NOTES
UPPER GASTROINTESTINAL CONGENITAL MALFORMATIONS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Upper gastrointestinal tract structural/functional anomalies during embryonic development; present at birth

CAUSES
- Genetic, environmental factors

SIGNS & SYMPTOMS

- May be asymptomatic/complete dysfunction of gastrointestinal (GI), life incompatibility

DIAGNOSIS

DIAGNOSTIC IMAGING
- Prenatal ultrasound; MRI
- X-ray/CT scan
  - Avoid if possible due to teratogenicity

TREATMENT

SURGERY
- See individual disorders

OTHER INTERVENTIONS
- Nasogastric intubation

CLEFT LIP & PALATE

osms.it/cleft-lip-and-palate

PATHOLOGY & CAUSES

- Group of congenital malformations in upper lip, oral cavity roof
- Result of improper fusion of facial bones, associated tissues

TYPES
- Based on severity

Cleft lip (CL, cheiloschisis)
- Unilateral, bilateral "hare lip"

Cleft palate (CP, palatoschisis)
- Commonly uvula also split

Combination (CLP, cheilopalatoschisis)
- Most severe forms; split alveolar ridge, uvula (cheilognathopalatoschisis)

RISK FACTORS
- Other inherited genetic disorders (e.g. Patau syndrome, Stickler syndrome)
- Environmental teratogenic factors (e.g. intrauterine hypoxia, pesticides, anticonvulsant medication, folate deficiency)

COMPLICATIONS
- Speech impediments, hearing issues/recurrent otitis media, difficulty eating

SIGNS & SYMPTOMS

- Velopharyngeal insufficiency
  - Inability to temporarily stop physical communication between oral, nasal cavities
- Dysphonia
  - Air leaks to nasal cavity → hypernasal vocalization
- Dysarthria
  - Abnormal structure increases speech difficulty → distorted word structure
- Nasal cavity infection
  - Food trapped in nasal cavity → predisposes infection

DIAGNOSIS

DIAGNOSTIC IMAGING

Prenatal ultrasound

- Evaluation of integrity of nares, upper lip, hard and soft palate
- 3D reconstruction and surface rendering allow for better diagnosis and help parents prepare psychologically

MRI

- Evaluation of associated extra/intracranial abnormalities
- Prenatal MRI aids in confirmation and characterization/integrity of maxillary arch

CT scan/X-ray

- Not typically used; 3D reconstructions can aid in surgical planning

OTHER DIAGNOSTICS

- Clinically evident at birth

TREATMENT

SURGERY

- Surgical closure of cleft lip by three months of age
- Timing for surgical closure of palate is variable; usually done by one year of age

OTHER INTERVENTIONS

- Temporary prosthetic implants, until surgery
- Speech-language therapy
- Folate supplementation during pregnancy decreases risk

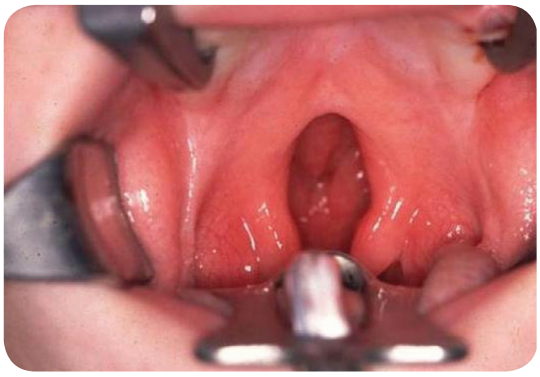

Figure 43.1 A cleft hard palate in an infant.

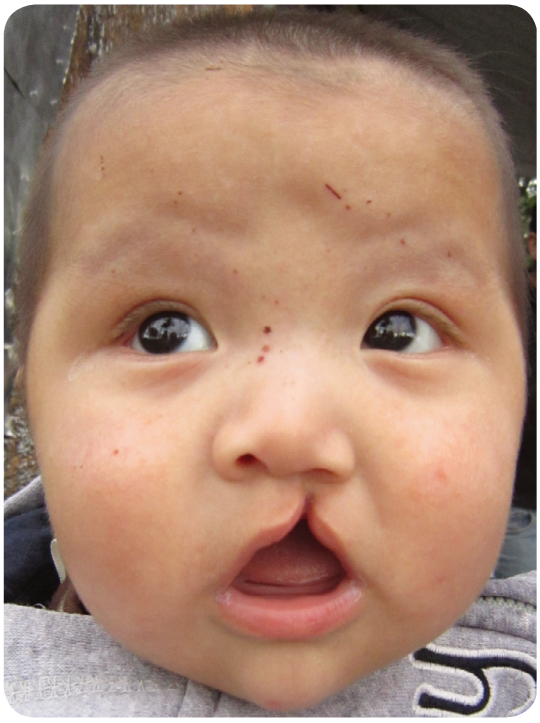

Figure 43.2 A child with a unilateral, incomplete cleft lip.

CONGENITAL DIAPHRAGMATIC HERNIA (CDH)

osms.it/congenital-diaphragmatic-hernia

PATHOLOGY & CAUSES

- Protrusion of abdominal viscera into chest cavity
- Results from abnormal development of diaphragm in utero
- High mortality rate
- Incomplete diaphragm formation → abdominal organs protrude into chest cavity → physical obstruction of heart, lung formation/function → pulmonary hypoplasia, surfactant deficiency, pulmonary hypertension, arrhythmia

TYPES

Bochdalek hernia

- Posterolateral diaphragmatic hernia; most common CDH
  - Viscera protrude through posterolateral segment of diaphragm
  - Left kidney, perinephric fat, stomach, small intestine

Morgagni hernia

- Retrosternal, parasternal diaphragmatic hernia
  - Viscera protrude through foramina of Morgagni (form sternocostal angle)

CAUSES

- Genetic, environmental factors

SIGNS & SYMPTOMS

- Dyspnea, tachypnea, central cyanosis, tachycardia, retractions, nasal flaring, decreased/absent breath sounds on affected side, scaphoid abdomen

MNEMONIC: 5Bs

Bochdalek hernia features

Bochdalek hernia

Big

Back and medial, usually left side

Baby

Bad: associated with pulmonary hypoplasia

DIAGNOSIS

DIAGNOSTIC IMAGING

Prenatal ultrasound

- Polyhydramnios
- Cardiomediastinal shift with possible abnormal cardiac axis
- Lack of visualization of normal stomach bubble
- Absent bowel loops in abdomen; stomach and small bowel in thorax
- Intrathoracic herniation of liver (seen in 85%, poor prognosis)
- Peristaltic bowel movements in thorax
- Reduced abdominal circumference

X-ray

- indistinct diaphragm, opacification of hemithorax (typically left-sided)

MRI

- Helpful in further assessment of pulmonary hypoplasia
- Measurement of fetal lung volumes

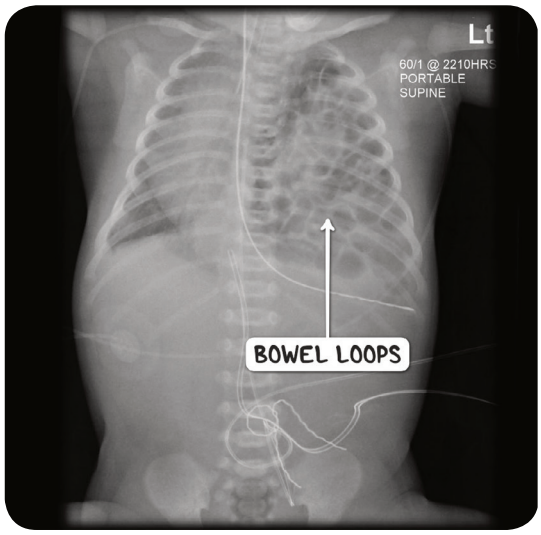

Figure 43.3 A plain X-ray of a newborn demonstrating visible bowel loops in the thoracic cavity.

TREATMENT

SURGERY

- Surgical repair of hernia

OTHER INTERVENTIONS

- Planned delivery after week 37 of gestation → immediate intubation, mechanical ventilation
- Inhaled nitric oxide for severe pulmonary hypertension
- Nasogastric, pulmonary intubation

ESOPHAGEAL WEB

osms.it/esophageal-web

PATHOLOGY & CAUSES

- Rare narrowing of esophagus due to thin membrane of esophageal tissues (mucosa, submucosa)
- Most commonly appear in lower third of esophagus
- Can be congenital/acquired
- May occur as solitary disease

RISK FACTORS

- Plummer–Vinson syndrome
  - Sideropenic dysphagia, iron-deficiency anemia, glossitis, cheilosis, esophageal webs

COMPLICATIONS

- Food impaction, perforation by solid food/esophageal probe insertion

SIGNS & SYMPTOMS

- May be asymptomatic (if small)
- *Dysphagia:* difficulty in swallowing
- *Odynophagia:* painful swallowing
- *Retrosternal pain:* can be mistaken for angina pectoris

DIAGNOSIS

OTHER DIAGNOSTICS

Fluoroscopy/barium swallow

- Visualized when esophagus is fully distended with contrast
- "Jet effect" of contrast being ejected distally from web

TREATMENT

OTHER INTERVENTIONS

- Endoscopic dilation via inflated balloon

HYPERTROPHIC PYLORIC STENOSIS

osms.it/hypertrophic-pyloric-stenosis

PATHOLOGY & CAUSES

- Constriction of pylorus due to pyloric sphincter hypertrophy → gastric outflow obstructed
- Autosomal dominant/multifactorial

RISK FACTORS

- Firstborn, biologically male, parents had hypertrophic pyloric stenosis, macrolide exposure

COMPLICATIONS

- Dehydration, malnourishment, acid-base imbalance

SIGNS & SYMPTOMS

- Projectile nonbilious vomiting at/soon after birth
- Visible peristalsis
- Dehydrated, undernourished

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Distended stomach, minimal intestinal gas

Ultrasound

- Modality of choice; but cannot exclude midgut volvulus
- Pyloric muscle thickness

Fluoroscopy

- Delayed gastric emptying
- Elongated pylorus with narrow lumen
- Entrance to pylorus may be beak shaped

OTHER DIAGNOSITCS

- Abdominal olive palpable on physical examination

TREATMENT

SURGERY

- Pyloromyotomy

OTHER INTERVENTIONS

- Rehydration
- Regulate acid-base status, correct electrolyte abnormalities

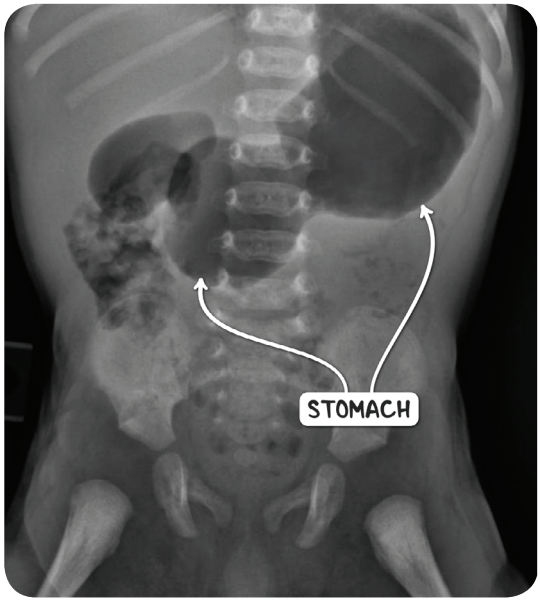

Figure 43.4 An abdominal radiograph demonstrating a grossly dilated stomach, secondary to obstructive pyloric stenosis.

THYROGLOSSAL DUCT CYST

osms.it/thyroglossal-duct-cyst

PATHOLOGY & CAUSES

- Benign cyst; epithelium of unclosed thyroglossal duct
- Thyroid cells migrate from foramen cecum downward → leave thyroglossal duct → thyroid duct stays open → fills with mucus → cyst forms

COMPLICATIONS

- Infection (spread from respiratory system), inflammation, discharging sinus with skin (secondary to inflammation/trauma), thyroid gland malformation (if thyroid cells remain in thyroglossal duct/cyst), extrathyroid thyroid carcinoma (from leftover thyroid cells)

SIGNS & SYMPTOMS

- Painless mass in front of neck, moves when swallowing; inflammation, pain; dysphagia; dyspnea

DIAGNOSIS

DIAGNOSTIC IMAGING

Ultrasound

- Fluctuant mass filled with anechoic fluid, thin walled, without vascularity

CT scan

- Thin-walled, well-defined homogeneous, fluid dense lesions, anterior midline/paramedian location
- May demonstrate capsular enhancement
- Sternocleidomastoid muscle may be displaced posteriorly/posterolaterally
- May be embedded in infrahyoid muscles

OTHER DIAGNOSTICS

- Fluctuant mass palpable at a anterior midline/paramedian location
- Draining sinus may be visible

TREATMENT

SURGERY

- Surgical excision (Sistrunk procedure)

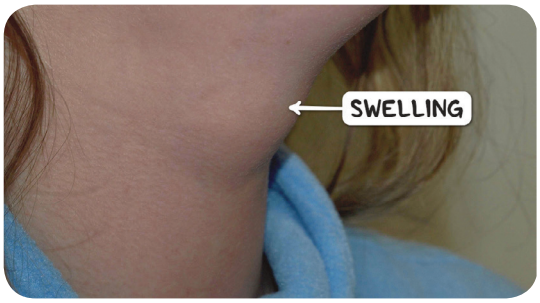

Figure 43.5 The clinical appearance of a thyroglossal duct cyst. There is a vague, fluctuant swelling in the midline of the neck.

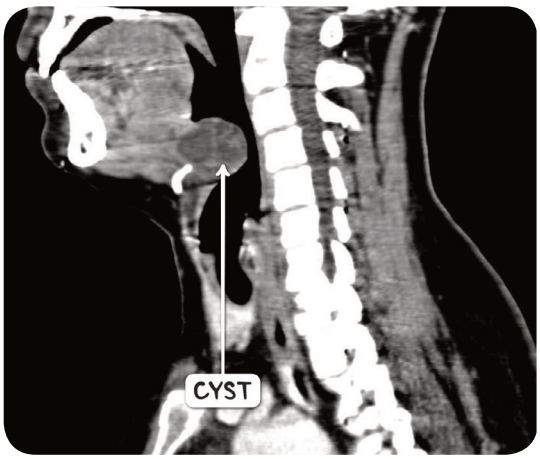

Figure 43.6 A CT scan of the head and neck in the sagittal plane demonstrating a thyroglossal duct cyst adjacent to the hyoid bone.

TRACHEOESOPHAGEAL FISTULA

osms.it/tracheoesophageal-fistula

PATHOLOGY & CAUSES

- Pathologic communication between trachea, esophagus
- Results from tracheoesophageal ridge fusion failure
- Occurs as congenital malformation/surgery complication (later in life)
- VACTERL association; see mnemonic

MNEMONIC: VACTERL

Group of malformations with common, unknown cause

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

TYPES

Type A
- Middle esophageal segment missing

Type B
- Proximal esophagus communicates with trachea

Type C (most common)
- Distal esophagus communicates with trachea, proximal esophagus atresia

Type D
- Proximal, distal esophageal segments communicate with trachea, middle segment atresia

Type E (AKA Type H)
- Complete esophagus, additional part communicates with trachea

COMPLICATIONS

- Atresia (due to hydrochloric acid accumulation), gastroesophageal reflux, dysphagia, frequent respiratory infections

SIGNS & SYMPTOMS

- Hypersalivation/drooling, choking, vomiting, central cyanosis upon feeding

DIAGNOSIS

DIAGNOSTIC IMAGING

Chest X-ray
- Nasogastric tube coiled in proximal esophagus (usually sufficient for diagnosis)

Fluoroscopy/Barium swallow
- If difficult to diagnose, may require contrast swallow study to visualize contrast passing into tracheobronchial tree
  - Barium is contrast medium of choice (ionic iodinated medium can cause chemical pneumonitis)

CT scan
- Useful for preoperative planning

OTHER DIAGNOSTICS

- Inability to pass gastric tube
- Neonates drool, choke, vomit during first feeding

TREATMENT

SURGERY

- Surgical closing of pathologic communication, fusion of esophageal buds

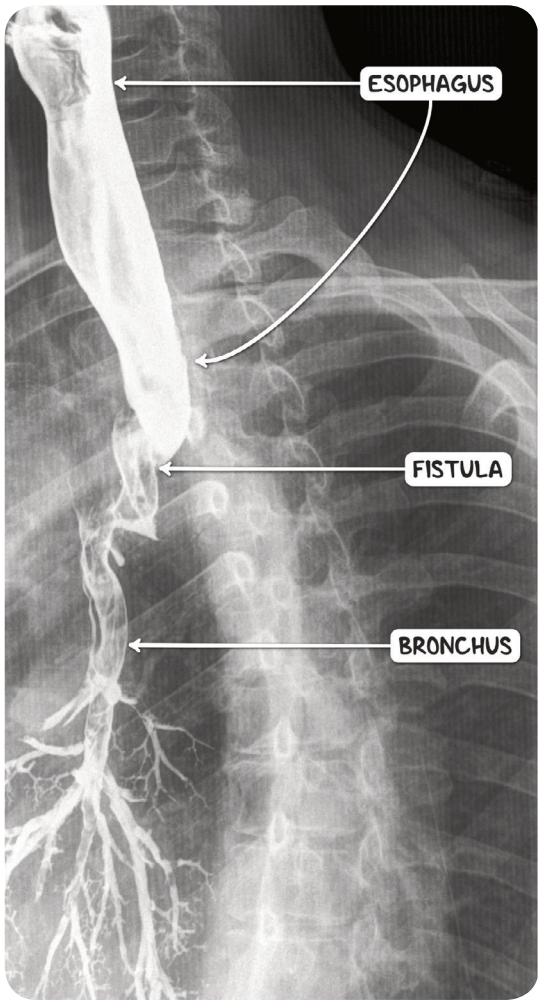

Figure 43.7 An acquired tracheo-esophageal fistula.