The Digestive System

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Divisions of the Gut Tube

Formation of the Primitive Gut

- Due to cephalocaudal and lateral folding of the embryo:
    - A portion of the endoderm-lined yolk sac cavity is incorporated into the embryo → forms the primitive gut.
    - Remaining parts of the endoderm-lined cavity:
        - Yolk sac
        - Allantois
          → Stay outside the embryo.

Subdivisions of the Primitive Gut

- Foregut
    - Blind-ending tube at cephalic end.
- Hindgut
    - Blind-ending tube at caudal end.

- Midgut
    - Middle portion, temporarily connected to yolk sac via vitelline duct (yolk stalk).

Detailed Divisions

1. Pharyngeal Gut (Pharynx)
    - From oropharyngeal membrane → respiratory diverticulum.
    - Part of foregut, important for head & neck development.
2. Remainder of Foregut
    - From caudal to pharyngeal tube → liver outgrowth.
3. Midgut
    - From caudal to liver bud → junction of right 2/3 and left 1/3 of transverse colon (in adult).
4. Hindgut

- From left 1/3 of transverse colon → cloacal membrane.

Germ Layer Derivatives

- Endoderm →
  - Epithelial lining of digestive tract.
  - Parenchyma (functional cells) of glands: hepatocytes, pancreatic exocrine & endocrine cells.
- Visceral Mesoderm →
  - Stroma (connective tissue) of glands.
  - Muscle, connective tissue, peritoneal components of gut wall.

Mesenteries

Definition & Function

- Mesenteries = Double layers of peritoneum:
  - Enclose an organ.
  - Connect organ → body wall.
  - Provide pathways for vessels, nerves, and

lymphatics to abdominal viscera.

- Intraperitoneal organs
    - Completely enclosed by mesentery → e.g., stomach, intestines.
- Retroperitoneal organs
    - Lie against posterior body wall.
    - Covered by peritoneum only on anterior surface → e.g., kidneys.
- Peritoneal ligaments
    - Specialized mesenteries.
    - Double layers of peritoneum that pass organ ↔ organ or organ ↔ body wall.

Development of Mesenteries

- Initially:
    - Foregut, midgut, hindgut in broad contact with posterior abdominal wall mesenchyme.

- By 5th week:
    - Connecting tissue bridge narrows.
    - Caudal foregut, midgut, and major hindgut → suspended by dorsal mesentery.

Dorsal Mesentery

- Extends: Lower end of esophagus → cloacal region.
- Region-specific names:
    - Stomach → Dorsal mesogastrium (greater omentum).
    - Duodenum → Dorsal mesoduodenum.
    - Colon → Dorsal mesocolon.
    - Jejunum & ileum loops → Mesentery proper.

Ventral Mesentery

- Exists only in:
    - Terminal part of esophagus.
    - Stomach.
    - Upper part of duodenum.
- Origin: Derived from septum transversum.

- Liver growth into septum transversum → divides ventral mesentery into:

    1. Lesser omentum

        - From lower esophagus, stomach, upper duodenum → liver.

    2. Falciform ligament

        - From liver → ventral body wall.

✅ Clinical Correlate (Exam Tip):

- Malrotation or abnormal fixation of mesenteries → can cause intestinal volvulus (life-threatening twisting of intestines).

- Knowledge of mesentery is vital in surgical approaches (e.g., ligation of vessels within mesenteric folds).

Foregut Derivatives

Esophagus

Development

- ~4th week:
    - Respiratory diverticulum (lung bud) arises from ventral foregut (border with pharyngeal gut).
    - Tracheoesophageal septum partitions diverticulum from dorsal esophagus.
- Result → Foregut divides into:
    - Ventral portion = Respiratory primordium (future trachea, lungs).
    - Dorsal portion = Esophagus.

Growth

- Initially short, but lengthens rapidly as heart & lungs descend.

Muscle & Nerve Supply

- Muscular coat from splanchnic mesenchyme:
    - Upper 2/3 → Striated muscle (innervated by Vagus nerve).
    - Lower 1/3 → Smooth muscle (innervated by

Splanchnic plexus).

Clinical Correlates: Esophagus

- Esophageal Atresia ± Tracheoesophageal Fistula
    - Caused by abnormal partitioning of foregut (tracheoesophageal septum deviation).
    - Most common type: Upper esophagus → blind sac, distal esophagus → connected to trachea near bifurcation.
    - Consequence: Polyhydramnios (amniotic fluid not swallowed).
- Esophageal Stenosis
    - Usually lower 1/3.
    - Causes: Incomplete recanalization, vascular abnormalities, ischemic accidents.
- Congenital Hiatal Hernia
    - Failure of esophagus to lengthen sufficiently → stomach pulled into thorax via esophageal hiatus.

Stomach

Initial Appearance

- 4th week → Fusiform dilation of foregut.

Growth & Rotation

1. Longitudinal Axis Rotation (90° clockwise)
    - Left side → anterior, Right side → posterior.
    - Vagus nerves shift:
        - Left vagus → anterior wall.
        - Right vagus → posterior wall.
    - Posterior wall grows faster → Greater curvature.
    - Anterior wall slower → Lesser curvature.
2. Anteroposterior Axis Rotation
    - Pyloric end (caudal) → moves right & upward.
    - Cardiac end (cranial) → moves left & downward.
    - Final axis runs from above-left → below-right.

Effect on Mesenteries

- Stomach attached by:
    - Dorsal mesogastrium (→ greater omentum).

- Ventral mesogastrium (→ lesser omentum + falciform ligament).

- Longitudinal rotation:

  - Pulls dorsal mesogastrium left → forms Omental bursa (lesser peritoneal sac).
  - Pulls ventral mesogastrium right.

Spleen & Pancreas Development

- Spleen primordium:

  - Appears as mesodermal proliferation in dorsal mesogastrium (~5th week).
  - With rotation:
    - Connected to left kidney by Lienorenal ligament.
    - Connected to stomach by Gastrolienal ligament.

- Pancreas:

  - Initially grows into dorsal mesoduodenum.
  - Tail extends into dorsal mesogastrium.
  - Fusion of dorsal mesogastrium with posterior abdominal wall → tail becomes secondarily retroperitoneal (covered only on anterior surface).

✅ Clinical Correlates: Stomach & Mesenteries

- Malrotation of stomach → abnormal position of omental bursa & curvatures.
- Pancreas anomalies (e.g., annular pancreas) often linked to abnormal rotations.

⚡ Exam Tips

- Always mention two axes of stomach rotation (longitudinal + anteroposterior).
- Stress nerve supply shift (vagus nerves) → favorite MCQ/short note question.
- "Secondarily retroperitoneal" = organ initially intraperitoneal → fuses with posterior abdominal wall (Pancreas, parts of duodenum, ascending/descending colon).

Mesenteries & Omenta

Greater Omentum

- Rotation of stomach (anteroposterior axis) → dorsal mesogastrium bulges down.

- Continues to grow → double-layered sac hanging over transverse colon & small intestine like an apron.

- Later → layers fuse → single sheet hanging from greater curvature = Greater omentum.

- Posterior layer of greater omentum fuses with transverse mesocolon.

Ventral Mesogastrium → Lesser Omentum & Falciform Ligament

- Derived from septum transversum mesoderm.

- Liver cords grow into septum → thins → forms:

    1. Peritoneum of liver

    2. Falciform ligament (liver → ventral abdominal wall)

        - Free margin contains umbilical vein → after birth → Ligamentum teres hepatis (round ligament).

3. Lesser omentum (stomach & duodenum → liver)

    - Subdivisions:
        - Hepatogastric ligament
        - Hepatoduodenal ligament (free margin).

- Hepatoduodenal ligament contents (Portal Triad):

    - Bile duct
    - Portal vein
    - Hepatic artery

- Forms roof of Epiploic Foramen of Winslow (connection between omental bursa [lesser sac] & greater sac).

Clinical Correlates – Stomach Abnormalities

- Pyloric Stenosis

    - Hypertrophy of pyloric circular (±longitudinal) muscle.
    - One of the most common gastric abnormalities in infants.
    - Presentation: 3–5 days after birth, projectile, non-bilious vomiting.

    - Risk ↑ with neonatal erythromycin exposure.
    - Severe → pyloric atresia (rare).

- Other rare anomalies:

    - Gastric duplications.
    - Prepyloric septum.

Duodenum

Origin

- Formed from:

    - Terminal foregut + Cranial midgut.

- Junction → just distal to liver bud origin.

Rotation

- With stomach rotation → duodenum takes C-shaped loop, rotates to right.

- Growth of pancreatic head further shifts duodenum rightward.

Fixation

- Duodenum & head of pancreas pressed against dorsal body wall.

- Dorsal mesoduodenum fuses with posterior peritoneum → disappears.

- Duodenum + pancreatic head → secondarily retroperitoneal.

- Exception → small part near pylorus (duodenal cap) retains mesentery → remains intraperitoneal.

Lumen Changes

- 2nd month: epithelial proliferation → lumen obliterated.

- Soon after → recanalized (failure → duodenal atresia/stenosis).

Arterial Supply

- Dual blood supply (reflects dual origin):

    - Celiac artery (foregut part).
    - Superior mesenteric artery (midgut part).

✅ Exam High-Yield Summary

- Greater omentum = dorsal mesogastrium.
- Lesser omentum + falciform ligament = ventral mesogastrium (septum transversum origin).
- Epiploic foramen of Winslow → opening between greater & lesser sac, bounded by hepatoduodenal ligament.
- Duodenum = secondarily retroperitoneal, except duodenal cap.
- Pyloric stenosis = projectile, non-bilious vomiting (common neonatal surgical emergency).

Liver & Gallbladder Development

Origin & Early Development

- Appears mid-3rd week as an outgrowth from distal foregut endoderm → Hepatic diverticulum (Liver bud).
- Liver bud growth:

- Proliferates into septum transversum (mesoderm between pericardial cavity & yolk sac stalk).
- Connection with foregut narrows → bile duct.
- Small ventral outgrowth from bile duct → Gallbladder + Cystic duct.

Differentiation

- Endodermal liver cords intermingle with vitelline & umbilical veins → form hepatic sinusoids.

- Derivatives:

  - Endoderm → Hepatocytes (liver cells) & Biliary epithelium.
  - Mesoderm (septum transversum) → Hematopoietic cells, Kupffer cells, Connective tissue.

Peritoneal Relations

- Liver invades septum transversum → divides it into:

  - Lesser omentum (between liver & foregut).
  - Falciform ligament (between liver & ventral body

wall).
    - Together = Ventral mesentery.

- Mesoderm on liver surface → Visceral peritoneum, except:
    - Bare area (cranial surface in contact with septum transversum → future diaphragm).
    - Septum transversum mesoderm here → Central tendon of diaphragm.

Functional Development

- 10th week:
    - Liver = ~10% of fetal body weight.
    - Cause: Extensive hematopoiesis (major site of blood cell formation until last 2 months IU life).
- At birth → liver = ~5% body weight (hematopoiesis subsides).
- 12th week: Hepatocytes begin bile secretion.
    - Bile duct + cystic duct + hepatic duct → Bile duct system established.

    - Bile enters duodenum → contents turn dark green (meconium).

- Duodenal rotation → shifts bile duct entrance from anterior → posterior position (bile duct passes behind duodenum).

Clinical Correlates

- Normal Variants:
    - Accessory hepatic ducts.
    - Gallbladder duplication (usually asymptomatic).

- Extrahepatic biliary atresia (1/15,000 births):
    - Failure of bile ducts to recanalize.
    - 15–20%: patent proximal ducts → surgically correctable.
    - Others → fatal unless liver transplant.

- Intrahepatic biliary duct atresia / hypoplasia (1/100,000 births):
    - Often due to fetal infection.
    - May be lethal or follow benign chronic course.

✅ Exam High-Yield Pointers

- Hepatic diverticulum = endodermal origin.
- Kupffer cells, hematopoietic cells, connective tissue = mesodermal origin.
- Bare area of liver = contact with septum transversum (forms diaphragm's central tendon).
- Liver = main hematopoietic organ till late gestation.
- Bile secretion begins ~12th week → meconium green.
- Extrahepatic biliary atresia = surgical emergency in neonates.

Pancreas Development

Origin

- Develops from 2 buds (endodermal in origin, from duodenal lining):
    - Dorsal pancreatic bud → grows into dorsal

mesentery.
    - Ventral pancreatic bud → develops near bile duct.

Rotation & Fusion

- Duodenum rotates right → C-shaped.
- Ventral pancreatic bud moves dorsally → lies below & behind dorsal bud.
- Buds fuse → form definitive pancreas.

Derivatives

- Ventral bud → Uncinate process + Inferior part of pancreatic head.
- Dorsal bud → Remaining gland (body, tail, superior head).

Pancreatic Duct System

- Main pancreatic duct (of Wirsung):
    - Formed from distal part of dorsal duct + entire ventral duct.

    - Opens into major duodenal papilla with bile duct.

- Accessory pancreatic duct (of Santorini):

    - From proximal dorsal duct (if persists).
    - Opens into minor duodenal papilla.

- Failure of fusion (10% cases) → double duct system persists.

Endocrine Development

- 3rd month → Islets of Langerhans develop from parenchymal tissue.

- 5th month → Insulin secretion begins.

- Other endocrine cells (glucagon, somatostatin) also differentiate from parenchyma.

- Connective tissue → from surrounding visceral mesoderm.

Clinical Correlates

1. Annular Pancreas

- Ventral bud has two components. Normally, both rotate together → fuse below dorsal bud.
- If one rotates opposite direction → duodenum encircled by pancreatic tissue.
- May → duodenal obstruction (vomiting, polyhydramnios in fetus).

2. Accessory Pancreatic Tissue

- Can occur anywhere from esophagus → Meckel's diverticulum.
- Most common site = stomach mucosa or intestinal mucosa.
- Histologically resembles normal pancreas.

✅ Exam High-Yield Pointers

- Dorsal bud = main contributor.
- Ventral bud = uncinate + lower head.
- Main duct = fusion product; Accessory duct = remnant of dorsal duct.
- Insulin secretion begins ~5th month (important MCQ).

- Annular pancreas → duodenal obstruction (vomiting after birth).

- Accessory pancreatic tissue → can mimic other lesions but histology = pancreas.

Midgut Development

Basic Facts

- In 5-week embryo:
    - Midgut suspended by short dorsal mesentery.
    - Communicates with yolk sac via vitelline duct (yolk stalk).

- Extent in adult:
    - Begins → distal to bile duct opening in duodenum.
    - Ends → proximal 2/3 of transverse colon.

- Blood supply: Entire midgut = Superior Mesenteric Artery (SMA).

Primary Intestinal Loop

- Forms due to rapid elongation of midgut + mesentery.
- Loop apex connected to yolk sac → vitelline duct.
- Cephalic limb → distal duodenum, jejunum, part of ileum.
- Caudal limb → lower ileum, cecum, appendix, ascending colon, proximal 2/3 transverse colon.

Physiological Herniation

- 6th week: Rapid growth of midgut + large liver = abdomen too small.
- Loops herniate into umbilical cord (extraembryonic cavity).
- Called physiological umbilical herniation.

Rotation of Midgut

- Occurs around axis of SMA.
- Counterclockwise (viewed from front).

- Total = 270° rotation:
    - 90° during herniation.
    - 180° during return to abdominal cavity.
- Jejunum & ileum → form coiled loops.
- Large intestine → elongates, but no coiling.

Retraction of Herniated Loops

- 10th week: Loops return to abdomen due to →
    - Regression of mesonephric kidney.
    - Reduced liver growth.
    - Expansion of abdominal cavity.
- Order of return:
    - Jejunum → first (to left side).
    - Ileum → later (to right side).
    - Cecal bud = last to return (initially RUQ, later descends to RLQ).
    - Cecal bud forms → appendix as diverticulum.

- Appendix position: often retrocecal or retrocolic.

Mesenteries & Fixation

- Primary mesentery = mesentery proper.
- During rotation, dorsal mesentery twists around SMA.
- Fusion & fixation:
    - Ascending & descending colon → fuse with posterior wall → become retroperitoneal.
    - Appendix, cecum (lower part), sigmoid colon → retain free mesentery.
    - Transverse mesocolon → fuses with greater omentum → retains mobility.
- Final attachment of jejunoileal mesentery → from duodenojejunal junction to ileocecal junction.

Exam High-Yield Points

- Physiological herniation: 6th week → 10th week.
- Rotation total = 270° counterclockwise around SMA.

- Cecal bud last to re-enter; initial RUQ → final RLQ.
- Retroperitoneal: ascending + descending colon.
- Mobile: transverse colon, sigmoid colon, appendix, cecum.
- Appendix positions: retrocecal (most common), retrocolic.

Clinical Correlates – Midgut Development

1. Abnormalities of Mesenteries

- Normal: Ascending colon (except caudal 1 inch) → fuses with posterior abdominal wall → peritoneum only on front & sides.
- Mobile cecum:
  - Cause → Persistence of portion of ascending mesocolon.
  - Cecum remains mobile → abnormal movements.
  - Extreme form → entire ascending colon mobile → predisposes to volvulus.

- Retrocolic pockets:
    - Cause → Incomplete fusion of mesentery with posterior wall.
    - May trap small intestine → retrocolic hernia.

2. Body Wall Defects

A. Omphalocele

- Definition: Herniation of abdominal viscera through enlarged umbilical ring.
- Covering: Amnion + peritoneum (sac).
- Cause: Failure of bowel to return to abdominal cavity during 6th–10th weeks (physiological herniation persists).
- Contents: Liver, intestines, stomach, spleen, gallbladder (variable).
- Incidence: 2.5 / 10,000 births.
- Associations:

- High mortality (~25%).
- Cardiac anomalies (50%).
- Neural tube defects (40%).
- Chromosomal abnormalities (~15%).

B. Gastroschisis

- Definition: Protrusion of abdominal viscera directly into amniotic cavity.
- Site: Lateral to umbilicus (usually right).
- Cause: Defective closure of body wall near connecting stalk.
- Covering: No covering (not covered by amnion/peritoneum).
- Effect: Viscera exposed to amniotic fluid → damage to bowel.
- Incidence: 1 / 10,000 births (↑ frequency, especially in young mothers <20 years).
- Associations:

- Not linked with chromosomal abnormalities.
- Usually not associated with other anomalies.
- Good survival rate.

3. Volvulus

- Definition: Abnormal twisting of intestine → compromises blood supply.

- Complication: Ischemia → necrosis of large segments → may cause fetal death.

Exam Pointers

- Omphalocele vs Gastroschisis

Feature	Omphalocele	Gastroschisis
Site	Umbilical ring	Lateral to umbilicus (right)
Covering	Amnion + peritoneum sac	No covering

Cause	Failure of return of bowel (6–10 wk)	Defective closure of body wall
Associations	Chromosomal anomalies, cardiac, NTDs	No chromosomal anomalies
Prognosis	Poor (25% mortality)	Good survival

Clinical Correlates – Midgut (continued)

1. Vitelline Duct Abnormalities (2–4% of population)

Normally → vitelline duct obliterates by week 7.
Persistence → anomalies:

A. Meckel's Diverticulum (Ileal diverticulum)

- Outpouching of ileum on antimesenteric border, ~ 40–60 cm from ileocecal valve.

- Rule of 2's (exam favorite 📝):

    - 2% population
    - 2 feet from ileocecal valve
    - 2 inches long
    - Symptoms in 2% cases
    - 2 types of ectopic tissue: gastric & pancreatic

- Usually asymptomatic.

- If ectopic gastric/pancreatic tissue → ulceration, bleeding, perforation.

B. Vitelline Cyst (Enterocystoma)

- Central portion of duct persists → cyst formation.

- Both ends → fibrous cords.

- Risk: bowel strangulation/volvulus around fibrous strands.

C. Vitelline Fistula (Umbilical fistula)

- Entire duct remains patent.

- Direct communication between ileum & umbilicus.

- Clinical sign → fecal discharge at umbilicus.

2. Gut Rotation Defects

A. Malrotation

- Normal rotation = 270° CCW.
- Malrotation = only 90° rotation.
- Colon + cecum → return first → settle on left side (left-sided colon).
- Can predispose to volvulus → ischemia.

B. Reversed Rotation

- Rotation 90° clockwise.
- Effect: Transverse colon lies behind duodenum & SMA (instead of anterior).

3. Intestinal Duplications

- Cysts/diverticula anywhere along gut (MC in ileum).
- Range: long duplicated segment → small diverticulum.

- Symptoms: obstruction, bleeding (early in life).
- 33% associated with other defects: atresias, imperforate anus, gastroschisis, omphalocele.
- Cause: uncertain (possibly abnormal proliferation of gut parenchyma).

4. Gut Atresias & Stenoses

- Incidence: 1 in 1,500 births.
- Sites:
  - MC → duodenum
  - Rare → colon
  - Equal → jejunum & ileum

A. Causes

1. Duodenal atresia (proximal) → failure of recanalization.
2. Jejunal/ileal/colonic atresias → vascular accidents (malrotation, volvulus, gastroschisis, omphalocele).

3. New evidence: defective gut differentiation → misexpression of HOX, FGF genes.

B. Types of Atresia/Stenosis

1. Segmental loss (50%) → portion of bowel absent.
2. Fibrous cord (20%) → blind ends connected by cord.
3. Membranous stenosis (20%) → thin diaphragm partially obstructs lumen.
4. Multiple atresias (5%).
5. Multiple stenoses (5%).
6. Apple Peel Atresia (10%) → jejunal atresia with distal small bowel coiled around a short mesenteric remnant; short gut syndrome.

C. Clinical Impact

- Severity depends on site & length of involved bowel.
- Large involvement → low birth weight, malabsorption, high morbidity.

Summary Table (High-Yield)

Abnormality	Cause	Key Feature	Clinical Significance
Meckel's diverticulum	Persistence of vitelline duct	2 feet from IC valve, antimesenteric	Painless bleeding, ulcer, perforation
Vitelline cyst	Middle duct persists	Cyst with fibrous cords	Risk of volvulus/obstruction
Vitelline fistula	Entire duct patent	Ileum ↔ umbilicus	Fecal discharge at umbilicus
Malrotation	Incomplete 90° rotation	Left-sided colon	Volvulus risk
Reversed rotation	Clockwise 90°	Transverse colon behind SMA	Obstruction

Duplications	Abnormal proliferation	Ileum MC site	Early obstruction /bleeding
Atresias/Stenoses	Recanalization failure / vascular insult / genetic	Duodenum MC site	Obstruction, vomiting, low birth weight
Apple Peel atresia	Vascular accident	Jejunum, distal bowel spiraled	Short gut, malabsorption

Hindgut Development

I. Derivatives of Hindgut

- Distal 1/3 of transverse colon
- Descending colon
- Sigmoid colon
- Rectum
- Upper part of anal canal
- Endoderm of hindgut also contributes to → bladder & urethra (internal lining)

2. Cloaca and Partitioning

- Cloaca = common endoderm-lined cavity at embryo's caudal end
    - Posterior part → primitive anorectal canal (hindgut)
    - Anterior part → primitive urogenital sinus (allantois entry)
- Cloacal membrane = endoderm + surface ectoderm, forms ventral boundary
- Urorectal septum:
    - Derived from mesoderm (yolk sac covering + tissue around allantois)
    - Grows caudally to separate cloaca into:
        - Anterior = urogenital sinus
        - Posterior = anorectal canal
    - Tip of septum → forms perineal body (important landmark in obstetrics)
- By end of week 7 → cloacal membrane ruptures, creating:

- Anal opening (hindgut)
- Urogenital opening (urogenital sinus)

3. Development of Anal Canal

- Upper 2/3 → from hindgut endoderm
- Lower 1/3 → from ectoderm (proctodeum)

Steps:

1. Ectoderm around proctodeum proliferates → forms anal pit.
2. Anal pit deepens.
3. Anal membrane (former cloacal membrane) degenerates.
4. Continuity established between upper (endodermal) & lower (ectodermal) parts.

4. Blood Supply & Nerve Supply

Region of Origin	Blood Supply	Innervation	Epitheliu m
Anal			

Canal

Upper 2/3	Endoderm (hindgut)	Superior rectal artery (IMA)	Autonomic (insensitive to pain)	Columnar
Lower 1/3	Ectoderm (proctodeum)	Inferior rectal arteries (Internal pudendal)	Somatic (inferior rectal nerve, painful)	Stratified squamous

- Junction = Pectinate line (just below anal columns).
    - Landmark for epithelium, blood, lymph, innervation, clinical conditions (hemorrhoids, cancer spread).

Clinical Correlates – Hindgut Abnormalities

1. Rectourethral and Rectovaginal Fistulas

- Incidence: ~1/5,000 live births
- Cause: Abnormal cloaca/urorectal septum

development

- Cloaca too small OR urorectal septum fails to descend fully
- Result: Hindgut opens anteriorly → into urethra (male) or vagina (female)

- Clinical importance: Abnormal fecal passage through urinary/reproductive tract.

2. Rectoanal Fistulas & Atresias

- Spectrum of severity:
  - Narrow tube to surface
  - Fibrous remnant with no lumen

- Cause: Misexpression of genes disrupting epithelial–mesenchymal signaling

- Exam Pearl:
  - Imperforate anus = anal membrane fails to break down.

3. Congenital Megacolon (Hirschsprung Disease)

- Definition: Aganglionic megacolon due to absence of parasympathetic ganglia in bowel wall.

- Origin of ganglia: Neural crest cells (migrate into gut wall).

- Genetic cause: Often RET gene mutation (tyrosine kinase receptor guiding crest migration).

Clinical Features:

- Failure to pass meconium
- Abdominal distension
- Severe constipation

Distribution:

- Rectum always involved
- 80%: Extends up to sigmoid midpoint
- 10–20%: Involves transverse & right colon
- 3%: Whole colon affected

Exam Pointers 🎯

- Imperforate anus = anal membrane persists.

- Fistulas = urorectal septum maldevelopment.
- Hirschsprung's = neural crest migration failure → RET gene.
- Always correlate with blood supply & pectinate line (upper vs lower anal canal).

1) Polyhydramnios + frothy secretions + respiratory distress at birth

Most likely diagnosis:

Esophageal atresia with tracheoesophageal fistula (TEF) — classically proximal blind pouch with distal TEF.

Embryological basis: Abnormal partitioning of the foregut by the tracheoesophageal septum → esophagus ends blindly and/or connects to trachea.

Why polyhydramnios? Fetus can't swallow amniotic fluid → fluid accumulates.

Key complications: Aspiration, pneumonia, abdominal distension with ventilation.

2) 20-week scan: midline, membrane-covered mass containing bowel

Diagnosis: Omphalocele.

Embryological basis: Failure of physiologically herniated midgut (6–10 wks) to return to the abdominal cavity; viscera remain herniated through enlarged umbilical ring and are covered by amnion.

Prognosis: Guarded vs gastroschisis — high association with other anomalies (≈50% cardiac, ≈40% NTDs) and chromosomal defects (~15%); mortality ≈ 25%. Outcome depends mainly on associated anomalies and size of defect.

3) Newborn girl: meconium per vagina + no anal opening

Diagnosis: Rectovaginal fistula with imperforate anus.

Embryological basis: Abnormal cloacal partitioning — urorectal septum fails to descend/fuse adequately (or cloaca too small) → hindgut opens anteriorly into vagina; persistence of anal membrane → imperforate anus.