

Abnormalities of the Female Reproductive Organs: Adenomyosis, Uterine Fibroids, Macromastia, and More

[See online here](#)

Abnormalities of the female reproductive organs have been a matter of concern in females of reproductive age. These abnormalities can be congenital or acquired. Environmental or genetic anomalies can result in congenital abnormalities in the female organs of infants. Acquired abnormalities imply infections, physical damage during delivery or surgery, or hormonal imbalances. Amenorrhea and infertility are the important representing symptoms of these abnormalities. The diagnosis and identification of abnormalities such as adenomyosis, uterine fibroids, etc., along with their exact location, allows for accurate treatment.



Hymenal Atresia (Imperforate Hymen)

Definition and etiology of hymenal atresia

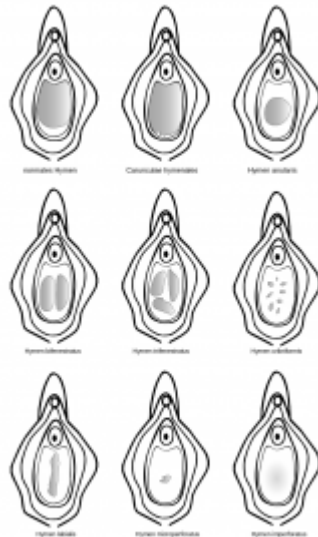


Image: Different forms of the hymen. By Typy_panenských_blan.svg, License: [CC BY-SA 3.0](https://creativecommons.org/licenses/by-sa/3.0/)

Imperforate hymen is a rare congenital malformation characterized by the **failure of the distal canalization of the vaginal plate at the junction between the urogenital sinus and vagina**. An imperforate hymen results in complete occlusion of the outflow from the female genital tract.

Patients are usually asymptomatic until puberty. At puberty, there is primary amenorrhea accompanied by abdominal pains, which occur at monthly intervals and are called menstrual colic. The pains are the result of a build-up of menstrual blood in the vagina (hematocolpos), which cannot drain away due to atresia of the hymen. As the condition develops, **blood can also collect in the uterus (hematometra) and in the fallopian tubes (hematosalpinx)**. Additionally, there can be micturition and defecation as well as flatulence. Some cases of the imperforate hymen can present during the neonatal period. The typical presentation of the imperforate hymen during the neonatal period includes a whitish bulging mass filling the vaginal introitus, ectrodactyly, and the presence of other genitourinary tract anomalies.

Diagnosis of hymenal atresia

Many patients visit doctors due to an absence of menstruation during puberty. A diagnosis can be achieved by gynecological and sonographic investigation. The **hymen can be seen to be distended and lividly colored** due to the blood collecting behind it. Additionally, hematocolpos can be detected as a large swelling using digital rectal examination and can be visualized via ultrasound. Hematometra and hematosalpinx can also be observed via ultrasound.

Treatment for hymenal atresia

The treatment for imperforate hymen is hymenectomy. A transverse incision is made in the hymen with subsequent digital expansion. As prophylaxis, antibiotics should be administered before the procedure.

Breast Abnormalities

Polythelia and polymastia

Polythelia presents with **additional nipples which lie along the mammary ridge**. The mammary ridge is formed during embryonic development but normally regresses afterward.

Due to incomplete regression, polythelia can develop, although it is not classified as a disease. The various forms include:

- **Polythelia completa:** Areola and nipple (mammilla) form on the axilla or below the mammary
- **Polythelia mamillaris:** Areola lies within or outside of the mammilla
- **Polythelia areolaris:** Areola without a nipple

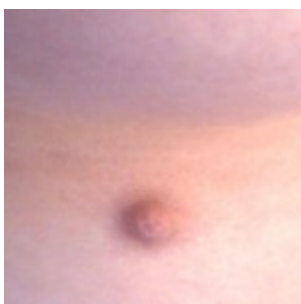
Note: Familial polythelia might be associated with an increased risk of kidney and urinary tract malformations, certain cancers, and Becker nevus.



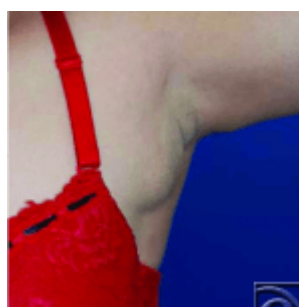
[Image:](#) "Natural milk lines" by The Geneva Foundation for Medical Education and Research. License: [CC BY-SA 3.0](#)



[Image:](#) "Congenital abnormality known as nipple dichotomy or intra-areolar polythelia" by Openi. License: [CC BY 2.0](#)



[Image:](#) "Additional nipple" by IraeLuna. License: Open access



[Image:](#) "Additional breast tissue" by Openi. License: [CC BY 2.0](#)

Additional breast tissue is known as polymastia and is also found along the

mammary ridge. The differences lie between accessory breast tissue (mamma aberrata), in which only gland tissue is affected, and polymastia completa, where in addition to extra glandular tissue, there are also areola and nipples present. This form, also known as mamma accessoria, is much rarer.

The accessory mammary gland is a soft bulge of tissue, most often felt in the area of the axilla or the vulva. It can **swell premenstrually, during pregnancy or during the lactation period, which can be painful. This can also lead to plugged milk ducts and result in mastitis.** In addition to this, a degeneration of the ectopic tissue is possible, in which case, operative removal of the excess mammary tissue is strongly recommended as a therapeutic option.

A missing nipple is known as **athelia** and a missing breast is termed **amastia**.

Macromastia and Micromastia

Macromastia (sometimes also known as gigantomastia) describes the **excessive hypertrophy of the breast tissue.**

Patients experience psychological stress in addition to physical complaints. The weight of the breasts can lead to neck and back pain, as well as postural problems. Breast reduction via plastic surgery is a possible operative strategy.

The **opposite of this condition is micromastia.** This is a hypoplasia of the breast. There are numerous possible causes of this condition, ranging from genetic disposition such as [congenital disturbances in sexual development](#) (i.e., Ullrich-Turner syndrome), to psychiatric diseases such as [anorexia nervosa](#). If there is considerable psychological distress, breast enlargement (augmentation plastic surgery) is an option.

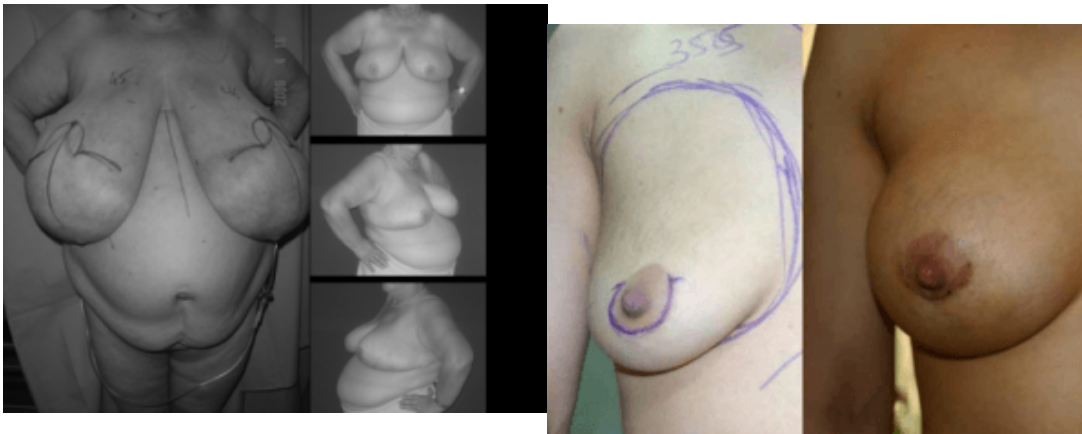


Image: “Patient with severe symptomatic macromastia.” by Openi. License: [CC BY 2.0](#)

Image: “breast augmentation – before and after the operation” by Openi. License: [CC BY 3.0](#)

Abnormalities of the Uterus

Common uterine abnormalities

1. Uterine fibroids (Leiomyoma)
2. Adenomyosis
3. Endometrial thickening

Uterine fibroids

A uterine fibroid is a **heterogeneous hypoechoic mass arising from the uterine myometrium**. A 'fibroid' that progressively enlarges in size or is greater than 8 cm should be suspected as a sarcoma and an MRI should be performed to help differentiate both.

Fibroids are:

- Benign tumors of the smooth muscle cells of the uterus
- Very common during the reproductive years
- Mostly asymptomatic but can cause pain, menorrhagia, and **infertility**
- Imaging findings: hypoechoic mass, possible calcifications, may have cystic components or fat.
- Location: submucosal (bleeding and infertility), intramural, or subserosal (mass effect on adjacent structures)

Adenomyosis

Adenomyosis is characterized by:

- Presence of heterotypic endometrial stroma within the myometrium
- Clinical presentation often reveals **dysmenorrhea** or menorrhagia
- Imaging findings: enlarged uterus, myometrial cystic spaces, widened posterior uterine wall on ultrasound, best visualized on ultrasound and MRI

Endometrial thickening

Focal	Diffuse
<ul style="list-style-type: none">• Polyp• Endometrial carcinoma• Submucosal fibroid	<ul style="list-style-type: none">• Normal secretory phase• Endometrial hyperplasia• Endometrial carcinoma

Endometrial thickening is difficult to distinguish based on imaging alone. A sonohysterogram helps differentiate focal from diffuse endometrial thickening and in localizing a focal lesion.

Endometrial atrophy occurs in postmenopausal women due to a lack of hormonal stimulation. The endometrium is found to be less than 4 mm in size. It is the second most common cause of postmenopausal bleeding.

Types of uterus abnormalities

Abnormalities of the uterus **occur as a result of the disrupted fusion of the Mullerian ducts during development**. Based on the period when incomplete fusion occurs, the resultant clinical pictures have a range of severity:

- **Arcuate uterus:** Least serious form where there is a concave contour towards the fundus.
- **Subseptate uterus:** Externally normally shaped, however, there is a projection of the medial septum causing a partial separation of the uterus.

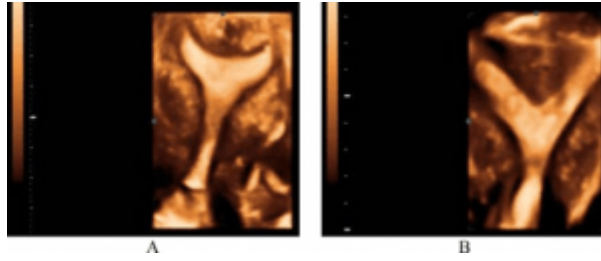


Image: Real-time 3D (4D) view of subseptate uterus. By Openi, License: [CC BY 3.0](https://creativecommons.org/licenses/by/3.0/)

- **Septate uterus:** Externally normally shaped, however, there is a projection of the medial septum that completely divides the uterus internally.
- **Uterus bicornis unicollis:** 2 uterus bodies, 1 cervix.

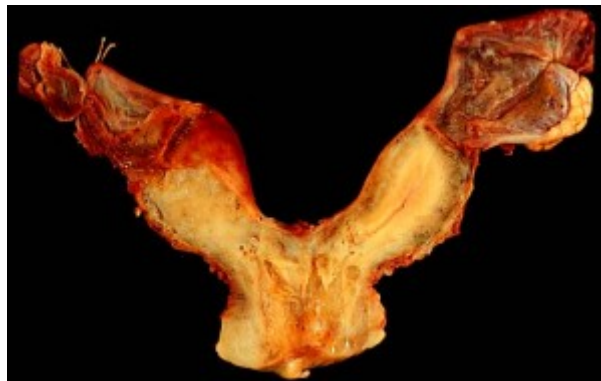


Image: Uterus bicornis. By Ed Uthman, License: [Public Domain](https://creativecommons.org/licenses/publicdomain/)

- **Uterus bicornis bicollis:** 2 uterus bodies, 2 cervixes.
- **Duplex uterus (uterus didelphys):** Most serious form; there are 2 uteri, 2 cervixes, and 2 vaginas.
- **Unicornuate/bicornuate uterus:** Rudimental horn formation as a result of the incomplete development of one of the Müllerian ducts and incomplete formation of the other.

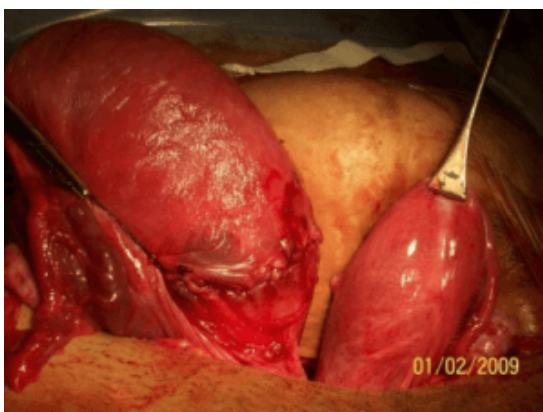


Image: “Bicornuate uterus during Caesarean Section.” by Openi. License: [CC BY 2.0](https://creativecommons.org/licenses/by/2.0/)



Image: “Unicornuate uterus with right-sided rudimentary horn” by Openi. License: [CC BY 2.0](https://creativecommons.org/licenses/by/2.0/)

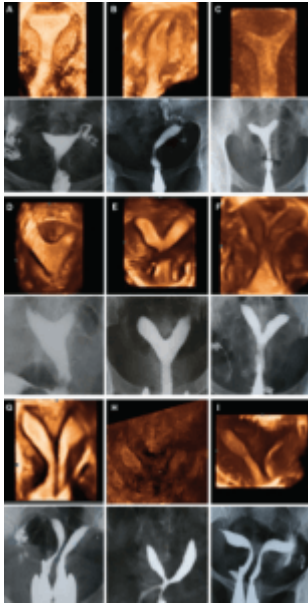


Image: “Comparison of three-dimensional ultrasound and HSG imaging in cases of uterine malformation using AFS: A. Normal uterus, B. Unicornuate uterus, C. Arcuate uterus, D-G. Different subtypes of septate uterus (partial to complete septum), H. Bicornuate uterus, I. Didelphys.” by Openi.
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Note: Abnormalities of the uterus usually accompany abnormalities of the vagina, the kidneys, and the efferent urinary tracts!

Symptoms, diagnosis, and therapy of uterine abnormalities



Image: Hysterosalpingogram demonstrates a communicating septate uterus, cervix duplex. By Openi, License: [CC BY 2.5](https://creativecommons.org/licenses/by/2.5/)

Abnormalities of the uterus can lead to [abortions](#), abnormal fetal position, birth complications, premature birth, [dysmenorrhea](#), and [sterility](#).

Several imaging procedures can be used to achieve a diagnosis: sonography, endoscopy, and hysterosalpingography (contrast X-rays of the uterus and fallopian tubes). These should precede a clinical examination with inspection and palpation.

Treatment involves the hysteroscopic separation of the septum in a single (sub) septate uterus, or in the case of a bicornuate uterus, the surgical separation of the septum with the subsequent unification of the 2 halves of the uterus (Strassmann's operative procedure).

Mayer-von-Rokitansky-Küster-Hauser Syndrome

In Mayer-von-Rokitansky-Küster-Hauser (MRKH) syndrome, the **uterus is only rudimentary (uterine aplasia)**. Additionally, there is hypoplasia or aplasia of the vagina, while the ovaries develop normally and therefore fulfill their hormonal function.



Image: Vaginal agenesis. By Dr. Antonio Jose Marrero Ochoa, Josea69, License: [CC BY-SA 2.5](https://creativecommons.org/licenses/by-sa/2.5/)

The abnormality develops in approx. the 2nd embryonic month. Those affected have **female gonadal sex and female chromosomal sex (46, XX)** and are normally symptom-free until they reach puberty. At this point, the leading symptom is primary amenorrhea. Additionally, there can be difficulties with intercourse. Because the uterus only has a rudimentary form, the patient is infertile.

This syndrome often arises in combination with abnormalities in the urinary system (e.g., renal agenesis, ectopic kidneys).

Ten percent of women who suffer from primary amenorrhea are **diagnosed with MRKH syndrome**. This can be diagnosed through clinical examination and with the aid of various imaging procedures (e.g., MRI, see images).



Image: "MRI showing pelvic kidney and absent uterus (Arrow)" by Openi.
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Image: "MRI showing pelvic kidney in relation to bladder" by Openi.
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Image: "Sagittal T2W image in mid sagittal plane. No uterus can be made out and only the lower vagina is seen" by Openi. License: [CC BY 2.0](https://creativecommons.org/licenses/by/2.0/)



Image: "Axial T2W image at the level acetabulum showing absent vagina between the bladder and rectum." by Openi. License: [CC BY 2.0](https://creativecommons.org/licenses/by/2.0/)

This condition can be **surgically treated by creating an artificial vagina (neovagina)**, allowing the patient to lead largely normal lives as women, including being able to carry out sexual intercourse. However, sterility cannot be treated.



Image: The stent is seen after securing to body with intravenous infusion set tubes which tied to the belt in the patient. By Openi, License: [CC BY 2.0](#)



Image: 3 months after McIndoe vaginoplasty, neovagina. By Openi, License: [CC BY 2.0](#)

References

Decherney, A. H., Lauren, N., Goodwin, T. M. & Neri, L., 2007. *Current diagnosis and treatment in Obstetrics and Gynaecology*. New York: McGraw-Hills.

Dutta, D., 2014. *DC Dutta's Textbook of Gynaecology*. New Delhi: Jaypee Brothers.

Kumar, V., Abbas, A. K. & Caster, J. C., 2013. *Robbins basic pathology*. Philadelphia: Elsevier Saunders.

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