Optimising Diagnosis and Management of Obstructive Müllerian Anomalies

Editor's Pick

Obstructive Müllerian anomalies can present with significant pain and reproductive morbidity, yet their diagnosis and management remain complex, particularly in adolescents. This insightful feature brings together the embryological basis of these anomalies with practical guidance on recognising key presenting symptoms, choosing appropriate imaging, and navigating treatment. Particularly valuable is the emphasis that Müllerian obstruction is not necessarily a surgical emergency: menstrual suppression can provide effective symptom control while allowing time for accurate diagnosis, careful surgical planning, and referral to specialist centres. The article also looks beyond current practice to some of the most exciting areas for future development, including advances in neovaginal graft materials, fertility-preserving approaches to cervical agenesis, and more personalised strategies for menstrual suppression. By combining practical clinical guidance with a thoughtful discussion of where the field needs to go next, this feature offers an engaging and useful perspective on improving care for patients with these challenging and often under-recognised conditions.

Markus Peck-Radosavljevic
Klinikum Klagenfurt am Wörthersee, Austria

Authors: Julia Feinstein,[1] *Y. Frances Fei[1,2]

1. Department of Obstetrics and Gynecology, The Ohio State University College of Medicine, Columbus, USA
2. Division of Pediatric and Adolescent Gynecology, Nationwide Children's Hospital, Columbus, Ohio, USA

*Correspondence to Frances.Fei@nationwidechildrens.org

Conflicts of interest:	The authors declare there are no conflicts of interest.
Gen AI use:	Generative AI was not used in the preparation of this work.
Peer review:	This article was accepted following double-blind peer review.
Received:	15.07.26
Accepted:	20.08.26
Keywords:	Cervical agenesis, diagnosis, menstrual suppression, Müllerian anomalies, neovaginal reconstruction, obstructive anomalies, reproductive tract malformations, surgical management.
Citation:	EMJ. 2026;11[3]:82-86. https://doi.org/10.33590/emj/13R2U275

Abstract

Müllerian anomalies are congenital reproductive tract malformations affecting 5–7% of individuals with female reproductive organs, and they can be associated with significant gynaecologic and obstetric morbidity. This review summarises the embryology, classification, diagnosis, and management of obstructive Müllerian anomalies. Obstructive anomalies often present with primary amenorrhoea, cyclic pelvic pain, or dysmenorrhoea, requiring

timely evaluation with physical examination, ultrasound, and MRI. Definitive treatment is surgical reconstruction, while menstrual suppression provides effective symptom control and facilitates appropriate surgical planning. Emerging areas of research include optimisation of neovaginal graft materials, fertility-preserving management of cervical agenesis, and personalised approaches to menstrual suppression.

Key Points

1. Müllerian anomalies are congenital reproductive tract malformations affecting 5–7% of individuals with female reproductive organs and can be associated with significant gynaecologic and obstetric morbidity.

2. Obstructive anomalies often present with primary amenorrhoea, cyclic pelvic pain, or dysmenorrhoea, requiring timely evaluation with physical examination, ultrasound, and magnetic resonance imaging.

3. Definitive treatment of obstructive Müllerian anomalies is typically surgical reconstruction, but initiation of menstrual suppression provides effective symptom control and facilitates appropriate surgical planning.

INTRODUCTION

Müllerian anomalies are a group of anomalies of the reproductive tract resulting from incomplete or atypical development of the Müllerian ducts during embryologic development. Müllerian anomalies affect 5–7% of people born with female reproductive organs and can increase the risk of a range of obstetric and gynaecologic problems, including endometriosis, pelvic inflammatory disease, infertility, ectopic pregnancy, preterm birth, and fetal malpresentation.[1,2]

EMBRYOLOGY

Embryologically, a fetus at 5–6 weeks old has both mesonephric and paramesonephric ducts, otherwise known as Wolffian and Müllerian ducts. In XY fetuses, the *SRY* gene on the Y chromosome produces the sex-determining region Y (SRY) protein, which creates anti-Müllerian hormone, leading to resorption of the Müllerian ducts. In XX fetuses, the Müllerian ducts migrate towards the pelvis and form the female reproductive tract. The superior portions become the fallopian tubes, while the inferior portions fuse at around 10 weeks of gestation to give way to the uterus and the proximal one-third of the vagina. The septum connecting the previously separate ducts resorbs by 20 weeks of gestation. The distal two-thirds of the vagina originate from the sinovaginal bulbs, which are derived from the urogenital sinus. This then fuses with the proximal vagina and canalises to form a single vaginal canal.[1]

Failures in the formation, fusion, and resorption contribute to the development of Müllerian anomalies. For example, if the Müllerian ducts do not fuse, a uterine didelphys or a bicornuate uterus could result. Failure of resorption of the septum could lead to a septate uterus. If the vagina fails to canalise, a transverse vaginal septum or distal vaginal agenesis may result. Complete absence of development of the paramesonephric ducts leads to uterovaginal agenesis or Mayer-Rokitansky-Küster-Hauser syndrome.[2] Notably, the renal and anorectal systems are closely related to the development of the Müllerian structures, both in timing and geography, and therefore patients with renal anomalies or anorectal malformations more frequently have concomitant Müllerian anomalies and should undergo early screening.[1,3,4]

OBSTRUCTIVE VERSUS NON-OBSTRUCTIVE MÜLLERIAN ANOMALIES

There are several classification systems for Müllerian anomalies. Currently, the

most common classification systems used are the 2013 European Society of Human Reproduction and Embryology (ESHRE), the European Society for Gynaecological Endoscopy (ESGE), and the 2021 American Society for Reproductive Medicine (ASRM) classification systems.[5,6] Müllerian anomalies can be divided into two broad categories: obstructive and non-obstructive. Obstructive anomalies are defined as anomalies that prevent menstrual egress. Within this category, the anomalies can either be complete or partial. Patients with partial obstruction may have a fenestration that allows for incomplete menstrual egress, or a duplicated system where only one side is able to menstruate. Non-obstructive anomalies are often asymptomatic in early reproductive years and may be an incidental finding on imaging or diagnosed during infertility evaluations. The hymen originates from the urogenital sinus, and hymenal abnormalities are usually not categorised as Müllerian anomalies.

Obstructive anomalies are typically diagnosed earlier than non-obstructive anomalies due to the presence of symptoms, including primary amenorrhoea, severe dysmenorrhoea, cyclic abdominal pain without menstruation, prolonged menstrual periods, or foul-smelling menstrual discharge.[2] Delayed diagnosis can increase the risk of endometriosis (possibly related to retrograde menstruation), chronic pelvic pain, pelvic inflammatory disease, and infertility.[3,4] Examples of obstructive anomalies include cervicovaginal agenesis, obstructed uterine remnant, distal vaginal agenesis, transverse vaginal septum, and obstructed hemivagina and ipsilateral renal anomaly (OHVIRA).[2]

DIAGNOSIS OF OBSTRUCTIVE ANOMALIES

For patients presenting with symptoms that suggest an obstructive Müllerian anomaly, an age-appropriate physical exam is an important first step in diagnosis. An abdominal examination assesses areas of tenderness and can identify an abnormal uterine mass. An examination for sexual maturity can be helpful to rule out pre-pubertal aetiologies. As patients are often young and not yet mature, it is important to take any pelvic examination slowly. To proceed with a genital exam, providers can start with traction on the labia to visualise the vaginal introitus, which could reveal an imperforate hymen with haematocolpos or distal vaginal atresia. If the patient is able to tolerate it, a single digit examination of the vagina can be helpful in identifying the level of the obstruction. For example, a vaginal dimple or shortened vagina may be consistent with distal vaginal agenesis or transverse vaginal septum. A tender bulge along one wall of the vagina and a single cervix palpated at the vaginal apex can be diagnostic of OHVIRA. Some patients may be more comfortable with a rectal exam than with a vaginal exam. A rectal exam can similarly allow for palpation of haematocolpos and assessment of the uterine body.[2]

Ultrasound is recommended as the first step in diagnostic imaging. Abdominal ultrasound might be better tolerated than transvaginal ultrasound in young patients. Ultrasound, especially 3D ultrasound, can evaluate the uterine contour and can assess for evidence of haematometra or haematocolpos. Discussing the concern for Müllerian obstruction with radiology prior to imaging can allow for improved identification and diagnosis. For example, uterine remnants may be in an extra-pelvic location and missed on pelvic ultrasound if not specifically considered. MRI is currently the gold standard in diagnosis. It allows for assessment of the presence of a cervix as well as the presence of endometrium in uterine remnants, both of which might be hard to evaluate with other imaging modalities.[1] Recent studies have shown increased sensitivity and specificity with 3D ultrasound, which may be able to replace MRI for diagnosis in experienced hands.[7]

MANAGEMENT OF OBSTRUCTIVE ANOMALIES

Surgery offers definitive treatment of symptomatic obstructive anomalies. However, not every patient is initially ready for surgery. Depending on the diagnosis,

pre- and/or post-operative vaginal dilation may be recommended to avoid the risk of post-operative vaginal stenosis, which might be challenging for a young or immature patient. Inadequate post-operative dilation may necessitate future reconstructive surgeries, which may be made more difficult by vaginal scarring.

It is important to note that Müllerian anomalies are not surgical emergencies. Symptoms can be managed, in the short or long term, with menstrual suppression. This allows time for accurate diagnosis, appropriate surgical planning, and transfer to specialty centres, if needed. Menstrual suppression can be achieved with hormonal contraceptives, non-contraceptive hormonal suppression (i.e., norethindrone acetate), or gonadotropin-releasing hormone agonists. The goal of any surgical procedure should be definitive reconstruction, and interim measures, such as drainage of a haematocolpos, should be avoided, as these can lead to infection and scarring. The possibility of endometriosis or pelvic adhesions should also be considered in any surgical planning.[1,2]

For patients who are diagnosed with a Müllerian obstruction but are asymptomatic, they should be thoroughly counselled regarding associated symptoms to allow for timely management in the future, but there is no need for urgent surgery at the time of diagnosis.[1]

In some patients, hydrocolpos due to Müllerian obstruction may be diagnosed in the neonatal period due to residual maternal oestrogenisation. In the majority of cases, families can be counselled that the hydrocolpos will most likely resorb spontaneously and surgery should be delayed until puberty. Patients should then be closely monitored during the peri-pubertal years for pubertal progression and any signs of developing obstruction. Pelvic ultrasound should be performed around the age of expected menarche, or with any concerning symptoms to allow for timely management.[1]

Surgical options vary across obstructive Müllerian anomalies. Patients with vaginal septa (including transverse vaginal septum and OHVIRA) are often treated with septum resection, though this can be complicated by vaginal stenosis and the need for definitive management with hysterectomy. Patients with a thicker transverse vaginal septum or distal vaginal agenesis may need vaginoplasty with or without interposition graft. Patients with obstructed uterine remnants or cervical agenesis frequently undergo hysterectomy.[1,2]

FUTURE DIRECTIONS

While surgical and medical management are the mainstays of treatment for anomalies, there is room for improvement.

Limited data exist regarding the optimal timing and method of surgical intervention, as well as surgical complications. For example, many options exist for management of transverse vaginal septum or distal vaginal agenesis. Many experts recommend pre-operative vaginal dilation, which can thin the septum or shorten the atretic portion of the vagina. This can shorten the distance from obstruction to vaginal introitus, which may avoid the need for interposition graft. If needed, there are a variety of different options for graft tissue that can be utilised for the creation of neovaginas, each with different advantages and disadvantages. For example, skin grafts are common, offer fast recovery, and can create good vaginal length with stretch. However, skin grafts can also lead to additional scarring and might have hair growth.[8] Other options include abdominal grafting, using intestinal or peritoneal mucosa. In terms of graft texture and moisture, these options are more similar to a vagina. However, these options also require abdominal surgery, which might be more complicated, risk damaging surrounding structures, and have longer recovery.[8] Bowel neovaginas also suffer from copious discharge, neovaginal prolapse, and potential for the development of inflammatory bowel disease or malignancy.[9] Buccal or oral mucosa can also offer texture and moisture similar to the vagina, but involve a potentially painful oral procedure to harvest the tissue.[10,11] Autologous vaginal grafts are another

option that are associated with increased cost. Tilapia fish skin can be used to create stratified squamous epithelium to create a similar texture and length to a vagina.[12] Human amnion grafts offer a more readily available option at a more limited cost, offer another good texture, and also provide antifibrotic activity.[8]

Cervical agenesis is a rare diagnosis that would benefit from increased data and long-term follow-up. At most centres, cervical agenesis is treated with hysterectomy. There are small studies and case reports of fertility-sparing surgical management that may hold promise for the future. Several approaches have been described, including neocervix creation with vaginal anastomosis, uterovaginal anastomosis, and uterus or cervical canalisation. However, resulting pregnancy rates are low and may result in morbidity or mortality related to ascending infection and stenosis.[13] Additional research in this area and shared outcome data could create opportunity for fertility for the patients who present with these rare anomalies.

Finally, another path for improvement is in menstrual suppression. Menstrual management is often a trial-and-error process to find the method that works best for the patient. Different methods have different rates of amenorrhoea and breakthrough bleeding that might differ from patient to patient. Complete amenorrhoea is often difficult to achieve regardless of method, which can lead to breakthrough bleeding or pain. Improved understanding of the causes of inadequate suppression and/or personalised genomic medicine might allow for earlier identification of optimal menstrual suppression for each patient.

CONCLUSION

Overall, it is important for clinicians to be able to recognise signs and symptoms of Müllerian anomalies and their implications on obstetrical and gynaecologic outcomes. Clinicians should be aware of the primary methods and timing of treatment options. Further research should investigate the best methods for neovaginal grafts, neocervical creation for fertility, and how personalised medicine might allow for better personalisation of treatment.

References

1. Hare K, Childress KJ. Obstructive and non-obstructive müllerian anomalies. Semin Pediatr Surg. 2025;DOI:10.1016/j.sempedsurg.2025.151544.
2. American College of Obstetricians & Gynecologists (ACOG). Management of acute obstructive uterovaginal anomalies: ACOG committee opinion, number 779. Obstet Gynecol. 2019;133(6):e363-71.
3. Fei YF et al. Should we screen for Müllerian anomalies following diagnosis of a congenital renal anomaly? J Pediatr Urol. 2022;18(5):676.e1-e7.
4. Friedman MA et al. Screening for Mullerian anomalies in patients with unilateral renal agenesis: leveraging early detection to prevent complications. J Pediatr Urol. 2018;14(2):144-9.
5. Grimbizis GF et al. The ESHRE/ESGE consensus on the classification of female genital tract congenital anomalies. Hum Reprod. 2013;28(8):2032-44.
6. Pfeifer SM et al. ASRM müllerian anomalies classification 2021. Fertil steril. 2021;116(5):1238-52.
7. Graupera B et al. Accuracy of three-dimensional ultrasound compared with magnetic resonance imaging in diagnosis of Müllerian duct anomalies using ESHRE-ESGE consensus on the classification of congenital anomalies of the female genital tract. Ultrasound Obstet Gynecol. 2015;46(5):616-22.
8. Vatsa R et al. Evaluation of amnion in creation of neovagina in women with Mayer-Rokitansky-Kuster-Hauser syndrome. Fertil steril. 2017;108(2):341-5.
9. Griffin KL et al. Complications and long-term outcomes of patients with cloacal malformation after bowel neovagina creation. J Pediatr Surg. 2025;60(9):162396.
10. Macedo A et al. Buccal mucosa graft vaginoplasty: a viable option demonstrated step-by-step. J pediatr urol. 2023;19(4):485-6.
11. Oakes MB et al. Augmentation vaginoplasty of colonic neovagina stricture using oral mucosa graft. J Pediatr Adolesc Gynecol. 2010;23(1):e39-42.
12. Dias MTPM et al. Neovaginoplasty using nile tilapia fish skin as a new biologic graft in patients with Mayer-Rokitansky-Küster-Hauser Syndrome. J Minim Invasive Gynecol. 2020;27(4):966-72.
13. Mikos T et al. Current knowledge about the management of congenital cervical malformations: a literature review. Fertil steril. 2020;113(4):723-32.