NOTES
LOWER GASTROINTESTINAL CONGENITAL MALFORMATIONS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Lower gastrointestinal tract structural/functional anomalies during embryonic development; present at birth
- Malformations from duodenum to anus

CAUSES
- Genetic, environmental factors

SIGNS & SYMPTOMS

- *At birth:* may be asymptomatic
- *Malformations:* relatively benign (nausea, vomiting, difficulty passing stool) to life incompatibility

DIAGNOSIS

DIAGNOSTIC IMAGING
- Prenatal ultrasound, MRI, CT scan/radiography
- Avoid X-ray due to teratogenicity

TREATMENT

SURGERY
- See individual disorders

GASTROSCHISIS

osms.it/gastroschisis

PATHOLOGY & CAUSES

- Extrasomatic protrusion of intestines through hole in abdominal wall near umbilicus
- *Hernia:* affected organs exit cavity
- *Week 4 of gestation:* lateral folds fail to fuse → hole in abdominal wall → organs protrude
- Most common on right side
- Usually small intestine
- Stomach, liver may also protrude (rare)

CAUSES
- Genetic, environmental factors

RISK FACTORS
- Mother's young age
- Exposure to teratogenic substances (alcohol, tobacco)

COMPLICATIONS
- Intestinal inflammation due to intrauterine exposure to amniotic fluid, malabsorption, infarction of intestinal tube due to compressed blood vessels, infection

SIGNS & SYMPTOMS

- *During fetal life:* asymptomatic
- *At birth:* difficulty feeding/passing stool

DIAGNOSIS

DIAGNOSTIC IMAGING

Intrauterine ultrasound, MRI

X-ray, CT scan
- Post-op evaluation

LAB RESULTS
- Increased maternal serum alpha-fetoprotein (MSAFP)

OTHER DIAGNOSTICS
- Defect visible at birth

TREATMENT

- Fatal if untreated

MEDICATIONS
- Antibiotics for existing/potential infection
- IV fluid/nutrients

SURGERY
- Surgical repositioning of organs back into abdominal cavity, closure of abdominal wall defect
- Usually requires multiple surgeries

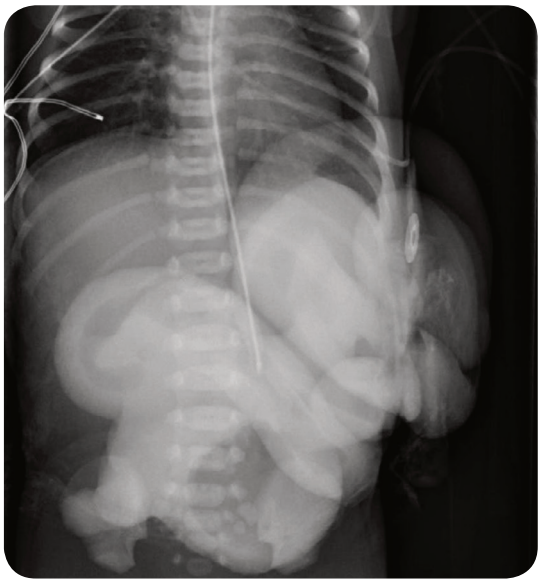

Figure 37.1 An abdominal X-ray of a newborn with gastroschisis. The abdominal contents are clearly visible outside the abdominal wall.

HIRSCHSPRUNG'S DISEASE

osms.it/hirschsprungs-disease

PATHOLOGY & CAUSES

- Myenteric (Auerbach), submucosal (Meissner) plexuses absent from intestinal wall in distal bowel
- AKA congenital aganglionic megacolon
- Absent plexuses (regulate bowel function) → intestine muscles permanently constricted → passing stool difficult, impossible

CAUSES

- Failure of neuroblasts to migrate from neural crest to intestine, form plexuses
- *Genetic:* *RET* proto-oncogene, *EDNRB*, etc.
- *RET proto-oncogene:* sporadic/autosomal dominant (familial) cases; associated with Down syndrome
- *Isolated:* sporadic/autosomal dominant
- *Present within syndrome:* Down syndrome, multiple endocrine neoplasia II, etc.

COMPLICATIONS

- Constipation/obstipation, malnutrition, enterocolitis, intestinal perforation, megacolon

SIGNS & SYMPTOMS

- *At birth:* asymptomatic
- Can be diagnosed in adulthood
- *First sign:* baby's inability to pass meconium, 48 hours postpartum
- Vomiting, abdominal distension, colics

DIAGNOSIS

DIAGNOSTIC IMAGING

- Barium assisted radiography

LAB RESULTS

- Rectal suction biopsy

OTHER DIAGNOSTICS

- Digital rectal exam

TREATMENT

SURGERY

- Surgical resection of intestine, subsequent fusion of remaining healthy tissue (pull-through technique)

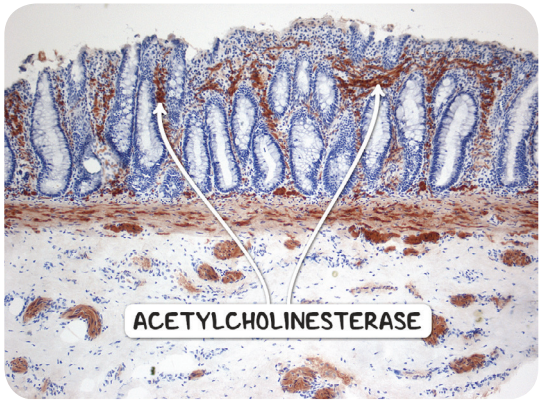

Figure 37.2 Immunohistochemical staining for acetylcholinesterase in the colon of an individual with Hirschprung's disease. Ganglia are absent resulting in overstimulation of nerves and increased levels of acetylcholinesterase.

IMPERFORATE ANUS

osms.it/imperforate-anus

PATHOLOGY & CAUSES

- Narrowed anal opening (anal stenosis)/ complete atresia
- AKA anal atresia
- Anus completely closed → colon ends in blind pouch in pelvis/opens into other pelvic structures (bladder, vagina) via fistulae
- All pelvic structures open into same channel → persistent cloaca
- Nerve, muscle tissue of missing parts of anus, rectum missing/malformed

CAUSES

- Mostly unknown genetic cause
- *HLXB9 gene:* only when imperforate anus is present within Currarino syndrome

COMPLICATIONS

- Megacolon, intestinal rupture, septic shock, incontinence/constipation (even after surgery)

MNEMONIC: VACTERL

Group of malformations with common, unknown cause

Vertebral anomalies
Anal atresia
Cardiovascular anomalies
Tracheoesophageal fistula
Esophageal atresia
Renal anomalies
Limb defects

SIGNS & SYMPTOMS

- Constipation (if anus narrowed), obstipation (if anus non-existent)
- Vomiting, abdominal distension

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI, ultrasound, X-ray/CT scan

- Determine internal extent of defect, plan corrective surgery

OTHER DIAGNOSTICS

- Physical exam at birth, defect visible

TREATMENT

SURGERY

- Anoplasty if possible, colostomy if not

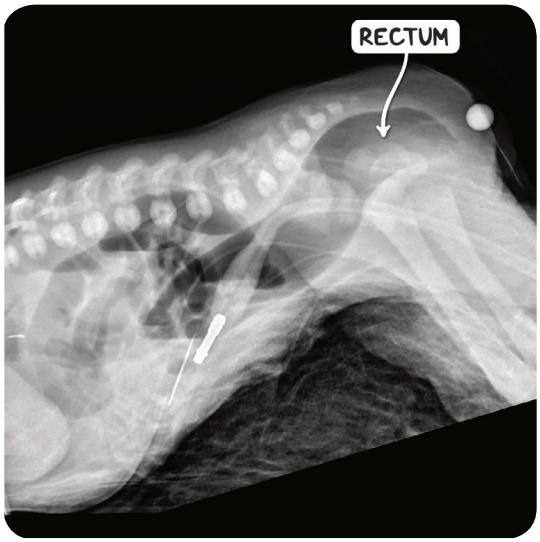

Figure 37.3 A lateral X-ray of a neonate demonstrating an imperforate anus. The rectum is dilated and the anal canal is absent.

INTESTINAL ATRESIA

osms.it/intestinal-atresia

PATHOLOGY & CAUSES

- Congenital malformation resulting in closed/absent part of small/large intestine
- Different from intestinal stenosis; in stenosis the passageway exists, and is just narrowed

TYPES

- *Named according to affected portion of intestine:* duodenal, jejunal, ileal, colonic
- Divided into duodenal/non-duodenal intestinal atresia due to different mechanism of origin
- Duodenal intestinal atresia is caused by failure in duodenal vacuolization
  - During fetal development duodenal epithelium proliferates rapidly → complete duodenal obstruction (AKA solid phase of vacuolization)→ apoptosis of excess cells → formation of small vacuoles which fuse → re-establish duodenal passageway (AKA recanalization phase)

CAUSES

- Duodenal intestinal atresia
  - Strongly associated with trisomy 21 (Down syndrome)
- Non-duodenal intestinal atresias
  - Intrauterine ischemic injury (small part of duodenum, entire jejunum, ileum, colon receive vascularization from superior mesenteric artery)

COMPLICATIONS

- Distension of stomach and duodenum caused by accumulated amniotic fluid which has nowhere to go
- Polyhydramnios (accumulation of amniotic fluid in amniotic sac)
  - Fetus swallows less fluid due to intestinal obstruction → more fluid accumulates in amniotic sac
- Intestinal perforation and pneumoperitoneum/meconium peritonitis

SIGNS & SYMPTOMS

- Bilious vomiting, abdominal pain, malnutrition

DUODENAL VS NON-DUODENAL INTESTINAL ATRESIA

	DUODENAL	NON-DUODENAL
PATHOLOGY	Error in vacuolization	Decreased perfusion → ischemic necrosis
CAUSES	Genetic	Unknown

DIAGNOSIS

DIAGNOSTIC IMAGING

Prenatal ultrasound

- To assess signs of obstruction; detectable in the third trimester
  - *Duodenal atresia:* dilated fluid-filled stomach adjacent to dilated duodenum
  - *Non-duodenal intestinal atresia:* Dilated fluid-filled bowel loops
  - Polyhydramnios

Postnatal X-ray

- *Duodenal atresia:* Double bubble sign (dilated stomach adjacent to dilated duodenum)
- *Non-duodenal intestinal atresia:* dilated bowel loops with air-fluid levels proximal to the obstruction

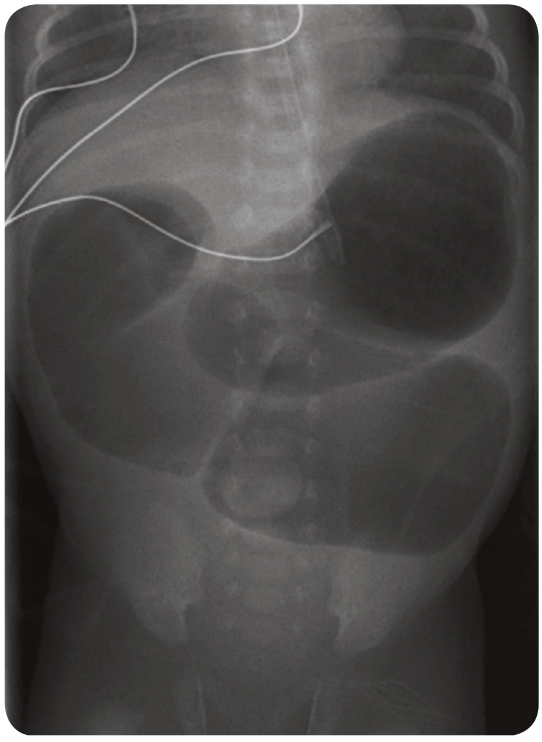

Figure 37.4 A plain abdominal radiograph of a neonate demonstrating the triple bubble sign of jejunal atresia.

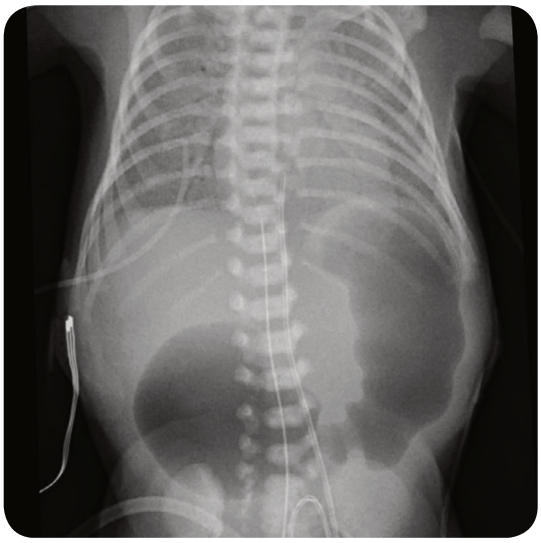

Figure 37.5 A plain abdominal radiograph of a neonate demonstrating the double bubble sign of duodenal atresia.

OTHER DIAGNOSTICS

- Physical examination
  - Apple peel (spiral) shape of intestines upon visual examination during surgery
- Amniocentesis to determine possible trisomy 21

TREATMENT

SURGERY

- *Gastric decompression:* removal of fluid from stomach
- IV fluid compensation
- Surgical reattachment of functional portions of intestines
  - In case of duodenal intestinal atresia → duodenoduodenostomy

INTESTINAL MALROTATION

osms.it/intestinal-malrotation

PATHOLOGY & CAUSES

- Improper rotation of midgut during embryogenesis
- Rapid midgut growth in restricted space → herniation into umbilical coelum → rotation 270° around SMA → error occurs = final anatomy description
  - Small intestine lodges into left abdominal cavity → cecum in lower right quadrant, first two thirds of colon lodge into right side over small intestine
- Due to error, several organs in incorrect anatomical position
  - Small intestine in right side
  - Coecum in epigastrium
  - Appendix follows coecum
  - Ladd's bands span over vertical duodenum, compressing from outside
  - Suspensory muscle of duodenum further right
  - Mesentery of small intestine narrower root

COMPLICATIONS

- Omphalocele, volvulus (part of intestine rotates around itself/part of mesenterium → blocks passage of intestinal content → compresses blood vessels → obstructs blood flow), ileus, ischemic bowel, malnutrition, hernias

SIGNS & SYMPTOMS

- May be asymptomatic
- Colic, bilous regurgitation, abdominal distension

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI/CT scan/barium-assisted radiography

- Detect improper organ position

TREATMENT

SURGERY

- Surgical repositioning of intestines, resection of Ladd's bands to remove duodenal obstruction
- Preventive appendectomy

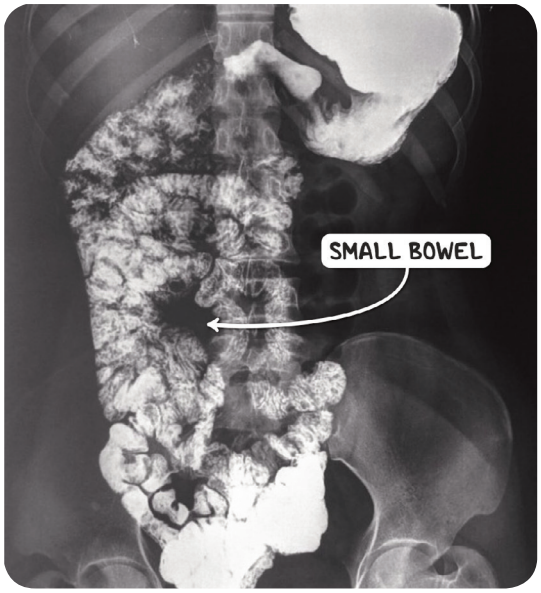

Figure 37.6 An abdominal X-ray with bowel contrast demonstrating intestinal malrotation. The entire small bowel is situated on the right side of the abdomen.

MECKEL'S DIVERTICULUM

osms.it/meckels-diverticulum

PATHOLOGY & CAUSES

- Abnormal pouch on antimesenteric side of ileum
- True diverticulum (contains all three layers of intestinal wall)
- *Early fetal life:* nutrients received from yolk sac into ileum via omphalomesenteric duct until it obliterates (week 5–6 of pregnancy)
- *If omphalomesenteric duct obliterates improperly:* Meckel's diverticulum
- May contain ectopic epithelia, omphalomesenteric duct lined with pluripotent cells

COMPLICATIONS

- Diverticulitis, ulcers from HCl secretion if gastric mucosa present, perforation of diverticulum, food impaction, lithiasis, peritonitis, peritoneal adhesions, intussusception, volvulus, neoplasms

MNEMONIC

Meckel's Rule of 2s

Symptomatic presentation before **2 years** of age

2% of population

Approximately **2 feet** from ileocecal valve

2 inches in length

2 types of ectopic mucosa (pancreatic, gastric)

SIGNS & SYMPTOMS

- Usually asymptomatic
- Abdominal pain/distension, melena, vomiting, constipation

DIAGNOSIS

DIAGNOSTIC IMAGING

Abdominal ultrasound/CT scan

- Incidental finding

Angiography

OTHER DIAGNOSTICS

Meckel's scan

- In children; technetium-99m procedure, detects gastric mucosa in diverticulum

Surgery

- Incidental finding

TREATMENT

SURGERY

- *Uncomplicated:* resection of diverticulum
- *Complicated:* resection of diverticulum, intestine

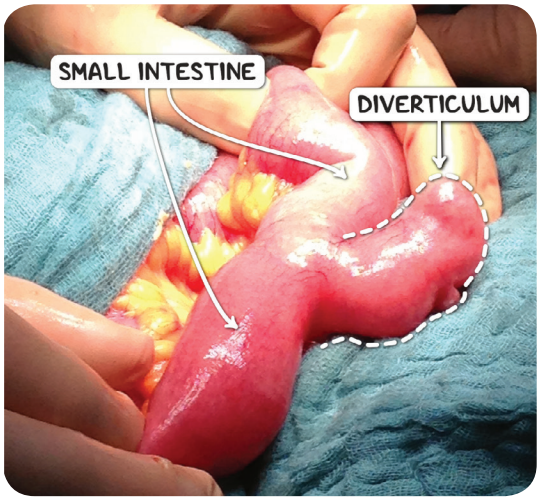

Figure 37.7 Intraoperative photograph of a Meckel's diverticulum.

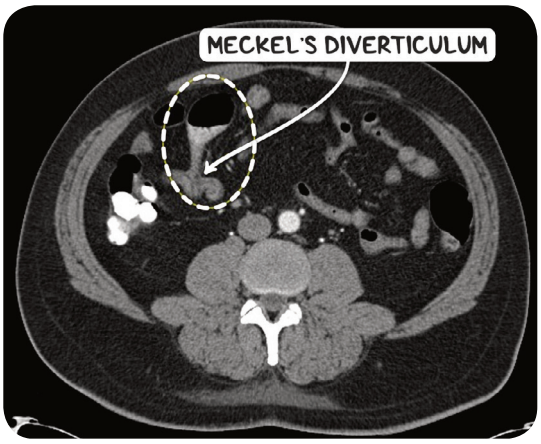

Figure 37.8 A CT scan in the axial plane demonstrating a Meckel's diverticulum.

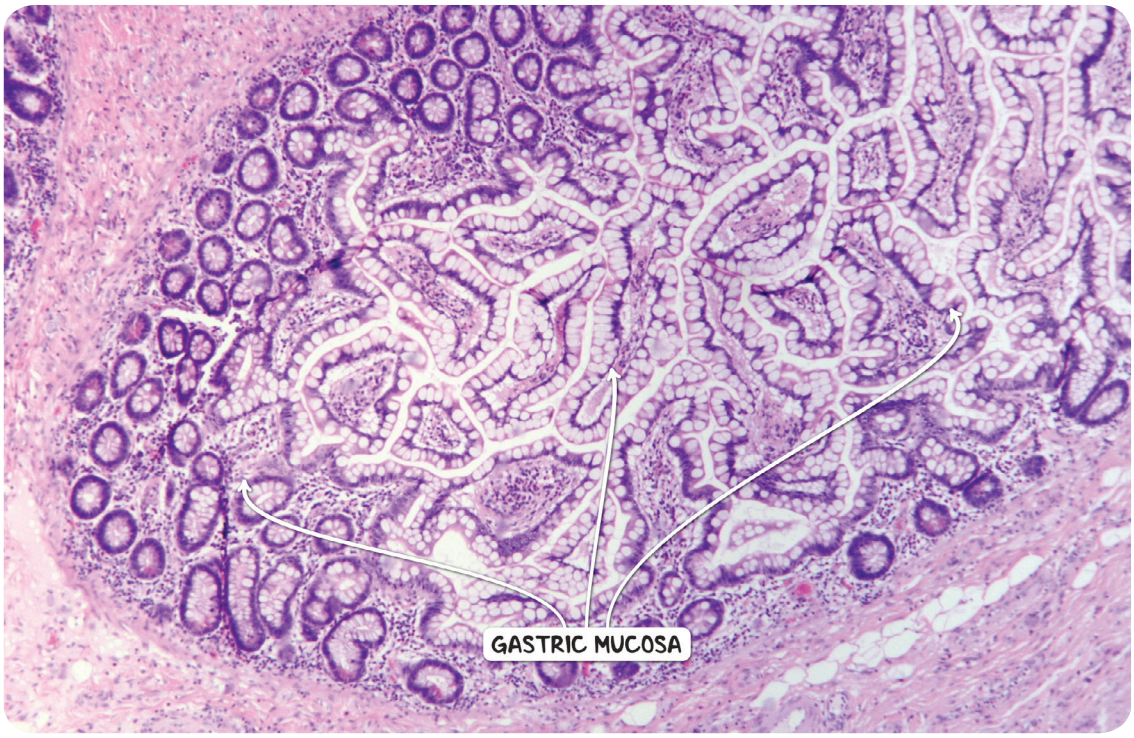

Figure 37.9 Histological appearance of a Meckel's diverticulum containing ectopic gastric mucosa.

OMPHALOCELE

osms.it/omphalocele

PATHOLOGY & CAUSES

- Persistent, pathological, herniation of intestines into umbilical cord
- Midgut herniates through umbilicus → pulls layer of peritoneum into umbilical cord in order to properly develop (grow, rotate) due to insufficient space in abdominal cavity → returns into abdomen
- *Midgut doesn't return:* omphalocele
- High mortality rate

CAUSES

- Genetic, environmental factors
- *Associated with:* trisomy 13 (Patau syndrome), trisomy 18 (Edward's syndrome), trisomy 21 (Down syndrome), Beckwith–Wiedemann syndrome

RISK FACTORS

- Consumption of alcohol/tobacco during pregnancy, certain medications (SSRIs), obesity

COMPLICATIONS

- Abdominal cavity malformation, volvulus, ischemic bowel

SIGNS & SYMPTOMS

- *Intrauterine:* asymptomatic
- *At birth:* visible defect

DIAGNOSIS

DIAGNOSTIC IMAGING

Intrauterine ultrasound

MRI

LAB RESULTS

- Blood test for MSAFP
- Amniocentesis

TREATMENT

SURGERY

- Surgical repositioning of protruding organs

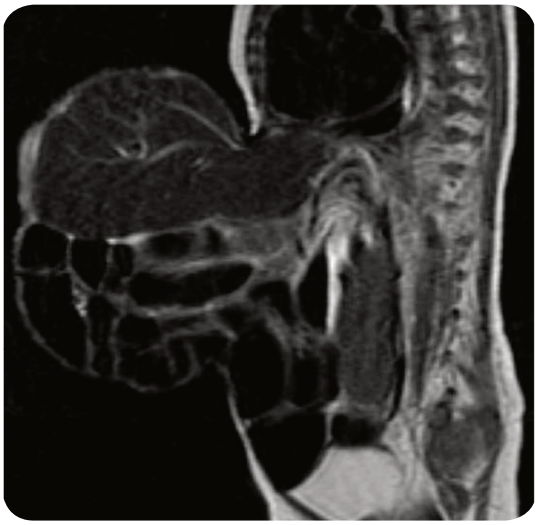

Figure 37.10 An MRI scan in the sagittal plane demonstrating a large omphalocele. The abdominal organs are clearly visible outside the abdominal wall.

GASTROSCHISIS VS OMPHALOCELE

GASTROSCHISIS	OMPHALOCELE
Herniated organs exposed to air	Herniated organs covered by peritoneum
Organs protrude through lateral opening in abdominal wall	Organs protrude through umbilicus
Small defect in abdominal wall	Large defect in abdominal wall