MAYER ROKITANSKY KUSTER HAUSER SYNDROME

Background:
- Mayer (1829): first to report vaginal agenesis in stillborn child
- Rokitansky (1838): 19 adult autopsy cases of uterovaginal agenesis
- First to relate vaginal agenesis and kidney abnormalities
- Kuster (1910): recognized urologic associations
- Hauser (1961): skeletal anomalies

Embryology:
- (mullerian aplasia) failure of the sinovaginal bulbs to develop and form the vaginal plate.
- Uterovaginal primordium
- Uterovaginal canal develops at week 5, along with other mesodermally derived organ systems.

Incidence: 1 / 5000 live female births

Definition: congenital absence of the proximal portion of the vagina in an otherwise phenotypically, chromosomally, and hormonally intact female (46XX, normal secondary sex characteristics, normal LH, FSH levels)

Usual presentation: primary amenorrhea (mean age 13 -18)

Differential diagnosis: gonadal dysgenesis (r/o with normal LH and FSH, Estrogen and Thyroid levels)

PE / Dx: absence of the vagina, normal breast and pubic hair. Normal female serum estradiol and testosterone.

Workup: Radiographic (sonogram) to delineate remnant mullerian structures and to evaluate renal and skeletal systems. Other modalities CT , MRI, cystoscopy, IVP, vaginoscopy

Classification:

typical: (type A) symmetrical uterine remnants, normal fallopian tubes, lack associated findings
Variable absence of cervix, uterus, fallopian tubes
25% lack uterus, 55% solid rudimentary uterus, 30% other uterine abnormalities
32% normal fallopian tubes, 50% rudimentary, 10% complete absence
ovaries usually present

Atypical: (type B) asymmetrical uterine buds, abnormal fallopian tubes
usually associated w/ **other abnormalities**
renal (unilateral renal agenesis or ectopia (74%)
secondary to relationship of mesonephros and paramesonephros
skeletal (10-20%) (congenital fusion of cervical vertebrae (Klippel-Feil)
Duncan (1977) MURCS association
Mullerian duct aplasia, renal aplasia, cervicothoracic somite dysplasia

Treatment strategies

1) counseling
    a. reassurance (fertility is potentially possible with assisted reproductive technologies and a gestational carrier)
    b. parents
        may seek early surg therapy
        best to wait until late teens mainly for psych reasons, but also less failure rates
2) nonoperative
    a. progressive invagination of vaginal dimple
        i. Frank (1938) – gradual glass dilators over 6 months – 2 years
            1. up to 90% success rates, avoids operation
            2. some females uncomfortable with self dilating
        ii. Ingram (1981) – dilator mounted on a bicycle seat on a stool
3) Operative
    a. Abbe-McIndoe vaginoplasty (1898)
        i. potential neovaginal space dissected between rectum and urethra
        ii. split thickness skin graft inserted mounted on mold
            1. requires 3 months of post op dilation to decrease stenosis
            2. risk of stenosis
    b. William vulvovaginoplasty
        i. full thickness skin graft from labia
            1. can create an awkward angle for intercourse
            2. does have reports of up to 90% success rate
    c. Vecchetti procedure (1979)
        i. Olive shaped marble placed in vaginal dimple
        ii. Two attached sutures surgically guided proximally thru neovaginal space out through anterior abdominal wall
        iii. Sutures on tension, marble pulled to create neovagina
        iv. Not FDA approved in USA
    d. Jejunal or ileal conduit
        i. High stenosis rate, not advocated anymore
    e. Colon (sigmoid or cecal) conduit (first described by Baldwin in 1907)
        i. No dilators needed
        ii. Well lubricated
        iii. Chronic vaginal discharge (need to wear pad and may need routine douching)
        iv. May have increased risk of HIV and hepatitis (less barrier than vaginal sc)

Ileal conduit

10-15cm usually taken 10-15cm proximal to ileo-cecal valve

contraindications: IBD, SBS, radiation enterititis – difficult for vaginal reconstruction for length reasons

Colon conduit

Transverse / sigmoid / ileocecal

Isolated on arterial pedicle

Mobilize attachments as needed (omentum, flexures, peritoneum)

Length of segment dictated by indication of operation

Can staple with GIA or close ends with Connell and Lembert 2 layer closure

Most common indication is for urinary diversion – not for vaginal agenesis

Conduit Complications (post urinary diversion)

	Ileal		Sigmoid		Transverse		Ileocecal	
Complication	early	late	early	late	early	late	early	late
Urine leak	2%	---	1%	---	8%	8%	6%	---
Wound infection	7%	2%	1%	---	5%	---	2%	---
Dehiscence	3%	---	1%	---	7%	---	7%	---
Acute pyelonephritis	3%	18%	---	7%	---	11%	---	14%
Prolonged ileus	6%	---	---	---	---	---	---	---
Bowel obstruction	3%	5%	---	6%	3%	2%	3%	10%
Ureteral stricture	2%	6%	---	9%	6%	17%	---	5%
Stones	---	7%	---	4%	---	11%	---	5%
Parastomal hernia	---	2%	---	3%	---	4%	---	5%
Stomal stenosis	---	30%	---	3%	---	2%	---	2%
Stomal prolapse	---		---		---	11%	---	16%
Fecal fistula	---	<1%	---	0.5%	---	2%	---	2%
Bowel leak	2%		---	---	---	---	3%	---
Metabolic acidosis	---	13%	---	---	---	---	---	12%
Conduit infarction		2%						
Volvulus		7%						
Excessive length		9%						

References:

Laufer MR. Congenital absence of the vagina: in search of the perfect solution. Curr Opin Obstet Gynecol 14:441-444.

Walsh: Campbell's Urology. 8th ed (2002) 2432-2436 and 3771-3775.

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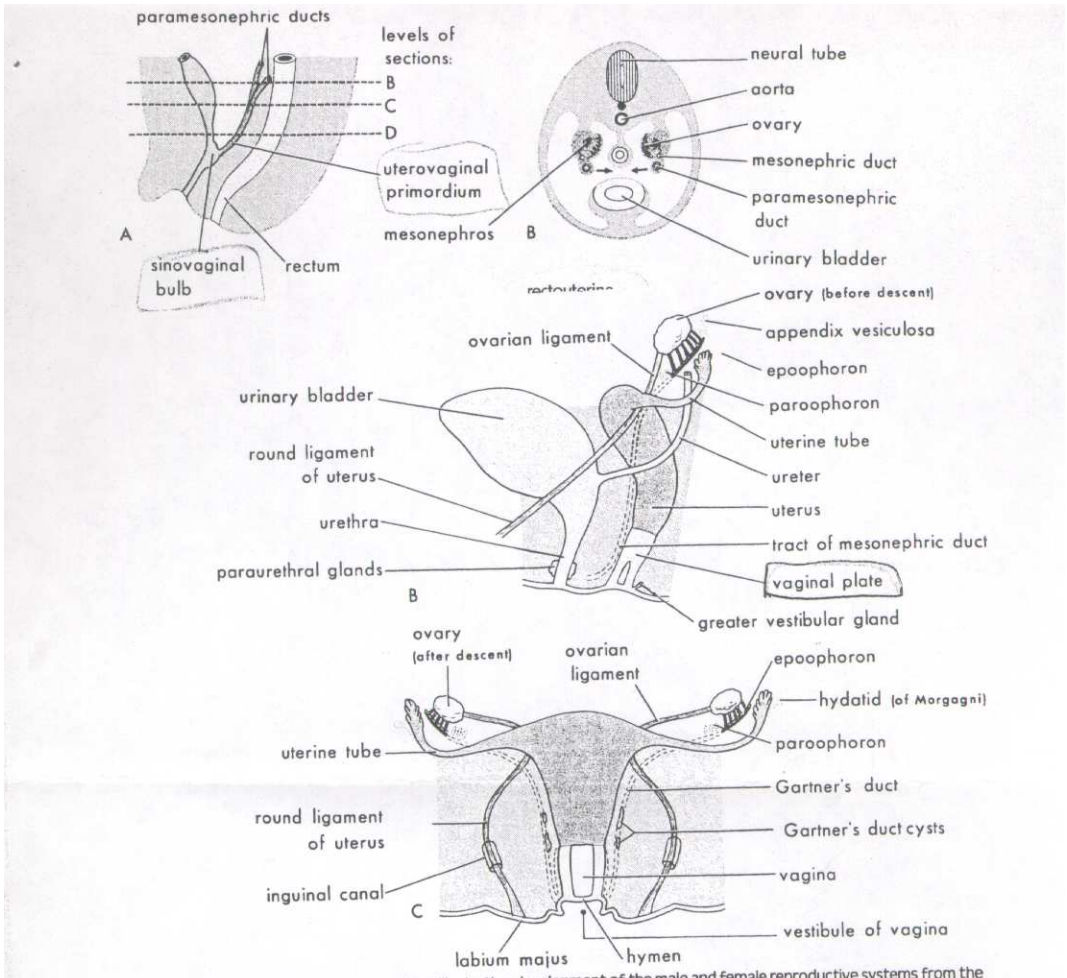

Figure 13–23. Schematic drawings illustrating development of the male and female reproductive systems from the genital ducts and the urogenital sinus. Vestigial structures are also shown. *A*, Reproductive system in a newborn male. *B*, Female reproductive system in a 12-week fetus. *C*, Reproductive system in a newborn female.

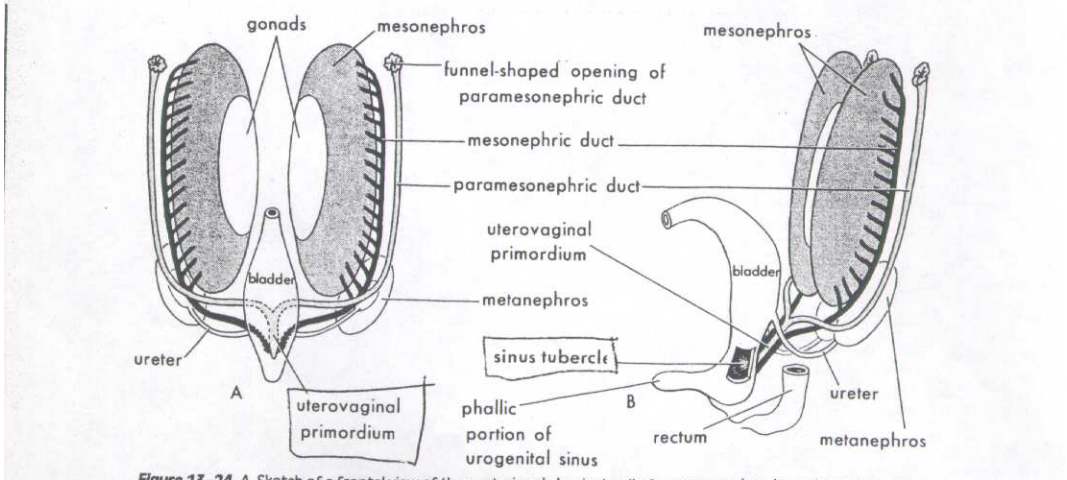

Figure 13–24. *A*, Sketch of a frontal view of the posterior abdominal wall of a seven-week embryo showing the two pairs of genital ducts present during the indifferent stage. *B*, Lateral view of a nine-week fetus showing the sinus tubercle (müllerian tubercle) on the posterior wall of the urogenital sinus. It becomes the hymen in females and the