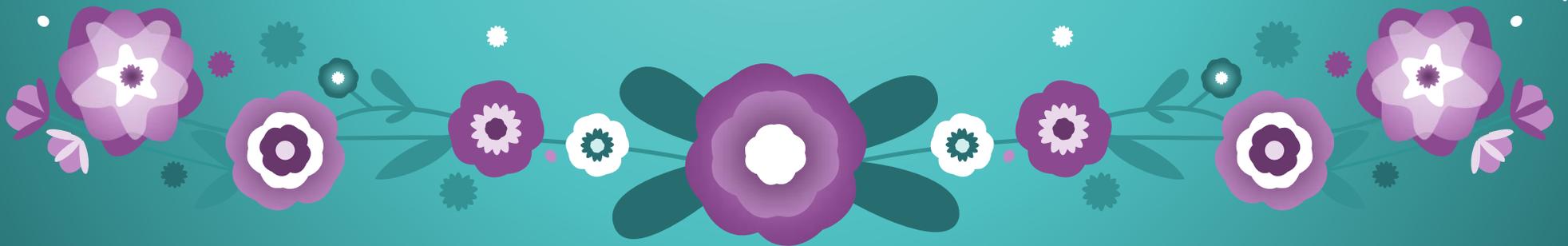
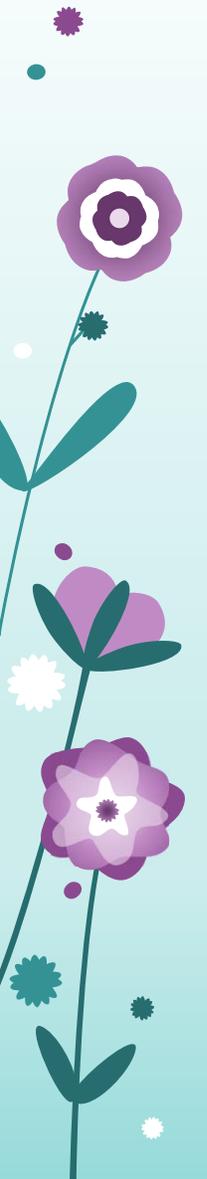


Intestinal Obstruction in Children

By Raed Al-Taher, M.D.

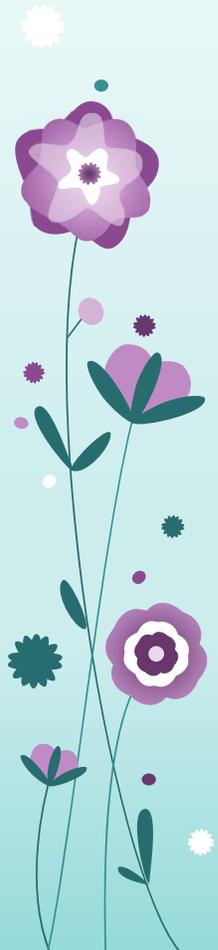


Malrotation



Malrotation

A spectrum of anomalies of rotation and fixation of the intestines, principally involving the midgut.



Malrotation

- Incidence ~1%
- Causes <5% of intestinal obstruction during childhood.
- Most (50–70%) are diagnosed in the neonatal period.
- M = F

Malrotation

Associated with:

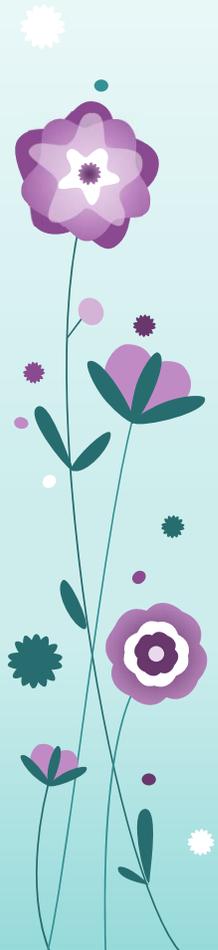
- Gastroschisis and omphalocele (exomphalos)
- Diaphragmatic hernia
- Duodenal atresia and biliary atresia
- Intussusception (Waugh's syndrome)
- Dysmotility and pseudo-obstruction syndromes

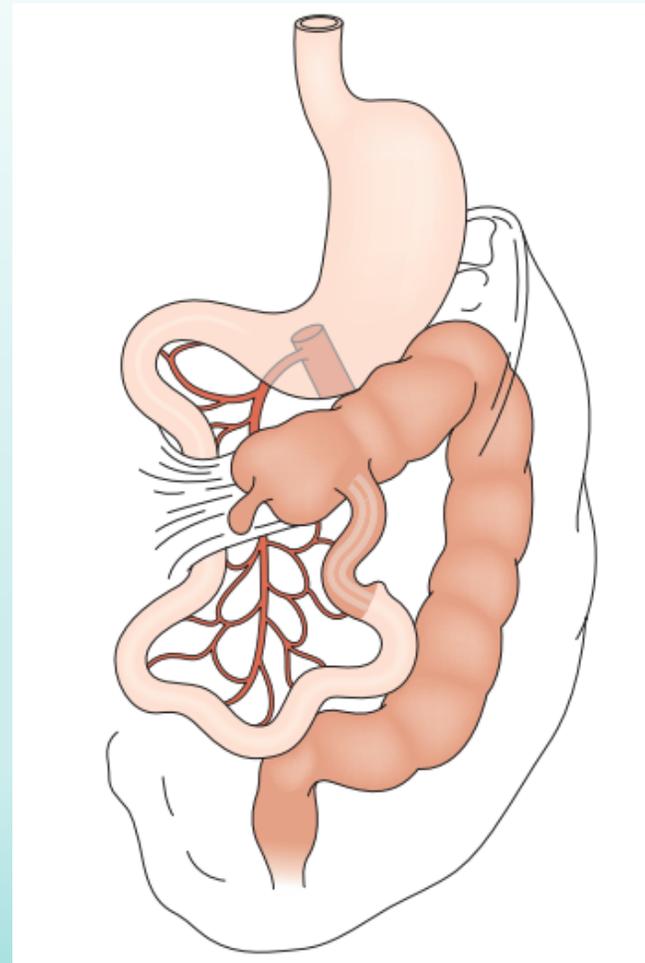
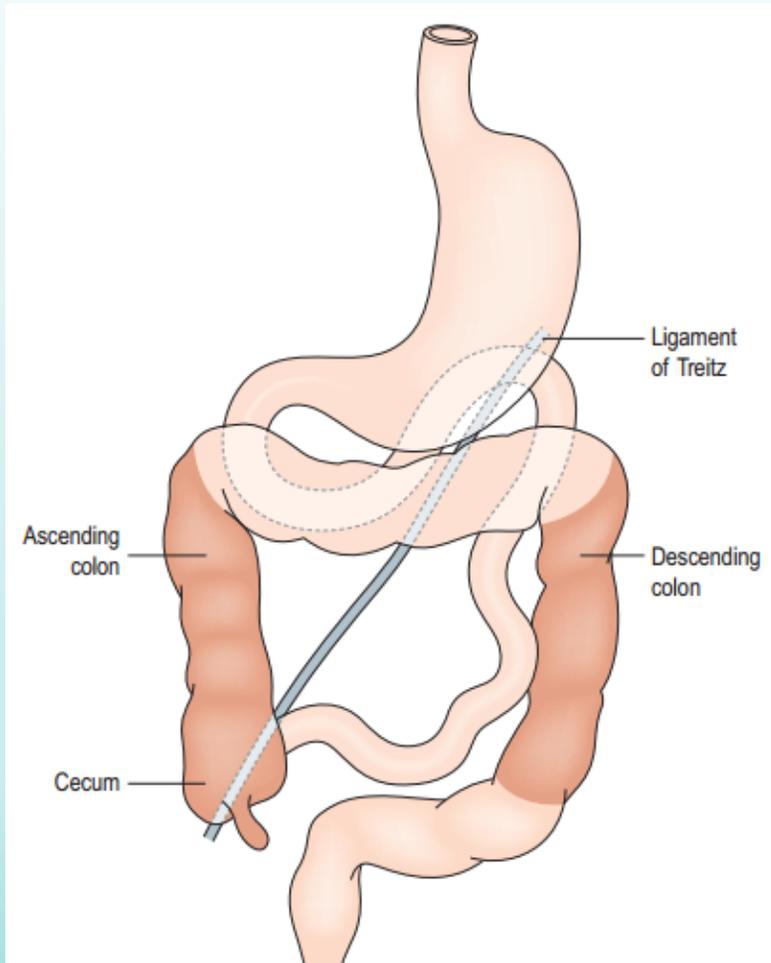
Malrotation

- Occurs when the normal process of rotation is not complete or is erroneous.
- The commonest variant:
 - failure of the final 90° anticlockwise rotation taking the cecum from the right upper quadrant to the right iliac fossa.
 - Cecum is fixed to the retroperitoneum by peritoneal bands running anteriorly to the second part of duodenum (Ladd's bands).

Malrotation

- The key pathology is:
 - The distance between the two ends of the small bowel mesentery (i.e., distance between DJ junction and IC valve).
- when diminished, then risk of volvulus increases.

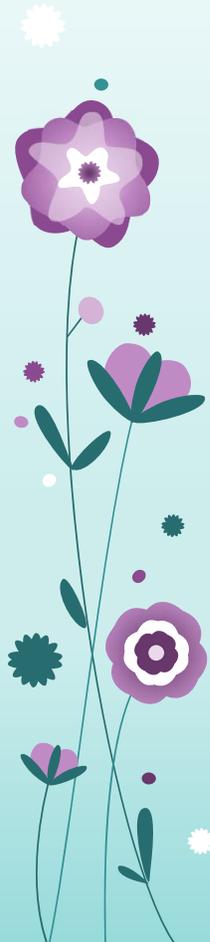




Malrotation

Clinical Features

- Can present at any age.
- Classic presentation: an infant with bile vomiting (~50%) due to duodenal obstruction (extrinsic due to Ladd's bands or by virtue of the twist of the volvulus).
- If delayed, then features are less specific but may include:
 - non-bile vomiting
 - intermittent or acute abdominal pain
 - diarrhea then constipation
 - failure to thrive
 - passage of altered blood
- Chronic midgut volvulus (<10% of cases) may cause mesenteric thickening, with lymphatic obstruction leading to chylous ascites and malabsorption.



Malrotation

Clinical Features

On physical examination:

- the abdomen is soft and nontender unless volvulus and bowel strangulation has occurred (~25% of all cases) leading to abdominal distension, tenderness, and blood-stained stools.
- 

Malrotation

Investigations

1. AXR

- probably “normal” in most cases.
- abnormal features may include:
 - Malposition of the bowel (“small bowel” to the right and “colon” to the left)
 - Lack of distal bowel gas (“gasless” abdomen or a “double bubble” appearance)
 - “Whirled” appearance of mid-abdominal bowel
 - Thick-walled, tubular bowel loops (suggesting chronic volvulus)

2. Upper GI contrast study

- investigation of choice (if time permits!)

Contrast studies in malrotation

	Normal features	Abnormal features
Overall	“C” shaped duodenum	Redundant duodenum, right-sided jejunal loops
Lateral appearance	Overlapping, posterior position of second and fourth parts	Anterior position of fourth part
Duodeno-jejunal flexure	Left of spinal pedicles; rising to at least level of L1/L2 disk space	Right-sided or centrally placed. Failure of ascent to level of pylorus

Malrotation

Management

- If volvulus is likely (or even a possibility), then every minute counts and the child needs urgent laparotomy for intestinal detorsion.
- Other elements to be considered meanwhile:
 - rapid fluid resuscitation
 - NG aspiration
 - correction of acid/base imbalance
 - IV broad-spectrum antibiotic
 - inotropic agents

Malrotation

Management: Surgery (Ladd's Procedure)

Untwist a volvulus and..

- a) Assess intestinal damage
- b) Prepare for reperfusion syndrome (hypotension, $\uparrow K^+$, \uparrow lactate)
- c) Resection (if unequivocal necrosis) \pm anastomosis (if safe)
- d) If, equivocal necrosis and short bowel is likely then consider leaving "ischemic" gut alone and perform second look laparotomy in 24–36 h.

Malrotation

Management: Surgery (Ladd's Procedure)

- The aim is to leave the midgut in a position of complete “non-rotation” with..
 - duodenum and small bowel on right
 - apex of midgut and hence SMA centrally
 - cecum and large bowel on left
- This achieves the widest possible base to the mesentery thus reducing propensity to volvulus.

Malrotation

Management: Surgery (Ladd's Procedure)

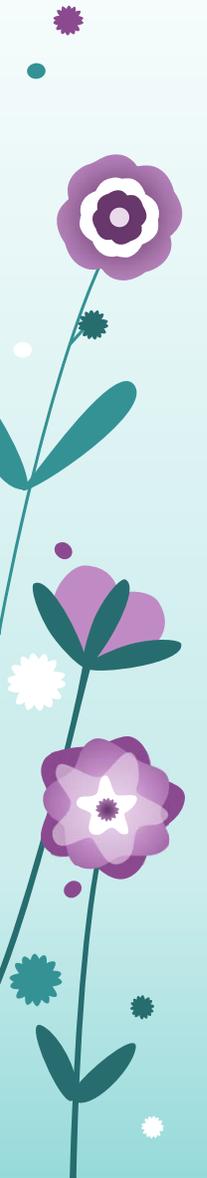
1. Division of Ladd's bands (lying across the duodenum from abnormal caecum in RUQ).
2. Widen mesenteric base (divide peritoneum overlying central mesenteric vessels).
3. Position bowel (small bowel right and large bowel left).
4. \pm Appendectomy

Malrotation

Outcome and Complications

- Midgut infarction (<5%)
- Recurrence of midgut volvulus post-Ladd's procedure (<2%)
- Adhesional intestinal obstruction (5%)

Intestinal Atresia



Intestinal Atresia

- 1 in 3,000 | black > white | jejunoileal
- 1 in 5,000 | F>M (slight) | duodenal
- 1 in 50,000 | colonic

Jejunoileal > duodenal > colonic

Intestinal Atresia

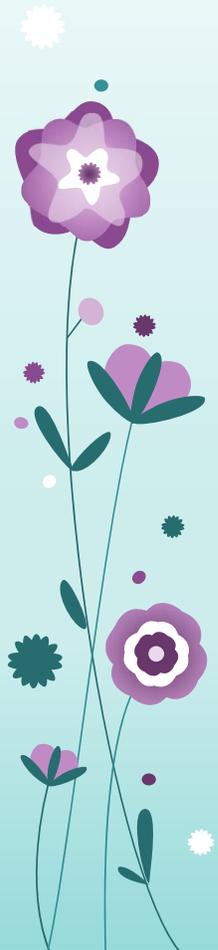
Associations

- Pyloric atresia
 - Epidermolysis bullosa
 - HMIA (Hereditary multiple intestinal atresia)
- Duodenal atresia
 - Trisomy 21, Down syndrome
 - Esophageal atresia
 - Malrotation
 - Cardiac anomalies
 - Vertebral anomalies
- Jejunal ileo colic atresia
 - Abdominal wall defects (esp. gastroschisis)
 - Maternal glomerulonephritis
 - Maternal cocaine use
 - Hirschsprung's disease (colon atresia)

Intestinal Atresia

Pathology

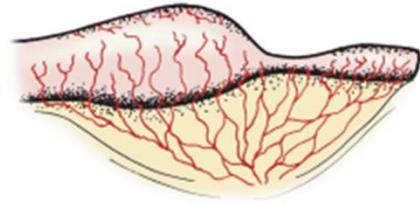
- Mostly unknown.
- **Theory:** intrauterine vascular insult to intestinal segment after being completely developed. Hence, meconium can be found in distal bowel beyond the site of obstruction.



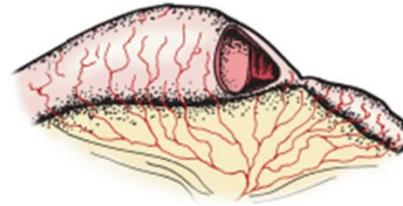
Intestinal Atresia

Classification (applicable to all parts of the intestine)

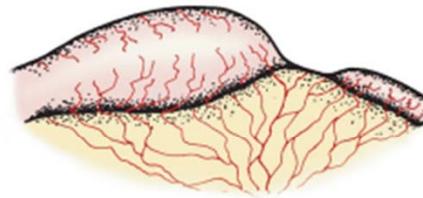
- Type I – Membrane or web
- Type II – Fibrous cord joins two blind ends of bowel
- Type III
 - IIIa – Gap between ends with a V-shaped mesenteric defect
 - IIIb – Large defect in the mesentery, significant intestinal loss and distal intestine winds round a single, tenuous vascular pedicle (“apple-peel” or “Christmas tree” atresia)
- Type IV – Multiple atresias (“string of sausages” appearance)



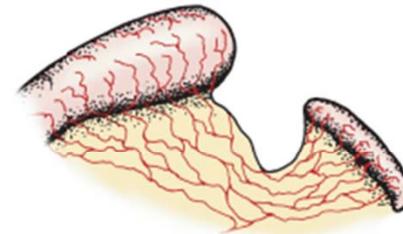
Stenosis



Type I



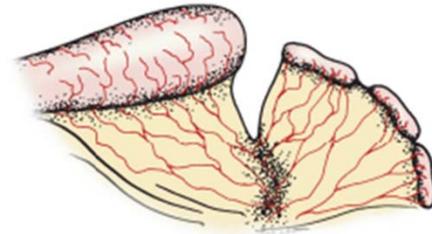
Type II



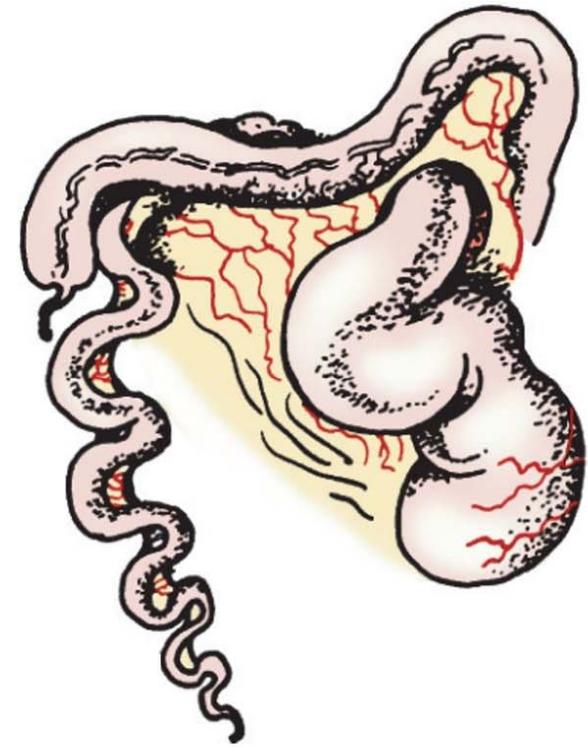
Type III (a)



Type III (b)



Type IV



Jejunal atresia – type IIIb
“Apple-peel” atresia complicated by necrosis.

Intestinal Atresia

Hereditary Multiple Intestinal Atresia (HMIA)

- Sporadic or familial condition with multiple atretic segments (from pyloric atresia down, often type 1, and >20).
- Associated with immunodeficiency and a dilated biliary tree.
- Almost uniformly fatal (survivors have often had both small bowel and bone marrow transplants).

Intestinal Atresia

Clinical Features

- Antenatal:
 - **US may show:**
 - polyhydramnios (↑ with the more proximal atresias).
 - “double bubble”
 - dilated proximal loops
 - echogenic bowel
- Postnatally:
 - **Bile-vomiting**
 - **Varying degrees of distension** (depending on level of obstruction)
 - **Delay in passage of meconium**

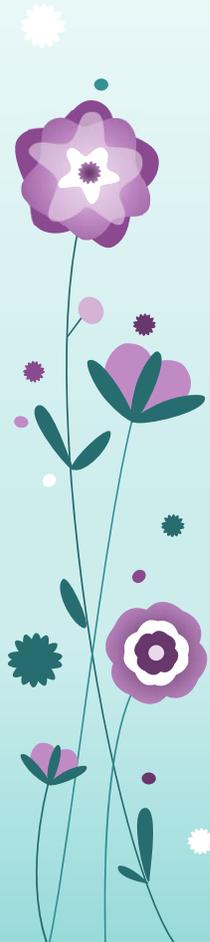
Intestinal Atresia

Investigations

- **AXR**
 - Features of obstruction (e.g., dilated bowel loops and absence of distal gas).
 - “Double bubble” and no distal gas (classical feature in duodenal obstruction).
 - More loops “bubbles” will be visible with distal obstructions.
 - Colon atresia (characteristic picture is a single, grossly dilated right-sided loop with a fluid level. This represents the obstructed colon with a competent ileo-cecal valve – a closed loop obstruction).
 - Intrauterine perforation and meconium cyst formation may be suggested by peritoneal calcification.
- **Contrast enema**
 - may show a microcolon and is also helpful in ruling out other causes (e.g., Hirschsprung disease, meconium plug).



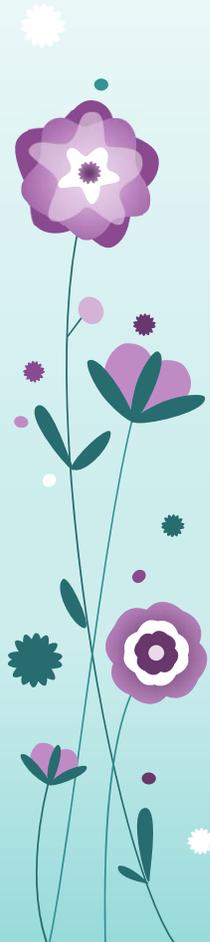
“Double bubble” and no distal gas
(classical feature in duodenal atresia)



Intestinal Atresia

Differential diagnosis

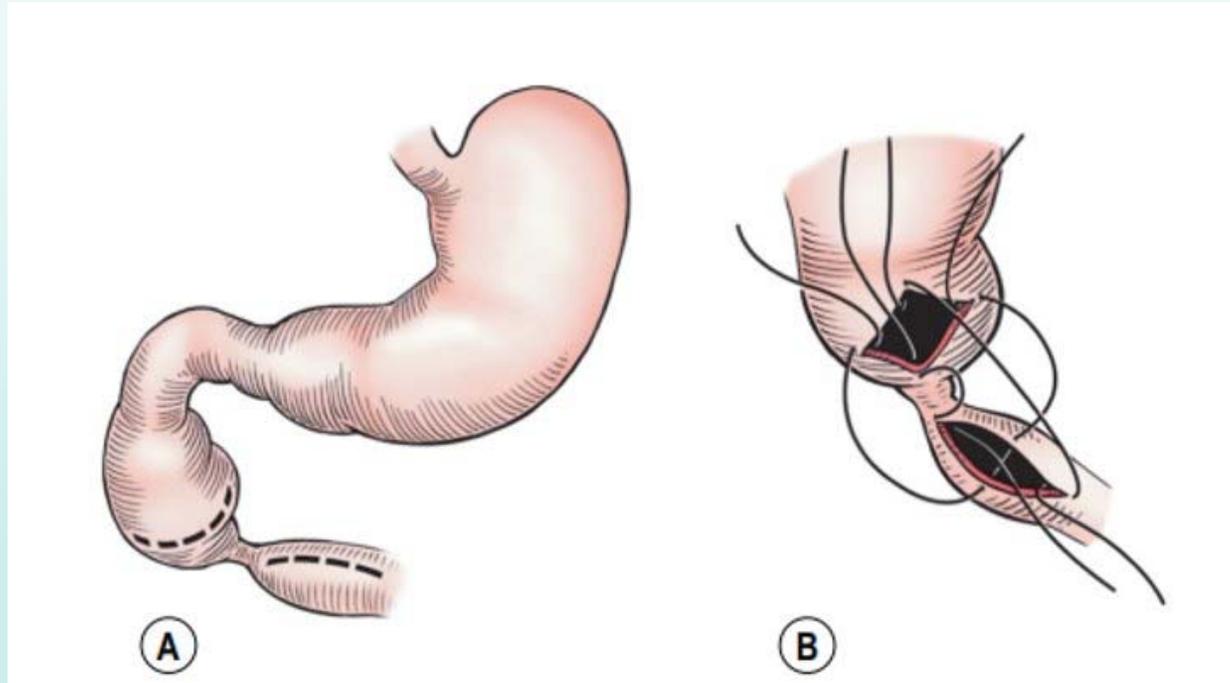
- Meconium ileus (“soap-bubble” sign, no fluid levels)
 - Hirschspung’s disease (↑ dilated bowel loops, transition zone on contrast enema)
 - Malrotation (double bubble ± distal gas).
- 



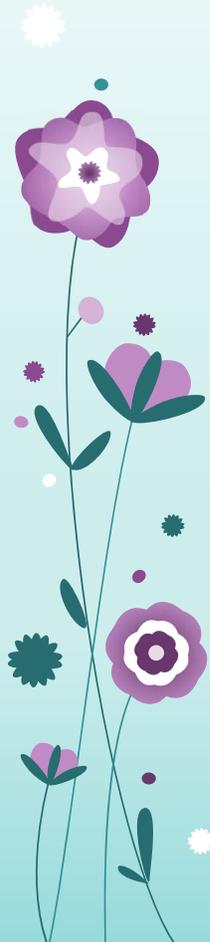
Intestinal Atresia

Surgery: Duodenal Atresia (Duodeno-duodenostomy)

1. Recognize and treat any associated malrotation.
 2. Approximate two ends.
 - Transverse incision proximal end.
 - Longitudinal incision distal end.
(Caution with area of ampulla)
- 



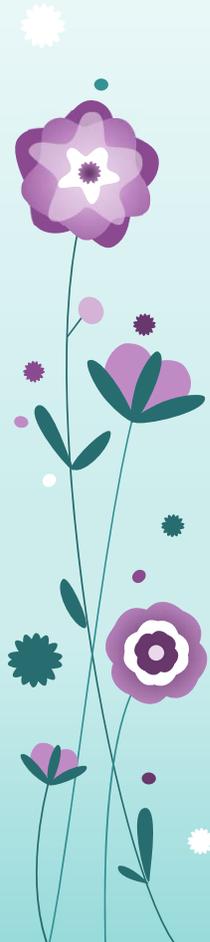
Duodeno-duodenostomy Procedure



Intestinal Atresia

Surgery: Jejunio-ileal Atresia

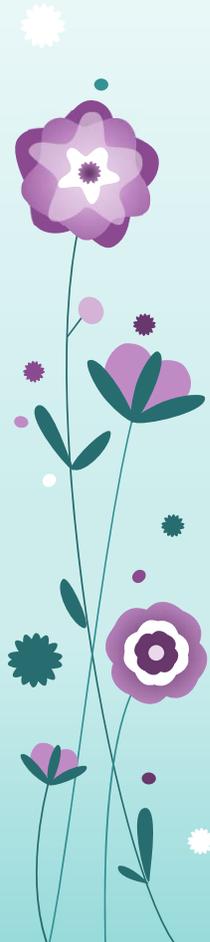
1. Assess viability and length of residual bowel.
 2. Assess patency of distal bowel lumen (look for other atretic segments).
 3. Resection of atretic segment(s) and re-anastomosis.
- 



Intestinal Atresia

Surgery: Colon Atresia

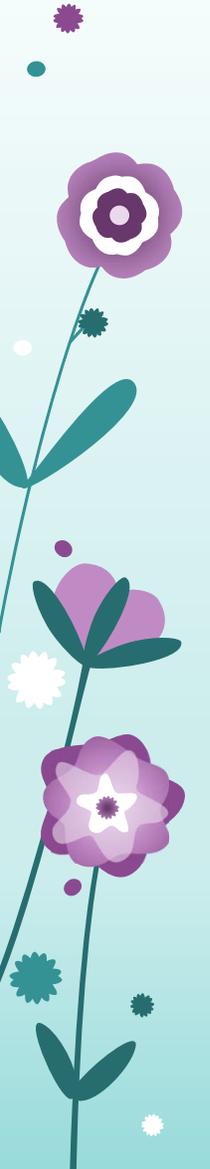
- Usually the site of atresia is two-thirds along the transverse colon.
 - Operative options include:
 1. Primary anastomosis (right hemicolectomy and ileo-transverse colostomy)
 2. Defunctioning colostomy and staged anastomosis
- 



Intestinal Atresia

Outcome

- Dependent on:
 - associated anomalies
 - length of residual small bowel
- 



Necrotizing Enterocolitis

(NEC)



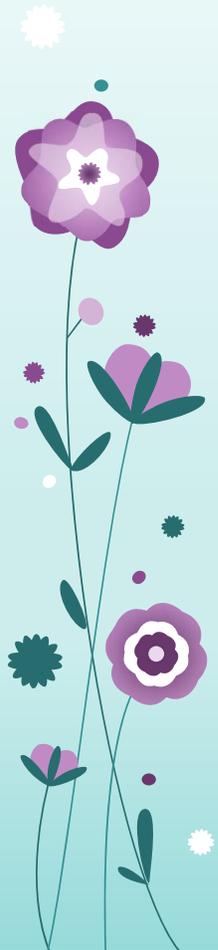
Necrotizing Enterocolitis

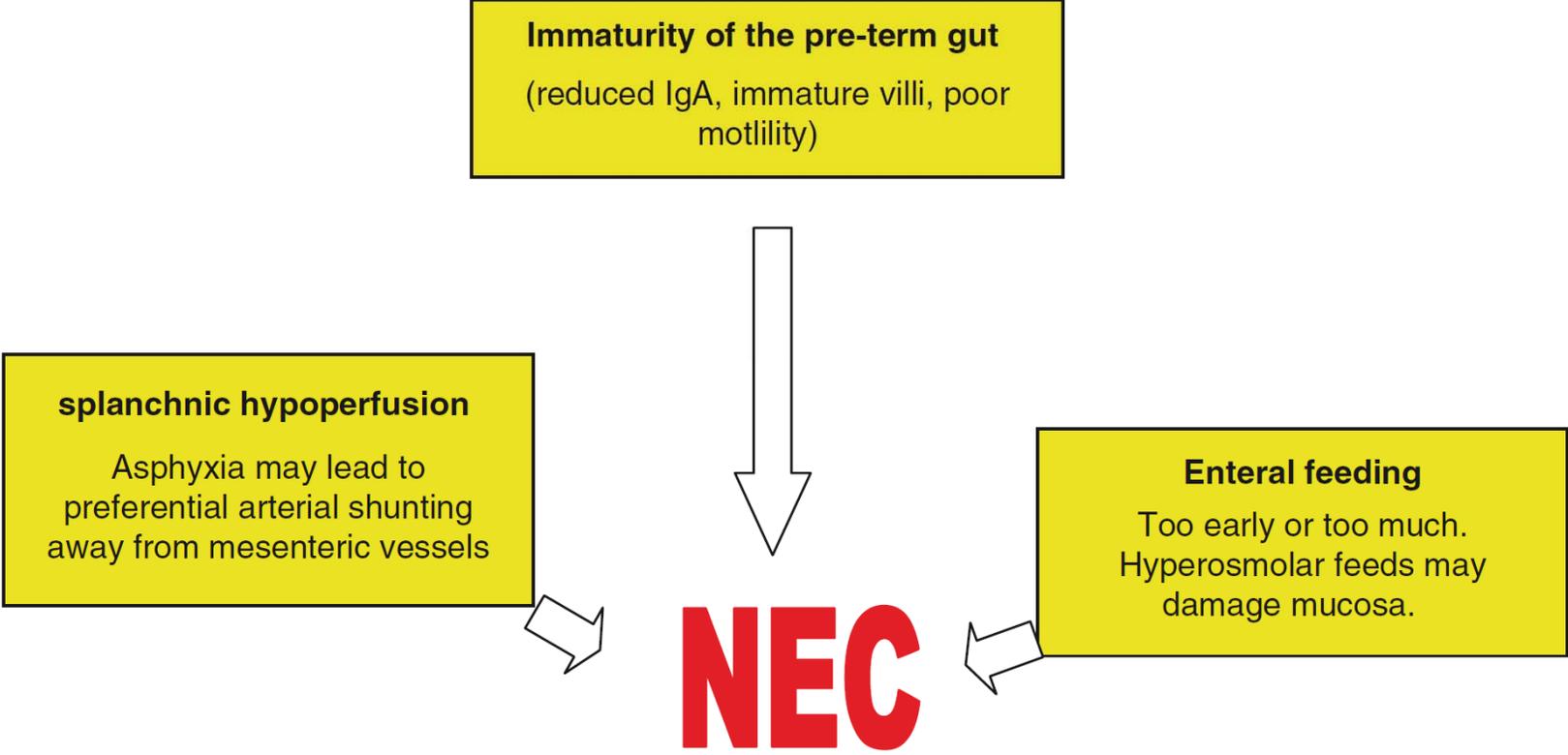
- 1–3/1,000 live-births | M:F = 2:1 | Black > white
- >90% occur in preterm infants
- Term infants – associated with congenital heart disease or birth asphyxia
- Reduced incidence in breast vs. formula-fed infants

Necrotizing Enterocolitis

Pathophysiology

- Multifactorial; seems a unique response in the immature, neonatal gut to “stress”.
- NEC may be focal or diffuse.
- Most commonly affected sites: terminal ileum and colon.





Possible causal factors in NEC

Necrotizing Enterocolitis

Pathophysiology

- Submucosal and subserosal gas-filled cysts may be obvious (N_2 or H_2), from action of gas-forming bacteria.
- Histologically:
 - Mucosal ulceration and transmural coagulative necrosis and gangrene (may result in perforation).
 - Microthrombi in mesenteric vessels.

Necrotizing Enterocolitis

Clinical Features

- Age of onset: between 7 and 10 days (inversely proportional to gestational age at birth).
- First-week NEC may be predisposed by:
 - maternal drug use
 - perinatal sepsis
- First-week NEC may be predicted by changes in umbilical artery Doppler characteristics.

Necrotizing Enterocolitis

Clinical Features

- **Nonspecific signs** related to sepsis and ischemia:
 - Tachycardia
 - Hypotension
 - Metabolic acidosis
 - Unstable body temperature
 - Increasing O₂ requirement
 - Thrombocytopenia
 - Coagulopathy
- **Specific local signs** related to the affected bowel loops:
 - Peritonism
 - Abdominal wall erythema
 - Bile-vomiting
 - GI bleeding
 - Abdominal mass formation



Abdominal wall erythema

Bell's Staging of NEC

	Clinical	Radiology
I – suspected	Irritable, apnea, bile-aspirates, abdo. Distension, +ve fecal occult blood	Intestinal distension
II – definite	GI bleeding ± Abdo. mass	Pneumatosis, “fixed” loops, portal venous gas
III – advanced	Shock, marked GI bleeding	Pneumoperitoneum, severe ascites

Necrotizing Enterocolitis

Investigations

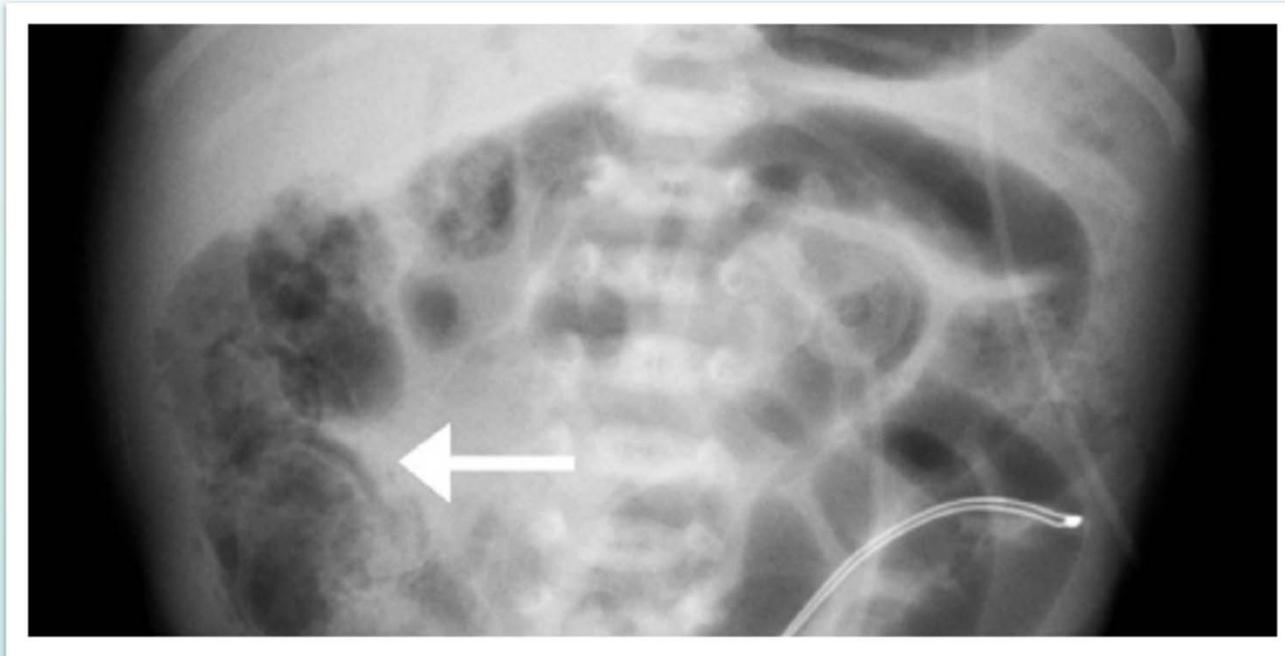
1. Blood work-up for signs of sepsis and intestinal necrosis
 - a) CBC (esp. platelet count)
 - b) Coagulation screen (INR, fibrinogen, D-dimers)
 - c) Lactate and arterial blood gases

Necrotizing Enterocolitis

Investigations

2. Radiology (supine AP \pm cross-table lateral)

- a) **Early signs – nonspecific distension**
- b) **Late signs:**
 - a) Pneumatosis (seen as linear radiolucent bands parallel to the wall of the bowel or “soap-bubble” appearance)
 - b) Portal venous gas (also detectable on US)
 - c) Extravisceral free air (most commonly seen between the liver and the diaphragm and anteriorly outlining falciform ligament “football sign”).
 - d) “Ground-glass” appearance – suggests free fluid.

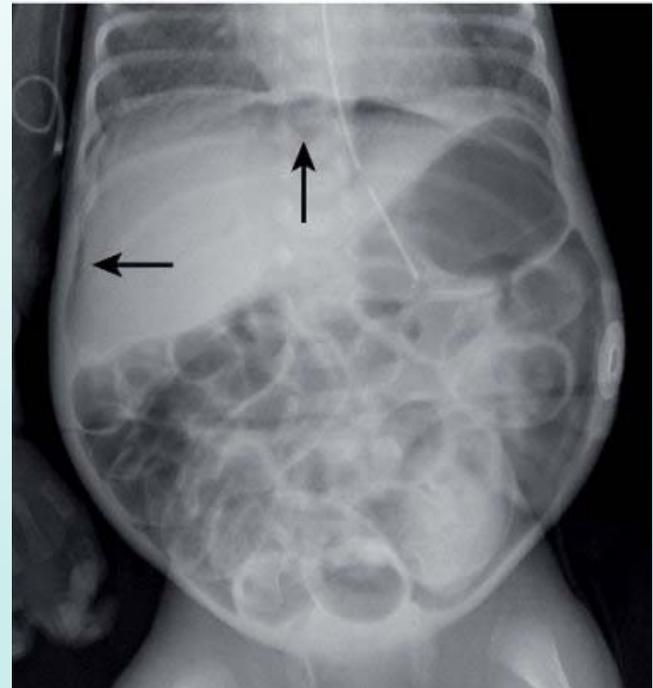


Pneumatosis Intestinalis

seen as linear radiolucent bands parallel to the wall of the bowel or “soap-bubble” appearance



Portal venous gas



Pneumoperitoneum
(Extravisceral free air)

Necrotizing Enterocolitis

Focal intestinal perforation

- different from **NEC**
- present with pneumoperitoneum without the associated necrosis
- good prognosis

Necrotizing Enterocolitis

Management

- The initial strategy should be supportive and aimed at resting the gut and optimizing the infant's hemodynamic and metabolic condition. This involves:
 - Nil by mouth (NPO) and naso-gastric tube
 - Restoration of..
 - fluid losses (including third space losses)
 - hematocrit
 - temperature instability
 - glucose instability
 - acid–base imbalance
 - Appropriate oxygenation
 - Broad-spectrum antibiotics
 - Analgesia

Necrotizing Enterocolitis

Management

- Monitoring should be combined with **serial radiography** to assess onset of complications (e.g., perforation).
- If improving, then continue for 7–10 days before restarting enteral nutrition.

Necrotizing Enterocolitis

Management

Indications for Surgery:

- Pneumoperitoneum (perforation) (Stage III Bell's)
- Failure to progress (after 24 h of full, supportive management)
- Obstructive features (↑ distension, ↑ bile-aspirates)
- “Fixed-loop” on serial imaging
- ↑ Abdominal wall erythema
- Palpable abdominal mass

Necrotizing Enterocolitis

Management

Surgery:

1. Assess site (ileocecal etc.), extent (focal, multifocal), and degree (ischemia, necrosis, perforation).
 - “Pan-intestinal” implies majority of small and large bowel is involved with <25% of viable bowel remaining.
2. Possible interventions include:
 - **Resection** (focal necrotic areas with either anastomosis (if good condition) or stomas (if not)).
 - **Proximal stoma only** (if grossly inflamed adherent RIF mass, makes resection dangerous).
 - “Clip and drop” technique (for pan-intestinal disease) (i.e., rapid resection of obviously dead areas with application of surgical clips to close separate ends. Then after 48 h, second-look laparotomy to fashion stoma and if appropriate re-anastomose distal salvaged segments).



“Pan-intestinal” NEC

Necrotizing Enterocolitis

Management

Other surgical option (esp. if unstable):

- **Primary peritoneal drainage (PPD)**
 - drain is inserted at cot-side under local anesthesia
 - temporizing measure for pneumoperitoneum (alternative to laparotomy)
 - can be the definitive treatment



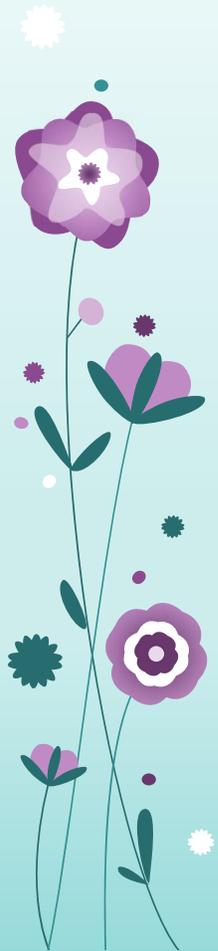
Part of surgical gloves
used as a drain for open
peritoneal drainage

Primary peritoneal drainage (PPD)
in an ELBW newborn

Necrotizing Enterocolitis

Outcome and Complications

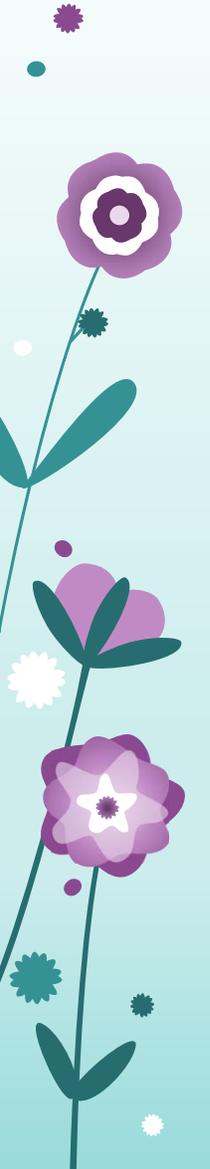
- Mortality ranges from 20 to 50% (higher in VLBW and ELBW)
- Recurrence (~5%) (usually within a month of initial presentation)
- Short-gut syndrome (~25%)
- Strictures (20%)



Necrotizing Enterocolitis

Possible Prevention Strategies (little evidence yet)

1. Breast milk (fourfold reduction in NEC incidence vs. formula)
2. Avoidance of indomethacin/ibuprofen/ranitidine and maintenance of gastric acidity
3. Probiotics (e.g., lactobacillus/bifidobacterium)
4. Oral antibiotics
5. Oral/IV immunoglobulin (IgA/IgG)
6. Amino-acid supplementation (glutamine)



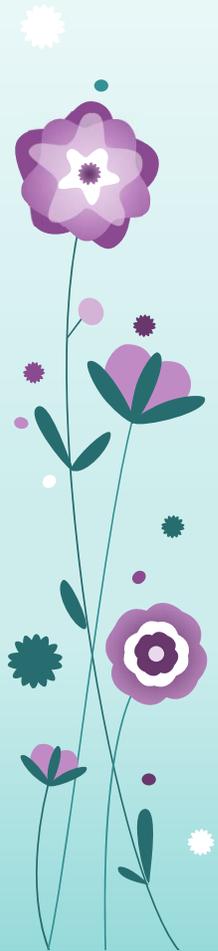
Meconium Ileus

(MI)



Meconium Ileus

Distal ileal intraluminal obstruction due to presence of abnormally viscid meconium.



Meconium Ileus

Meconium

- Normal meconium: contains bile pigments, desquamated epithelia and is black.
- CF meconium:
 - ↑↑ albumin
 - ↓ carbohydrate
 - ↓(HCO₃⁻)
 - ↓ fluid content resulting in ↑↑viscosity
 - There is also abnormal intestinal motility and mucin production

Meconium Ileus

- M=F
- ~90% of infants presenting with meconium ileus (MI) will have cystic fibrosis (CF).
- ~15% of CF patients will present with MI.
- MI is the cause of ~20% of neonatal intestinal obstruction.

Meconium Ileus

Cystic Fibrosis

- Most common (carrier 1 in 25) recessive disease in Caucasians.
- 1 in 3,000 live-births.
- Mutation in the CFTR (CF Transmembrane-conductance Regulator) gene – Ch 7 (q 31.2).
- Chloride channel defect:
 - Respiratory epithelium → viscid secretion and impairment of ciliary mucus clearance → bacterial colonization.
 - Causes ↑ (Cl⁻) in sweat.
 - Causes ↓ (Cl⁻) GI tract, pancreas, and liver → ↑ viscosity secretions → exocrine gland blockage.
 - Luminal obstruction (vas deferens).

Meconium Ileus

Clinical Features

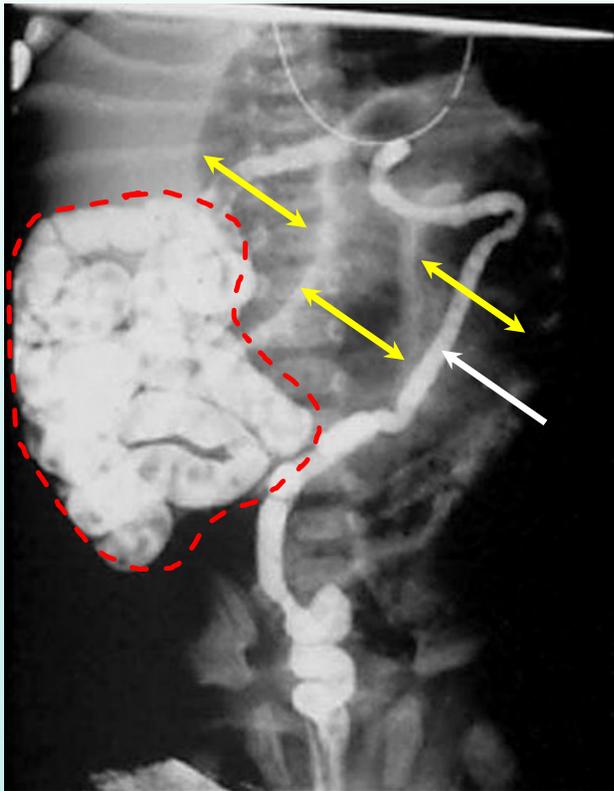
- Antenatal period:
 - Hyperechogenic dilated bowel, or nonvisualization of the gall bladder on US

(N/B. these features also seen in Down' syndrome, intestinal atresia, and the normal fetus → so, parental genotypes ± amniocentesis should be done to confirm dx)
- **MI** has been classified into **two types**:
 - Simple MI:
 - abdominal distension, and bile vomiting
 - failure to pass meconium
 - “Doughy,” palpable bowel loops on examination
 - Complicated MI:
 - Intrauterine perforation → meconium pseudocyst
 - Intestinal atresia
 - Local volvulus (usually ileum) → gangrene

Meconium Ileus

Investigation

1. AXR:
 - Dilated loops of bowel with a coarse granular (“soap-bubble”) appearance "Neuhauser's sign"
 - Calcification may also be seen in meconium peritonitis.
2. Contrast enema
 - will show micro-colon (may contain small pellets of mucus)
3. Postnatal US – if there is a palpable mass (meconium cyst).
4. Confirmation of CF:
 - a) Sweat-Chloride test (pilocarpine iontophoresis) (>1 month postnatal) (normal <40, diagnostic ≥ 60 mmol/L).
 - b) Gene mutation analysis.
 - c) Immunoreactive trypsinogen (basis for screening - $\uparrow\uparrow$ levels in CF).



Contrast Enema of two MI cases

- Micro-colon (contains small pellets of mucus – filling defects) [white arrow]
- Dilated proximal small bowel loops filled with gas (black shadow) [yellow arrows]
- Distal small bowel filled with inspissated meconium (“soap-bubble” appearance or “Neuhauser’s sign”) [red dash-lines]

Meconium Ileus

Management

- Water-soluble contrast enema (in absence of complications):
 - Gastrografin® ~1,900 mOsm/L (x5 osmotic pressure than normal)
 - Success 60–70% in simple MI
 - Bowel perforation rate ~3%

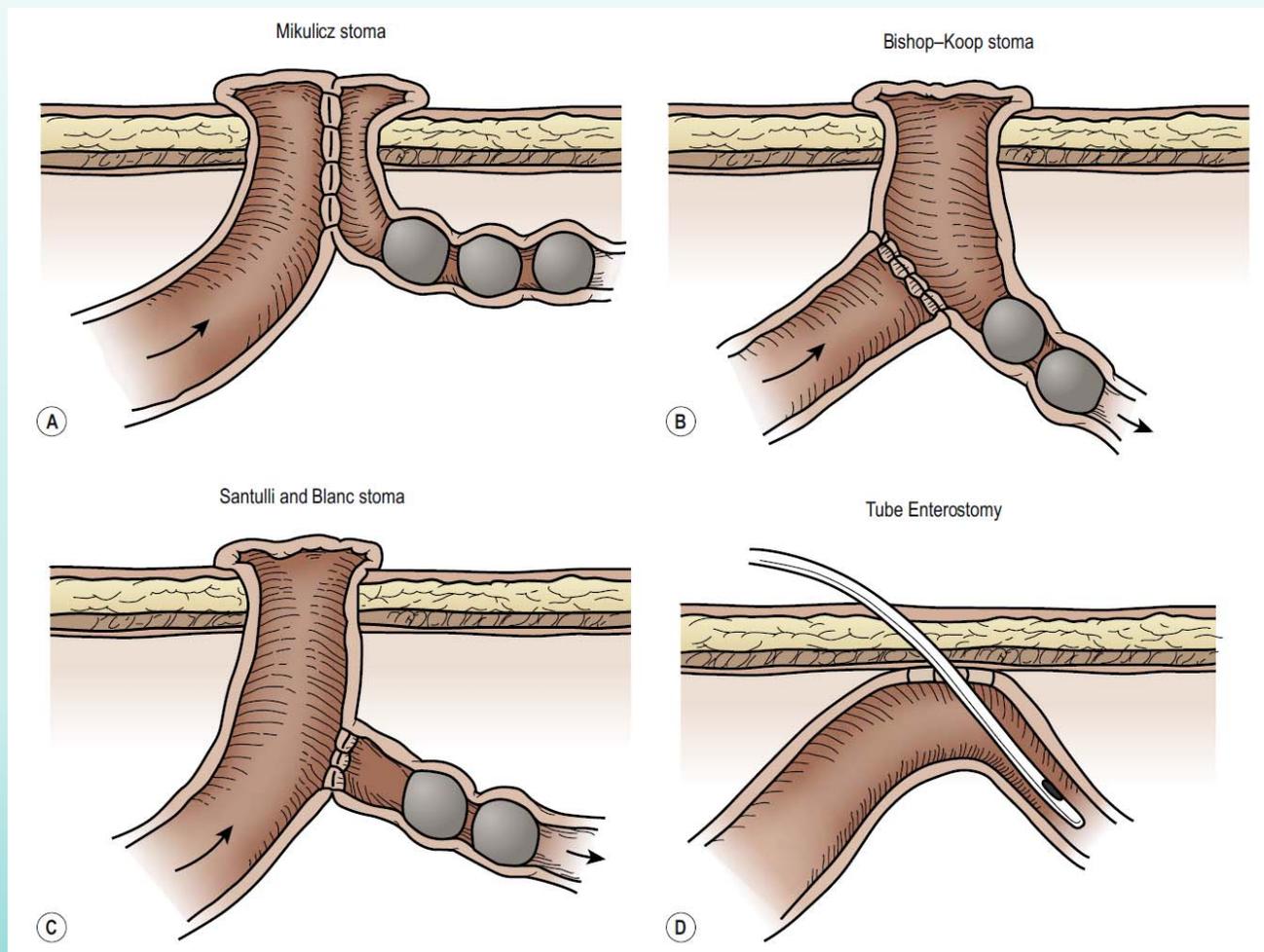
Meconium Ileus

Management

Surgery

- Simple MI:
 - Proximal ileum is dilated. Distal ileum is relatively narrow and filled with inspissated meconium. Colon is narrow (micro-colon).
 - Ileotomy and passage of catheter (proximal to the inspissated meconium), using N-acetylcysteine or normal saline and massage to break up obstruction.
 - Options then include:
 - Simple closure and return
 - Enterostomy tube
 - Double-barrelled Mikulicz ileostomy
 - Bishop-Koop ileostomy
 - Santulli ileostomy

Variations in ileostomy for meconium ileus



Meconium Ileus

Management

Surgery

- Complicated MI:
 - ± Resection of ischemic bowel (or cyst, atretic segment, etc.)
 - Diverting stoma or sometimes primary anastomosis

Meconium Ileus

Management

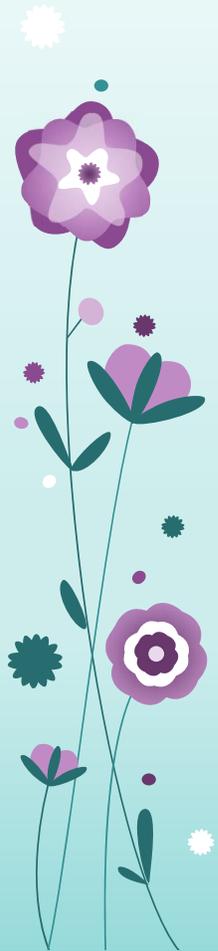
Surgery

- Postoperative care:
 - Parenteral nutrition
 - N-acetylcysteine (10%) enterally (5–10 mLs) if persisting functional obstruction
 - Enteral pancreatic enzymes (e.g., Creon®, Pancrease®)
 - Antibiotics
 - Involvement of CF team

Meconium Ileus

Outcome

- With aggressive respiratory care and improved nutritional awareness most children born today will expect to live into their adult years.
- Current average life expectancy ~35 years

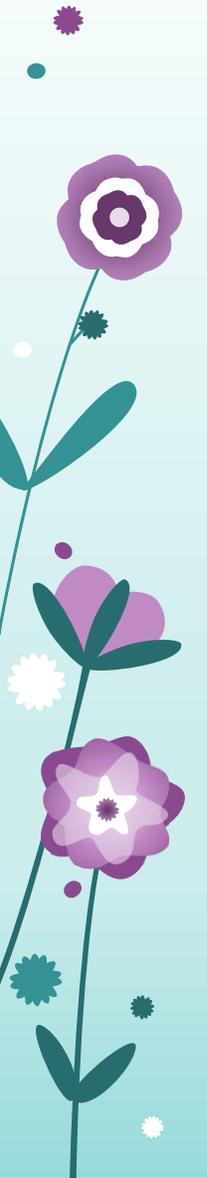


Meconium Ileus

Meconium Plug Syndrome (MPS)

- Intraluminal colonic obstruction with heterogeneous etiology.
- Associations:
 - Maternal factors (e.g., eclampsia and diabetes)
 - Neonatal factors (e.g., sepsis, prematurity, hypothyroidism, and neonatal intestinal dysmotility)
 - Specific (20%):
 - Hirschsprung's disease and CF
 - Small left colon syndrome (radiological entity with transitional zone at level of splenic flexure, associated with maternal diabetes. ± Histological features of immature ganglion cells).
- Neonates present with features of intestinal obstruction, with a differential of meconium ileus and Hirschsprung's disease.
- The key investigation is the diagnostic contrast enema followed by a therapeutic water-soluble contrast enema (>95% successful).
- Further investigations:
 - genotype and sweat test
 - ± rectal suction biopsy

Hirschsprung's Disease



Hirschsprung's Disease

- 1 in 5,000 live-births
- >90% of cases diagnosed in the neonatal period
- Two distinct clinical types with genetic differences:
 - *Short segment* (recto-sigmoid) 75% (M:F 4:1)
 - *Long segment* 25% (M = F)

Hirschsprung's Disease

Associated anomalies (variable incidence ~10%)

- **Down's syndrome (~5%)**
- Neurocristopathies as:
 - *Waardenburg-Shah* syndrome; white forelock, bicolored iris, deafness
 - *Hypoventilation syndrome* (Ondine's curse) – association with HD termed Haddad syndrome
- Mental retardation syndromes
 - *Smith-Lemli-Optiz* syndrome; mental retardation, polydactyl, defect in cholesterol metabolism
 - *Mowat-Wilson syndrome*; mental retardation, characteristic facies
- **Development colon anomalies**
 - Colon atresia, anorectal atresia
- Miscellaneous
 - *Kaufman-McKusick syndrome*; hydrometrocolpos, hypospadias, polydactyl

[N.B. MEN type 2B (Marfanoid habitus, medullary thyroid cancer, café au lait spots, mucosal neuroma) is associated with *hyperganglioneosis* (functionally similar to HD)]

Hirschsprung's Disease

Embryology

- Migration of neuroenteric cells from the neural crest to GI tract:
 1. Esophagus 5th week
 2. Mid-gut 7th week
 3. Distal colon by 12th week
- Some studies suggest that ganglion cells are guided to their destination by neural glycoproteins or fibers (e.g., fibronectin, hyaluronic acid).

Hirschsprung's Disease

Anatomy

- The normal intestine contains two distinct nerve plexi, between three muscle layers (longitudinal, circular, muscularis mucosae).
 1. Submucosal plexus (of Meissner)
 2. Myenteric or intermuscular plexus (of Auerbach)
- Each plexus contains a fine meshwork of neurons (ganglion, CD55^{+ve}) and supporting (glial, CD55^{-ve}) cells which control motility, absorption, secretion and blood flow.

Hirschsprung's Disease

Anatomy

- Ganglion cells (nested in groups of four to six cells) receive extrinsic cholinergic and adrenergic signals:
 1. **Intrinsic neuron stimulation causes muscle relaxation.**
 - a) Nitric oxide (NO) is prime mediator
 - b) Other mediators include VIP, Histidine, substance P, Neurokinin A, Enkephalin, Gastrin release peptide, isoleucin, and many others.
 2. **Extrinsic.**
 - a) Cholinergic neurons (contraction)
 - b) Adrenergic neurons (relaxation)
 3. **Nonadrenergic and noncholinergic (NANC) nervous system.**
 - a) Controlled by interstitial cells of Cajal also seem to play an important role in peristalsis.

Hirschsprung's Disease

Etiology (hypotheses)

1. Failure of migration

- a) Distal aganglionosis occurs in chick embryos, when the hind gut is transected.
- b) Abnormal glycoproteins have been found in the distal aganglionic gut.

2. Hostile environment

- a) Loss of neural cell adhesion molecules (NCAM) leads to inability of normal ganglion cells to adhere to smooth muscle cells.

3. Immunologic attack

- a) Abnormal immune response mounted by fetus against ganglion cells may lead to destruction of ganglion cells.

Hirschsprung's Disease

Pathology

- Lack of progression of peristaltic wave into the aganglionic segment of intestine and absent or abnormal internal anal sphincter relaxation is the hallmark of HD.
- The gross appearance of intestine varies with age of the child.
 - In the neonatal period the proximal intestine may appear normal but with the passage of time the proximal intestine distends and hypertrophies.

Hirschsprung's Disease

Variable Affected Segment

- Short segment (recto-sigmoid)
- Long segment:
 - Total colonic +/- ileal involvement
 - Total intestinal aganglionosis (incompatible with life)
- Ultra-short segment disease (rare)
- Segmental disease or “skip” lesions (extremely rare)

Hirschsprung's Disease

Genetics

- Strong evidence of genetic predisposition.
- Risk in siblings is 3–4% (↑ with long segment disease).
- Gene mutation (50% familial and 15–35% isolated)
 - **RET gene** (Ch 10q11 | associated with Down's syndrome)
 - Other genes (SOX10, EDNRB, GDNF , EDN3, ECE1, NTN, SIP1).

Hirschsprung's Disease

Clinical Features

Two overlapping scenarios

1. Neonatal bowel obstruction:

- Delayed passage of meconium
- Abdominal distension
- Bile vomiting
- ± Enterocolitis (variable incidence)

2. Chronic constipation (not encopresis/soiling)

- ± Enterocolitis (variable incidence)
- Failure to thrive

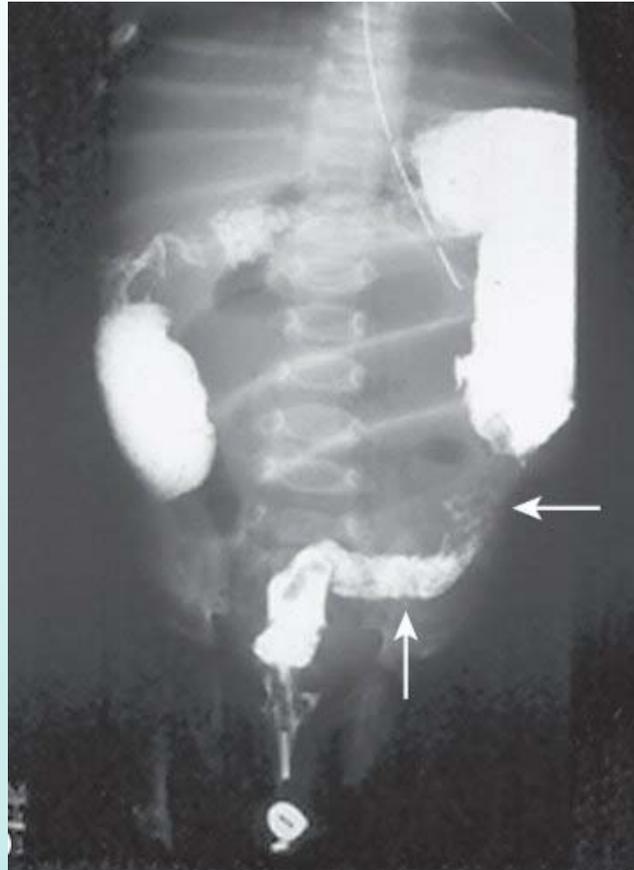
• Perforation may complicate HD.

• Explosive discharge of fecal matter after rectal examination is a valuable sign and may indicate enterocolitis.

Hirschsprung's Disease

Investigations

1. AXR (multiple dilated intestinal loops, absence of gas in rectum)
2. Contrast enema
 - Ideally before rectal exam
 - Looking for transitional zone
 - Delayed films may show contrast retention
3. Submucosal rectal biopsy (suction or open under GA)
 - 1, 2, and 3 cm above dentate line
 - **Characteristic features:**
 - Absence of ganglion cells
 - Hypertrophied nerve bundles
 - ± Acetyl cholinesterase staining (90% accurate, less so in neonates and LS HD)
 - Absence of Calretinin staining
 - ± Immunohistochemistry (e.g., LDH, S100, SDH, etc.)
4. Anorectal manometry
 - Shows absence of Recto-Anal Inhibitory Reflex
 - Not widely available & operator-dependent

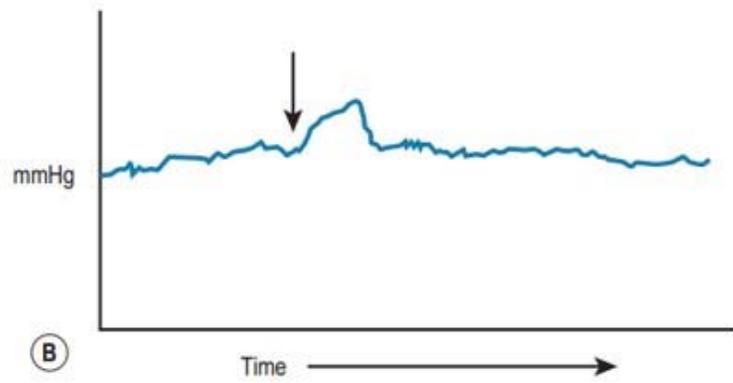
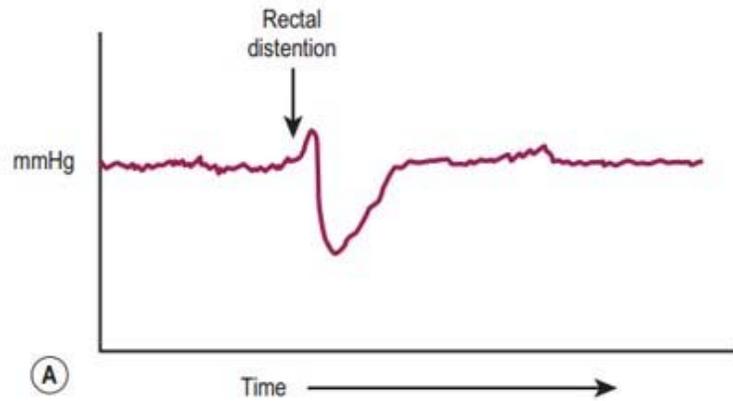


Contrast Enema

showing dilated proximal segment and distal narrow segment with transitional zone in between



Contrast Enema
showing narrow colon, suggesting "Total Colonic HD"



Anorectal Manometry

- A. Normal response to rectal distention (presence of Recto-Anal Inhibitory Reflex).
- B. Absence of Recto-Anal Inhibitory Reflex.

Hirschsprung's Disease

Differential Diagnosis

- Mechanical causes of neonatal bowel obstruction:
 - Ileal and colon atresia
 - Anorectal malformations
 - Meconium ileus
 - Meconium plug syndrome (10% have HD)
- Functional hypoperistalsis:
 - Prematurity
 - Sepsis and electrolyte imbalance
 - Small left colon syndrome
 - Hypothyroidism
- For the older child:
 - Idiopathic constipation
 - Hypothyroidism
 - Intestinal neuronal dysplasia
 - Hyperganglionosis

Hirschsprung's Disease

Management

- The aim always in HD is to decompress obstructed bowel and may be attempted even before definitive investigation.
- If enterocolitis is suspected (sepsis, fever, diarrhea, bloody stool) then further active intervention is required including:
 - Rectal washout (10–20 mL/kg of normal saline is instilled via a rectal tube in small volumes ensuring all fluid inserted is returned. Repeat up to 3× daily)
 - Antibiotics (e.g., vancomycin, metronidazole)
 - ± Colostomy

Hirschsprung's Disease

Surgery

- Currently, most infants can be managed by parents at home with daily rectal washouts until they are considered suitable for a single-stage primary pull-through procedure.
- Colostomy (indications):
 - Laparotomy for neonatal intestinal obstruction (in absence of diagnosis)
 - Low birth weight and preterm infants
 - Late diagnosis with hugely distended proximal bowel (especially in older children)
 - Repeated episodes of enterocolitis (especially in LS disease)
- Colostomy (transverse/sigmoid) or ileostomy (LS disease) should be performed in proximal, ganglionic bowel (ideally confirm by frozen section).

Hirschsprung's Disease

Surgery (Pull-Through Procedure)

- The aim is to resect the aganglionic segment, bringing the ganglionic bowel through the pelvis and anastomosing it either at the anus or somewhere close.
- Techniques:
 1. **Swenson's pull-through:**
 - Removal of all aganglionic bowel up to 1 cm from dentate line posteriorly and 2 cm of dentate line anteriorly.
 - Colo-anal anastomosis from outside.
 - Potential for pelvic nerve (incontinence) and anterior structure (vas, bladder, vagina) damage.
 2. **Duhamel's pull-through**
 - Dissection behind rectum (to minimize pelvic nerve damage) to create tunnel.
 - Ganglionic bowel is brought through ~1 cm above dentate and side-to-side anastomosis created (with GIA or EndoGIA stapler).
 - Anterior blind pouch may lead to fecaloma and recurrent obstruction.

Hirschsprung's Disease

Surgery (Pull-Through Procedure)

3. Soave's endorectal pull-through

- At pelvic reflection, the colon dissection continues in the submucosal plane to ~1 cm from dentate line.
- Ganglionic bowel is pulled through the rectal muscle sleeve and anastomosed to anal mucosa.
- Avoids potential for nerve damage.
- Retained aganglionic muscle cuff may cause functional obstruction and constipation or sleeve abscess.

4. Laparoscopy-assisted trans-anal pull-through

- Any of the above can be performed under laparoscopic vision for the pelvic dissection.

5. Trans-anal endorectal pull-through

- Dissect entirely from below (submucosa or full-thickness) into the peritoneal cavity, removing the aganglionic bowel and achieving a safe anastomosis.

Hirschsprung's Disease

Outcome

- **Early complications:**
 - Enterocolitis (even post-op.)
 - Anastomotic leak and stricture
 - Intestinal adhesion obstruction
 - Perianal excoriation
- Duhamel and Soave procedures have a higher rate of constipation.
- Swenson's have higher incidence of incontinence.

**All the patients need long-term follow-up,
as they are seldom cured of their pathology**

Anorectal Malformations



Anorectal Malformations

The terminal part of the hindgut is abnormally placed and lies outside (partially or completely) the sphincter mechanism.

Anorectal Malformations

- Incidence ~1 in 5,000
- More common in Down's syndrome and Cat-eye syndrome.
- Male>female (60:40)
- Second child involvement rare (<1%)
- 5% of babies have no fistula (usually associated with Down's syndrome)
- Majority has a connection between the distal rectum and the genitourinary tract.

Anorectal Malformations

Embryology

- By 21 days there is a common chamber (cloaca) occluded by a membrane, but visible from the outside as an ectodermal pit – the proctodaeum.
- At ~33 days, the posterior hindgut is then separated from the anterior urogenital sinus by mesenchymal ingrowth of the urorectal septum.
- Cloacal membrane breaks down at ~46 days.
- The process is regulated by differential expression of the gene Sonic Hedgehog3 (SHH), and other target genes (BMP-44 and HOX5).

Anorectal Malformations

Anatomical classification

Males	Females
Anteriorly displaced anus	Anteriorly displaced anus
Recto-perineal fistula	Recto-perineal fistula
Recto-bulbar urethral fistula (most common)	Recto-vestibular fistula (most common)
Recto-membranous urethral fistula	Recto-vaginal fistula
Recto-prostatic urethral fistula	Imperforate anus without fistula
Recto-bladder neck fistula	Persistent cloaca
Imperforate anus without fistula	Rectal atresia
Rectal atresia	

Anorectal Malformations

Clinical Features

- Clinical examination is the most important part of management (makes the diagnosis in 90% of cases).
- High anomalies:
 - flat perineum
 - passage of meconium per urethra
 - short sacrum
 - little sphincter muscle contraction
 - bifid scrotum or a sphincter close to the scrotum --> often prostatic fistula
- In female babies, a careful examination will tell about the position of the opening (vestibular, perineal, vaginal, or cloacal).
- Cloaca is diagnosed by a single perineal opening.
- Low anomalies are suggested by an opening in the perineum and a “bucket-handle” defect.



“Bucket-handle” deformity,
suggesting a low type ARM

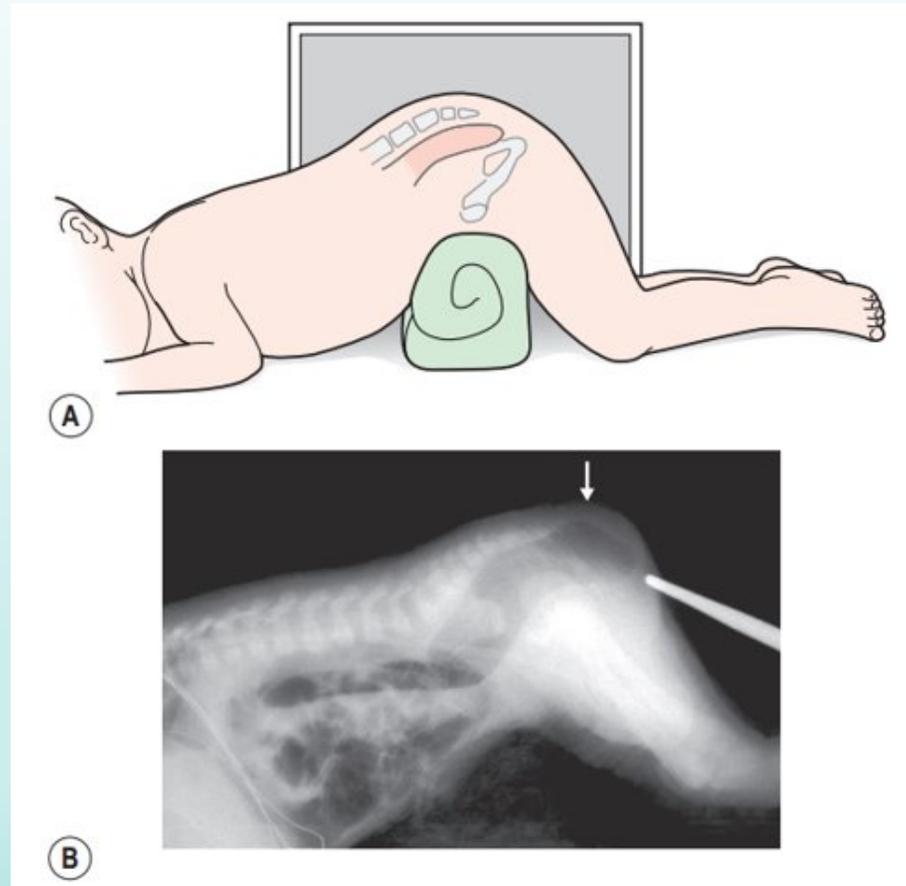


Imperforate anus with flat perineum,
suggesting a high type ARM

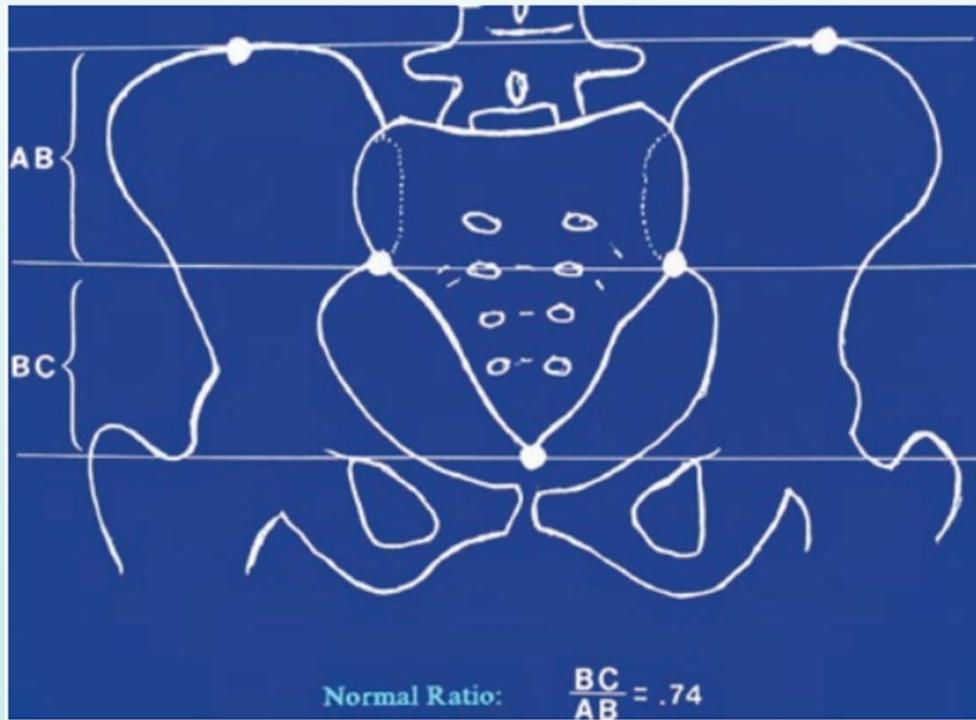
Anorectal Malformations

Management

- AXR
 - A cross-table lateral film (after 12–24 h) is useful in localizing the distance of rectal gas from perineum, if physical examination is unable to detect a rectal fistula.
 - Sacral ratio (Normal=0.74 (0.7–0.8) | Lower values (<0.4) are prognostic for low potential for continence)
- US
 - To detect associated renal anomalies, vertebral anomalies, and hydrocolpos (in cloaca).
 - Echocardiogram to rule out cardiac anomalies.



Cross-table lateral film (after 12–24 h of life) in Jackknife position
to localize the distance of rectal gas from perineum

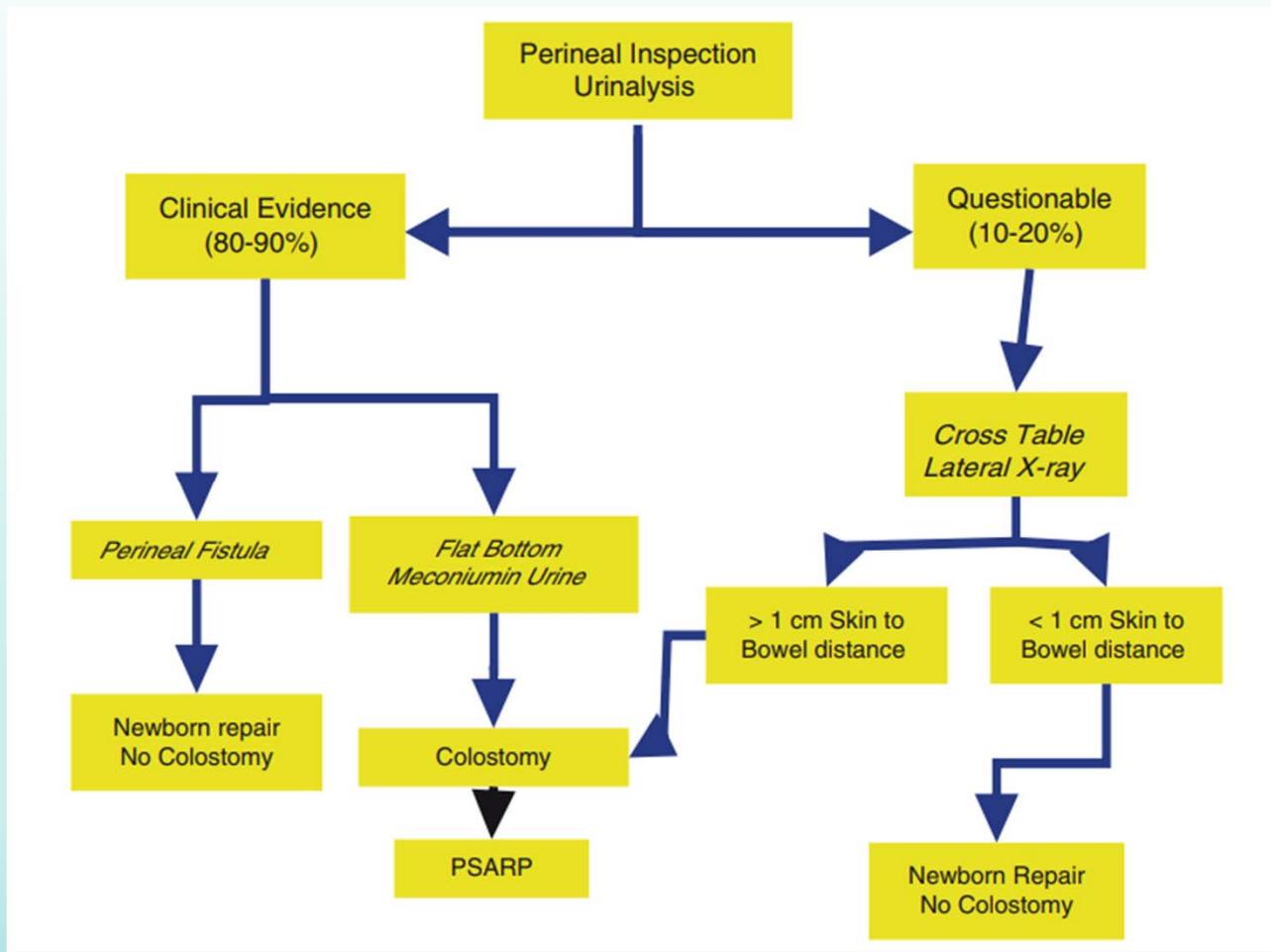


Sacral ratio

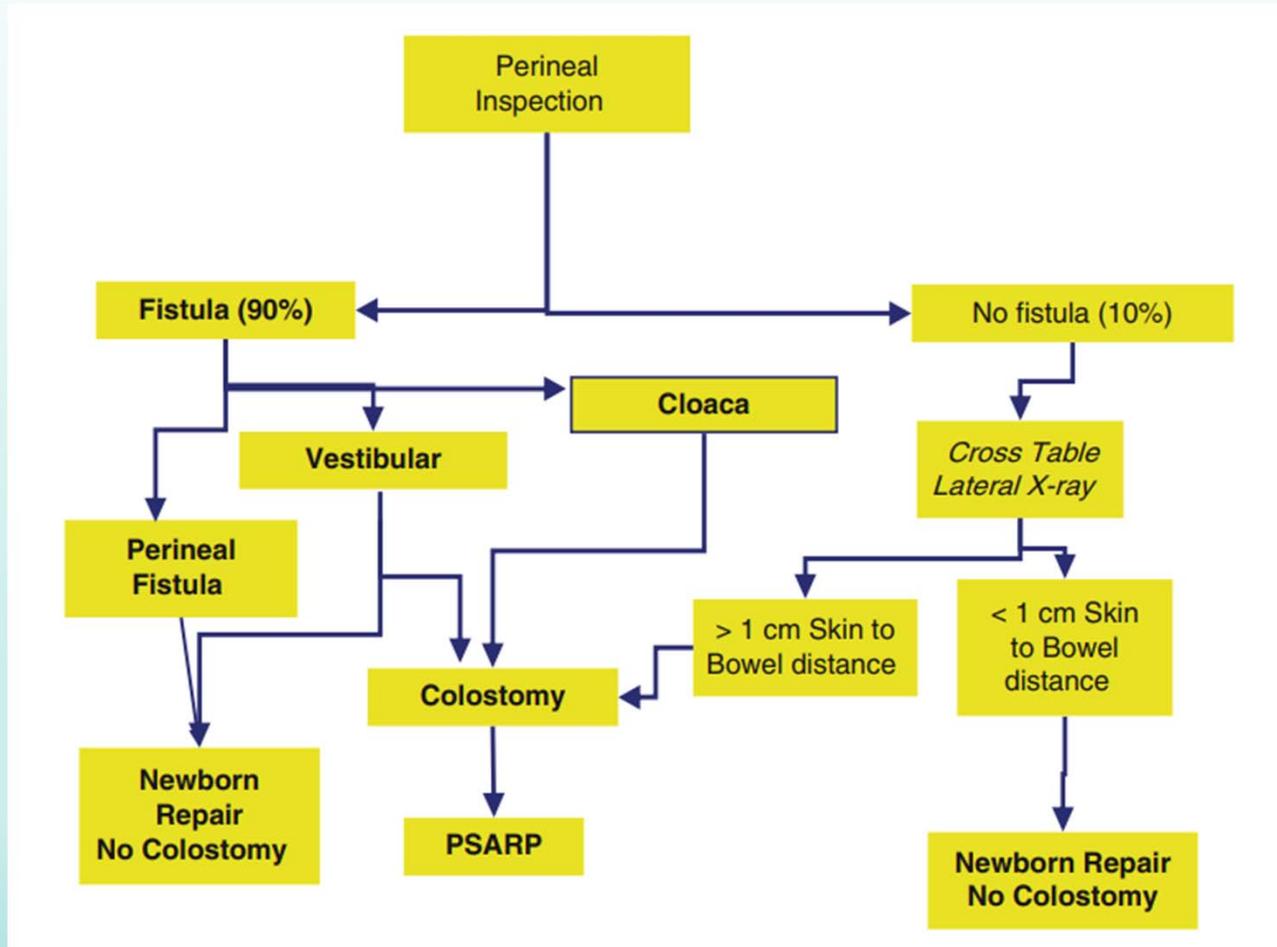
Anorectal Malformations

Management

- Initial management includes:
 - Nasogastric tube (for decompression, but if difficult to insert --> CXR to rule out esophageal atresia)
 - Nil by mouth (NPO)
 - Intravenous fluids
 - Antibiotic prophylaxis
 - Watchful waiting (for about 24 h) while proper assessments/investigations are done
 - Rule out VACTERL associations



Algorithm for male infant



Algorithm for female infant

Anorectal Malformations

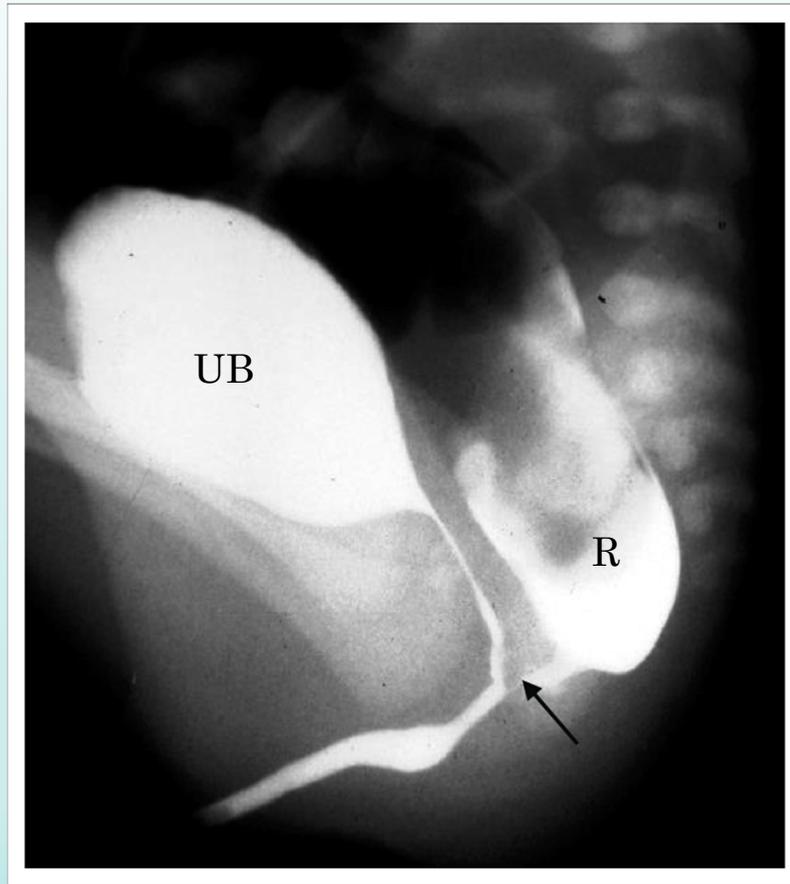
Management

- Babies with a high anomaly are usually treated with initial colostomy, followed by definitive surgery and then colostomy closure.
- Defunctioning divided colostomy in the fixed (most proximal) part of sigmoid colon.

Anorectal Malformations

Management

- In suspected high types, after colostomy creation, distal high-pressure colostogram must be done:
 - To check fistula type
 - To help choosing the appropriate surgical approach
- Definitive surgery can be done around 6–12 weeks.



Distal Colostogram

Shows that contrast is passing through a recto-membranous urethral fistula (black arrow) filling the urinary bladder and urethra

Anorectal Malformations

Management

Posterior Sagittal Anorectoplasty (PSARP) (Male)

1. Identify sphincter position:
 - An appropriate muscle stimulator (e.g., Pena model Radionics Inc.) is crucial in identifying the center of anal sphincter..
2. Midline skin incision:
 - rectum may be too high to reach from below, and a combined abdominal approach will be needed (can be done laparoscopically).
3. Identify and ligate the fistula.
4. Pull the rectum to create neoanus (inside anal sphincter complex).
5. Reconstruct anal sphincter complex around mobilized rectum.

Anorectal Malformations

Management

Posterior Sagittal Anorectoplasty (PSARP) (Female)

1. Identify “true” sphincter position with stimulator.
2. Midline sagittal incision.
3. Identify and ligate fistula (if present)
4. Create neoanus within muscle complex.
5. Reformation of anterior perineal body.

Anorectal Malformations

Management

Definitive single-stage surgery without colostomy can be done in newborn during 24–48 h after birth if:

- Perineal fistula is present.
- Rectum is within 1 cm from skin (on cross-table lateral film).
- Vestibular fistula (if an experienced surgeon is available).

Anorectal Malformations

Outcome

- The higher the ARM anomaly, the lower the achieved continence.
- Sacral anomaly – ↓ achieved continence.
- Lower malformations tend to suffer more from constipation.

Thank You

