate uterus can expect a successful pregnancy outcome in 55–60 and 14% of women have poor reproductive outcome. As with other uterine anomalies, preterm delivery occurs in 20–60 and 28% have miscarriage.

HSG is usually the initial diagnostic modality, but confirmation is done by sonography or laparoscopy, which is especially useful to differentiate from septate uterus. Presently, no surgical reconstruction is recommended unless there is evidence of poor reproductive outcome and no other causative factors identified. Strassman's technique of unification can be done abdominally and laparoscopically in women with recurrent pregnancy losses. Caesarian delivery is indicated after metroplasty due to high risk for uterine rupture.

Uterine Didelphys

Failed fusion of Müllerian ducts results in two separated uterine cavities each with a cervix known as uterus didelphys. The women are asymptomatic and in fact of all the major anomalies it has the best reproductive performance with foetal survival rate of 75% and a spontaneous abortion rate of 21%. Pregnancies are located more commonly (76%) in the right uterus. This anomaly is usually diagnosed incidentally on USG or HSG or dyspareunia due to the presence of vaginal septum. No surgical intervention is recommended unless repeated late trimester losses or premature delivery has occurred without any other cause.

Uterine Septum

Failure of resorption of the medial segment after lateral fusion of Müllerian ducts creates a uterus with smooth but transversely broad outer surface on the fundus and a fibrous or fibromuscular septum in the endometrial cavity which can be minimal only at the fundus or can extend half way in the cavity or a complete uterine septum up to the internal os (Fig. 5). Septate uterus is associated with a significantly more pregnancy loss rate (88%) and first trimester abortion rate (42%) than bicornuate uterus. Diagnosis is made on HSG and confirmed on sonography and laparoscopy. Metroplasty should be done in patients with poor reproductive outcome or in women with infertility [25]. Traditionally abdominally metroplasty was done to remove septum but recently with the advent of endoscopy, hysteroscopic septal resection under laparoscopic guidance is an effective and safe alternative. Apart from being less invasive hysteroscopic septoplasty showed improved reproductive outcome, reduced risk of pelvic adhesions and obviates the mandate for Caesarian section.

Compliance with Ethical Standards

Conflict of interest The authors declare that there is no conflict of interest.

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