Female Congenital Malformations

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Objectives

• Identify which female reproductive organs have the highest rate of congenital defects
• Illustrate the congenital vaginal and cervical malformations
• Describe the major cause for outflow tract abnormalities
• Define imperforate hymen and its cause
• Summarize the clinical features by age of an imperforate hymen
• Describe the complications of an imperforate hymen in adolescence
• Define vaginal adenosis, its pathology and its cause
• Describe the pathogenesis of vaginal adenosis and its complication
• Describe the association of DES and clear cell adenocarcinoma of the vagina
• Determine the congenital anomalies of the uterus
• Differentiate between a bicornis uterus and a uterus didelphys
Female Congenital Malformations

- Relatively rare conditions which affect the vagina and endometrium most often.
- A congenital absence of the vagina is generally associated with anomalies of the uterus. In the presence of a functional uterus, the absence of a vagina may lead to an accumulation of menstrual blood within the uterus.
- Congenital absence of the uterus (agenesis) reflects failure of the mullerian ducts to develop and is usually associated with other anomalies of the urogenital tract.
Congenital Vaginal Malformations

1. Septate Vagina: results from the failure of the embryonic mullerian ducts to fuse properly and the resulting median wall does not resorb.

2. Imperforate Hymen: an outflow tract abnormality. Female genital malformations occur in 5% of the general population, with Imperforate Hymen being the most common genital outflow tract defect.

3. Vaginal Adenosis due to Diethylstilbesterol (DES)
Causes of Outflow Tract Abnormalities

• Embryologically, the lower 2/3 of the vagina develop from the urogenital sinus. The upper vagina, cervix, uterus, fallopian tubes and ovaries form from the Mullerian (paramesonephric) Duct

• Failure of vertical fusion or canalization of these two systems in utero results in these abnormalities

• The hymen originates from the embryonic vagina buds from the urogenital sinus. It becomes perforated forming a central canal with communication between the upper vaginal tract and the vestibule of the vagina
Embryology of Female Reproductive Tract

[Diagram showing the development of the female reproductive organs, including the mesonephros, Müllerian duct, mesonephric duct, Müllerian tubercle, urogenital sinus, and female reproductive structures like the ovary, fimbriae, suspensory ligament of the ovary, ovarian ligament proper, mesovarium, corpus (body), cervix, fornix, and vagina.]
Imperforate Hymen

• The most common and most distal form of vaginal outflow obstruction, occurring in 0.1% of infant girls

• Normally, the central portion of the hymenal membrane is usually absent, creating the typical configuration of a ring-like structure at the level of the vaginal vestibule (annular)

• Although a congenital anomaly that may be detected at birth or early in life, it may also remain undetected until puberty in some patients
Intact Vascular Hymen
Imperforate Hymen
Imperforate Hymen in the Neonate

• At birth, the presence of increased mucus secretions in the vagina secondary to maternal estrogen effects may result in a Mucocolpos appearing as a bulging hymenal membrane between the labia. The membrane may be white because of the trapped mucoid material and may lead to urinary tract infections or bladder obstruction due to urethral compression.
Imperforate Hymen in the Adolescent

• The adolescent usually presents with primary amenorrhea (most common clinical presentation), so the presence or absence of secondary sexual characteristics should be noted

• Other symptoms include lower abdominal or pelvic pain that may be cyclic, back pain, urinary retention and constipation

• The diagnosis is often made on examination when a distended, bluish hymenal membrane is observed in the introititis, due to collected menstrual blood (hematocolpos)
Imperforate Hymen in the Adolescent

• The trapped menstrual secretions may back up into the uterus (hematometrocolpos)
• Reflux of endometrial tissue and blood through the fallopian tubes (hematosalpinx) may result in secondary endometriosis
• An accumulation of infected material within the vaginal cavity (pyocolpos) may cause ascending genital tract infections
Vaginal Adenosis Due to DES

- Occurs in daughters exposed in Utero to DES
- Refers to failure of the normal glandular epithelium that lines the embryonic vagina to be replaced by squamous epithelium during fetal development
- DES (morning after pill), a synthetic estrogen, was used in the 1940’s and 1950’s for high-risk pregnancies (used to prevent abortion)
- In the 1970’s, there was a substantial increase in the incidence of this disease in young daughters of women who received DES during pregnancy
Vaginal Adenosis Due to DES

- At the 10\textsuperscript{th} week of gestation, the upgrowth of squamous epithelium derived from the urogenital sinus replaces the glandular (mullerian) epithelium lining the vagina and exocervix.
- DES exposure anywhere from the 10\textsuperscript{th} to about the 18\textsuperscript{th} week of gestation, arrests this transformation process and glandular tissue remains within the vagina (adenosis).
- Manifests grossly as red, granular patches on the vaginal mucosa which usually disappear as the woman gets older.
Red, Patchy Vaginal Adenosis
Vaginal Adenosis due to DES

- Microscopically, there is mucinous columnar cells, similar to the endocervix, along with ciliated cells with eosinophilic cytoplasm, similar to the lining cells of the fallopian tube and endometrium.
- The glandular cells ultimately undergo squamous metaplasia.
- Rare cases of clear cell adenocarcinoma of the vagina have also occurred in the daughters of women treated with DES.
Clear Cell Adenocarcinoma due to DES

- A rare tumor of the vagina encountered exclusively in women exposed to Diethly-stilbesterol (DES).
- Develops most frequently on the anterior wall of the upper third of the vagina.
- Most common between ages 17 and 22.
- Abundant glycogen account for the clear nature of the cytoplasm and they are essentially curable when small and asymptomatic.
Clear Cell Adenocarcinoma due to DES

- Although almost all cases of clear cell adenocarcinoma are associated with vaginal adenosis, very few women with adenosis develop this cancer.

- In more advanced stages, they may spread by hematogenous or lymphatic routes.
Clear Cell Adenocarcinoma
Clear Cell Carcinoma due to DES
Vaginal Atresia

• A congenital anomaly of the female genital tract that presents as a deformed, obstructed and nonfunctional vagina that occurs in 1 in 4-5000 live female births
• Often unnoticed until adolescence, when pain and lack of normal menstrual flow indicated the condition, which is due to an obstruction
• Frequently associated with Rokitansky-Mayer-Kuster-Hauser Syndrome which is a congenital lack of a uterus, vaginal atresia or agenesis, despite normal ovaries and external genitalia
• Surgery may repair the atresia
Vaginal Atresia
Vaginal Septum

• A congenital longitudinal or transverse partition within the vagina, both corrected with surgery
  1. Longitudinal Septum: develops during embryogenesis due to incomplete fusion of the two mullerian ducts, resulting in a double vagina, with a chance of a double cervix and uterus
  2. Transverse Septum: Also develops during embryogenesis when the two mullerian ducts fuse improperly to the urogenital sinus. A complete septum blocks menstrual flow and is a cause of primary amenorrhea
• Both may cause obstruction to flow and dyspareunia
Embryology of Female Reproductive Tract
Cervical Stenosis

• A congenital or acquired abnormality that occurs at the level of the internal os,
• The most common acquired cause is due to prior diathermy excision of cervical lesions
• Congenital stenosis is usually associated with dilation of the lower uterine segment, and may be an obstruction with menstrual flow.
• Examination reveals an elongated, rigid cervix with no elasticity and a small contracted os
Cervical Atresia

Atresia of cervix
Congenital Anomalies of the Uterus

1. Bicornis (septate) Uterus: is a uterus with a common fused wall between two distinct uterine cavities. Due to a failure of the common wall between the apposed mullerian ducts to degenerate, forming a single uterine cavity.

2. Uterus Didelphys: refers to a double uterus with a double vagina. Again due to failure of the two mullerian ducts to fuse during embryonic life.

3. Uterus Septae: refers to a single uterus with a partial remaining septum, owing to failure of the wall of the fused mullerian ducts to fully resorb.
Embryology of Female Reproductive Tract
Uterus Bicornis
Bicornis (Septate) Uterus
Uterus Didelphys

Uterine tube

Uterus didelphys with double vagina
Uterus Didelphys
Uterus Septae
Slide References

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