

# CONGENITAL MALFORMATION OF FEMALE GENITAL ORGANS

*DR. SHEEBA.S,  
ASSISTANT PROFESSOR,  
(DEPT OF OBG),  
SKHMC.*



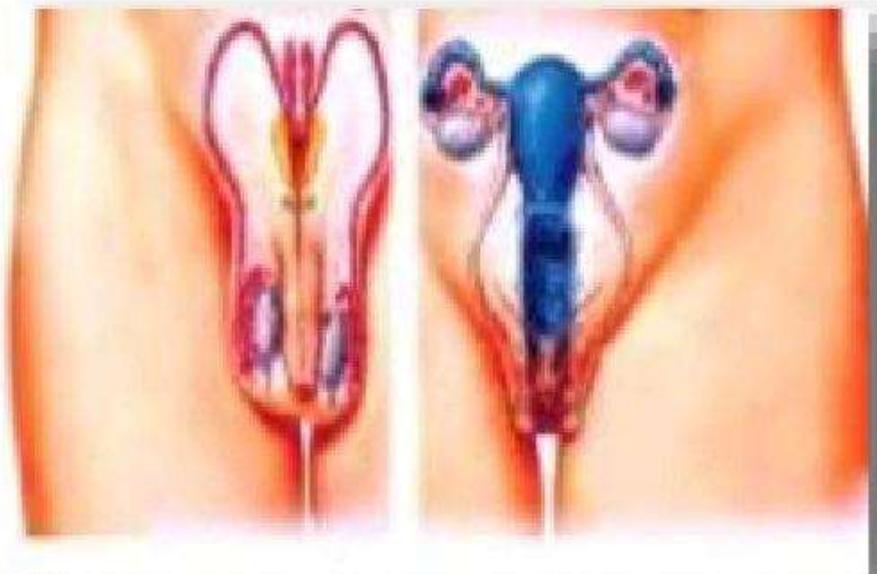
# INTRODUCTION

From the embryological considerations, the following facts can be deduced.

- ◆ Developmental anomalies of the external genitalia along with ambiguity of sex are usually genetic in origin
- ◆ Major anatomic defect of the genital tract is usually associated with urinary tract abnormality (40%), skeletal malformation (12%), and normal gonadal function
- ◆ While minor abnormality escapes attention, it is the moderate or severe form, which will produce gynecologic and obstetric problem.

# Definition

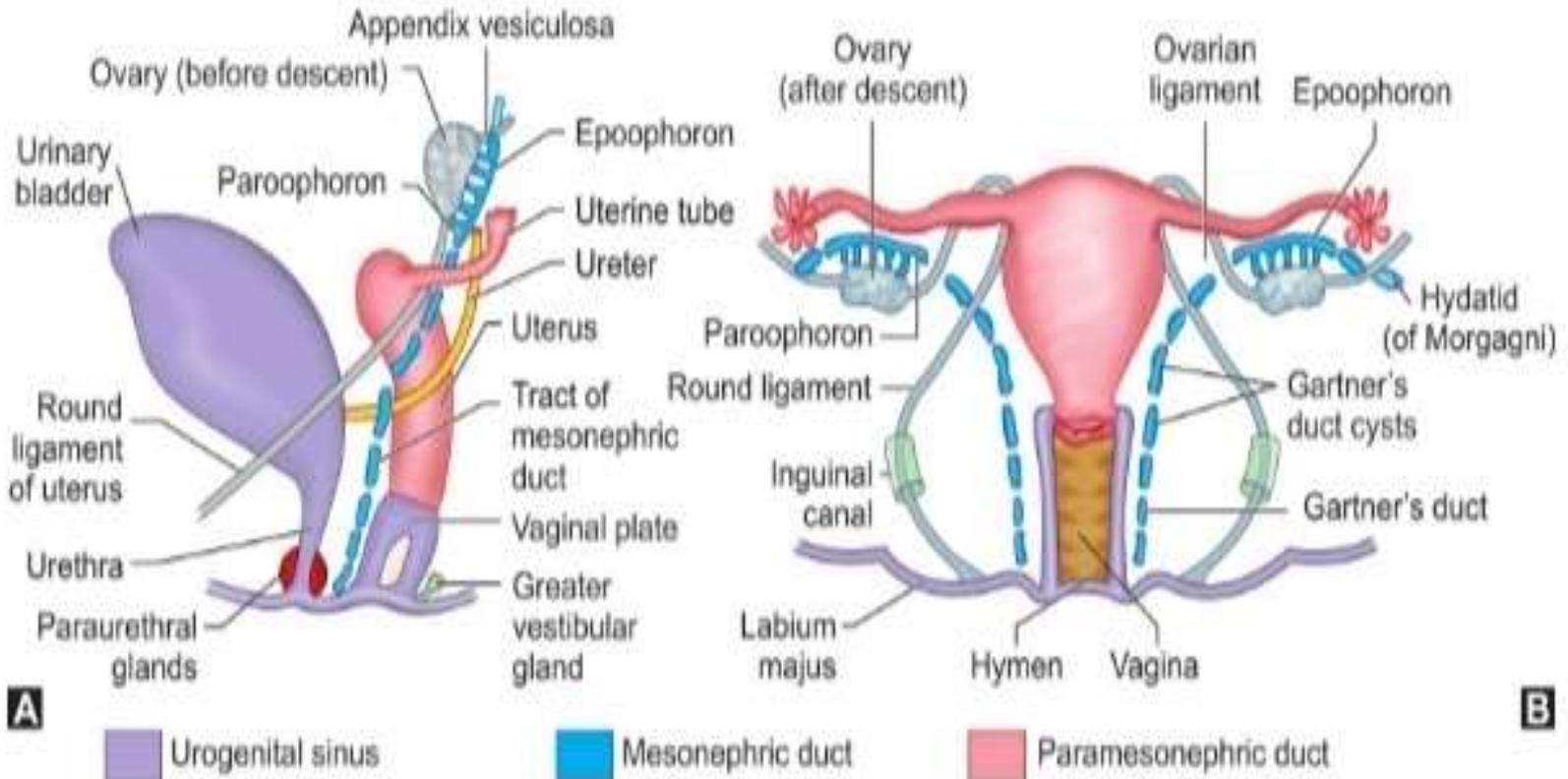
Congenital anomaly of the genitalia is a medical term referring to any physical abnormality of the **male** or **female internal** or **external** genitalia present at birth.



# Congenital malformations of female genital organs

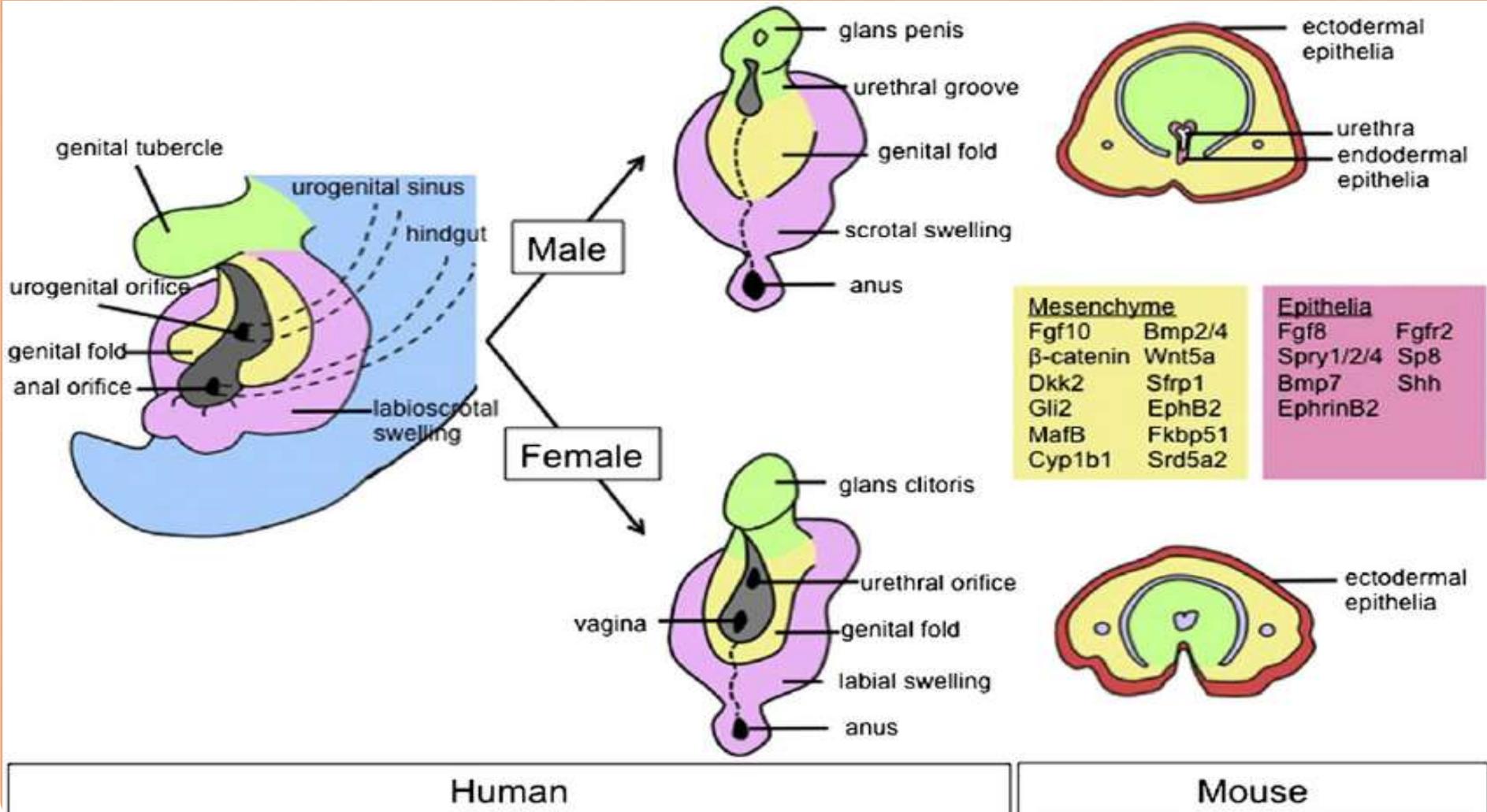
<b>internal</b>	Uterine malformation	Müllerian agenesis Cervical agenesis Unicornuate uterus Uterus didelphys Bicornuate uterus Uterine septum Arcuate uterus
	Vaginal malformation	Vaginal septum Vaginal hypoplasia Vaginal atresia
<b>external</b>	Clitoromegaly Imperforate hymen Progestin-induced virilisation Pseudohermaphroditism	

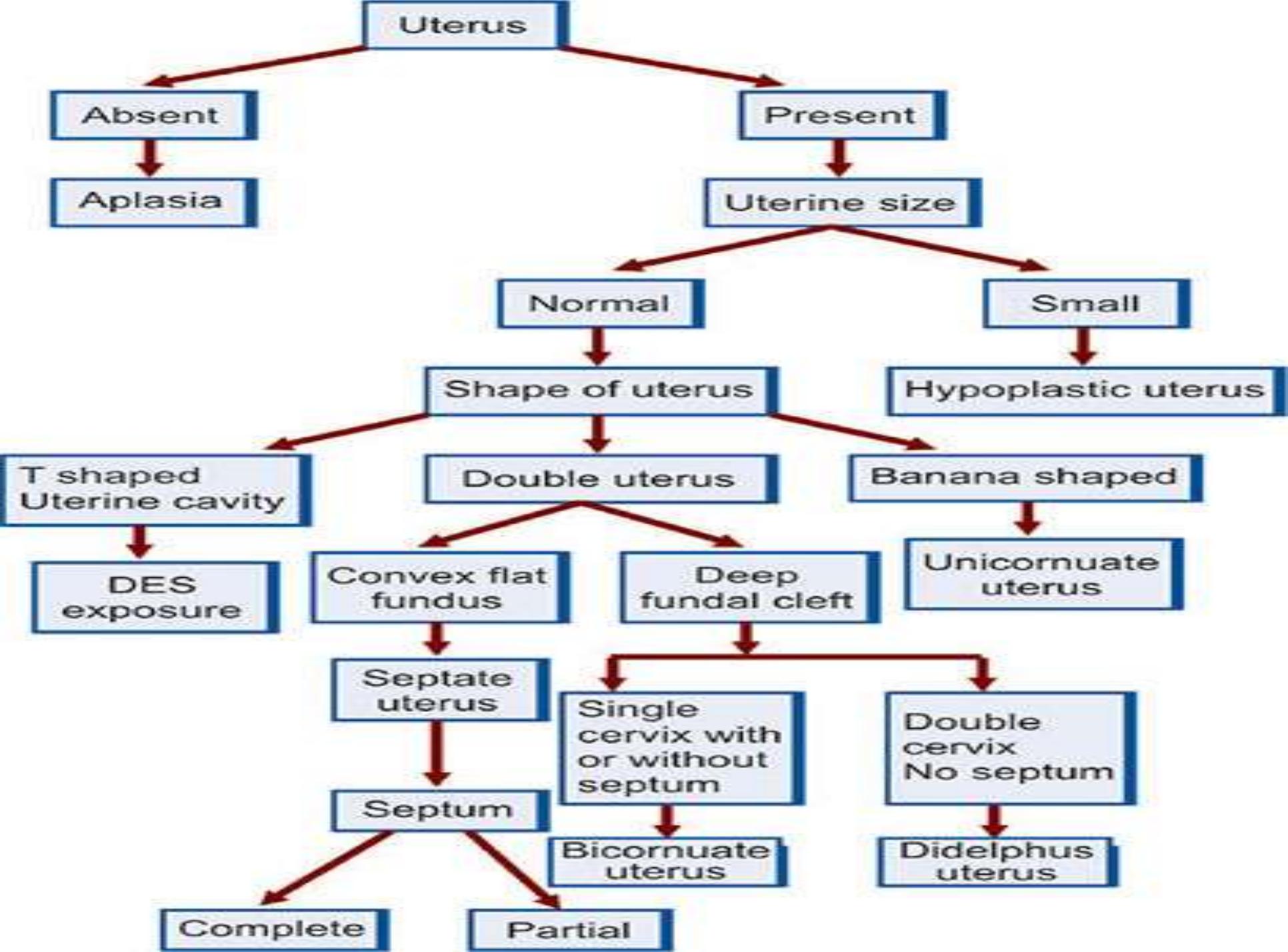
# DEVELOPMENT

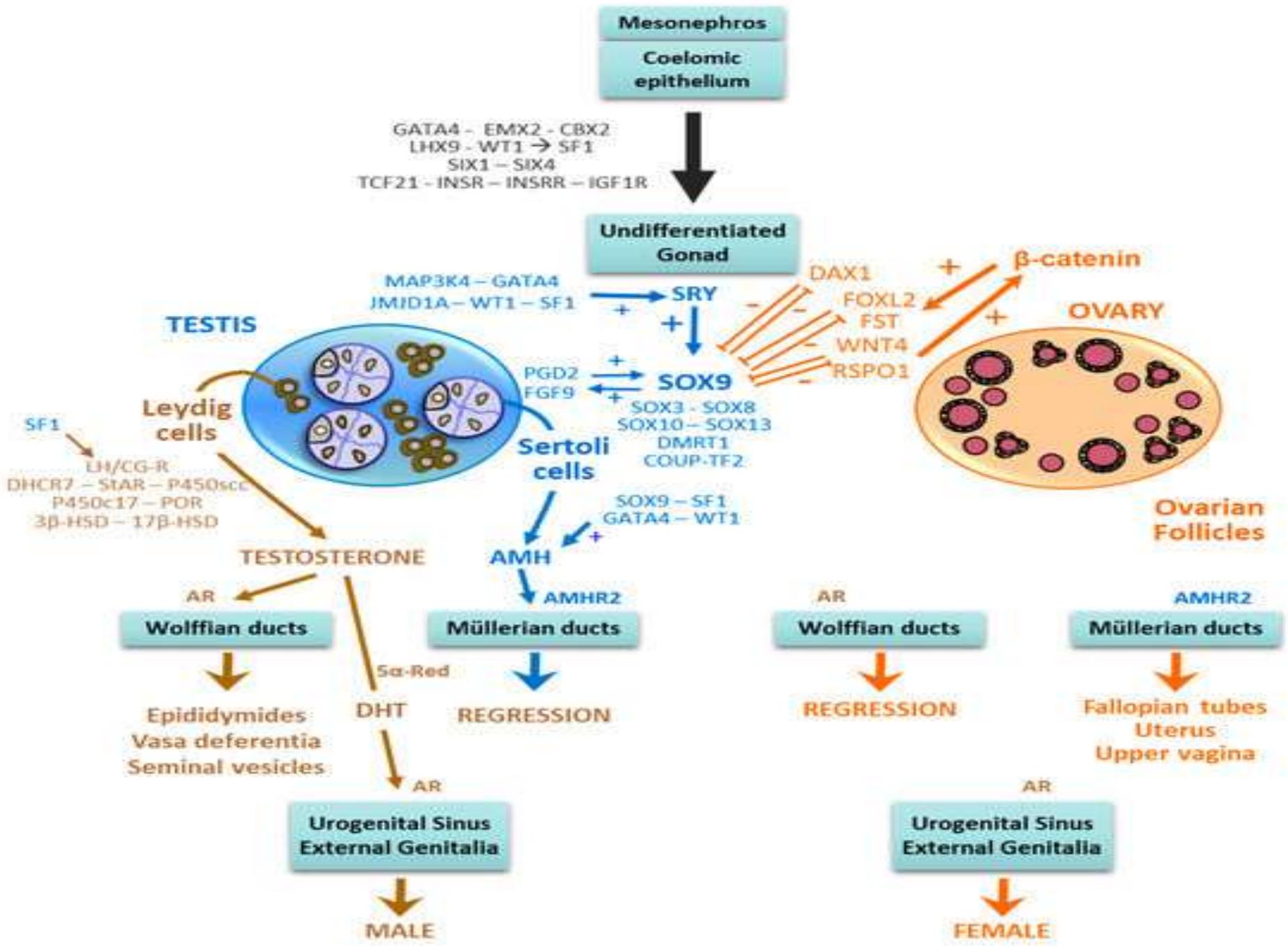


**Fig. 3.3:** Diagrammatic representation showing development of female reproductive systems from the primitive genital ducts. Vestigial structures are also shown. (A) Female fetus at 12 weeks; (B) Newborn female

# DEVELOPMENTAL ANOMALIES OF THE EXTERNAL GENITALIA

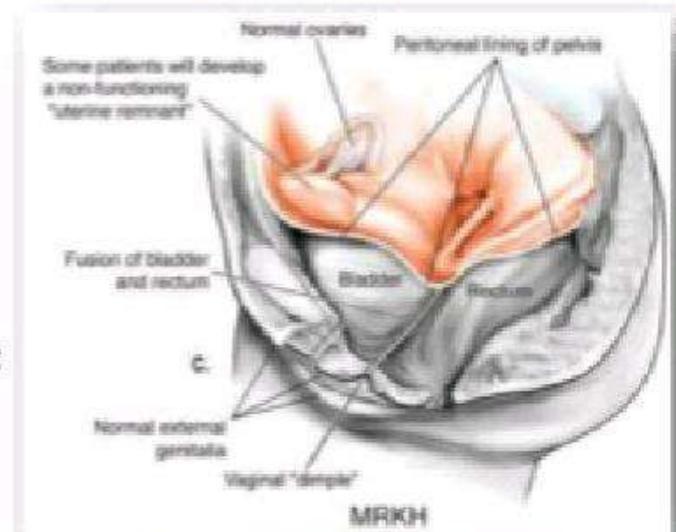






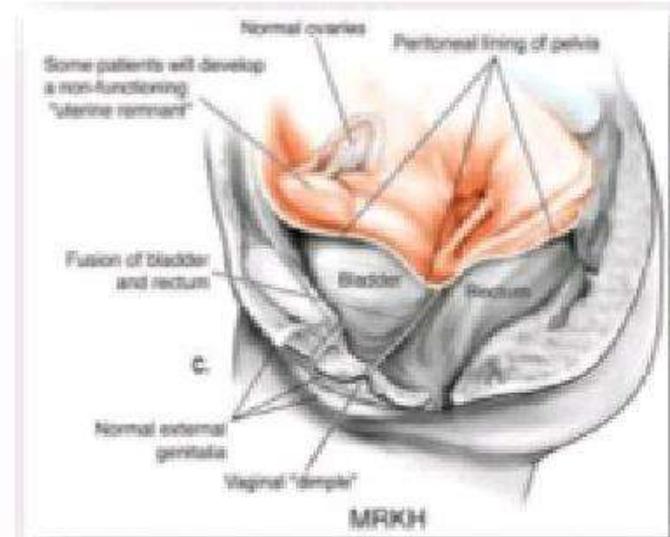
# Müllerian agenesis = Mayer-Rokitansky-Küster-Hauser Syndrome (MRKH)

- Congenital absence of the uterus and vagina (small rudimentary uterine bulbs are usually present with rudimentary fallopian tubes)
  - Menstruation does not appear at the usual age because the uterus is absent
- Normal ovarian function, including ovulation
- Phenotypic sex: female (normal development of breasts, body proportions, hair distribution, and external genitalia)



# Müllerian agenesis = Mayer-Rokitansky-Küster-Hauser Syndrome (MRKH)

- Genetic sex: female (46, XX karyotype)
- Frequent association of other congenital anomalies (skeletal, urologic, and especially renal)
- 15% of primary amenorrhea
- Normal female range testosterone level
- First seen by a gynecologist at age 14 to 15 years, when the absence of menses causes concern



# UTERINE ANOMALIES

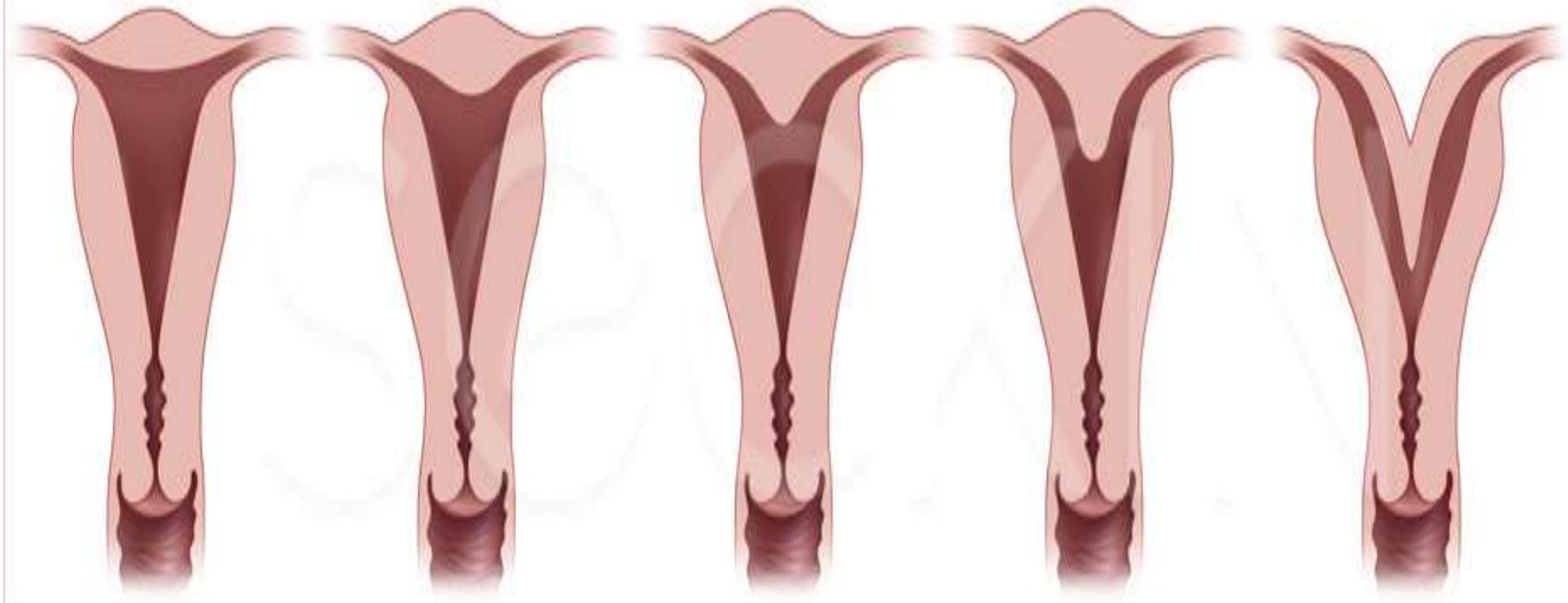
Normal

Mild Arcuate

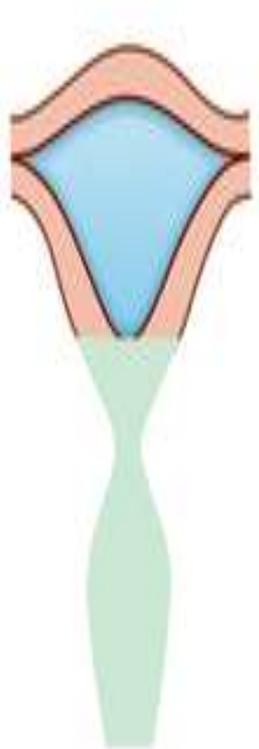
Moderate Arcuate

Severe Arcuate

Bicornuate



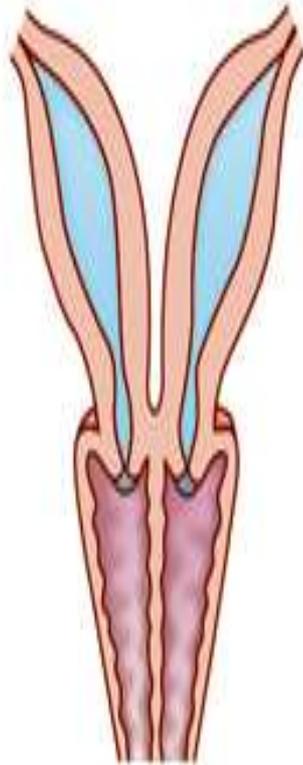
# UTERINE ANOMALIES



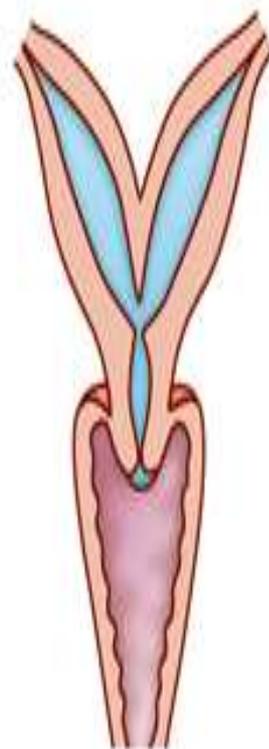
Cervicovaginal  
agenesis



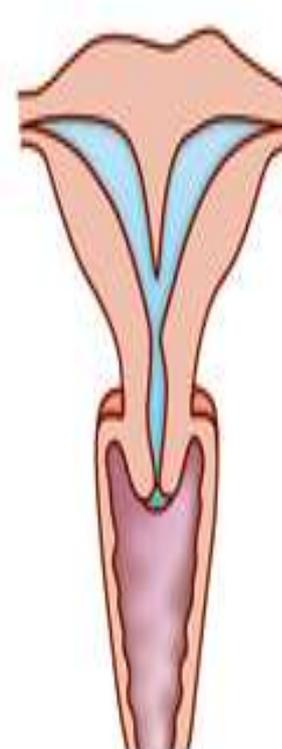
Uterus  
unicornis



Uterus  
didelphys



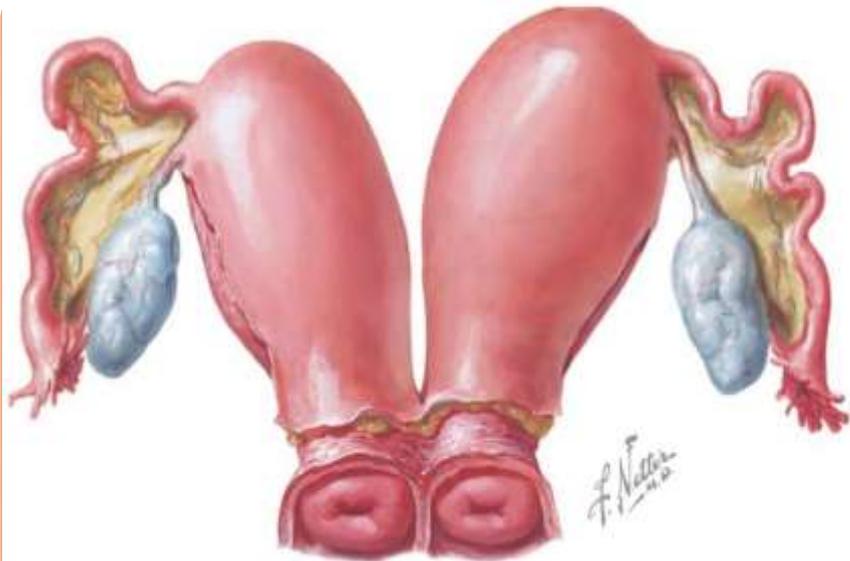
Uterus  
bicornis unicollis



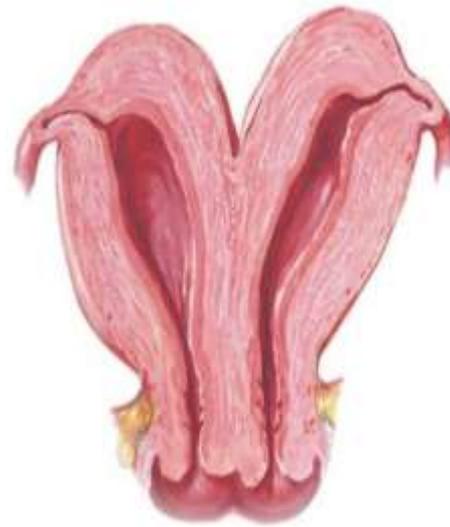
Uterus  
septus



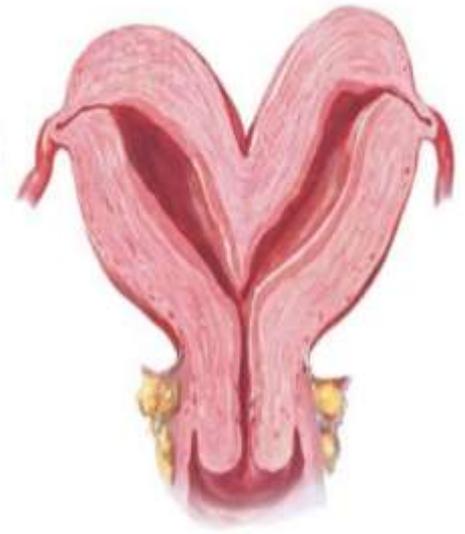
Rudimentary  
horn  
(non-communicating)



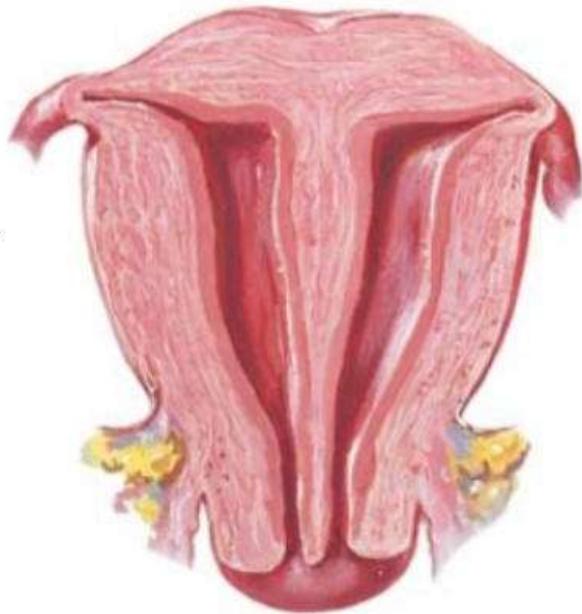
Uterus didelphys (uterus duplex separatus)



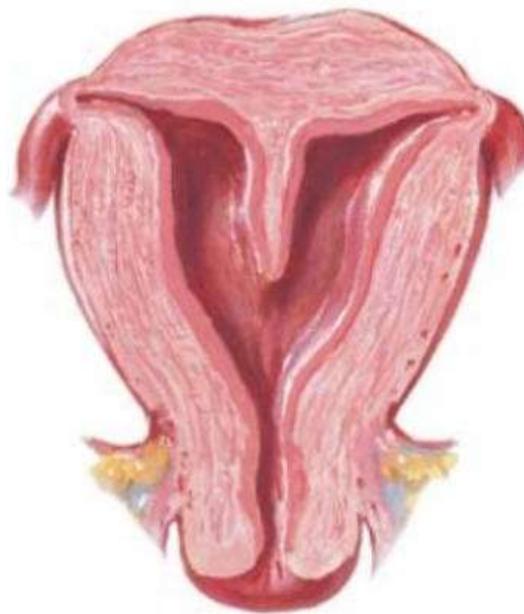
Uterus duplex bicornis (septus)



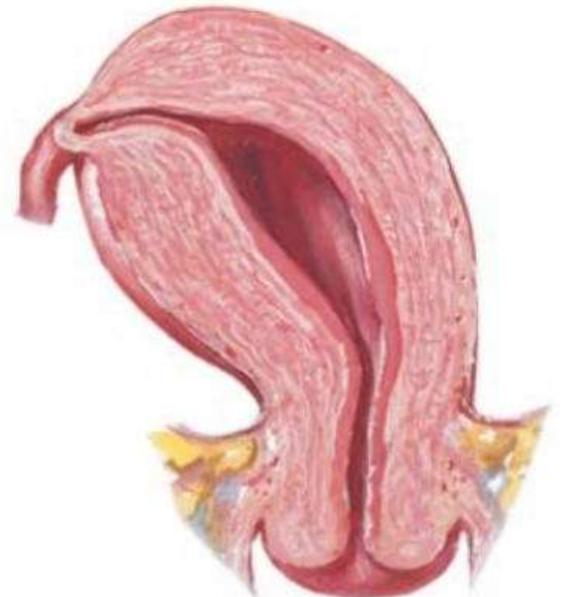
Uterus bicornis unicollis



Uterus septus



Uterus subseptus



Uterus unicornis

- Class I: Müllerian agenesis/Hypoplasia segmental
- Class II: Unicornuate uterus
- Class III: Didelphys uterus
- Class IV: Bicornuate uterus,
- Class V: Septate uterus
- Class VI: Arcuate uterus,
- Class VII: Diethylstilbestrol (DES)-related abnormality.

# UTERINE ANOMALIES

**Complete**

**Incomplete**

**Formation**



**Complete Agenesis**



**Unicornuate**



**Unicornuate with horn**

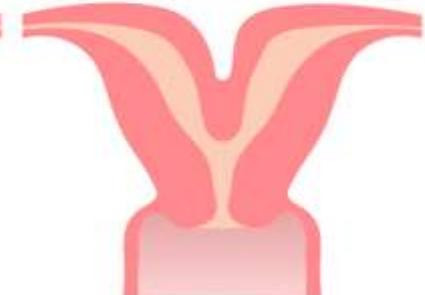
**Fusion**



**Uterus Didelphys**

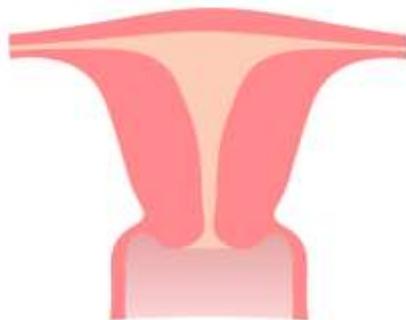


**Complete Bicornuate**

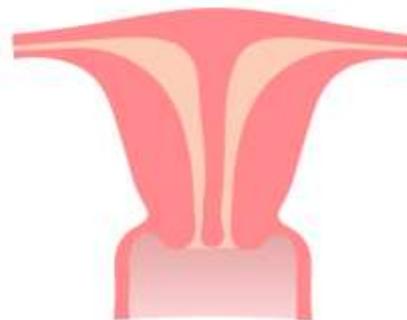


**Partial Bicornuate**

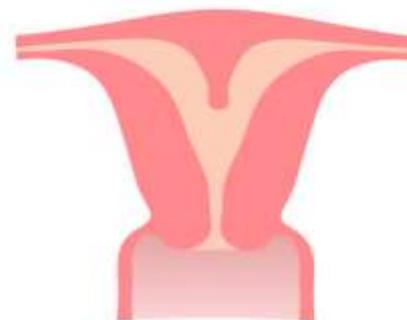
**Resorption**



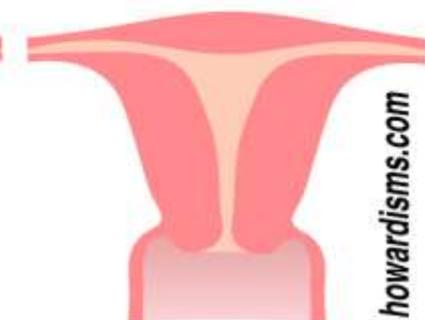
**Normal**



**Complete Septate**



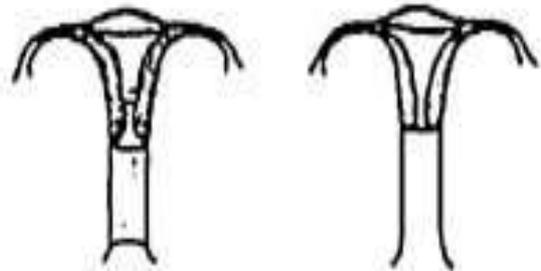
**Partial Septate**



**Arcuate**

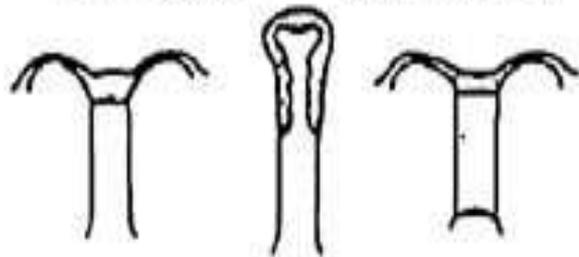
Uterine morphology	Fundal contour	External contour
Normal	Straight/ convex	Uniformly convex or indentation <10 mm
Arcuate	Concave fundal indentation with central point of indentation at obtuse angle (>90)	Uniformly convex or indentation <10 mm
Subseptate	Presence of septum which does not extend to cervix with central point of septum at acute angle (<90)	Uniformly convex or indentation <10 mm
Septate	Presence of uterine septum that completely divides cavity from fundus to cervix	Uniformly convex or indentation <10 mm
Bicornuate	Two well-formed uterine cornua	Fundal indentation >10 mm dividing the two cornua
Unicornuate with or without rudimentary horn	Single well-formed uterine cavity with a single interstitial portion of fallopian tube and concave fundal contour.	

### I Hypoplasia/agenesis



(a) Vaginal

(b) Cervical

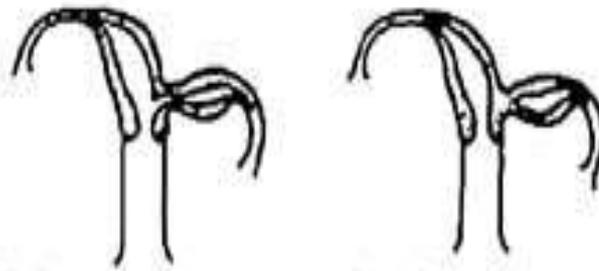


(c) Fundal

(d) Tubal

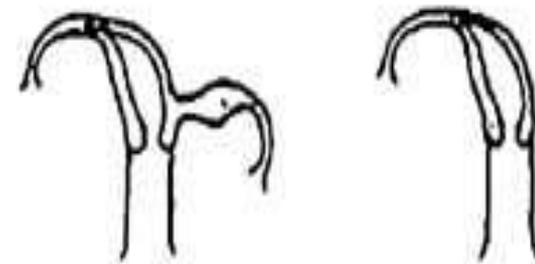
(e) Combined

### II Unicornuate



(a) Communicating

(b) Non Communicating



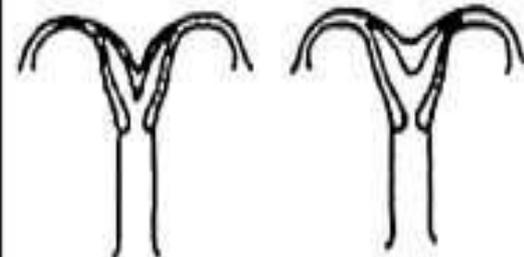
(c) No cavity

(d) No horn

### III Didelphus



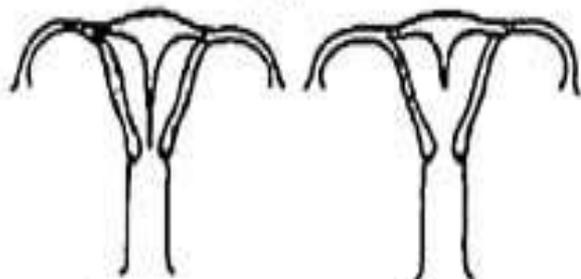
### IV Bicornuate



(a) Complete

(b) Partial

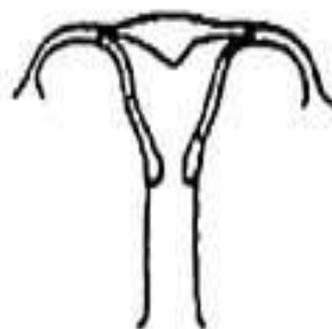
### V Septate



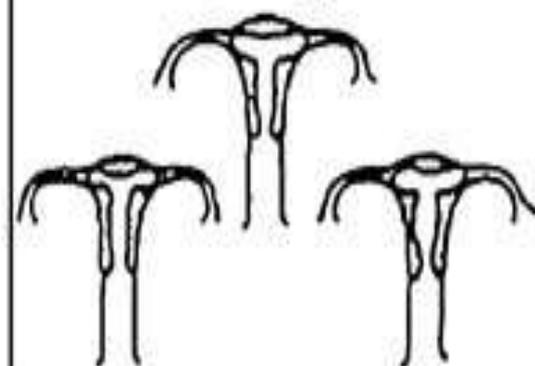
(a) Complete

(b) Partial

### VI Arcuate

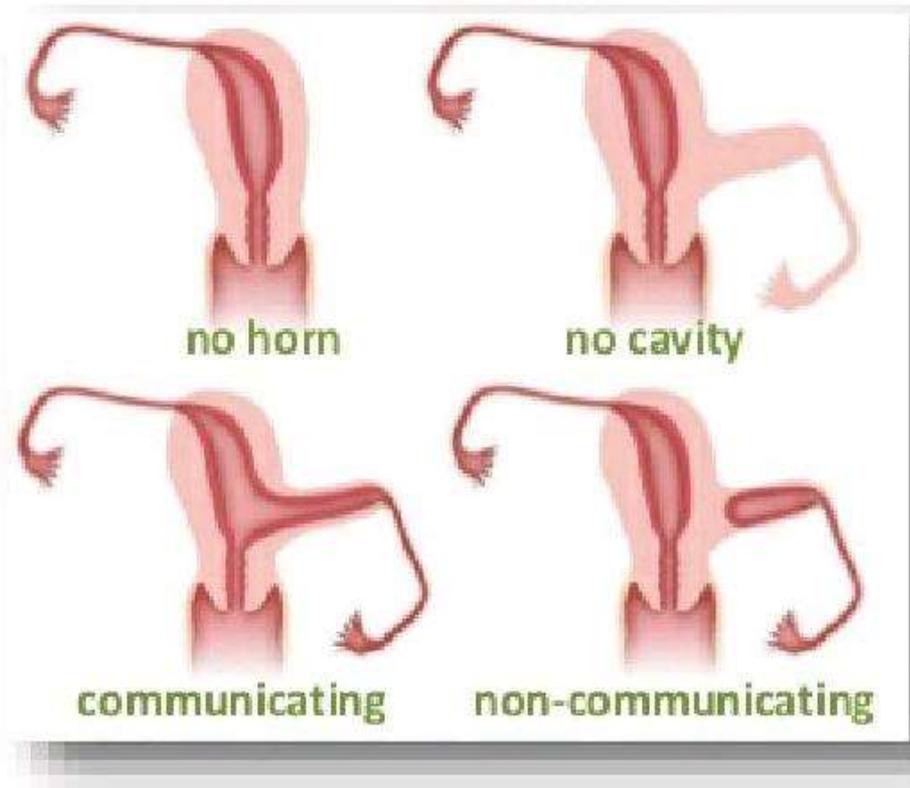


### VII DES drug related



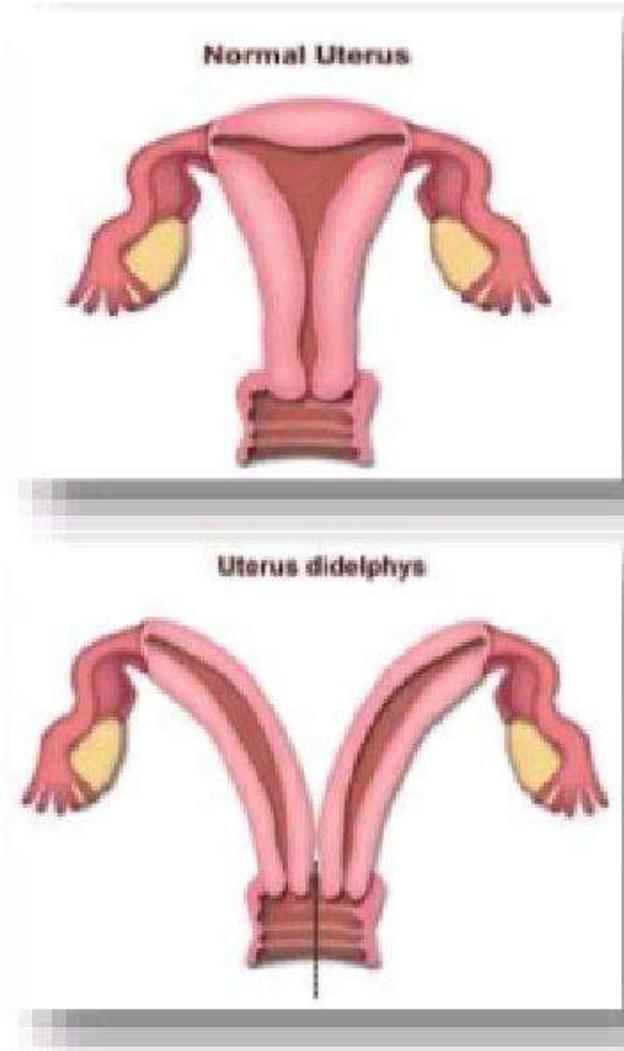
# Unicornuate uterus

- Seen in 1% cases and is associated with renal abnormality on the same side
- Recognized by the absence of round lig. and fallopian tube on the opposite side
- Can cause abortion, preterm labor and IUGR



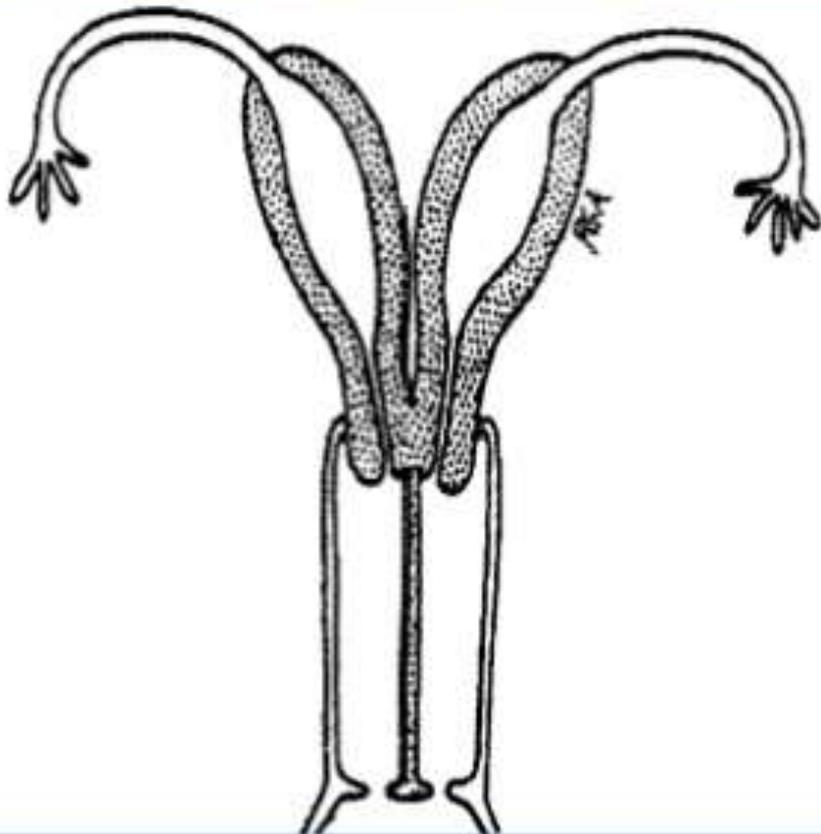
# Uterus didelphys

- Two Mullerian Ducts fails to fuse along the whole of their lengths
- Develop normally and remain separate
- Two vaginas open at the vulva
- A vaginal Septum can be seen
- Two cervixes seen





Didelphys uterus. Note the complete separation but full development of each müllerian duct.



Uterine didelphys with complete vaginal septum

# Bicornuate uterus

- Two Mullerian ducts fuses at the body of the uterus
- **Uterus bicornis bicollis:** Vagina is single but the two cornua remain separated and two cervixes project into the vagina.
- **Uterus bicornis unicollis:** Vagina is single with a single cervix

Normal Uterus

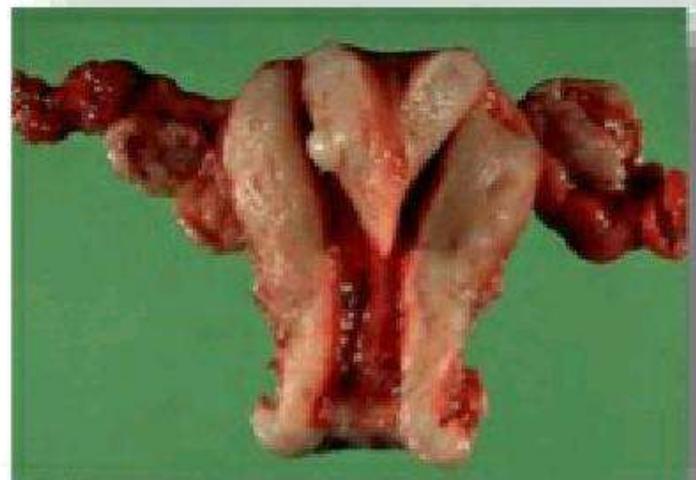
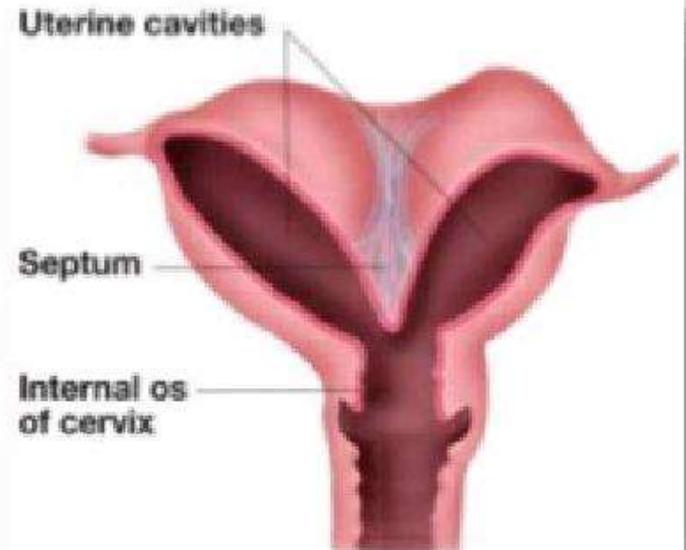


Bicornuate Uterus



# Uterine septum

- Although the two Mullerian ducts have fused, a median septum passes from the fundus of the uterus to the cervix and may extend into the vagina
- In Uterus subseptus, the septum is retracted to the body of the uterus



# CLINICAL FEATURES OF UTERINE ANOMALIES

## Gynecological

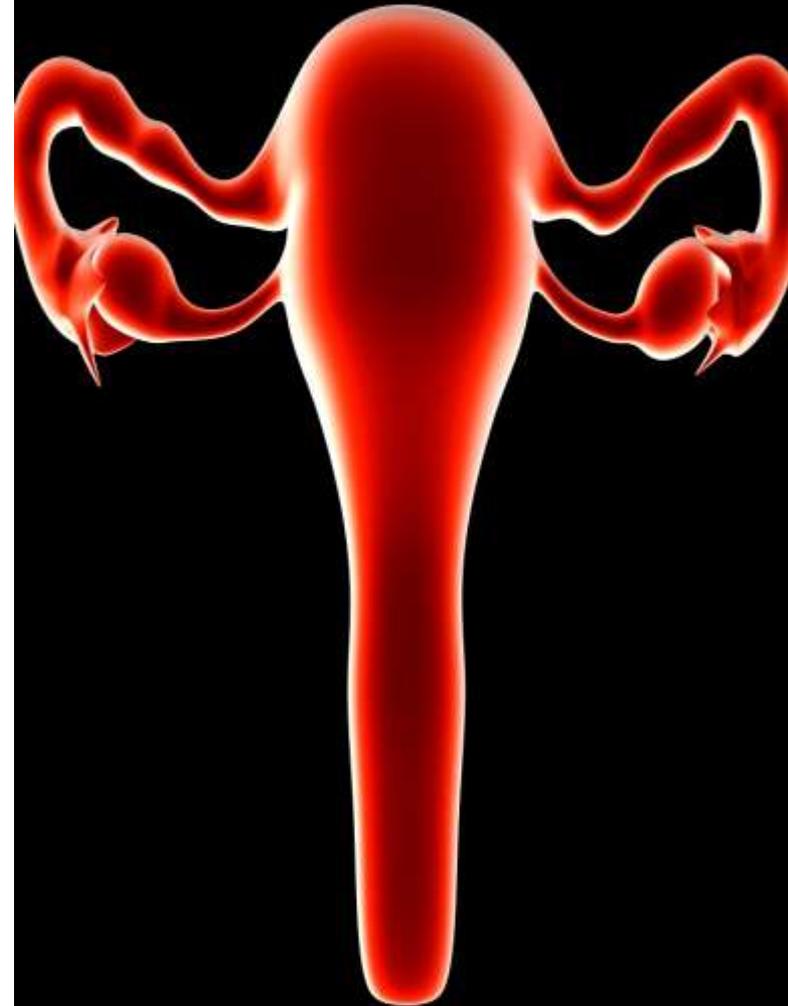
- Infertility and dyspareunia.
- Dysmenorrhea in bicornuate uterus or due to cryptomenorrhea
- Menstrual disorders (menorrhagia, cryptomenorrhea)

## Obstetrical

- Midtrimester abortion
- Rudimentary horn pregnancy
- Cervical incompetence

# CLINICAL PRESENTATION

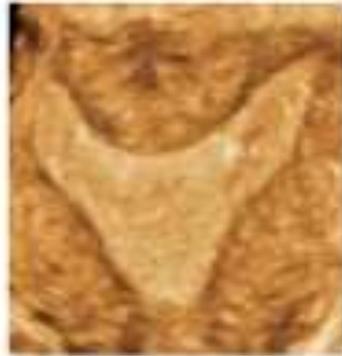
- Spontaneous Abortion
- Premature Delivery
- Infertility
- Abnormal Fetal Lie
- Dystocia at delivery
- Dysmenorrhea, endometriosis
- Cervical incompetence



# RADIOLOGY



Normal uterus



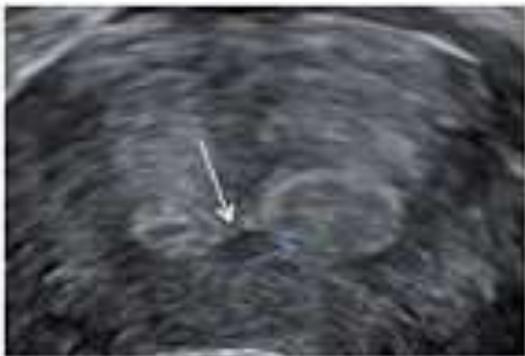
Heart shaped (arcuate) uterus



Septate uterus



Double uterus



Intra-uterine adhesions (arrow)

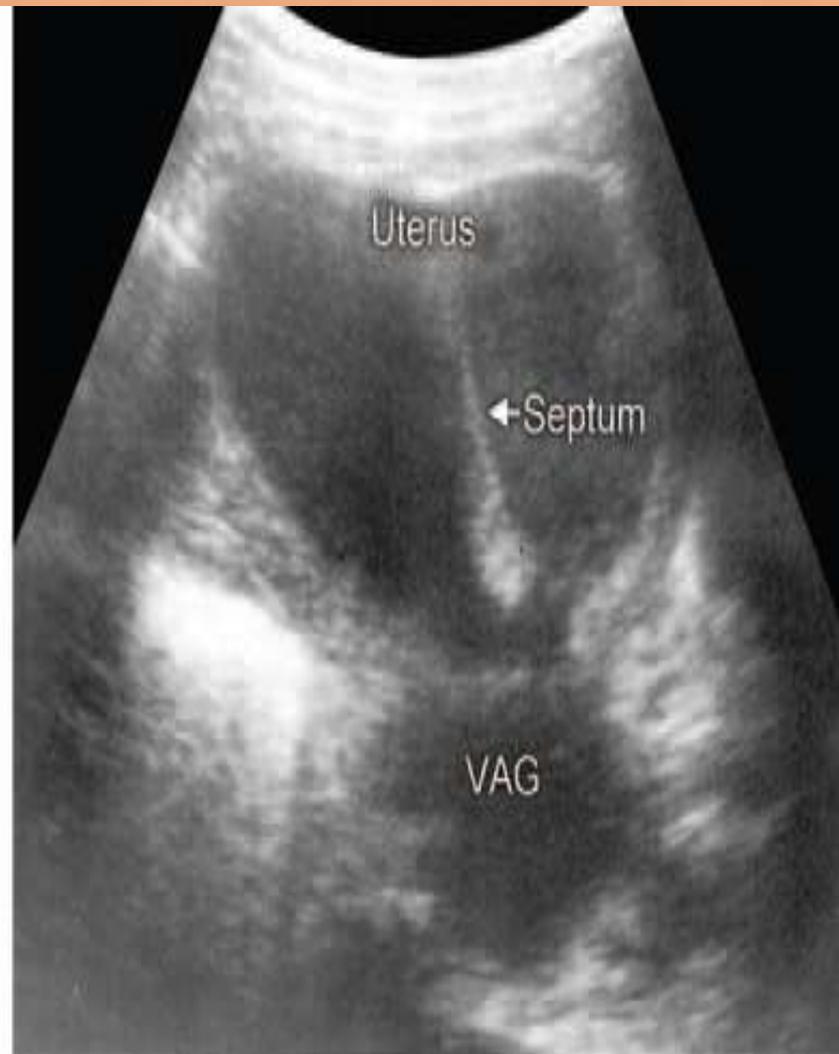


Intra-uterine adhesions on 3D scan (arrow)

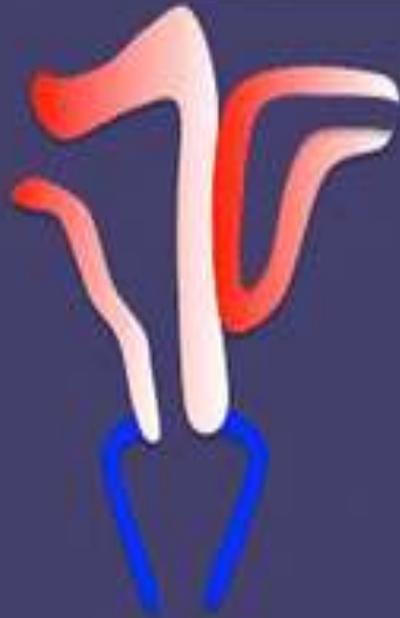


Severe intra-uterine adhesions blocking menstrual flow

# HYSTEROSCOPIC VIEW OF A SEPTATE UTERUS



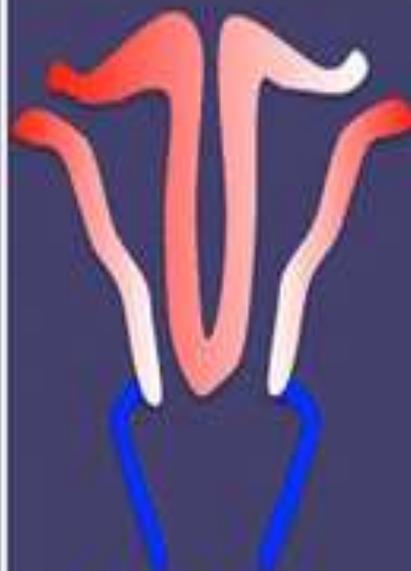
Abnormalities can occur at various stages of embryological development



Incomplete migration/  
development of one  
Mullerian duct



Unicornuate uterus with  
rudimentary horn



Didelphys

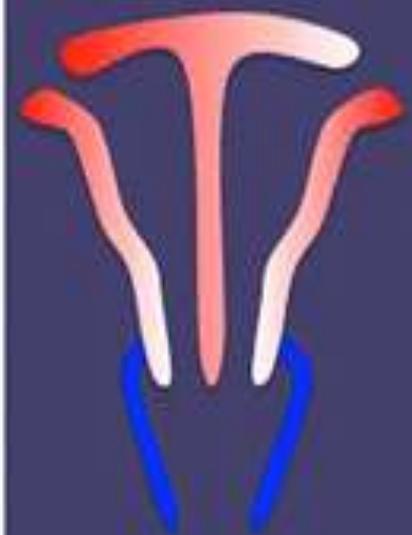


Bicornuate

Complete or incomplete failure of  
Mulleian duct fusion



Didelphys/Bicornuate  
Uterus



Failed resorption of  
median dividing wall  
between two  
Mullerian ducts

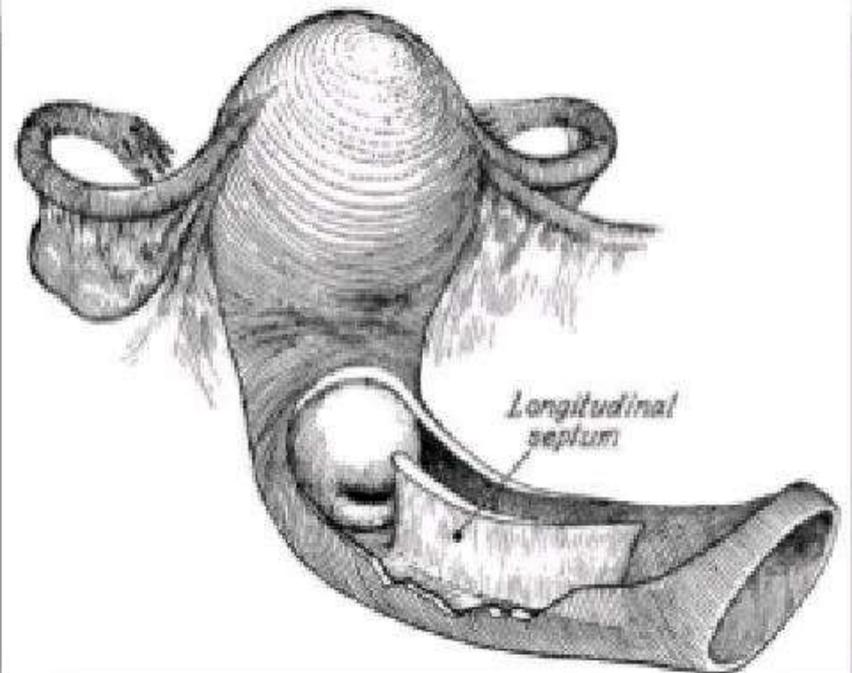
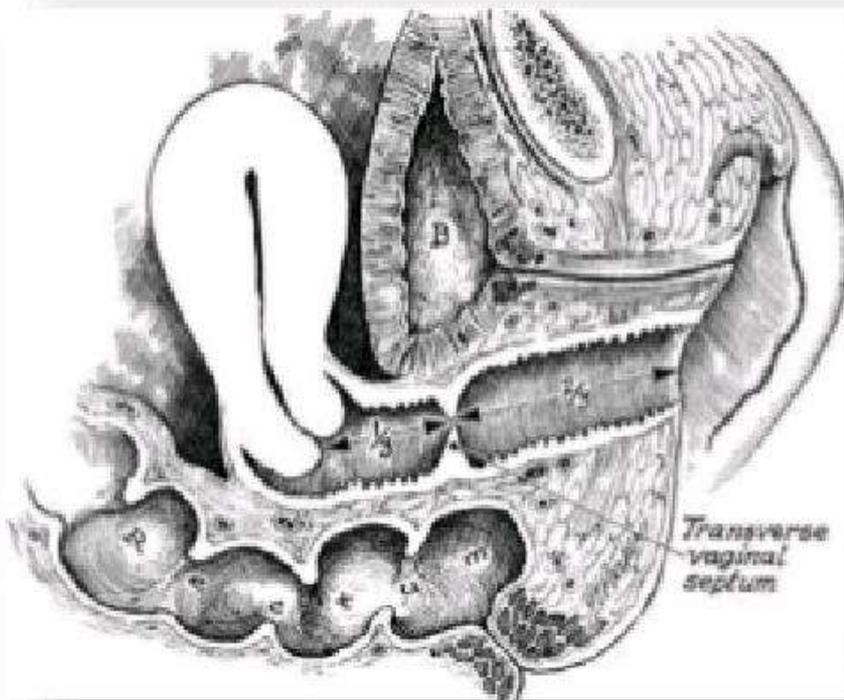


Septate Uterus

# Vaginal septum

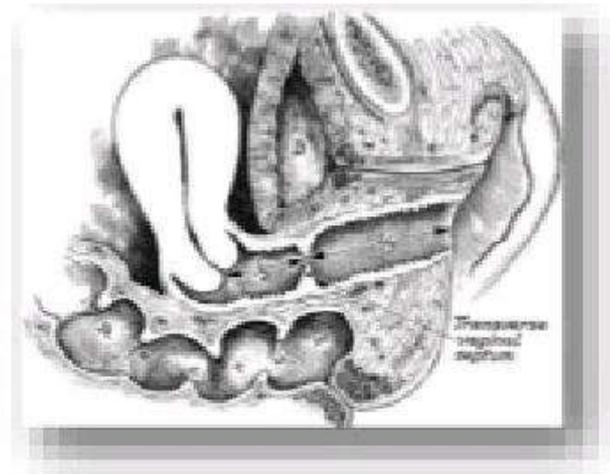
**Transverse Vaginal Septum**

**Longitudinal vaginal septum**



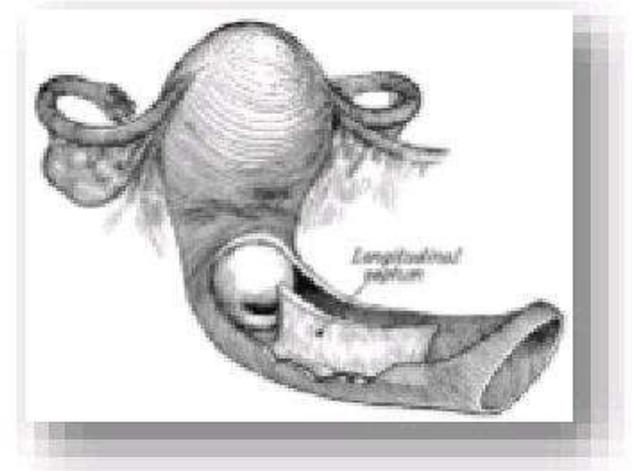
# TRANSVERSE VAGINAL SEPTUM

- Vertical fusion - complete cavitation of the vaginal plate between the sinovaginal bulbs and uterovaginal canal.
- Transverse vaginal septum may be caused by a failure of this process
- incidence of 1 in 70,000 females
- The septum may be obstructive, with accumulation of mucus or menstrual blood, or may be non-obstructive, allowing for flow of mucus and blood.



# Longitudinal vaginal septum

- Results from defective lateral fusion and incomplete reabsorption of the paired müllerian ducts.
- These septa are generally seen with partial or complete duplication of the cervix and uterus.
- Pt complain of difficulty with intercourse or with complaints of vaginal bleeding



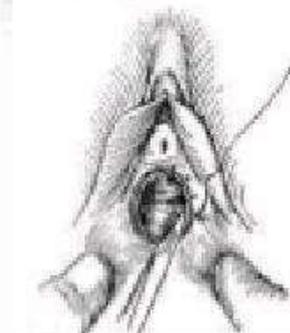
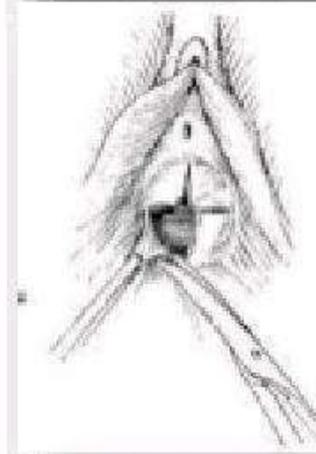
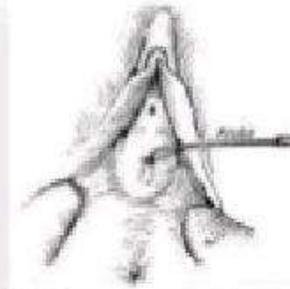
# Imperforate hymen

- By the 12th week, the paramesonephric ducts and/or upper vagina joins with the vaginal plate, which canalizes beginning caudally and creates the lower vagina.
- By the fifth month of gestation, the canalization of the vagina is complete.



# Imperforate hymen

- Clinical manifestations:
  - Classic appearance of bulging, blue-domed, translucent membrane
  - Cyclic pelvic pain due to hematocolpos, hematometria
- Diagnosis:
  - Pelvic/ Rectal exam
  - Ultrasound
  - MRI
- Treatment:
  - Cruciate incision



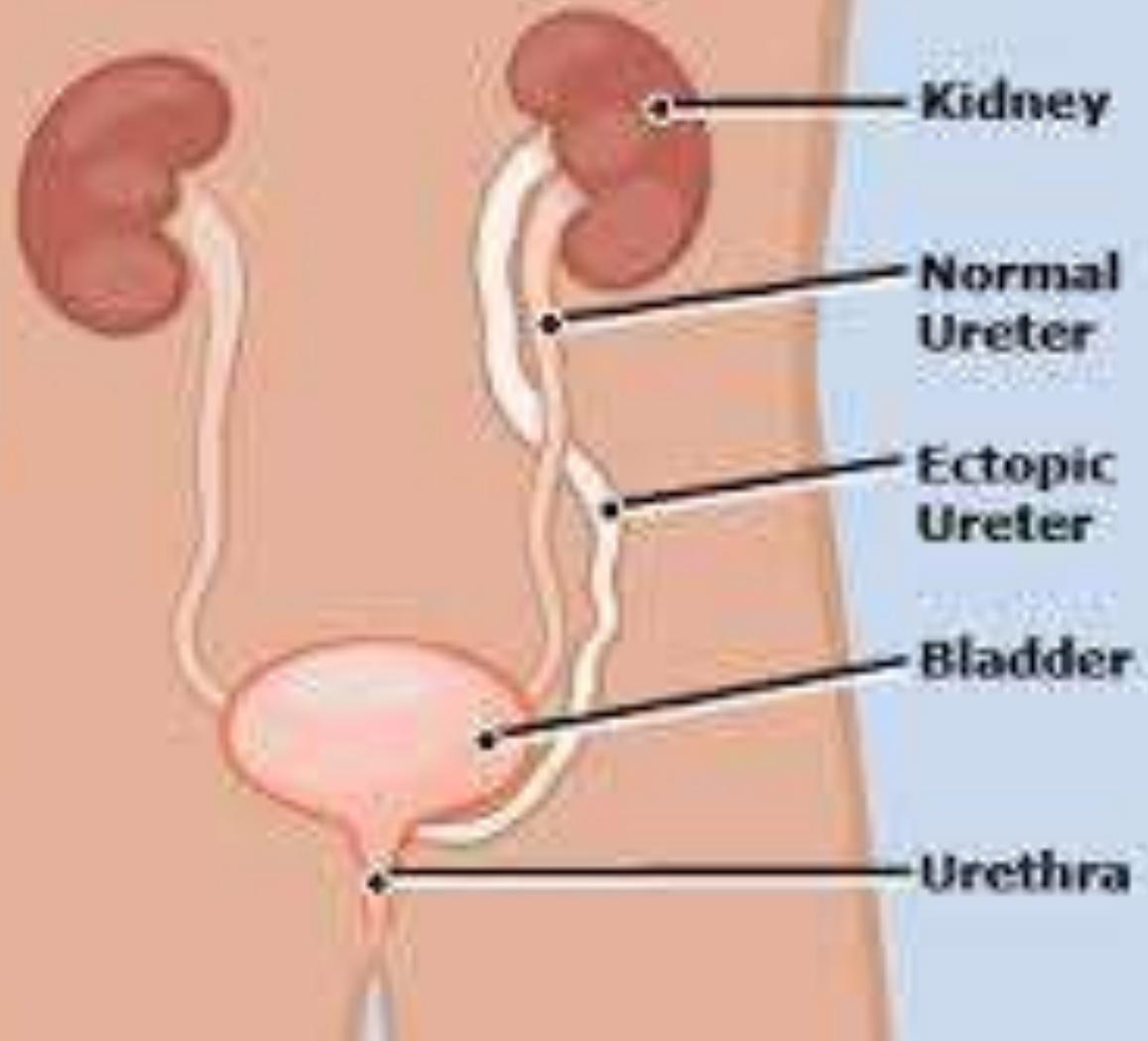
# PERINEAL OR VESTIBULAR ANUS

## PERINEAL OR VESTIBULAR ANUS

---

- Detected at the time of birth
- Usual anal opening site is evidenced by anal pit
- Either anus will be situated close to the posterior end of the vestibule or in the vestibule.
- In vagina (rarely) → (congenital rectovaginal fistula)
- Opening is usually sufficiently big
- Future reproduction → not a problem
- Ceaserian section advised in the future

# ECTOPIC URETER



## DEVELOPMENTAL ANOMALIES OF THE EXTERNAL GENITALIA

### PERINEAL OR VESTIBULAR ANUS

- ✓ The entity is detected at birth
- ✓ The anal opening is situated either close to the posterior end of the vestibule or in the vestibule.
- ✓ There is no problem in future reproduction
- ✓ The delivery should be by cesarean section

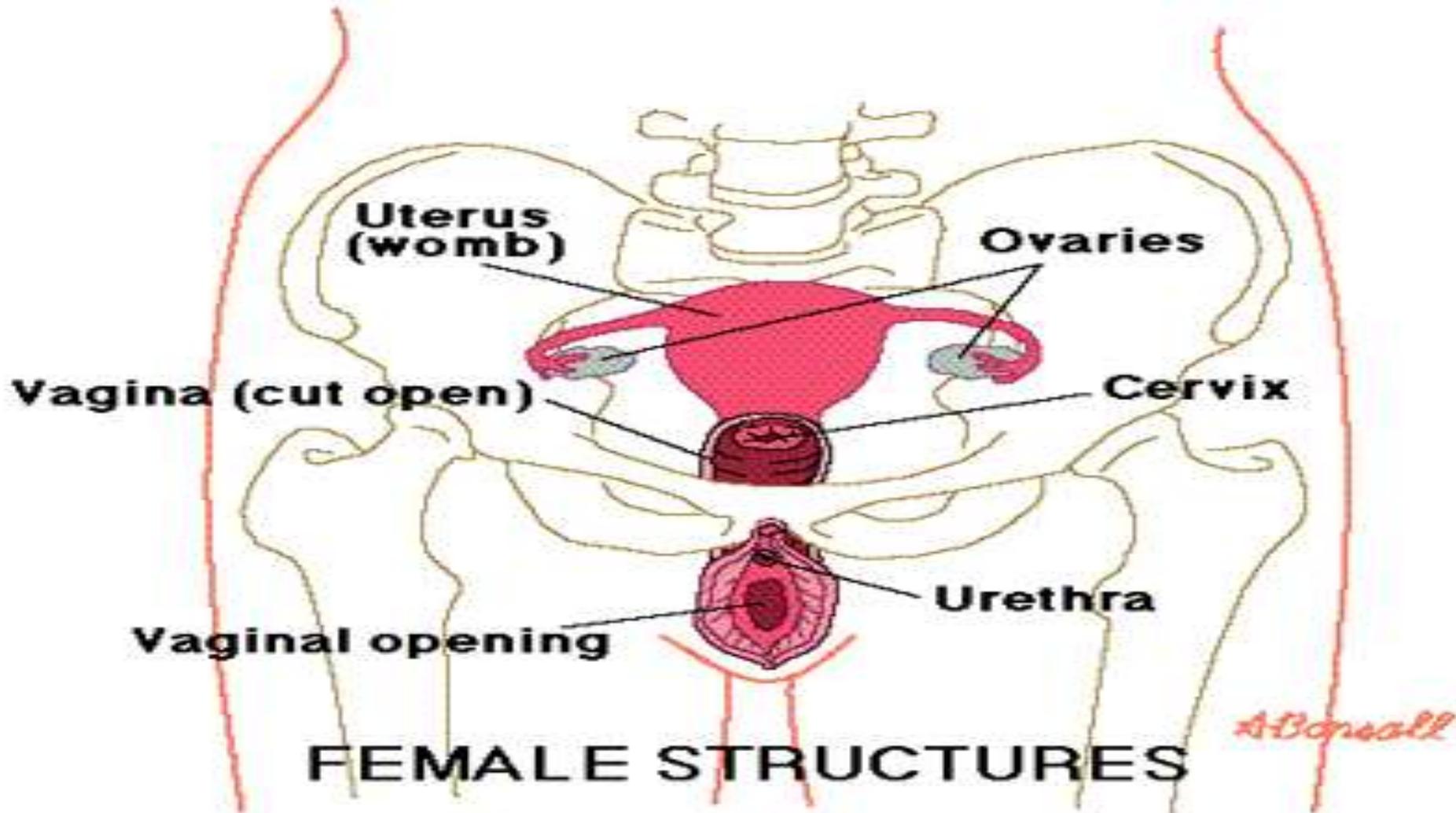
### ECTOPIC URETER

- ❑ The additional ureteric opening is usually in the vestibule close to the urethra or in the vagina
- ❑ Symptom is uncontrollable wetness
- ❑ Partial nephrectomy and ureterectomy may be indicated

# VAGINAL ABNORMALITIES

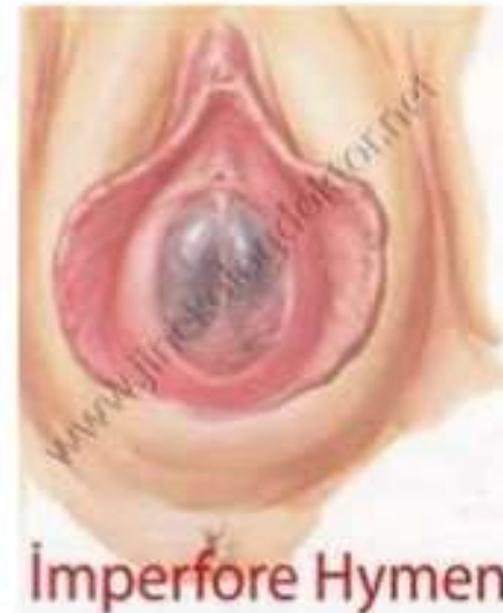
- Narrow introitus
- Hymen abnormality
- Septum
- Agenesis
- Associated abnormalities

# NARROW INTROITUS



# HYMEN ABNORMALITY

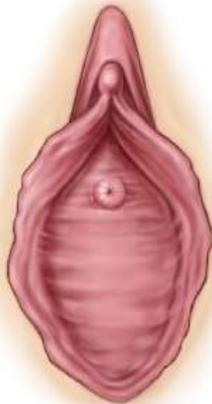
- Imperforate hymen → of significance abnormality.
  - Always unnoticed until 14 – 16 yrs.
  - Uterine functioning is normal → menstrual blood is pent up inside vagina behind the hymen (**CRYPTOMENORRHOEA**)
  - Depending upon the amount of blood so accumulated, it first distends the vagina (**HAEMATOCOLPOS**)
  - The uterus is next involved and cavity dilated (**HAEMATOMETRA**)
  - If late and neglected, tubes may also be distended after the fimbrial ends becoming closed by adhesions (**HAEMATOSALPINX**)



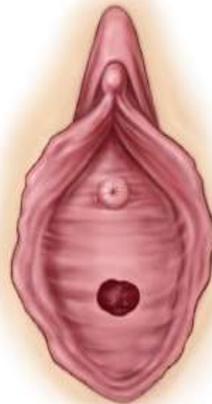
# HYMEN ABNORMALITY

## Hymen types

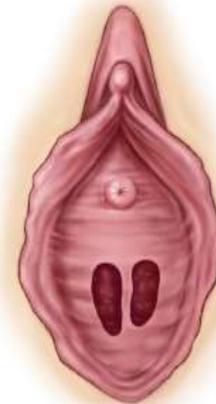
Imperforate hymen



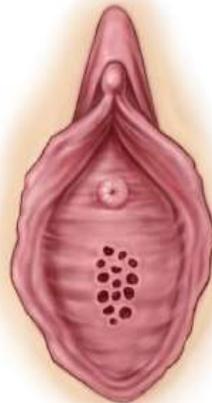
Annular hymen



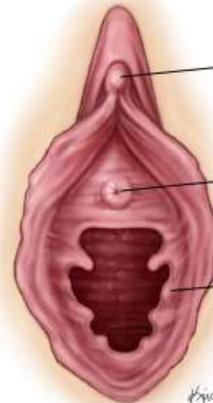
Septate hymen



Cribiform hymen



Parous introitus hymen



Clitoris

Urethral opening

Hymen

*Kristina DeRycke*

# TYPES



**Normal**



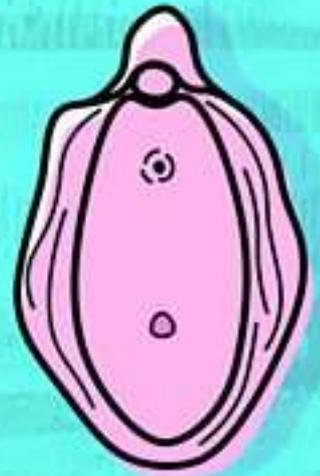
**Septate**



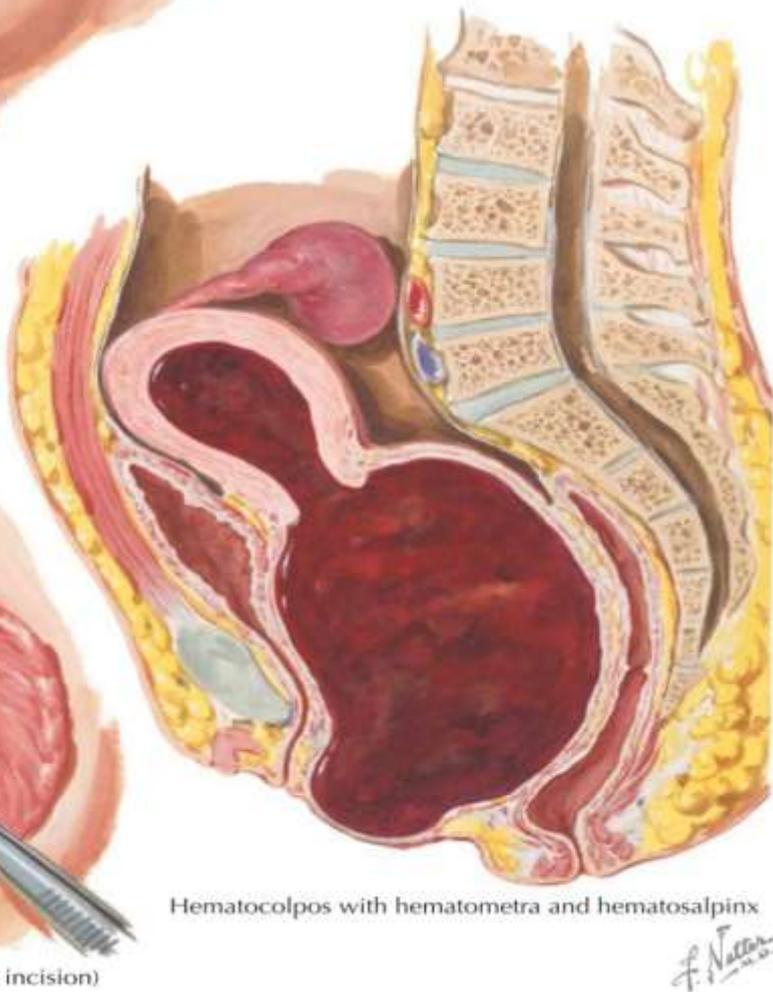
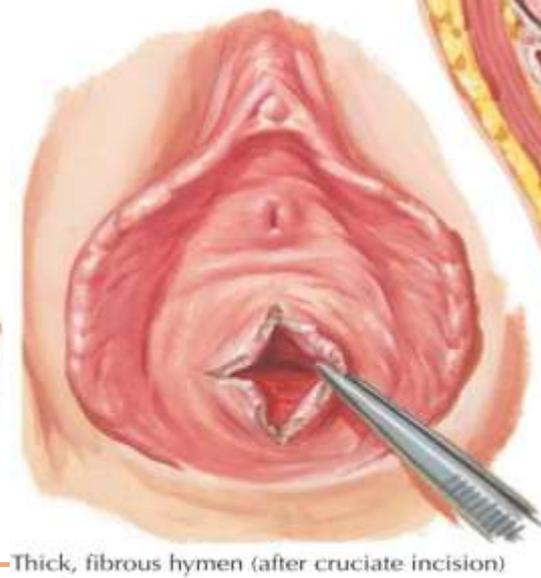
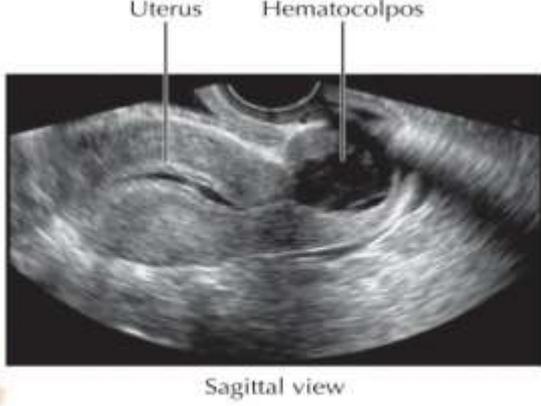
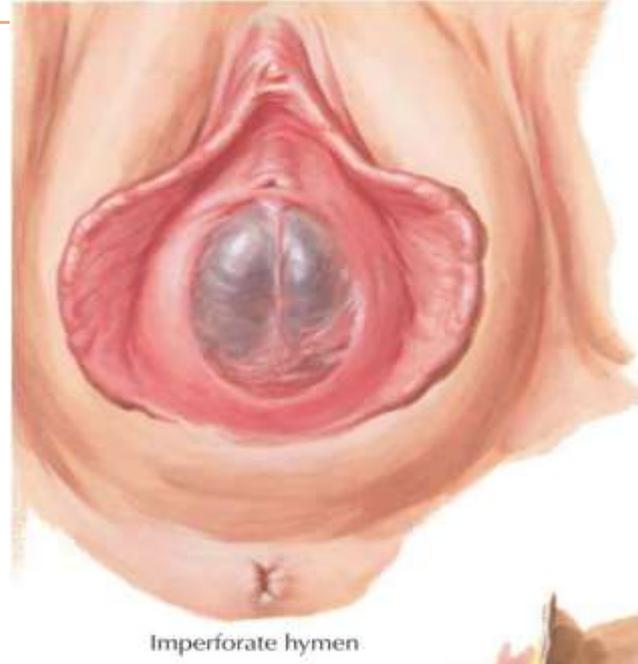
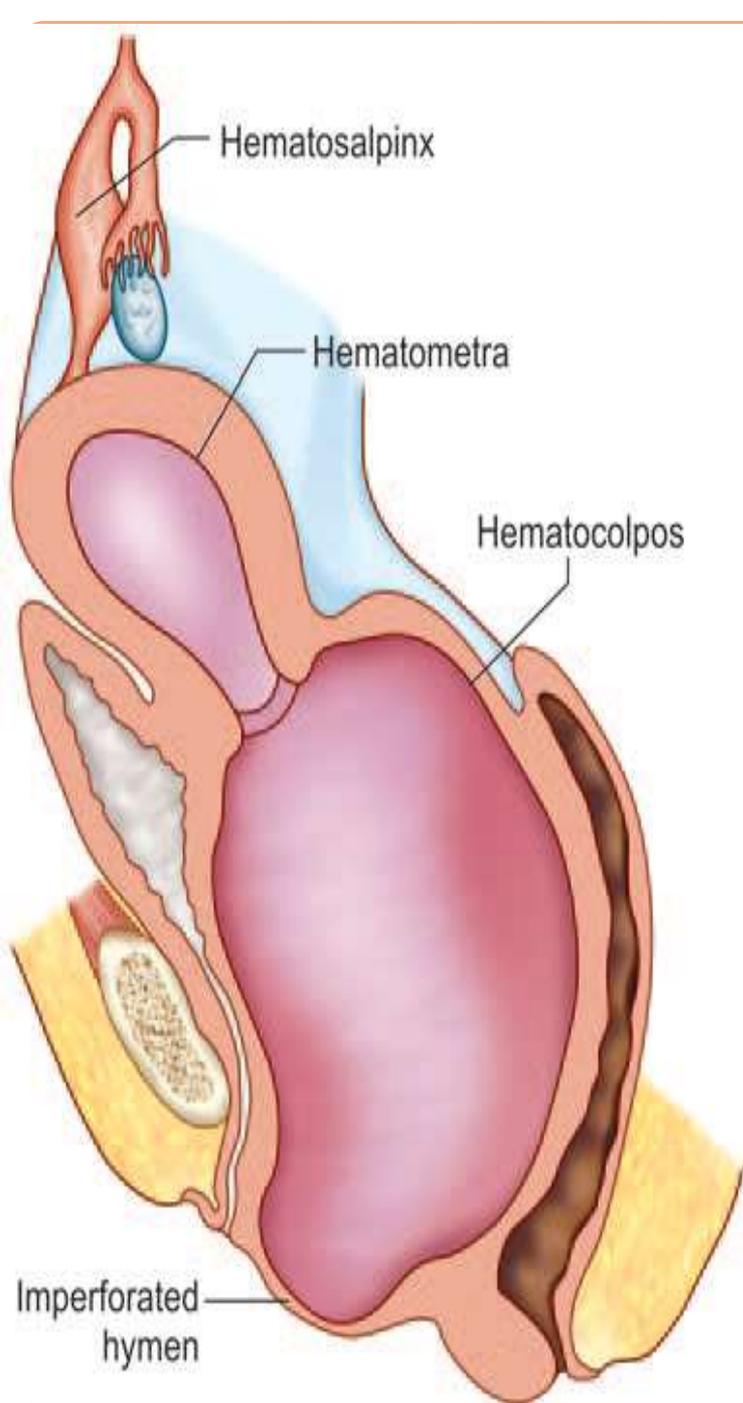
**Cribriform**



**Imperforate**

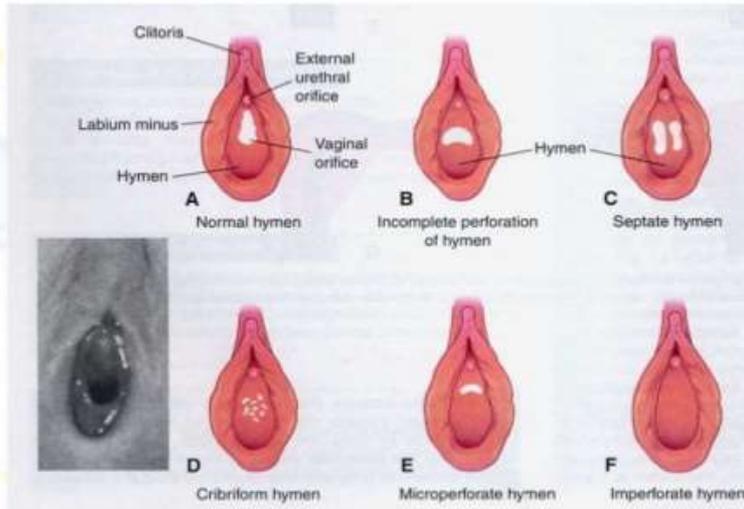


**Microperforate**



F. Netter M.D.

## Congenital anomalies of hymen

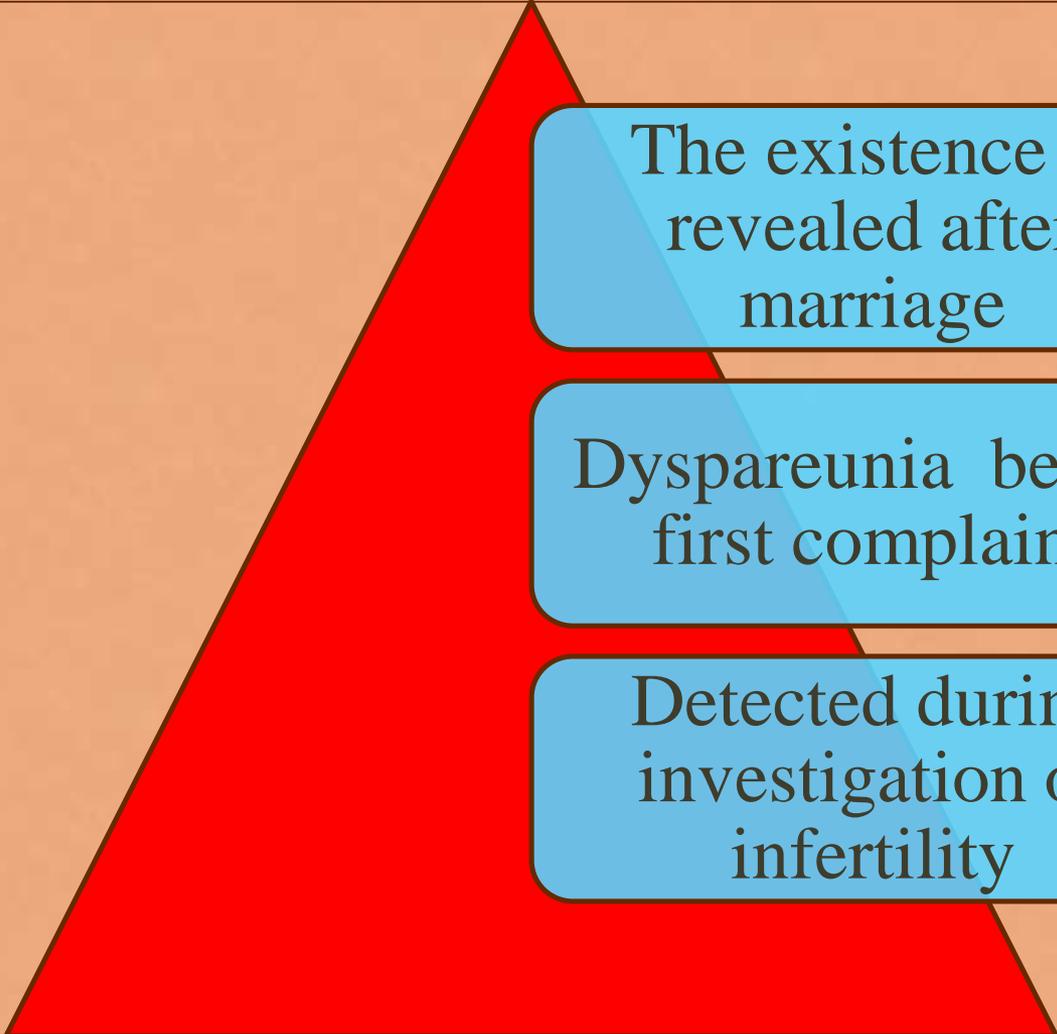


### imperforate hymen

- is a congenital disorder where a hymen without an opening completely obstructs the vagina.
- It is caused by a failure of the hymen to perforate during fetal development.
- It is the most common **obstructive** anomaly of the female reproductive tract.
- occurs in approximately 1 /1000 newborn girls.



# REASONS



The existence is revealed after marriage

Dyspareunia be the first complaint

Detected during investigation of infertility

**Imperforate hymen; due to failure of disintegration of the central cells of the Müllerian eminence that projects into the urogenital sinus**

**The girl is aged about 14–16 years**

**The chief complaints are periodic lower abdominal pain, which may be continuous, primary amenorrhea and urinary symptoms**

## C/F:

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- Age – 14 -16
- Periodic lower abdominal pain which may be continuous.
- Primary amenorrhoea
- Urinary symptoms

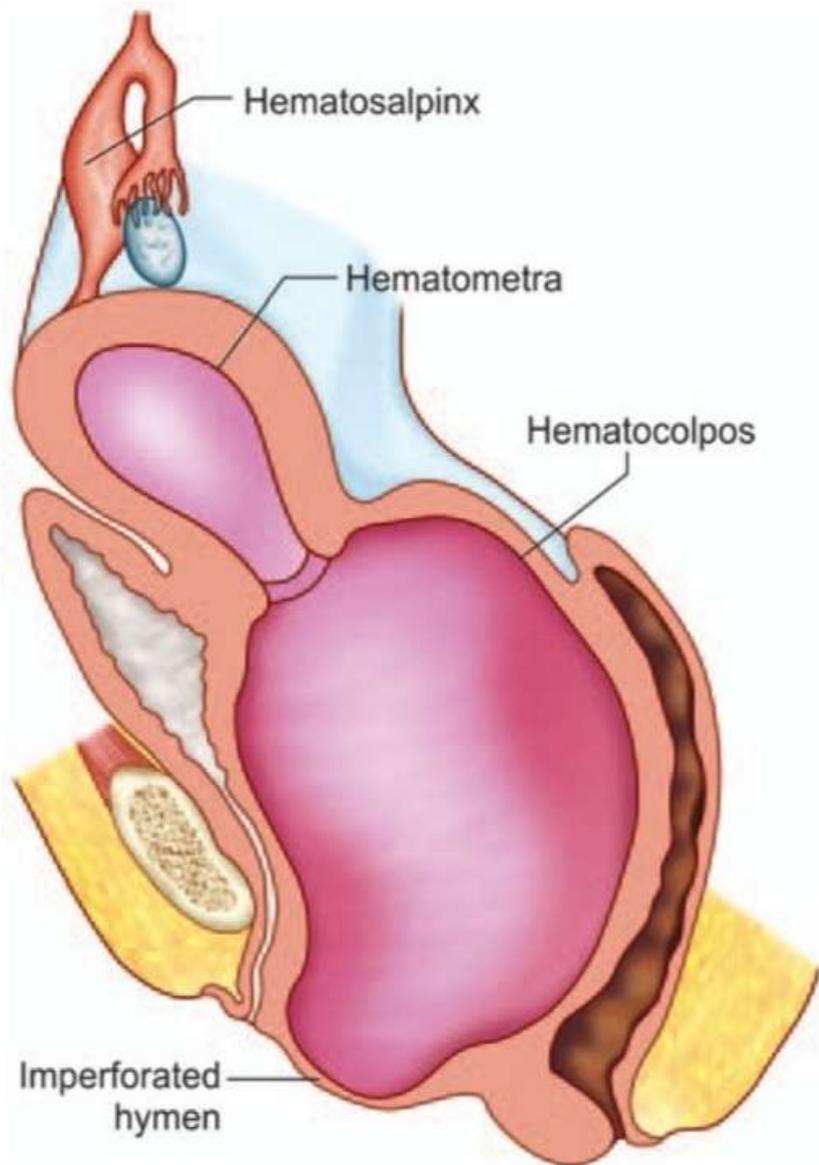
## On examination

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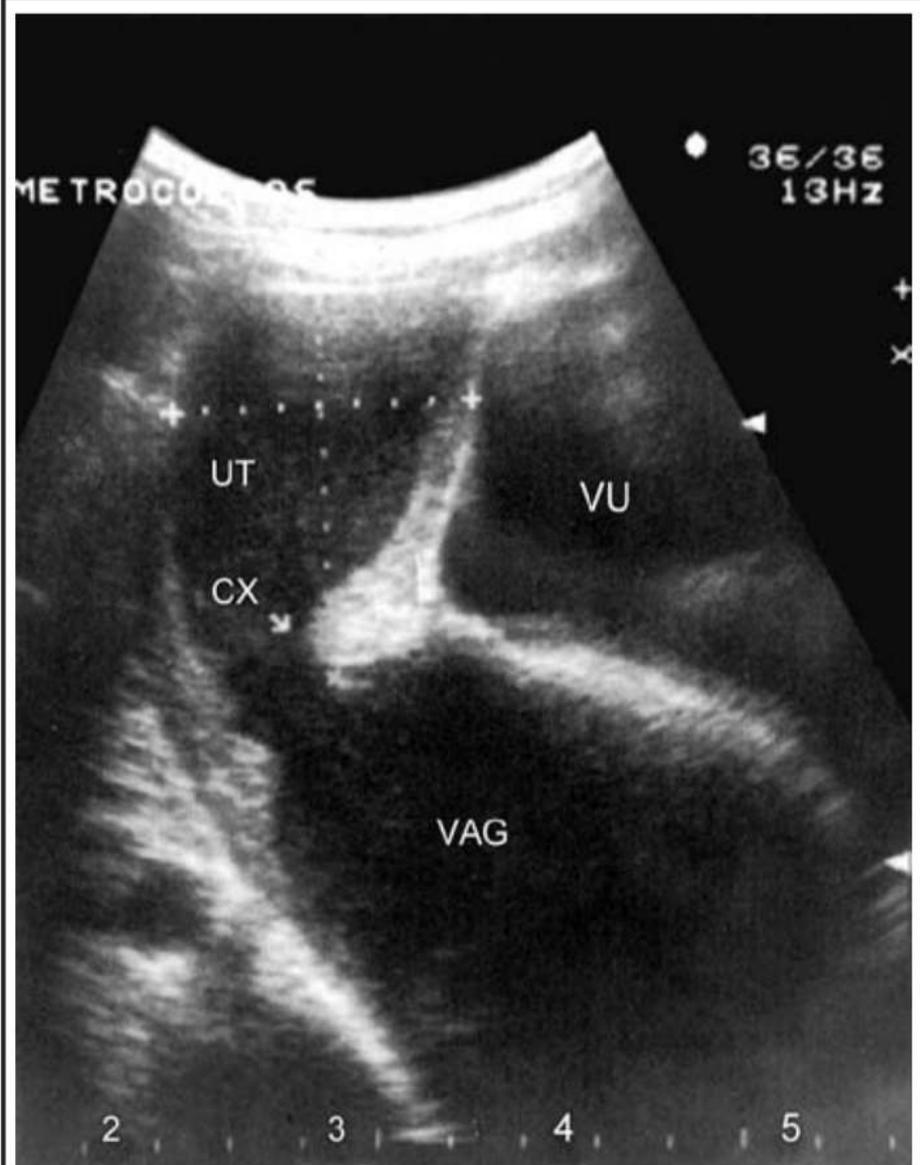
- Abdominal examination
  - Suprapubic swelling
- Vulval inspection:
  - Tense bulging membrane of bluish colouration
- Rectal examination:
  - Bulged vagina



**Fig. 4.3:** Tense bulging of the hymen  
in hematocolpos



**Fig. 4.1:** Hematocolpos and hematometra due to imperforate hymen. Note the elongation of the urethra due to distension of the vagina by blood



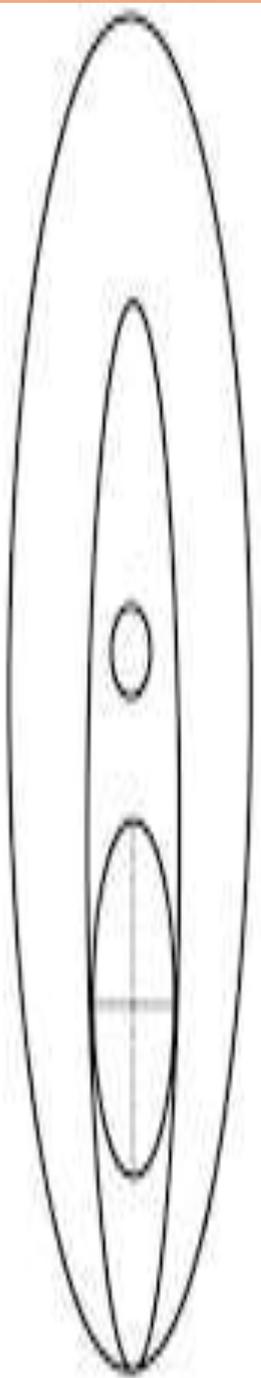
**Fig. 4.2:** Ultrasonographic view of hematometra and hematocolpos in a girl with imperforate hymen

## Treatment

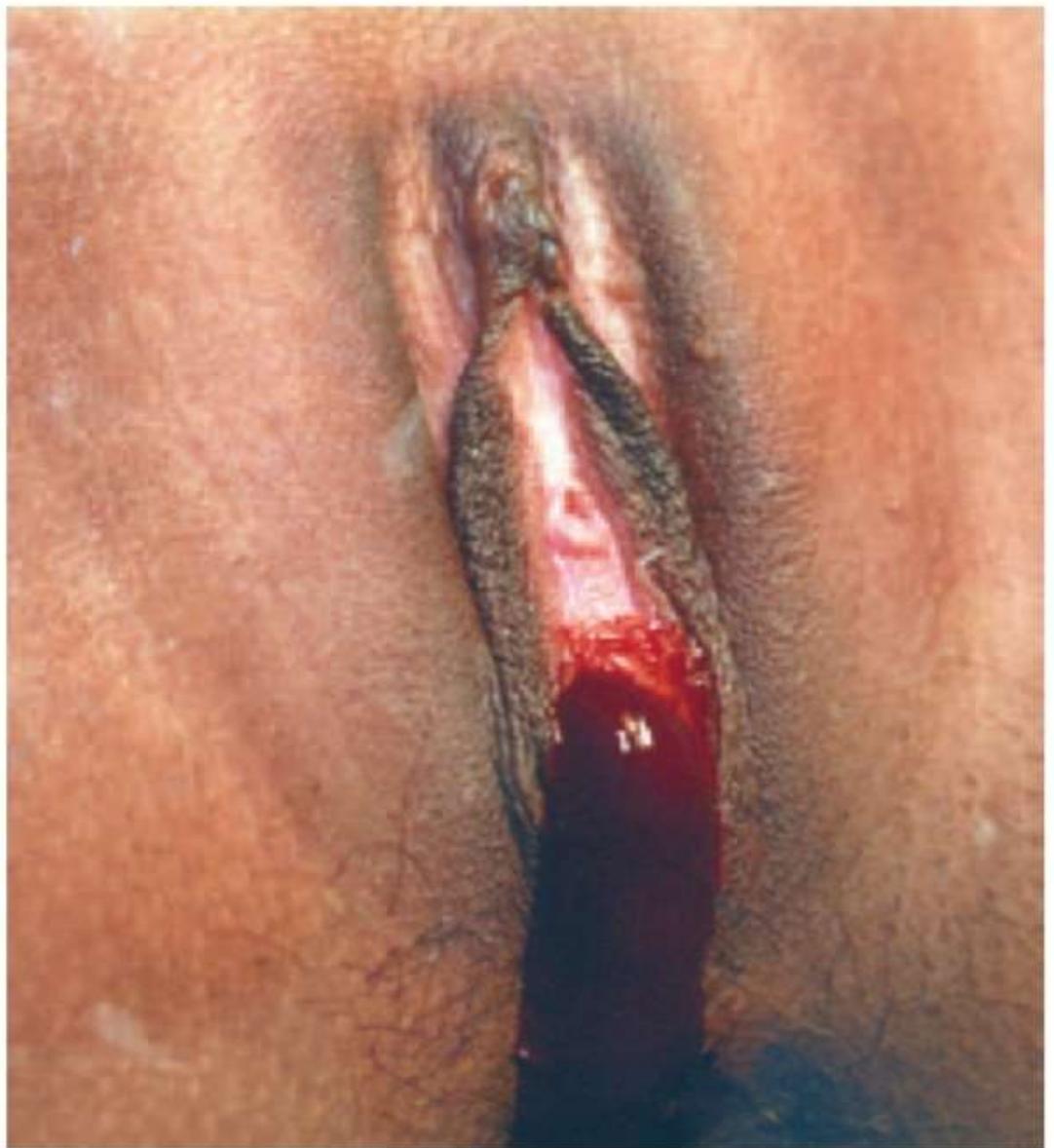
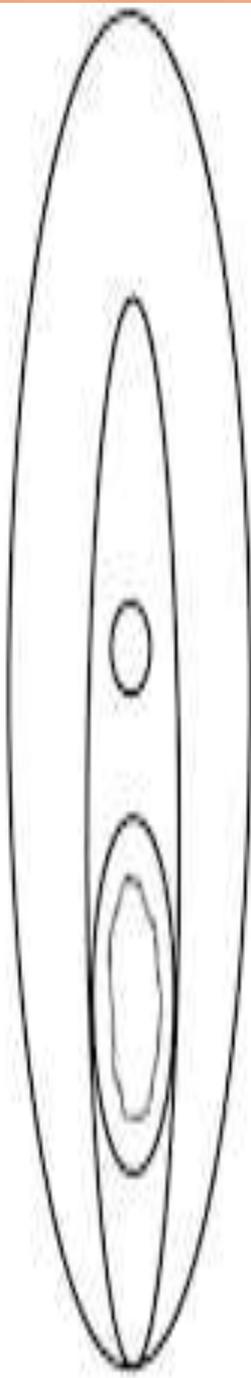
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- Cruciate incision is made in the hymen.
- Quadrants of the hymen are partially excised not too close to the vaginal mucosa
- Spontaneous escape of dark tarry coloured blood is allowed.
- Patient should be made to lie down with the head end raised.

(a)



(b)



**Fig. 4.4:** Spontaneous escape of dark tarry blood following incision

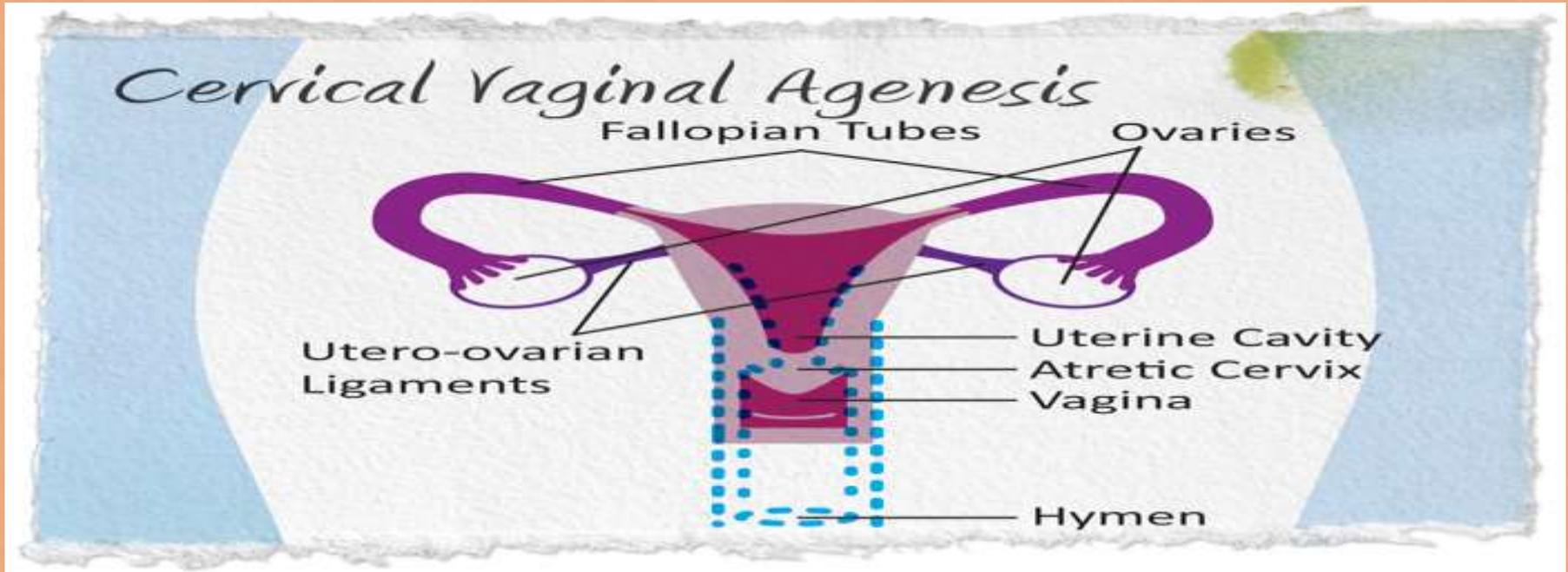
The uterus is next involved and the cavity is dilated (hematometra)

In the late and neglected cases, the tubes may also be distended after the fimbrial ends are closed by adhesions (Hematosalpinx)

The uterus is functioning normally, the menstrual blood is pent up inside the vagina behind the hymen (cryptomenorrhea)

Depending upon the amount of blood so accumulated, it first distends the vagina (hematocolpos)

# VAGINAL MALDEVELOPMENTS



## Common variations of vaginal maldevelopments

- ◆Agenesis of vagina
- ◆Failure of vertical fusion
- ◆Failure of lateral fusion

# VAGINAL MALDEVELOPMENTS

FIGURE 236.

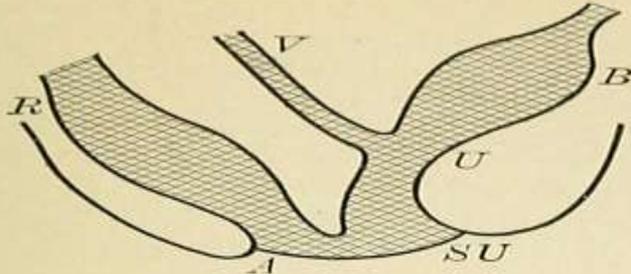


FIGURE 237.

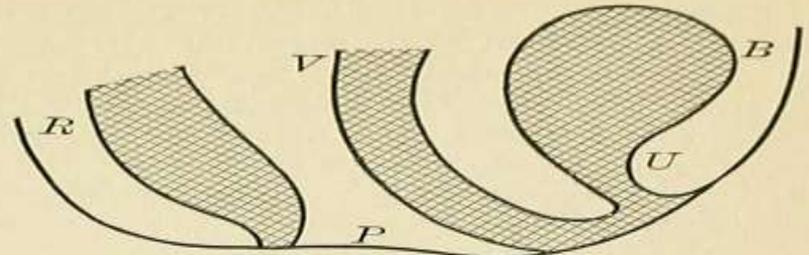
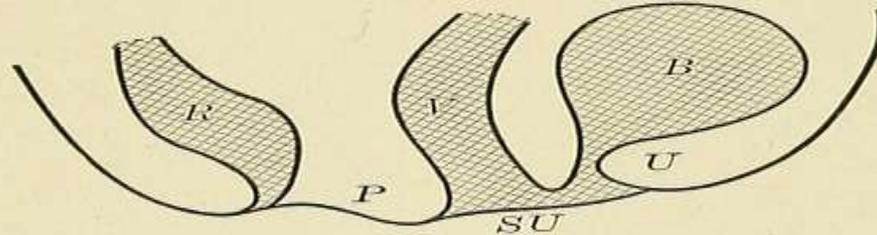


FIGURE 236.—The cloaca is becoming divided into the urogenital sinus, *SU*, and anus, *A*, by the downward growth of the perineal septum.  
 FIGURE 237.—The perineum is completely formed. *P*, perineum. The ducts of Mueller have united the lower portion forming the vagina.

FIGURE 238.



The upper part of the urogenital sinus has contracted into the urethra; the lower portion, *SU*, now becomes the vulva. *P*, perineum. *R*, rectum. *V*, vagina. *B*, bladder. *U*, urethra.

FIGURE 239.

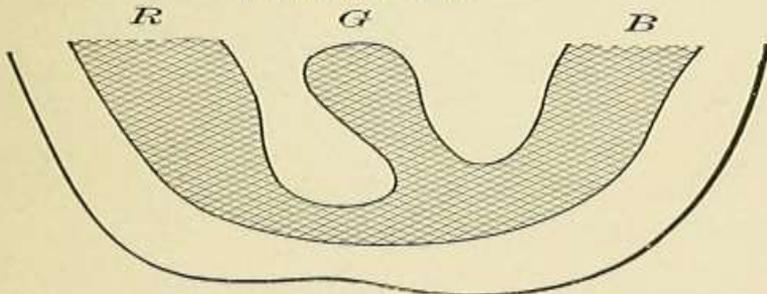


FIGURE 240.

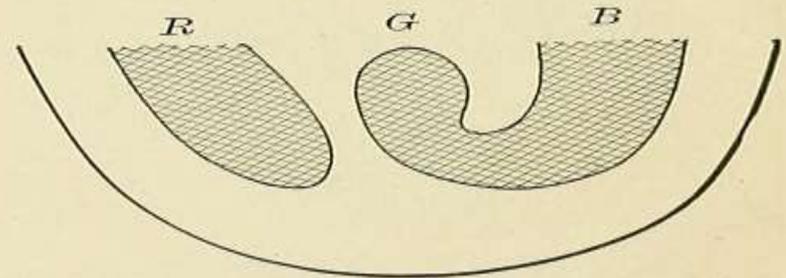
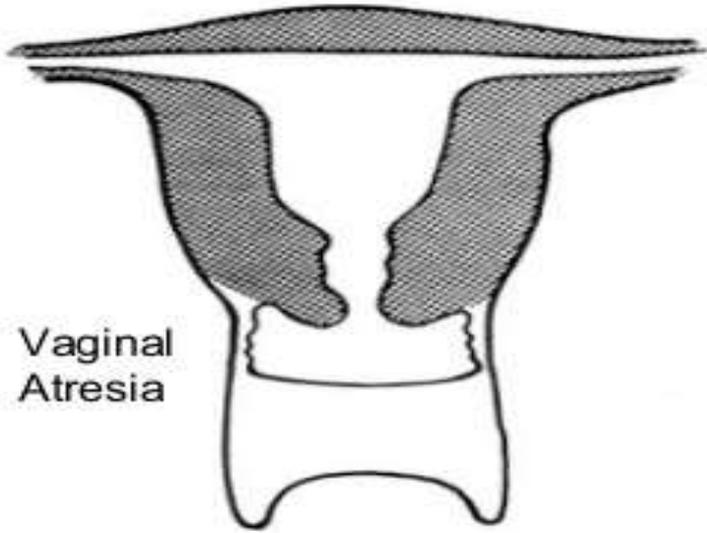


FIGURE 239.—Absence of cloacal division. Perineal septum wanting. *R*, rectum. *G*, genital canal. *B*, bladder.  
 FIGURE 240.—Absence of cloacal division. Perineal septum present.

# VAGINAL ABNORMALITIES



Vaginal Atresia



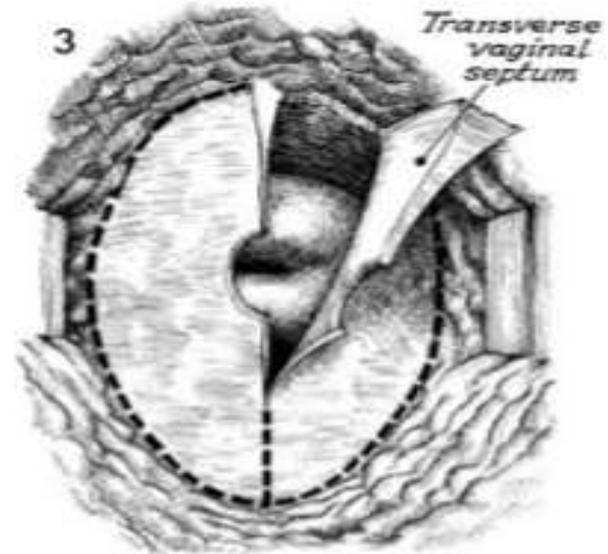
Fig. I. Ovaries adhaerent exteriorly.



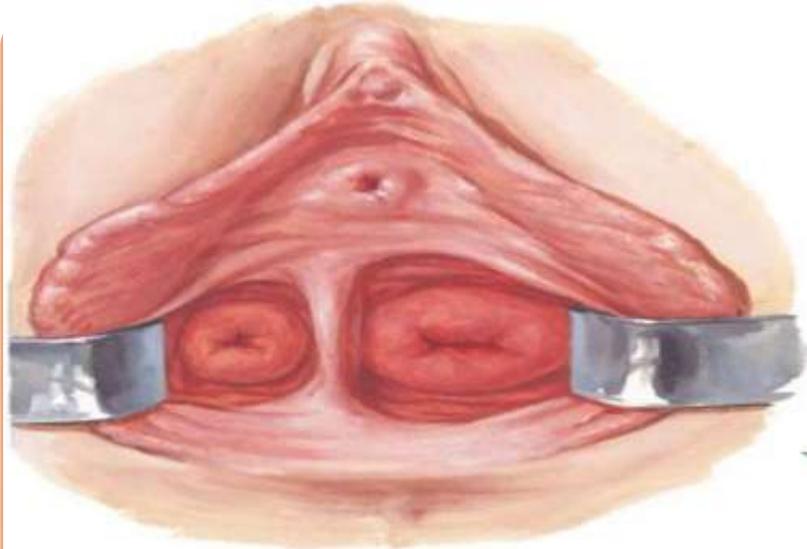
Fig. II. Coupe transversale et verticale du bassin. On trouve de haut en bas : 1° la vaine; 2° les deux vagins accolés comme des masses de foin; 3° le rectum.



Longitudinal vaginal septum

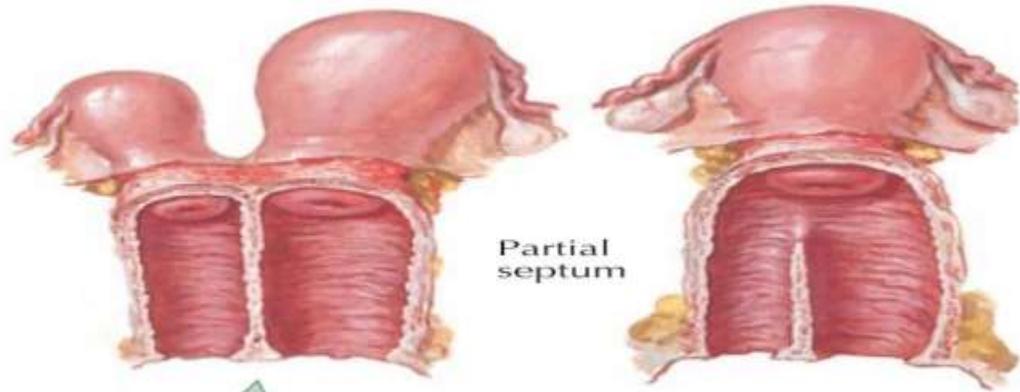






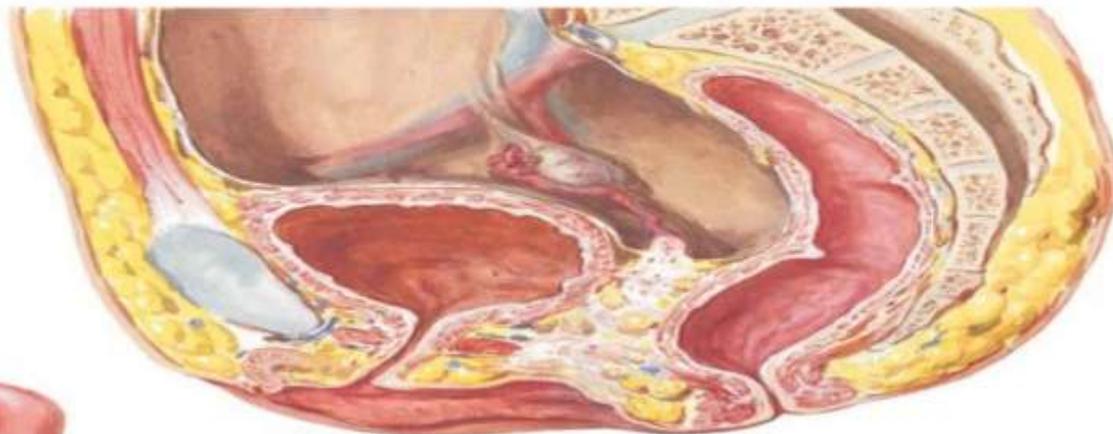
Double vagina

Septum is usually less than one centimeter in thickness and may be complete or incomplete. Location is variable

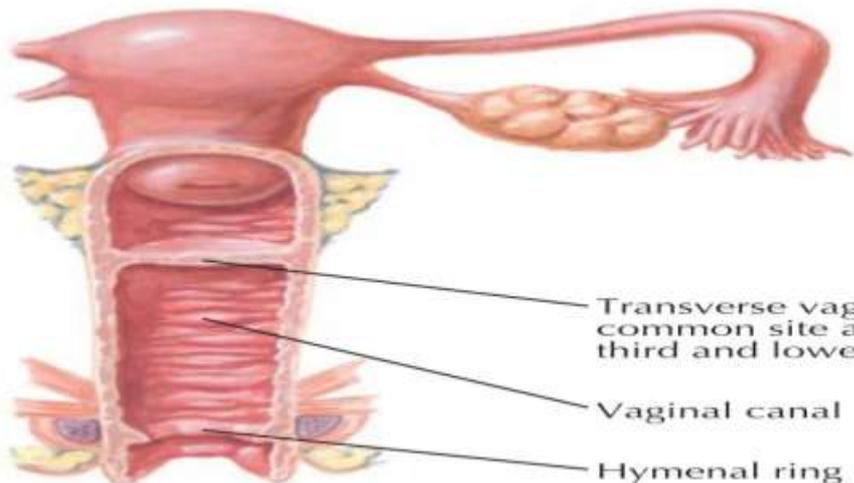


Partial septum

Complete septum with double uterus



Absence of vagina and uterus



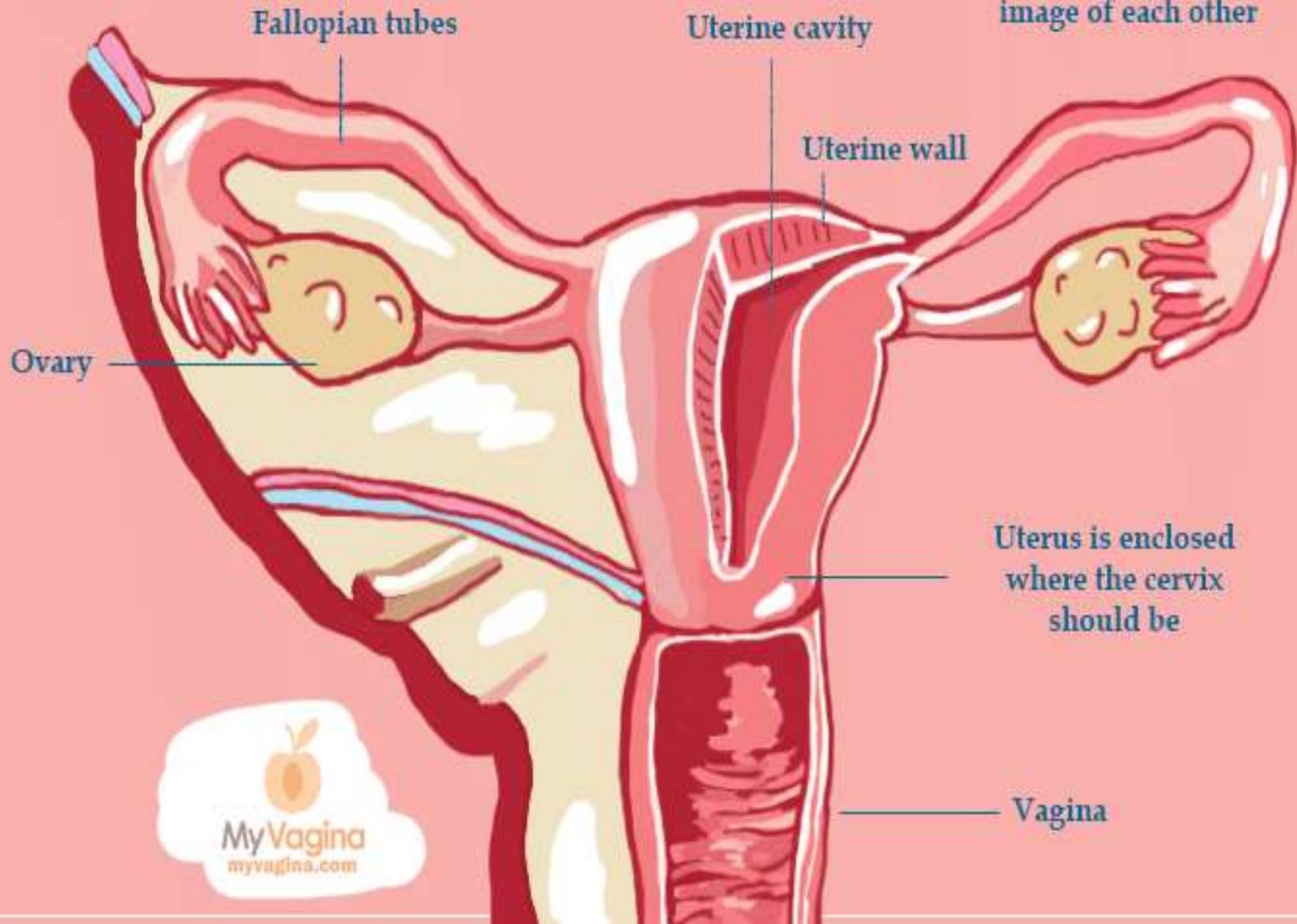
Transverse vaginal septum—most common site at junction of upper third and lower two-thirds of vaginal canal

Vaginal canal

Hymenal ring

# Cervical Agenesis (Missing Cervix)

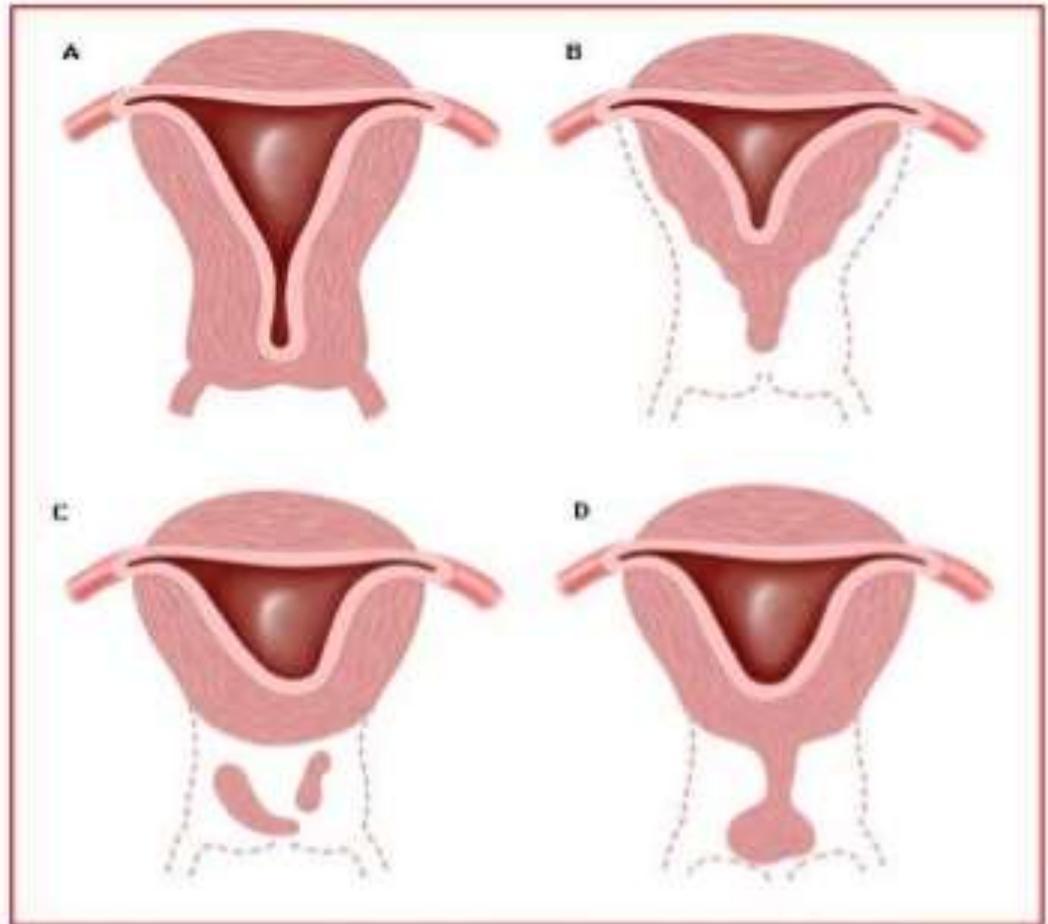
\*Both sides are a mirror image of each other



# Cervical agenesis and hypoplasia



**Cervical agenesis** The fundus of the uterus is present without the body of the cervix. Dotted lines represent missing cervix and upper vagina.



**Congenital cervical dysgenesis** The cervical body is intact (A) with obstruction of the cervical os. The cervical body (B) consists of a fibrous band of variable diameter that may contain endocervical glands. Fragmented portions of the cervix are noted (C) with no connection to the uterine body. Hypoplasia of the uterine cavity may be associated with cervical cords or fragmentation. The midportion of the cervix (D) is hypoplastic with a bulbous tip. No cervical lumen is identified.

## ABNORMALITIES OF THE FALLOPIAN TUBES

Unduly elongated

May have accessory ostia or diverticula

Rarely, the tube may be absent on one side.

These conditions may lower the fertility or favor ectopic pregnancy.

**Class U0/normal uterus**



**Class U1/Dysmorphic uterus**



a. T-shaped



b. Infantilis

c. Others

**Class U2/septate uterus**



a. Partial



b. Complete

**Class U3/Bicorporeal uterus**



a. Partial



b. Complete



c. Bicorporeal septate

**Class U4/Hemi uterus**



a. With rudimentary cavity



b. Without rudimentary cavity

**Class U5/Aplastic uterus**



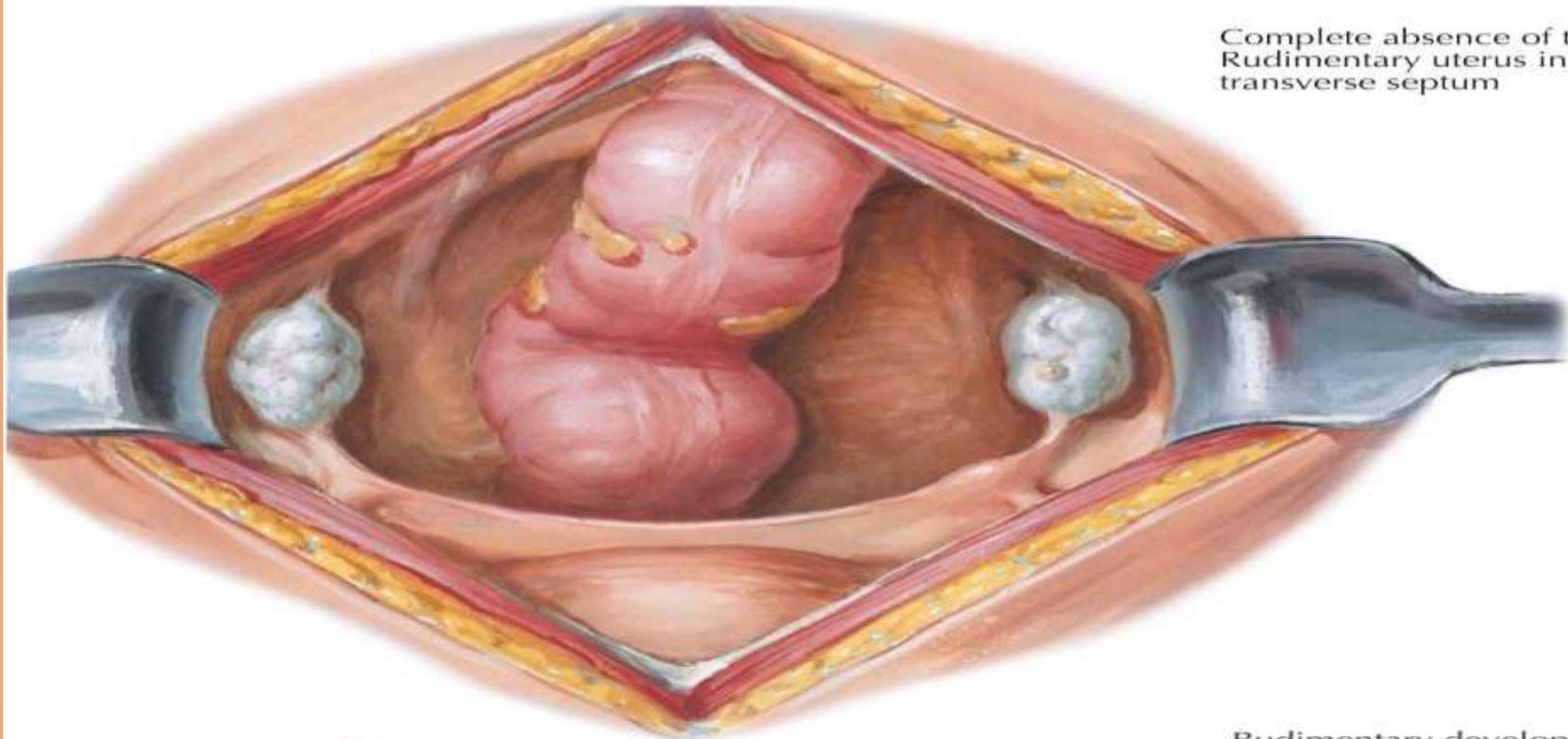
a. With rudimentary cavity



b. Without rudimentary cavity

**Class U6/Unclassified Cases**

# ABNORMALITIES OF THE FALLOPIAN TUBES

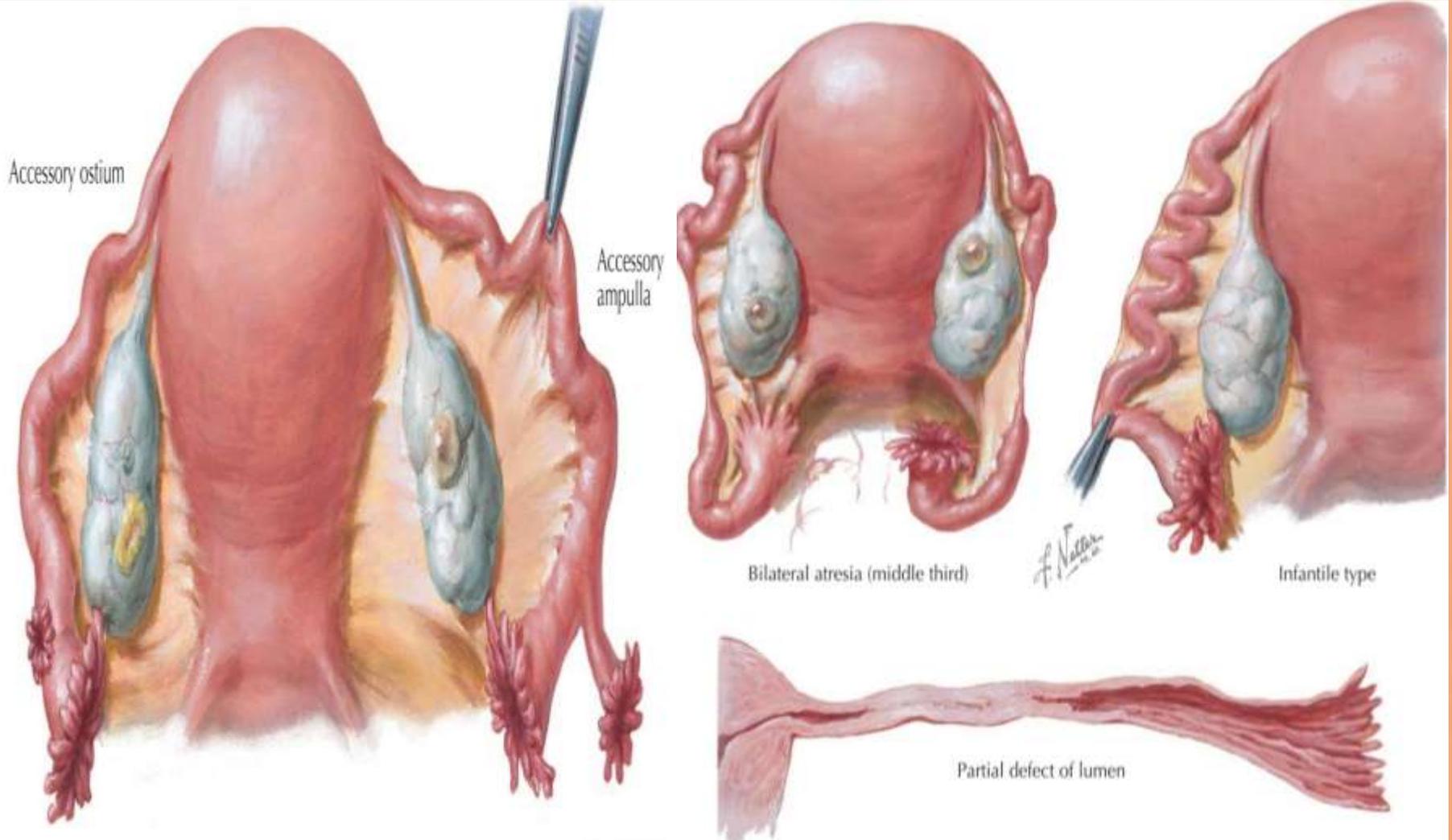


Complete absence of tubes.  
Rudimentary uterus in  
transverse septum



Rudimentary development  
of one tube

# ABNORMALITIES OF THE FALLOPIAN TUBES



## **Diagnostic Evaluation:**

- ✓ Medical history or family History
- ✓ physical exam and developmental and neurological exam.
- ✓ CT scan or MRI
- ✓ Laboratory testing for Genetic or Chromosomal Disorders.

# Diagnostic Evaluation

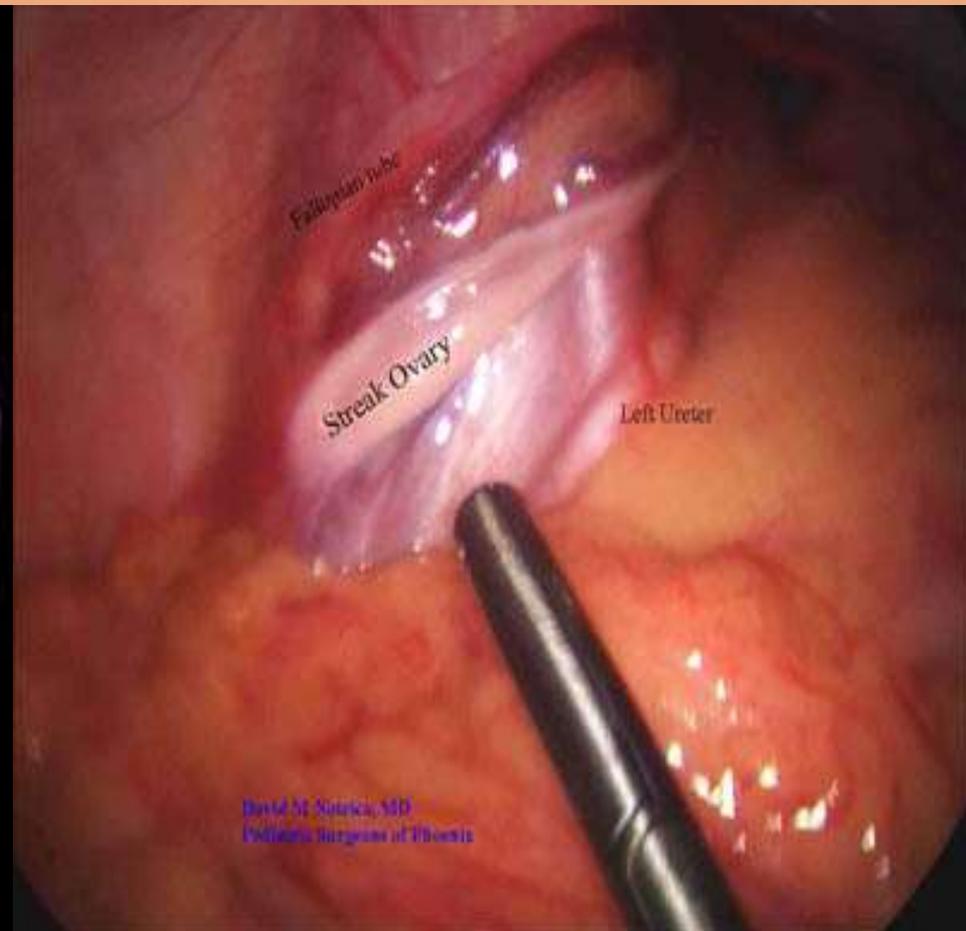
- pelvic **ultrasound**, **CT**, **MRI**, sonohistogram, **hysterosalpingogram**, **hysteroscopy**, and **laparoscopy**
- Keep in mind that **uterine septa** and **bicornuate** uteri may appear identical on hysteroscopic evaluation
- Because there is an increased incidence of **renal anomalies** (unilateral renal agenesis, pelvic or horseshoe kidneys, or irregularities in the collecting system), additional radiologic evaluation should be pursued in the setting of a congenital Müllerian anomaly

# ANOMALIES OF THE OVARIES

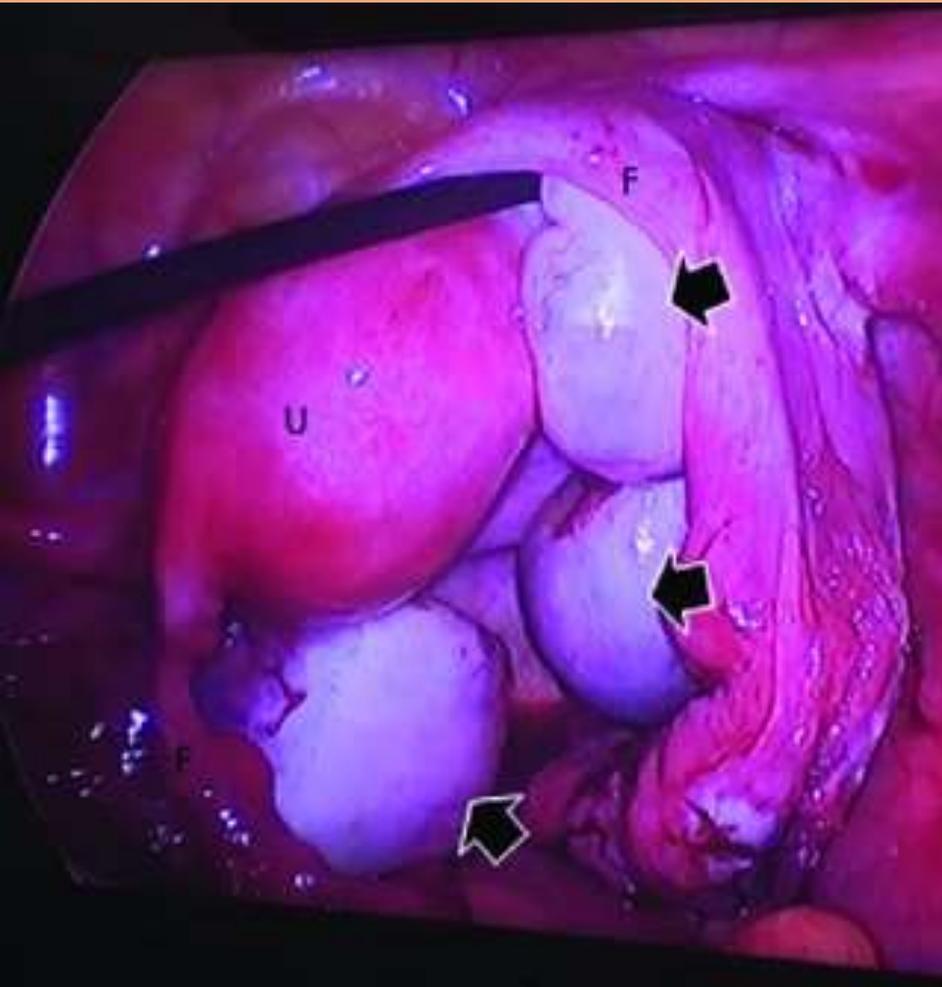
- **Streak Gonads**
- **Accessory ovary**
- **Supernumerary ovaries**

# ANOMALIES OF THE OVARIES

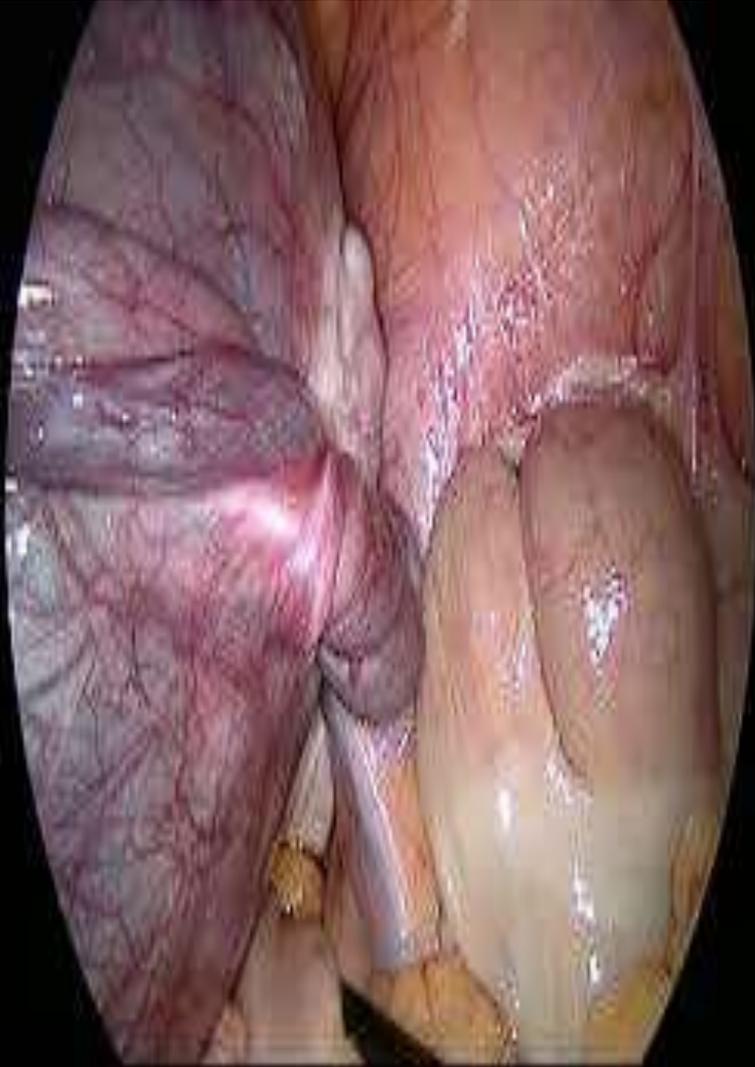
## STREAK GONADS OR GONADAL DYSGENESIS



# ACCESSORY OVARY



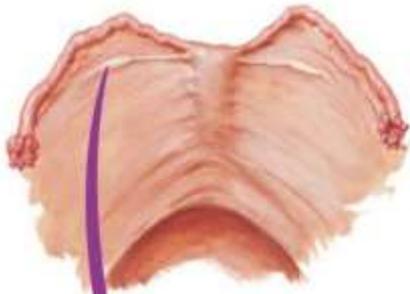
# SUPERNUMERARY OVARIES



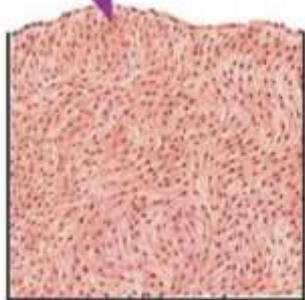
# Congenital anomalies of ovaries and fallopian tubes

- **Duplication**
- **Absence**
- **Ectopic ovarian tissue** : Accessory or supernumerary ovaries are extremely rare, and may also be associated with other congenital genitourinary abnormalities
- **Sexual bipotentiality** “Ovotestis “**true hermaphrodite**”  
In which combined ovarian and testicular tissues seen “
- **Genetic chromosomal disorders** :
- 1. Turner syndrome (45 XO) “**streaked ovaries** that are a hallmark of the disorder ”

Ovarian agenesis



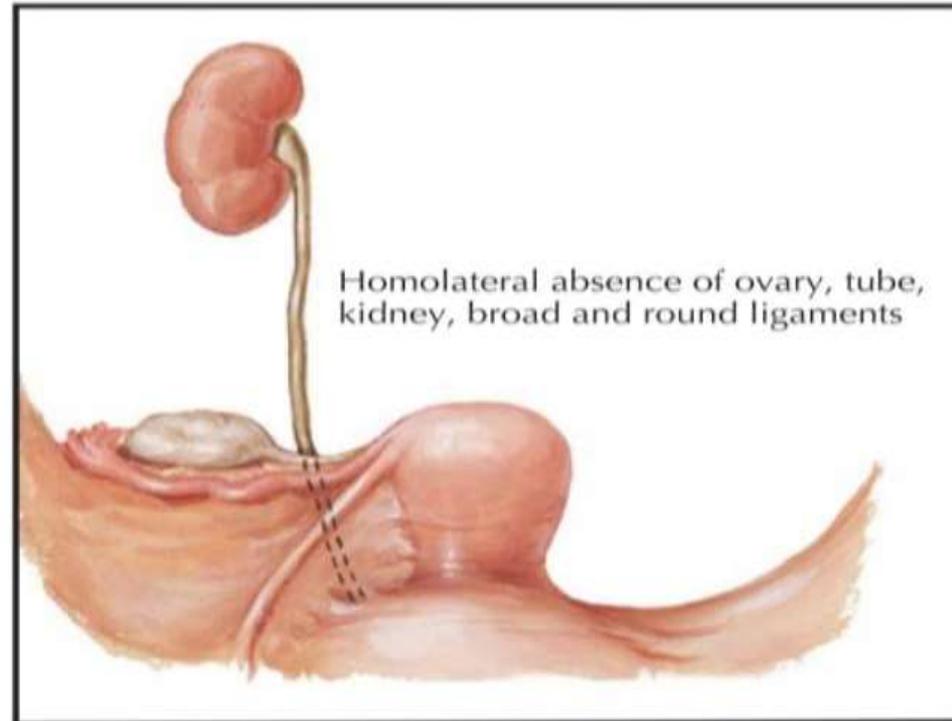
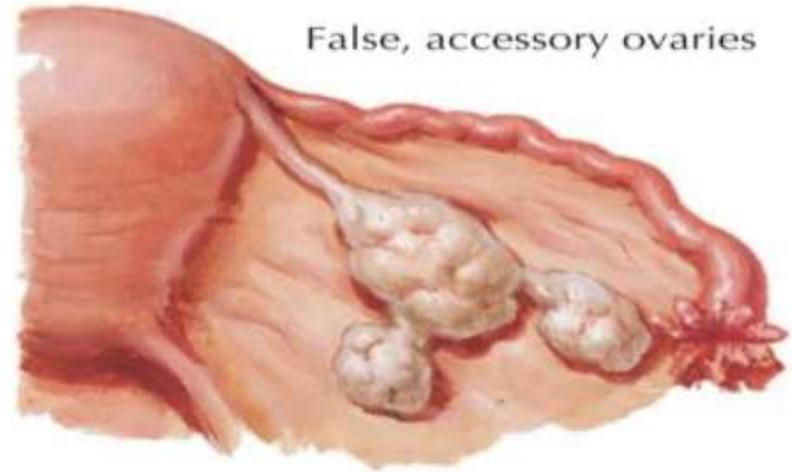
Rudimentary ovaries or primitive genital streaks



Microscopic section: complete absence of follicular elements

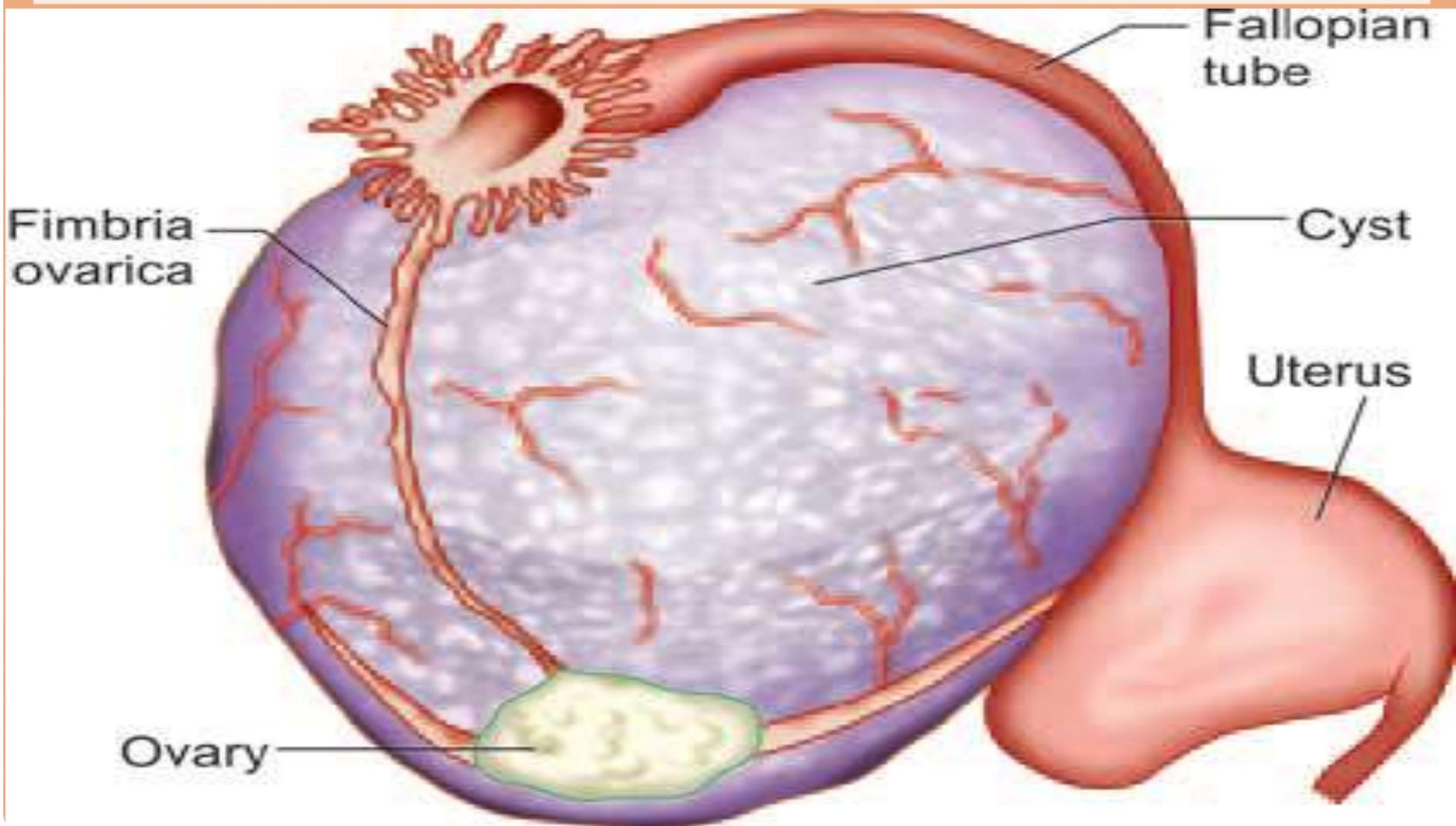
Short stature, absence of secondary sex characteristics, infantile genitalia, sparse pubic hair, high gonadotropin level, estrogen deficiency, and multiple congenital abnormalities (web neck, shieldlike chest, cubitus valgus)

False, accessory ovaries



Homolateral absence of ovary, tube, kidney, broad and round ligaments

# PAROVARIAN CYST



# WOLFFIAN REMNANT ABNORMALITIES

The tubules of the Gartner's duct may be cystic, the outer ones are Kobelt's tubules, the middle set, the epoophoron and the proximal set, the paroophoron. Small cyst may arise from any of the tubules.

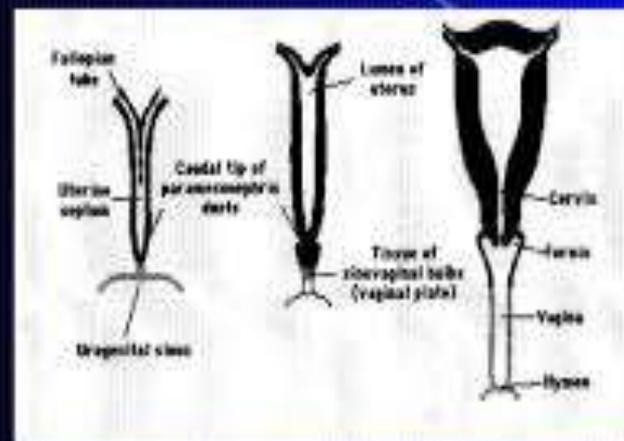
A cystic swelling from the Gartner duct may appear in the anterolateral wall of the vagina, which may be confused with cystocele.

The outer end of the Wolffian (Gartner) duct may be cystic, size of pea, often pedunculated (hydatid of Morgagni) and attached near the outer end of the tube.

## Vertical Fusion Defects: obstructive and non-obstructive

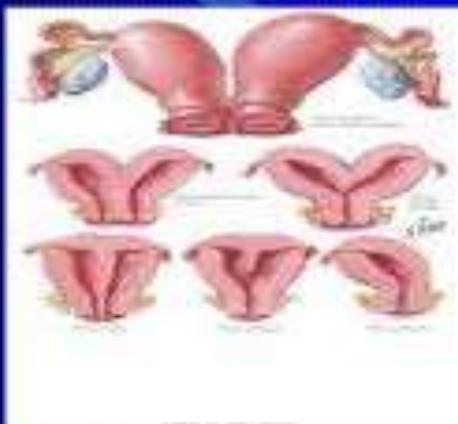
- Incomplete cavitation of the vaginal plate formed by the down-growing müllerian ducts and the up-growing urogenital sinus. Can be considered in two categories:
- 1. Imperforate Hymen
- 2. Transverse Vaginal Septum

## Vertical Fusion Defects

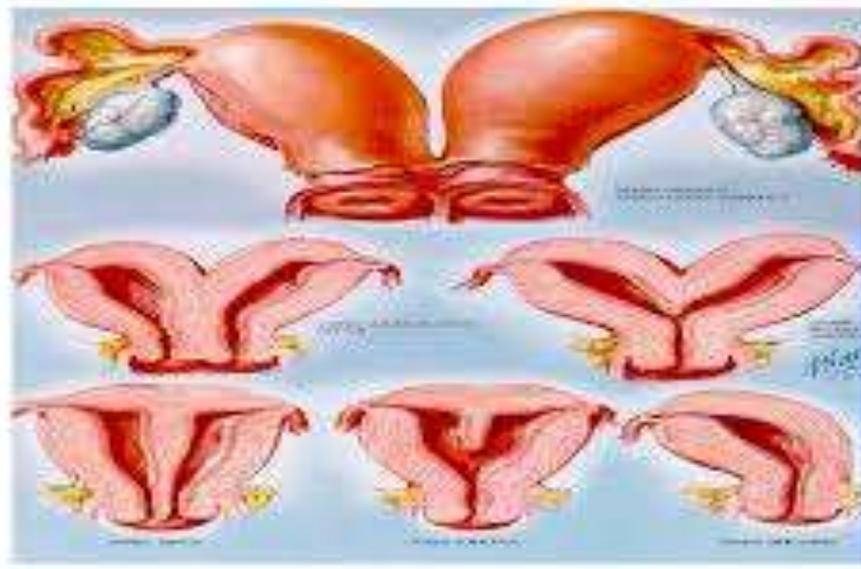


## Lateral Fusion Defects

- Result from failure of fusion of the müllerian ducts
- , or failure of absorption of the septum.

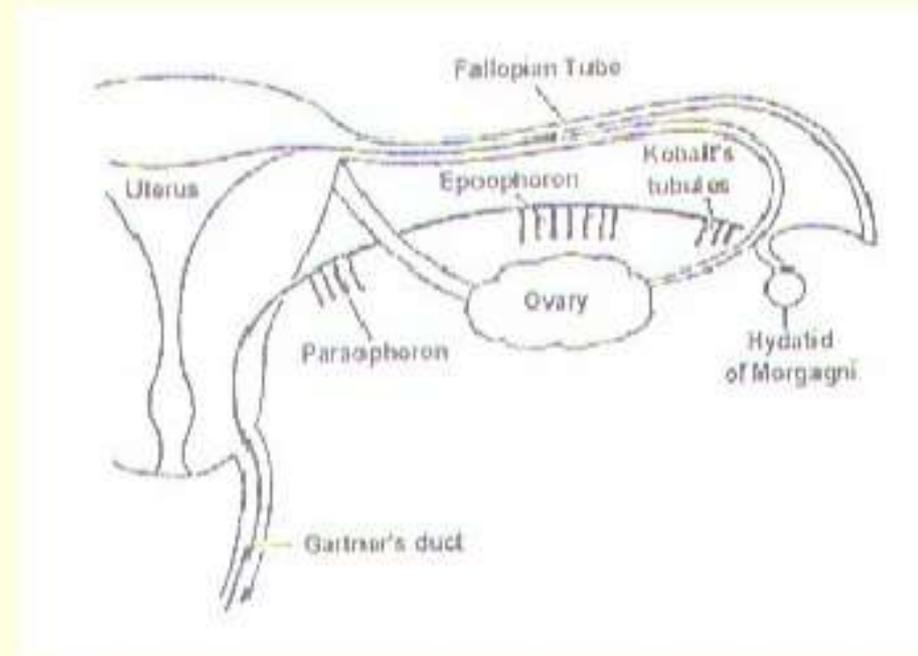


## Lateral Fusion Defects



# The Wolffian remnants in the female

- in female, the wolffian ducts undergo atrophy leaving remnants between two layers of B.L. and in the anterolateral wall of the vagina
- **These remnants are:**
  - Hydated cyst of morgagni: lateral to the tube
  - Koblet's tubules: in the outer part of B.L.
  - Epoophoron: between the ovary and F.T
  - Paroophoron: between the ovary and uterus.
  - Gartner's duct: may undergo cystic dilatation in the B.L. gives rise to paraovarian cyst or in the anterolateral wall of the vagina, gives rise to Gartner's cyst



It arises from the vestigial remnants of Wolffian tissue situated in the mesosalpinx between the tube and the ovary.

**The wall consists of connective tissue lined by single layer of low columnar epithelium**

**The cyst is unilocular; the wall is thin and contains clear translucent fluid**

**The ovarian fimbria with the ovary is stretched over the cyst**

**Increases in size**