Hirschsprungs & ARM

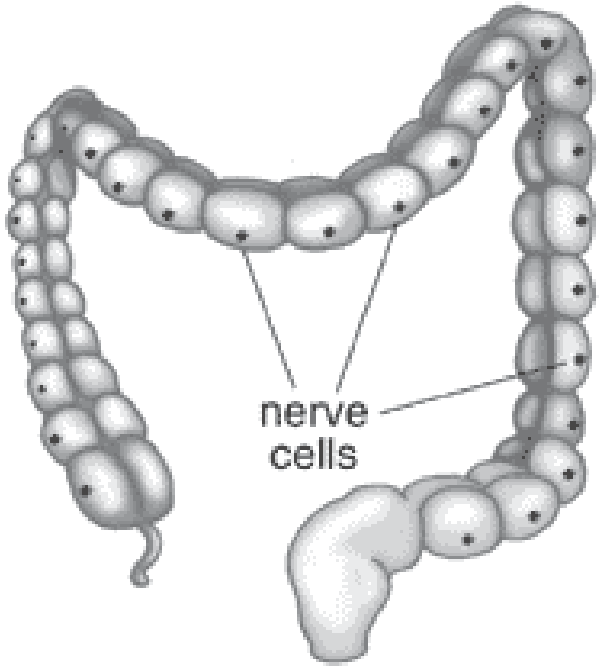
Hirschsprung's disease (Aganglionic mega colon)

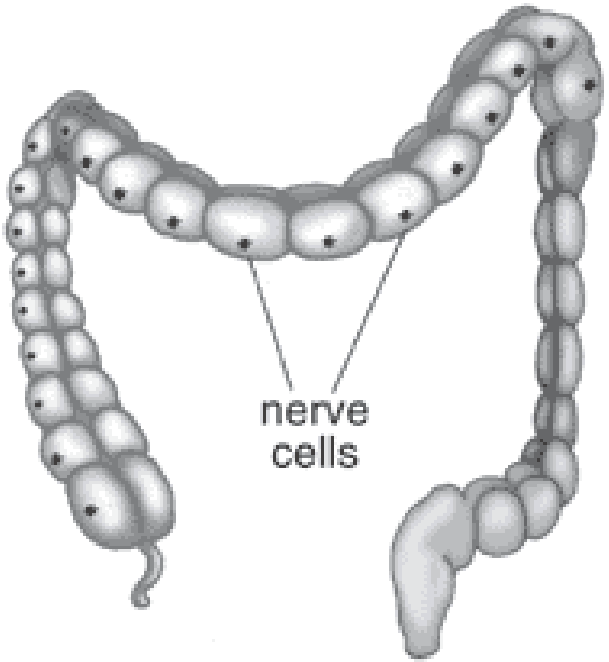
It is the commonest cause of intestinal obstruction in infancy

- Failure of caudal migration of neuroblasts derived form the neural crest
- There is absence of ganglions in the submucous and the myenteric plexuses

- Prevalence: 1/5000 births
- 3-5% of pts have Down's syndrome
- 80% affected are boys
- Most cases are sporadic
- Long segment and total colonic aganlionosis have strong familial association (15% &25%)
- >95% cases are full term babies

Pathogenesis





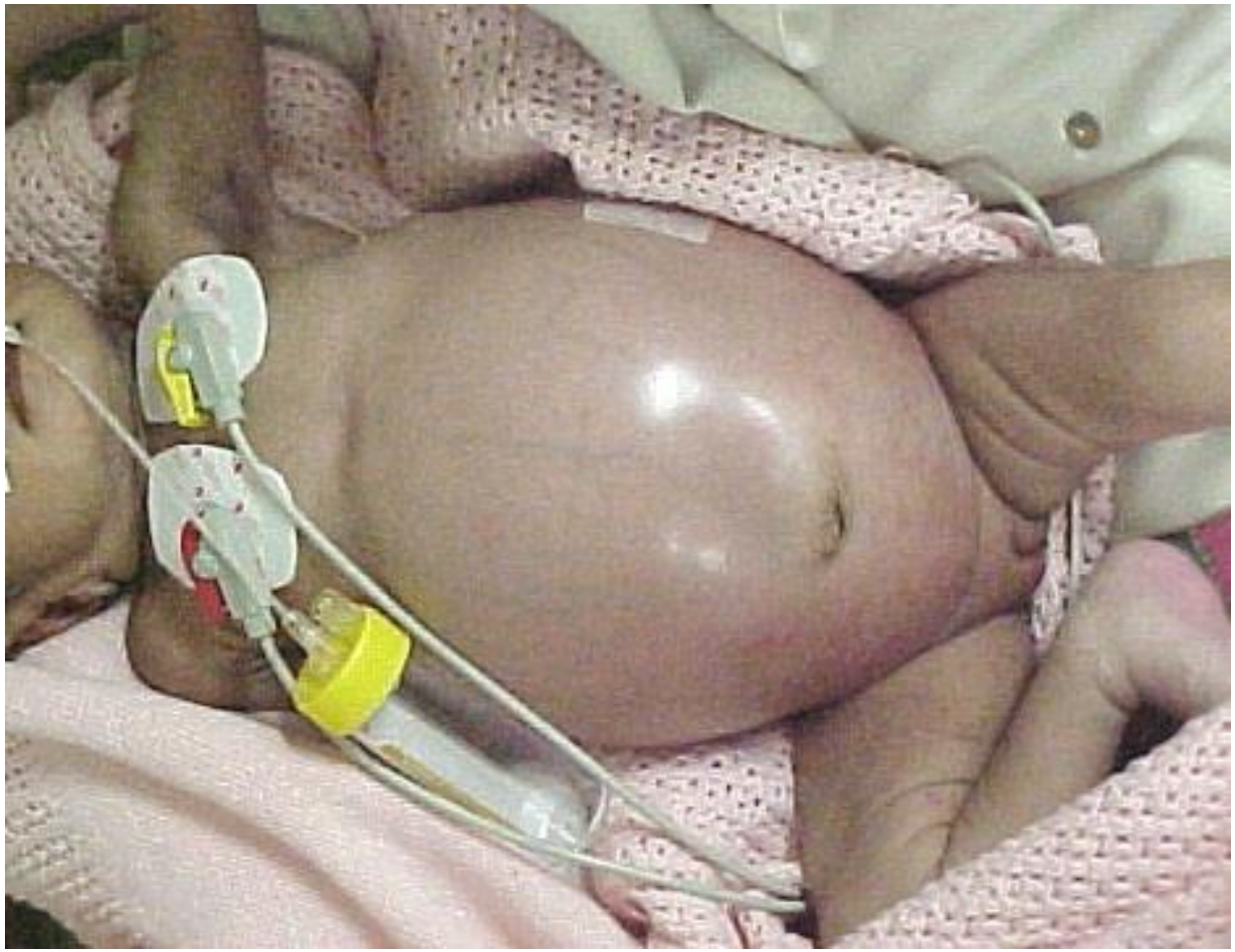
Pathogenesis

- Failure of neural crest cells to migrate caudally
- Aganglionosis begins at anorectal line
- 80% involve only rectosigmoid area
- 10% extend proximal to splenic flexure
- 10% involves the entire colon and part of small bowel
- Rarely involves entire gastrointestinal tract

Pathogenesis—genetics

- 10th chromosome
- RET-protooncogene
- Endothelin B gene

Presentation

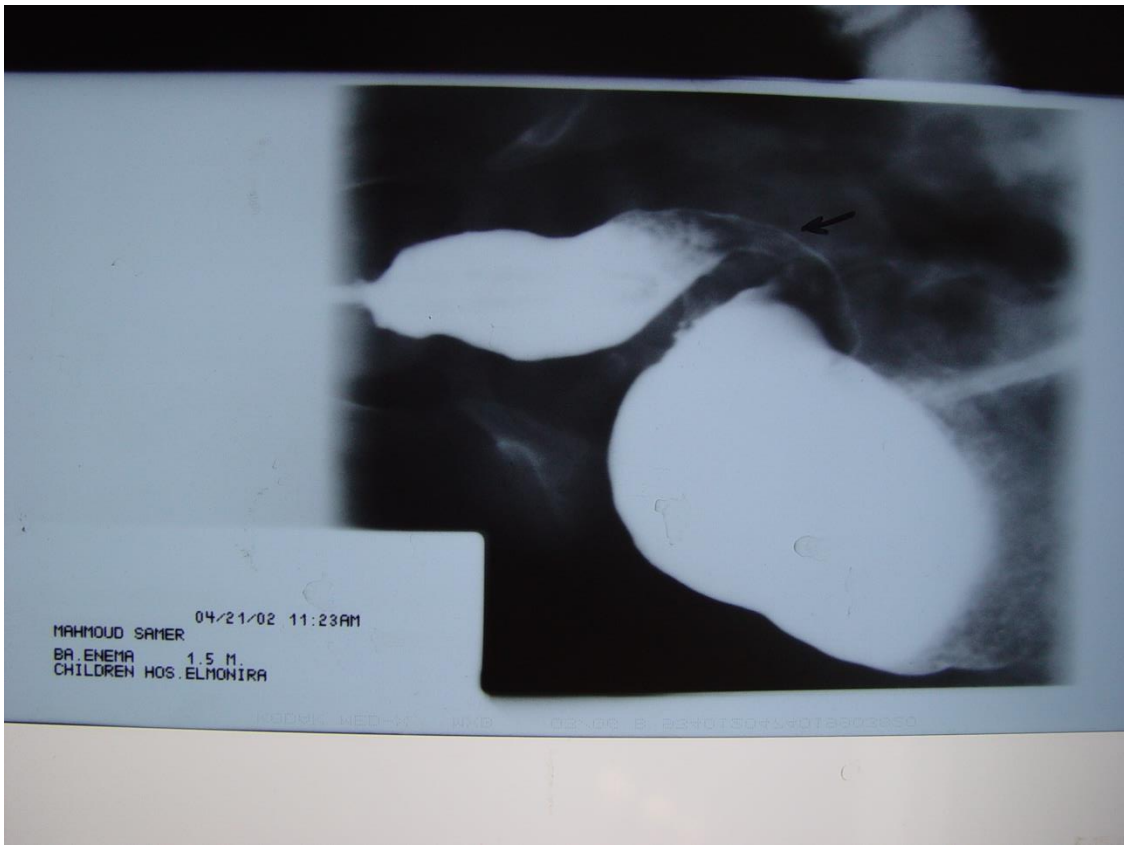


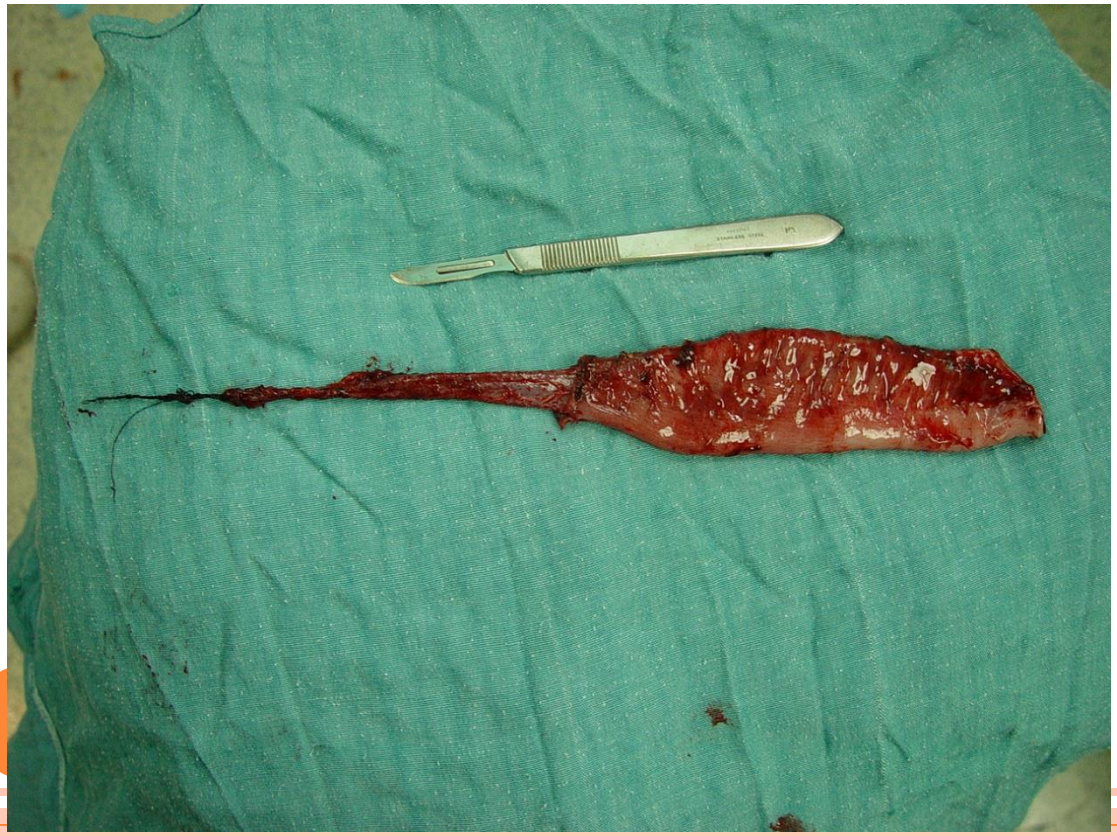
Presentation

- Severe abdominal distention
- 95% - failure to pass meconium in first 24 hours life
- Rectal examination reveals a tight spastic rectum
- Bilious vomiting
- Older children - constipation, failure to thrive
- 10-15% - severe diarrhea alternating w/ constipation—*enterocolitis of Hirschsprung's disease*

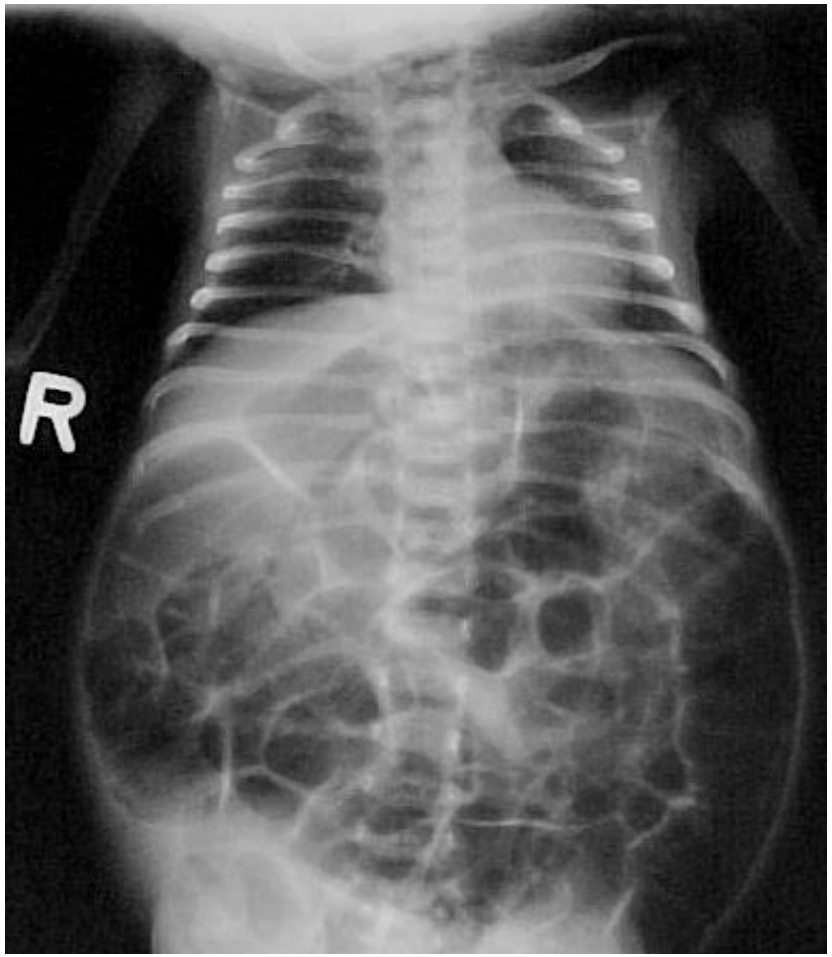
Diagnosis

- Abdominal plain X-rays
- Barium Enema
- Rectal Biopsies gold standard
- Anal manometry

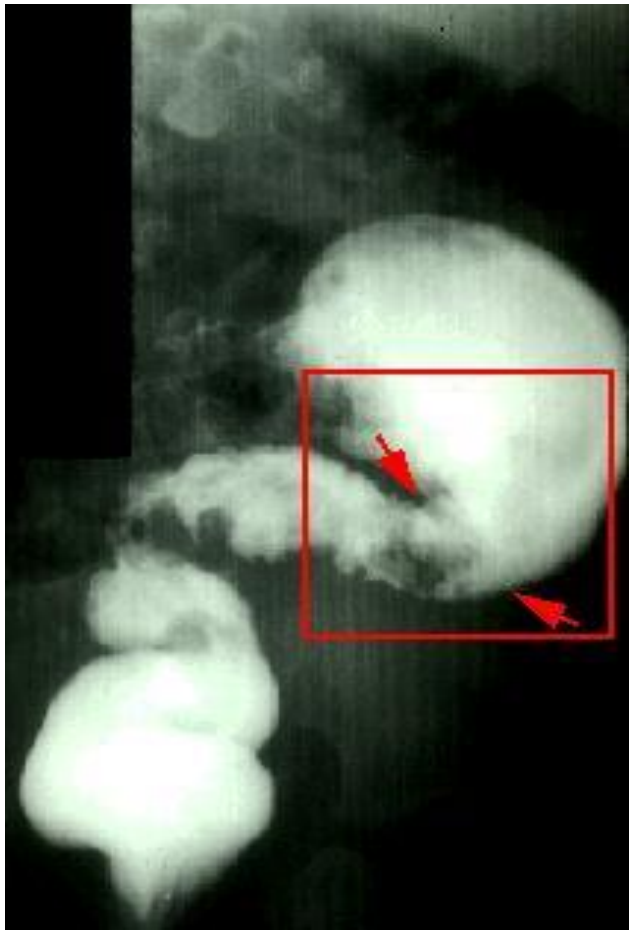

04/21/02 11:23AM
MAHMOUD SAMER
BA.ENEMA 1.5 M.
CHILDREN HOS.ELMONIRA

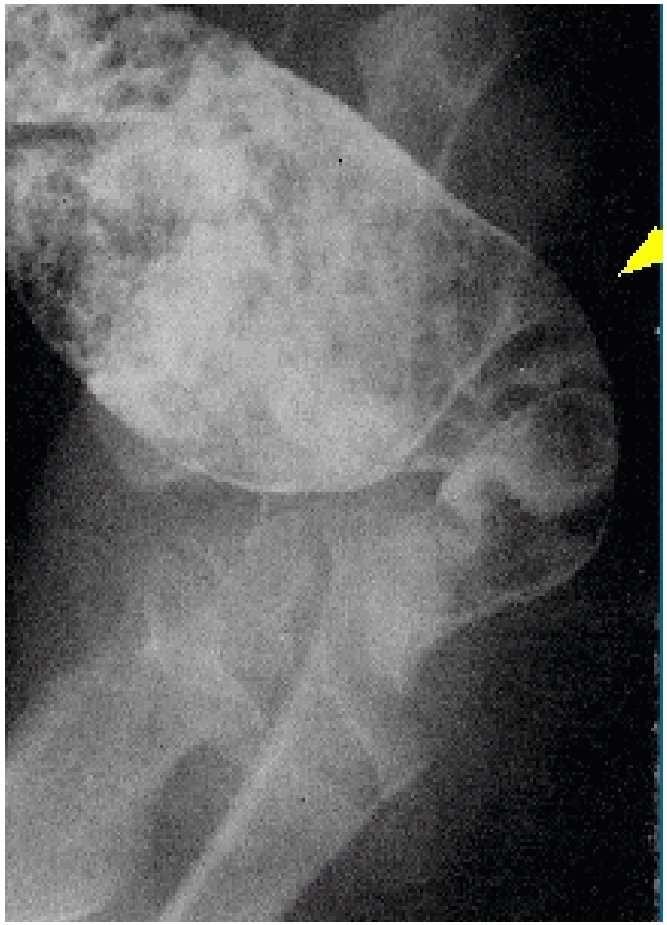


Abdominal X-ray



Barium Enema





Barium Enema

- Less sensitive for detecting short lesions, total colon aganglionosis, and disease of the newborn
- Many newborns do NOT show definitive transition zone
- Delayed evacuation of contrast

Rectal biopsy

- Submucosal suction biopsy
  - Meissner's submucosal plexus
- Full thickness rectal biopsy
  - Auerbach's myenteric plexus
- Acetylcholinesterase staining
  - increased staining of neurofibrils

Anorectal manometry

- Absent rectoanal inhibitory reflex
- Lack of internal anal sphincter relaxation in response to rectal stretch

Treatment:

- Temporary measures include laxatives and enemas.
- Colostomy may be a life saving procedure in some cases
- Definitive treatment is surgical excision of the spastic segment an re-anastomosis to the anal canal. Various procedures can be done e.g Swenson's, Duhamel's and Soave's procedures

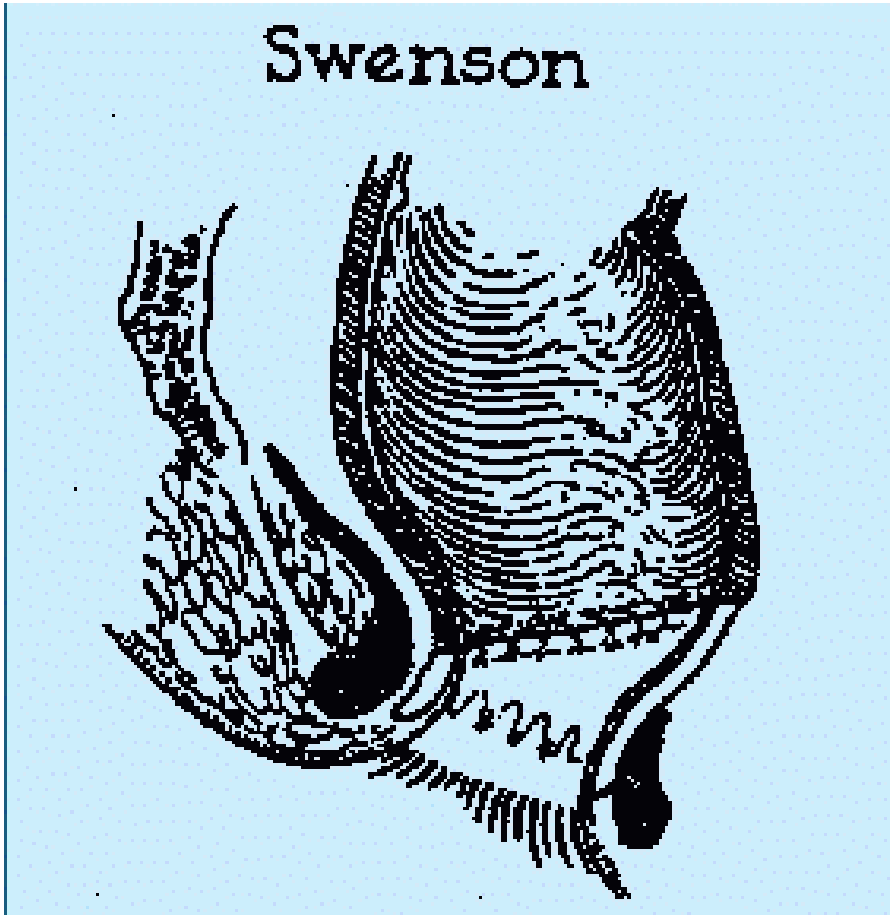
Surgical Options

- Swenson Procedure (1948)
- Duhamel Procedure (1960)
- Soave Procedure (1963)

Swenson Procedure

- Sharp extrarectal dissection down to 2 cm above the anal canal
- Aganglionic colonic segment resected
- End-to-end anastamosis of normal proximal colon to anal canal
- Completely removes defective aganglionic colon

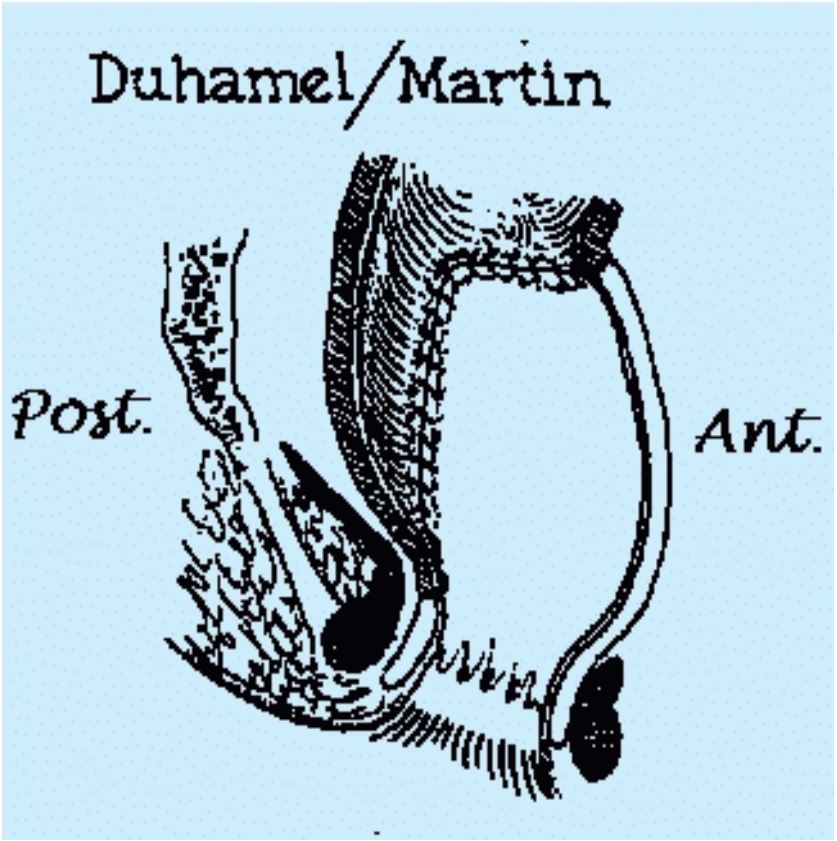
Swenson Procedure

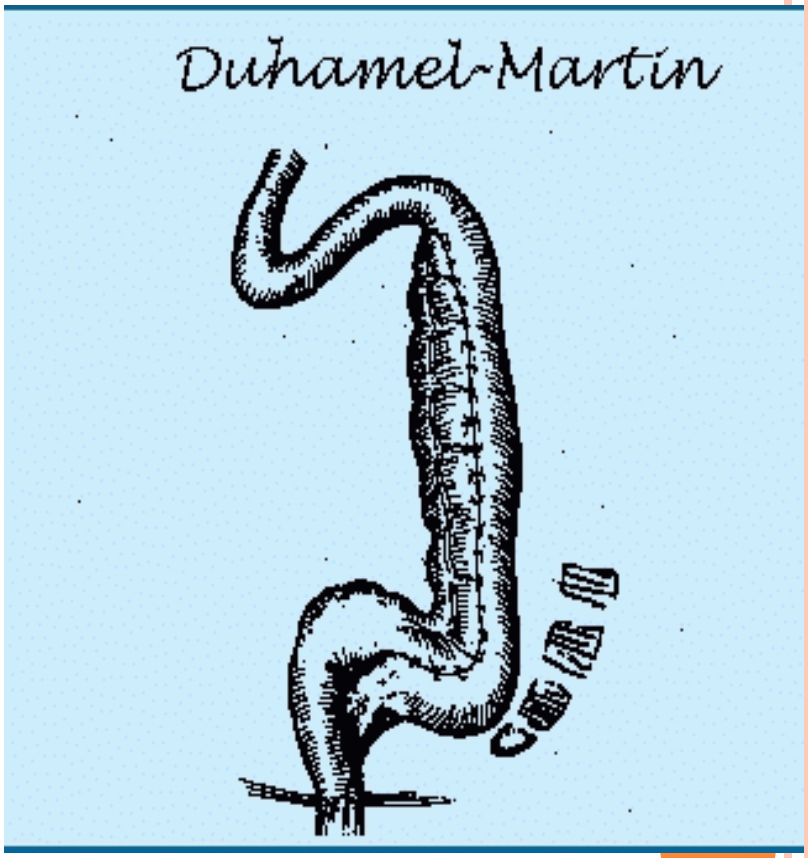


Duhamel Procedure

- Posterior portion of defective colon segment resected
- Side to side anastamosis to left over portion of rectum
- Constipation a major problem d/t remaining aganglionic tissue
- Simpler operation, less dissection

Duhamel Procedure

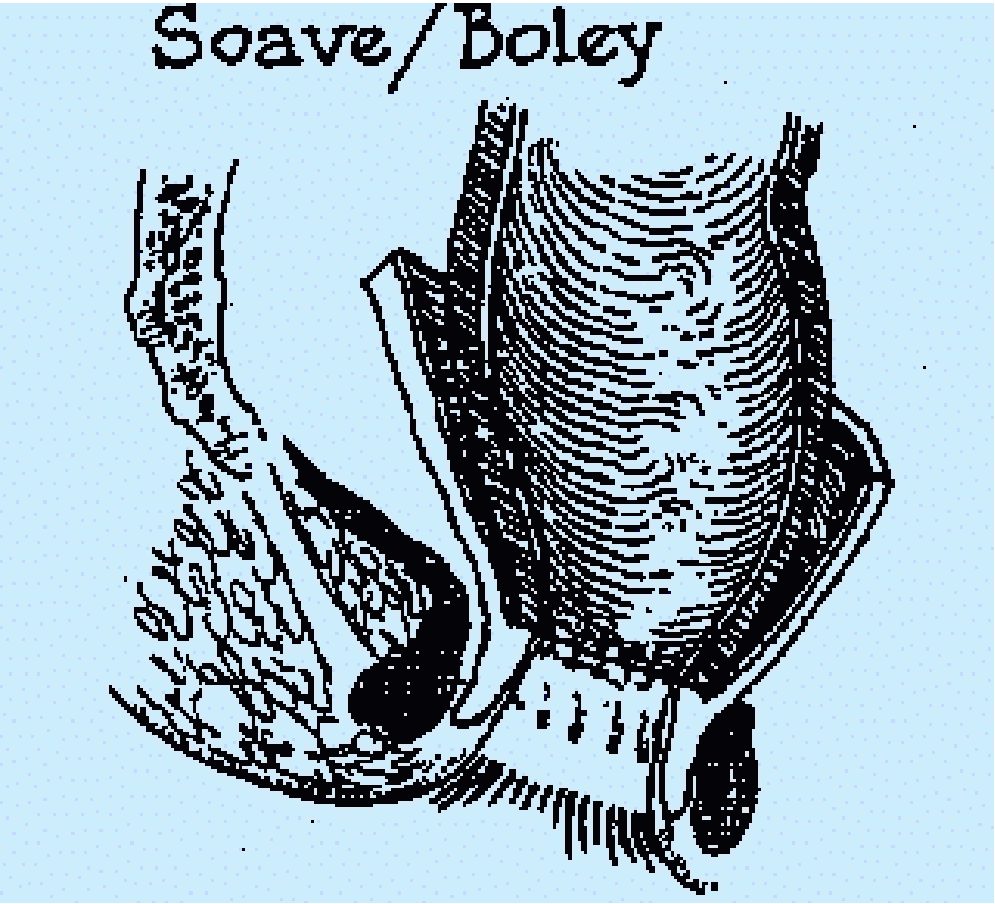




Soave Procedure

- Circumferential cut through muscular coat of colon at peritoneal reflection
- Mucosa separated from the muscular coat down to the anal canal
- Proximal normal colon is pulled through retained muscular sleeve
- Telescoping anastamosis of normal colon to anal canal

Soave Procedure



Soave Procedure

- Advantage: rectal intramural dissection ensures no damage to pelvic neural structures
- Higher rate enterocolitis, diarrhea
- Problems w/ cuff abscesses, often requires repeated dilations

Overall Mortality

- Swenson procedure: 1-5%
- Duhamel procedure: 6%
- Soave procedure: 4-5%

Operative complications

- Leak at anastamosis: 5-7%
- Postop Enterocolitis: 19-27%
- Constipation
- Stricture Formation
- Incontinence

One vs Two Stage procedure

- Historically, two stage procedure performed: preliminary colostomy, then completion pull through
- Delicate muscular sphincters of newborn may be injured
- 1980s, 1 stage procedures became more popular

One vs Two Stage procedure

- Early complications: No difference in incidence of anastomotic leak, pelvic infection, prolonged ileus, wound infection, wound dehiscence
- Late complications: No difference in incidence of anastomonic stricture, late obstruction, constipation, incontinence, urgency
- Postoperative enterocolitis higher in 1 stage (42% vs 22%)

Laparoscopic techniques

- Small studies of laparoscopic pull through procedures
- Excised aganglionic tissues removed through anal canal, no abdominal incision
- Better results in terms of pain, return of bowel function, hospital stay
- Similar incidence of leaks, pelvic abscesses, enterocolitis, postop bowel function

ARM

Anorectal malformations (ARMs) are a complex group of congenital anomalies involving the distal anus and rectum, as well as the urinary and genital tracts in a significant percentage of cases.

Urogenital abnormalities are the most frequently observed and appear in up to 60% of patients, with vesicoureteral reflux and hydronephrosis the most common findings.

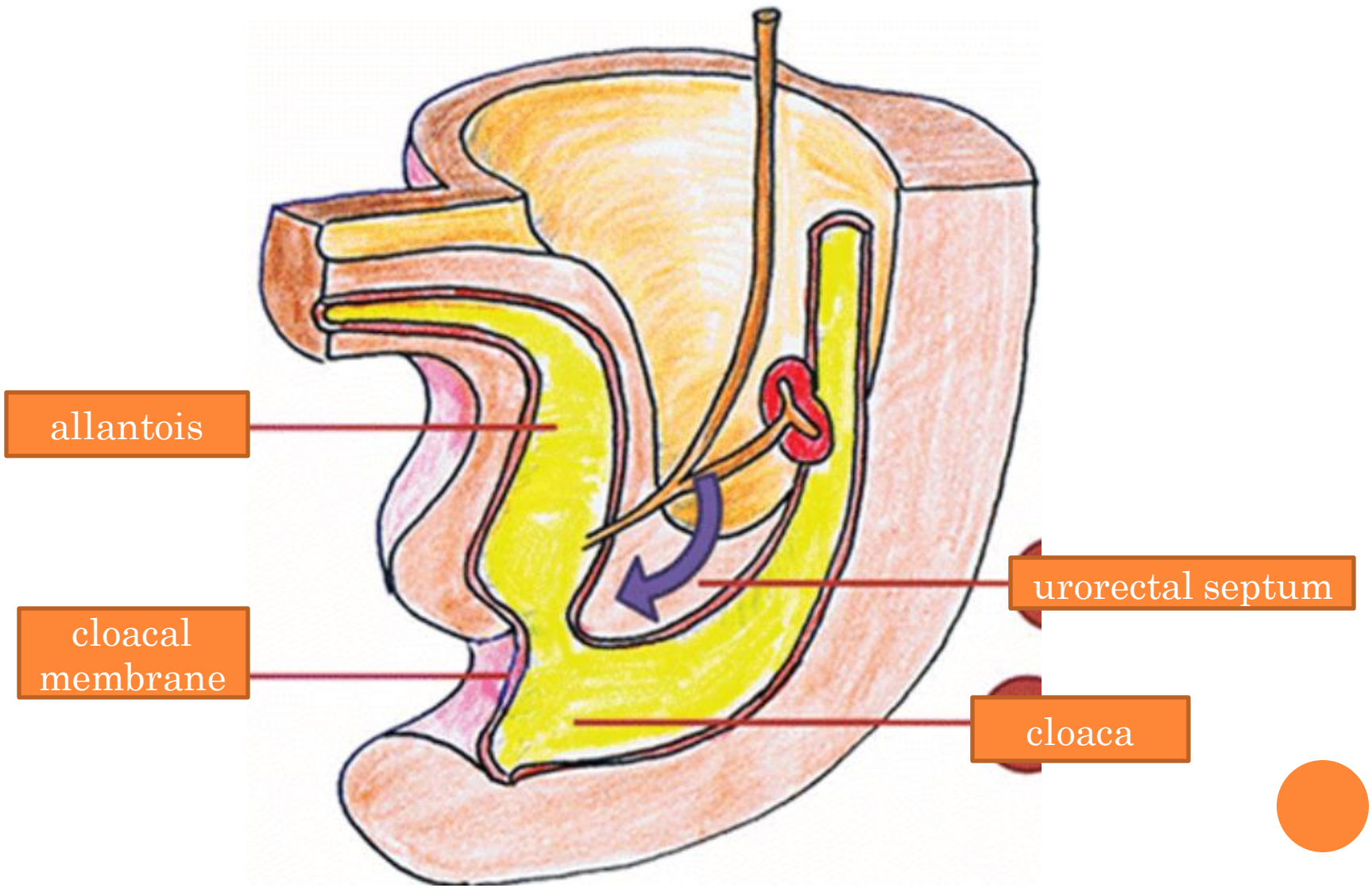
The spine and spinal cord are also often involved, with agenesis and dysplasia of the sacrum, vertebral dysplasia, and tethered cord syndrome the most frequently detected problems.

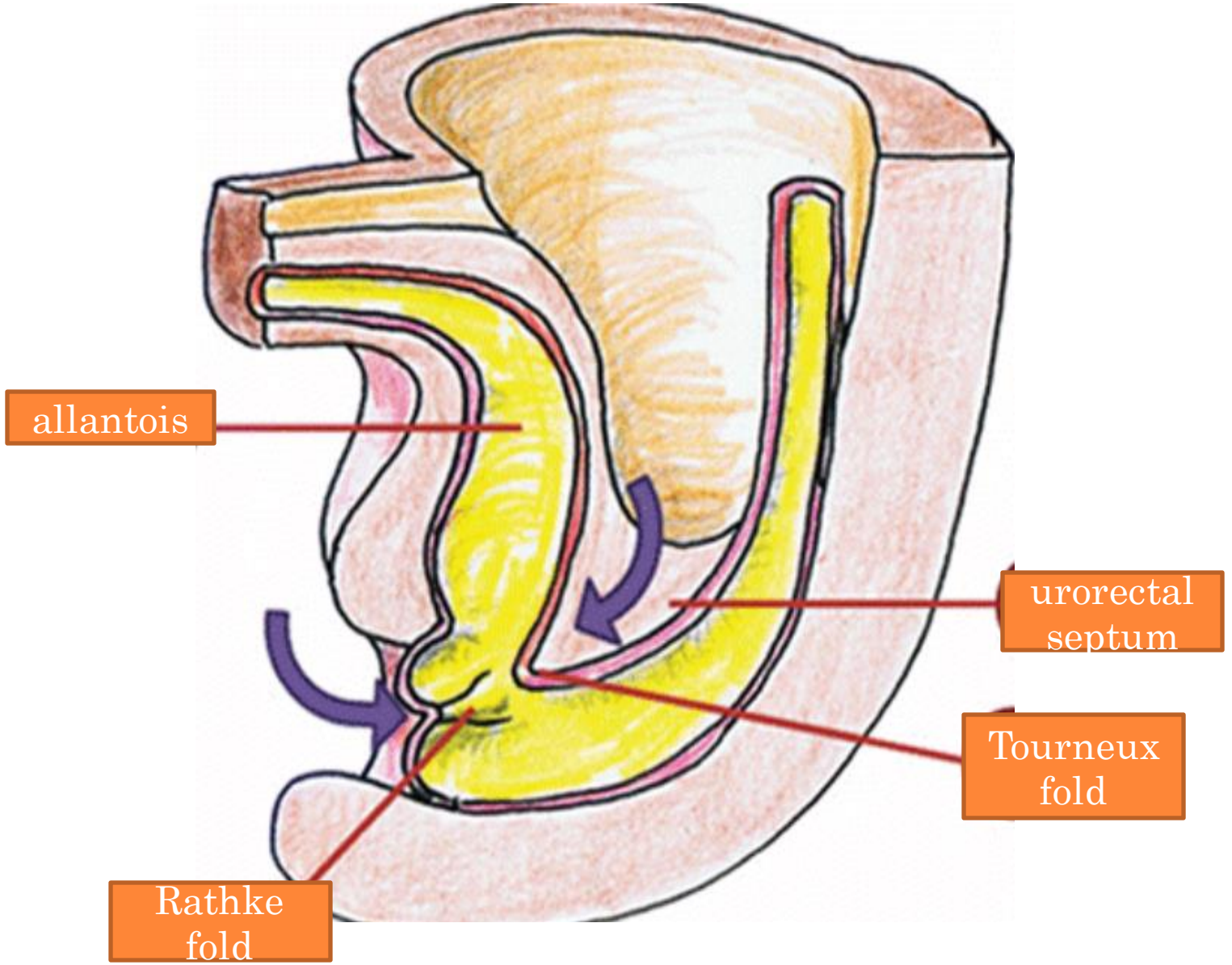
ARMs are also present in a great number of syndromes and associations of multisystemic congenital anomalies

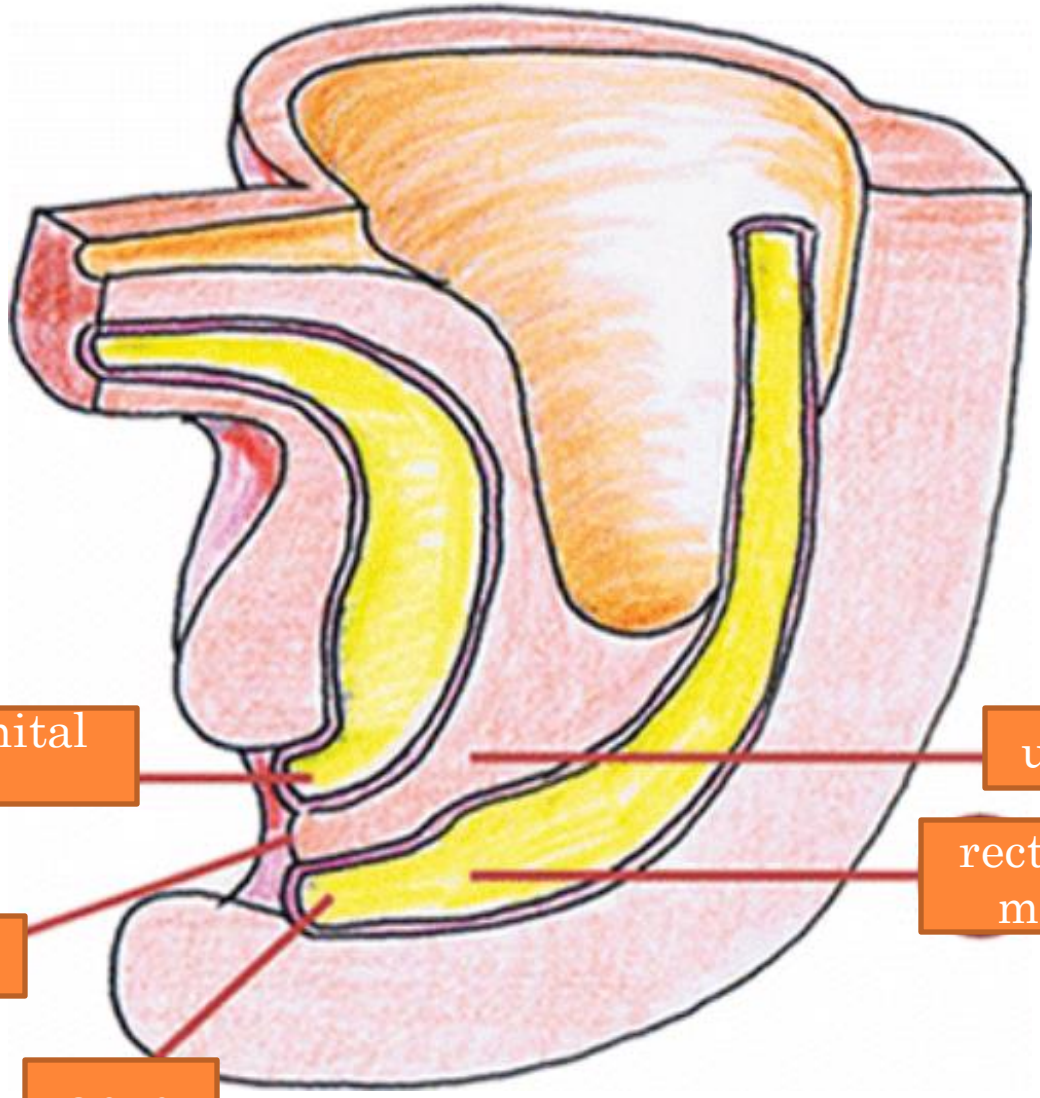
Affected System	Anomalies
Cardiovascular	Tetralogy of Fallot, atrial septal defect, ventricular septal defect, dextrocardia, coarctation of the aorta
Gastrointestinal	Esophageal atresia; duodenal, jejunal, or ileal atresia; absent colon; intestinal malrotation; volvulus; Meckel diverticulum
Musculoskeletal	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

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

Embryology




allantois
urorectal septum
Tourneux fold
Rathke fold


primitive urogenital sinus
urorectal septum
rectu m
perineal area
anus

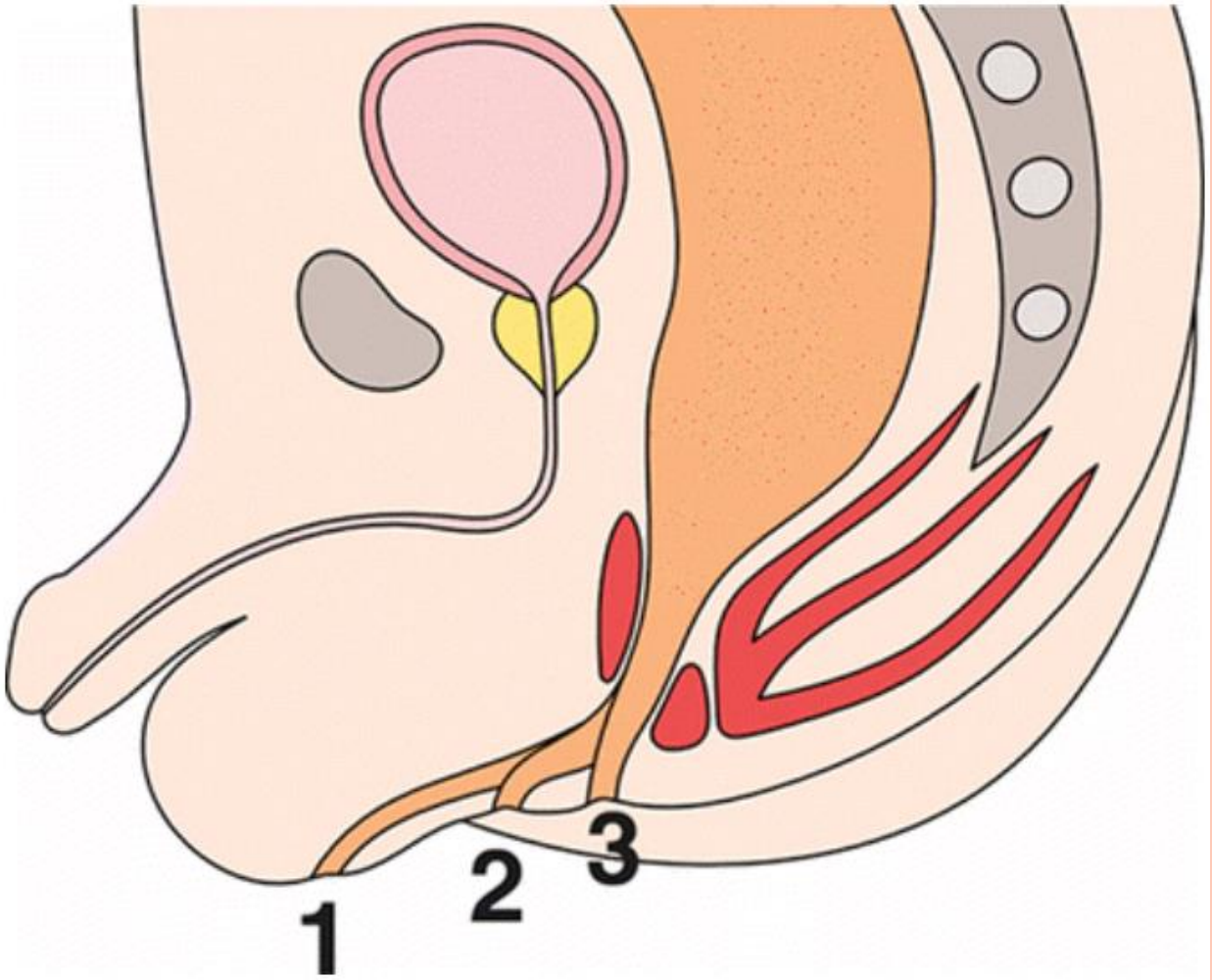
Embryologically, ARMs can thus be subdivided into two main groups according to when the disturbances occur:

Those manifesting as an ectopic anal orifice or fistula are due to early abnormal development of the dorsal part of the cloaca and the cloacal membrane (at weeks 4–7),
whereas those manifesting as an abnormal anus in a normal position are due to later defective recanalization of the secondary occluded anal orifice (at weeks 7 and 8).

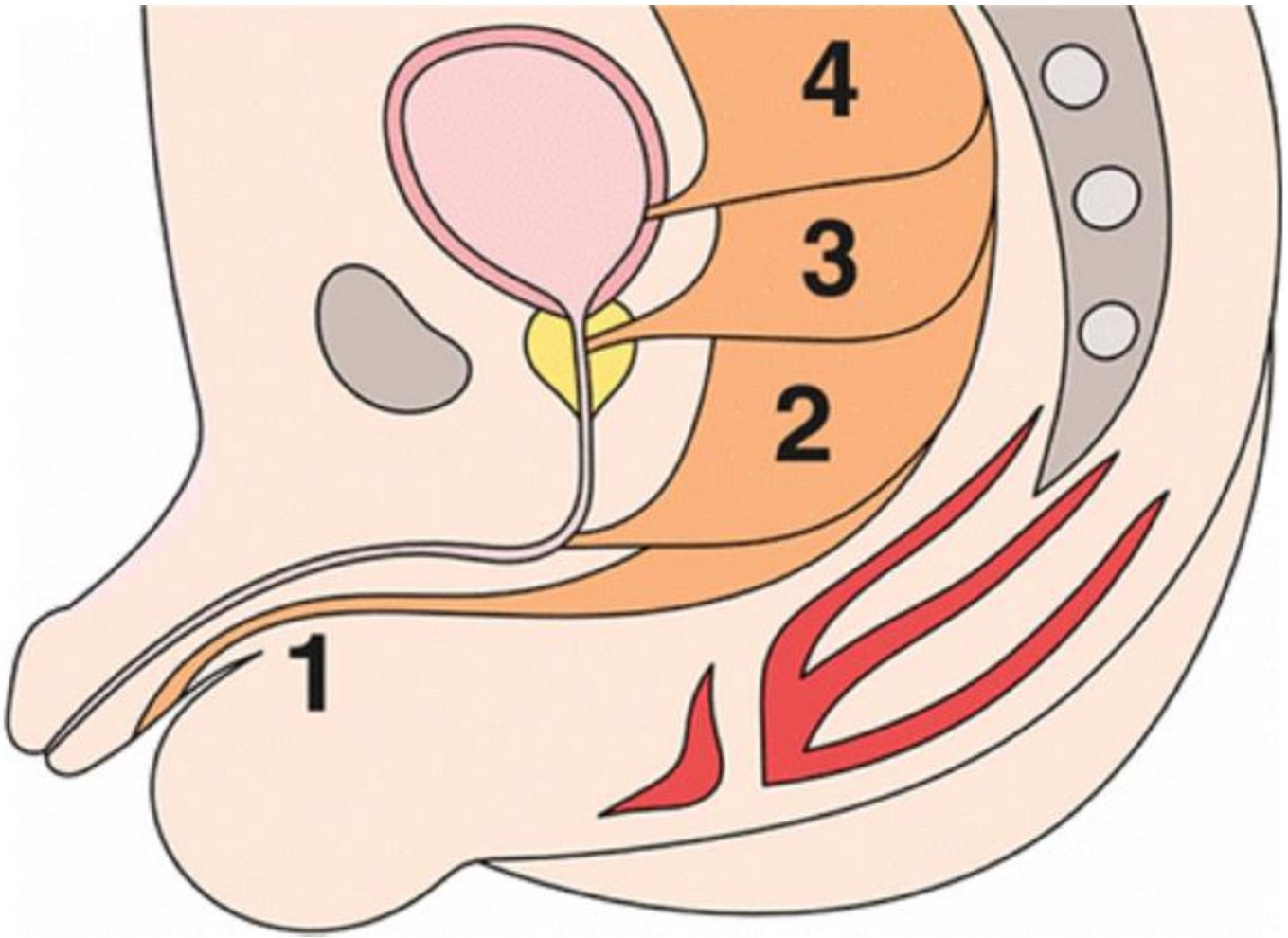
Classification of ARMs

Possible locations of fistulas in males with ARMs according to the Krickenbeck classification.

(a) Low-type ARMs have an external anocutaneous opening in the scrotum *(1)* or perineum (*2*, *3*).

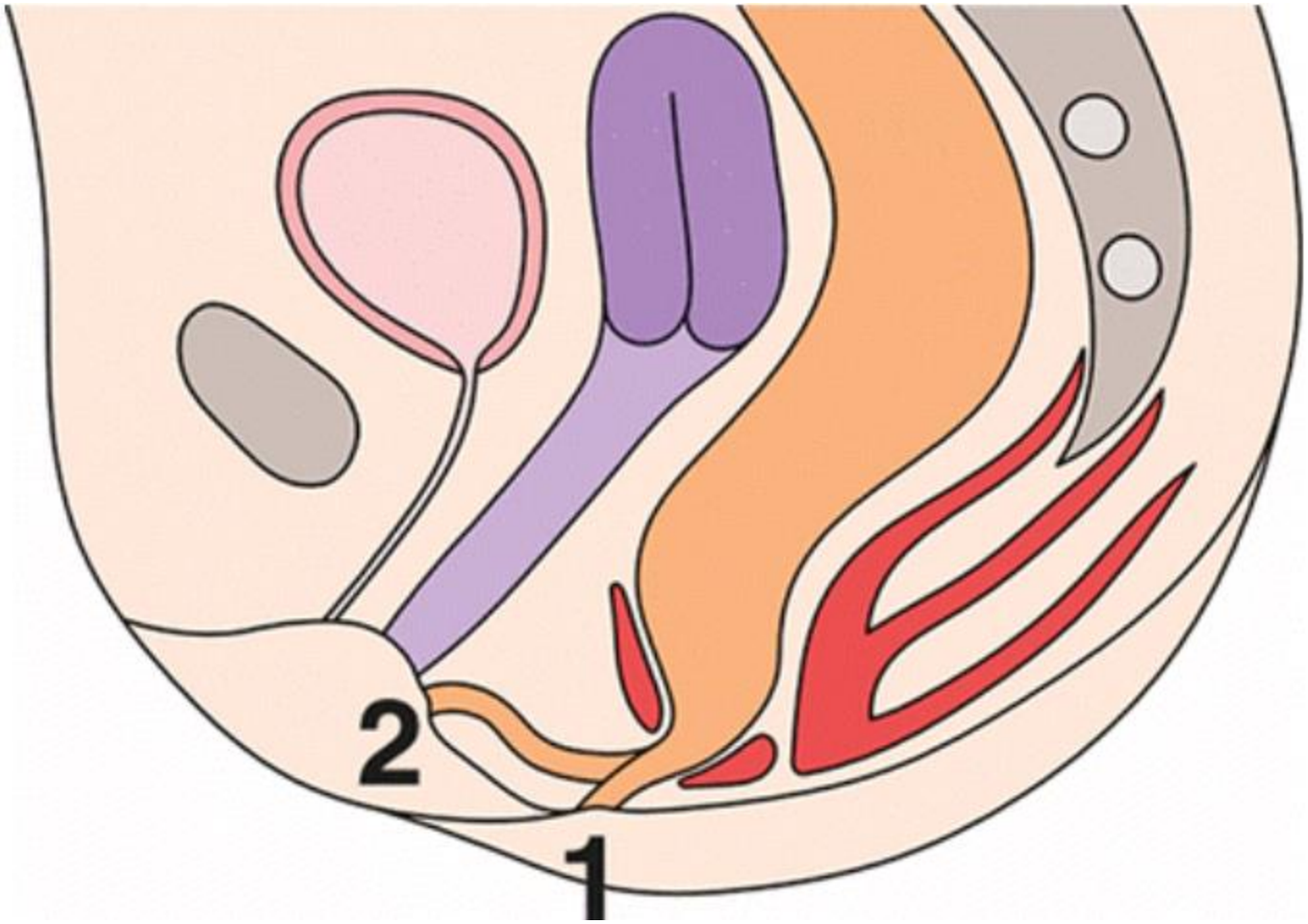


(b) Intermediate- and high-type ARMs extend anteriorly to the base of the penis *(1)*, the bulbar *(2)* or prostatic *(3)* urethra, or the urinary bladder *(4)*.

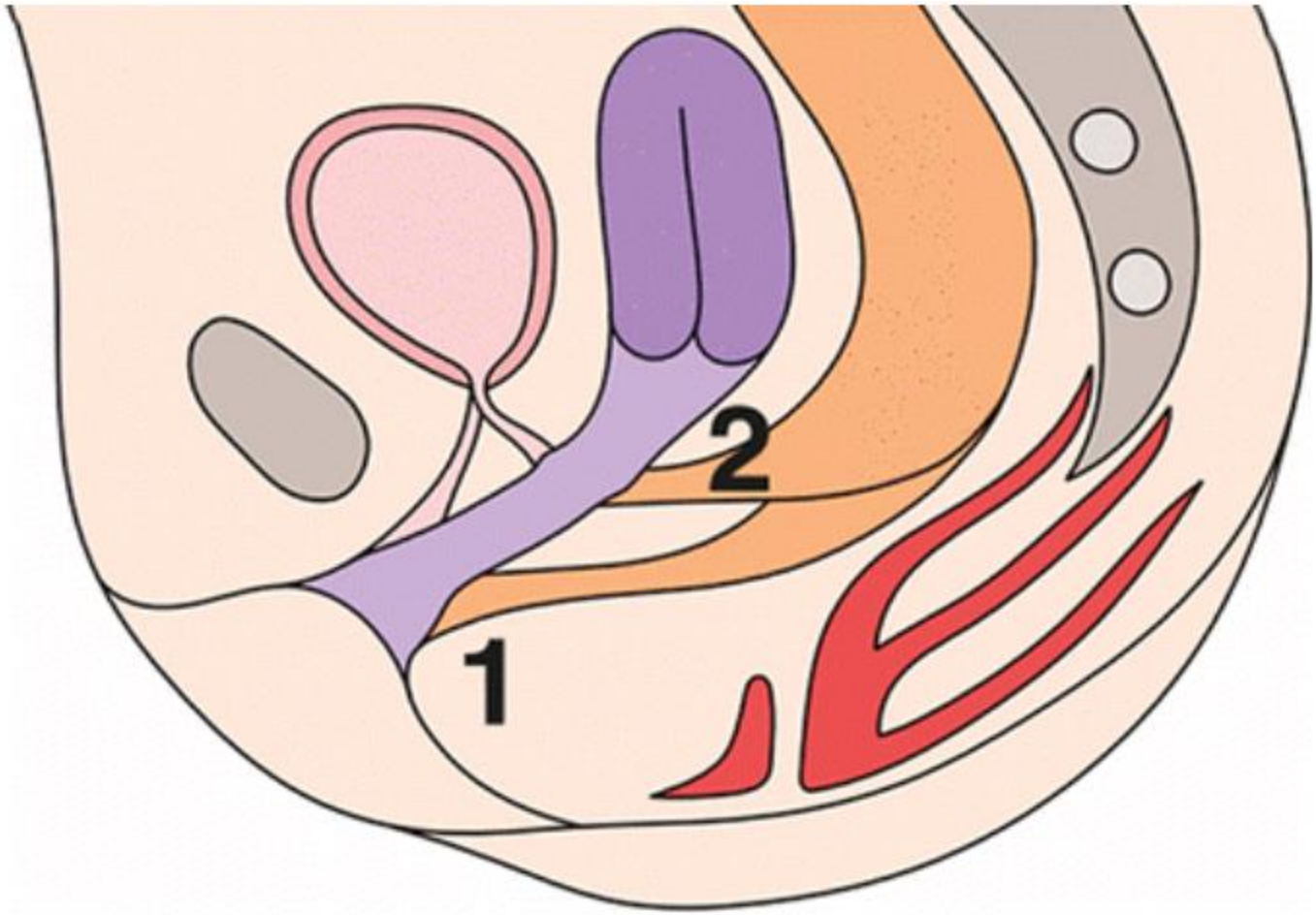


Possible locations of fistulas in females with ARMs according to the Krickenbeck classification.

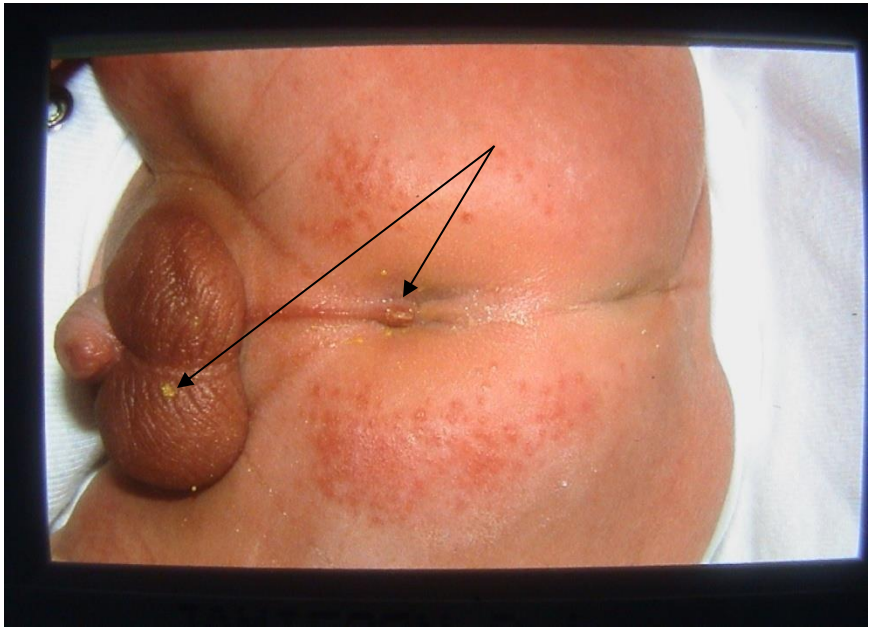
(a) Low-type ARMs have an external opening in the perineum *(1)* or vestibular area *(2)*.

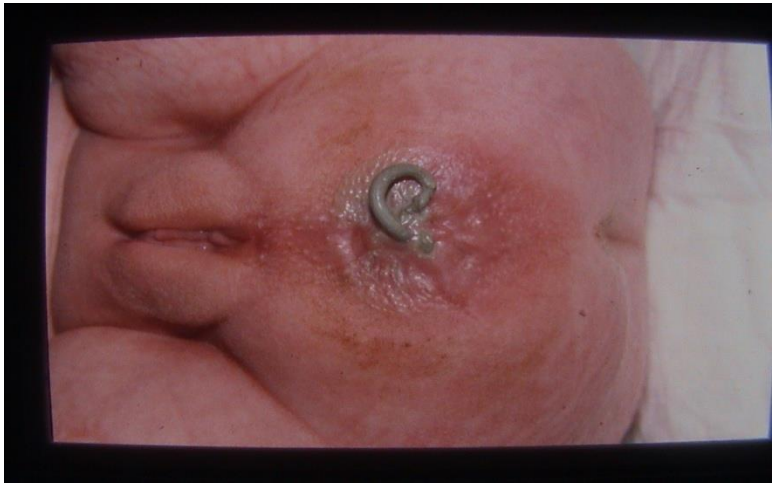


(b) Cloacal anomaly is a complex anatomic disorder that manifests as a unique external perineal opening with a short *(1)* or long *(2)* common canal for the genital, urinary, and digestive systems. Isolated rectovaginal fistulas are extremely rare and are considered a variant of cloacal anomaly.



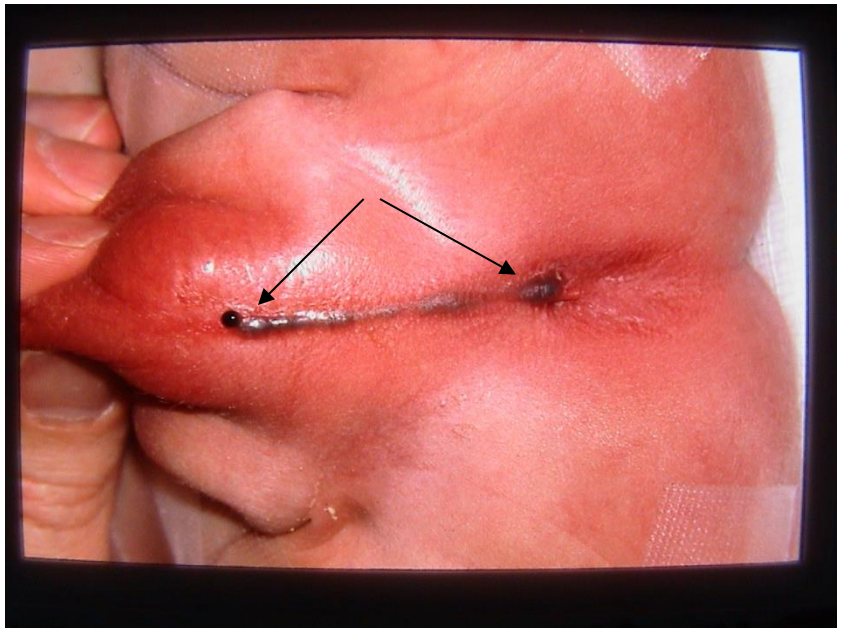
Classification of Anorectal Malformations	
Males	***Females***
Perineal fistula	Perineal fistula
Rectourethral fistula	Vestibular fistula
Bulbar	Persistent cloaca
Prostatic	≤3 cm common channel
Rectobladder neck fistula	>3 cm common channel
Imperforate anus without fistula	Imperforate anus without fistula
Rectal atresia	Rectal atresia
Complex defects	Complex defects





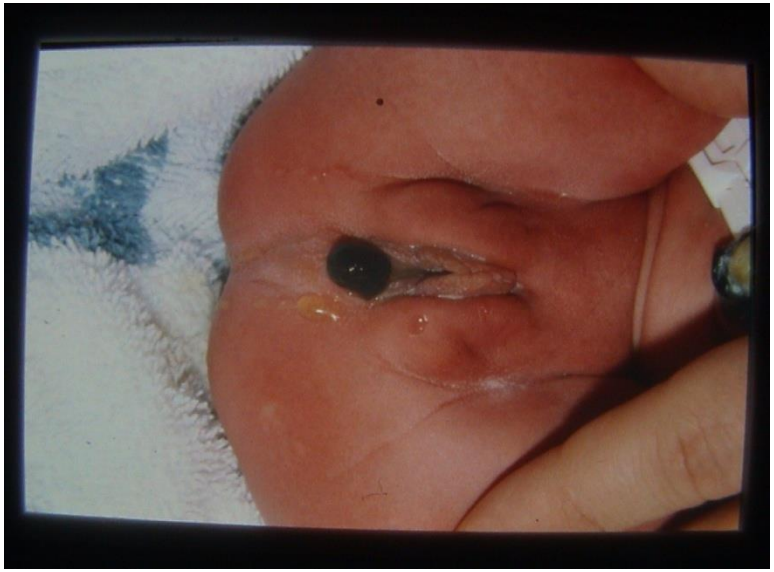
Above Right: Anal stenosis

Above Left
Covered Anus (Bucket handle anomaly)
Note stool particles (arrows)



Left
Low Imperforate Anus
Note translucent membrane & ano-cutaneous fistula showing Meconium (arrows)

High Anomalies (Male & Female)

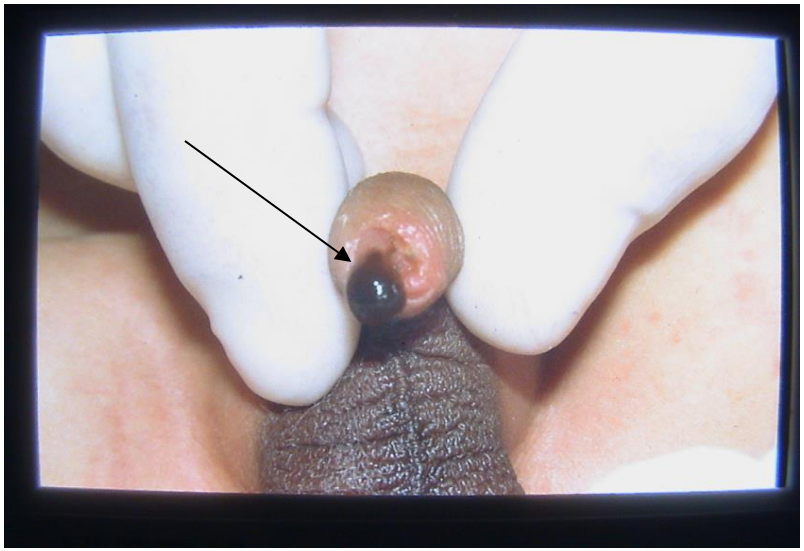


Left

Vesibular Anus

Note meconium coming

Out of vaginal vestibule



Right

Note meconium coming out

Of the urethra (arrow),

Indicating recto-urinary fistula

- The pubococcygeal line extending from the upper border of the pubis to the coccyx corresponds with the attachment of levator ani muscles to the pelvic wall, separating high-type malformations lying above the levator muscle and intermediate and low forms of anorectal agenesis lying below this anatomic line.

Management

As there are frequent association with other anomalies these should be also looked for, diagnosed or excluded

- **Clinical examination:**
  Usually the clinical diagnosis is made a birth either by inspection or by failing to pass a thermometer easily into the anus

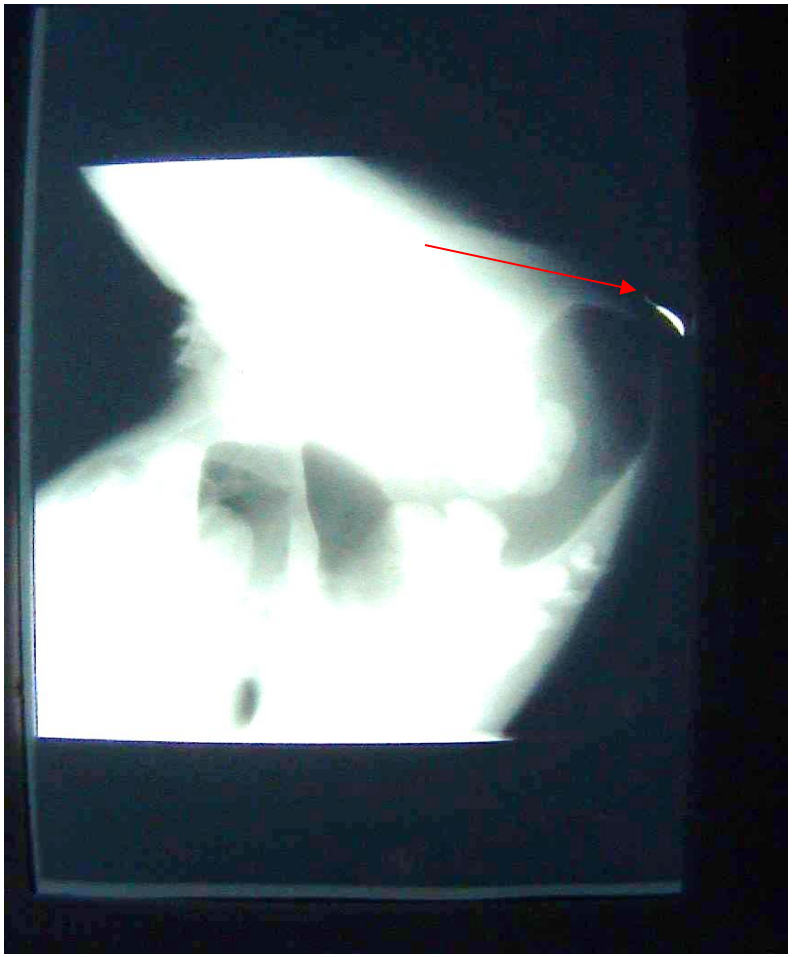
- **Radiography:**
  Invertogram, done after six hours of birth
  Contrast studies (distal loopogram): in a colostomized child to diagnose the site of any fistulous connection to the genito-urinary system

Imaging studies in the first 2 days of life should include radiography of the thorax, spine, and pelvis along with cardiac, perineal, abdominal, pelvic, and spine US to detect possible associated anomalies.

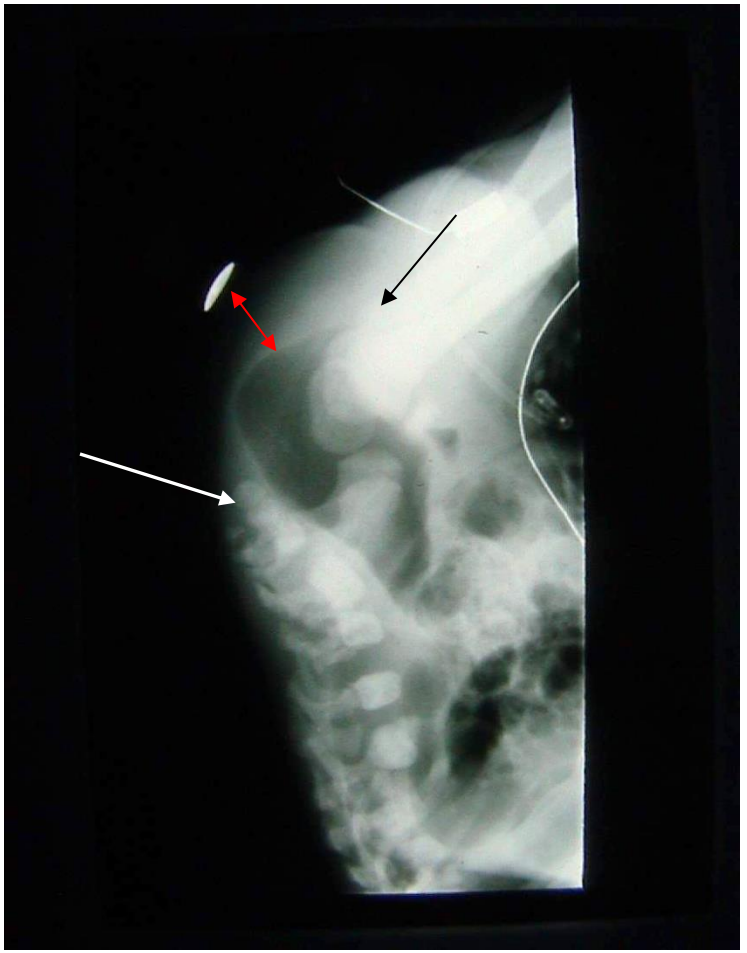
In cases of vertebral or sacral anomalies, spinal MR imaging should be performed.

Invertogram

Low Anomaly



High Anomaly



- **Treatment:**

  -*Colostomy:* early in the neonatal period for high anomalies

  -*Local procedure:* for low anomalies

  -Full correction of high anomalies is done later in life starting from the 6th month after which the colostomy maybe closed

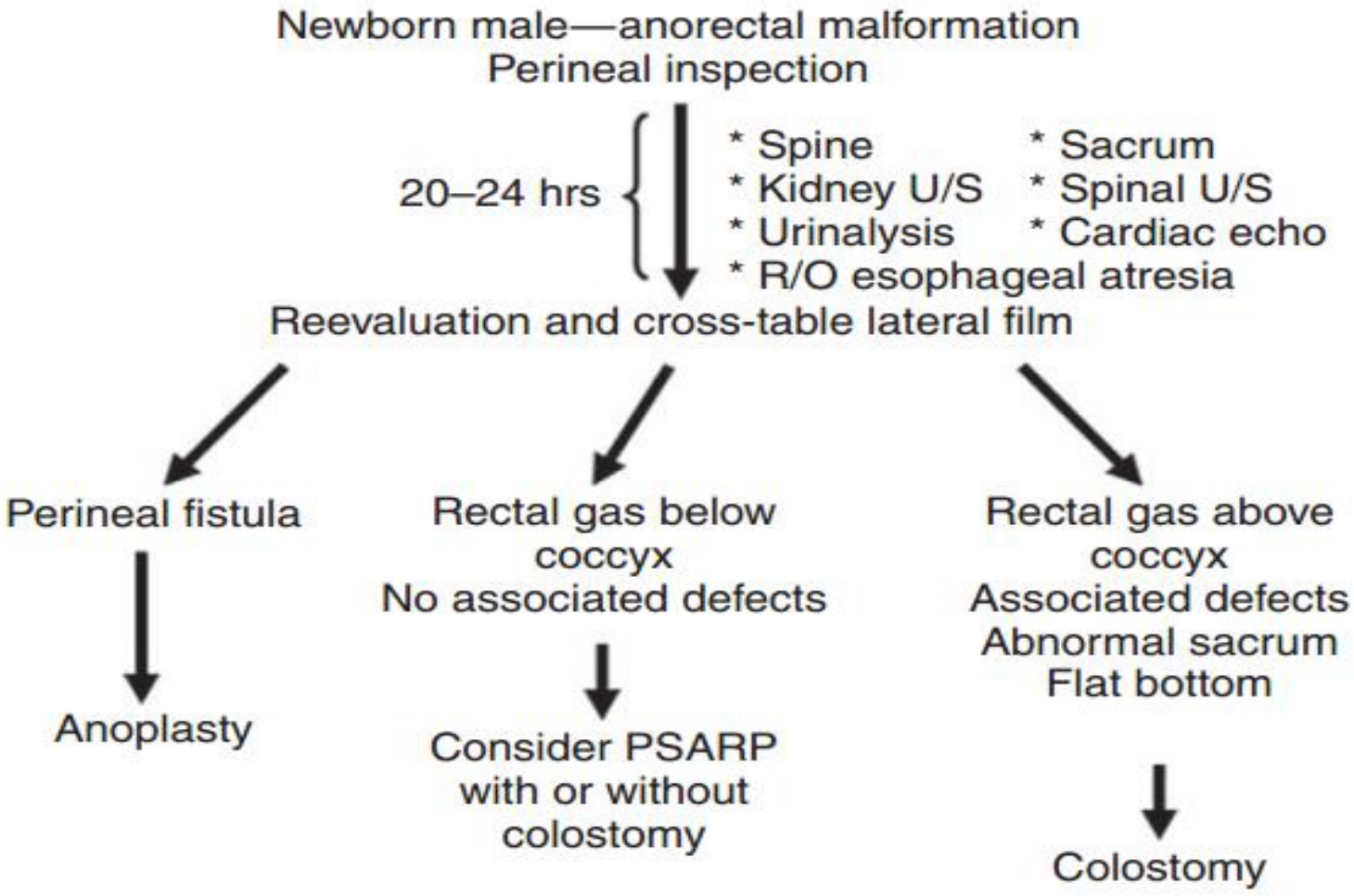
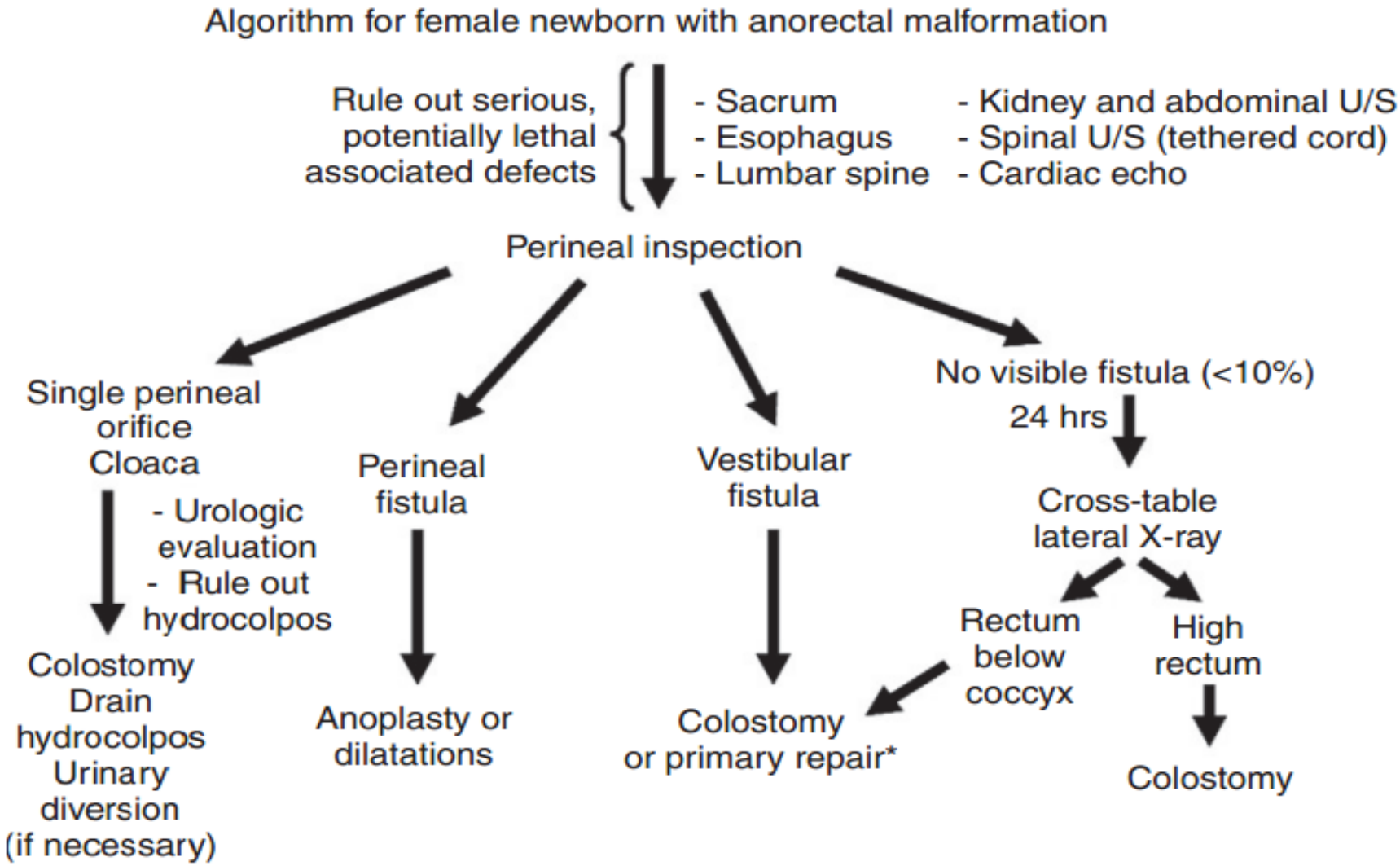


FIGURE 103-2 Algorithm for the treatment of a male newborn with an anorectal malformation.

PSARP —— posterior sagittal anorectoplasty



*Depending on the experience of the surgeon and general condition of the patient