CONGENITAL DISORDERS OF THE DIGESTIVE TRACT

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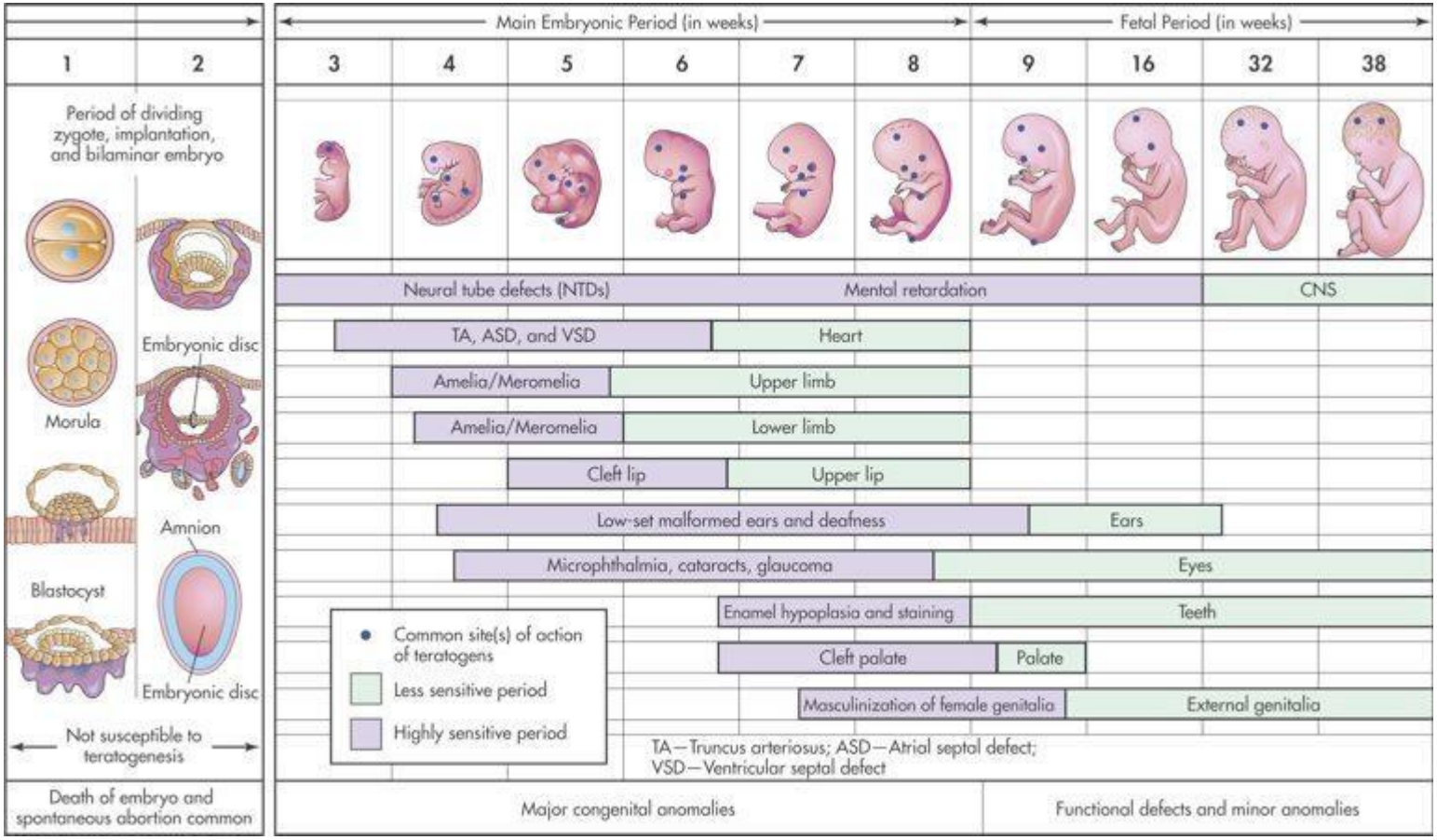
Embryologic development



http://2.bp.blogspot.com/-L9ngeyYTBpg/UUptASbN2BI/AAAAAAAAB5I/8dDZLapkFMs/s1600/Developmental-stages.jpg Accessed 11/11/2019

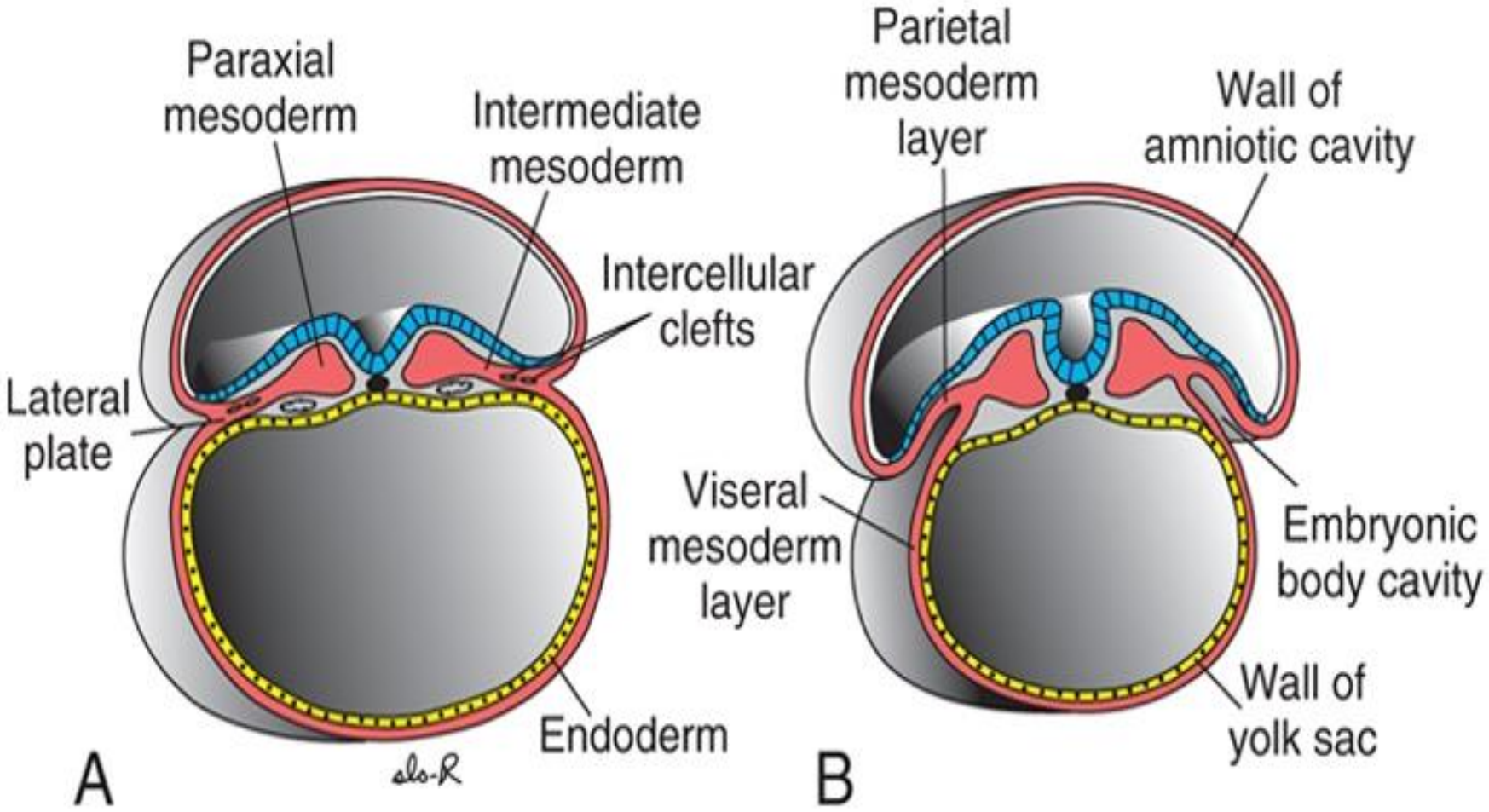
Molecular regulation of embryogenesis

- A gradient of retinoic acid (high caudally, low cranially) causes differential expression of regulatory genes
- The regulatory genes define the structure of the different regions of the gut tube
- SOX2 esophagus and stomach
- PDX3 duodenum
- CDXC small intestine
- CDXA large intestine and rectum

Molecular regulation of embryogenesis

- The lower gut tube produces a gradient of Sonic hedgehog (SHH)
- SHH induces differential expression of Hox genes by the surrounding mesoderm
- The differential Hox expression creates the regional differences between the small intestine, cecum, colon, and cloaca

Gastrulation



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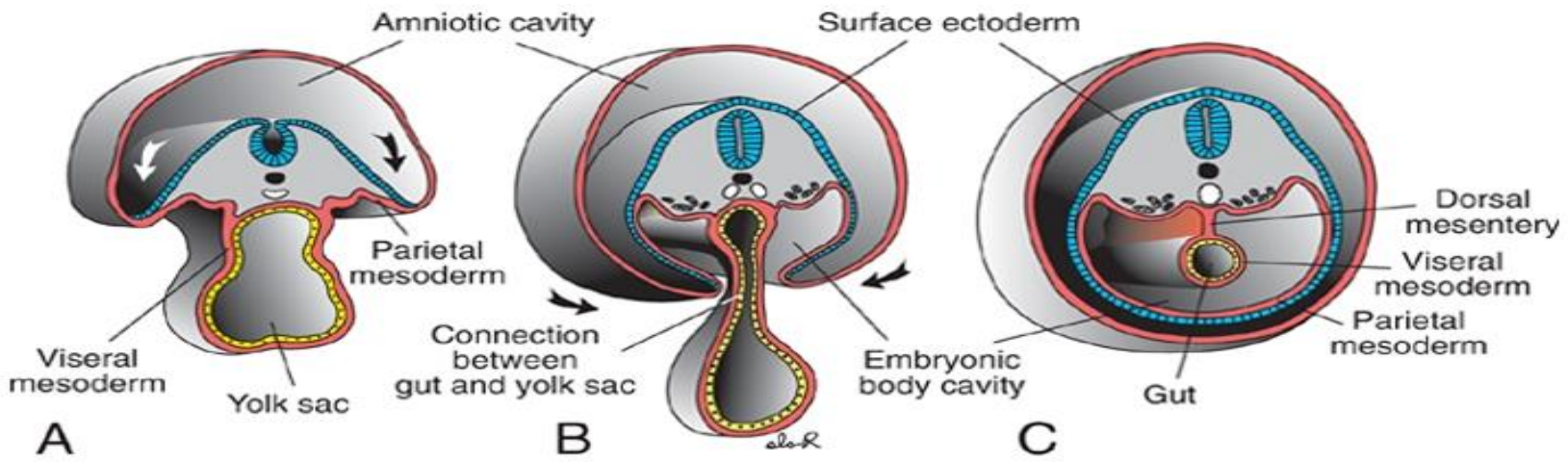
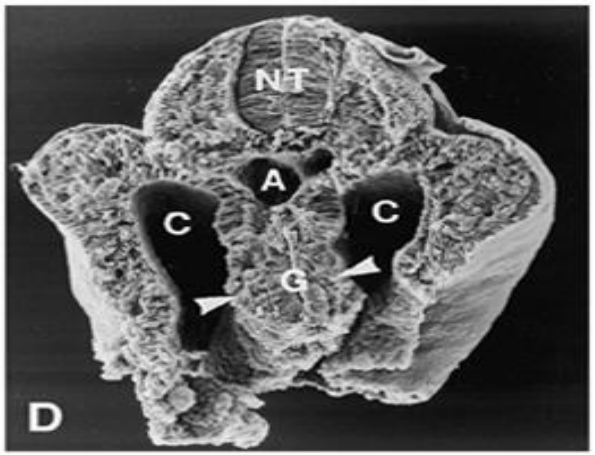
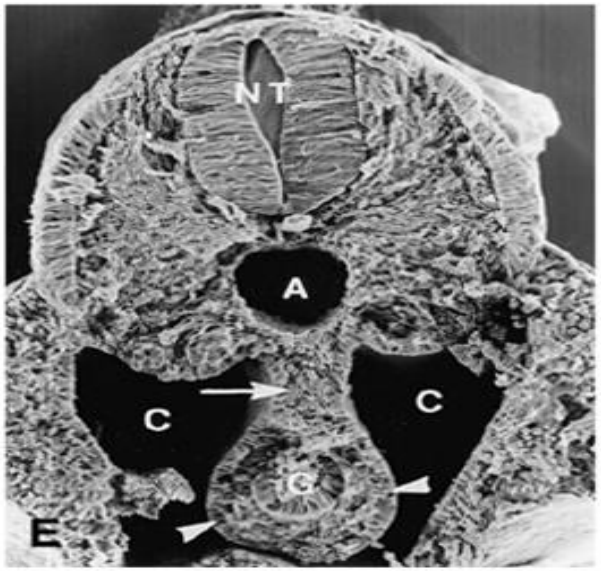
Embryogenesis

- The rostral and caudal ends of the embryo gut tube end blindly.
- The yolk sac and allantois remain outside the embryo.
- The central opening of the gut tube remains in communication with the yolk sac through the vitelline duct .
- At the 5^{th} week, the coelom fuses, separating the embryo from the yolk sac, creating the intracoelomic space.

5th week of gestation







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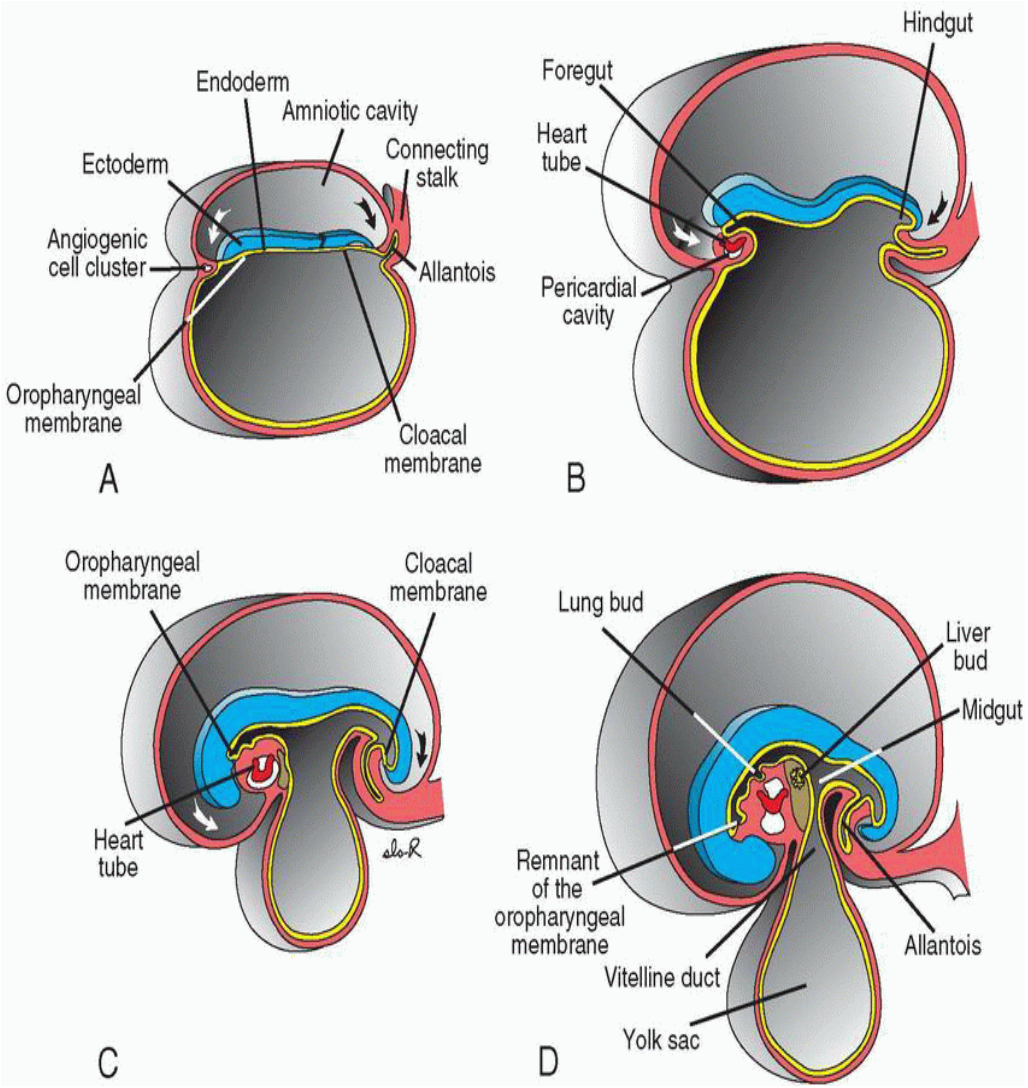
Embryogenesis

- By the 7th week, abdominal and thoracic cavities are separate.
- The tracheo-bronchial diverticulum gives rise to the tracheo-esophageal system.
- Paired lateral ridges fuse, creating the tracheo-esophageal septum, separating respiratory and digestive system.
- The thoracic cavity partitions further with the formation of the cardiac and pulmonary systems.

Embryogenesis

- The transverse septum gives rise to the central tendon of the diaphragm.
- The somatic mesoderm gives rise to the muscular part of the diaphragm.
- The phrenic nerve is the only motor nerve to the diaphragm (C3-C5).

7th week of gestation



Embryogenesis

- Later the pharyngeal pouches begin to differentiate.
- Submandibular and sublingual glands arise from the gut tube.
- The parotid arises from ectoderm.
- The epithelium of the digestive tract from pharynx through the upper two-thirds of the anal canal arises from endoderm.
- Mucosa of the mouth and pharynx and the lower-third of the anal canal arise from surface ectoderm.

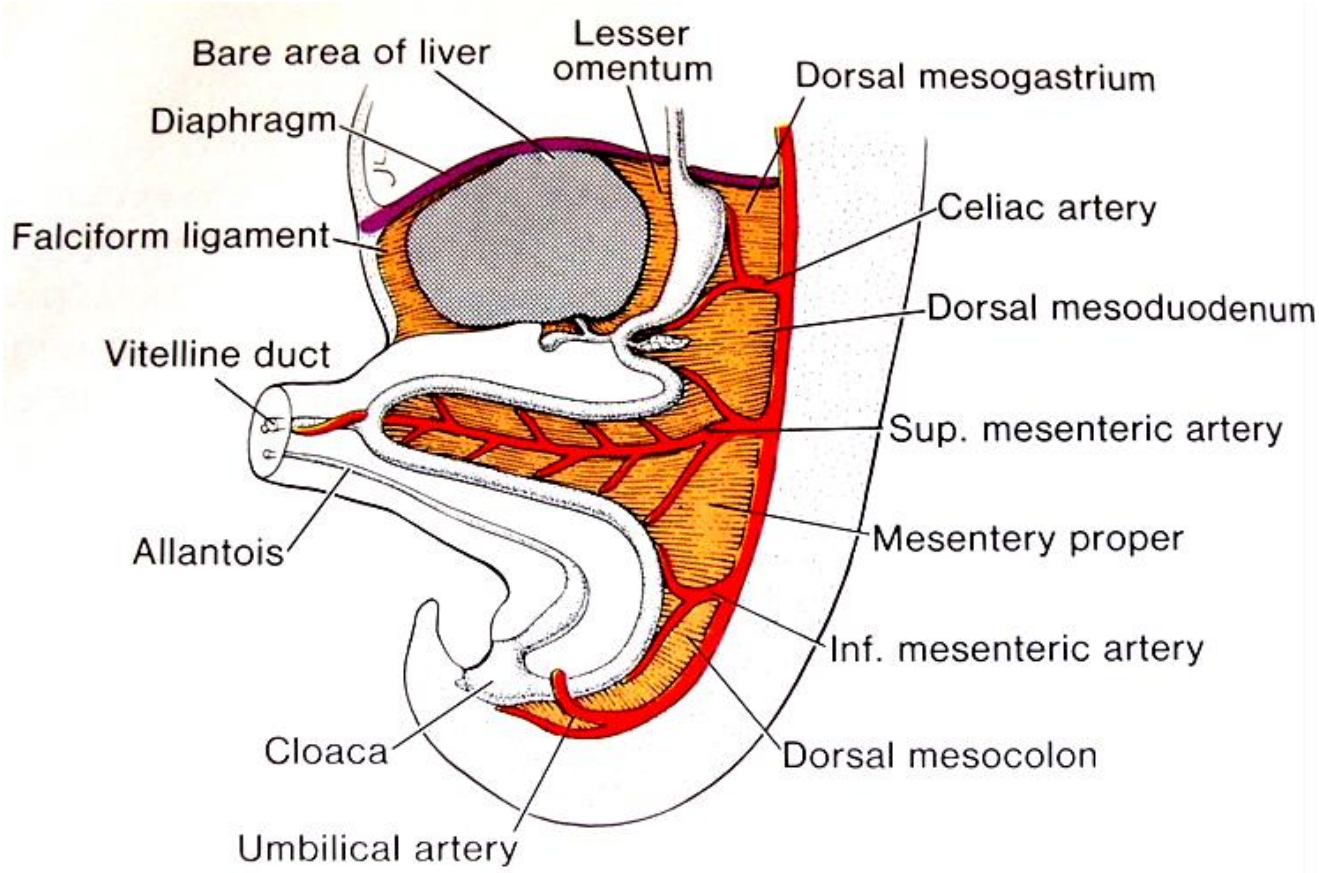
Embryogenesis

- The hindgut is continuous with the cloaca
- The region of contact between endoderm lined cloaca and surface ectoderm forms the cloacal membrane.
- The pectinate line marks this division between ectoderm (anus) and endoderm (rectum).
- The anal pit is proctoderm.

Embryogenesis

- The uro-rectal septum is formed from two folds:
- Tournoux and Rathke
- These serve to divide the cloaca into a urogenital sinus (bladder and urethra)
- And divide the cloaca into a recto-anal canal.
- Failure to close leads to fistula formation.

Blood supply to the gut



The foregut is supplied by the celiac artery.
The midgut is continuous with the yolk sac (and supplied by the superior mesenteric artery).
The hindgut is supplied by the inferior mesenteric artery.

Abdominal wall anomalies

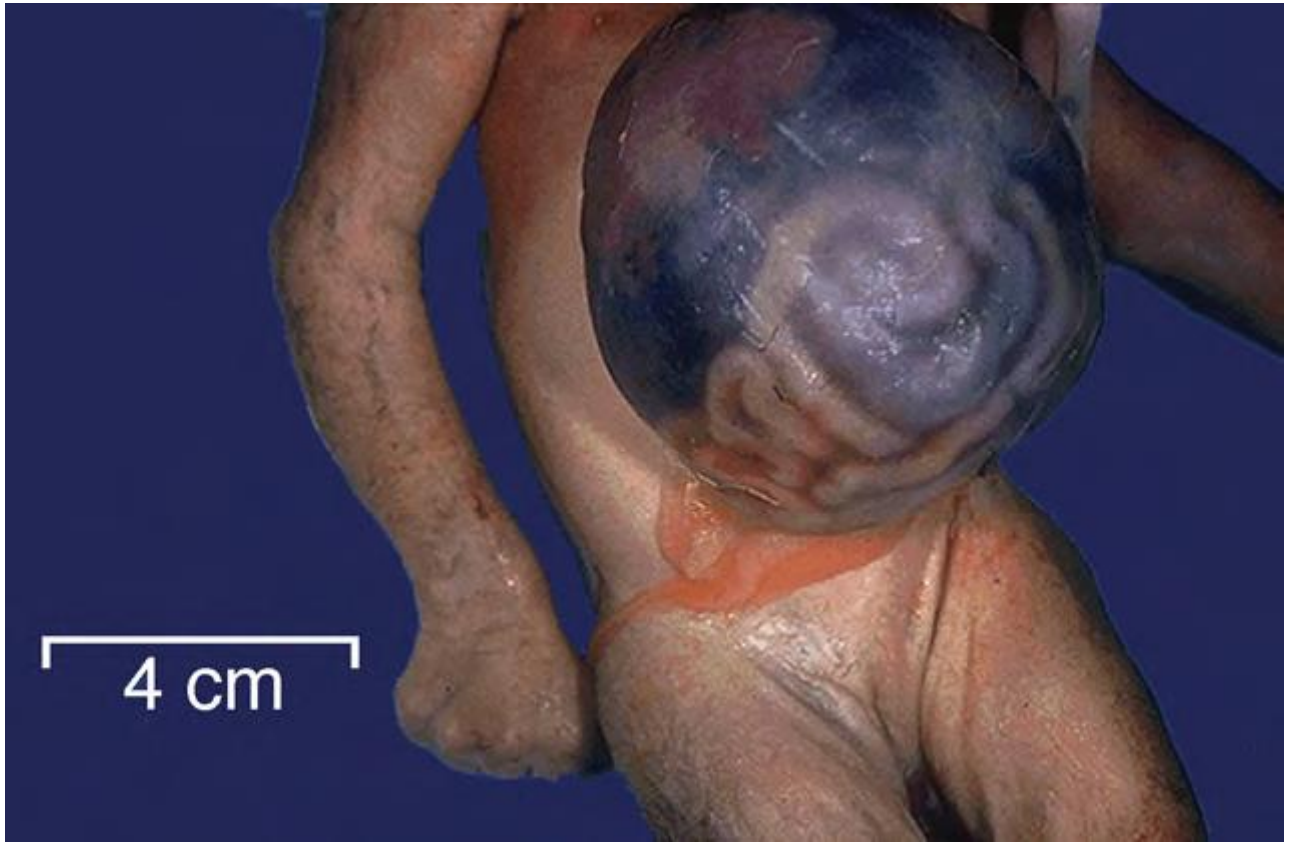
- Failure of the bowel to return to the abdomen in the 10th week of gestation following herniation of abdominal contents into the base of the umbilical cord during the 6th week of life produces an omphalocele.
- The herniated intestine is covered by peritoneum.
- May be repaired surgically
- Associated with other birth defects in 40% of cases
- With gastroschisis there is no peritoneal covering. It results from a full thickness abdominal wall defect.

Abdominal wall anomalies

- Prune-belly syndrome involves a failure of hyperaxial myotome.
- There is lack of an abdominal wall.
- Pulmonary hypoplasia may be associated as a result of elevated intra-abdominal pressure.

Omphalocoele

Klatt, EC, Robbins and Cotran Color Atlas of Pathology. Elsevier. Philadelphia. 2015. Figure 7-59 Accessed 11/07/2019



Gastroschisis

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Hernias

- Diaphragmatic hernia
- Incomplete diaphragm development and herniation of abdominal organs into the thorax
- Results in pulmonary hypoplasia
- A sliding hiatal hernia is due to a defect in the right crus and/or a short esophagus.
- The upper part of the stomach is retained in the thorax and the stomach is constricted at the level of the diaphragm.
- A Bockdelek hernia involves a defect in the left crus.

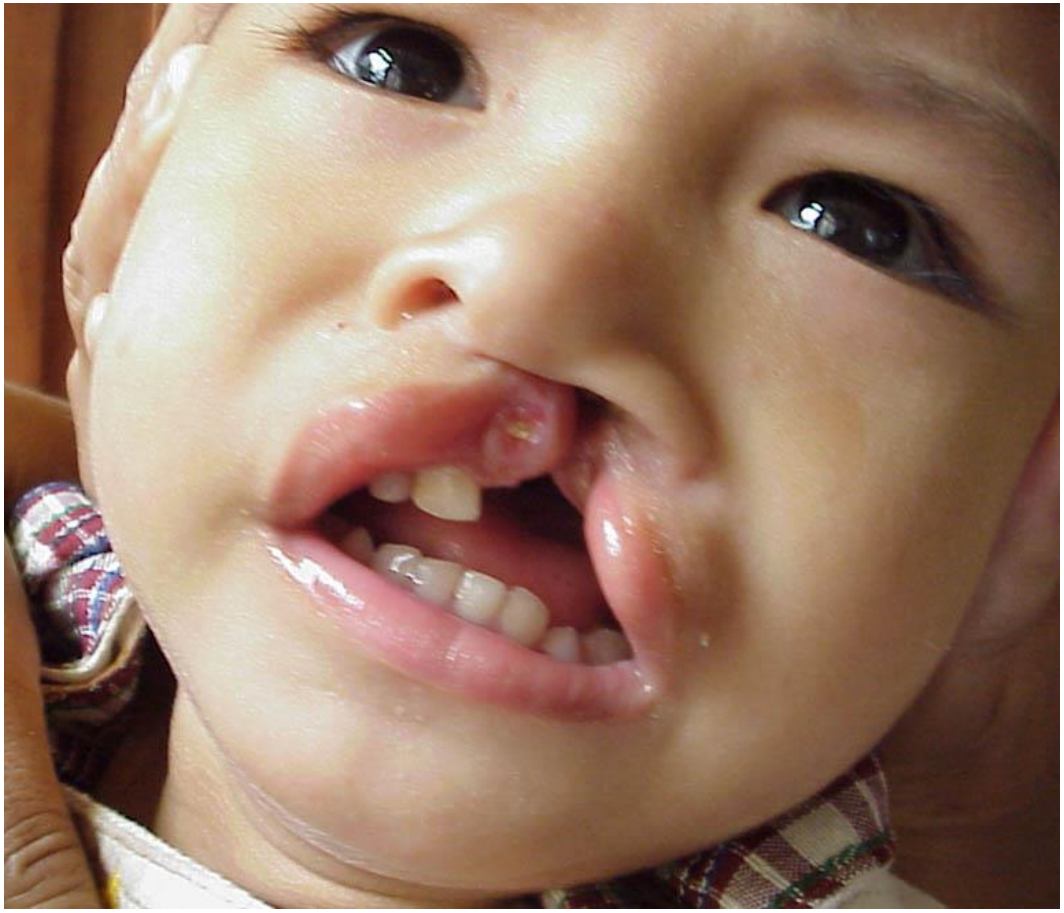
Cleft palate

- Most common congenital disorder of oral cavity
- 1 in 800 births
- 3% recurrence in subsequent siblings
- More common in whites than in blacks
- Cleft lip and palate account for 50% of lesions
- Cleft lip alone (25%), more common in males
- Cleft palate alone (25%), more common in females
- Failure of fusion of facial processes
- Cleft palate alone differs in genetic origin from cleft lip alone or cleft lip and palate

Cleft palate

- Complications
- Eustachian tube dysfunction (chronic otitis media)
- Speech difficulties
- Surgical correction
- Folate supplementation in pregnancy diminishes incidence of cleft palate

Cleft lip and palate



Cleftlipcleftpalate5.blogspot.com
Accessed 11/11/2019

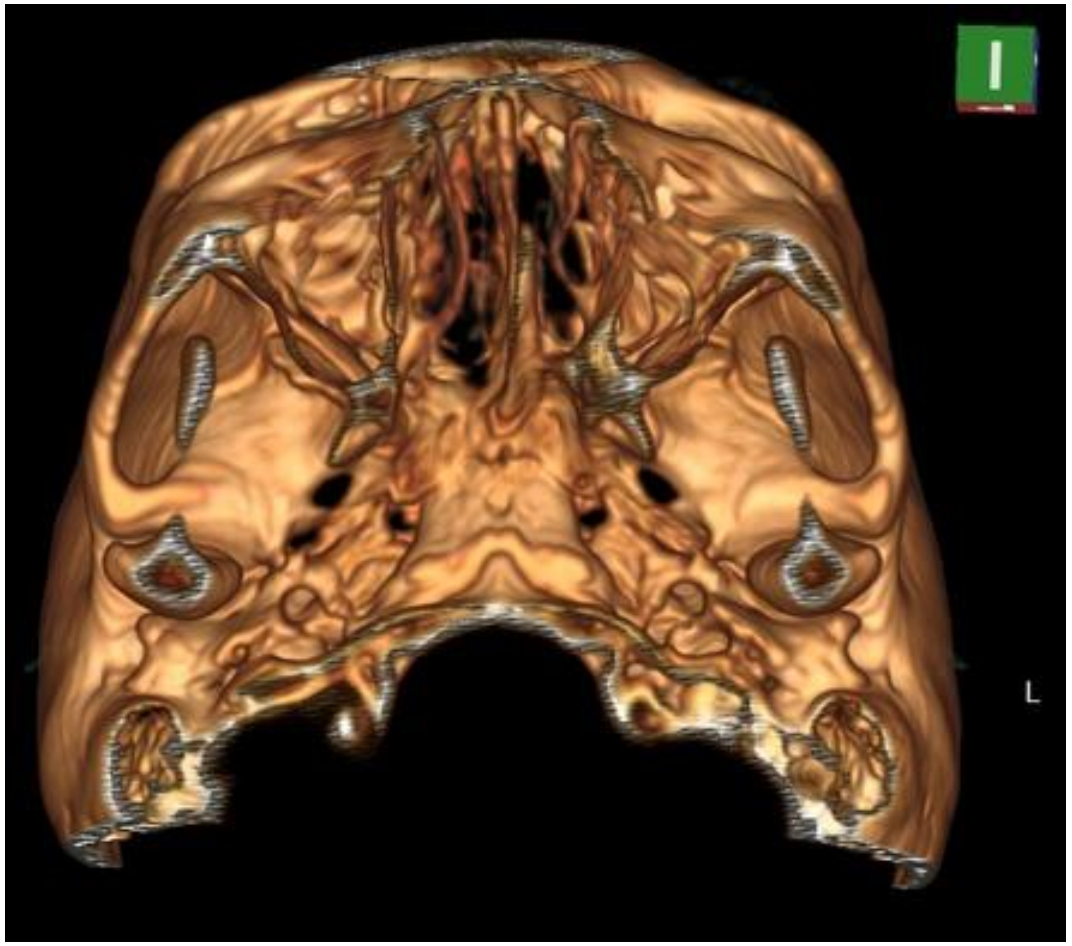
Neonatal presentations

- Choanal atresia
- Duodenal atresia
- Pyloric stenosis
- Tracheoesophageal fistula
- Meckel’s diverticulum
- Intestinal atresia
- Anal atresia

Neonatal presentations

- Hirschsprung's disease may also present at birth with:
- Obstipation
- Abdominal distention
- Feculent vomitus.
- Meconium ileus also presents with feculent vomitus.
- Bowel sounds diminished (ileus).
- May note rectal prolapse.
- Consider cystic fibrosis.

Choanal atresia



Presents as cyanosis with feeding.

Bone window on CT. Nasal cavity and sinuses filled with (hyaline) secretions.

https://radiopaedia.org/images/31435372
Accessed /01/10/20

Duodenal atresia

- Failure of the lumen of the duodenum to recanalize by apoptosis shortly after the obliteration of its lumen in the 9^{th} week of embryogenesis.
- Poor feeding

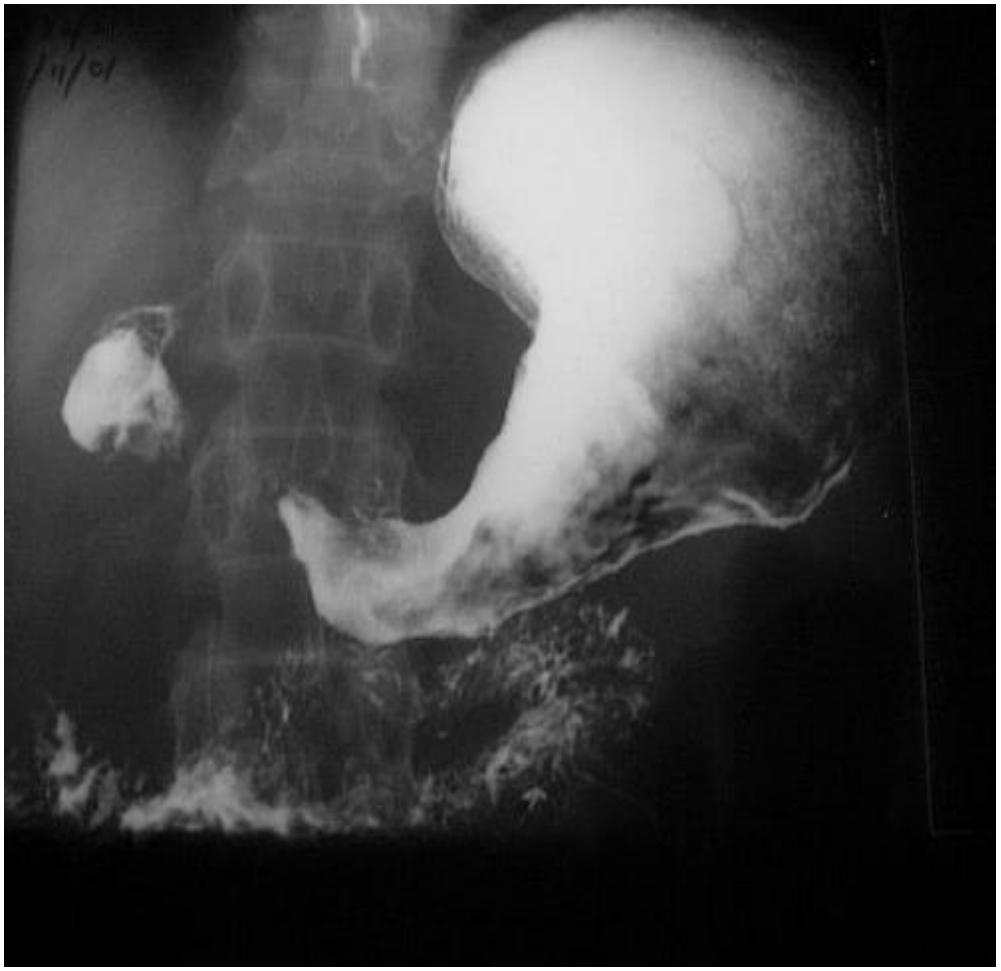
Pyloric stenosis

- More common in males
- 1 in 300-900 live births
- High rate of concordance in monozygotic twins (200 times), while concordance lower (20 times) in dizygotic twins and in siblings of affected individuals
- Deficiency of nitrous oxide synthetase
- Turner syndrome and Trisomy 18 also associated with pyloric stenosis
- Macrolide antibiotic exposure in first two weeks of life linked to increased disease incidence

Pyloric stenosis

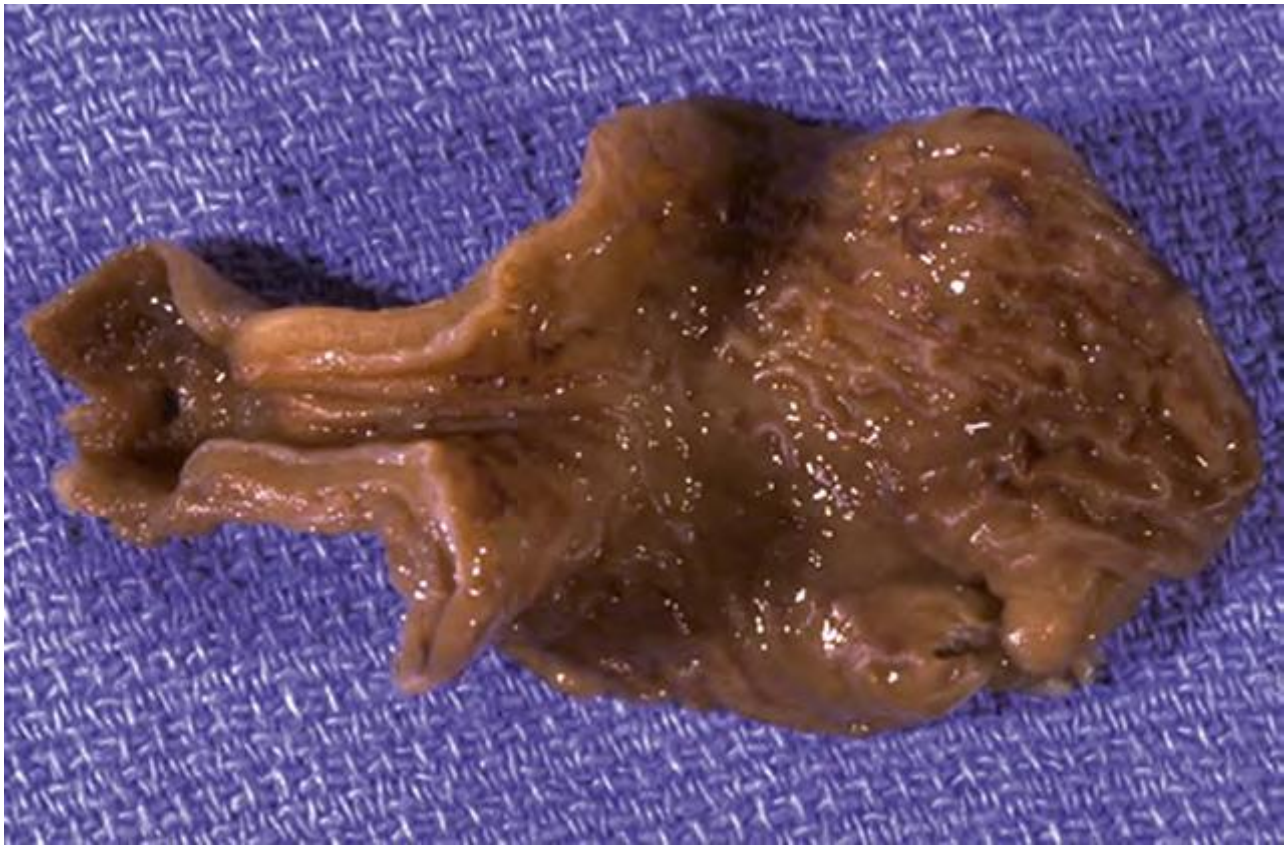
- Presents between third and sixth weeks of life
- New onset regurgitation, projectile, non-bilious vomiting after feeding, and frequent demands for refeeding
- Abnormal left to right hyperperistalsis may be seen during or immediately after feeding
- Myotomy is curative

Pyloric stenosis



Barium column stops at pylorus. Small amount of barium in small intestine.

Pyloric stenosis

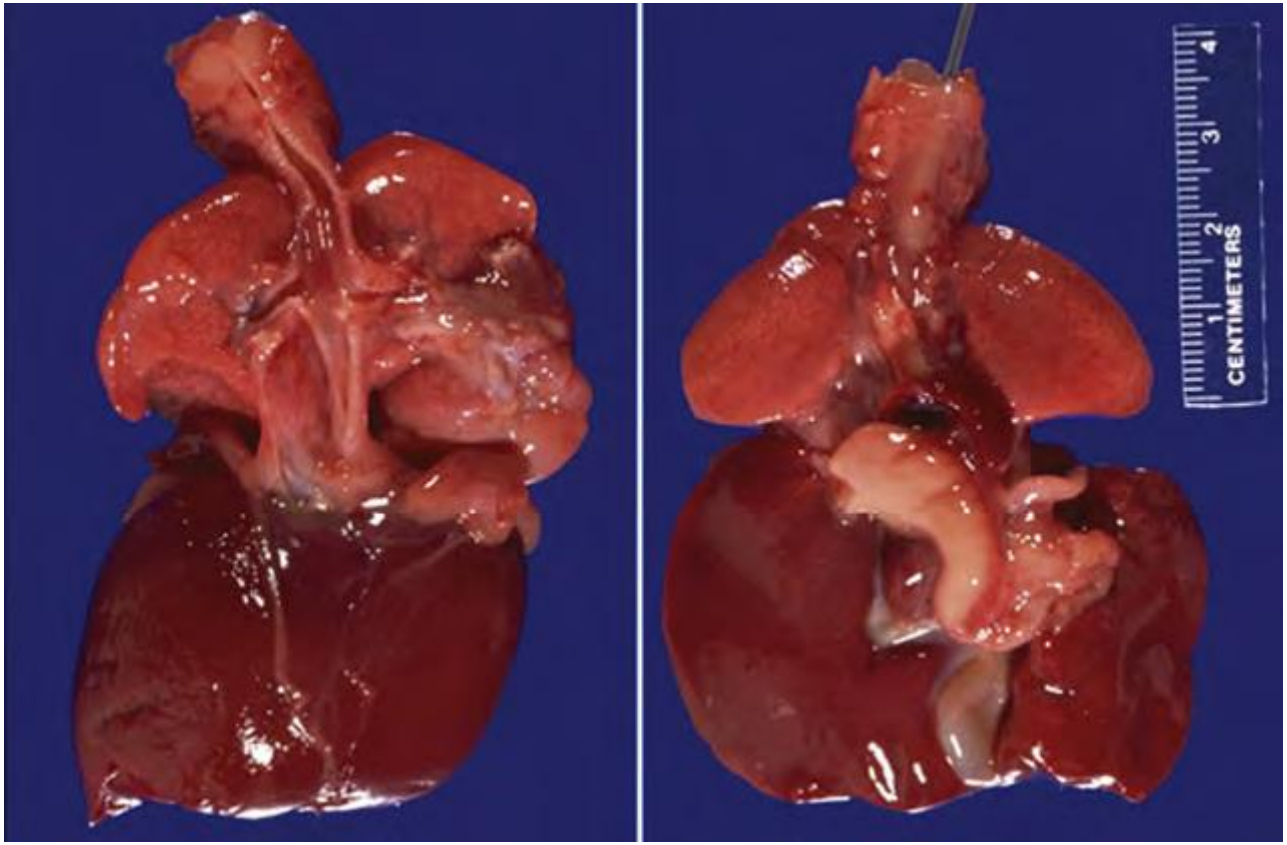


Klatt, EC, Robbins and Cotran Color Atlas of Pathology.
Elsevier. Philadelphia. 2015. Figure 7-32 Accessed 11/07/2019

Tracheoesophageal (TE) fistula

- Thoracic and intestinal organs separate at 7th week of gestation.
- Principal foregut abnormality
- Proximal esophagus ends blindly
- Distal esophagus arises from trachea
- Polyhydramnios (excess amniotic fluid)
- Swallowed amniotic fluid cannot be reabsorbed in small intestine.

Tracheo-esophageal fistula



Klatt, EC, Robbins and Cotran Color Atlas of Pathology. Elsevier. Philadelphia. 2015. Figure 7-4 Accessed 11/07/2019

Meckel diverticulum

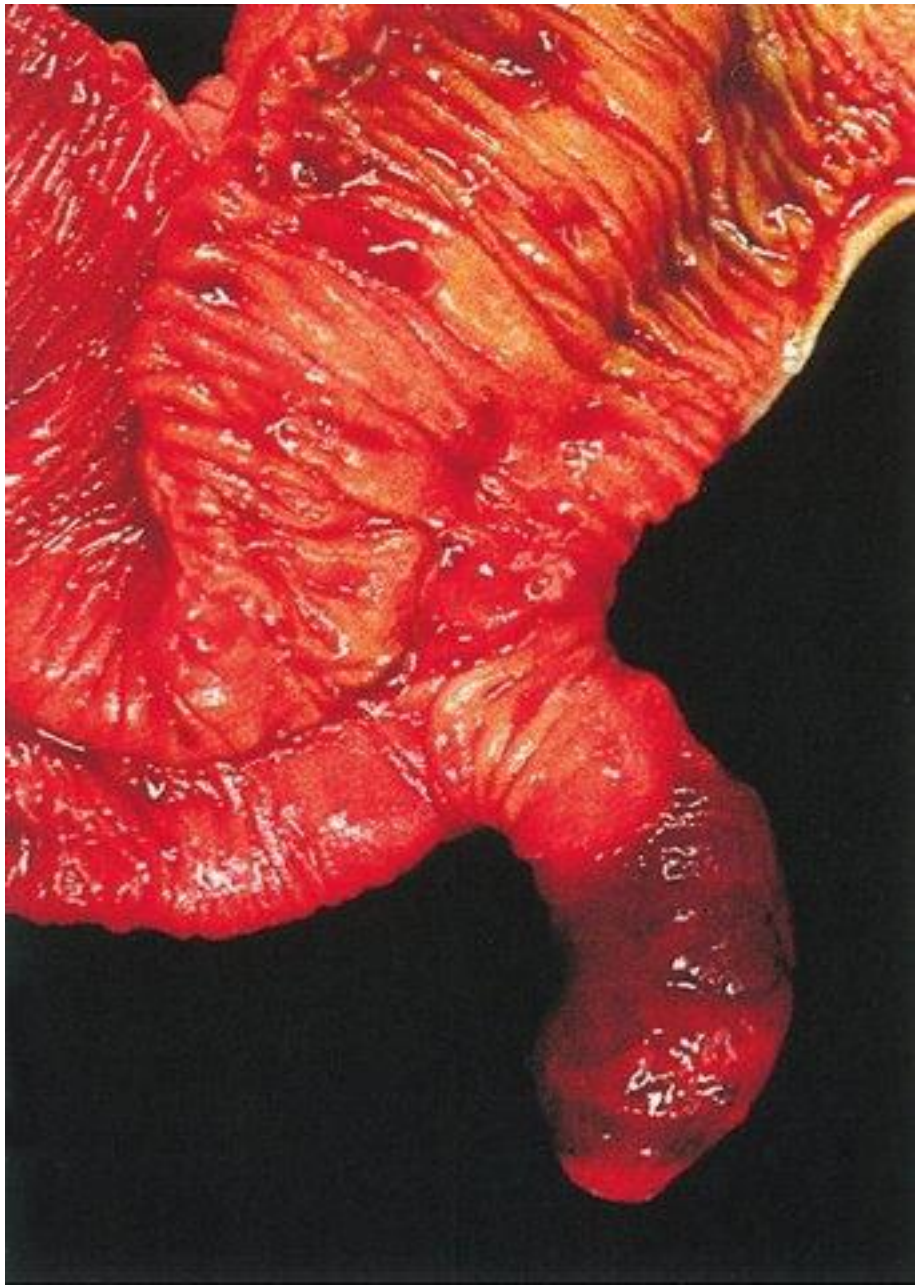
- This is the most common congenital abnormality of the gastrointestinal tract.
- Failed involution of vitelline duct which connects the lumen of the developing gut to the yolk sac
- Mucosal lining may resemble that of normal intestine (true diverticulum in which all three layers of bowel are present) but ectopic gastric or pancreatic tissue may be present

Meckel diverticulum

- Found in the Ileum
- Bleeding most common presentation in young child
- Newborn may have fecal material in umbilical area due to persistence of vitelline duct

Meckel diverticulum

- 2 inches long
- 2 feet from ileocecal valve
- 2% population
- 2 times more common in males
- 2% symptomatic
- May be symptomatic by age 2
- Older children and adults may present with obstruction or diverticulitis (mimics appendicitis)

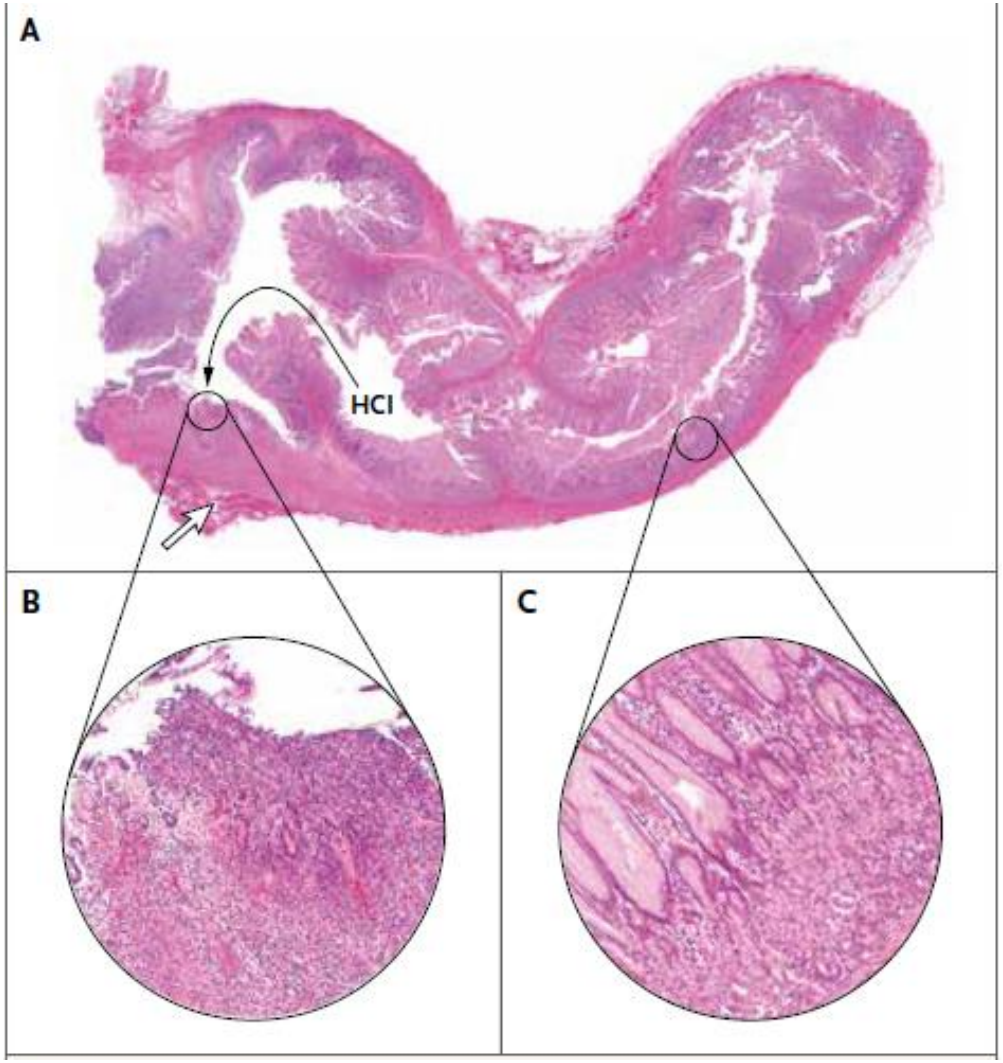


Meckel's diverticulum

Armed Forces Institute of Pathology, Public Domain, https://commons.wikimedia.org/w/index.php?curid=10645588

Accessed 12/10/2019

Meckel's diverticulum



NEJM.org Cases in Primary Care (2017)
Accessed 12/20/2019

Present are all three layers of bowel wall (Panel A); serosal fibrinous exudate is present (white arrow); peptic ulceration is present in the adjacent intestinal mucosa (black arrow).

The intestinal mucosa at the opening of the diverticulum shows mucosal erosion, acute inflammation, and underlying granulation tissue, findings consistent with an ulceration (Panel B).

Heterotopic gastric mucosa lines the entire diverticulum and is composed of surface foveolar cells, mucous neck cells, parietal cells, and basal chief cells (Panel C).

Intestinal atresia

- Imperforate anus as most common form of intestinal atresia
- Hindgut abnormality
- Failure of cloacal diaphragm to involute
- Anal atresia may present with feculent vomitus
- Double bubble sign on x-ray

Hirschsprung disease

- Presents as obstruction because unable to pass meconium.
- Coordinated peristalsis is lost
- Distal intestinal segment lacks both the Meissner submucosal and the Auerbach myenteric plexes.
- Defect always begins at the rectum

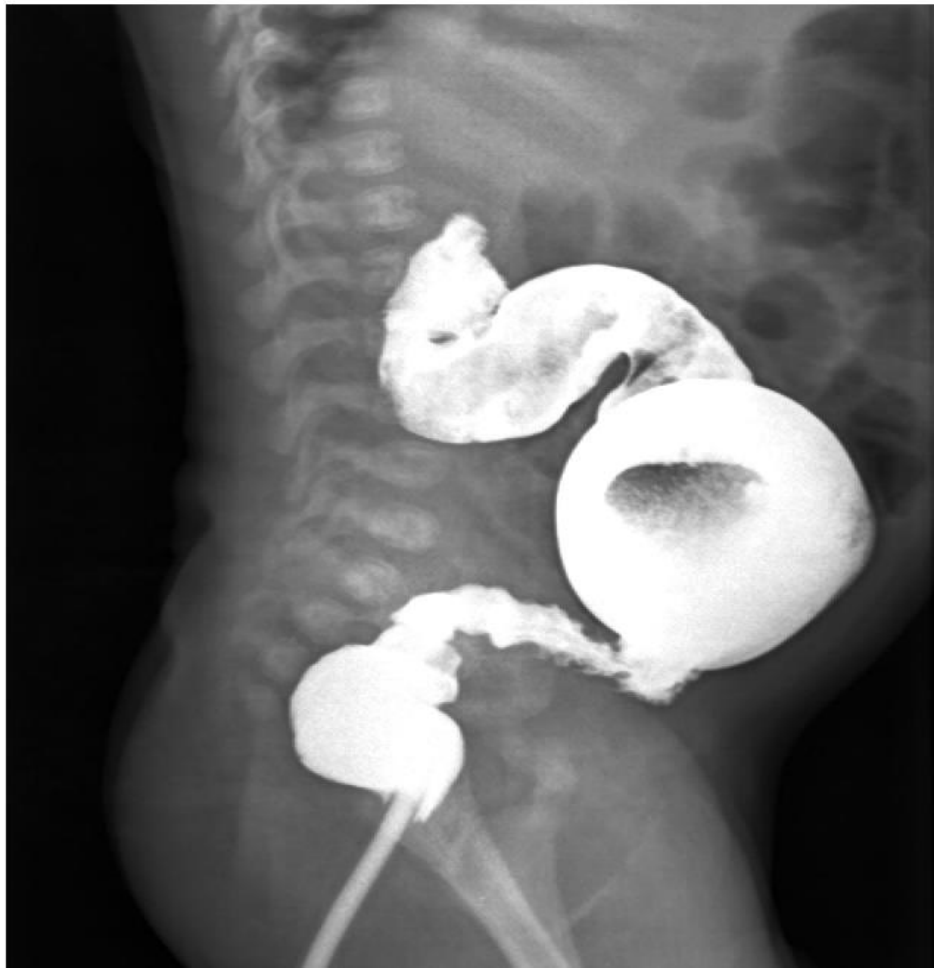
Hirschsprung disease

- Short segments of bowel involved in male
- Long segments of bowel involved in female.
- Surgical resection of aganglionic segment with end to end anastomosis of the normal proximal colon to the rectum as treatment.
- Normal bowel function and continence develop over time.

Hirschsprung disease

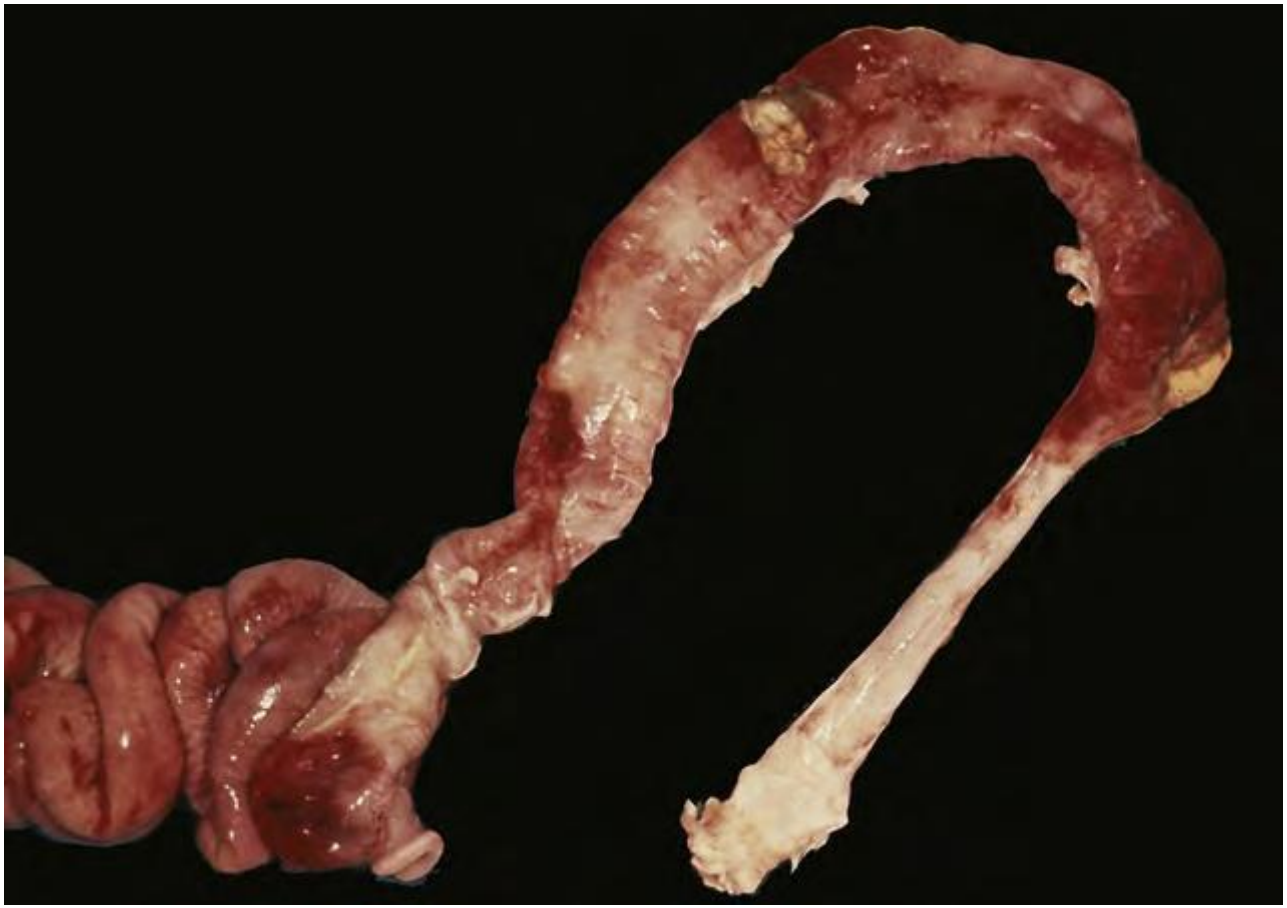
- One in 5000 live births
- 4% of patient siblings affected
- 10% of all cases occur with Down's syndrome
- 5% of all other cases associated with serious neurologic abnormalities
- Loss of function mutation in RET accounts for majority of familial cases and 15% of sporadic cases
- Failure of migration of neural crest cells into the bowel wall during embryogenesis

Hirschsprung disease



Dilated bowel segment proximal to narrowing.

Hirschsprung's disease



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Hernias

- The external oblique fascia is continuous with the external spermatic fascia.
- The inferior margin of the aponeurosis of the external oblique becomes the inguinal ligament. It serves as a retinaculum.
- A hernia may present as a mass worsened by increase in abdominal pressure.
- Groin pain may also be present.

Hernias

- The internal oblique fascia is continuous with the cremasteric fascia and muscle.
- The transversalis fascia is continuous with the internal spermatic fascia.
- The tunica vaginalis of testis is derived from the processus vaginalis (peritoneum).

Hernias

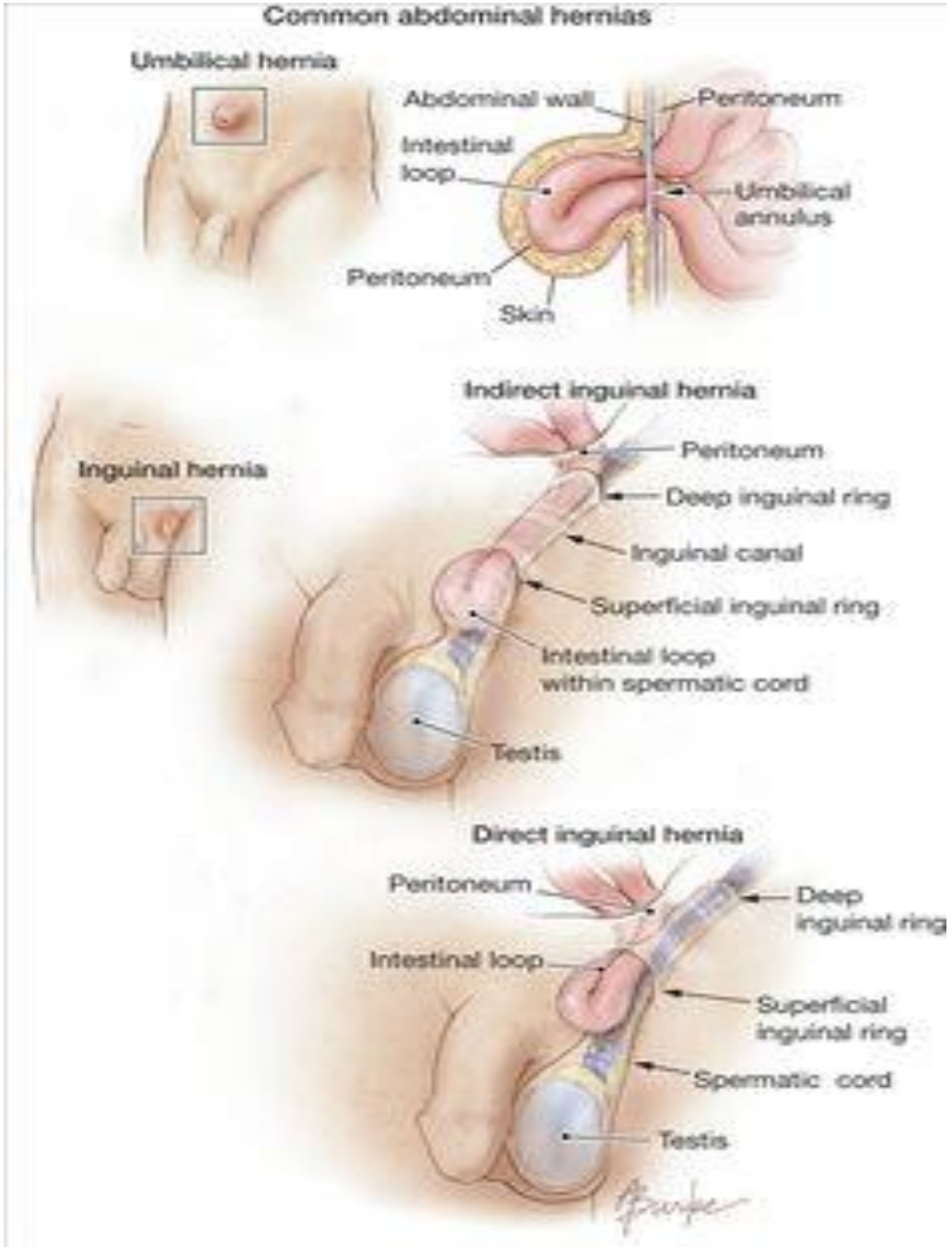
- Incarcerated if contents cannot be pushed back through hernia ring.
- Strangulated if blood supply impaired.
- Omentum is the most commonly herniated object.

Direct inguinal hernia

- Protrusion of abdominal contents through transversalis fascia.
- Hesselbach's triangle is bounded by the rectus abdominus muscle, the inferior epigastric vessels, and the inguinal ligament.
- Hernia medial to inferior epigastric artery.
- Rarely reaches scrotum.
- No sac.

Indirect inguinal hernia

- Protrusion of abdominal contents through an enlarged deep inguinal ring and a patent processus vaginalis.
- Hernia lateral to inferior epigastric artery.
- Follows spermatic cord to reach scrotum or labia (patent processus vaginalis).
- More common than a direct hernia.
- More common on the right.
- Sac.



Pinterest.com
Accessed
10/25/2019

Hernias

- Pantaloon hernia
- Hernia has both direct and indirect components.
- Femoral hernia
- Through the saphenous opening of the femoral ring to the anterior thigh (below the inguinal ring).
- More common in women.
- No sac.
- More likely to incarcerate.

Hernias

- Spigelian hernia:
- Along the lateral inferior border of the rectus muscle.
- Abdominal pain.
- Epigastric hernia:
- In the midline above the umbilicus. Abdominal pain.
- Richter's hernia:
- Incarcerated or strangulated hernia that involves only one side of bowel wall.
- Repair involves closing defect with the incorporation of a mesh for reinforcement.