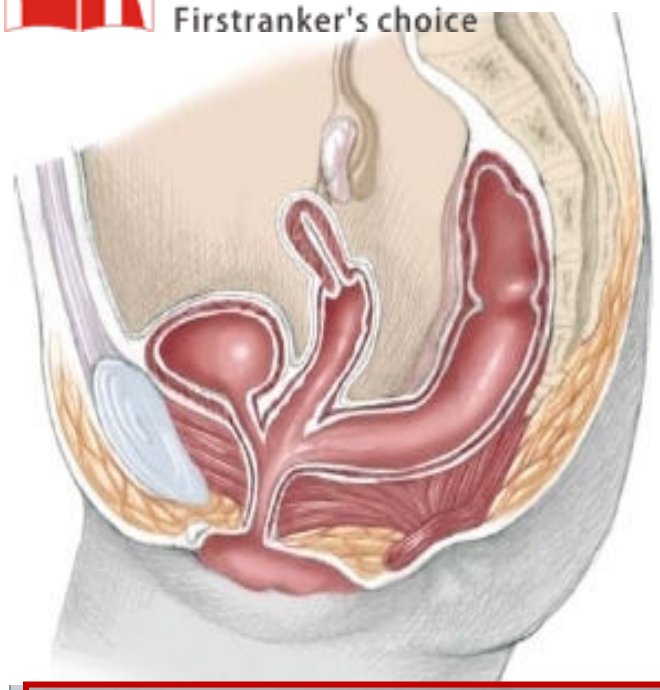
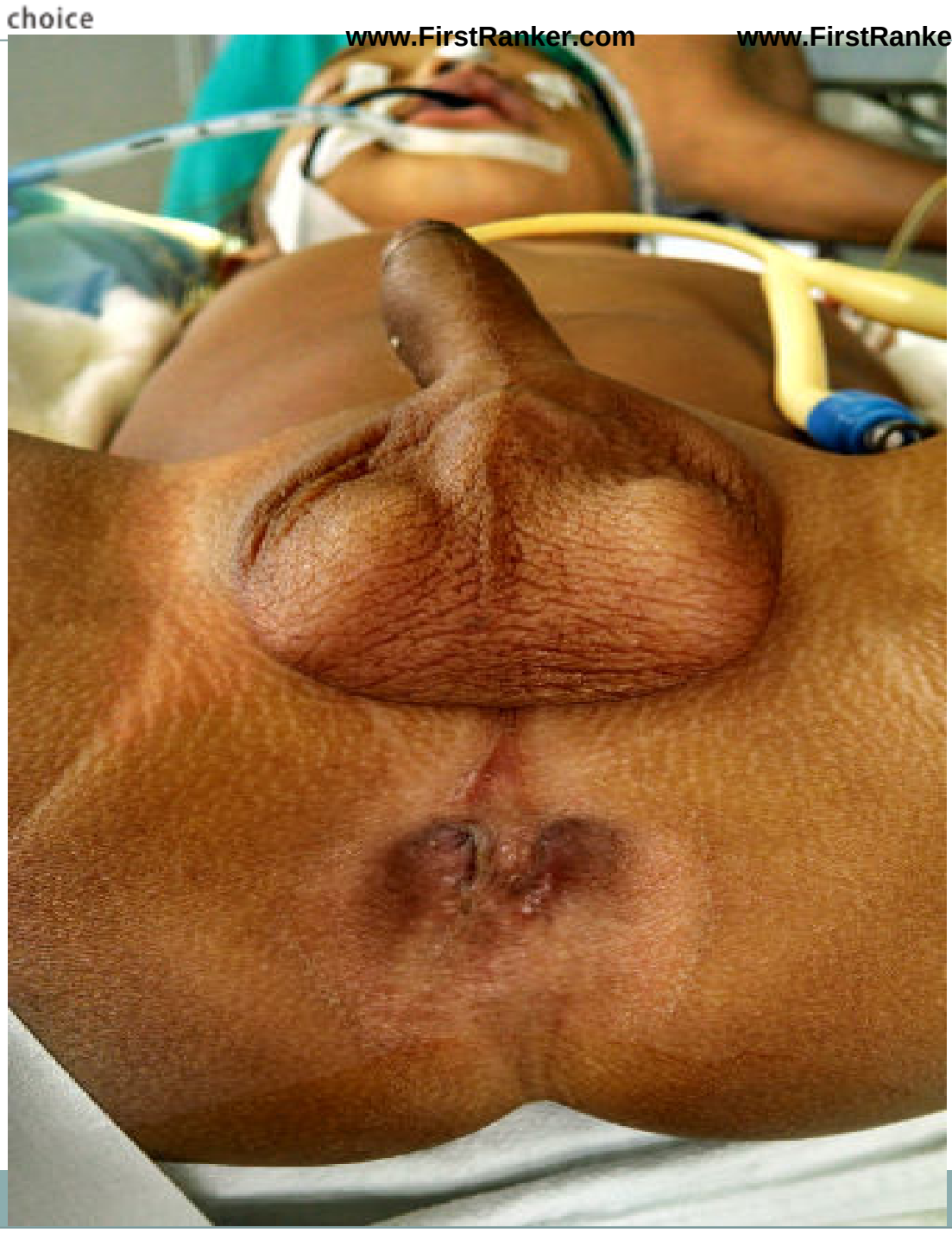
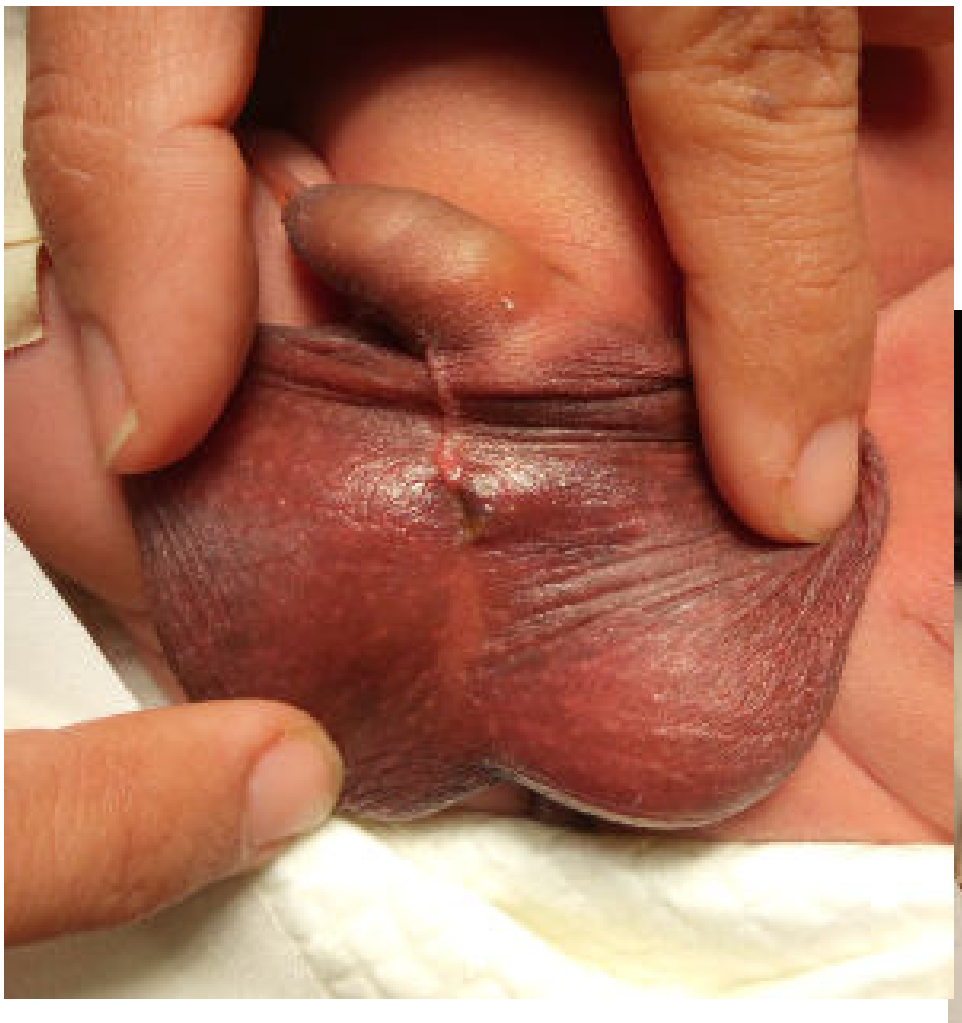


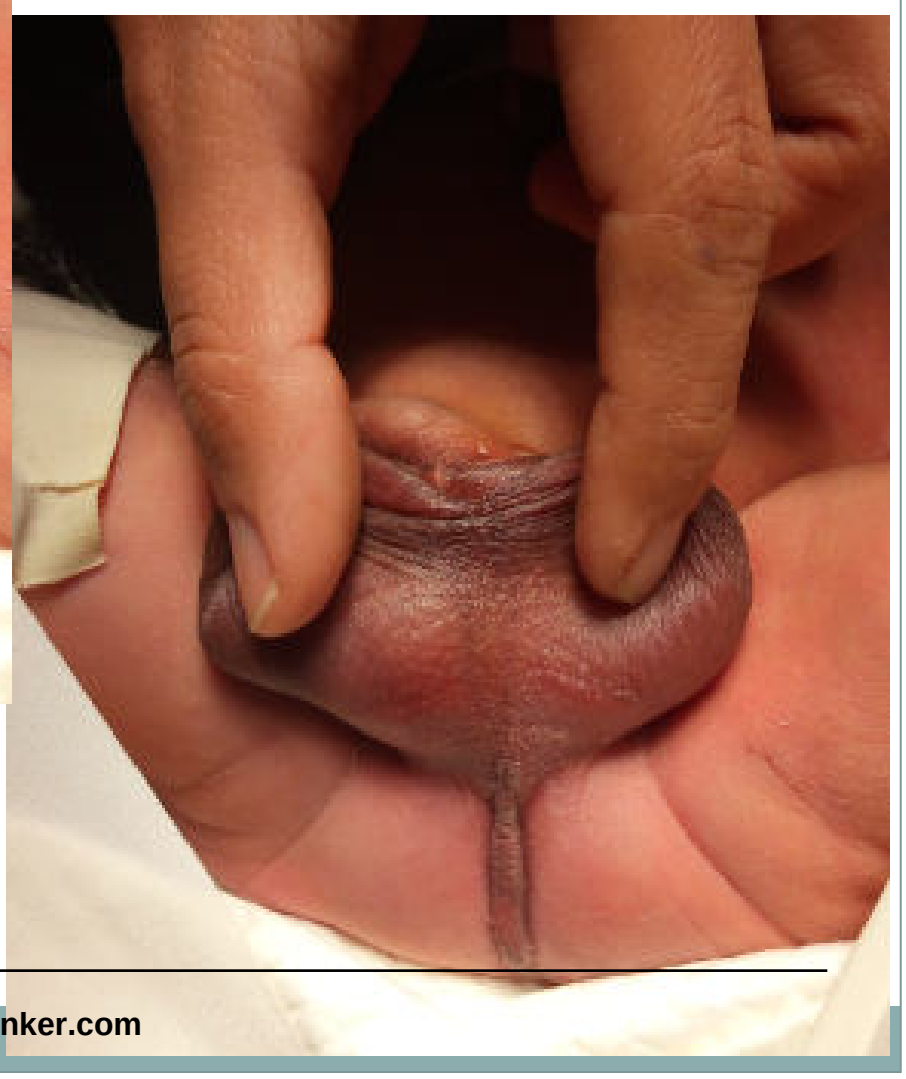
ANORECTAL MALFORMATION (ARM)

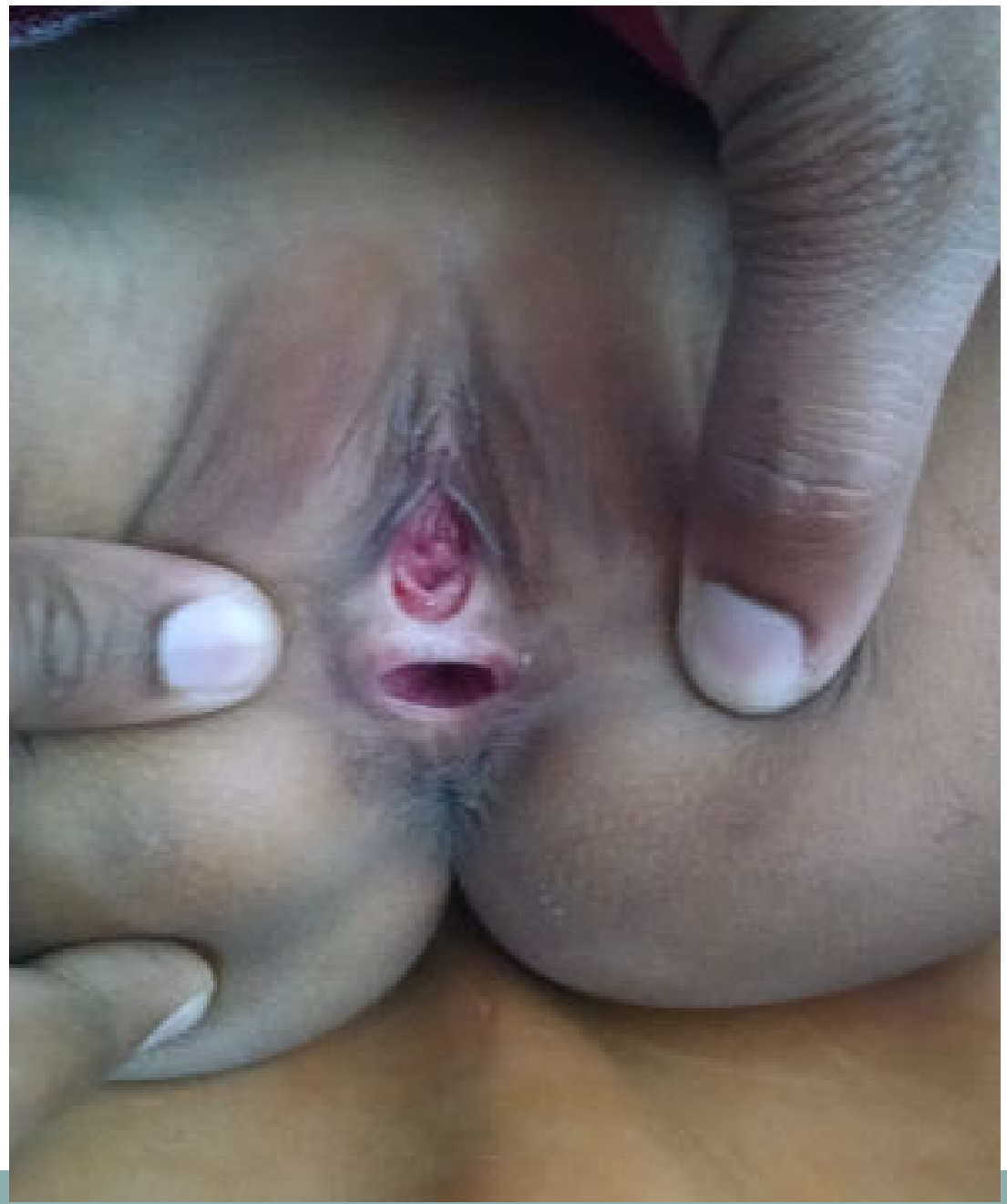
CONTENTS

- Introduction
- Embryology
- Genetics and associations
- Incidence
- Classification
- Approach to a patient



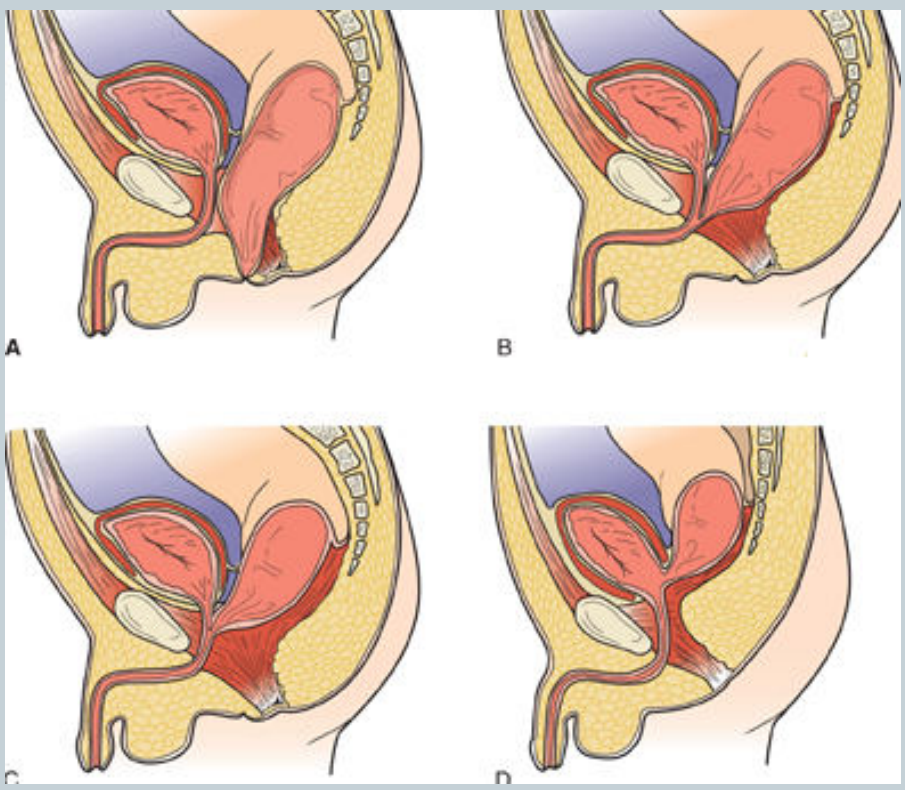






ANORECTAL MALFORMATION

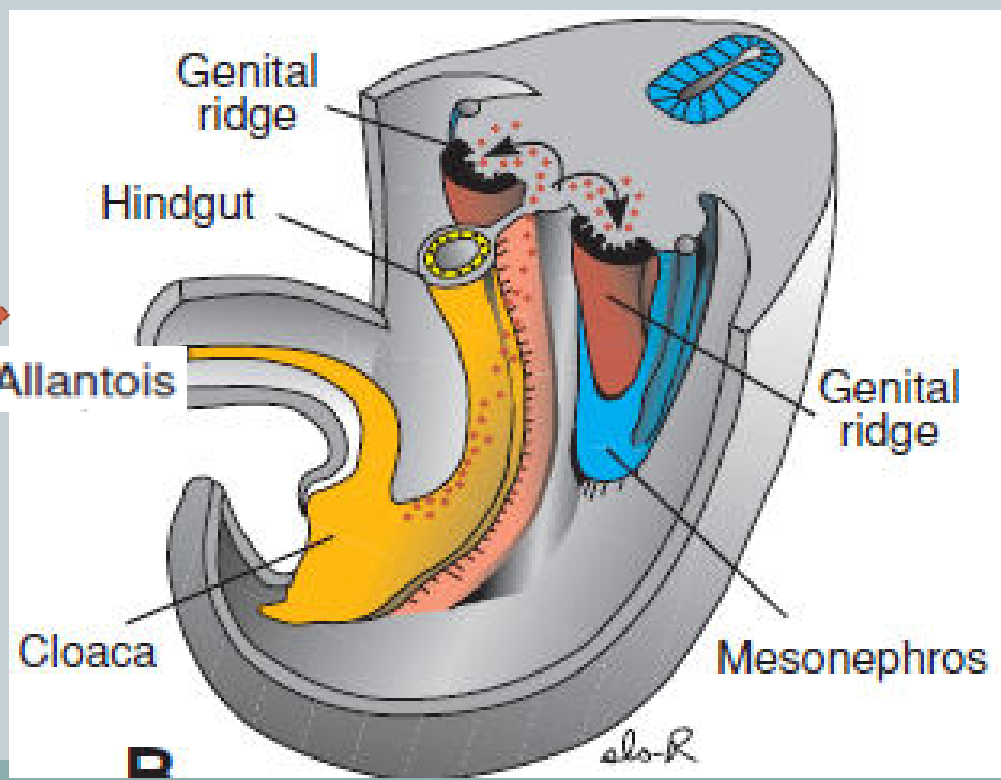
- Complex group of congenital anomalies involving the **anus and rectum**, as well as the urinary and genital tracts in a significant number of cases.



EMBRYOLOGY

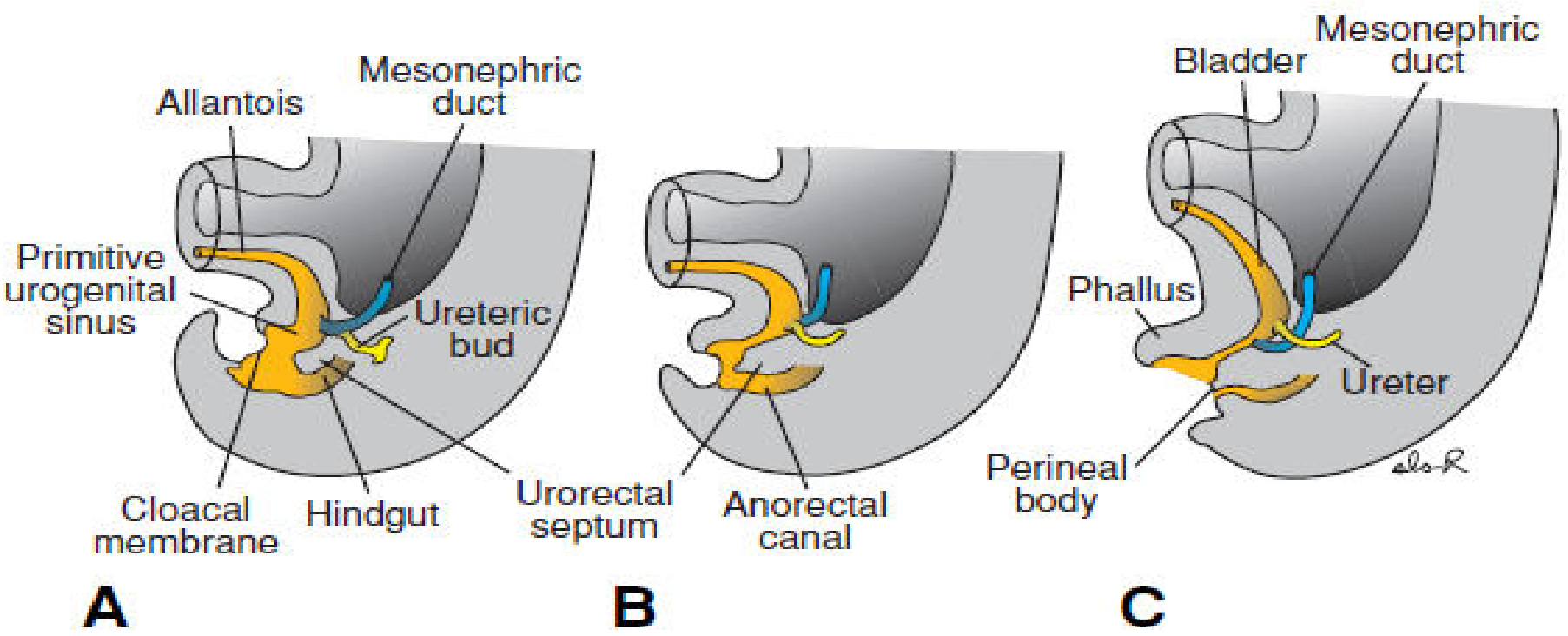
- Abnormal development of the **urorectal septum** in early fetal life.

- At 3 weeks



EMBRYOLOGY

- At 5,7,8 weeks



GENETICS

- ARM
  - NON-SYNDROMIC
    - SPORADIC
    - FAMILIAL
  - SYNDROMIC

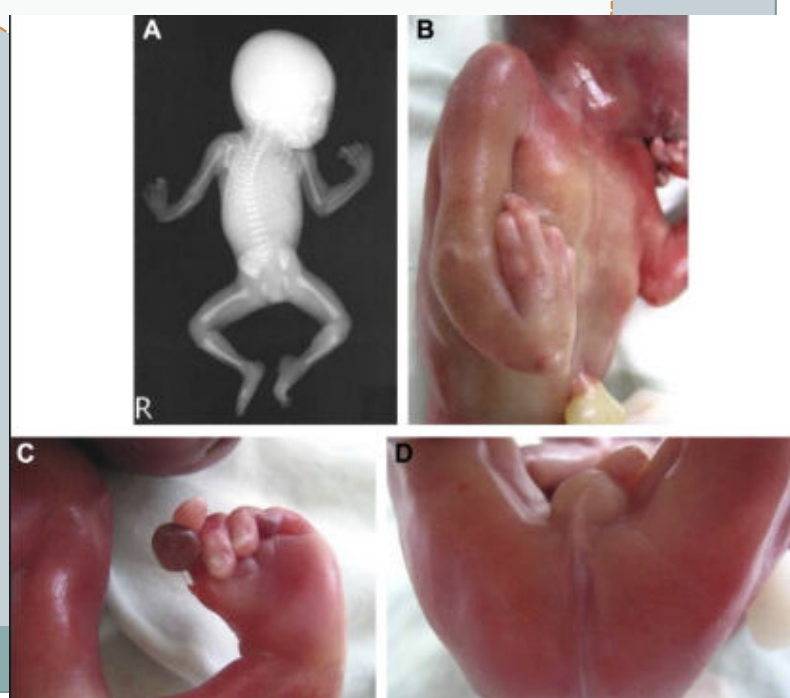


Table 1
Most Common Associated Anomalies in Patients with ARMs

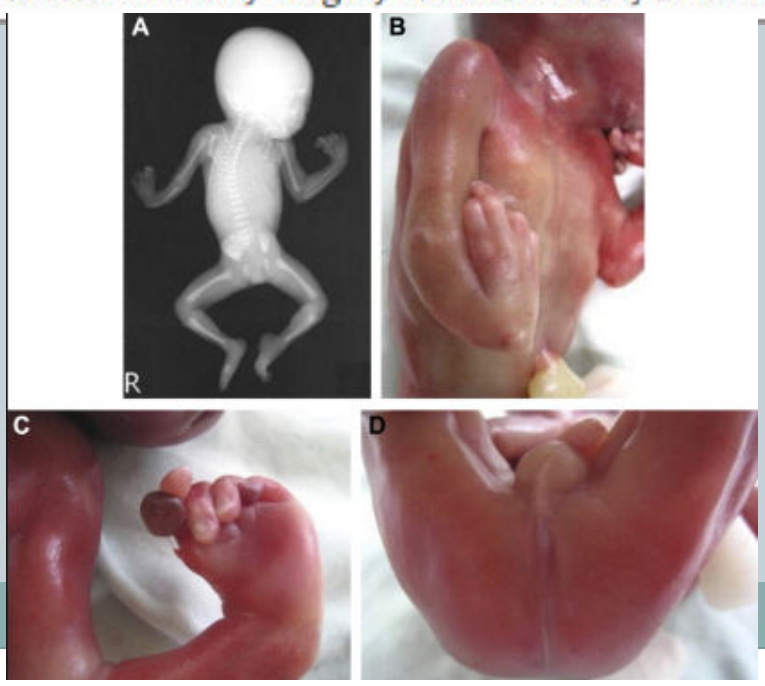
up to 70% of cases

Affected System	Anomalies
Cardiovascular	Tetralogy of Fallot, atrial septal defect, ventricular septal defect, dextrocardia, coarctation of the aorta
Gastrointestinal	Esophageal atresia; duodenal, jejunal, or ileal atresia; absent colon; intestinal malrotation; volvulus; Meckel diverticulum
Musculoskeletal	Hip dislocation or dysplasia, fusion of iliac bones, Madelung deformity, arthrogryposis, clubfoot, polydactyly, syndactyly, limb deficiency
Spinal cord and spine	Sacral agenesis, vertebral dysplasia, spina bifida, tethered cord, myelomeningocele
Urogenital	Vesicoureteral reflux, hydronephrosis, bilateral or unilateral renal agenesis, renal dysplasia, renal ectopia, horseshoe kidney, polycystic kidney, renal duplication, megaureter, exstrophy of the bladder, micropenis, hypospadias, double uterus or double vagina, vulvovaginal atresia, ambiguous genitalia

up to 60% of patients,

Table 2
Most Common Syndromes or Multisystemic Conditions Associated with ARMs

Type of Associated Entity	Syndromes or Multisystemic Conditions
Associations of congenital anomalies	VACTERL (*V*ertebral anomalies, *A*nal atresia, *C*ardiac malformations, *T*racheo-*E*sophageal fistula, *R*enal and *L*imb anomalies), OEIS (*O*mphalocele, *E*xstrophy, *I*mperforate anus, *S*pinal defects), MURCS (*MÜ*llerian duct aplasia, *R*enal aplasia, *C*ervicothoracic *S*omite dysplasia)
Chromosomopathies	Trisomy 13, 18, and 21; parental unidisomy 16; deletion of 22q11.2 and 13q; heterotaxia
Syndromes	Baller-Gerold, cat-eye, caudal regression, Christian, Currarino triad, Down, facio-auriculo-vertebral, Feingold, fetal alcohol, FG, Fraser, Ivemark, Johanson-Blizzard, kabuki, Klippel-Feil, Lowe, MIDAS, Okihiro, Opitz, Pallister-Hall, Pallister-Killian, Rieger, Townes-Brock, ulnar-mammary, Walker-Warburg

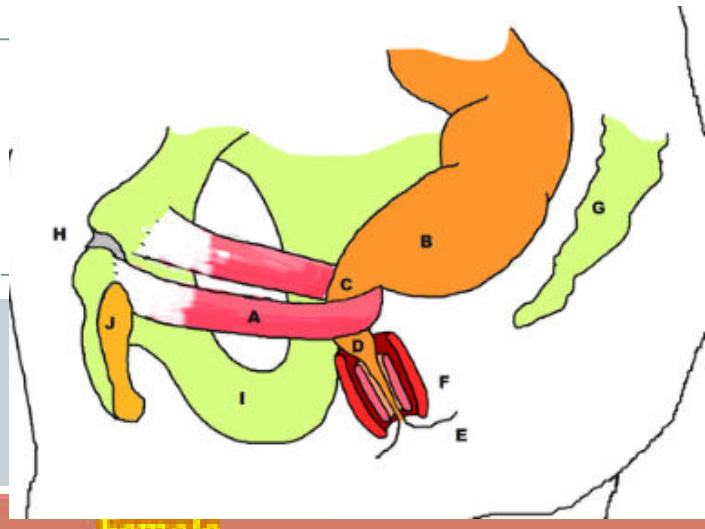


LADD AND GROSS CLASSIFICATION- 1934

Type	Anomaly
I	Anal and anorectal stenosis
II	Imperforate anus
III	Imperforate anus with blind ending pouch with fistula
IV	Rectal Atresia

WINGSPREAD CONFERENCE

CLASSIFICATION- 1984



Level of anomaly	Male	Female
High (supralevator)	1. Anorectal agenesis A. Rectovesical fistula B. Without fistula 2. Rectal Atresia	1. Anorectal agenesis A. Rectovaginal fistula B. Without fistula 2. Rectal Atresia
Intermediate	1. Rectourethral fistula 2. Anal agenesis without fistula	1. Rectovestibular Fistula, 2. Rectovaginal fistula 3. Anal agenesis without fistula
Low (infralevator)	1. Anocutaneous (perineal) fistula 2. Anal stenosis	1. Anovestibular (perineal) fistula, 2. Anocutaneous (perineal) fistula 3. Anal stenosis
Miscellaneous	Rare malformations	Persistent cloacal anomaly Rare malformations

High (Supra-levator)- Male

(a) Without Fistula

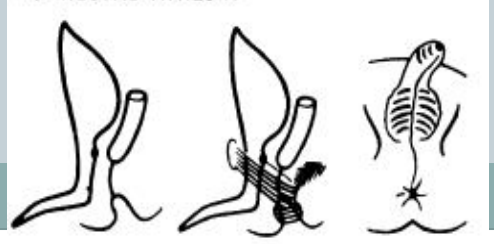
1. ANORECTAL AGENESIS (Without Fistula)

2. RECTOVESICAL FISTULA

(ii)

3. RECTOURETHRAL FISTULA

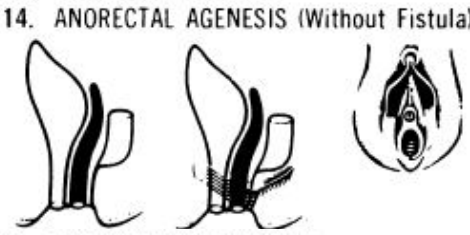
4. RECTAL ATRESIA



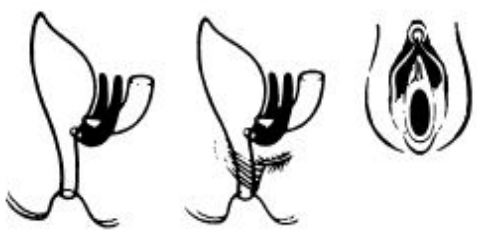
High (Supra-levator)- Female

(a) Without Fistula

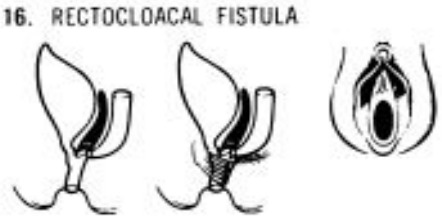
14. ANORECTAL AGENESIS (Without Fistula)



15. RECTOVESICAL FISTULA



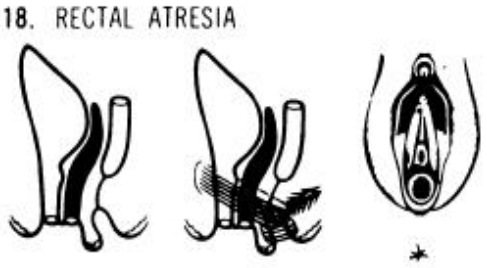
16. RECTOCLOACAL FISTULA



(iii)

17. RECTOVAGINAL FISTULA High

18. RECTAL ATRESIA



Intermediate- Male

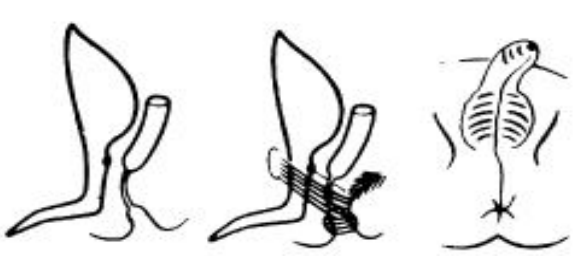
(a) Without Fistula

5. ANAL AGENESIS (Without Fistula)

(b) With Fistula

6. RECTOBULBAR FISTULA

7. ANORECTAL STENOSIS



Intermediate- Female

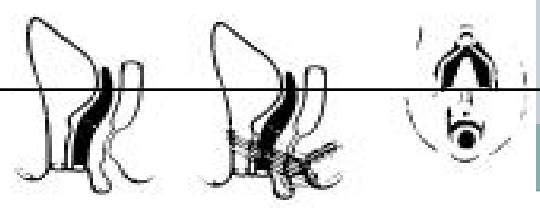
(a) Without Fistula

19. ANAL AGENESIS (Without Fistula)

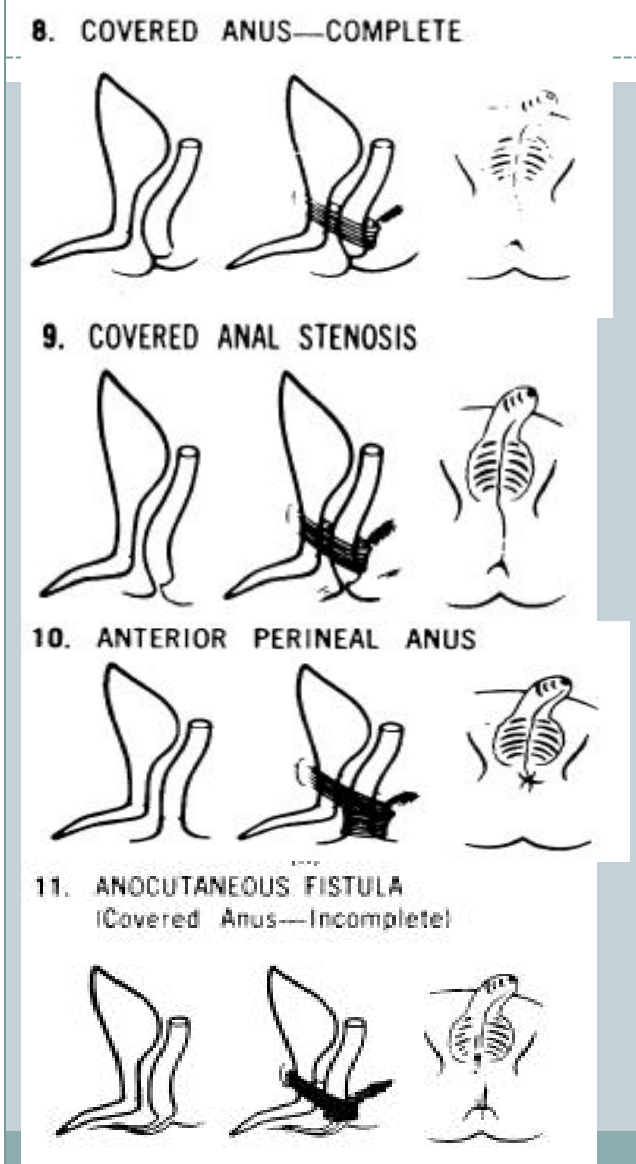
20. RECTOVAGINAL FISTULA (Low)

21. RECTOVESTIBULAR FISTULA

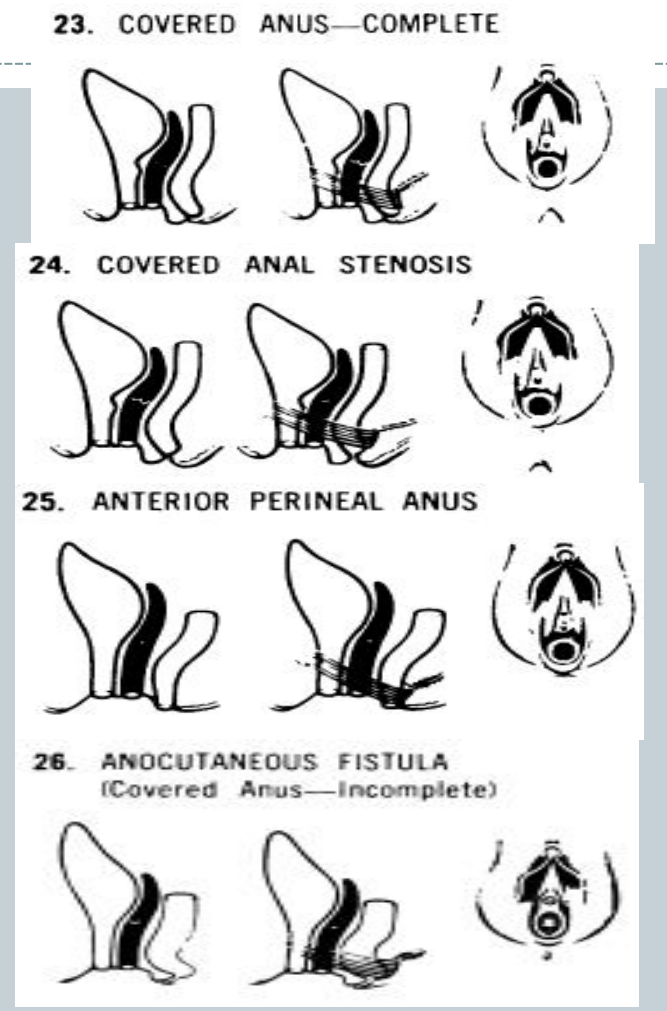
22. ANORECTAL STENOSIS

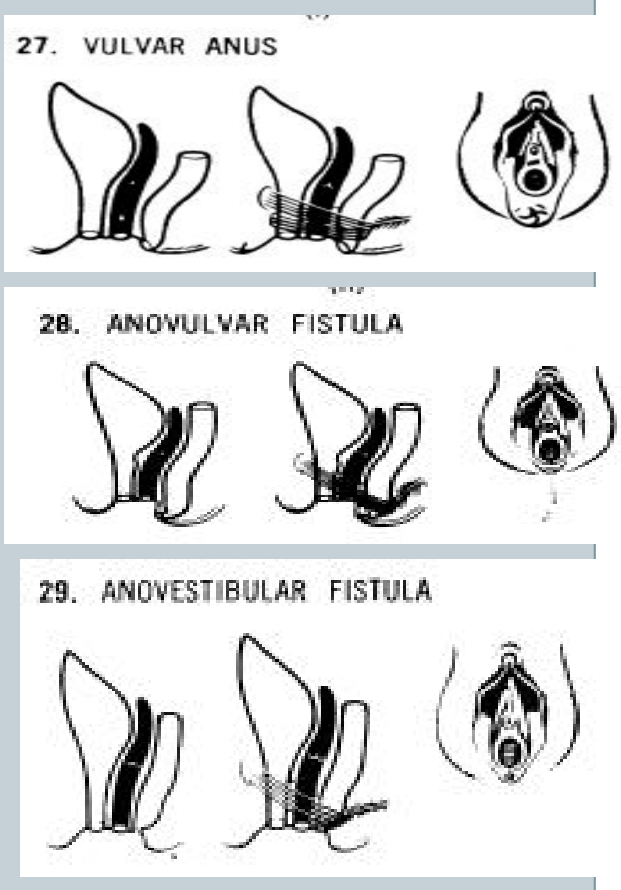


Low (Trans-levator)- Male



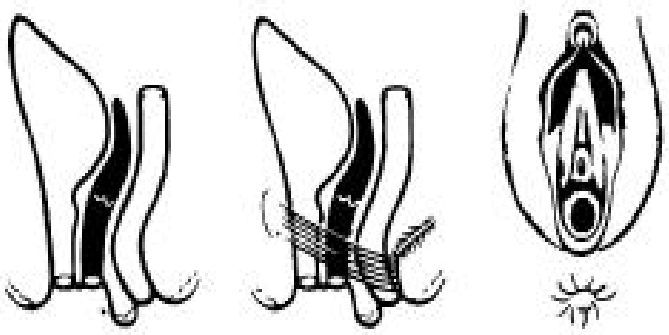
Low (Trans-levator)- Female



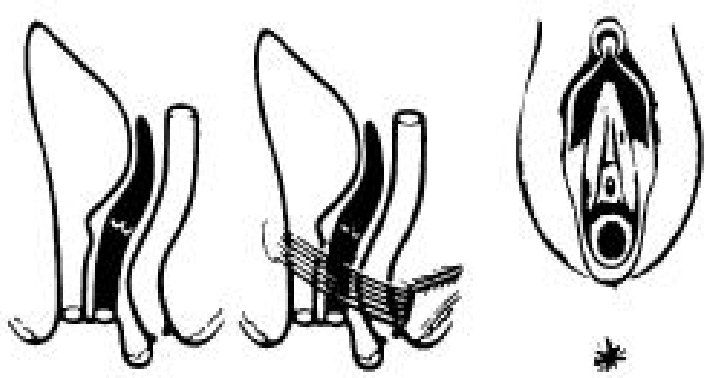


Miscellaneous- Female

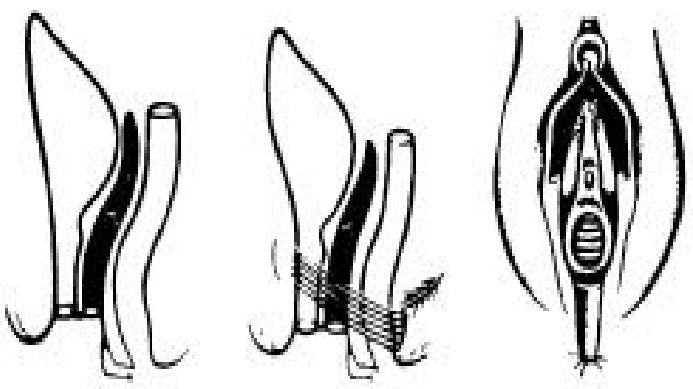
30. IMPERFORATE ANAL MEMBRANE*



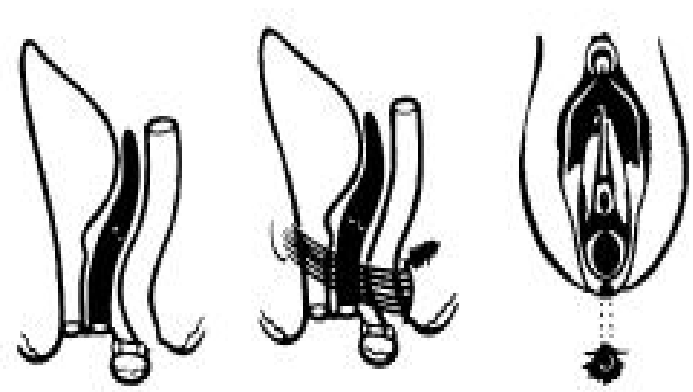
31. ANAL MEMBRANE STENOSIS**



32. PERINEAL GROOVE

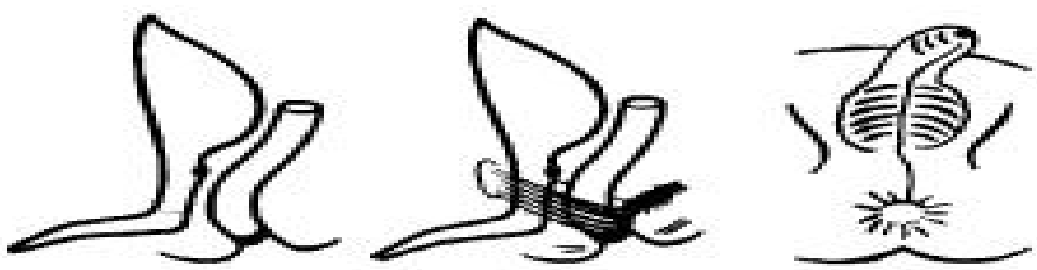


33. PERINEAL CANAL

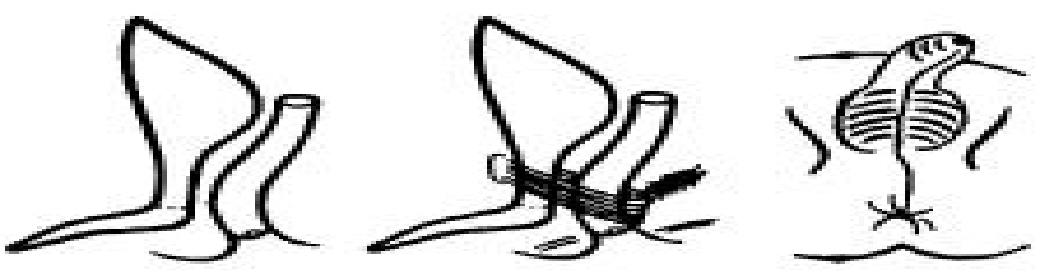


Miscellaneous- Male

12. IMPERFORATE ANAL MEMBRANE*

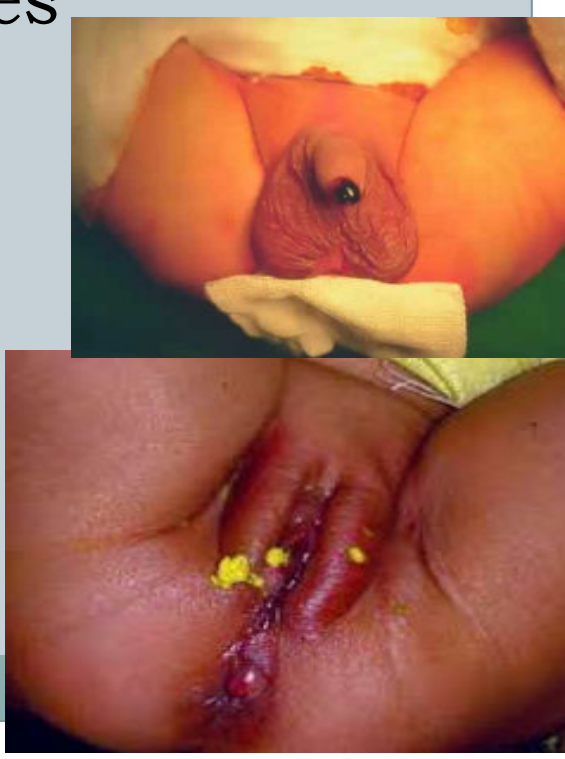


13. ANAL MEMBRANE STENOSIS**



INCIDENCE & FREQUENCY

- 2-2.5 per 10,000 live births
- M:F ratio= 1.5:1
- Supra-levator lesions more in males
- Associated anomalies more in males
- Commonest-
  - recto-urethral fistula
  - ano-vestibular fistula (female)

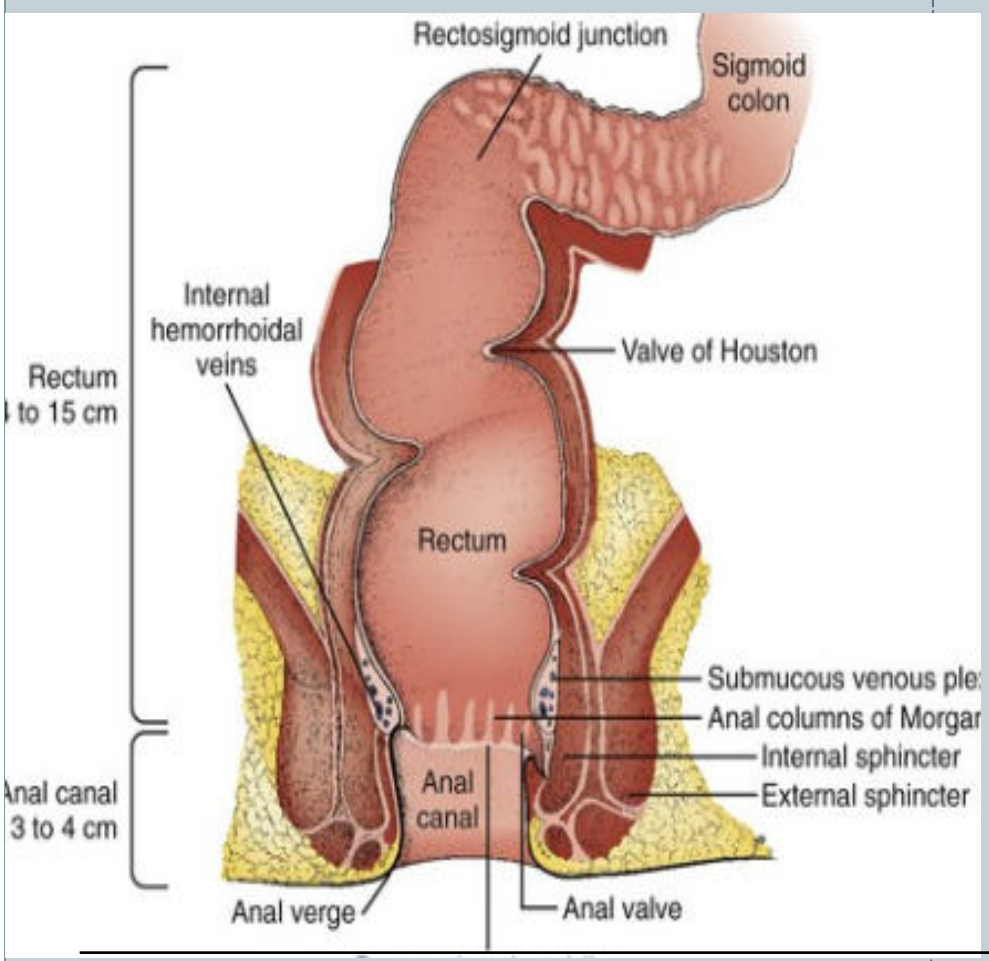


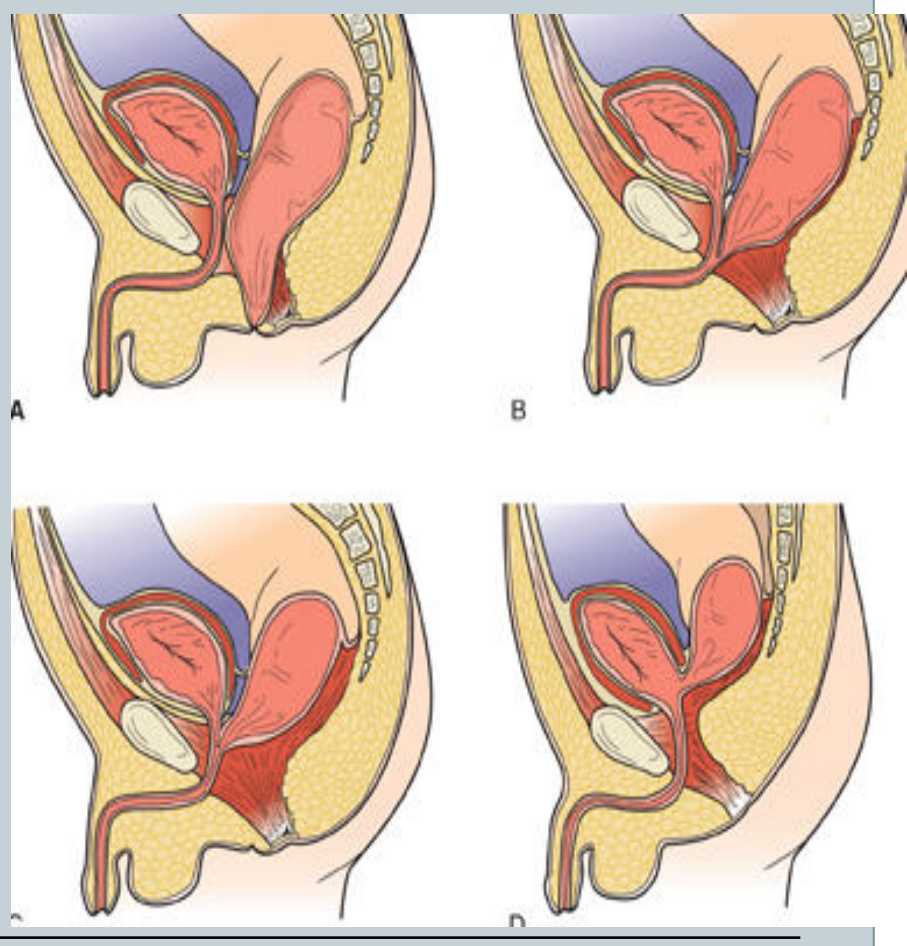
RISK FACTORS

- Mothers >35 years old
- Maternal recreational drug use
- Parental consanguinity
- Maternal residence at high altitude
- Paternal occupation in vehicle manufacture
- Consumption of raw vegetables (pesticides)
- Geographical (pouch colon)

ANATOMY

NORMAL -VS -ARM





APPROACH TO A PATIENT OF ARM

Prenatal Diagnosis- USG

(Low sensitivity and specificity)

- Dilated colon
- Oligohydramnios and a highly distended vagina (in females)
- Absence of circular rim of hypoechogenicity (Anus).
- Enterolithiasis
- Polyhydramnios if associated with UGI obstruction.
- Associated anomalies.

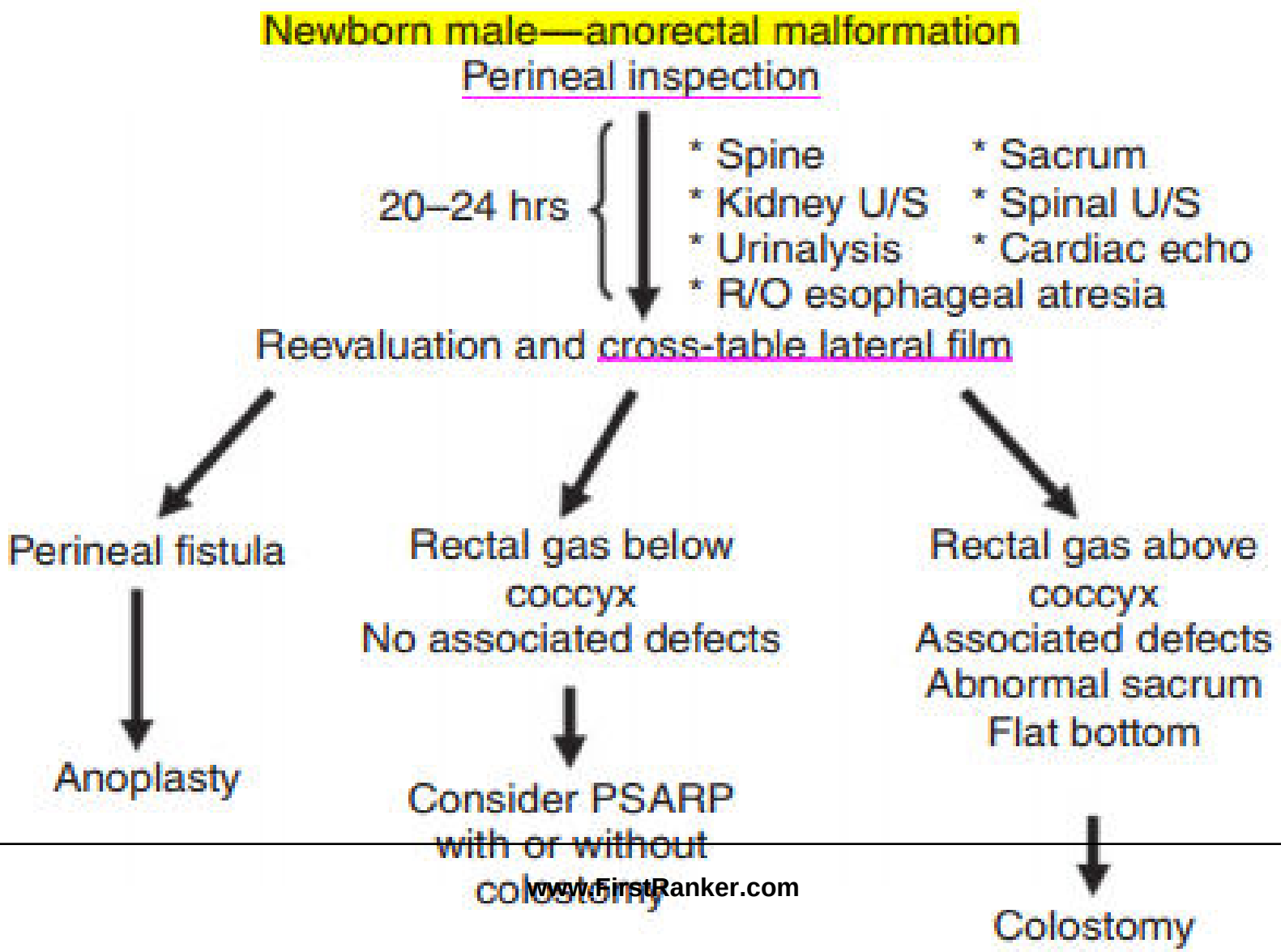
GOALS OF INITIAL ASSESSMENT

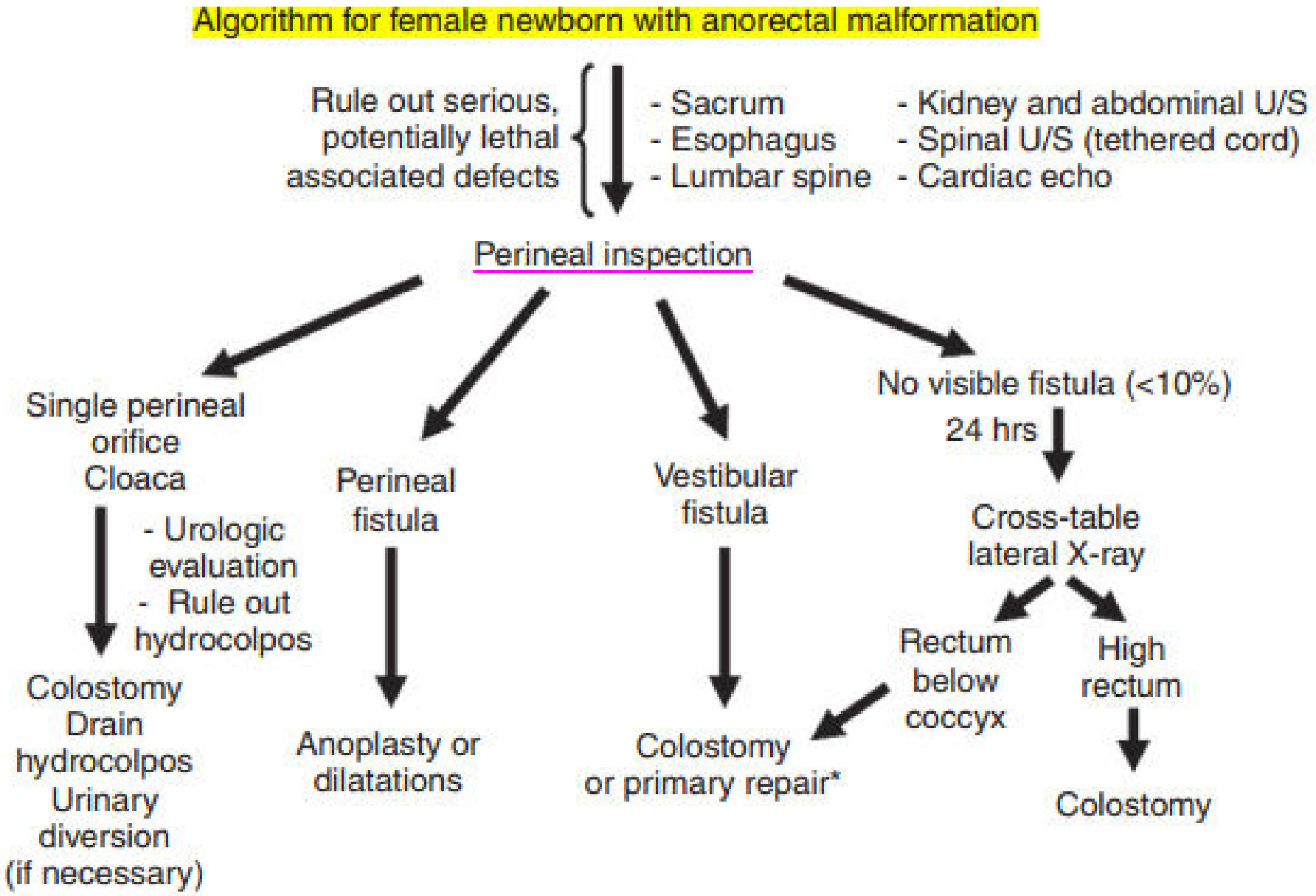
1. To **determine the level of the malformation** in relation to the muscular sphincters and the site of any fistulous communications.

2. To determine the **integrity of sphincters and their nerve supply**.

3. To document any **associated anomalies** that may affect survival.

ALGORITHM FOR MALE ARM



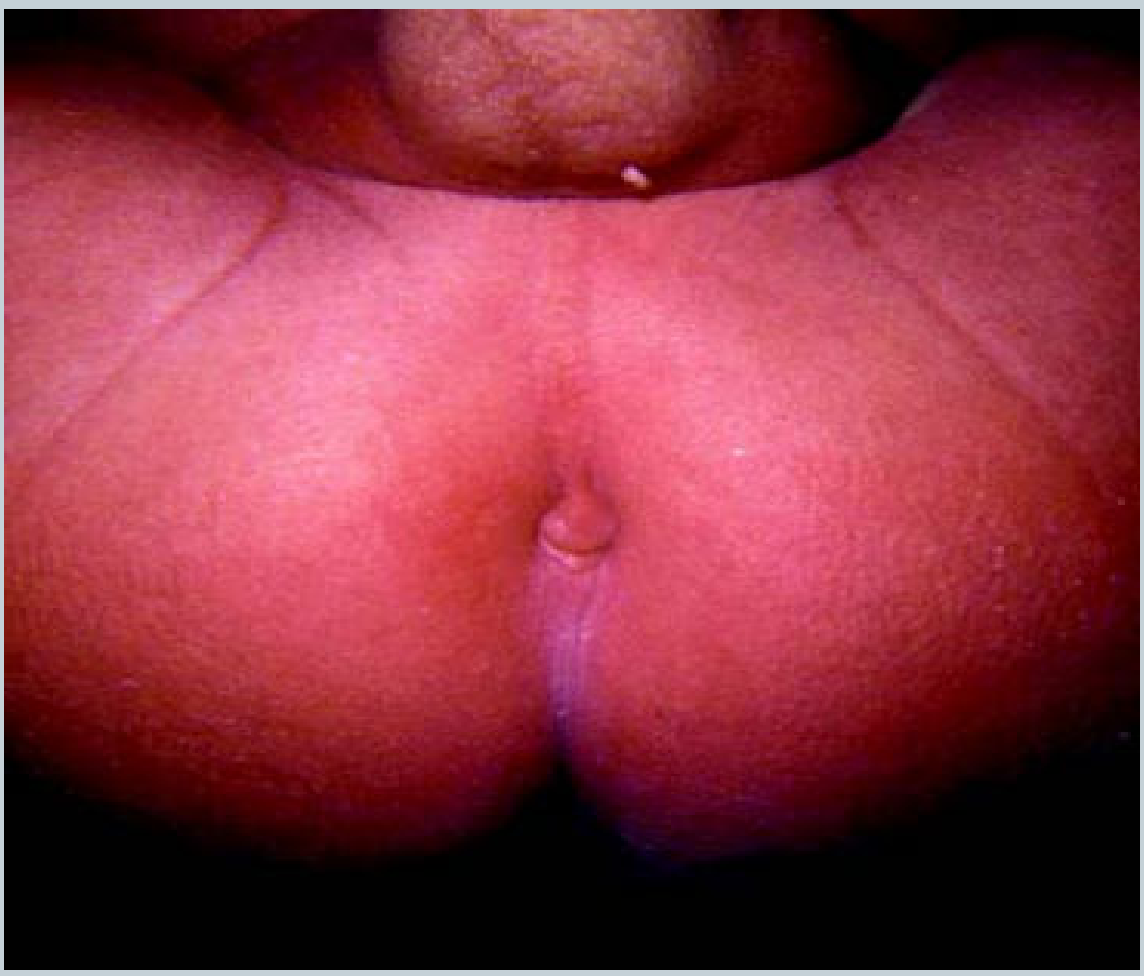


I. LEVEL OF MALFORMATION
Neonatal examination

1. **Good perineal examination** under a good light
2. Position of the **anus or its absence; fistula.**
3. **Perineal shape** (midline raphe)- parasagittal fibers and **gluteal development.**
4. **Passage of a soft catheter** greater >2 cm into the rectum and the presence of meconium passage rules out atresia

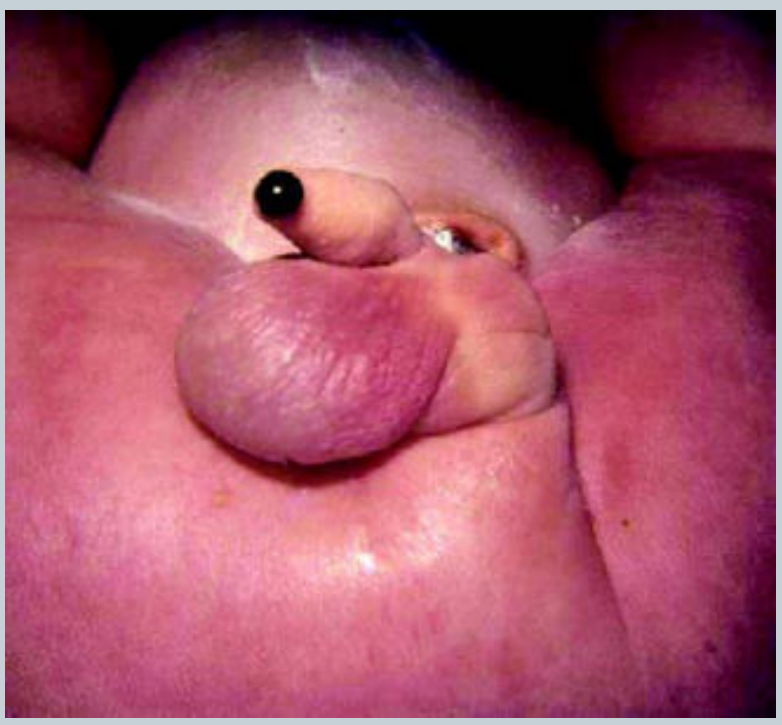
- Normal size of the anus- 1.3 + (3 × birth wt in kg) in mm

HIGH ARM-FLAT GLUTEUS



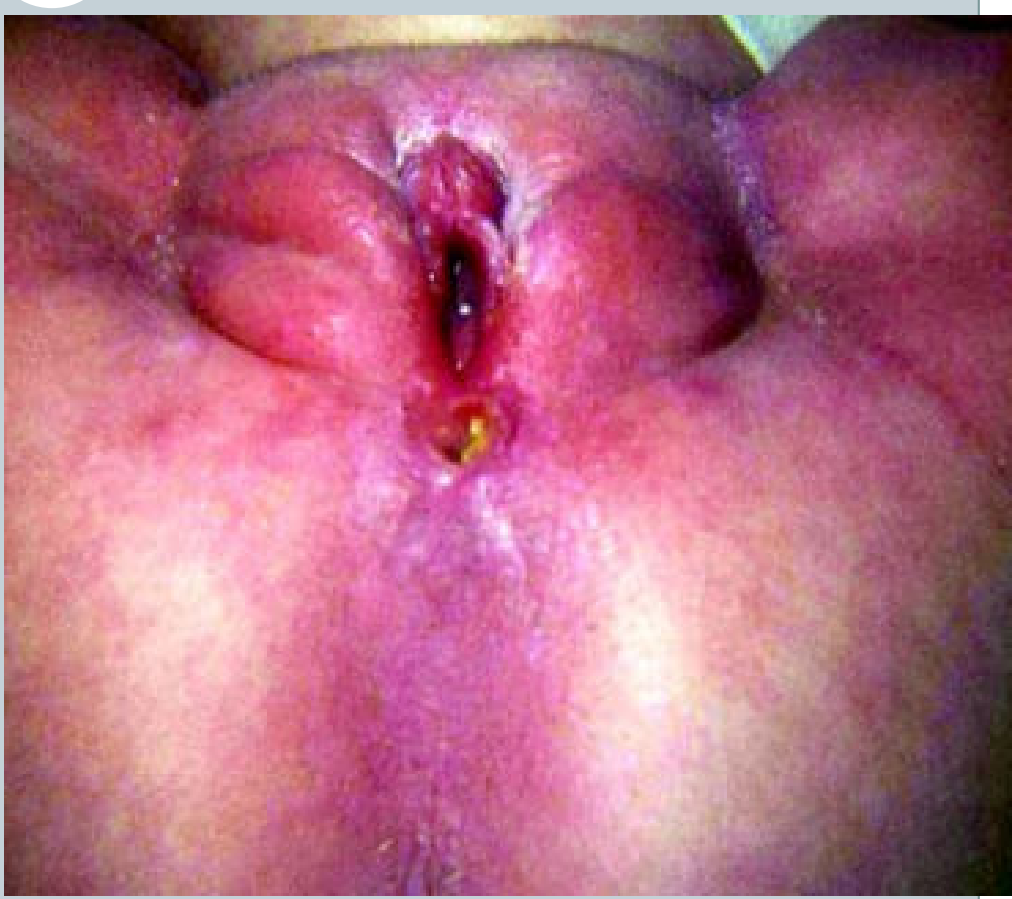
EVIDENCE OF FISTULA

- H/O meconuria
- Meconium at the tip of meatus
- Meconium/squamous epithelium in urine microscopy

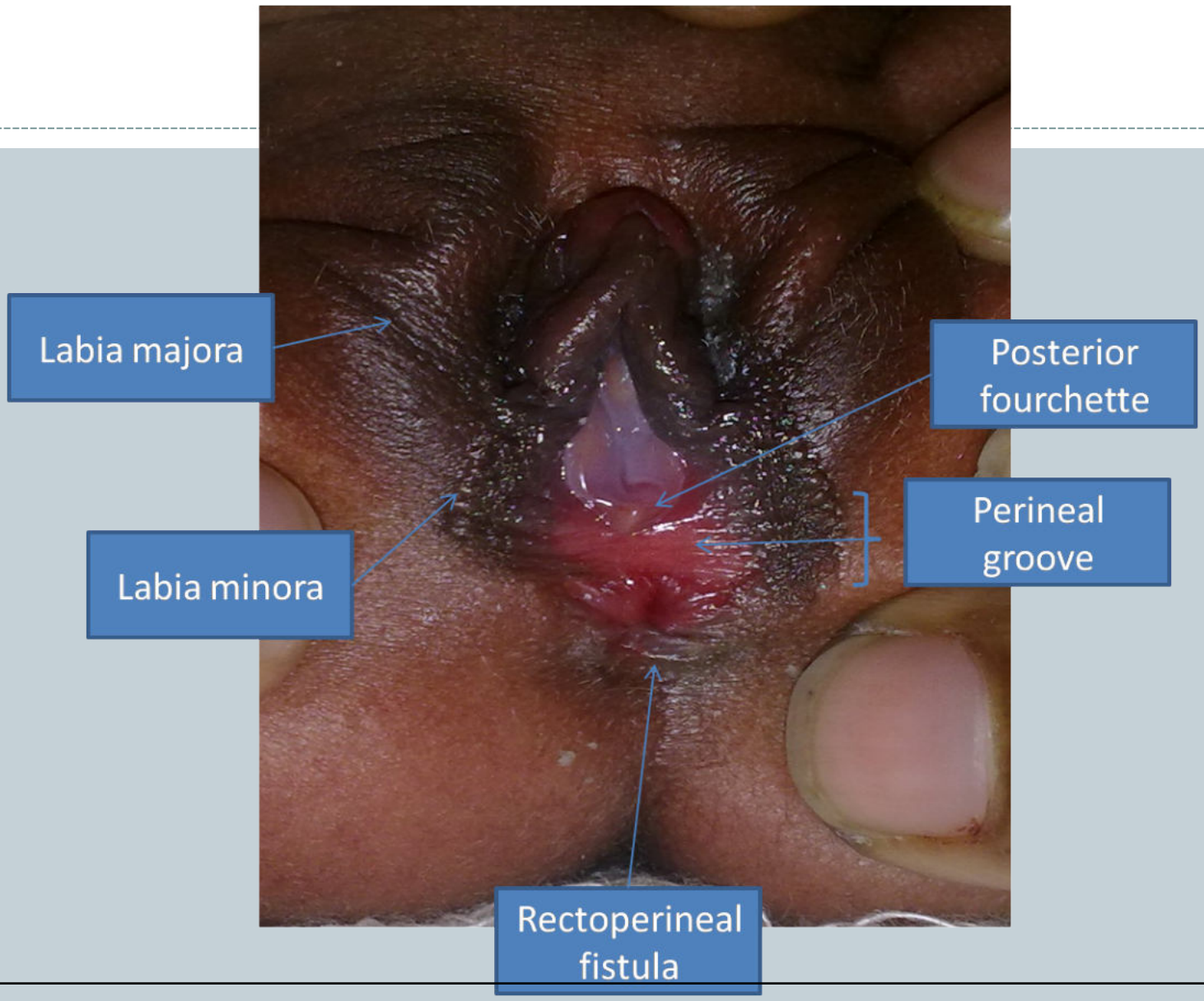


ANTERIOR ECTOPIC ANUS

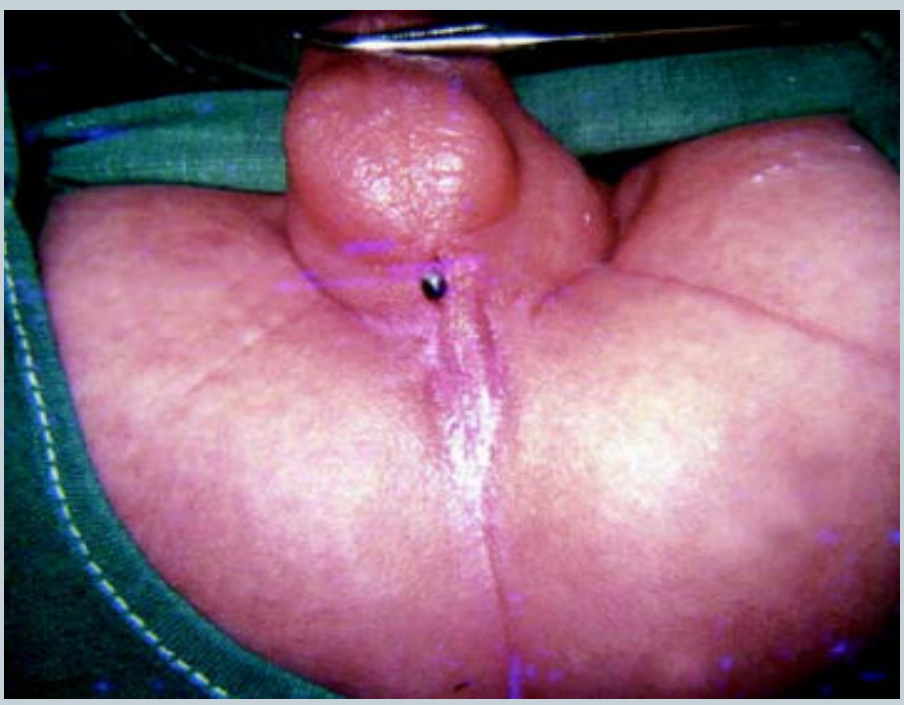
- Anal index of < **0.34** in girls and < **0.46** in case of boys

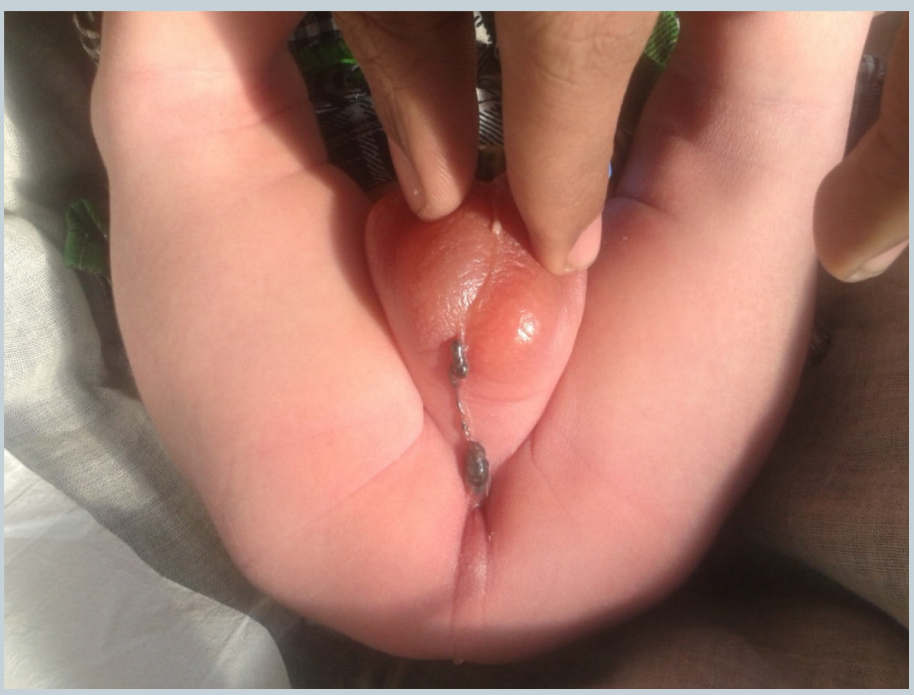


- **Anal index-**
- Ratio of scrotal-anal distance to the scrotal-coccygeal distance in males and
- fourchette-anal distance to fourchette-coccygeal distance in females.

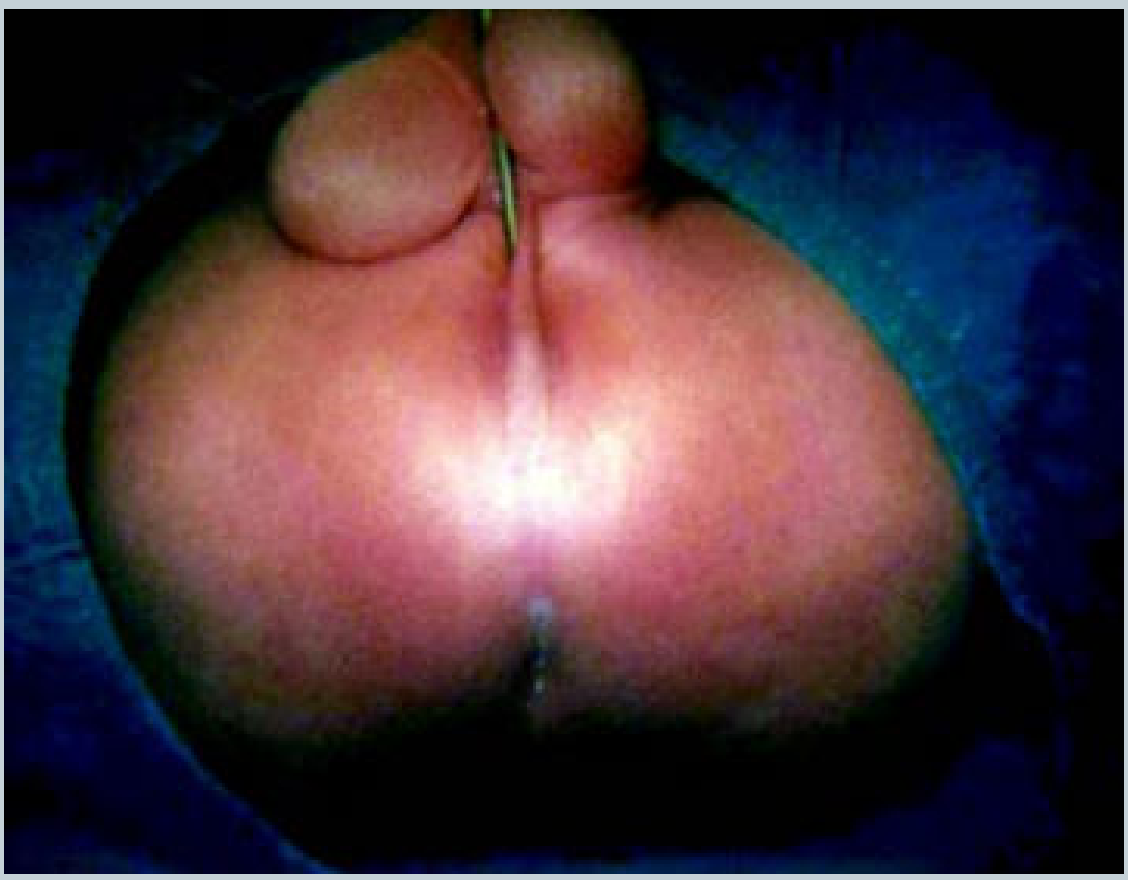


LOW ARM WITH FISTULA IN SCROTUM, MECONIUM TRACT IN PERINEUM

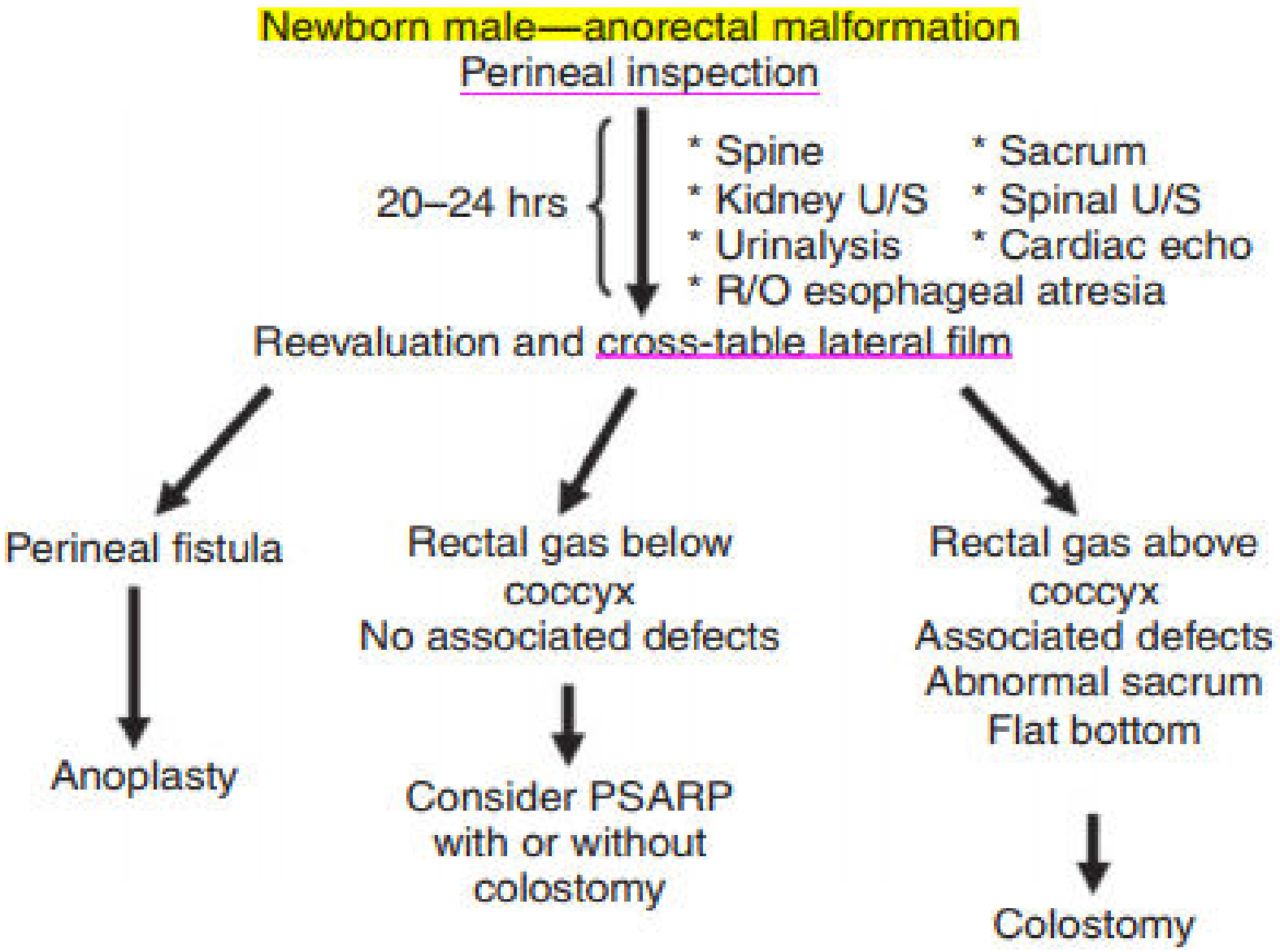




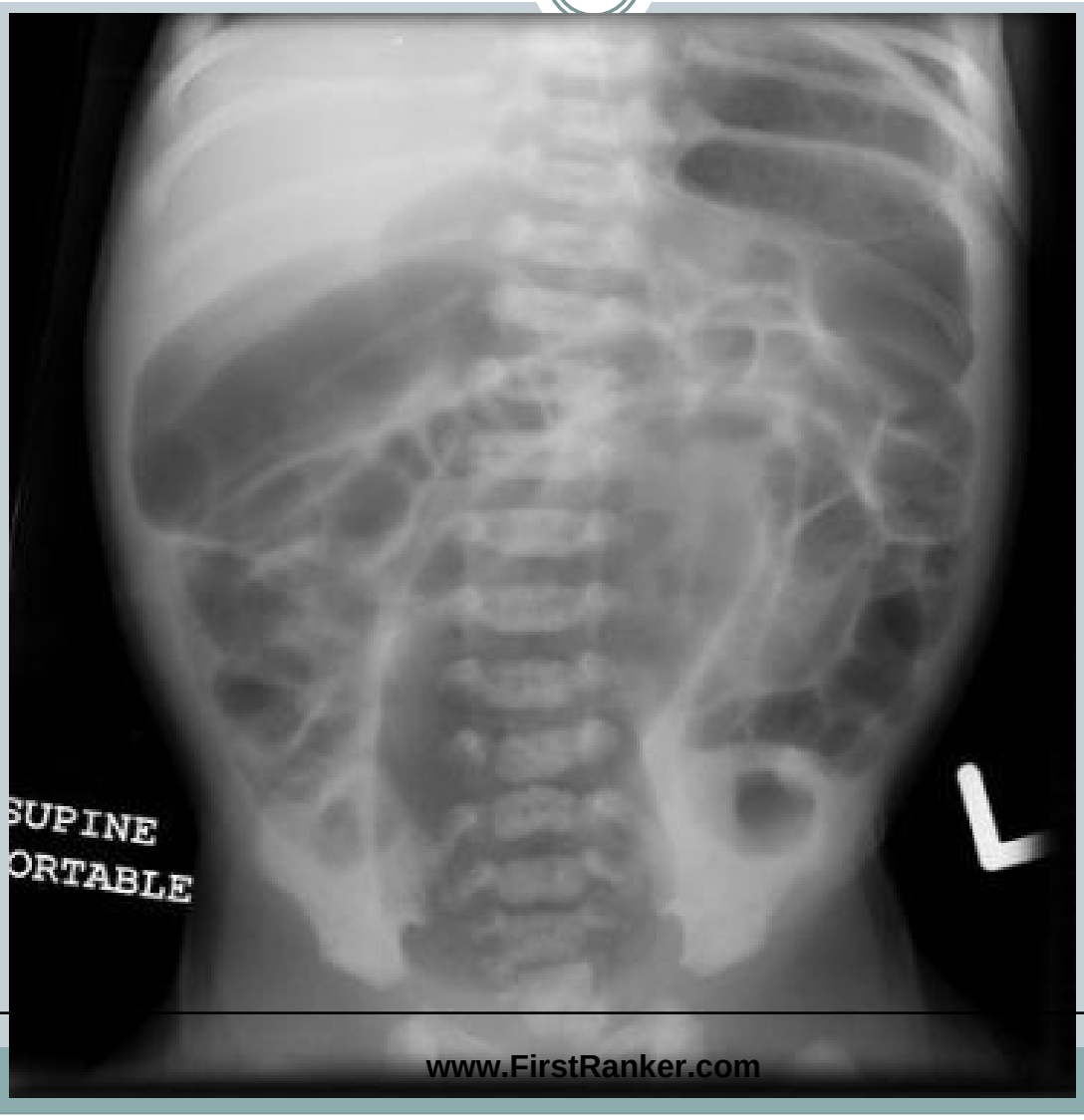
PERINEAL FISTULA WITH PROBE IN SITU



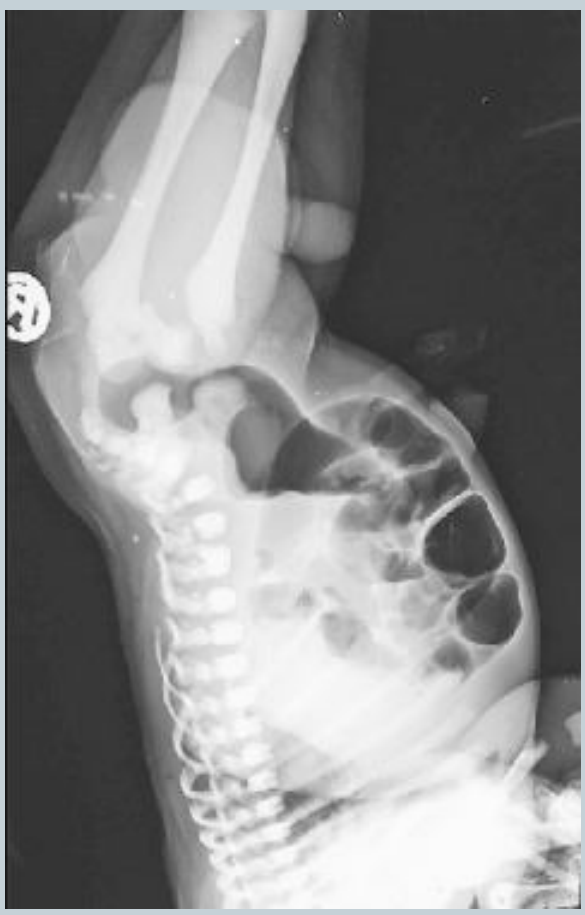
ALGORITHM FOR MALE ARM

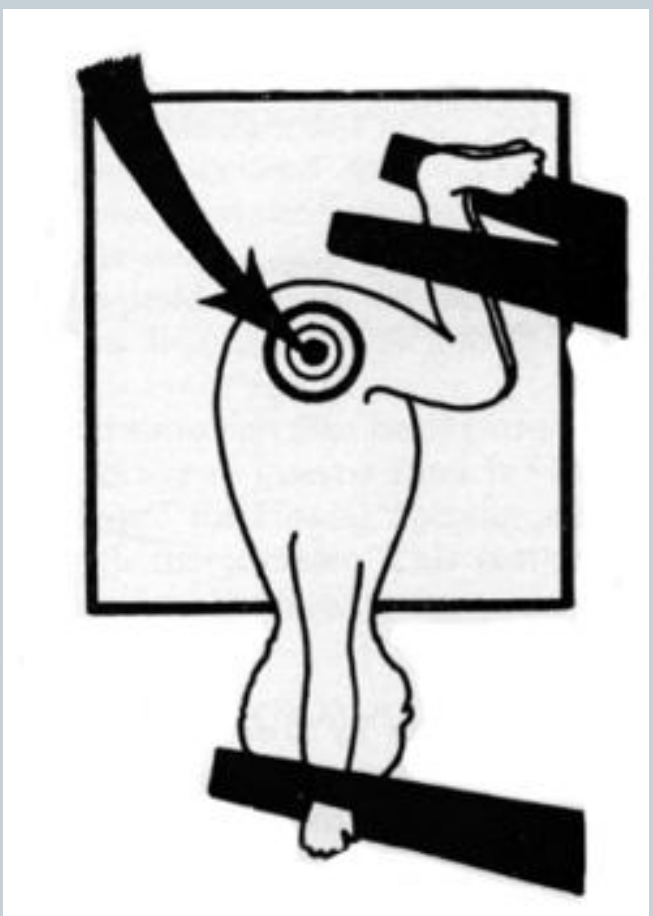


AXR



INVERTOGRAM





Prone Cross-Table Lateral View: An Alternative to the Invertogram in Imperforate Anus

K. L. Narasimharao[1]
G. R. Prasad[1]
S. Katariya[2]
K. Yadav[1]
S. K. Mitra[1]
I. C. Pathak[1]

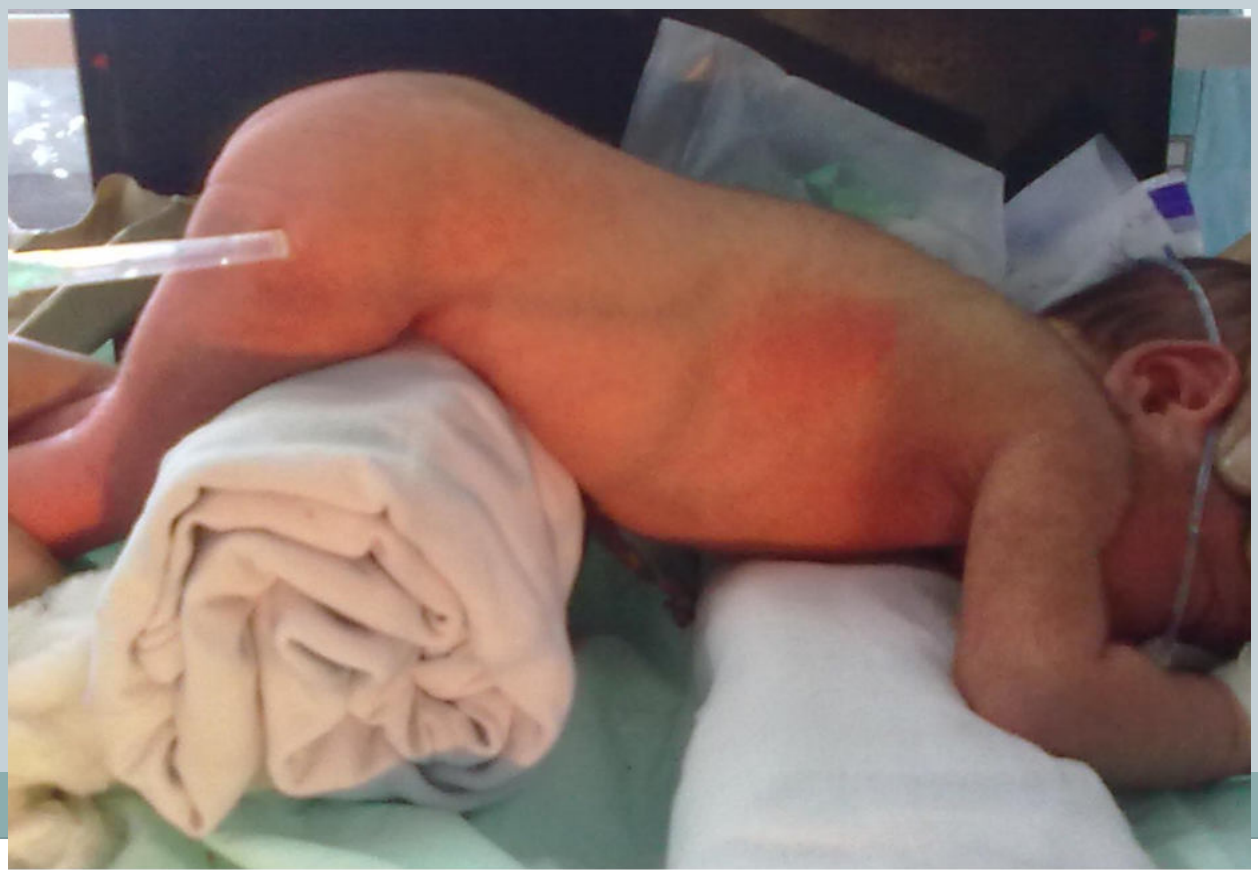
The prone cross-table lateral radiograph provides equal or sometimes better information, compared to the invertogram, for demonstration of the level of rectal atresia in neonates. Easy positioning, better cooperation of the patient, elimination of the effect of gravity, and better delineation of the rectal gas shadow are the advantages of the prone lateral view. Among the 45 cases compared, equal findings were noted in 37, but in eight babies the level of rectal atresia was more caudal in the prone radiograph than in the invertogram.

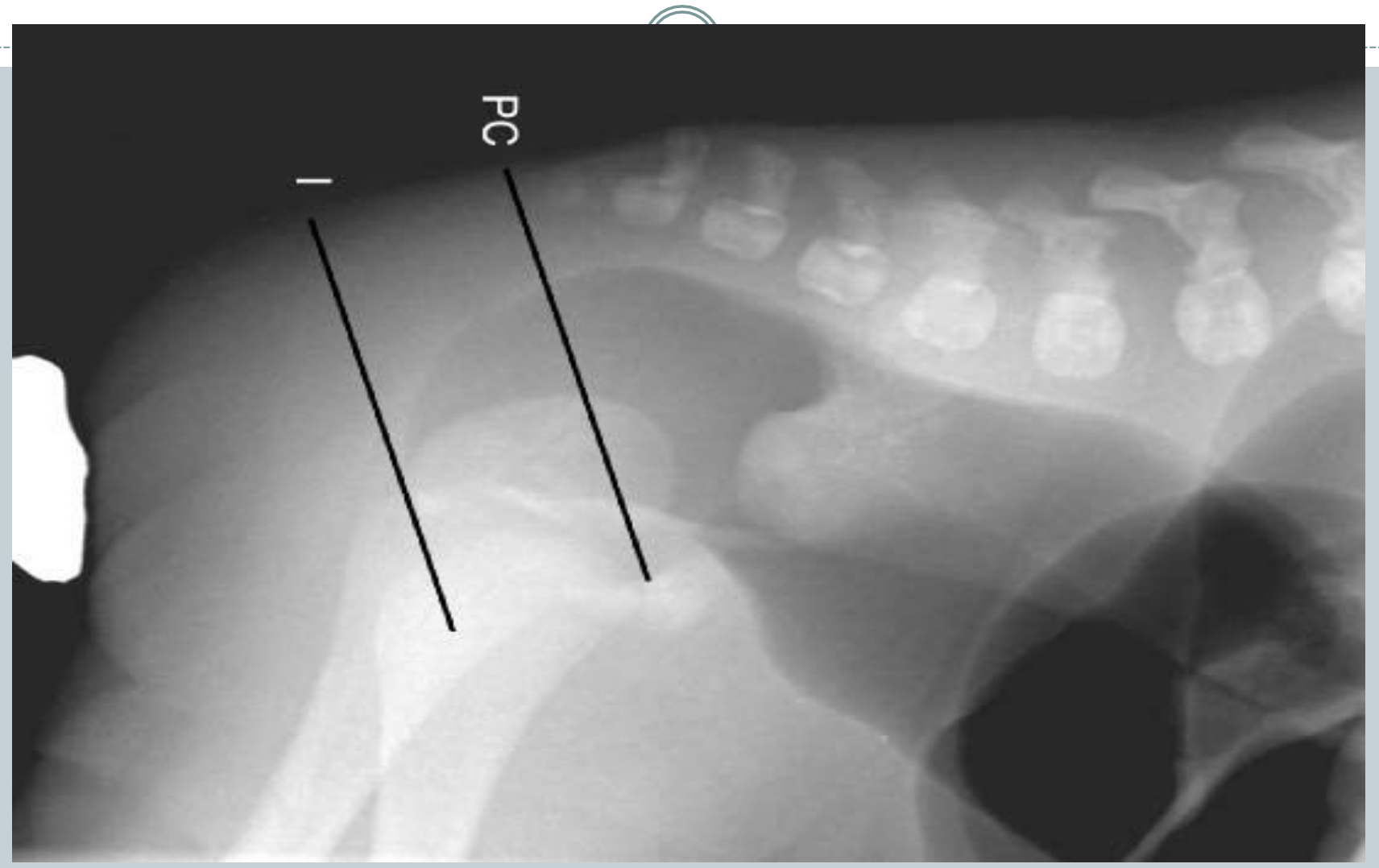
In the radiologic evaluation of the neonates with imperforate anus, the invertogram, first described by Wangensteen and Rice [1], has stood the test of time. The precautions in the technique and the pitfalls in its interpretation have been well documented [2, 3]. Since the rectum is the highest part of the bowel in the prone position [4], the evaluation of the rectal gas shadow in a prone cross-table lateral view should help in distinguishing the supralevator from the translevator lesions. Our study was conducted to test this hypothesis and to consider whether the prone lateral radiograph, which has certain advantages over the conventional invertogram, can replace the latter in the diagnosis of anorectal malformations.

*** Narsimharao KL et al. Am J Roentgenol 1983; 140: 227-229**

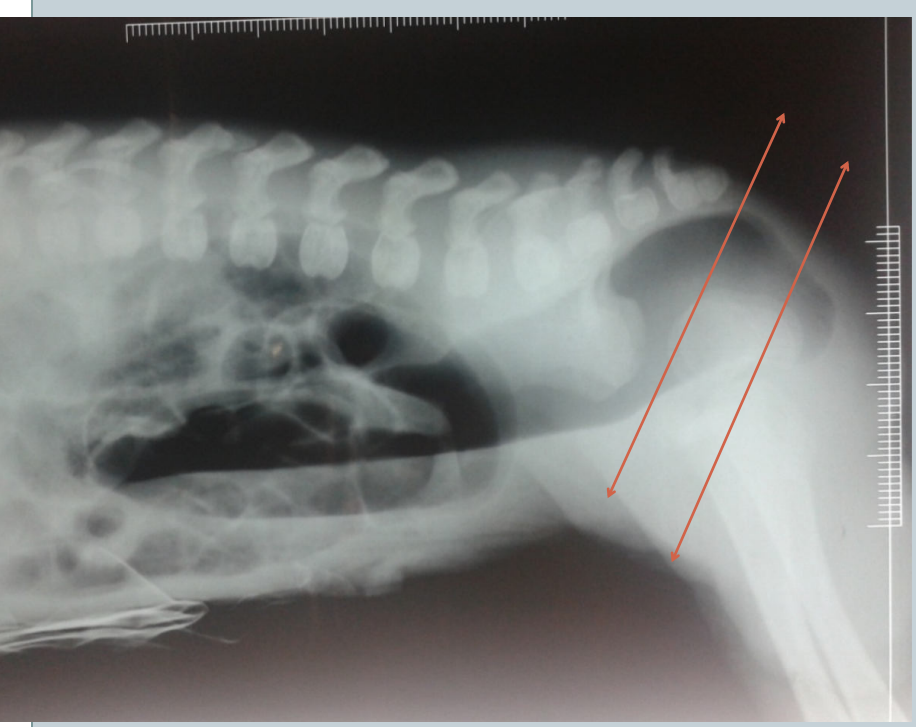
Cross-table Prone lateral x-ray (CTPL)

- 12–24 hrs after birth
- Prone position, hips- slightly flexed-3 mins
- Centering on the greater trochanter

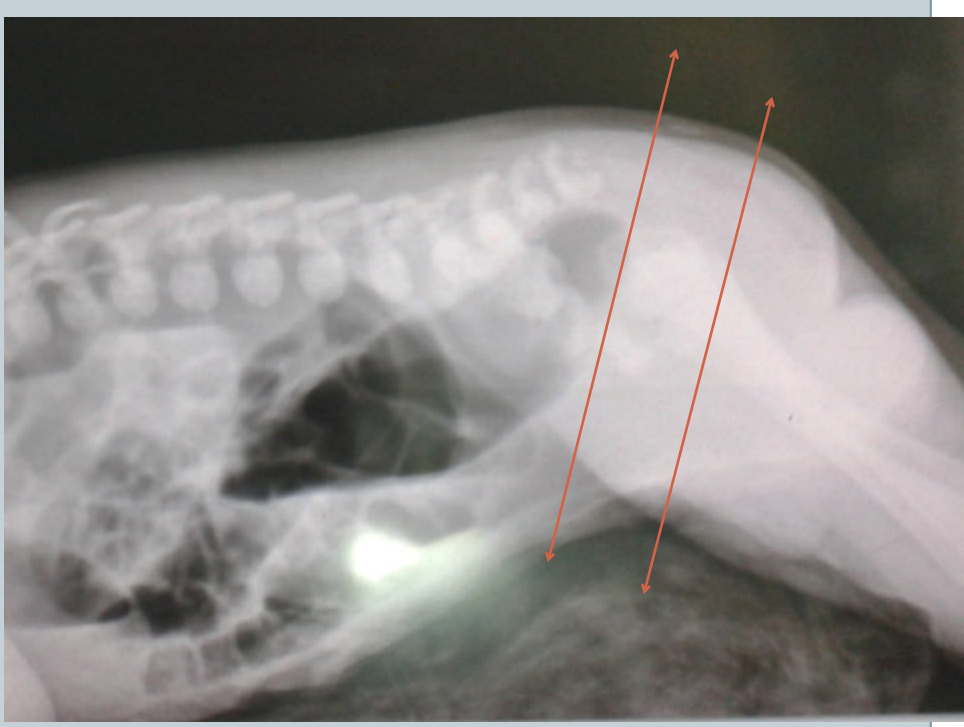




LOW



HIGH



ALGORITHM FOR MALE ARM

Newborn male—anorectal malformation

Perineal inspection

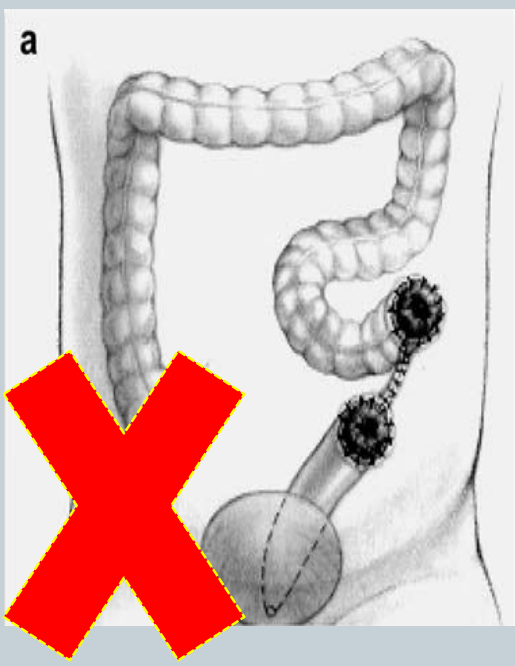
20–24 hrs

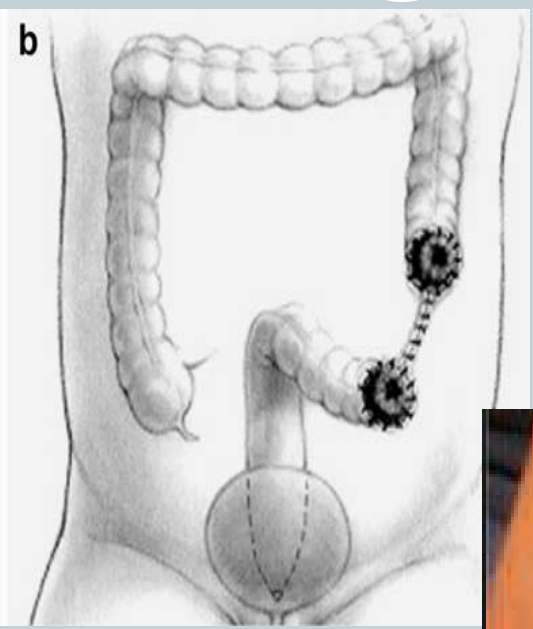
* Spine
* Kidney U/S
* Urinalysis
* R/O esophageal atresia
* Sacrum
* Spinal U/S
* Cardiac echo

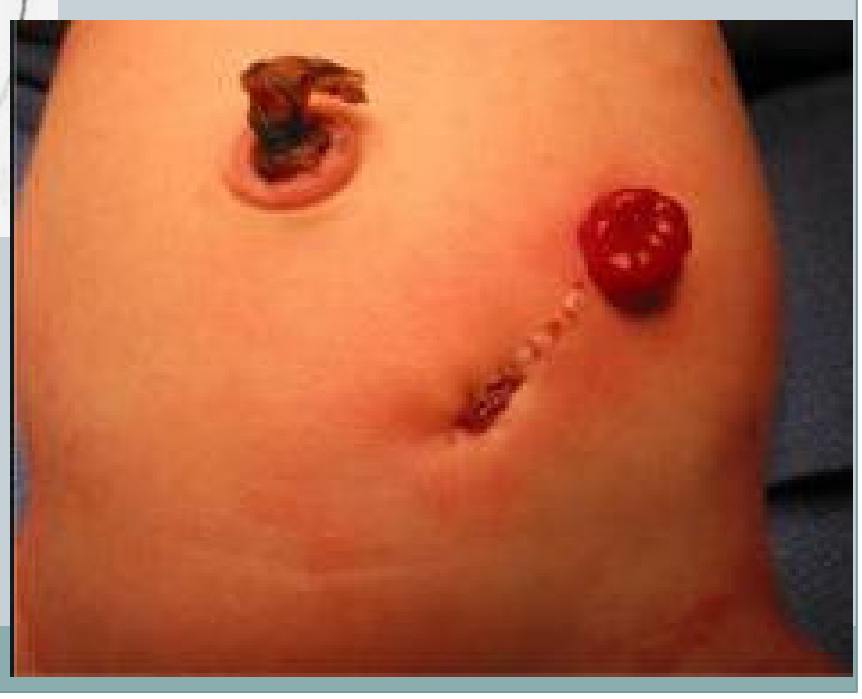
Reevaluation and cross-table lateral film

- Perineal fistula → Anoplasty
- Rectal gas below coccyx, No associated defects → Consider PSARP with or without colostomy
- Rectal gas above coccyx, Associated defects, Abnormal sacrum, Flat bottom → Colostomy

COLOSTOMY



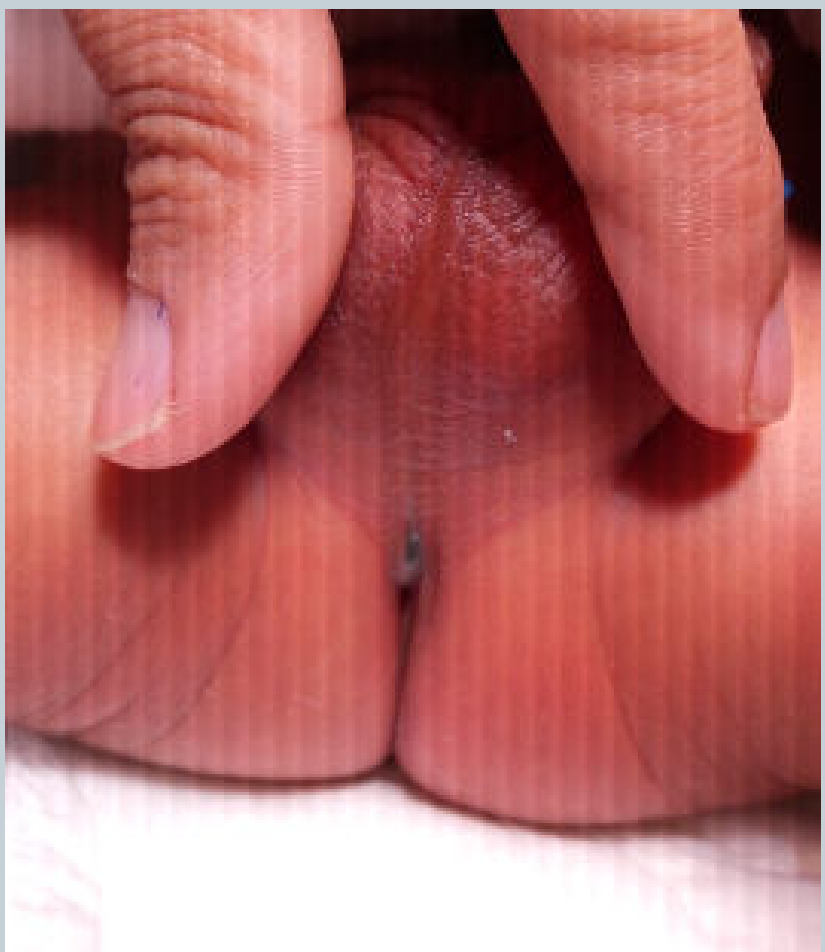


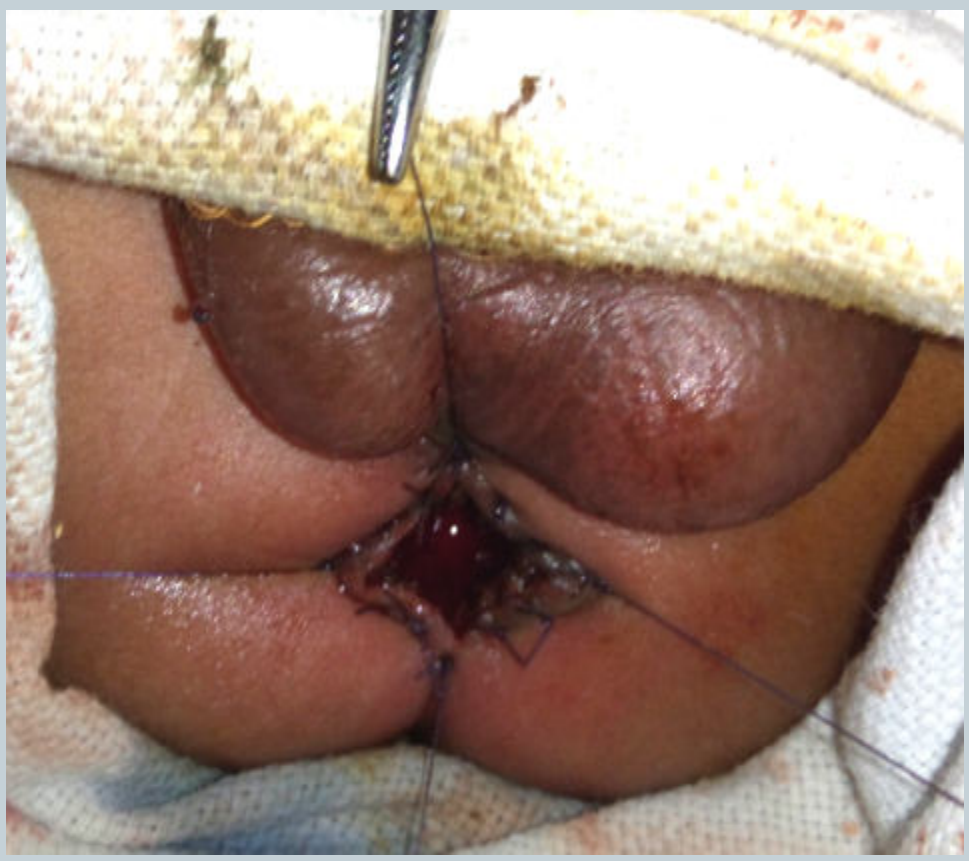


HIGH DIVIDED SIGMOID COLOSTOMY (HDSC)

- Bowel decompression and protection for the final repair
- Facilitates the distal colostogram
- Relatively short segment of defunctionalized distal colon- less chances of microcolon
- Mechanical cleansing
- Large rectourethral fistula- Less metabolic acidosis
- Doesn't permit stool distally
- Easy pull through

ANOPLASTY



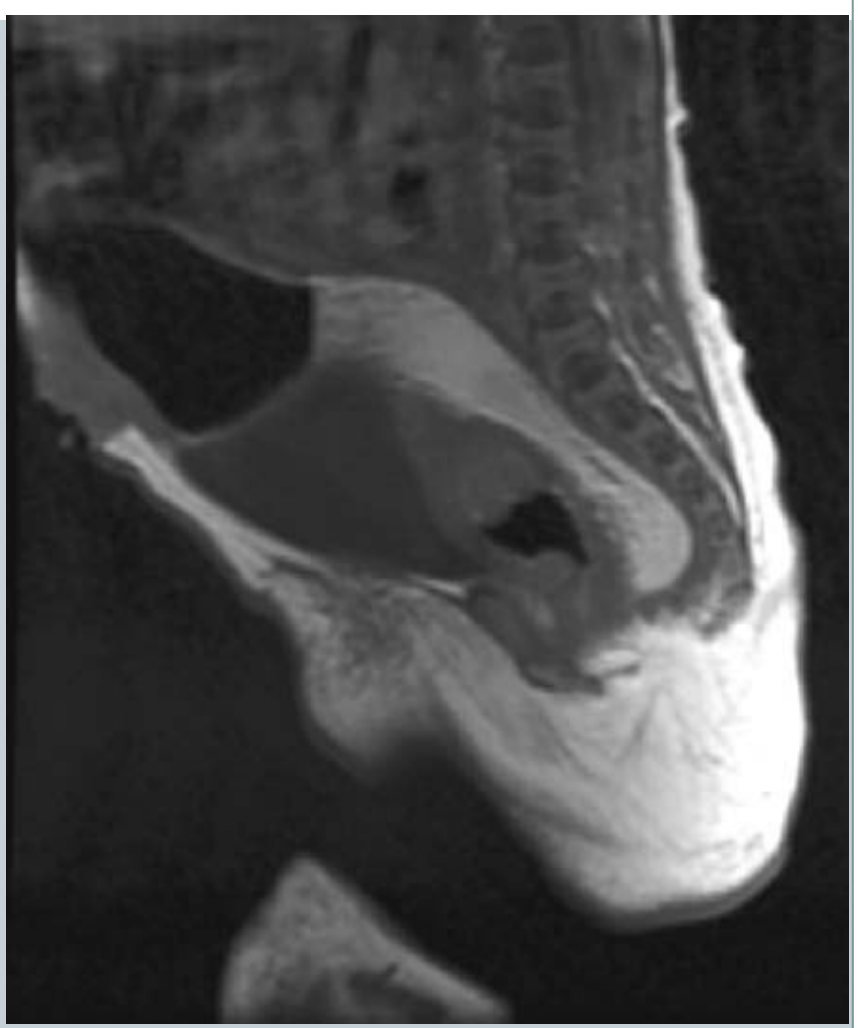


Computed Tomography

- Demonstrate the level of the terminal colon
- Increased detail - osseous structures musculature.
- Difficulty in distinguishing meconium from the rectal wall and adjacent musculature- and so the site of fistula.
- Radiation cost to the patient.

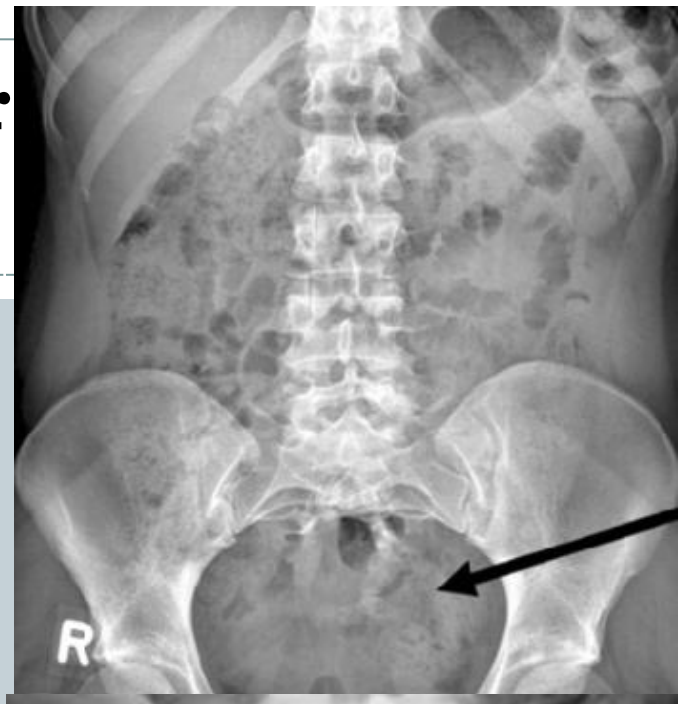
MRI

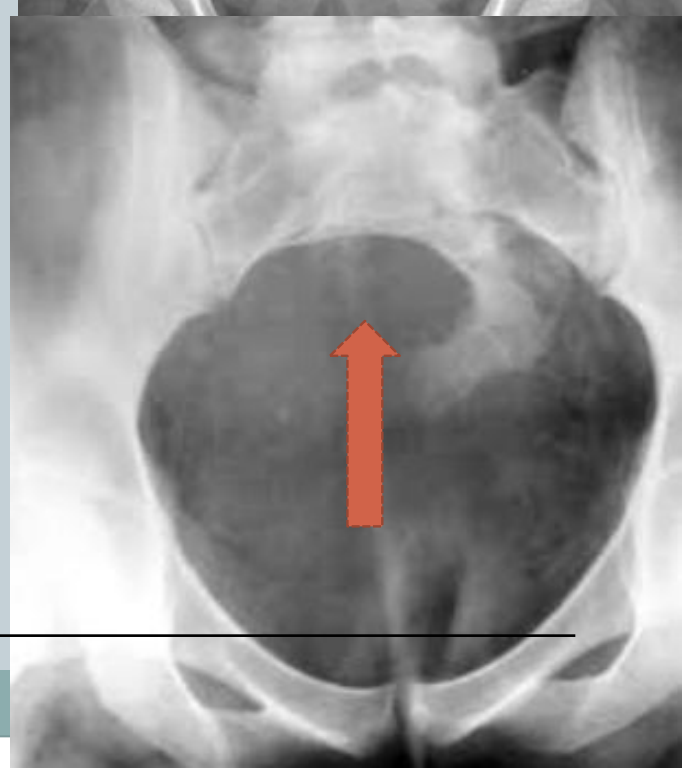
- No radiation burden
- Easy differentiation of meconium from the rectal wall and levator musculature
- Level of the terminal bowel, and the state of the pelvic floor musculature
- Fistulae (difficult in neonates)



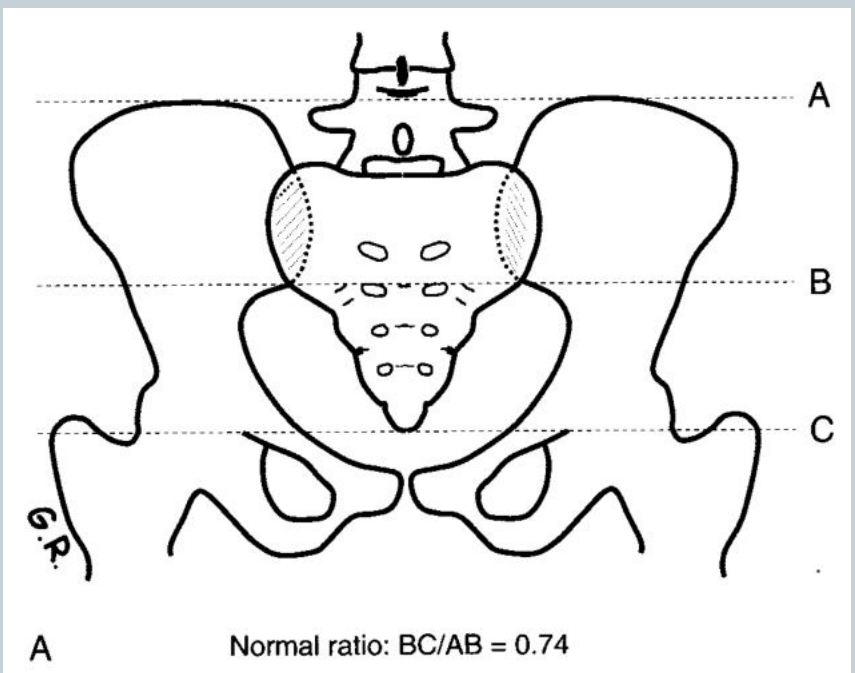
II. Assessment of the Sphincter complex and Nerve supply

- Absent S4 AND S5- **Normal** variant
- Absent S3,4,5 - **Variable incontinence**
- Absent S1 AND S2 - **always incontinent**
- Hemisacral defects- Results unpredictable
- Meningomyeloceles.

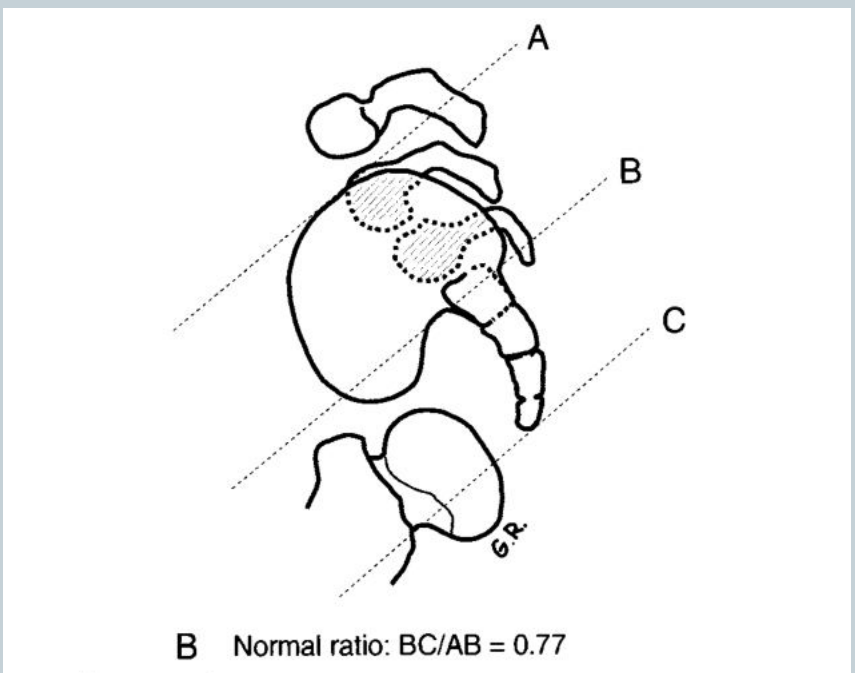




- The sacral ratio is a valuable prognostic tool
- It quantifies degree of sacral agenesis
- Patient with ratio < 0.5 are universally incontinent
- Ratio that approach 1 usually predict a good prognosis



A Normal ratio: BC/AB = 0.74



B Normal ratio: BC/AB = 0.77

KRICKENBECK SURGICAL PROCEDURES

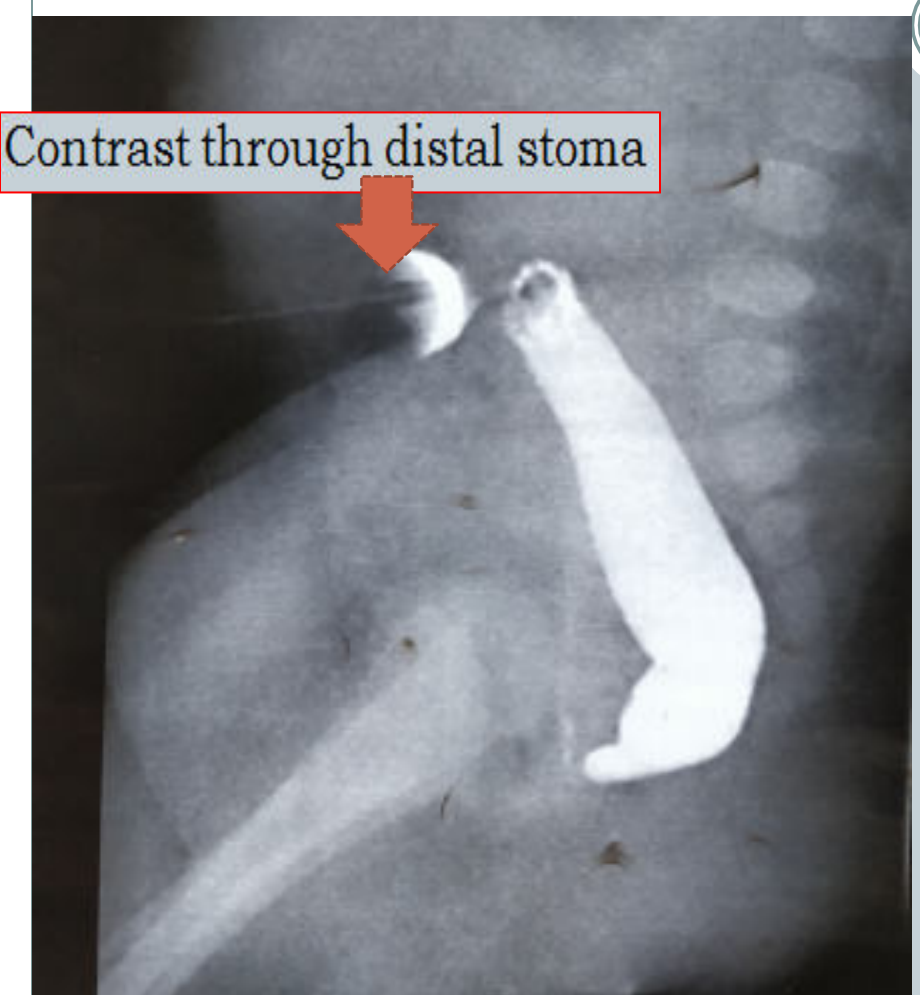
Operative procedures

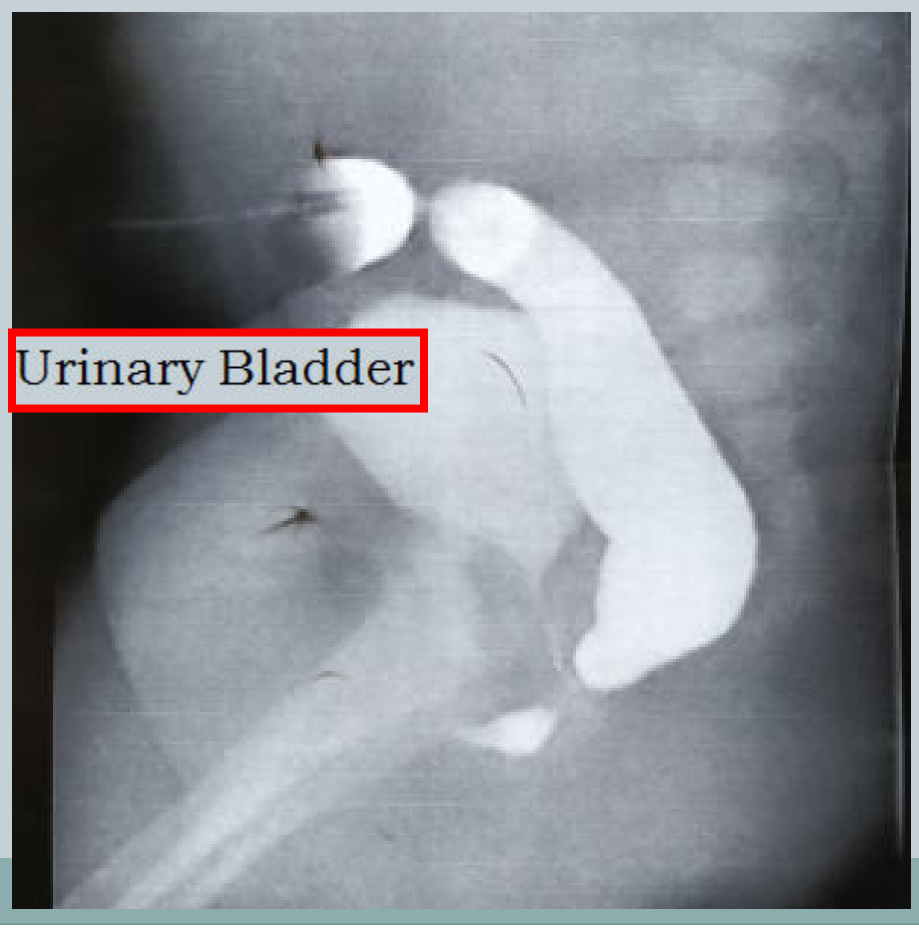
- Perineal operation
- Anterior sagittal approach
- Sacroperineal procedure
- PSARP
- Abdominosacroperineal pull-through
- Abdominoperineal pull-through
- Laparoscopic-assisted pull-through

Associated conditions

- Sacral anomalies
- Tetherd cord

Pressure-augmented colostogram



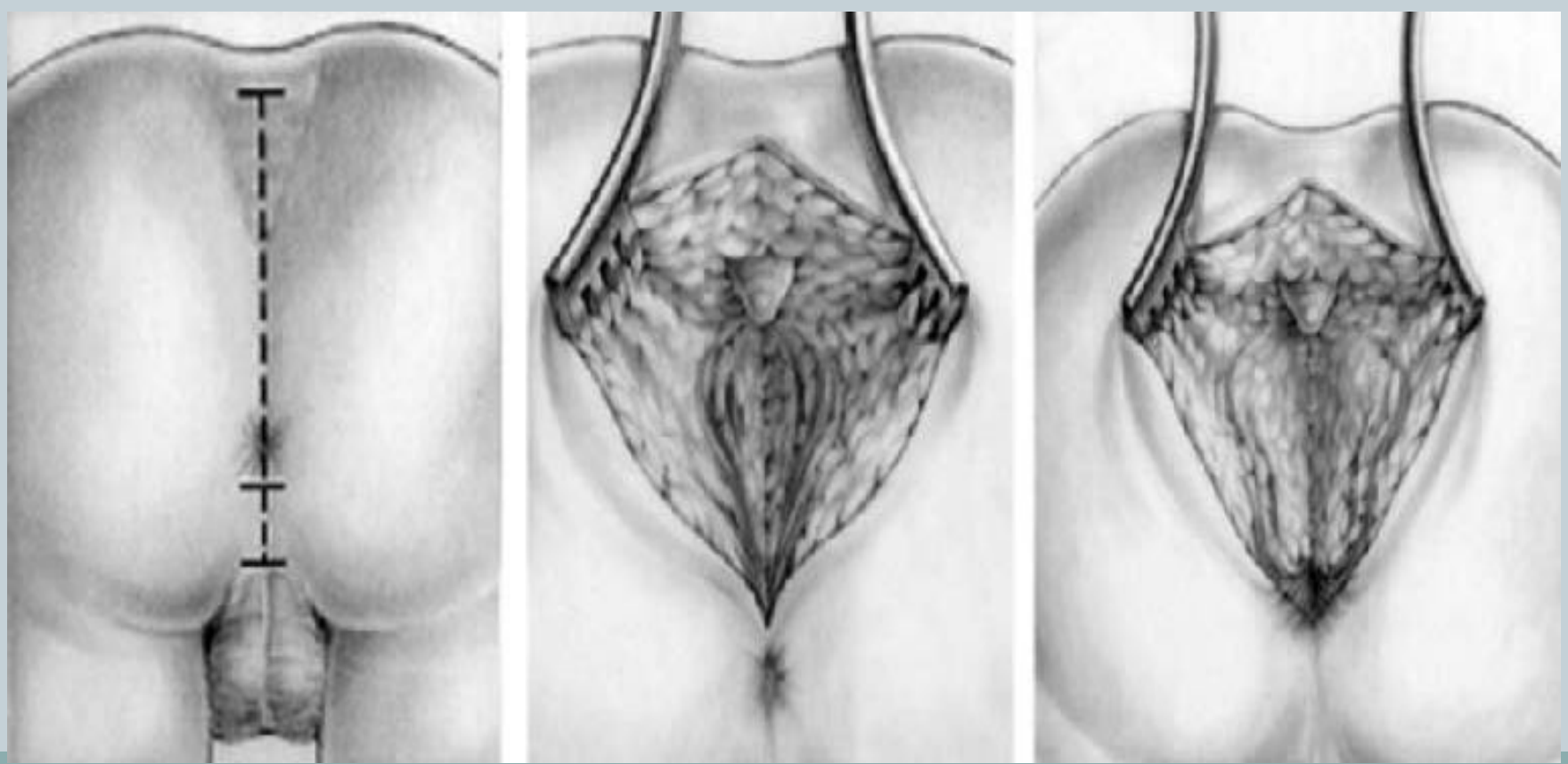


Posterior sagittal Anorectoplasty (PSARP)

- Jack knife position (Prone)

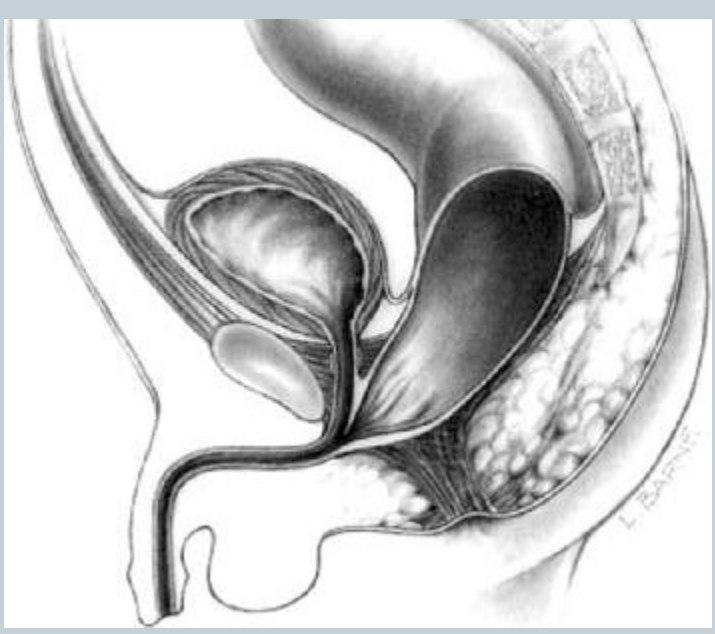
PSARP

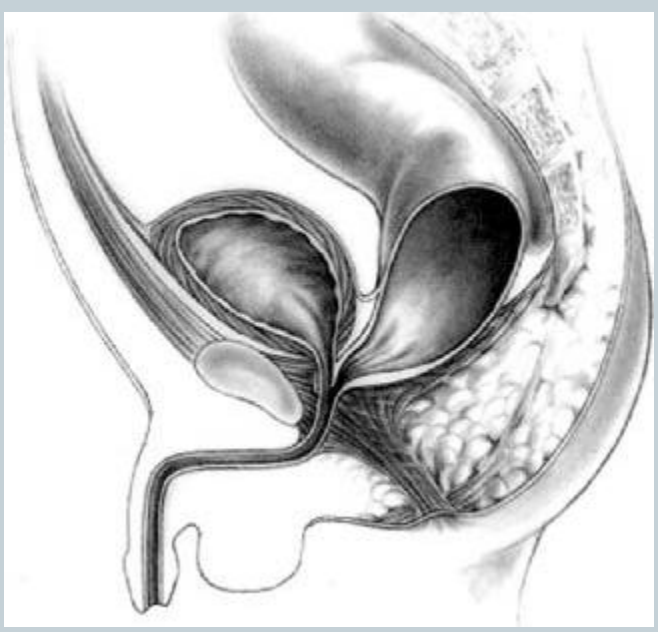
- Electrical stimulator- sphincter location.



Rectourethral fistulas

Rectourethrobulbar fistula	Rectourethroprostatic fistula





-Most frequent defect in male patients

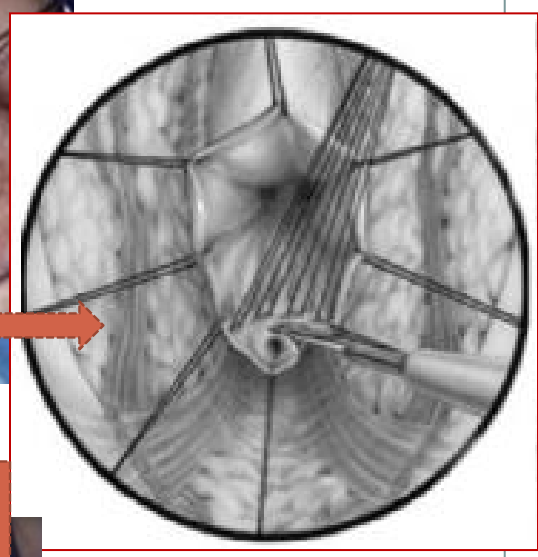
-Rectum identified and opened posteriorly in between stay sutures

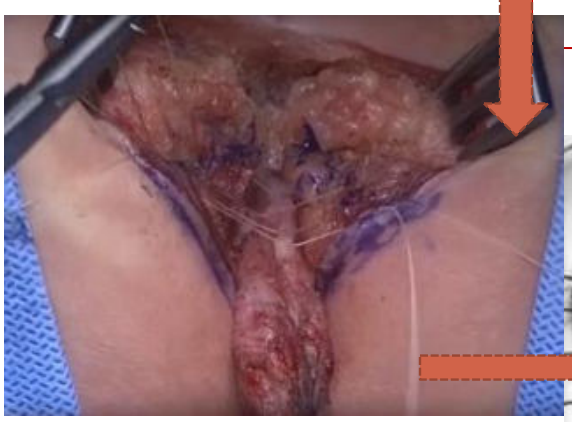
-Fistula visualized

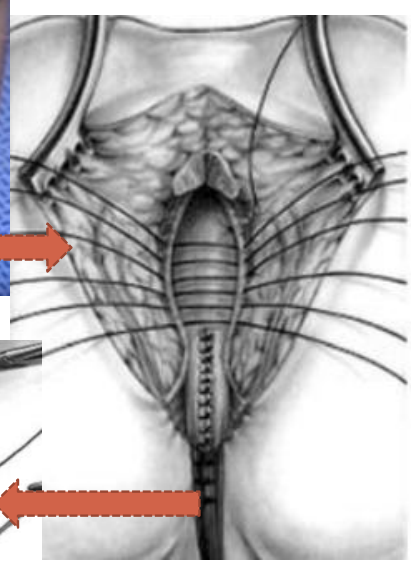
-Rectum separated from the urethra, and mobilised.

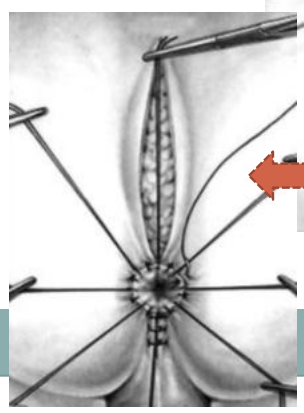
-Rectum passed in front of levator and within sphincter.

-Anoplasty done.



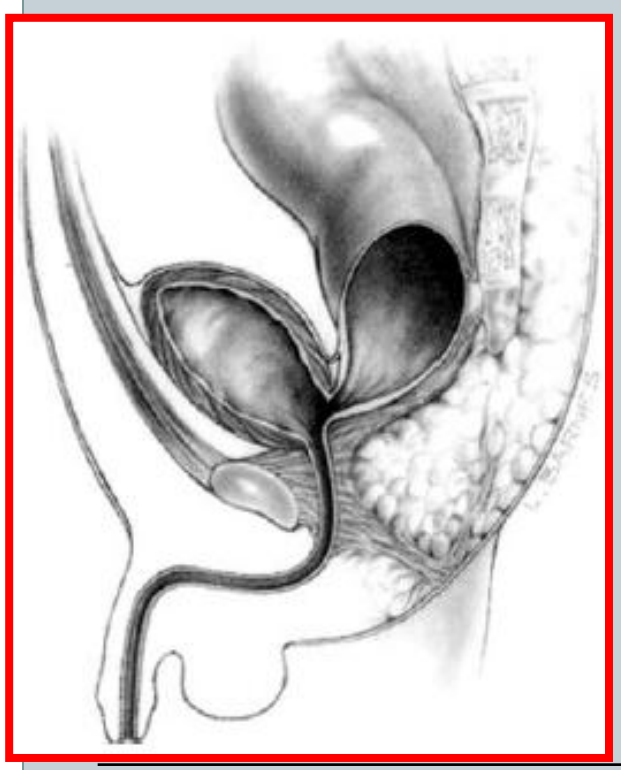


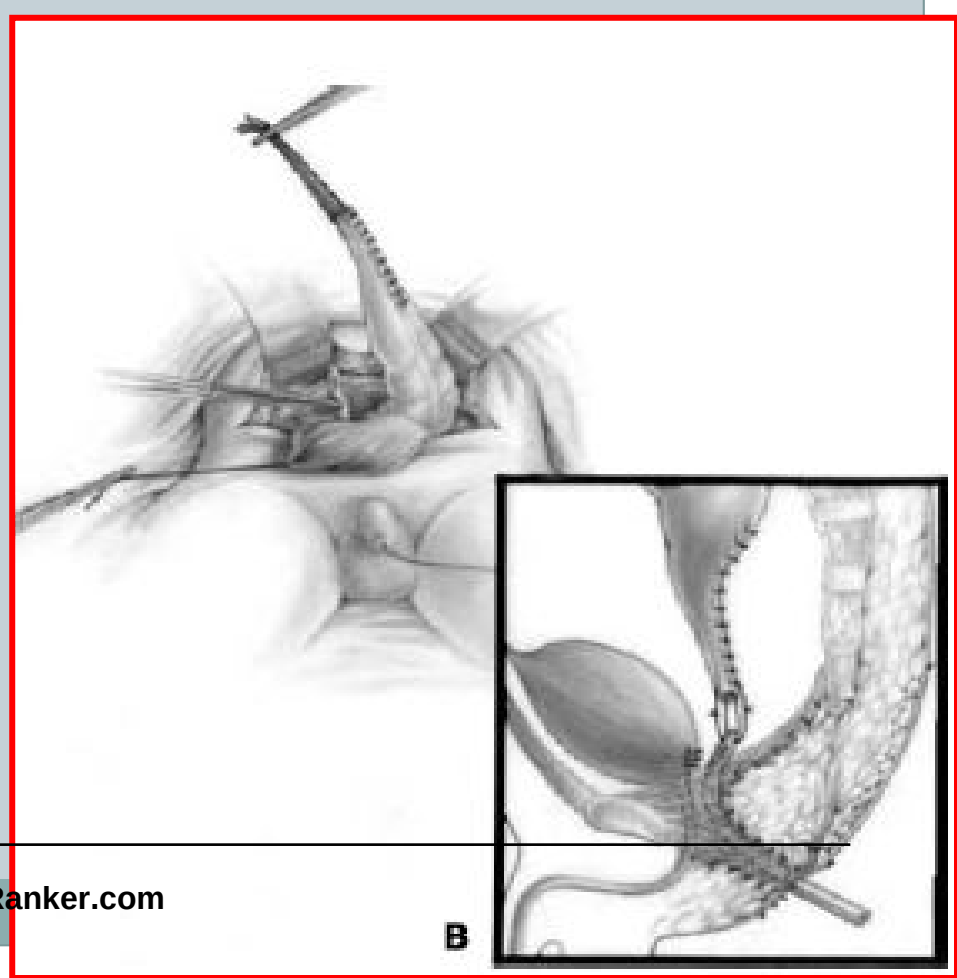




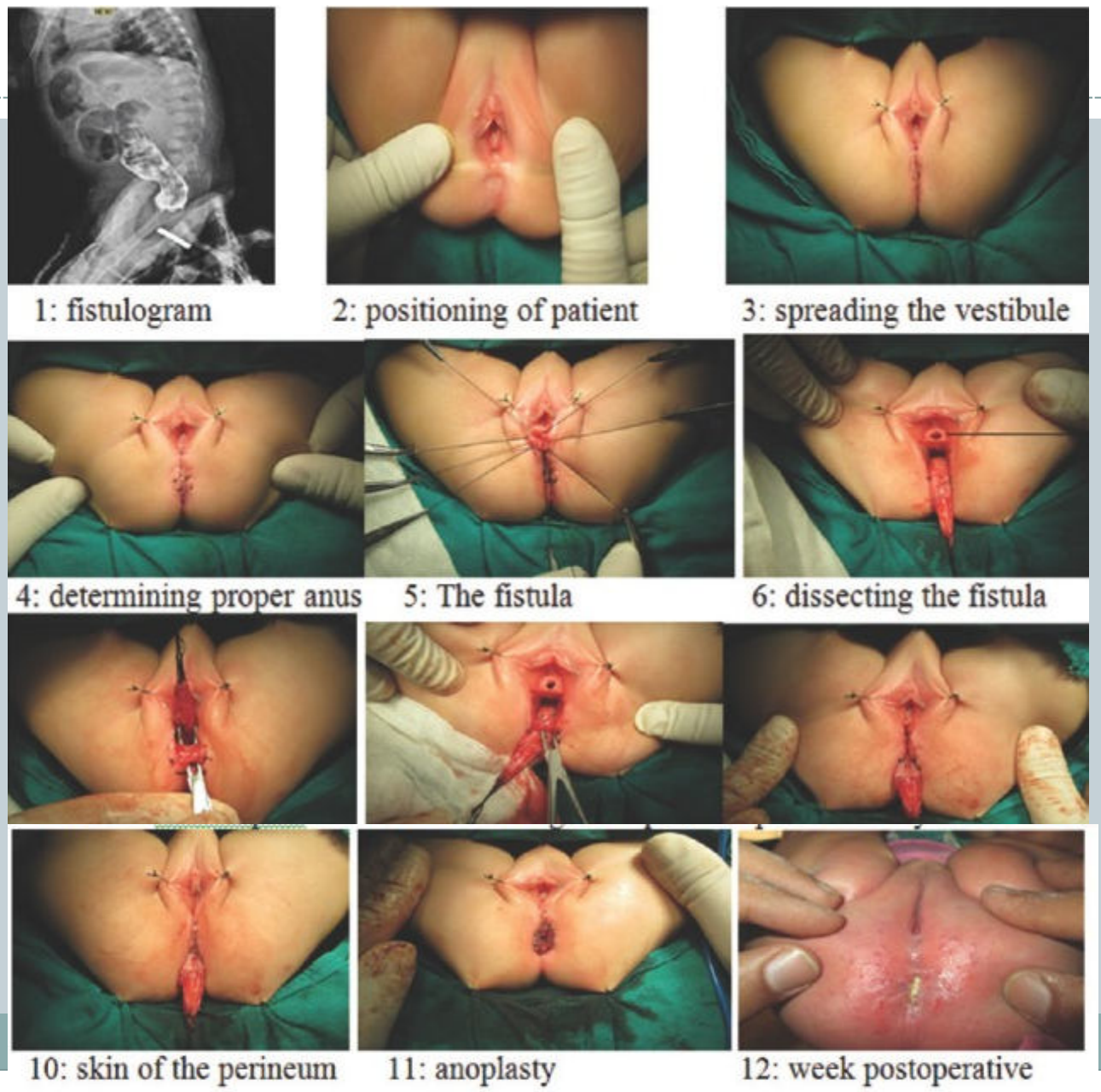
Recto-bladder neck fistula

- Combined abdomino-perineal approach
- Poor prognosis





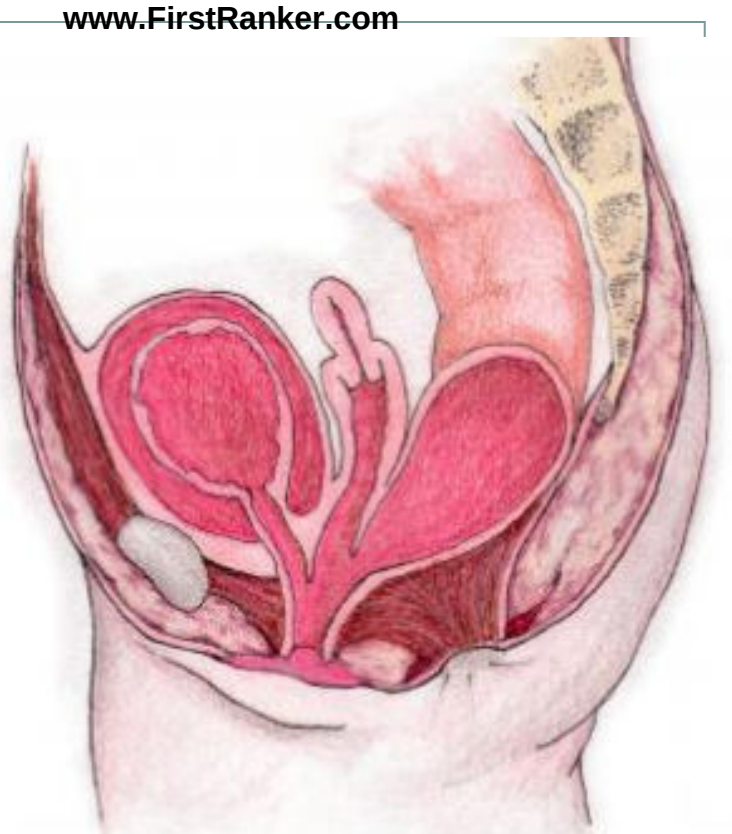
ANTERIOR SAGITTAL ANORECTOPLASTY(ASARP)

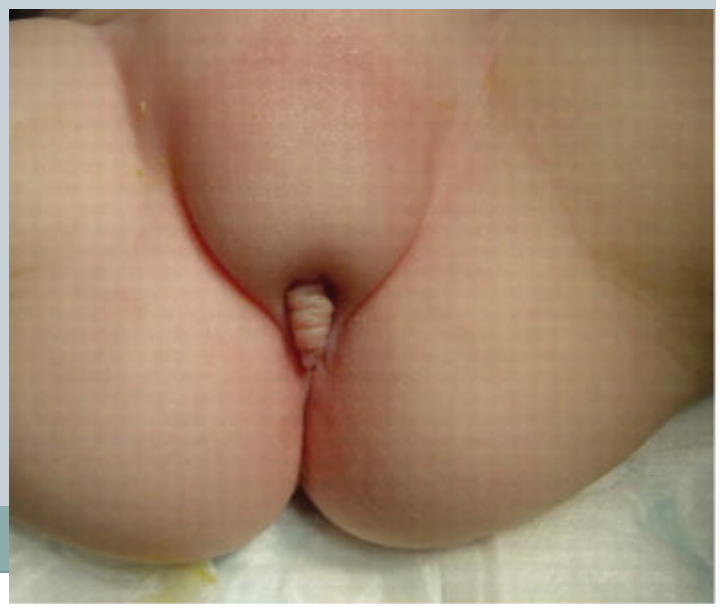


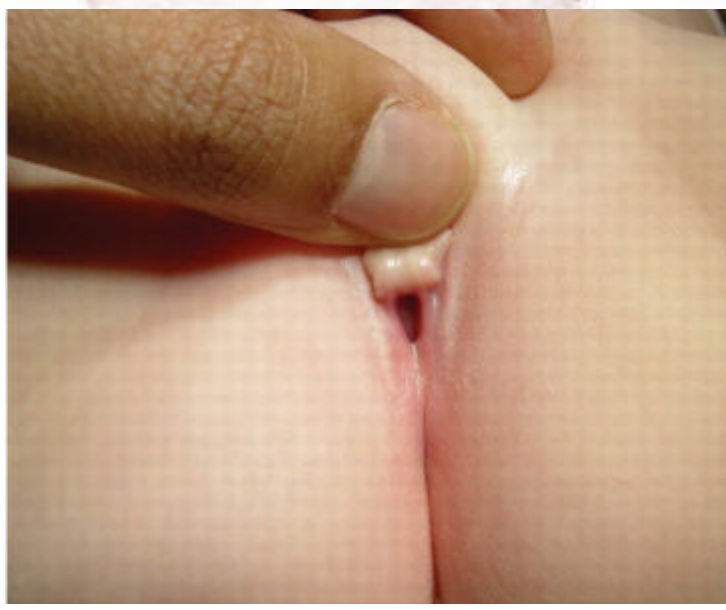
ARM VARIANTS

I.PERSISTENT CLOACA

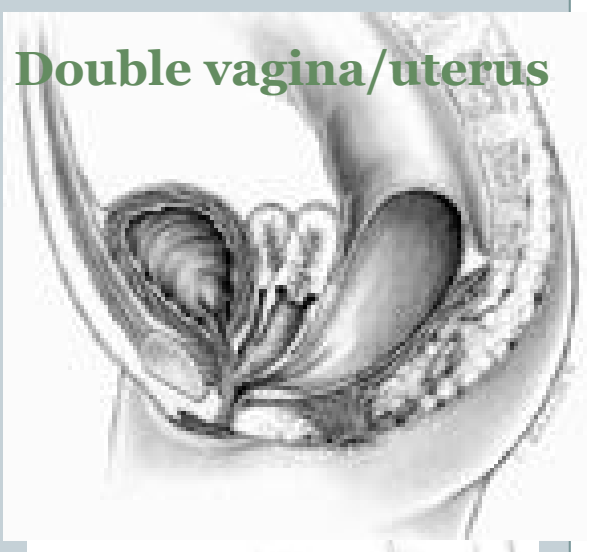
- Defect in which the rectum, one or two vaginas and the urinary tract converge into one common channel.

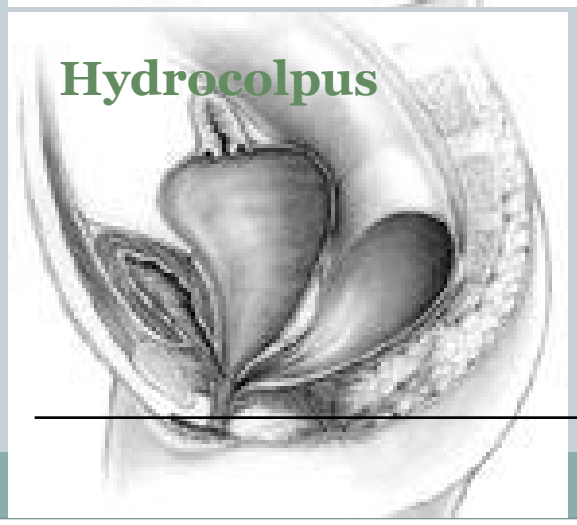


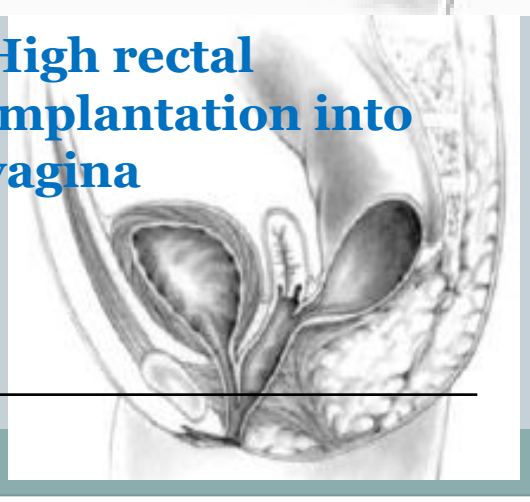




Cloacal anomalies

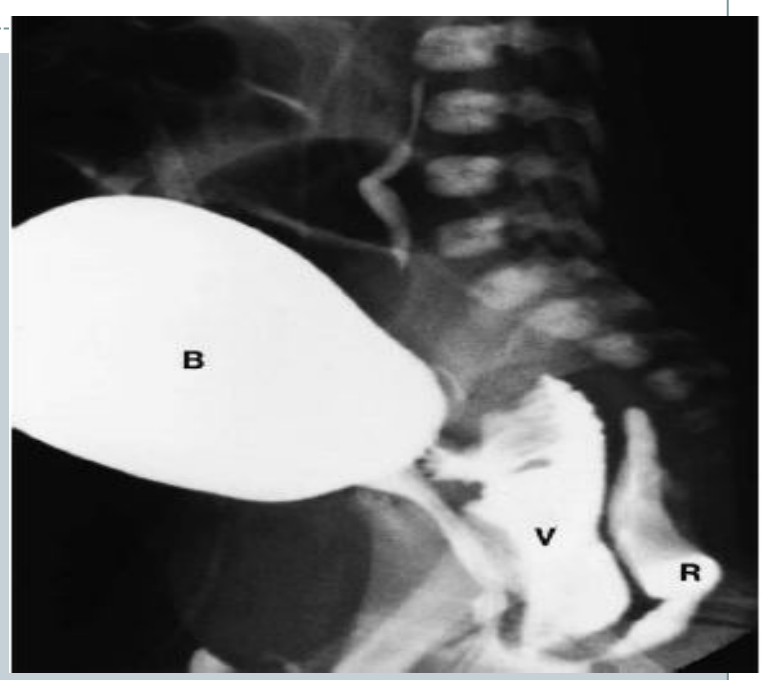


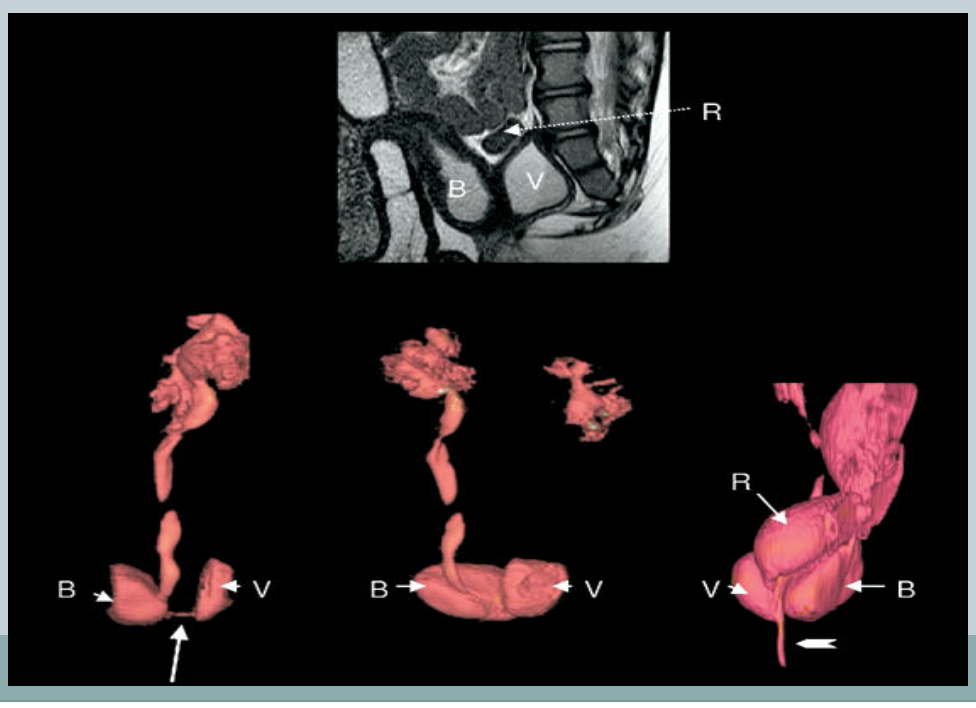




DEFINITIVE MANAGEMENT

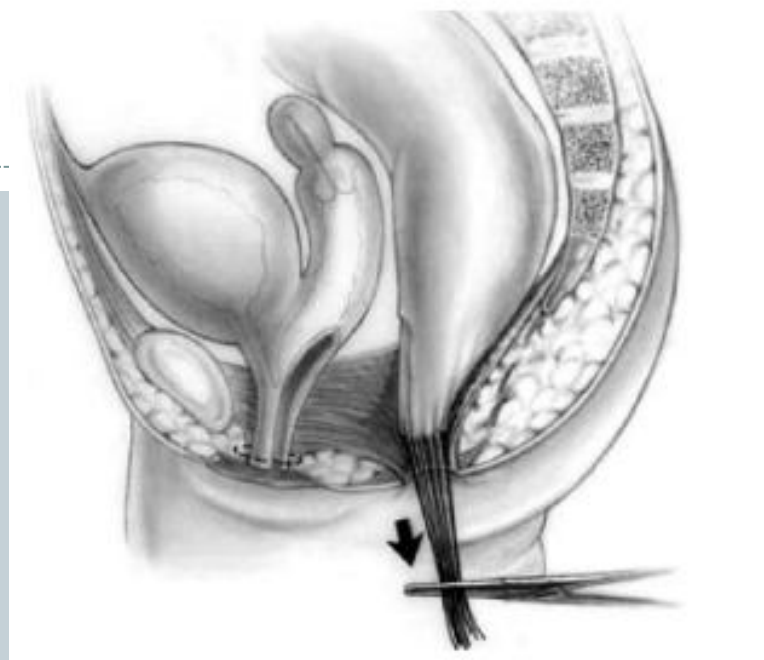
- Genitogram
- Genitoscopy
- 3D MR Genitography



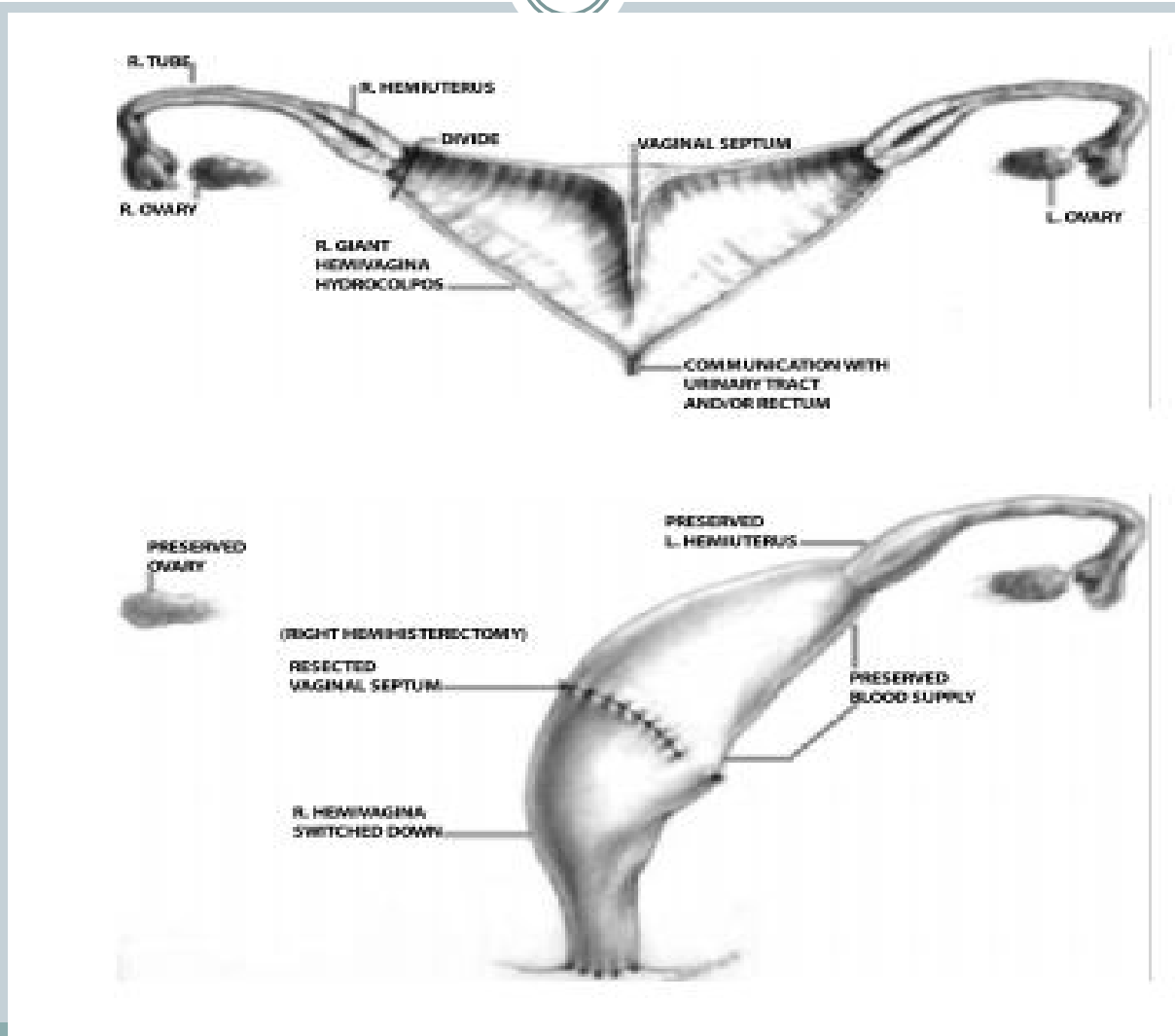


SURGERY

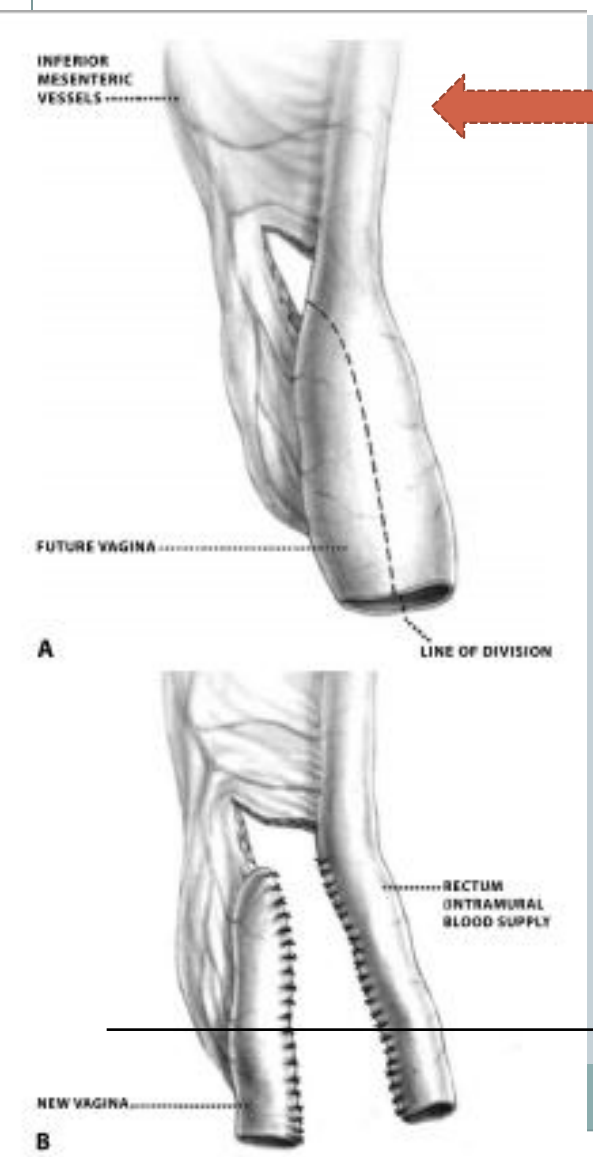
- Posterior sagittal approach
- Rectal mobilization (sub serosally)
- Total Uro Genital mobilization
- Vulval & perineal reconstruction
- Abdomino- perineal approach if long common channel.



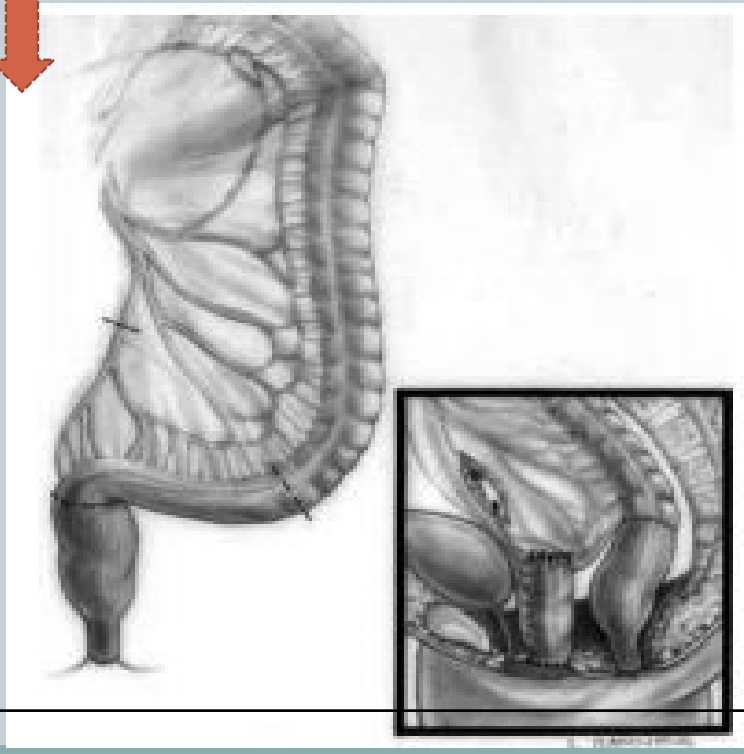
VAGINAL SWITCH



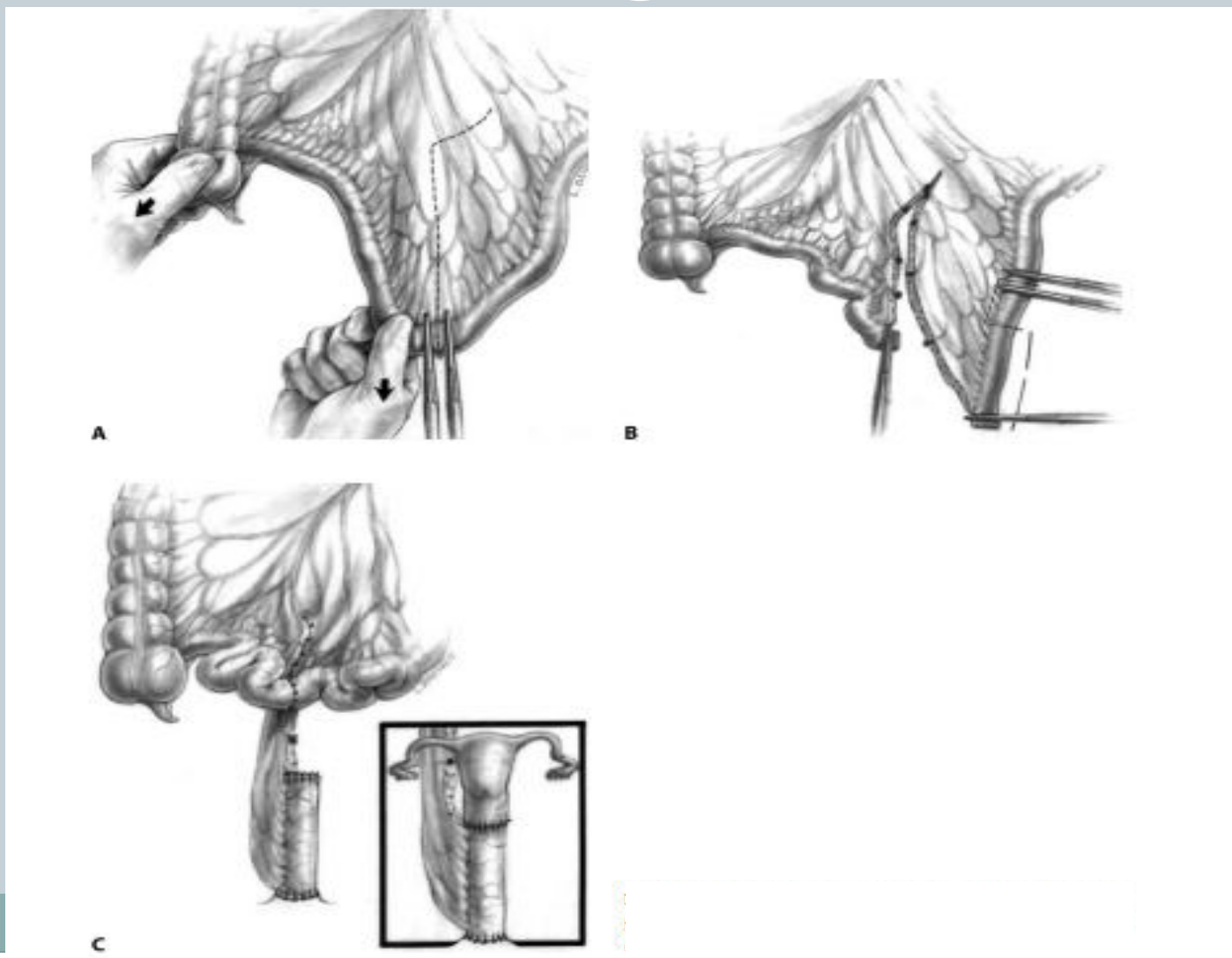
VAGINAL AUGUMENTATION



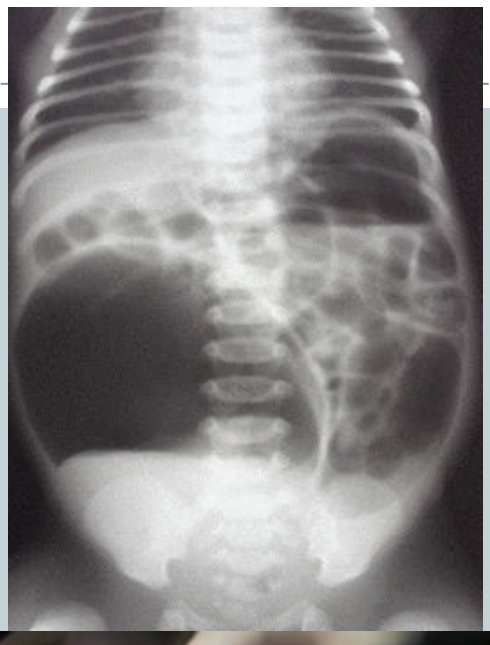
- From rectum
- From colon



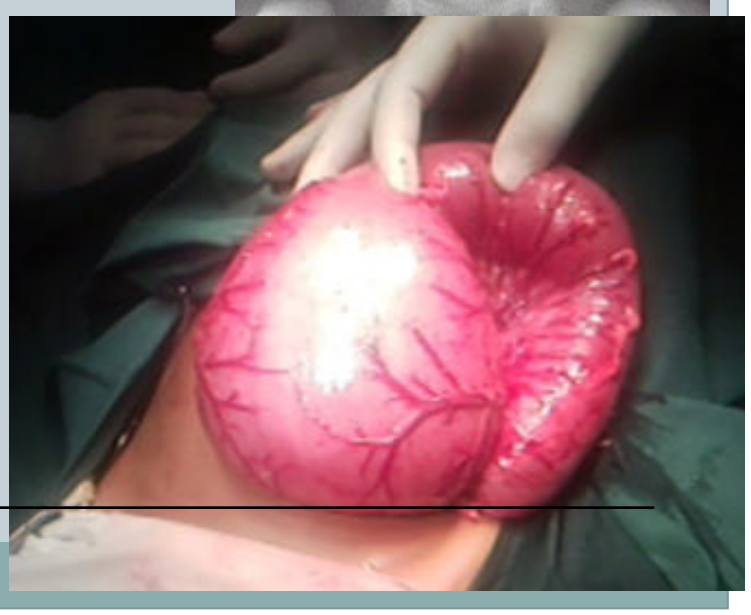
From small intestine



2. CONGENITAL POUCH COLON

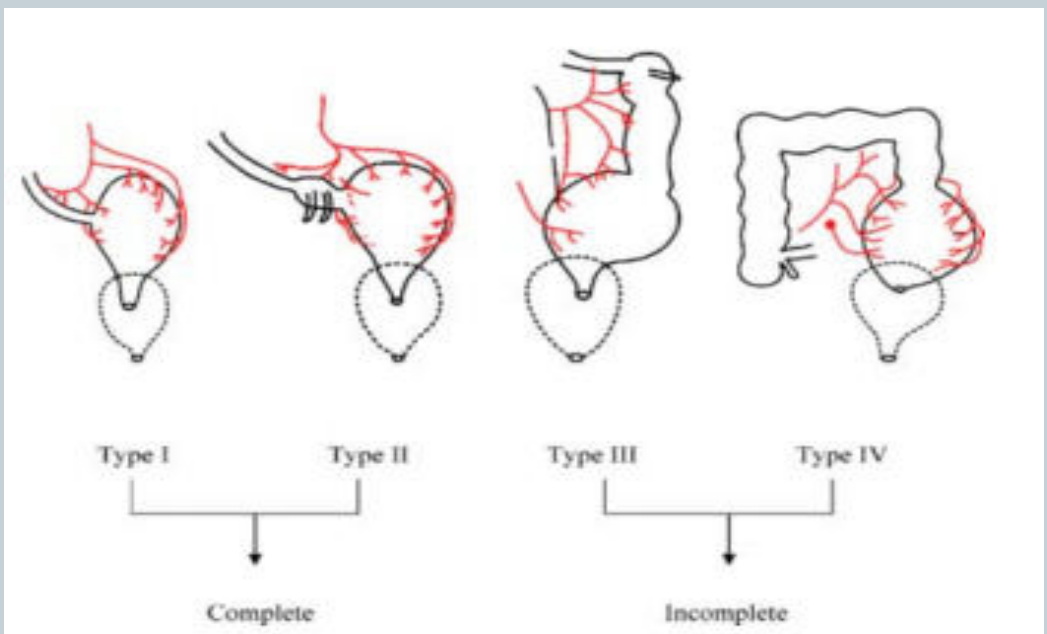


- 2.5 – 9 % of ARM in North India.
- All or part of the colon is replaced by a pouch-like dilatation, which communicates distally with the urogenital tract via a large fistula.



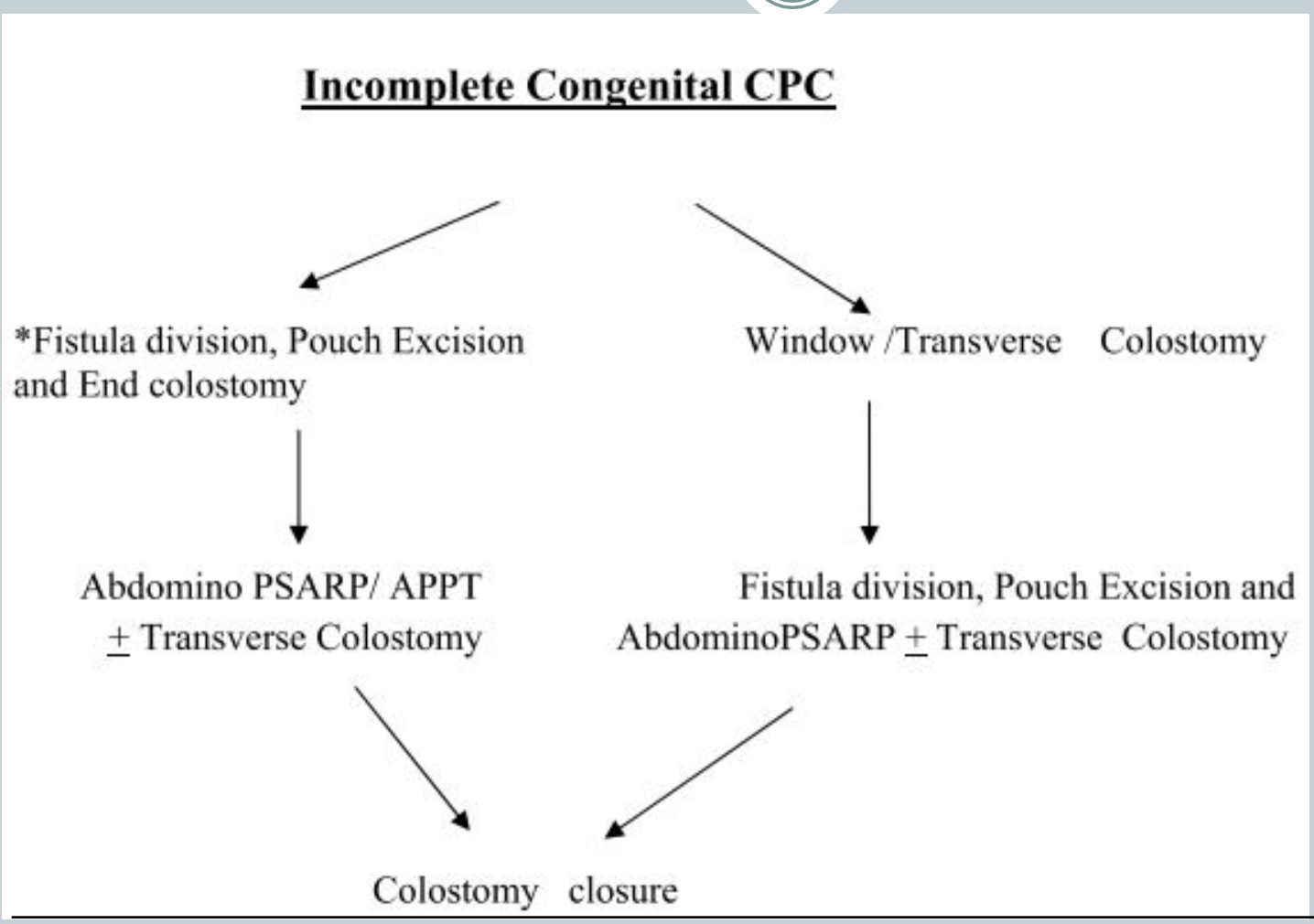
MODIFIED WAKHLU'S CLASSIFICATION

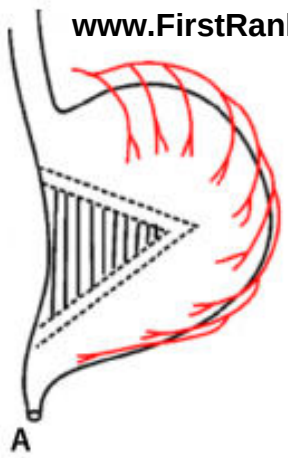
- Depending on length of available colon

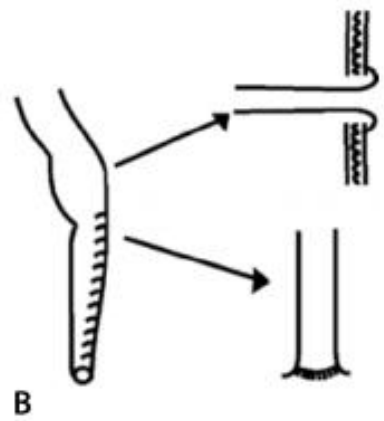


• **Wakhlu AK, Tandon RK, Kalra R (1982) Short colon with anorectal malformation. Indian J Surg 44:621–629.**
• **Narsimha Rao KL, Yadav K, Mitra SK, Pathak IG (1984) . Congenital short colon with imperforate anus (CPC syndrome). Ann Pediatr Surg 1:159**

MANAGEMENT







Complete Congenital CPC

*Fistula division, Coloplasty and End colostomy

Window Colostomy / Proximal ileostomy

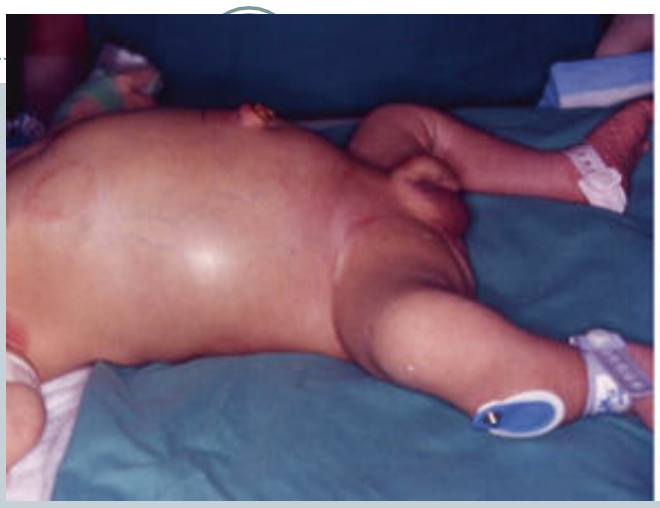
Abdomino PSARP/ APPT ± Proximal ileostomy

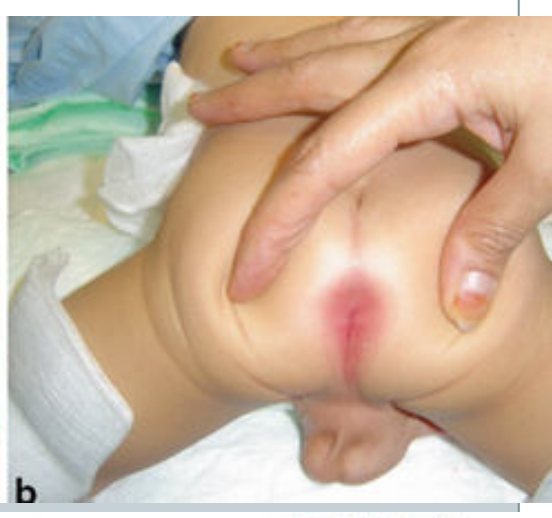
Fistula division, Coloplasty and AbdominoPSARP ± Proximal ileostomy

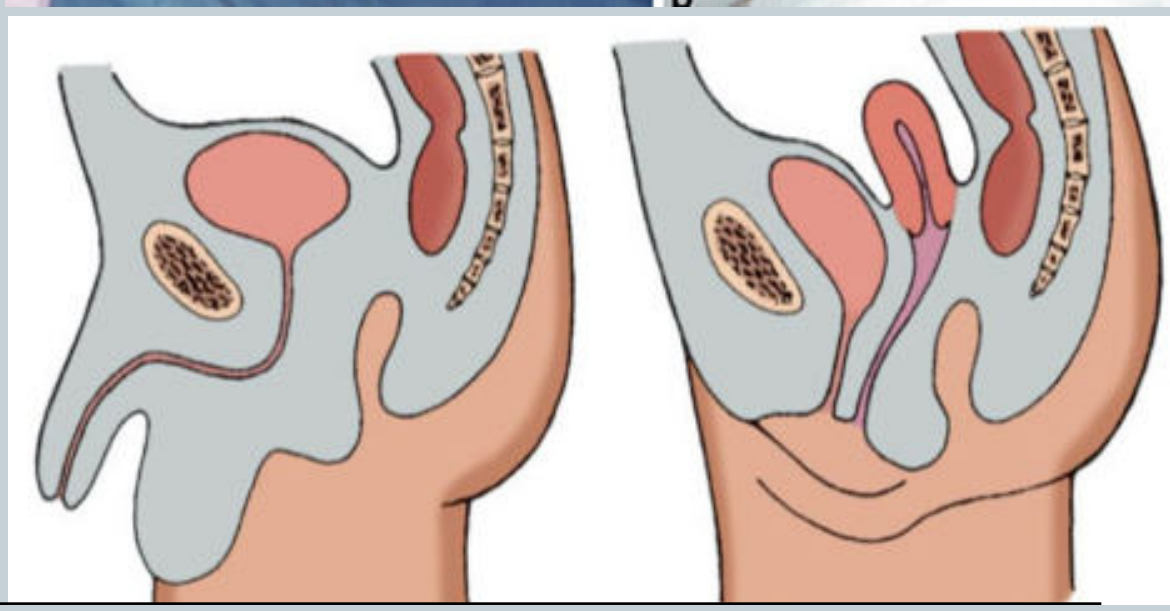
Ileostomy closure

3. RECTAL ATRESIA

- 1-2% of ARM
- Common in South India
- End to end anastomosis via PSARP route.







Postoperative management

- Foleys catheter
- IV antibiotics and wound care
- Anal dilatation (after 2 weeks) till desired size
- Colostomy closure

Age	Hegar dilator (no.)
1–4 months	12
4–12 months	13
8–12 months	14
1–3 years	15
3–12 years	16
>12 years	17

COMPLICATIONS

Group A	Patients with fecal incontinence requiring reoperation
Group B	Perioperative complications • Wound infection • Femoral nerve palsy • Rectal problems: dehiscence, retraction, infection and/or acquired atresia • Rectourinary fistula • Rectovaginal fistula • Persistent urogenital sinus • Acquired vaginal atresia • Acquired urethral atresia • Posterior urethral diverticulum • Urologic injuries • Neurogenic bladder • Complications involving the laparoscopic approach • Rectal prolapse
Group C	Complications from mismanagement of constipation

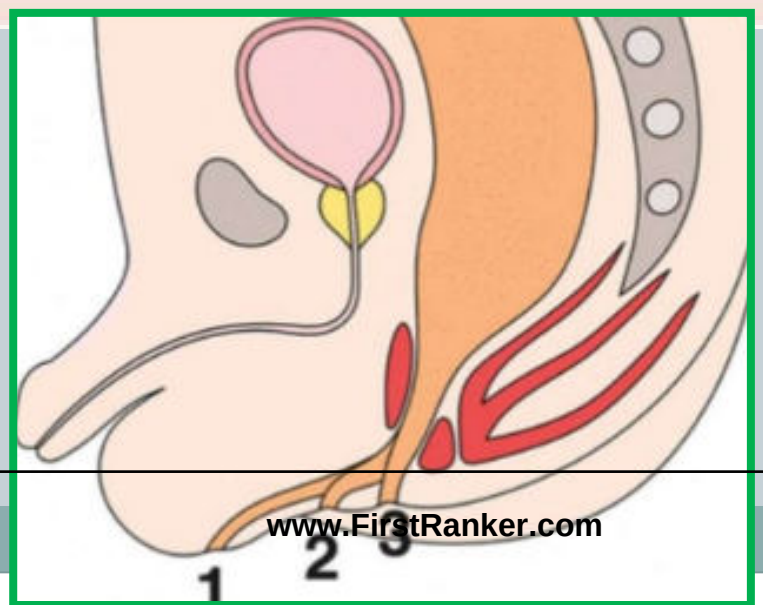
EVALUATION OF OUTCOMES

Table 27.8 Method for assessment of outcome established in Krickenbeck 2005 (patient age > 3 years, no therapy; Holschneider et al. [9])

1. Voluntary bowel movements	**yes/no**
Feeling of urge	
Capacity to verbalize	
Hold the bowel movement	
2. Soiling	**yes/no**
Grade 1 Occasionally (once or twice per week)	
Grade 2 Every day, no social problem	
Grade 3 Constant, social problem	
3. Constipation	**yes/no**
Grade 1 Manageable by changes in diet	
Grade 2 Requires laxatives	
Grade 3 Resistant to diet and laxatives	

FUNCTIONAL OUTCOME DURING CHILDHOOD- LOW ARM

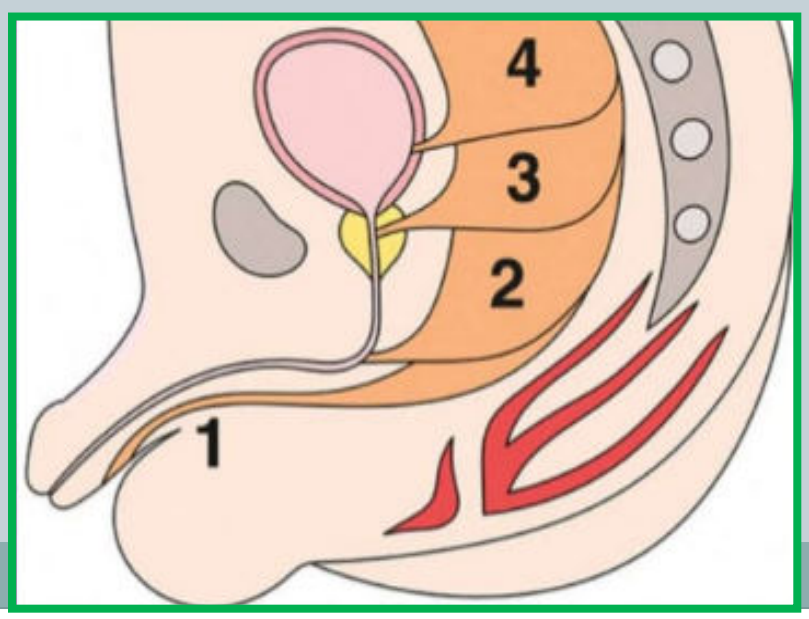
Recent series	N	Normal	Constipated	Soil/smear
Yeung and Kiely [23]	35	47%	53%	28%
Rintala et al. [57]	40	52%	42%	10%
Laboure et al. [59]	27	52%	48%	
Javid et al. [60]	28	53%	47%	



Functional outcome during childhood – high malformations: PSARP

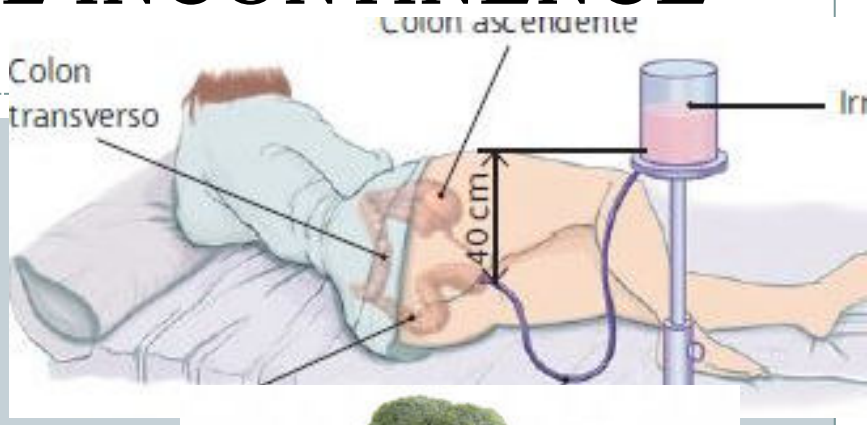
	Total continence	Significant soiling	Constipation
Peña [8]	36%	41%	43%
Rintala and Lindahl [28]	35%	30%	60%
Langemeijer and Molenaar [29]	7%	56%	5%
Rintala and Lindahl [70]*	50%	22%	9%

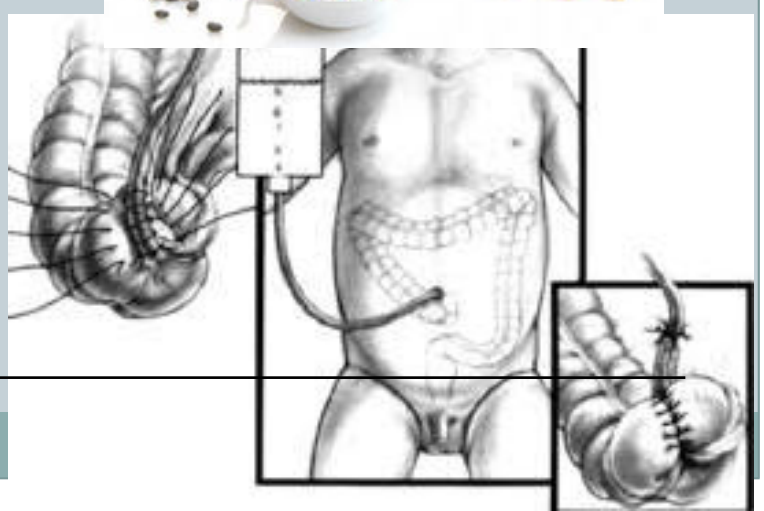
*Adolescents



TREATMENT OF FECAL INCONTINENCE

- Proper bowel management program
- Dietary modifications
- Surgical procedures to improve continence





OTHER OUTCOMES

- Urinary function
- Sexual function
- Long-term growth and development