Anorectal malformation:
Rare Variants

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Outline

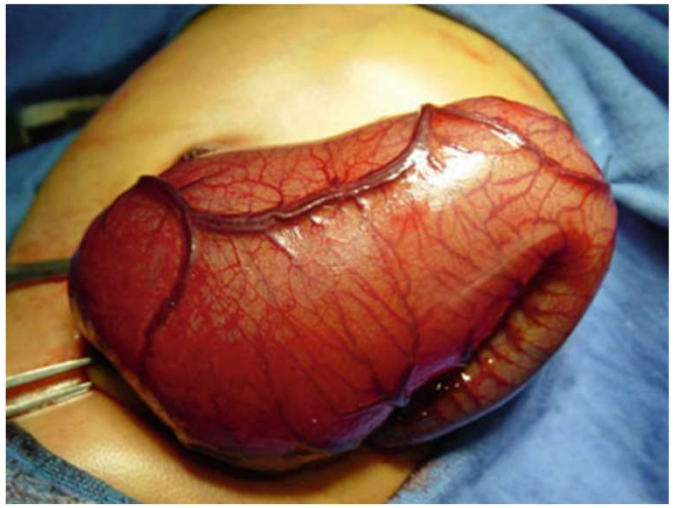
1. Congenital pouch colon
2. Rectal atresia
3. H-type fistula
4. Vaginal fistula
5. Penile fistula
6. Perineal groove
7. Anterior Anus
8. Posterior cloaca

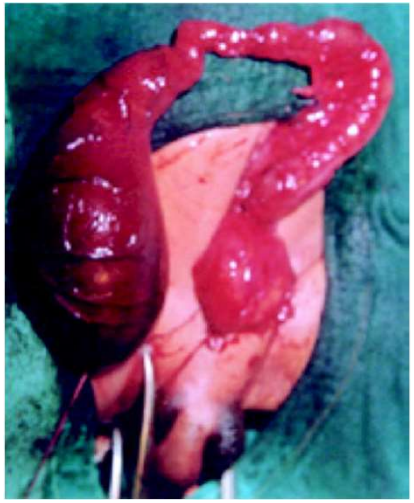
1 - Congenital pouch colon

Definition and spectrum

- Abnormal colon associated with ARM (usually high with large, long fistula)
  - Abrupt change (no transition with normal bowel)
  - Shortened
  - pouch-like dilatation
  - Abnormal blood supply
  - Abnormal wall
- Rare (more common in Northern India 5-18% of ARM)
- More common in males (4:1) and associated with colovesical fistula
- In females its ass. with cloaca, vaginal or vestibular fistula. Double vagina with a wide intervaginal bridge

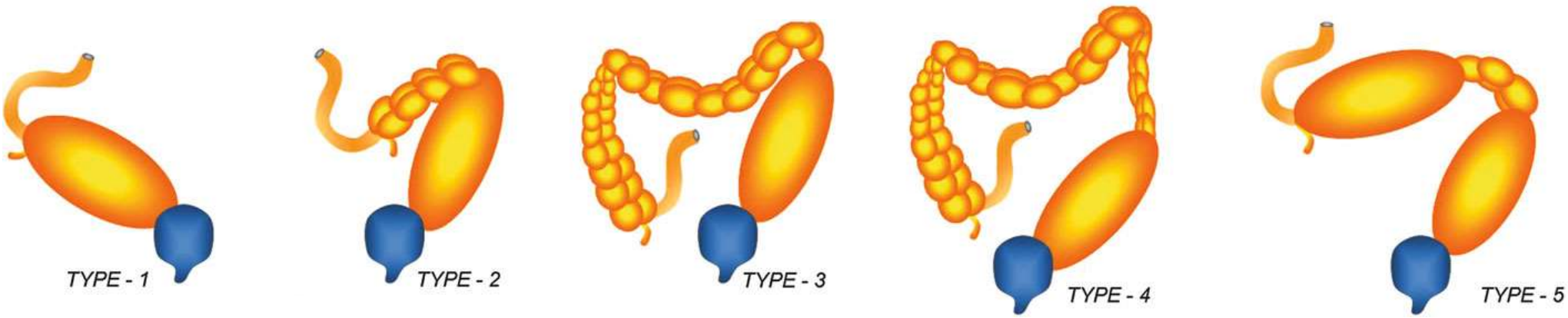
Definition and spectrum





- colonic pouch of variable **size** (5–15 cm in diameter). The **mesentery** of this pouch is short and poorly developed, the **wall** is very thick, the taenia coli are absent or ill defined, haustration and the appendices epiploicae are absent. The main pouch is supplied by the branches arising from the **superior mesenteric artery**, which form a leash of vessels around it. The inferior mesenteric artery is present in only 50% of cases of distal CPC, and it is also quite insignificant.

Definition and spectrum



- **Saxena-Mathur classification (2008)**: 1- Normal colon absent, 2- normal cecum, 3-normal transverse, 4-normal descending, 5-normal colon interposition (double pouch)

Associated anomalies

Genitourinary anomalies	Gastrointestinal anomalies	Other organ anomalies
Hydronephrosis	Absent appendix	Sacral agenesis
Vesicoureteral reflux	Double appendix	Congenital heart disease
Bicornuate uterus	Malrotation	Myelomeningocele
Cryptorchidism	Colon duplication	Prune belly syndrome
Hydroureteronephrosis	Meckel's diverticulum	Hemivertebrae
Hypospadias	Double Meckel's diverticulum	Congenital talipes equinovarus
Renal aplasia/agenesis	Esophageal atresia	Perineal teratoma
Renal dysplasia	Small intestinal duplication	Absent ribs
Double uterus	Rectal atresia	Down's syndrome
Double vagina		
Septate vagina		
Ectopic kidney		
Urethral duplication (males)		
Urethral diverticula		
Bifid penis		
Megalourethra		
Urethral strictures		
Bladder exstrophy		
Duplicate bladder exstrophy		

- major associated malformations are relatively uncommon. A large number of associated anomalies are found mostly genitourinary followed by gastrointestinal.

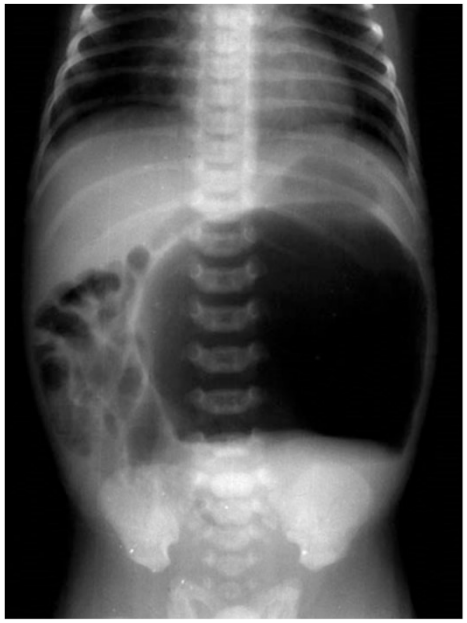
Etiology

- **Environmental factors** – high incidence in northern belt of india (also known as the stone belt due to the deficient nutritional factors in the diet and iodine deficiency in the water there)
- **Embryologic theories**
  - chronic obstruction theory (Trusler et al. 1959) - not accepted
  - Interference of hindgut growth and migration theory (Dickinson 1967)
  - altered hindgut stimulation theory (Chatterjee 1991).
  - faulty rotation and fixation theory (Wu 1990)
  - vascular insult theory (Chadha 1994)

Pathology

- **Disorganization of the muscle coat** (no inner longitudinal, outer circular + arranged in decussating pattern)
- responsible for the absence of normal peristaltic activity, requiring the removal of the dilated pouch and retaining only the normal bowel.

Management

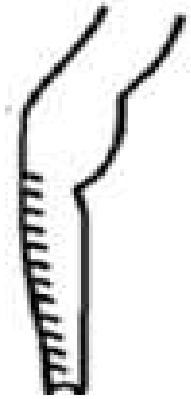


- **Plain x-ray**: classic grossly dilated loop of bowel occupying the left abdomen, and displacing the small intestines toward the right abdomen. Similar X-ray may be found in a perforated or significantly dilated sigmoid; and in females, with rectouterine fistula when severe dilatation of the meconium filled uterus

Management

- **Incomplete CPC** (adequate length)
  - fistula division + pouch excision + end colostomy >> abdomino-PSARP ± transverse colsotomy
  - transverse / Window colostomy >> fistula division + pouch excision + abdomino-PSARP ± stoma
- **Complete CPC** (inadequate for pullthrough)
  - fistula division + coloplasty + end colostomy >> abdomino-PSARP ± ileostomy
  - ileostomy / Window colostomy >> fistula division + coloplasty + abdomino-PSARP ± stoma
- **Type 5 CPC**
  - Excise distal pouch with coloplasty on proximal one
  - Coloplasty on both

Management



- **Coloplasty** procedure to lengthen colon while preserving vascular arcade. performed after mobilizing the pouch completely by division of the inferior mesenteric artery (if present) and incising the pouch on the antimes nteric border, thus preserving the vascularity. The tube is fashioned over a red rubber catheter to obtain a uniform diameter

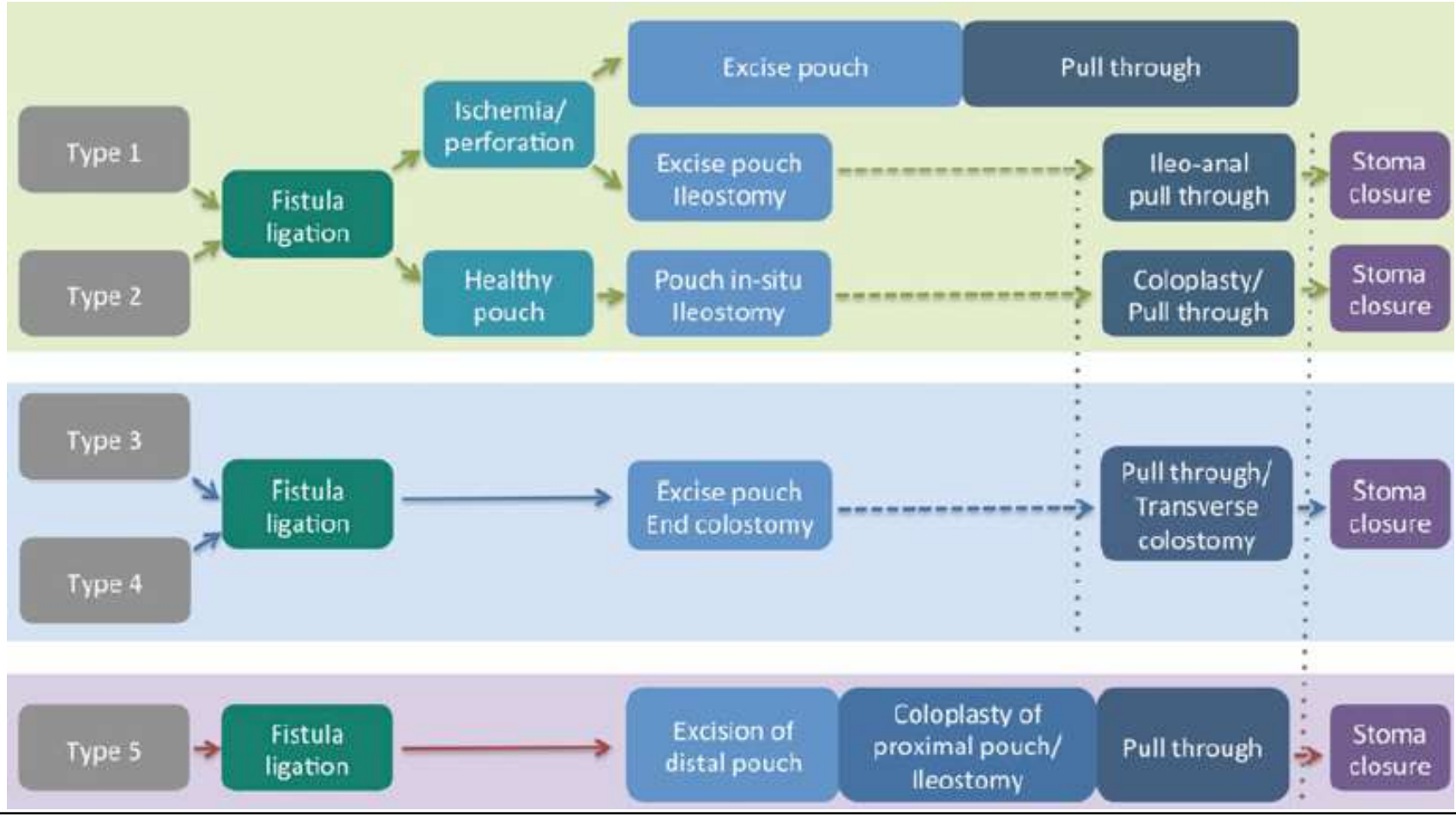
Management considerations

- ***Appendectomy** should be performed at the time of pull-through to prevent misdiagnosis
- ***Window colostomy:** (colostomy on pouch) not preferred but favored in the sick neonate
  - **Complications** include massive prolapse, retraction/stenosis, pouchitis/enterocolitis, ischemia, bleeding, FTT, UTI, VUR
- **Coloplasty**
  - **Complication** include leak & rupture (now reduced due to ileostomy), redilatation of the tube coloplasty

Outcome

- **Depends on** length of normal bowel (watery diarrhea, poor weight gain)
    - suffer from increased frequency the initial 3–6 months,
    - decreases with the growth of the child and dietary modifications.
- **Mortality** previously as high as 30–40%, but has now come down to 10–20%

Summary

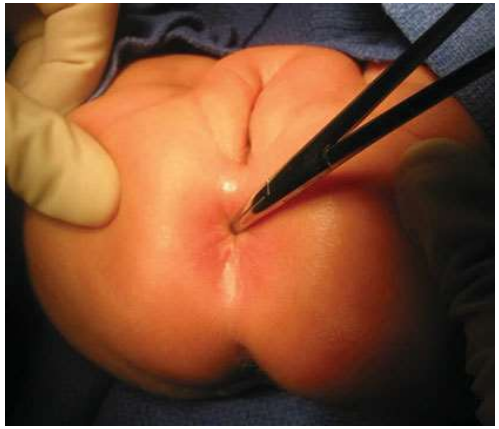

Type 1
Type 2
Fistula ligation
Ischemia/ perforation
Healthy pouch
Excise pouch
Pull through
Excise pouch Ileostomy
Pouch in-situ Ileostomy
Ileo-anal pull through
Coloplasty/ Pull through
Stoma closure
Stoma closure
Type 3
Type 4
Fistula ligation
Excise pouch End colostomy
Pull through/ Transverse colostomy
Stoma closure
Type 5
Fistula ligation
Excision of distal pouch
Coloplasty of proximal pouch/ Ileostomy
Pull through
Stoma closure

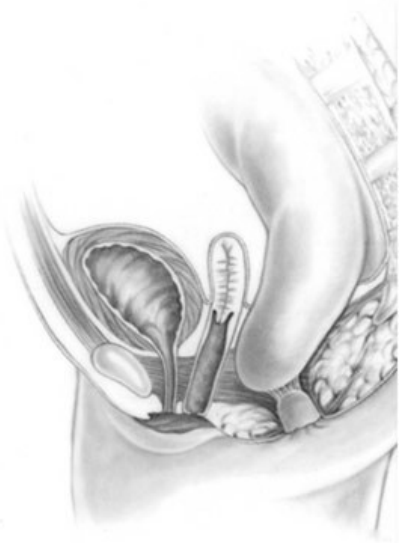
2 - Rectal atresia

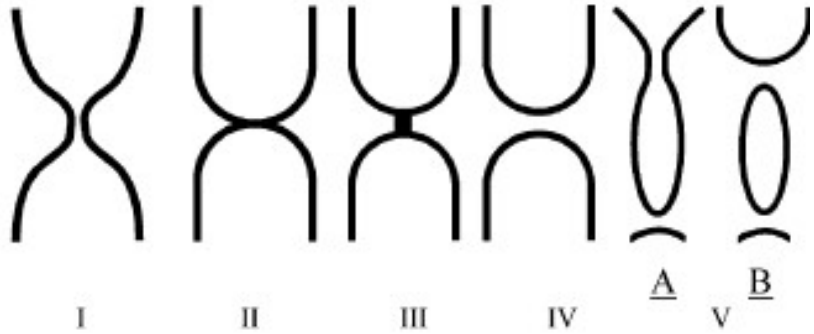
Definition and spectrum

- **1% of ARM** (used to be more common in southern India= 14%)
- More common in males
- anus seems completely normal (have dentate line), but there is an atresia or stenosis
- sphincter mechanism is excellent in most cases.
- do not have typical association with other defects like in other ARM
- Workup should specifically evaluate for **Currarino syndrome** and an associated **presacral mass**

Definition and spectrum







- External appearance of normal anus, but there is atresia at the junction with rectum. Sometimes it's a thin septum that can be perforated, sometimes very thick, connected by fibrous strand or significant gap. Rarely atresia with stenosis or multiple atresias can occur.

Etiology

- **Embryologic –** vascular accident ~13-14 wk
- **Genetic –** high incidence with consanguineous marriages (south India)

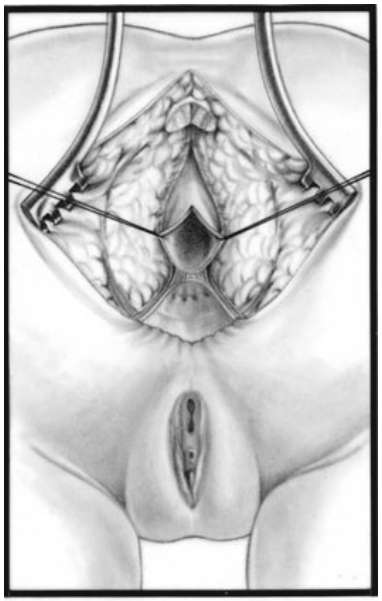
Presentation

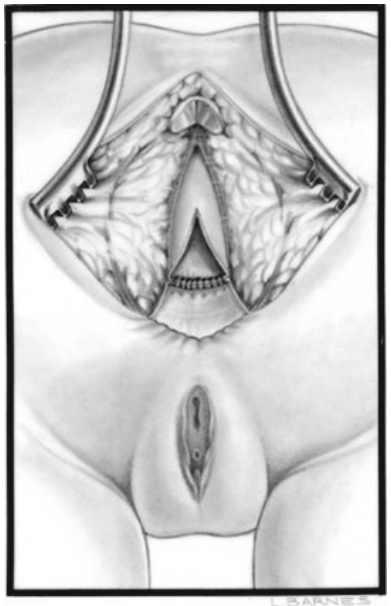
- Usually missed and present with obstruction 3-5 days
- forceful anal catheterization may easily perforate the bowel resulting in peritonitis
- thermometer, is passed and stops 1.5–3 cm depth from the anal verge.

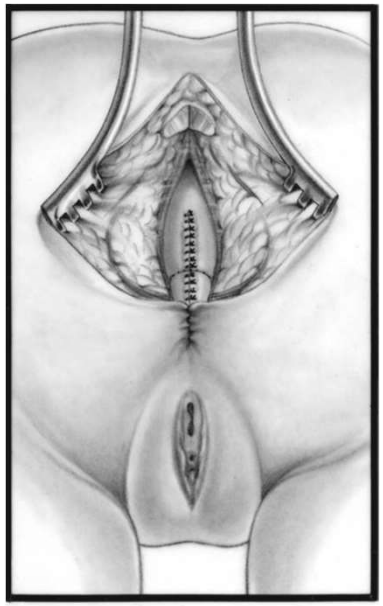
Management

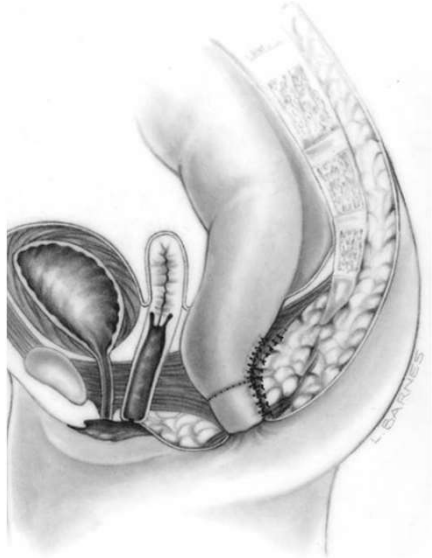
- Rectal stenosis is managed with dilatation
- Rectal atresia
  - Approach – preserve dentate line
    - **Transanal** = for septum/memb (pushed from colostomy and divided transanaly)
    - **Posterior sagitally** = for gap (satisfactory result for most)
    - **Transabdominal mobilization** = rarely required
  - Timing
    - Colostomy & rectal biopsy => gap assesment (colstogram/MRI) (holschnider)
    - Primary surgery (pena)
- Postop dilatation is required like other ARM

Repair



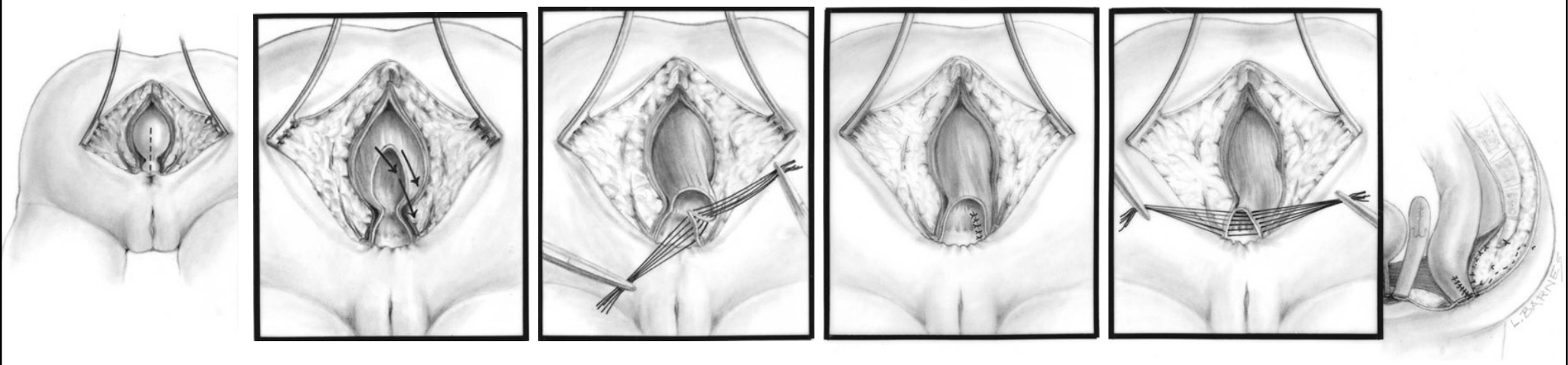






- **Posterior sagittal incision:** Remove septum and ano-rectal anastomosis

Repair



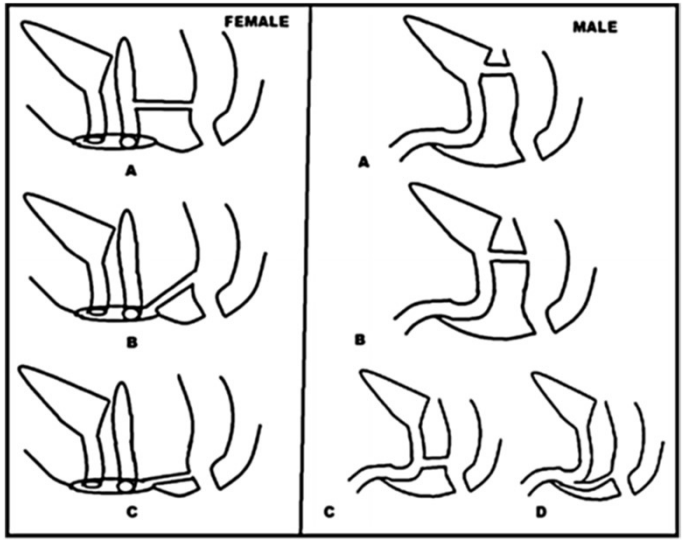
- **Posterior sagittal incision (Technical variant for size discrepancy)**: Rectum anastomosed to anus anteriorly, but pulled down to skin posteriorly. This enables enlarging circumference of anus, while maintaining enough sensation.

Outcome

- Excellent bowel control but suffer from constipation

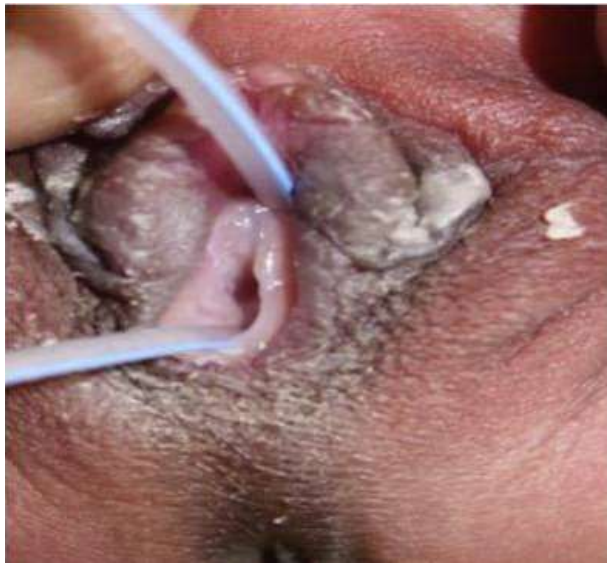
  b/c of dilated rectum

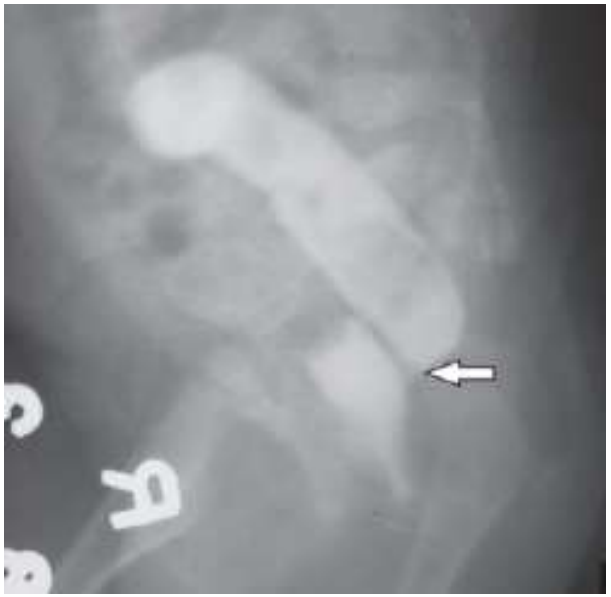
3. H type fistulas



- Congenital rectourogenital connection and an external anal opening in a normal or ectopic position. Can be **Urethral/vesical** in males and **vaginal/vestibular** in females. The anomaly in males is usually associated with a stricture of the anterior urethra as the baby uses the fistula to pass urine through the rectum. Variety of procedures have been described including PSARP/ASARP, transanal excision of fistulous tract, vestibulo anal pullthrough, endorectal pullthrough. Urethral hypoplasia can be managed with diltation or urethroplasty

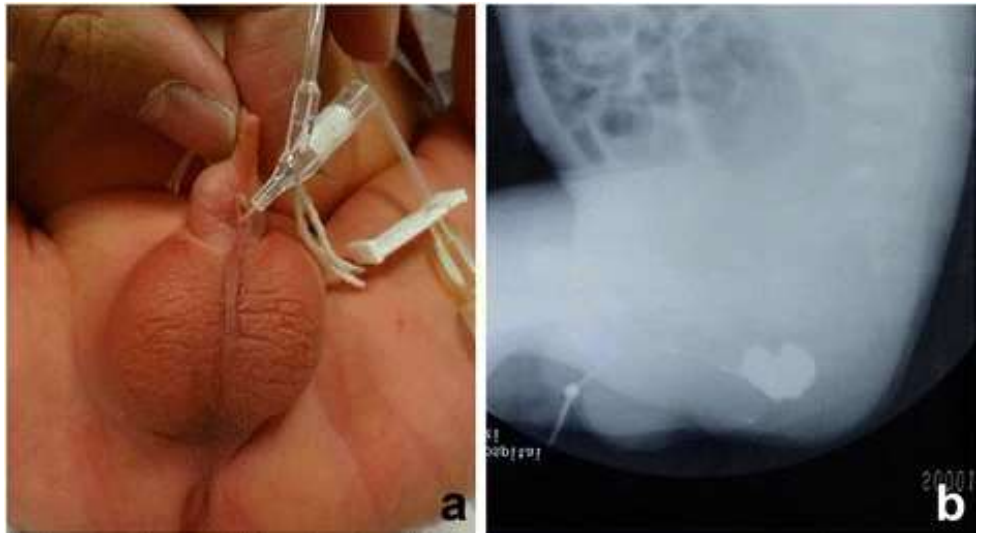
4. Recto-vaginal fistula





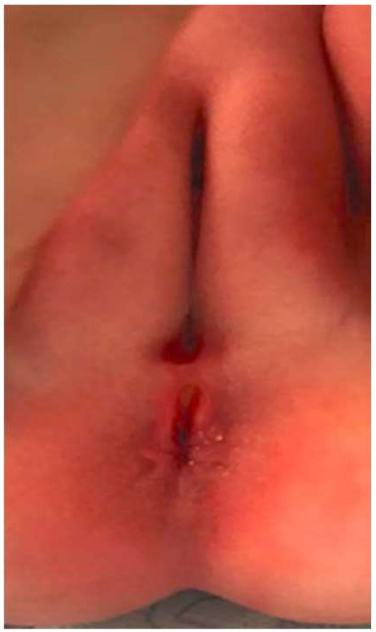
- In the past many vestibular and cloacal fistulas had been erroneously labeled as vaginal fistulas. Rectum can open low (lower vagina) and fistula may be visualized inside hymen during perineal inspection. Rectum can be high (posterior fornix) and suggested by only two openings in interoitus. Can be managed by PSARP

5. Recto-penile fistula



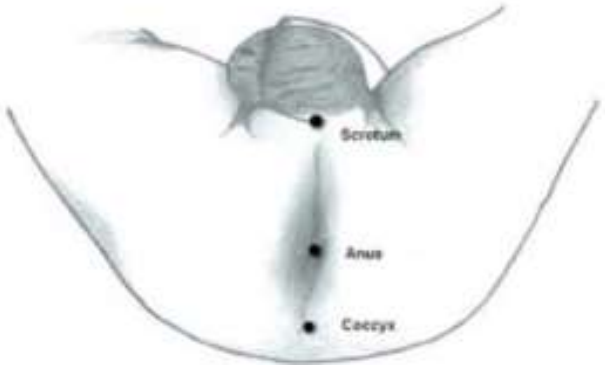
- Unlike the common varieties of rectourethral fistula, the bowel in these anomalies terminates in the penile urethra distal to the bulbar portion. There may be fistula opening in ventral penile skin without communication to the urethra (tract extend in spongiosum). Some performed staged procedure with colostomy while others performed primary surgery with aid of fistulography.

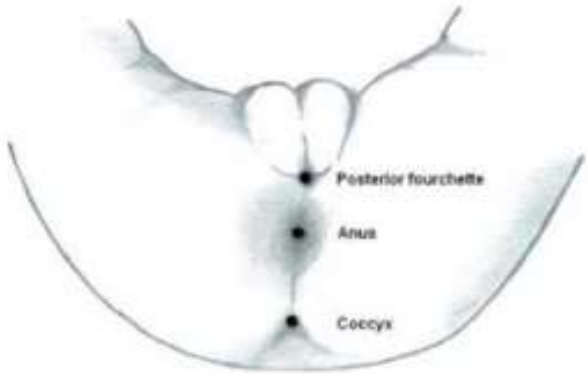
6. Perineal groove

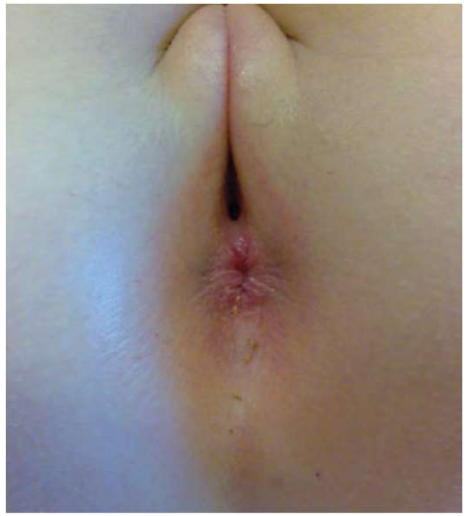


- anal opening is normal but there is an exposed wet sulcus with non-keratinized mucous membrane that extends from the posterior vaginal fourchette to the anterior ridge of the anal orifice. The lesion can be misdiagnosed as contact dermatitis, trauma, or even sexual abuse. No surgery is indicated.

7. Anterior Anus

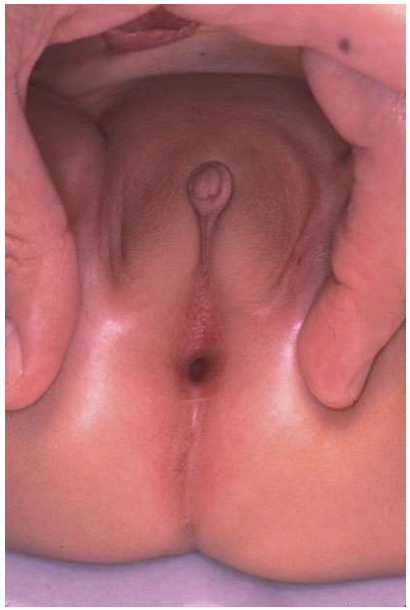


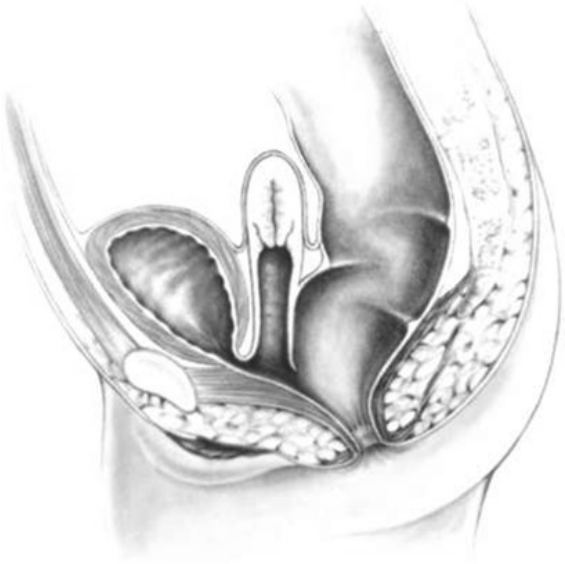


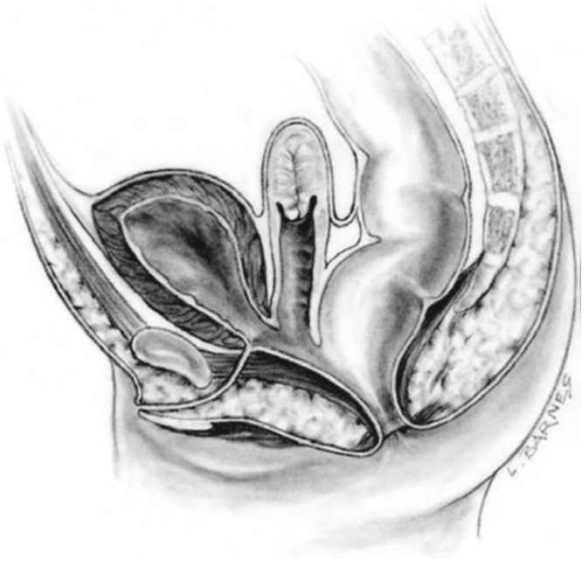


- Anus in **anterior perineum** (anal index of less than 0.34 in girls and less than 0.46 in boys), but with of **adequate caliber** and **surrounded by muscle complex** over 360 degrees. The condition is more common in girls (2:1) and not associated to other anomalies. The relations with constipation is controversial. Some authors recommend surgery if constipation is severe. The anomaly has not been seen either by Peña or Holschnider (believe they are *mislabeled perineal fistulas that don't traverse the whole of the striated muscle complex)*

8. Posterior cloaca

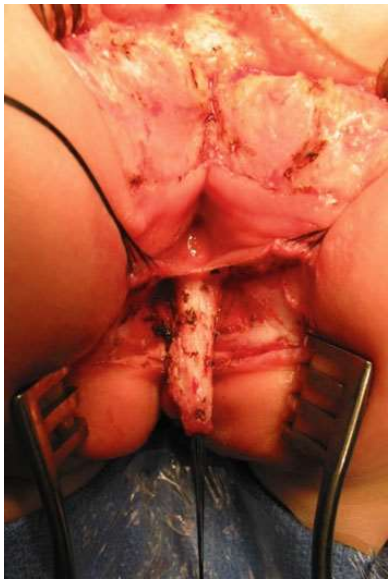


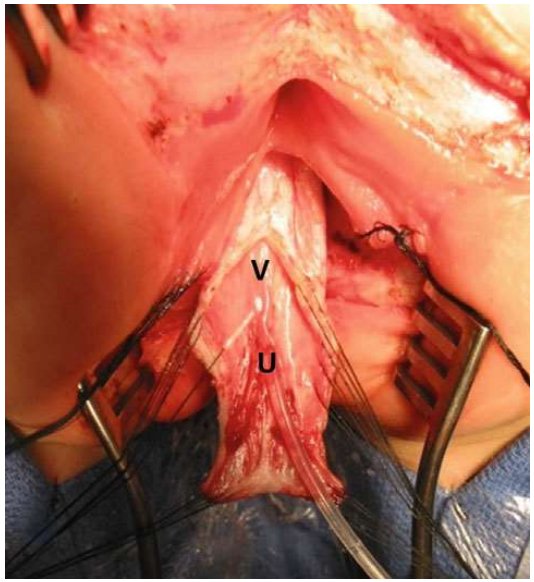


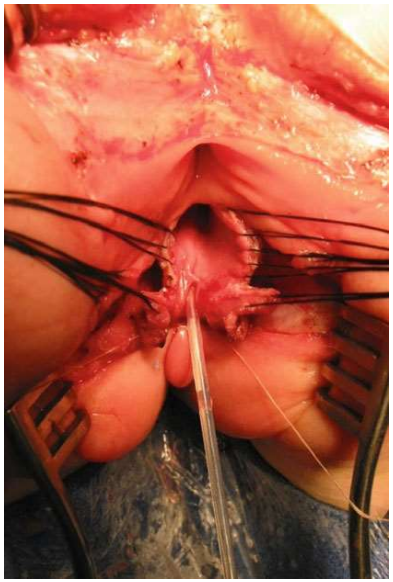


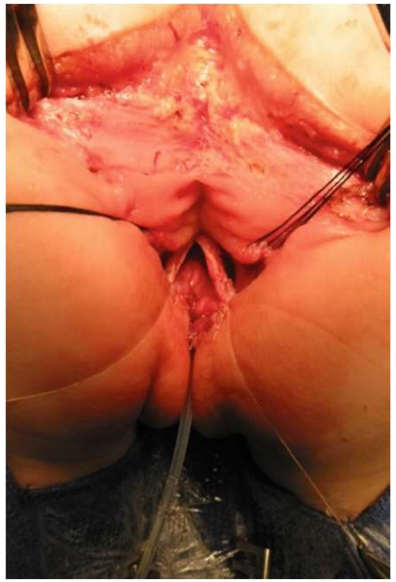
- **Posterior cloaca** A single perineal orfice located in the same location as normal anus. urogenital sinus, posteriorly deviated and connected to the anterior rectal wall. pubis is extremely thick many of these patients, when examined externally, look normal. It takes special interest to separate the labia of the genitalia in order to see the anomaly. Variants can have an accessory quasi-atretic urethra opening at the tip of a pseudophallus (clitoris)

8. Posterior cloaca



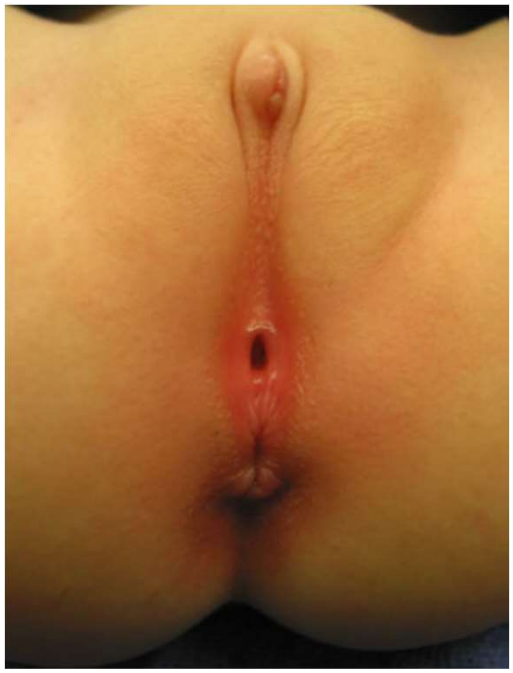


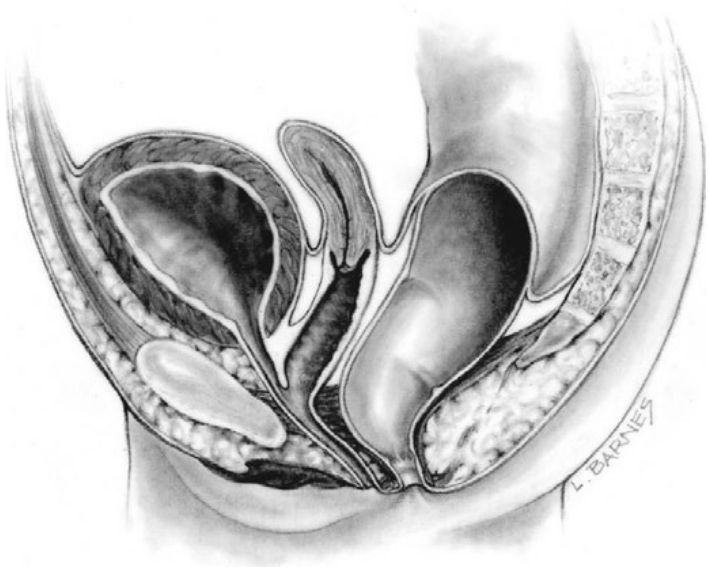




- **Repair**: perineum anterior to the anus is also divided in the midline all the way up to the clitoris. The urogenital sinus that is connected to the anterior rectal wall, usually about 1 or 2 cm from the anal verge. Pubic carving for thick pubis. Then total urogenital mobilization. The anterior rectal wall and the anal wall are sutured with two layers of interrupted longterm absorbable sutures. The posterior rectal wall is reconstructed in the same manner followed by a meticulous reconstruction of a normal sphincter mechanism

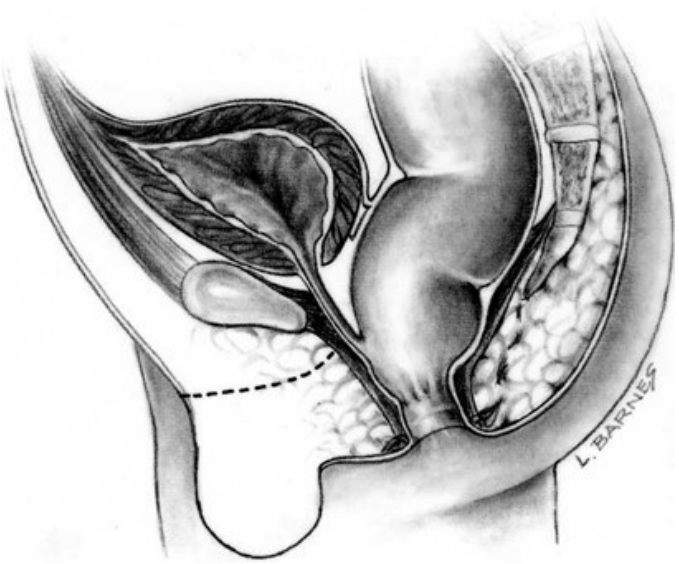
8. Posterior cloaca

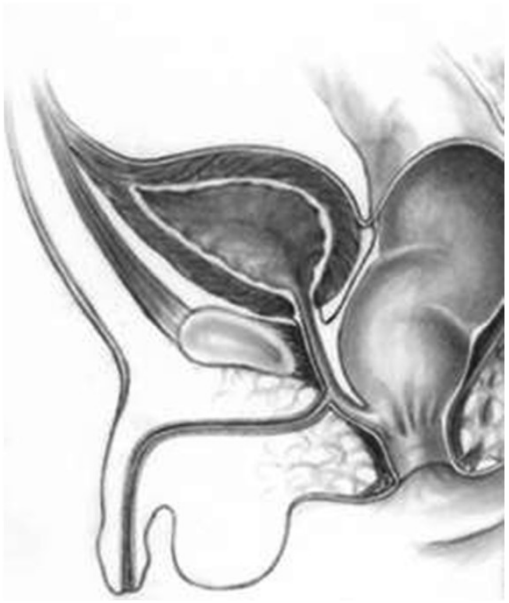


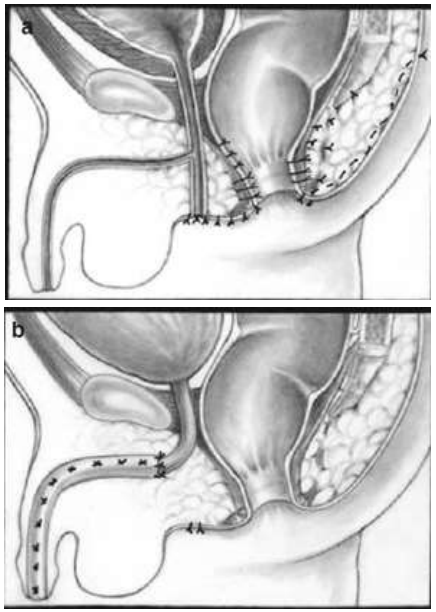


- **2 orfice variant of Posterior cloaca** : urogenital sinus opens just anterior to anus, far away from clitoris. Pena still uses same approach for the repair inorder not to compromise the repair trying to spare opening the anorectum

8. Posterior cloaca







- **Absent penis**: chromosomally males, have testicles, and are born without a phallus. The posterior urethra is posteriorly deviated and opens into the anterior rectal wall or immediately anterior to the anus similar to posterior cloaca in females. Most tend to be raised as females (orchiectomy and neovagina). A quasi-atretic penile urethra may also be found. This can be corrected in 2 stages. Creating perineal hypospadias in the first and reconstructing penile urethra in the second