

Female genital abnormalities

What are female genital abnormalities?

Congenital malformations of the female genital tract are defined as deviations from normal anatomy, resulting from embryonic maldevelopment of the Müllerian or paramesonephric ducts^[1]. Female genital abnormalities often do not present until, or well after, puberty^[2].

Congenital malformations of the female genital tract may be the result of a clear disturbance in one stage of embryonic development, or result from disturbances in more than one stage of normal formation^[3]. There are therefore extremely wide anatomical variations and a large number of combinations of congenital malformations of the female genital tract^[4]. Therefore, although female genital abnormalities may be isolated, careful assessment for possible underlying disorders, particularly chromosomal or metabolic, is essential.

Different classification systems have led to some uncertainty regarding epidemiology with some concerns that the classifications may lead to underdiagnosis or overdiagnosis^[5].

Surgical techniques to correct genital malformations depend on the type of anomaly, its complexity, the patient's symptoms and the correct embryological interpretation of the anomaly. Most anomalies can be resolved vaginally or by hysteroscopy but laparoscopy or laparotomy is often also required^[6].

Uterine abnormalities

Anomalies in the Müllerian ducts have a prevalence from 0.5 to 6.7% in the general population and up to 16.7% in women with recurrent miscarriage^[7]. The most common types of uterine abnormalities are caused by incomplete fusion of the Müllerian or paramesonephric ducts:

- Complete failure is rare and results in double vagina, double cervix and double uterus. Variants may occur depending on the degree of fusion of the Müllerian ducts.
- More extensive fusion of the Müllerian ducts results in a single vagina, single cervix and double single-horned uteruses which are partially fused.
- Other abnormalities include septate uterus (uterus with midline septum), arcuate uterus (uterus slightly indented in the middle) and unicornuate uterus (second blind-ending rudimentary horn).
- An important limitation of the current classification of septate uterus, is the high proportion of unclassifiable cases^[8] . The high rate of overdiagnosis of septate uterus according to ESHRE/ESGE-2016 may lead to unnecessary surgery and therefore unnecessary risk in these women, and may impose a considerable financial burden on healthcare systems.
- Investigations:
 - Ultrasound.
 - Hysterosalpingography, which allows evaluation of the uterine cavity and tubal patency.
 - MRI scan, which is considered the best imaging technique for uterine abnormalities^[9] .

- Complications:
 - Dysmenorrhoea.
 - Haematometra.
 - Complications during pregnancy and labour: late miscarriage, uterine rupture, premature labour, malpresentation, obstructed labour, retained placenta, postpartum haemorrhage.
 - Fertility is usually unaffected.
 - Congenital renal abnormalities often coexist - 48.4% in one review and these were observed most frequently in women having a duplex uterus with obstructed hemivagina^[10]. The most common renal abnormality was unilateral renal agenesis, which was observed in 58 of 90 women (64.4%).
- Management:
 - Decision for surgical intervention will depend on the effect of the abnormality on enabling a viable pregnancy.
 - A septate vagina and the rudimentary horn of a bicornuate uterus are usually removed.
 - Uterine reconstruction is recommended for a bicornuate or septate uterus which is considered to be the cause of recurrent miscarriages.

Vaginal abnormalities

- Vaginal agenesis:
 - Usually occurs with absent uterus but ovaries present.
- Vaginal atresia:
 - The lower portion of the vagina consists of fibrous tissue with a well-differentiated uterus.

- Müllerian aplasia:
 - Nearly all of the vagina and most of the uterus are absent.
 - Accounts for most cases of absence of vagina with normal external genitalia.
 - Can be associated with other anomalies including fused cervical vertebrae and middle-ear defects.
- Transverse vaginal septa:
 - Can be present as single or multiple in the upper or lower segments and may be patent or perforated; can be the cause of haematometra or other fluid collections.
 - Longitudinal vaginal septa have also been seen.
- Associated anomalies:
 - The urethra can open into the vaginal wall or the vagina can open into a persistent urogenital sinus. Associated rectal abnormalities include vaginorectal fistula, vulvovaginal anus, rectosigmoidal fistula.

Abnormalities of the hymen

- The incidence of imperforate hymen is estimated of 1 in 1,000 female births^[11] .
- This may first present with obstruction of menstrual flow after puberty.
- Because the hymen is not a Müllerian structure, urinary tract anomalies are unlikely.

Cloacal dysgenesis^[12]

- Rectocloacal fistula with persistent cloaca is a common outlet for the urinary, genital and intestinal tracts.

- With a rectovaginal fistula, the vestibule can appear normal but the anus is found in the perineum.
- Can also be persistent urogenital sinus with a single external orifice without an anorectal defect.

Abnormalities of external genitalia

- Labia minora abnormalities: can have labial fusion or hypertrophy in otherwise normal females. Hypertrophy can be unilateral or bilateral and may occasionally require surgical correction.
- Abnormalities of labia majora: can be hypoplastic or hypertrophic. Abnormal fusion is usually associated with ambiguous genitalia of female pseudohermaphroditism due to congenital adrenal hyperplasia.
- Clitoral abnormalities: these are generally rare; agenesis is extremely rare and is double clitoris or bifid clitoris. Hypertrophy can be associated with a number of intersex disorders.

Ovarian agenesis/dysgenesis

- This includes Turner syndrome and a wide range of chromosomal anomalies characterised by the absence of two X chromosomes with the required critical zones.
- This results in the production of streak ovaries and is associated with a number of other somatic abnormalities.
- Neonates with streak ovaries often have oedema of the hands and feet as a first presenting sign. However, many present in adolescence with short stature.

See also the separate [Turner Syndrome](#) article.

True hermaphroditism

- This is now called ovotesticular disorder of sex development and is characterised by the presence of both ovarian and testicular tissue in a single patient. The testes will develop in the presence of a single Y chromosome even with more than a single X chromosome^[13].

- No single **clinical** feature can distinguish true hermaphroditism from other forms of intersexuality with firm diagnosis possible after ultrasound and hormone assay.
- Often patients have been reared as males because of appearance of external genitalia but, with early diagnosis, most should be reared as females, with many developing female-type breasts. This distinguishes them from male hermaphrodites.
- Many menstruate and some who have had testicular tissue removed have become pregnant.
- Many cases have an apparently normal pair of X chromosomes, many of which have Y-specific sequences.
- Treatment is to remove contradictory organs and reconstruct external genitalia corresponding to sex of rearing. Unless there are pressing medical reasons, surgery can be delayed until the person is able to voice whether they feel male or female and gender can be appropriately assigned.

Female hermaphroditism due to congenital adrenal hyperplasia

See the separate [Congenital Adrenal Hyperplasia](#) article.

Ambiguous genitalia

For discussion of disorders of sexual development (DSD), see the separate [Ambiguous Genitalia](#) article. Timing of surgery is more controversial than previously. Adverse outcomes have led to recommendations to delay unnecessary surgery to an age when the patient can give informed consent.

Further reading

- [Ludwin A, Martins WP, Nastri CO, et al](#); Congenital Uterine Malformation by Experts (CUME): better criteria for distinguishing between normal/arcuate and septate uterus? *Ultrasound Obstet Gynecol.* 2018 Jan;51(1):101–109. doi: 10.1002/uog.18923.

- [Brannstrom M](#); Uterus transplantation and beyond. J Mater Sci Mater Med. 2017 May;28(5):70. doi: 10.1007/s10856-017-5872-0. Epub 2017 Mar 29.
- [Imaeda K, Kobayashi Y, Nakamura K, et al](#); Usefulness of Endoscopy for Female Genital Plastic Surgery: A Series of 4 Cases with Genital Malformations. Am J Case Rep. 2021 Nov 7;22:e934363. doi: 10.12659/AJCR.934363.

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