Abnormalities of the Female Reproductive Organs

Abnormalities and disorders which can affect the female body are by no means rare and are not just of cosmetic relevance. A malformed uterus can cause a woman to be infertile, ectopic breast tissue can degenerate malignantly, and atresia of the hymen leads to amenorrhea. The following article gives prospective doctors an overview of the abnormalities of the reproductive organs – in order to allow for these to be diagnosed securely in clinical or general practice examinations - and to allow a treatment strategy to be developed with the patient.

Hymenal Atresia (Imperforate Hymen)

Definition and etiology of hymenal atresia
A physiological imperforation of the hymen underlies atresia of the hymen. The hymen is a thin sheet of tissue which is responsible for the separation of the vagina and the urogenital sinus during development. The hymen can take different forms and imperforate hymen results in a complete closure of the vagina.

Symptoms and clinical picture of hymenal atresia

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 molimina. The pains are the result of a build-up of menstrual blood in the vagina (hematocolpos) which cannot drain away due to the 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.

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 a digital rectal examination and can be visualized via ultrasound. Hematometra and hematosalpinx can also be observed via ultrasound.

Treatment for hymenal atresia

Treatment for hymenal atresia is done by making a surgical opening: there is a transverse incision made in the hymen with a subsequent digital expansion. As a 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 classed as a disease. The various forms include:

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

**Note**: Polythelia is usually clinically unremarkable!

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 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 and 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 the operative removal of the excess mammary tissue should be 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 an excessive hypertrophy of the breast tissue.

Patients suffer from psychological stress in addition to physical complaints. The weight of the breasts can lead to neck and back pain, as well as postural problems. A 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 for 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.

### Abnormalities of the Uterus

#### Types of uterus abnormalities

Abnormalities of the uterus occur as a result of disrupted fusion of the müllerian ducts during development. Based on the time period when the 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](https://example.com/subseptate uterus.png)

  *Image: “Real time 3D (4D) view of subseptate uterus,” by Openi. License: CC 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**: Two uterus bodies, one cervix.

  ![Image](https://example.com/uterus bicornis unicollis.png)

  *Bild: “Uterus bicornis,” by Ed Uthman. License: Public Domain*

- **Uterus bicornis bicornis**: Two uterus bodies, two cervixes.

- **Duplex uterus (uterus didelphys)**: Most serious form; there are two uteri, two cervixes and two 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](https://example.com/unicornuate uterus.png)

  *Image: “Unicornuate uterus with right-sided rudimentary horn” by Openi. License: CC BY 2.0*

  ![Image](https://example.com/unicornuate uterus during Caesarean Section.png)

  *Image: “Bicornuate uterus during Caesarean Section.” by Openi. License: CC BY 2.0*
**Note:** Abnormalities of the uterus usually accompany abnormalities of the vagina, the kidneys and the efferent urinary tracts!

Symptoms, diagnosis and therapy of uterus abnormalities
Abnormalities of the uterus can lead to abnormalities of the uterus can lead to **abortions**, abnormal fetal position, birth complications, premature birth, dysmenorrhea and sterility.

A number of 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 can be done by 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 subsequent unification of the two halves of the uterus (Strassmann’s operative procedure).

**Mayer-von-Rokitansky-Küster-Hauser Syndrome**

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

The abnormality develops in approximately the second **embryonic month**. Those affected have a female **gonadal** sex and a 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**. Due to the uterus only having 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 with primary **amenorrhea** are diagnosed with Mayer-von-Rokitansky-Küster-Hauser syndrome. This can be diagnosed through clinical examination and with the aid of various **imaging** procedures (e.g., **M.R.I.**, see images).
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. The sterility cannot be treated, however.
Review Questions

The answers can be found below the references.

1. **Which symptom or finding is not typical of hymenal atresia?**
   - A. Hematocolpos
   - B. Amenorrhea
   - C. Hematosalpinx
   - D. Hemothorax
   - E. Hematometra

2. **Which of the following abnormalities does not accompany an abnormality of the uterus?**
   - A. Polythelia
   - B. Uterus bicornis unicollis
   - C. Mayer-von-Rokitansky-Küster-Hauser syndrome
   - D. Arcuate uterus
   - E. Uterus didelphys

3. **Which statement is true? Mayer-von-Rokitansky-Küster-Hauser syndrome...**
A. ...is accompanied by karyotype 46, XY.
B. ...can be accompanied by abnormalities of the urinary system.
C. ...is diagnosed in 1 % of all women who suffer from amenorrhea.
D. ...can be treated with neovaginal surgery, which leads to a regression of the sterility.
E. ...has the lead symptom that the patient has difficulties in having intercourse.

References


Correct answers: 1D, 2A, 3B

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