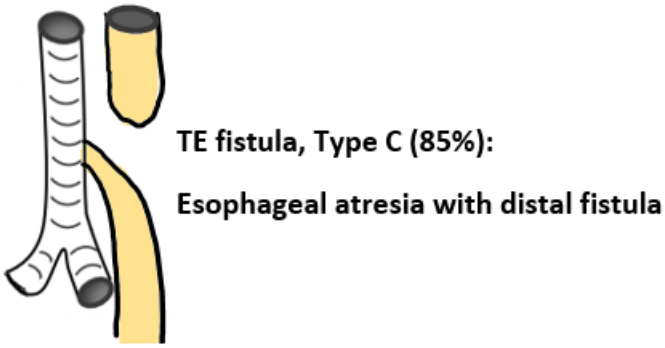
GI tract: Esophagus

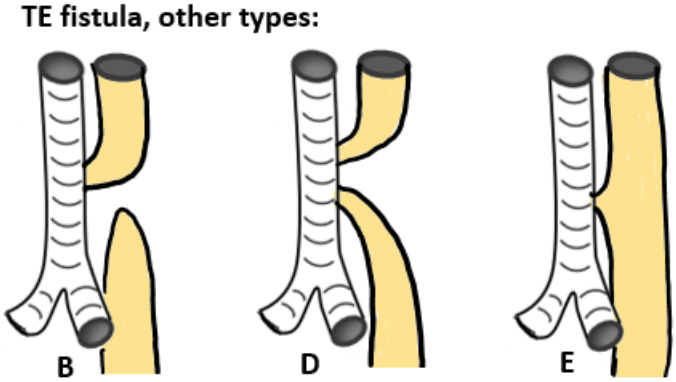
Introduction:

- The commonest presentation of esophageal disease is heartburn from reflux esophagitis. This does not need investigation unless there are alarm symptoms, such as new onset after age >60 years, evidence of blood loss, weight loss or dysphagia.
- Dysphagia history requires differentiation of difficulty swallowing both solids and liquids (motor disorder) versus solids only (mass such as carcinoma).

Congenital/hereditary disease: **Tracheal-esophageal fistula (TE fistula)**

- Pathophysiology:
    - Embryologic defect in the lateralization of the trachea and the esophagus in the foregut





Tracheal-esophageal fistulas

- Presentation:
    - Symptoms:
        - Regurgitation with first feed
        - Respiratory distress is an uncommon variant with aspiration

Vomiting in the newborn

Vomiting is the forceful ejection of gastric contents by the reflex of reverse peristalsis coordinated with relaxation of the lower esophageal sphincter. **Regurgitation** is the passive upwelling of food as would occur from the esophagus.

- Tracheoesophageal fistula: regurgitation on first feed
- Pyloric stenosis: non-bilious vomiting after 4 weeks
- Duodenal atresia/ annular pancreas: bilious vomiting after 1-2 days

    - P/E: non-contributory
    - Testing: imaging
- Natural history: surgical correction

Acute diseases

Gastroesophageal reflux disease (GERD)

Presentation

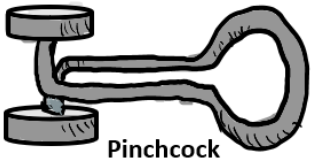
- Symptoms:
    - Usually heartburn. Other important but less common symptoms include
        - Wheezing
        - Metallic taste in the mouth
        - Chest pain
- Signs:
    - Physical examination is non-contributory
- Diagnosis:
    - For the classic presentation, the diagnosis is clinical, and the patient can be placed on PPI medication
    - Investigation is indicated with ***Alarm Symptoms***.
    - Endoscopy and biopsy
        - Inflammatory changes in the deep epithelium
    - Esophageal pH monitoring
        - Episodes of acidosis

Alarm symptoms for GERD
- New onset >60 years
- Weight loss
- Hematemesis
- Dysphagia
- Others…

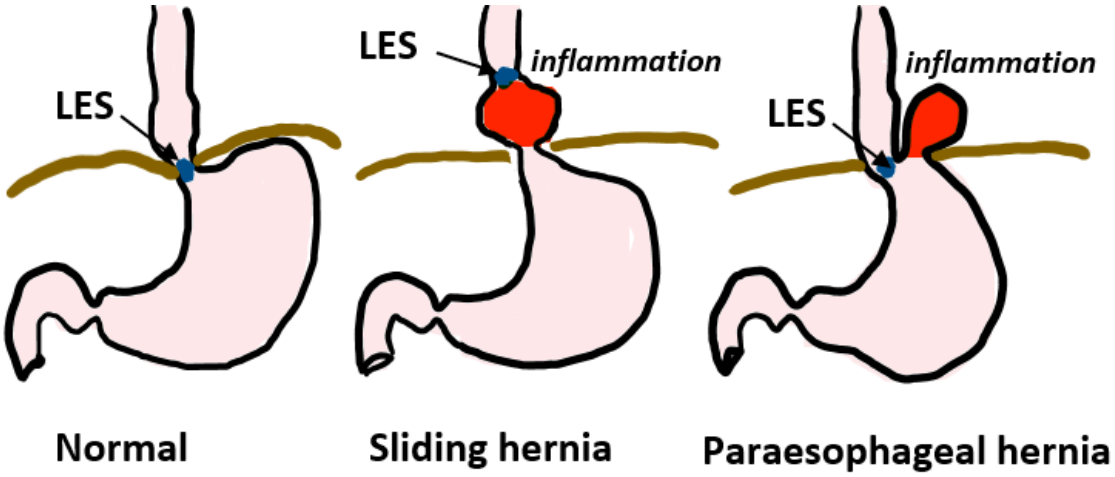
Pathophysiology

- Gastroesophageal junction incompetence is central to GERD with 3 possible abnormal processes:

- Abnormal lower esophageal sphincter (LES) relaxation for burping
    - Burping is a complex function that includes esophageal peristalsis to clear acid. Transient esophageal relaxation that is not coordinated with other functions fails to clear the acid.
- Poor tone in the LES
    - This can be associated with medications, certain foods and obesity.
    - Fibrosis of the esophagus is seen in systemic rheumatic diseases like scleroderma and rheumatoid arthritis.
- Hiatal hernias
    - There is a ligamentous attachment of the lower esophagus at the LES to the diaphragmatic crura. The crura act as a "pinchcock". With age, laxity of the ligament leads to rise of the esophagus into the chest, called a **sliding hernia**. Acid gets trapped in the supradiaphragmatic segment of the esophagus between the LES and the diaphragm. Sliding hernias constitute 95% of hiatal hernias and have a high prevalence.



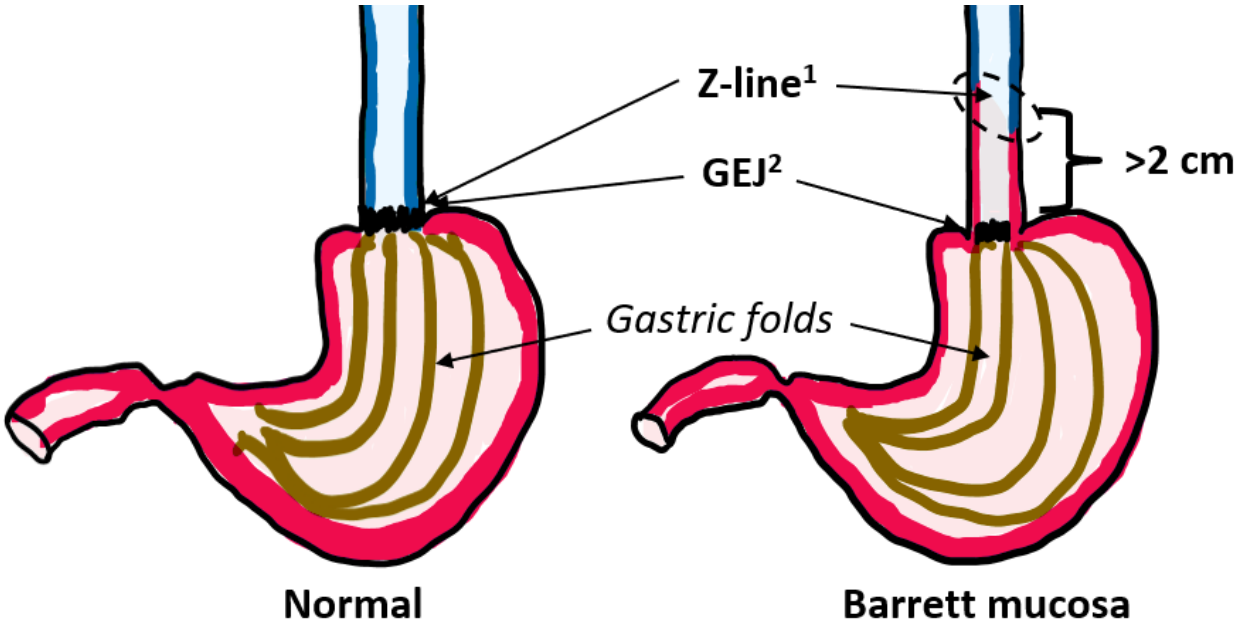
    - The second type of hiatal hernia is **paraesophageal hernia**. In this, the fundus of the stomach slides through the diaphragmatic hiatus into the thorax. Acid and gastric contents get trapped in the hernia causing inflammation. While only 5% of hiatal hernias, these are more clinically dangerous as the fundal segment is at risk of incarceration and necrosis



Natural history

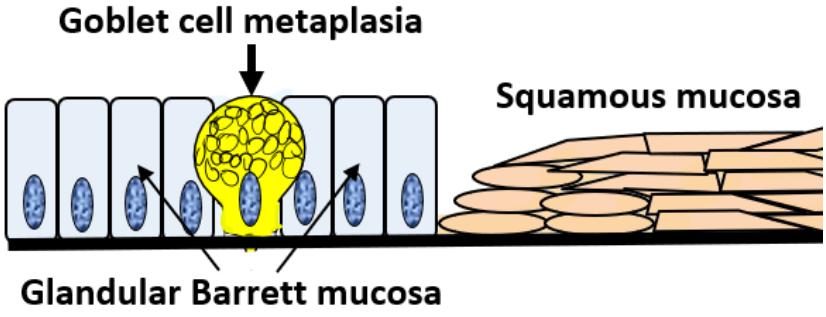
- Treatment
    - Lifestyle and diet modification
        - Weight loss as obesity causes an increase in intra-abdominal pressure
        - Raising the head of the bed
    - Acid inhibition: H2 blockers are cheaper; PPIs are more effective but are more expensive and have more side effects
- Complications:

- Barrett mucosa, goblet cell metaplasia and the risk of adenocarcinoma
    - Barrett mucosa is a metaplasia in which the squamous mucosa becomes a glandular mucosa. It is diagnosed when the histologic squamocolumnar junction, called the Z-line, is > 2 cm above the gastroesophageal junction, defined by the end of the gastric folds.



1. Squamocolumnar junction, 2. Gastroesophageal junction

    - Goblet cell metaplasia
        - Barrett mucosa is a glandular metaplasia (i.e. the normal squamous epithelium is regrown as a glandular epithelium)
        - Goblet cell metaplasia is a specific type of glandular metaplasia in which the new cell is a mucin-producing cell with a microscopic shape suggestive of a drinking glass, a “goblet”. It is this type of metaplasia that is thought to carry the highest risk of adenocarcinoma.

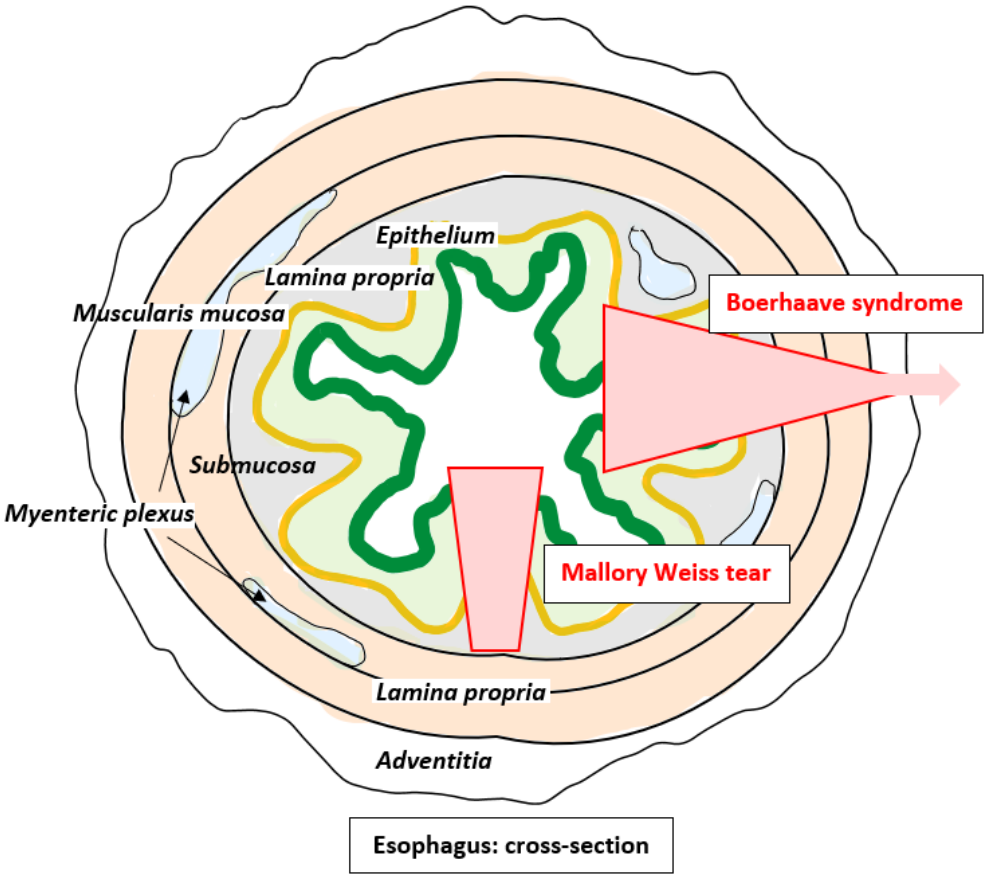


Mallory-Weiss syndrome

- Pathophysiology:
  - Longitudinal tears in the mucosa around the gastroesophageal junction can be caused by forceful vomiting. Bleeding is from torn submucosal blood vessels
  - This type of vomiting is seen with alcohol use and bulimia
- Presentation:
  - Symptoms: hematemesis ± pain
  - Physical examination: assess severity of blood loss
  - Testing: endoscopy when stable
- Natural history:
  - Prognosis: rapid healing as tearing only involves the mucosa
  - Complications: hemodynamic instability from acute blood loss
  - Treatment:
    - Endoscopic control of severe bleeding.
    - Proton pump inhibitors

Boerhaave syndrome

- Pathophysiology:
  - Effort rupture of the distal esophagus seen in vomiting or severe straining. Perforation of the esophagus leaks contents into the mediastinum with life-threatening acute mediastinitis.
- Presentation:
  - Symptoms: acute onset of chest pain that may have radiate into the back. Less than 50% of patients can describe an antecedent event.
  - P/E: tactile crepitus of the chest wall due to pneumomediastinum
  - Testing: imaging to identify the perforation and leakage.



- Natural history:
  - Prognosis: fatal without treatment
  - Treatment: surgical repair of esophageal perforation, antibiotics for mediastinitis.

Degenerative / autoimmune diseases

Achalasia

- Presentation

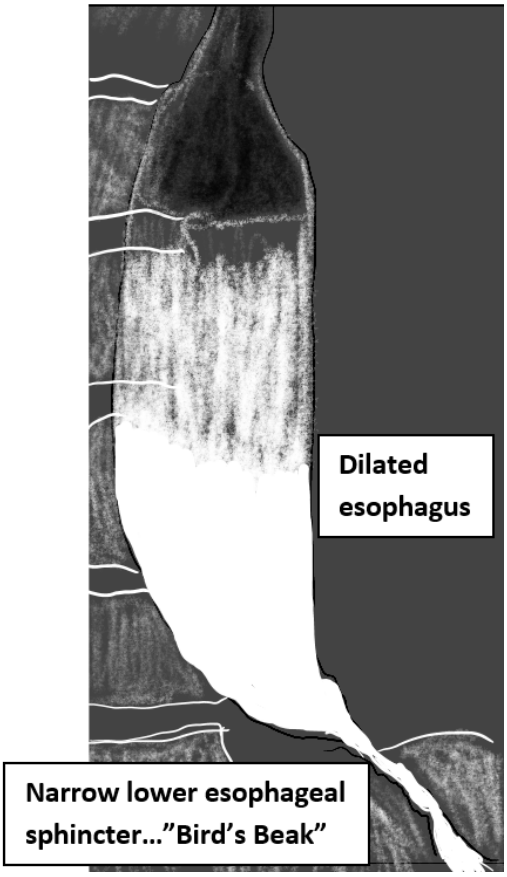
- Symptoms:
    - Dysphagia of both **liquids and solids**
        - The buildup of food in the esophagus may lead to aspiration
        - The inability of relax the lower esophageal sphincter (LES) leads to bloating as the patient cannot burp.
- Signs: the physical examination is non-contributory
- Diagnosis:
    - Barium swallow: not widely used anymore as it is non-specific. It shows dilation of the proximal esophagus and narrow gastroesophageal junction (EGJ) causing the "bird's beak sign".
    - Esophagoscopy: to rule out other disease such as cancer.
    - Motililty studies (manometry): loss of peristalsis in the distal 2/3 of the esophagus with inabiliity to relax the LES

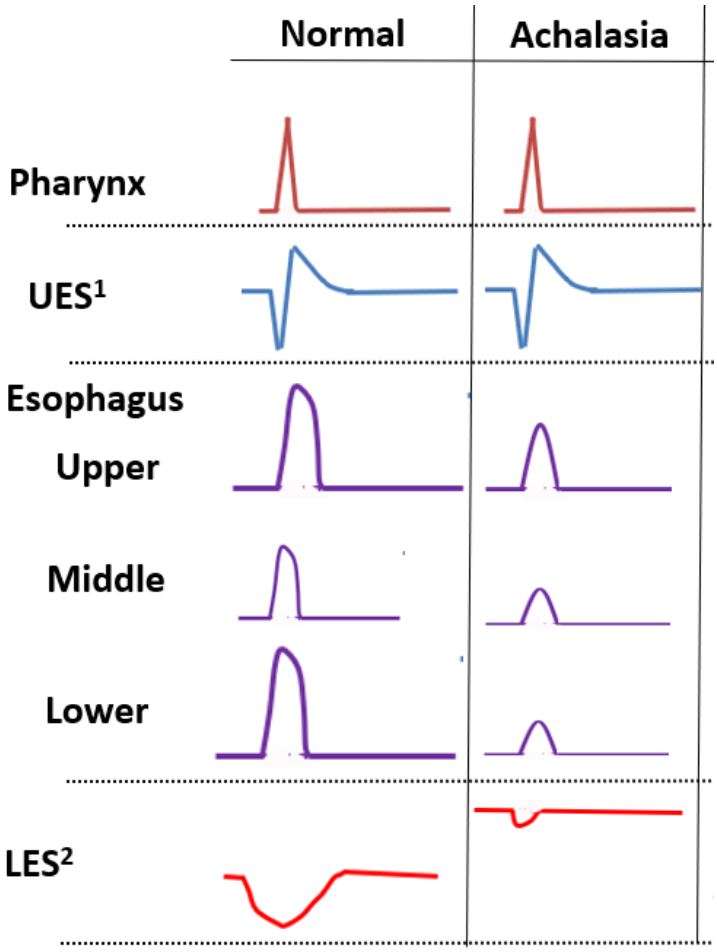
The History in Dysphagia

It is important to differentiate difficulty swallowing solids versus difficulty swallowing both solids and liquids.

a. Solids only : mechanical defect. Endoscopy to R/O cancer
b. Solids and liquids: motor defect. Motility studies such as manometry.

Achalasia: barium swallow





1. Upper esophageal sphincter,
2. Lower esophageal sphincter

Manometry

The propulsive pressure wave of peristalsis starts in the pharynx and ends with relaxation of the LES.

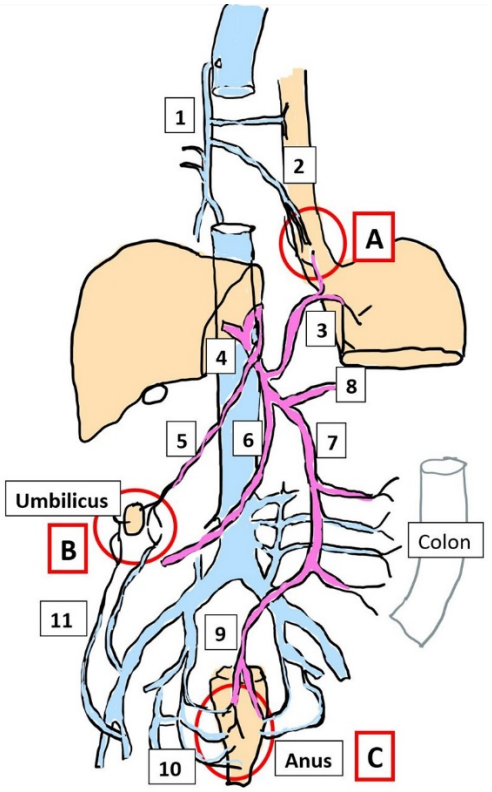
In achalasia, there is loss of peristalsis in the distal esophagus and a high pressure LES that cannot relax

- Pathophysiology
  - Achalasia is caused by a loss of neurons in the myenteric plexus of the esophagus. There is selective loss of inhibitory neurons, which allow for relaxation. This causes increased resting tone in the LES and its inability to relax.
  - Loss of neurons is presumed to be autoimmune.
    - The one known cause of myenteric nerve loss is infection with *Trypanosoma cruzi* which causes **Chagas disease**.
      - There should be a history of travel to rural Latin America
- Natural history
  - Without treatment there is progressive dilation of the esophagus
  - Treatment consists of manual dilation or Botox injections of the LES.

Esophageal varices

- Pathophysiology:

- Porto-systemic anastomosis arise with portal hypertension. The increased volume and pressure dilate submucosa veins in the lower third of the esophagus. These are prone to rupture.



	Vein
1	Azygous
2	Esophageal
3	Left gastric
4	Portal
5	Paraumbilical
6	Superior mesenteric
7	Inferior mesenteric
8	Splenic
9	Superior rectal
10	Inferior rectal
11	Epigastric
A	Esophageal varices
B	Caput medusa
C	Internal hemorrhoids

- Presentation:
  - Symptoms: hematemesis
  - P/E: cirrhosis (portosystemic anastomoses, splenomegaly, and ascites) and elevated estrogen (feminization of males, palmar erythema, spider angiomas)
  - Testing: endoscopy
- Natural history
  - Prognosis: one-third of patients die with each bleeding episode
  - Treatment:
    - Hemodynamic support (IV fluids, blood transfusion)
    - Urgent endoscopy for bleeding control
    - Drugs to decrease portal blood pressure (e.g. octreotide, vasopressin, somatostatin)

Neoplasia

- Most malignancies of the esophagus arise in the epithelium and therefore are carcinomas. **Squamous cell carcinomas (SCC)** arise from the squamous epithelium and are mostly found in the mid-esophagus. **Adenocarcinomas** mostly arise in Barrett mucosa (see GERD, above) and found around the gastroesophageal junction.

	Squamous cell carcinoma	Adenocarcinoma
Epidemiology	·More common outside of the US ·M > F ·Age > 50 years	·More common in US ·M > F ·Age >40 years
Risk factors	·Smoking & alcohol (synergistic) ·HPV ·Achalasia	GERD → Barrett mucosa
Location	Mid-esophagus	Distal esophagus

- Presentation:
  - Symptoms: dysphagia and weight loss

> **Dysphagia: solids versus liquids**
>
> When the pathology is mechanical, such as a malignant mass, patients describe difficulty swallowing solids but not liquids as the fluid can flow around the mass. When the pathology is due to a problem with peristalsis, such as achalasia, then both liquids and solids are difficult to swallow.

  - P/E: non-contributory
  - Testing: endoscopy for mass with biopsy confirmation
- Natural history:
  - Prognosis: depends on stage. >50% of patients present with incurable disease due to early spread to paraesophageal lymph nodes
    - Survival drops below 50% 5 year survival when disease is ≥ Stage II (extension into adventitia or lymph node involvement)
  - Treatment: surgical resection for early disease. Palliative for the rest.