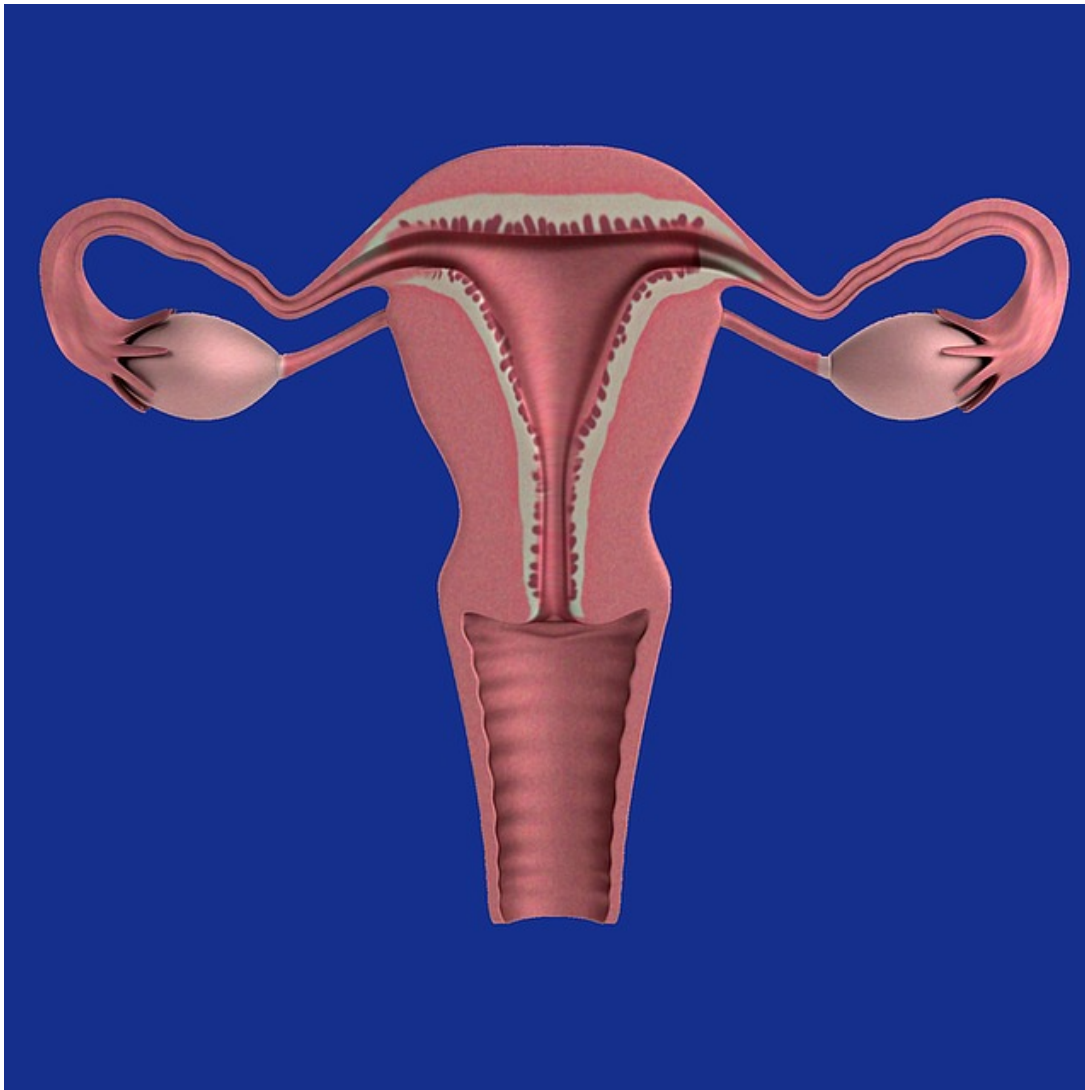


Mullerian Duct Anomalies (MDAs) — Classification and Development

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Mullerian duct anomalies (MDAs) present with developmental malformations of the vagina, cervix, uterus, or fallopian tubes. These malformations can involve a single organ or a combination of organs. Precise etiology is unknown. The estimated incidence of MDAs ranges between 2% and 10% depending on the studied population. The most common presentation of MDA is primary amenorrhea. Infertility and cyclic abdominal pain can also be presenting features.



Overview

The **Mullerian ducts form the female reproductive tract**. The fallopian tubes, uterus, cervix, and superior aspect of the vagina originate from the Mullerian ducts; therefore, Mullerian duct anomalies (MDAs) can present with a wide range of anatomical disruptions in any of these reproductive organs.

In addition to the fallopian tube, uterus, cervix, and vaginal anomalies, **MDAs are also associated with renal and axial skeletal malformations**. Patients with MDAs usually have normal-looking external genitalia and functional ovaries.

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a particular form of MDA that is characterized by the absence of the proximal two-thirds of the vagina in addition to vertebral, cardiac, urologic, and otologic anomalies. With assisted-fertility techniques, fertility and pregnancy may be achievable in some women with MRKH syndrome.

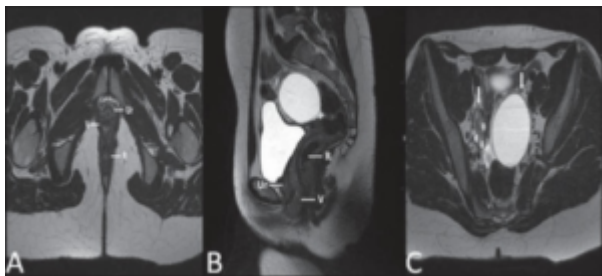


Image: “Mayer-Rokitansky-Kuster-Hauser syndrome (complete presentation). Multiplanar T2-weighted sequence (A, B, C) showing absence of the uterus and of the upper third of the vagina (V) between the rectum (R) and the urethra (Ur). The pelvic images confirm the presence of normal ovaries and large cystic mass in the left ovary” by Ferreira DM. License: [CC BY 4.0](https://creativecommons.org/licenses/by/4.0/)

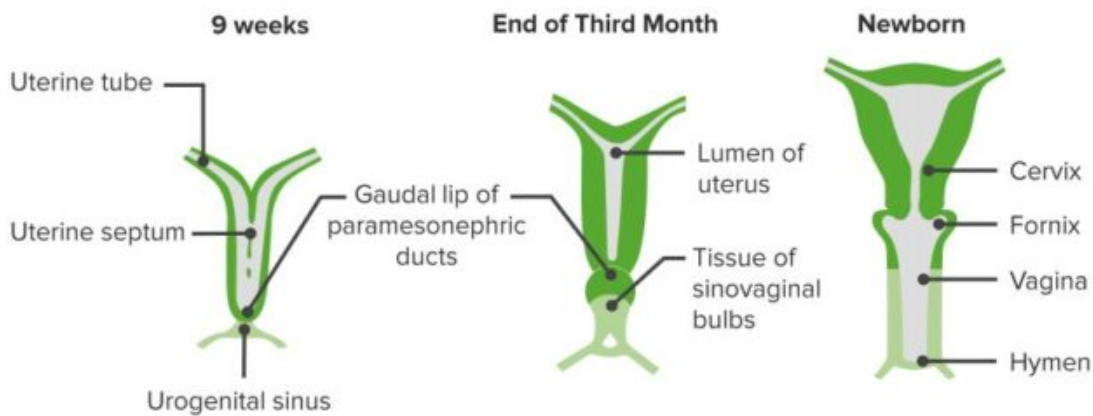
Epidemiology

The **exact incidence or prevalence of MDAs in the general population is unknown**, but estimates range from 0.1%–3.5%. Other studies that focus on uterine malformations alone reported an estimated incidence of 4.3%.

The **incidence of uterine malformations depends on the studied population**. For example, women with fertility problems have an estimated incidence of approximately 6%. The incidence of Mullerian duct defects, however, can be as high as 10% in women with recurrent abortions. Estimated prevalence, therefore, ranges between 2% and 10% depending on the studied population. The **most commonly identified forms of MDAs are septate, arcuate, didelphys, unicornuate, and hypoplastic uteri**.

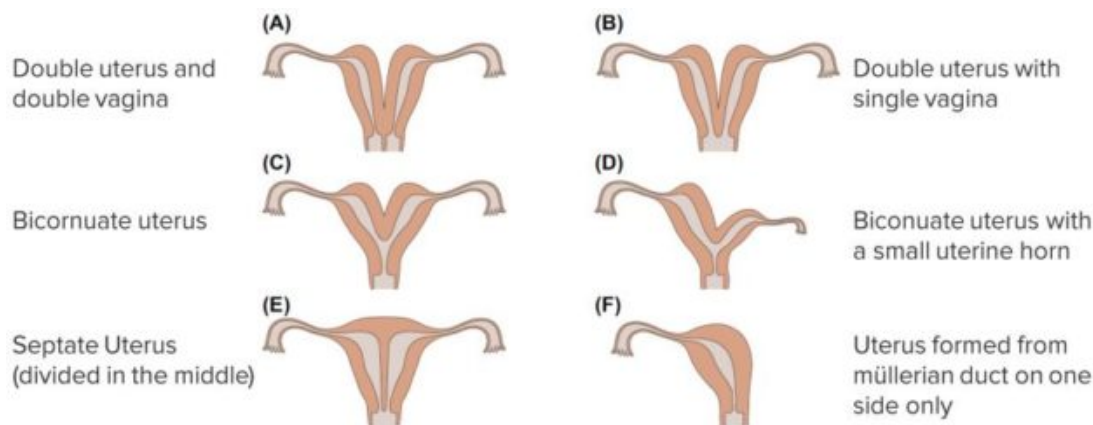
Pathogenesis

Mullerian duct anomalies are believed to result from a disruption in the normal development of the Mullerian ducts in the female. At 6 weeks of fetal development, the Mullerian ducts and Wolffian ducts form. These 2 duct systems are identical at this stage. **If the sex of the fetus is female, Wolffian ducts undergo degeneration and differentiation of the Mullerian ducts start**. Disruption of this differentiation process, which also involves a fusion of different structures, is responsible for the defects seen in MDAs.



Mullerian duct embryological development. Image created by Lecturio.

A family history of MDAs, exposure to diethylstilbestrol or thalidomide, and other intrauterine or extrauterine injuries are the most commonly identified causes of MDAs. A genetic predisposition to MDAs has also been documented. Different forms of inheritance include autosomal dominant, autosomal recessive, or x-linked.



Classification of Mullerian anomalies. Image created by Lecturio.

Based on the affected parts and the time of the insult, different forms of MDAs can occur. The following table summarizes the classes of MDAs and the description of the anomaly.

Class	Description
I	The most common example for this class of MDA is MRKH syndrome. Malformations of the cervix, uterine fundus, or fallopian tubes are also common in this class.
II	This class features the presence of a horn in the uterus that can be communicating with the uterine cavity or can be non-communicating. All patients with class II MDAs also have ipsilateral renal and ureter agenesis. The malformation is described as a unicornuate uterus with or without a horn.
III	This class of malformation is known as a didelphys uterus and is characterized by the complete or partial duplication of 1 of the female reproductive organs that originate from the Mullerian ducts, i.e. the vagina, cervix, or uterus.
IV	This malformation is known as a bicornuate uterus, which can be complete or partial. A septum separates the uterus into 2 chambers. The septum does not extend to the cervix or the vagina. The septum, in this case, extends from the uterine fundus to the cervical os.
V	This class is characterized by a complete or partial midline septum within the uterus and is known as a septate uterus.
VI	This malformation is characterized by a small septate indentation at the fundus of the uterus.

VII	This special class of MDA is known as a diethylstilbestrol-related abnormality. A T-shaped uterus which might be associated with horns is the most common pathology.
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Clinical Presentation

The **most common clinical presentation of MDAs is primary amenorrhea**. Cyclic abdominal pain can be also present. Patients usually have normal secondary sexual development characteristics and are phenotypically females. Infertility is another common presentation and recurrent abortions may be the only presentation of an MDA in an adult female. Patients with abnormal vaginas can present with difficulty with intercourse.

Diagnostic Workup

Patients presenting with any of the above-noted complaints usually undergo abdominal and pelvic ultrasonographical examination for structural evaluation of the vagina, cervix, uterus, fallopian tubes, and ovaries. In **more complicated cases, the patient may need a magnetic resonance imaging study to better visualize the different organs** known to be involved in MDAs.

Laboratory investigations usually show normal follicle-stimulating hormone and luteinizing hormone levels. **Testosterone levels are also normal in females with MDAs**. Chromosomal studies and testing for androgen insensitivity syndrome are also indicated.

Treatment

Isolated vaginal agenesis is usually repaired surgically with a simple vaginoplasty procedure. A stent may be placed within the newly created space to ensure patency after healing. Skin grafting can be used to provide tissue for the vaginoplasty.

Artificial dermis and absorbable adhesion barriers can be also used in vaginal reconstruction procedures, with excellent results. A bowel segment is sometimes used instead of a skin graft for the reconstruction of the vaginal cavity.

Surgical treatment of class II MDAs should be reserved for patients with rudimentary horns that have an endometrium. Laparoscopic excision of the horn can be done easily and with excellent results. Surgical interventions with bipolar coagulation of the pedicle of the horn can be also used to remove the accessory horn.

Patients with class III malformations usually present late compared with patients with other classes. **Surgical intervention is dependent on whether the duplicate uterus is obstructed with a unilateral or nonobstructed vagina**.

Patients with a nonobstructed uterus didelphys should undergo a Strassmann metroplasty procedure, in which the 2 uterine cavities are unified and the cervix is left intact. Patients with an obstructed duplicate uterus should undergo a full excision of the vaginal septum in addition to a hemihysterectomy. **If a hemihysterectomy is to be performed, it is reasonable to remove the ovary** and the fallopian tube on that side.

Class IV malformations can be treated with excision of the uterine horns and apposition of the myometrial edges. Class V malformations are best treated with hysteroscopic

metroplasty with laparoscopy. The treatment of class VI malformations is similar to that for class V malformations. The treatment of class VII malformations is dependent on the type and extent of the exact structural abnormalities.

References

Medscape; Lawrence S Amesse; Apr 13 2016; Mullerian Duct Anomalies.

<http://emedicine.medscape.com/article/273534>

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